LAVORI PER ESTESO PUBBLICATI SU RIVISTE RECENSITE

ANNO 2009

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Balconi Michela, Pozzoli Uberto (2009); AROUSAL EFFECT ON EMOTIONAL FACE COMPREHENSION. FREQUENCY BAND CHANGES IN DIFFERENT TIME INTERVALS; Physiology & Behavior, 97(3-4):455-462

I.F. 2008: 2,806

The study aimed to explore the modulation of frequency bands (delta, theta, alpha-1 and alpha-2, and gamma) in response to emotional face within different post-stimulus time intervals (50-450 ms). Twenty adults looked at emotional (happy, sad, angry, fearful) or neutral faces. EEG results showed that motivational significance of face can modulate frequency bands, specifically for theta and gamma. Moreover, gamma can be varied related to degree of arousing feature (high or low) of facial expression. As a function of time, ANOVA and regression analysis revealed that emotional discrimination by gamma and theta is observable mainly within 150-250 time interval and, as revealed also by coherence analysis, that it is more distributed on the anterior-right (theta) or right (gamma) side of the scalp for the emotional stimuli, whereas delta is maximally increased within 250-350 interval and it is more posteriorly (parietal site) distributed for all the stimulus type. We proposed that band modulations respond to variations in processing emotional face, and, whereas delta reflects updating of the stimulus, and theta responds to the emotional significance of face, gamma reflects differences in the arousing power of facial expression.

Baloch Hasan A., Brambilla Paolo, Soares Jair C. (2009); CORPUS CALLOSUM ABNORMALITIES IN PEDIATRIC BIPOLAR DISORDER; Expert Review of Neurotherapeutics, 9(7):949-955

I.F. 2008: 0,000

The corpus callosum (CC) is a midline white matter brain region that is important in interhemispheric communication and coordination. CC abnormalities are associated with a variety of psychiatric conditions, including increased vulnerability for psychotic illness, stressful early-life experiences, marijuana use, attention-deficit/hyperactivity disorder, obsessive-compulsive disorder, borderline personality disorder, dementia, schizophrenia and bipolar disorder. CC abnormalities in bipolar disorder have been identified in both pedi-

atric and adult populations. In adults, a consistent finding has been a reduction in CC size, as well as abnormal axonal orientation or structure. Axonal abnormalities have also been noted in pediatric populations, but overall CC size reductions have not thus far been demonstrated. Furthermore, there are unique gender differences in the expression of CC abnormalities in pediatric populations, possibly related to androgen changes during puberty. The protean number of conditions in which the CC is involved is reflective of its central role in normal brain function and its potential as an early marker of neuropathology in psychiatric illness. Specifically, in bipolar disorder it has the potential to be useful as an early preclinical marker of disease or disease risk.

Battaglia Marco, Pesenti-Gritti Paola, Medland Sarah E., Ogliari Anna, Tambs Kristian, Spatola Chiara A.M. (2009); A GENETICALLY INFORMED STUDY OF THE RELATIONSHIPS BETWEEN CHILDHOOD SEPARATION ANXIETY, SENSITIVITY TO CO2, PANIC DISORDER, AND THE IMPACT OF CHILDHOOD PARENTAL LOSS; Archives of General Psychiatry, 66(1):64-71

I.F. 2008: 14,273

CONTEXT: Childhood separation anxiety disorder can predate panic disorder, which usually begins in early adulthood. Both disorders are associated with heightened sensitivity to inhaled CO(2) and can be influenced by childhood parental loss. OBJECTIVES: To find the sources of covariation between childhood separation anxiety disorder, hypersensitivity to CO(2), and panic disorder in adulthood and to measure the effect of childhood parental loss on such covariation. DESIGN: Multivariate twin study. PARTICIPANTS: Seven hundred twelve young adults from the Norwegian Institute of Public Health Twin Panel, a general population cohort. MAIN OUTCOME MEASURES: Personal direct assessment of lifetime panic disorder through structured psychiatric interviews, history of childhood parental loss, and separation anxiety disorder symptoms. Subjective anxiety response to a 35% CO(2)/65% O(2) inhaled mixture compared with compressed air (placebo). RESULTS: Our best-fitting solution yielded a common pathway model, implying that covariation between separation anxiety in childhood, hypersensitivity to CO(2), and panic disorder in adulthood can be explained by a single latent intervening variable influencing all phenotypes. The latent variable governing the 3 phenotypes' covariation was in turn largely (89%) influenced by genetic factors and childhood parental loss (treated as an identified element of risk acting at a family-wide level), which accounted for the remaining 11% of covariance. Residual variance was explained by 1 specific genetic variance component

for separation anxiety disorder and variable-specific unique environmental variance components. CONCLUSIONS: Shared genetic determinants appear to be the major underlying cause of the developmental continuity of childhood separation anxiety disorder into adult panic disorder and the association of both disorders with heightened sensitivity to CO(2). Inasmuch as childhood parental loss is a truly environmental risk factor, it can account for a significant additional proportion of the covariation of these 3 developmentally related phenotypes.

Bellani Marcella, Marzi Carlo Alberto, Brambilla Paolo (2009); INTERHEMISPHERIC COMMUNICATION IN SCHIZOPHRENIA; Epidemiologia e Psichiatria Sociale, 18(1):19-22

I.F. 2008: 2,180

Abstract non disponibile

Bellani Marcella, Tomelleri Luisa, Brambilla Paolo (2009); EMOTION-BASED DECISION MAKING IN SCHIZOPHRENIA: EVIDENCE FROM THE IOWA GAMBLING TASK; Epidemiologia e Psichiatria Sociale, 18(2):104-106

I.F. 2008: 2,180

Abstract non disponibile

Bellani Marcella, Fagnani Corrado, Brambilla Paolo (2009); TWIN STUDIES IN PSYCHOTIC DISORDERS; Epidemiologia e Psichiatria Sociale, 18(3):195-199

I.F. 2008: 2,180

Abstract non disponibile

Bellani Marcella, Perlini Cinzia, Brambilla Paolo (2009); LANGUAGE DISTURBANCES IN SCHIZOPHRENIA; Epidemiologia e Psichiatria Sociale, 18(4):314-317

I.F. 2008: 2,180

Language disturbances represent a core feature of schizophrenia, affecting social interactions and quality of life. Here we summarize linguistic and pragmatic deficits and illustrate the role of brain imaging studies in delineating the neural substrates of language deficits in patients with schizophrenia.

Bellani Marcella, Yeh Ping-Hong, Tansella Michele, Balestrieri Matteo, Soares Jair C., Brambilla Paolo (2009); DTI STUDIES OF

CORPUS CALLOSUM IN BIPOLAR DISORDER; Biochemical Society Transactions, 37(Pt 5):1096-1098

I.F. 2008: 2,979

Although the pathogenesis of bipolar disorder is still not completely understood, there is evidence from imaging studies that abnormalities in interhemispheric communication may play a major role in the pathophysiology of bipolar disorder. In the present review, we discuss the most consistent findings from diffusion imaging studies exploring corpus callosum integrity in bipolar disorder.

Beretta Elena, Cimolin Veronica, Piccinini Luigi, Turconi Anna Carla, Galbiati Sara, Crivellini Marcello, Galli Manuela, Strazzer Sandra (2009); ASSESSMENT OF GAIT RECOVERY IN CHILDREN AFTER TRAUMATIC BRAIN INJURY; Brain Injury, 23(9):751-759

I.F. 2008: 1,116

OBJECTIVE: The aims were: (1) to quantify the functional limitation of children at the beginning of recovery of independent ambulation after Traumatic Brain Injury (TBI), using clinical-functional scales; (2) to evaluate the changes in gait pattern during rehabilitation (about 5 months later), using 3D Gait Analysis (GA) in post-acute phase; (3) to investigate the presence of correlation among parameters obtained by 3DGA, clinical assessment and measures connected with the trauma. METHODS: Fourteen children with hemiplegia after severe TBI were evaluated at independent gait recovery (S0) and 5.5 months later (S1) by clinical assessment (GOS, DRS, WeeFIM and GMFM) and 3D GA (spatio-temporal parameters, kinematics and kinetics). RESULTS: At S1 all clinical measures had improved. Regarding spatio-temporal parameters, velocity and step length improved. Significant progress was evident at the ankle joint, while an unchanged condition appeared at pelvis and hip in sagittal plane with a worsening of hip rotation which increased its internal rotation. Significant correlations were found between motor performance, clinical assessment and trauma-related measures. CONCLUSIONS: Repeated GA and clinical evaluations were useful in quantifying the motor recovery of children with TBI during rehabilitation underpinning the role of GA in quantifying these modifications in an objective and non-invasive way.

Bersano Anna, Del Bo Roberto, Lamperti Costanza, Ghezzi Serena, Fagiolari Gigliola, Fortunato Francesco, Ballabio Elena, Moggio Maurizio, Candelise Livia, Galimberti Daniela, Virgilio Roberta, Lanfranconi Silvia, Torrente Yvan, Carpo Marinella, Bresolin Nereo, Comi Giacomo Pietro, Corti Stefania (2009); INCLUSION BODY

MYOPATHY AND FRONTOTEMPORAL DEMENTIA CAUSED BY A NOVEL VCP MUTATION; Neurobiology of Aging, 30(5):752-758

I.F. 2008: 5,959

Hereditary inclusion body myopathy (IBM) with Paget's disease of the bone (PDB) and frontotemporal dementia (FTD) is a rare autosomal dominant disease caused by mutations in the valosin-containing protein (VCP) gene. We report a novel heterozygous VCP gene mutation (R159C) in a 69-year-old Italian patient presenting with slowly progressive muscle weakness of the distal upper and proximal lower limbs since the age of 50 years, 18 years later FTD supervened. No dementia or myopathies were revealed in the family history covering two generations. Degenerative changes and rimmed vacuoles together with VCP- and ubiquitin-positive cytoplasmic and nuclear aggregates were observed at the muscle biopsy. Several elements support the pathogenic role of the R159C VCP gene mutation: the occurrence at the same codon of a different, previously identified pathogenic mutation within a VCP gene mutational hot-spot, the histopathological and biochemical evidence of muscle VCP accumulation and the combined clinical presentation of IBM and FTD. These findings suggest VCP gene investigation even in apparently sporadic cases.

Bianco Fabio, Perrotta Cristiana, Novellino Luisa, Francolini Maura, Riganti Loredana, Menna Elisabetta, Saglietti Laura, Schuchman Edward H., Furlan Roberto, Clementi Emilio, Matteoli Michela, Verderio Claudia (2009); ACID SPHINGOMYELINASE ACTIVITY TRIGGERS MICROPARTICLE RELEASE FROM GLIAL CELLS; The EMBO Journal, 28(8):1043-1054

I.F. 2008: 8,295

We have earlier shown that microglia, the immune cells of the CNS, release microparticles from cell plasma membrane after ATP stimulation. These vesicles contain and release IL-1beta, a crucial cytokine in CNS inflammatory events. In this study, we show that microparticles are also released by astrocytes and we get insights into the mechanism of their shedding. We show that, on activation of the ATP receptor P2X7, microparticle shedding is associated with rapid activation of acid sphingomyelinase, which moves to plasma membrane outer leaflet. ATP-induced shedding and IL-1beta release are markedly reduced by the inhibition of acid sphingomyelinase, and completely blocked in glial cultures from acid sphingomyelinase knockout mice. We also show that p38 MAPK cascade is relevant for the whole process, as specific kinase inhibitors strongly reduce acid sphingomyelinase activation, microparticle shedding and IL-1beta release. Our results represent the

first demonstration that activation of acid sphingomyelinase is necessary and sufficient for microparticle release from glial cells and define key molecular effectors of microparticle formation and IL-1beta release, thus, opening new strategies for the treatment of neuroinflammatory diseases.

Bonaglia Maria Clara*, Giorda Roberto*, Massagli Angelo, Galluzzi Rita, Ciccone Roberto, Zuffardi Orsetta (2009); A FAMILIAL INVERTED DUPLICATION/DELETION OF 2P25.1 PROVIDES NEW CLUES ON THE GENESIS OF INVERTED DUPLICATIONS; European Journal of Human Genetics, 17(2):179-186

I.F. 2008: 3,925

**Autori che hanno contribuito in ugual misura al lavoro*

We studied a family in which the same 10 Mb inverted duplication of 2p25.3-p25.1 segregates in two children and their father, all showing a trisomy phenotype. As FISH analysis demonstrated that the duplication was inverted, we suspected that a contiguous terminal deletion was also present, according to the classical inv dup del type of rearrangements. Although FISH with 2p and 2q subtelomeric probes gave normal results, 100 kb resolution array-C/GH (aCGH) showed that, beside the duplication, a 273 kb deletion was also present. The presence of a single-copy region between the deleted and duplicated regions was further suspected through high-resolution aCGH analysis (approximately 20 kb), although only one informative spot having a normal log ratio was detected. The precise structure of the rearrangement was re-defined by real-time PCR and breakpoint cloning, demonstrating the presence of a 2680 bp single-copy sequence between deleted and duplicated regions and the involvement of a simple repeat with the potential for forming a non-B DNA structure. The rearrangement was not mediated by segmental duplications or short inverted repeats, and the double-strand break might have been repaired by non-homologous end joining or microhomology-mediated intrastrand repair. These data highlight the fact that concomitant deletions associated with inverted duplications are very likely to be more frequent than classical cytogenetic methods alone have been able to demonstrate. The phenotypic effects of the trisomy and of the terminal 2p deletion are discussed.

Bonaglia Maria Clara*, Giorda Roberto*, Beri Silvana, Bigoni Stefania, Sensi Alberto, Baroncini Anna, Capucci Antonella, De Agostini Cristina, Gwilliam Rhian, Deloukas Panos, Dunham Ian, Zuffardi Orsetta (2009); MOSAIC 22Q13 DELETIONS: EVIDENCE FOR CONCURRENT MOSAIC SEGMENTAL ISODISOMY AND GENE

CONVERSION; European Journal of Human Genetics, 17(4):426-433

I.F. 2008: 3,925

**Autori hanno contribuito in ugual misura al lavoro*

Although 22q terminal deletions are well documented, very few patients with mosaicism have been reported. We describe two new cases with mosaic 22q13.2-qter deletion, detected by karyotype analysis, showing the neurological phenotype of 22q13.3 deletion syndrome. Case 1 represents an exceptional case of mosaicism for maternal 22q13.2-qter deletion (45% of cells) and 22q13.2-qter paternal segmental isodisomy (55% of cells). This complex situation was suspected because cytogenetic, FISH and array-CGH analyses showed the presence of an 8.8 Mb mosaic 22q13.2-qter deletion, whereas microsatellite marker analysis was consistent with maternal deletion without any evidence of mosaic deletion. Molecular analysis led to the definition of very close, but not coincident, deletion and uniparental disomy (UPD) break points. Furthermore, we demonstrated that the segmental UPD arose by gene conversion in the same region. In Case 2, mosaicism for a paternal 8.9 Mb 22q13.2-qter deletion (73% of cells) was detected. In both patients, the level of mosaicism was also verified in saliva samples. We propose possible causative mechanisms for both rearrangements. Although the size of the deletions was quite similar, the phenotype was more severe in Case 2 than in Case 1. As maternal UPD 22 has not been generally associated with any defects and as the size of the deletion is very similar in the two cases, phenotype severity is likely to depend entirely on the degree of mosaicism in each individual.

Bonanni Paolo, Gobbo Annamaria, Nappi Sara, Moret Ornella, Nogarol Anita, Santin Michela, Randazzo Giovanna, Martinuzzi Andrea (2009); FUNCTIONING AND DISABILITY IN PATIENTS WITH ANGELMAN SYNDROME: UTILITY OF THE INTERNATIONAL CLASSIFICATION OF FUNCTIONING DISABILITY AND HEALTH, CHILDREN AND YOUTH ADAPTATION FRAMEWORK; Disability and Rehabilitation, 31(S1):S121-S127

I.F. 2008: 1,395

Purpose. Angelman syndrome (AS) accounts for upto 6% of all cases with severe mental retardation and epilepsy. Clinical findings include developmental delay, severely impaired findings include developmental delay, severely impaired expressive language, ataxic gait, tremulousness of limbs and a typical behavioral profile including a happy demeanour, hyperactive behavior and low attention span. Seizures, abnormal electroencephalogram, microcephaly and scoliosis are observed in 480% of patients. Cognitive, lan-

guage and orthopedic problems must be addressed with vigorous rehabilitation programs. Classification of functioning disability and health, children and youth adaptation (ICF-CY) can provide the most adequate framework to describe the condition of the persons towards whom rehabilitative efforts are concentrated. The aim of the study is to test whether the ICF-CY framework is effectively able to capture the various dimensions of health in AS.
Methods. We applied the ICF-CY, to the detail of second level codes, to a cohort of 11 patients with AS of various ages. The coding was obtained by the multi-professional team following these children for the rehabilitation program.
Results. The functional profile obtained applying the ICF-CY is complete and comparable with the characteristics of the syndrome described in literature. The possibility of highlighting not only the problems but also the points of strength appears as very helpful. The second level codes may be too broad to provide insight in the clinical and rehabilitative peculiarities, and
the use of the full power of the classification may be more advisable for clinical use.
Conclusions. This prelimiary study shows that ICF-CY is a valid tool to frame the clinical characteristics of a complex syndrome as AS, and may give a strong foundation for the rehabilitation programming.

Borgnolo Giulio, Soares Isabel Cristina, Dos Santos Soares Benilde, Gongolo Francesco, Vaz Paula, Meucci Paolo, Quintas Rui, Lembo Rosalba, Martinuzzi Andrea (2009); PRELIMINARY RESULTS OF ICF DISSEMINATION IN PRIMARY HEALTH CARE IN MOZAMBIQUE: SHARING THE ITALIAN EXPERIENCE; Disability and Rehabilitation, 31(S1):S78-S82

I.F. 2008: 1,395

Purpose. To present the preliminary results of training courses on the International Classification of Functioning, Disability and Health (ICF) prepared by ItalianWHO FIC CC and to report on strategies for ICF dissemination among primary health care (PHC) workers in Mavalane Health Area, Maputo, Mozambique.
Methods. A participant-observer description of ICF principles introduction and development of ICF-CY modified checklists in Mavalane health area according to a person-environment interaction classification (PEIC) tree.
Results. The first ICF courses were held atMavalane hospital and involved 25 health workers and 24 members of the health committee acting in the local area. The courses were prepared having as a framework the theoretical principles of the UN Convention of rights of persons with disabilities and the

ICF bio-psychosocial model. The trainers adapted and modified the course materials to increase the applicability of the protocol to the cultural and social reality of Mozambique, eventually applying the person-environment interaction classification (PEIC) tree.
Conclusions. The very first phase of this project indicates that the use of the ICF checklist is feasible also in a crowded and busy environment like some PHC units of a developing country. However, data collection require a simpler and lighter to use data compilation tool. The proposed new checklist, which includes a PEIC tree, makes easier the compilation and the collection of data.

Brambilla Paolo, Bellani Marcella, Yeh Ping-Hong, Soares Jair C. (2009); MYELINATION IN BIPOLAR PATIENTS AND THE EFFECTS OF MOOD STABILIZERS ON BRAIN ANATOMY; Current Pharmaceutical Design, 15(1):2632-2636

I.F. 2008: 4,399

In this review, we debate the evidence for abnormal white matter microstructure and myelination in bipolar disorder, as mainly detected by diffusion tensor imaging (DTI) studies. The effects of mood stabilizers on white matter are discussed, based on available findings from human and animal studies. Last, perspectives in this field of research are also drawn.

Brambilla Paolo, Bellani Marcella, Yeh Ping-Hong, Soares Jair C., Tansella Michele (2009); WHITE MATTER CONNECTIVITY IN BIPOLAR DISORDER; International Review of Psychiatry, 21(4):380-386

I.F. 2008: 1,563

There is evidence that intra- and inter-hemispheric white matter communication, mainly fronto-limbic and callosal connectivity, is impaired in bipolar disorder as reported in several magnetic resonance (MR) diffusion imaging studies. In this review we will discuss diffusion imaging studies that examined white matter integrity in patients with bipolar disorder, trying to outline future research strategies

Brambilla Paolo, Tansella Michele (2009); WHAT ARE THE PERSPECTIVES OF HUMAN BRAIN MAPPING IN THE FIELD OF BIPOLAR DISORDER?; International Review of Psychiatry, 21(4):295-296

I.F. 2008: 1,563

Abstract non disponibile

Bresolin Nereo, Zucca Claudio, Pecori A. (2009); EFFICACY AND TOLERABILITY OF EPERISONE AND BACLOFEN IN SPASTIC PALSY: A DOUBLE-BLIND RANDOMIZED TRIAL; Advances in Therapy, 26(5):563-573

I.F. 2008: 0,973

INTRODUCTION: Few trials have compared different central muscle relaxants in the treatment of spastic palsy. This head-to-head phase 3 trial compares oral eperisone, a central muscle relaxant with a promising activity in spasticity therapy, and oral baclofen. METHODS: Patients (>18 years) with moderate to severe spastic palsy were eligible in this double-blind, randomized study; they received eperisone 300 mg/ day or baclofen 60 mg/day for 6 weeks. The efficacy evaluations included: functional analysis (Pedersen's scale, muscular tone, joint range of motion, 10-meter walking time); physiological and pathological reflexes; and electromyography (Hmax/Mmax amplitude ratio and the Wartenberg test). Physicians and patients globally assessed treatment efficacy. RESULTS: Both eperisone (n=40) and baclofen (n=40) significantly improved functionality of lower limbs versus baseline (eperisone: -9.1%, P<0.01; baclofen: -8.3%, P<0.05), but only eperisone improved this parameter in the upper limbs (-7.8%, P<0.01 vs. -6.3%, P=NS). Both drugs reduced muscular tone from week 2. Only eperisone improved the joint range of motion (-32.5%, P<0.01 vs. -14.6%, P=NS). Both treatments reduced the 10-meter walking time (eperisone: -20.2%, P<0.01; baclofen: -24.0%, P<0.01); this effect was evident at week 2 with eperisone only. Both drugs improved reflexes. Eperisone and baclofen decreased the Hmax/Mmax amplitude ratio (eperisone: -30.0%, baclofen: -18.6%; P<0.01 for both). Eperisone increased the number of leg oscillations at the Wartenberg test (P<0.05) while baclofen increased the velocity of leg falling (P<0.01). For tolerability, no differences were observed between eperisone and baclofen in any parameters. Eperisone was judged as "good" by a higher number of physicians and patients than baclofen. Eighteen adverse events, most of mild intensity, were reported with eperisone and 27 with baclofen. CONCLUSION: Eperisone 300 mg/day and baclofen 60 mg/day, administered orally, are effective and well-tolerated drugs in the treatment of spastic palsy. However, eperisone might be associated with some additional clinical benefits when compared with baclofen.

Bresolin Nereo, Zucca Claudio, Pecori A. (2009); EFFICACY AND TOLERABILITY OF EPERISONE IN PATIENTS WITH SPASTIC PALSY: A CROSS-OVER, PLACEBO-CONTROLLED DOSE-RANGING TRIAL;

European Review for Medical and Pharmacological Sciences, 13(5):365-370

I.F. 2008: 0,000

BACKGROUND AND OBJECTIVES: Central muscle relaxants are a clinical option in patients with spastic palsy. Eperisone is a central muscle relaxant used in several conditions, but its therapeutic potential in spastic palsy needs to be verified. This dose-ranging trial compares two doses of eperisone in patients with spastic palsy associated to cerebral or spinal diseases. PATIENTS AND METHODS: In this randomized, placebo-controlled, double-blind, three-way cross-over study, patients (18-75 years) with spastic palsy received eperisone 150 mg/day, eperisone 300 mg/day, or placebo for 8 weeks. Treatment periods lasted for 14 days. Objective clinical parameters (intensity of spasticity and physiological reflexes) and functional parameters (walking capability, capability to climb stairs, rigidity) were measured. Tolerability was also evaluated. RESULTS: Eighteen patients were enrolled. The reduction in the intensity of spasticity versus the beginning of each treatment cycle was significant with eperisone 300 mg/day ($p = 0.004$). Similar findings were observed in the evaluation of patellar reflex ($p = 0.01$), while the other reflexes were not significantly different. Walking capability was significantly improved with eperisone 300 mg/day ($p < 0.05$). No significant differences were observed in the capability to climb stairs and in rigidity. A trend towards a reduction in pain was noted with eperisone 300 mg/day versus placebo. The incidence of adverse events was similar in all groups. DISCUSSION: Eperisone 300 mg/day might be an effective and well-tolerated treatment for spastic palsy. Larger studies are required to further characterize the efficacy of eperisone in this therapeutic area.

Cagliani Rachele, Fumagalli Matteo, Pozzoli Uberto, Riva Stefania, Cereda Matteo, Comi Giacomo Pietro, Pattini Linda, Bresolin Nereo, Sironi Manuela (2009); A COMPLEX SELECTION SIGNATURE AT THE HUMAN AVPR1B GENE; BMC Evolutionary Biology, 9:123

I.F. 2008: 4,050

BACKGROUND: The vasopressin receptor type 1b (AVPR1B) is mainly expressed by pituitary corticotropes and it mediates the stimulatory effects of AVP on ACTH release; common AVPR1B haplotypes have been involved in mood and anxiety disorders in humans, while rodents lacking a functional receptor gene display behavioral defects and altered stress responses. RESULTS: Here we have analyzed the two exons of the gene and the data we present suggest that AVPR1B has been subjected to natural selection in humans. In particular, analysis of exon 2 strongly suggests the action of balanc-

ing selection in African populations and Europeans: the region displays high nucleotide diversity, an excess of intermediate-frequency alleles, a higher level of within-species diversity compared to interspecific divergence and a genealogy with common haplotypes separated by deep branches. This relatively unambiguous situation coexists with unusual features across exon 1, raising the possibility that a nonsynonymous variant (Gly191Arg) in this region has been subjected to directional selection. CONCLUSION: Although the underlying selective pressure(s) remains to be identified, we consider this to be among the first documented examples of a gene involved in mood disorders and subjected to natural selection in humans; this observation might add support to the long-debated idea that depression/low mood might have played an adaptive role during human evolution.

Cagliani Rachele, Fumagalli Matteo, Pozzoli Uberto, Riva Stefania, Comi Giacomo Pietro, Torri Federica, Macciardi Fabio, Bresolin Nereo, Sironi Manuela (2009); DIVERSE EVOLUTIONARY HISTORIES FOR BETA-ADRENORECEPTOR GENES IN HUMANS; American Journal of Human Genetics, 85(1):64-75

I.F. 2008: 10,153

In humans, three genes--ADRB1, ADRB2 and ADRB3--encode beta-adrenoreceptors (ADRB); these molecules mediate the action of catecholamines in multiple tissues and play pivotal roles in cardiovascular, respiratory, metabolic, and immunological functions. Genetic variants in ADRB genes have been associated with widespread diseases and conditions, but inconsistent results have often been obtained. Here, we addressed the recent evolutionary history of ADRB genes in human populations. Although ADRB1 is neutrally evolving, most tests rejected neutral evolution for ADRB2 in European, African, and Asian population samples. Analysis of inferred haplotypes for ADRB2 revealed three major clades with a coalescence time of 1-1.5 million years, suggesting that the gene is either subjected to balancing selection or undergoing a selective sweep. Haplotype analysis also revealed ethnicity-specific differences. Additionally, we observed significant deviations from Hardy-Weinberg equilibrium (HWE) for ADRB2 genotypes in distinct European cohorts; HWE deviation depends on sex (only females are in disequilibrium), and genotypes displaying maximum and minimum relative fitness differ across population samples, suggesting a complex situation possibly involving epistasis or maternal selection. Overall, our data indicate that future association studies involving ADRB2 will benefit from taking into account ethnicity-specific haplotype distributions and sex-based effects. With respect to ADRB3, our data indicate that the gene has been subjected to a selective

sweep in African populations, the Trp64 variant possibly representing the selection target. Given the previous association of the ancestral ADRB3 Arg64 allele with obesity and type 2 diabetes, dietary adaptations might represent the underlying selective force.

Cimolin Veronica, Piccinini Luigi, Avellis Martino, Cazzaniga Andrea, Turconi Anna Carla, Crivellini Marcello, Galli Manuela (2009); 3D-QUANTITATIVE EVALUATION OF A RIGID SEATING SYSTEM AND DYNAMIC SEATING SYSTEM USING 3D MOVEMENT ANALYSIS IN INDIVIDUALS WITH DYSTONIC TETRAPARESIS; Disability and Rehabilitation: Assistive Technology, 4(6):422-428

I.F. 2008: 0,000

To improve postural stability in individuals with dystonic cerebral palsy, the concept of a dynamic seat has been suggested as a potential solution. An experimental set-up for the acquisition of movement during extensor thrusts while sitting on a seating system was defined and applied on a group of dystonic individuals, to compare a dynamic versus a rigid seat system, using quantitative movement analysis. The seating system in dynamic configuration is able to reduce the extensor thrust experienced by the consumers, as well as to increase range of motion in the anterior-posterior direction, limiting the sliding down of trunk and showing better upper limb smoothness during extensor thrusts. The procedures used in this study appear to provide a useful tool for better understanding how the concept of a dynamic back in a seat system may affect and influence position and stability of individuals with dystonia on the seat system.

Colussi Claudia, Gurtner Aymone, Rosati Jessica, Illi Barbara, Ragone Gianluca, Piaggio Giulia, Moggio Maurizio, Lamperti Costanza, D'Angelo Maria Grazia, Clementi Emilio, Minetti Giulia, Mozzetta Chiara, Antonini Annalisa, Capogrossi Maurizio C., Puri Pier Lorenzo, Gaetano Carlo (2009); NITRIC OXIDE DEFICIENCY DETERMINES GLOBAL CHROMATIC CHANGES IN DUCHENNE MUSCULAR DYSTROPHY; FASEB Journal (The), 23(7):2131-2141

I.F. 2008: 7,049

The present study provides evidence that abnormal patterns of global histone modification are present in the skeletal muscle nuclei of mdx mice and Duchenne muscular dystrophy (DMD) patients. A combination of specific histone H3 modifications, including Ser-10 phosphorylation, acetylation of Lys 9 and 14, and Lys 79 methylation, were found enriched in muscle biopsies from human patients affected by DMD and in late-term fetuses, early

postnatal pups, or adult mdx mice. In this context, chromatin immunoprecipitation experiments showed an enrichment of these modifications at the loci of genes involved in proliferation or inflammation, suggesting a regulatory effect on gene expression. Remarkably, the reexpression of dystrophin induced by gentamicin treatment or the administration of nitric oxide (NO) donors reversed the abnormal pattern of H3 histone modifications. These findings suggest an unanticipated link between the dystrophin-activated NO signaling and the remodeling of chromatin. In this context, the regulation of class IIa histone deacetylases (HDACs) 4 and 5 was found altered as a consequence of the reduced NO-dependent protein phosphatase 2A activity, indicating that both NO and class IIa HDACs are important for satellite cell differentiation and gene expression in mdx mice. In conclusion, this work provides the first evidence of a role for NO as an epigenetic regulator in DMD.

Combi Romina, Grioni Daniele, Contri Margherita, Redaelli Serena, Redaelli Francesca, Bassi Maria Teresa, Barisani Donatella, Lavitrano Maria Luisa, Tredici Giovanni, Tenchini Maria Luisa, Bertolini Mario, Dalprà Leda (2009); CLINICAL AND GENETIC FAMILIAL STUDY OF A LARGE COHORT OF ITALIAN CHILDREN WITH IDIOPATHIC EPILEPSY; Brain Research Bulletin, 79(2):89-96

I.F. 2008: 2,281

Epilepsies are characterized by genetic heterogeneity and by the possible coexistence of different phenotypes in one family. Moreover, in different epilepsies, mutations in the same gene have been reported. We aimed to collect data in a large Italian cohort of 81 families with children affected by partial or generalized epilepsies and to evaluate the prevalence of several ion channel mutations. In particular, a clinical and genetic survey was performed and DNA regions known to be associated with several epilepsies were analysed by sequencing. We observed genetic complexity in all phenotype groups: any epileptic type may be transmitted as either autosomal dominant or recessive. No significant phenotype identity among generations and no differences among genders could be observed. Two missense mutations in SCN1A were identified in two GEFS+ probands confirming the importance of this channel for this epilepsy. Moreover, a previously unreported CLCN2 mutation was detected in a proband showing CAE. In conclusion, even in this highly heterogeneous cohort, the complexity of the epileptic condition was highlighted and mutations in the analysed candidate region of ion channel genes appear to explain only a minority of cases.

Conclave Mario, Fusaro Guido, Sala Marina, Martinuzzi Andrea,

Russo Emanuela, Frare Mara, Gorini Giovanna, Leonardi Matilde, Raggi Alberto (2009); THE ICF AND LABOUR POLICIES PROJECT: THE FIRST ITALIAN NATION-WIDE EXPERIENCE OF ICF IMPLEMENTATION IN THE LABOUR SECTOR; Disability and Rehabilitation, 31(S1):S16-S21

I.F. 2008: 1,395

Purpose. To describe the first Italian experience of ICF implementation in the Labour Sector by a Ministerial body, and the development of the International Classification of Functioning, Disability and Health (ICF)-based worker checklist.
Methods. Nation-wide training was provided. ICF-based worker checklist was developed by linking Italian legislative procedures and schedules to the ICF, and by adding standard ICF checklist's categories. When a third-level ICF category was linked, the corresponding second-level one was included in the worker checklist too.
Results. Eighty-four ICF categories were linked and five were added due to rolling-up procedure: 15 from body functions, 65 from activities and participation and 4 from environmental factors. In total, the dedicated ICF-based worker checklist is composed of 183 ICF categories, 34 of whom are at the third level and 89 from the domain of activities and participation.
Conclusions. The inclusion of the standard ICF checklist's items aimed to complement the information contained in the ministerial schedule, in which ICF categories from environmental factors domain are underrepresented. Future directions include the development of an ICF-based company checklist and an application tool for matching the information derived from the worker and the company ICF-based checklists.

Corradi-Dell'Acqua Corrado, Tomasino Barbara, Fink Gereon R. (2009); WHAT IS THE POSITION OF AN ARM RELATIVE TO THE BODY? NEURAL CORRELATES OF BODY SCHEMA AND BODY STRUCTURAL DESCRIPTION; Journal of Neuroscience, 29(13):4162-4171

I.F. 2008: 7,452

Neuropsychological studies suggest that the human brain is endowed with two body representations: the body schema (BS), coding the orientation of one's body parts in space, and the body structural description (BSD), coding the location of body parts relative to a standard body. We used fMRI to disentangle the neural mechanisms underlying these putatively distinct body representations. Participants saw an arm or a pot's handle (stimulus: arm, handle) rotated at different angles (angle: 30-150 degrees). If the stimu-

lus was an arm, subjects were instructed to imagine (1) rotating their own arm until it matched the stimulus orientation (comparing the seen arm to their own) or (2) seeing the stimulus moving toward its appropriate position on a simultaneously presented human body [comparing the arm to the one of a standard body (strategy: motor, visual imagery)]. If the stimulus was a handle, subjects were instructed to imagine (1) placing the handle on its appropriate position on a simultaneously presented pot or (2) seeing it moving toward its pot's position. The analysis of the interaction stimulus x strategy revealed activation of left secondary somatosensory cortex (SII), specifically when comparing the stimulus arm to one's own. The analysis of the parameters describing the linear effect of angle revealed that neural activity of left posterior intraparietal sulcus was modulated by the stimulus's rotation, but only when relating the arm to a standard body. The results associate BS and BSD with differential neural substrates, thereby suggesting that these are independent body representations, and furthermore extend current concepts of SII function.

Corsi Fabio, De Palma Clara, Colombo Miriam, Allevi Raffaele, Nebuloni Manuela, Ronchi Silvia, Rizzi Giuseppina, Tosoni Antonella, Trabucchi Emilio, Clementi Emilio, Prosperi Davide (2009); TOWARDS IDEAL MAGNETOFLUORESCENT NANOPARTICLES FOR BIMODAL DETECTION OF BREAST-CANCER CELLS; Small, 5(22):2555-2564

I.F. 2008: 6,525

An increasing number of novel molecular markers based on nanomaterials for tumor diagnostics have been developed in recent years. Many efforts have focused on the achievement of site-targeted bioconjugated nanoparticles. In contrast, the mechanisms of toxicity, endocytosis, and degradation pathways are still poorly understood, despite their primary importance for clinical translation. In this study, three different model nanoscale magnetofluorescent particle systems (MFNs) are designed and fabricated. These nanoparticles are evaluated in terms of size, morphology, zeta potential, fluorescence efficiency, capability of enhancing T(2) relaxivity of water protons, and stability. Accordingly, two are developed and the mechanism of internalization, the intracellular fate, and the toxicity in MCF-7 adenocarcinoma cells are studied. Besides the well-documented size effect, the anionic charge seems to be a crucial factor for particle internalization, as MFN penetration through the cell membrane could be modulated by surface charge. Ultrastructural analysis of transmission electron micrographs combined with evidence from confocal microscopy reveals that MFNs are internalized by clathrin-mediated endocy-

tosis and macropinocytosis. Moreover, MFNs are found in EEA1-positive endosomes and in lysosomes, indicating that they follow a physiological pathway of endocytosis. Magnetorelaxometric analysis demonstrates that MFNs enable the detection of 5 x 10(5) cells mL(-1) after treatment with particle dosages as low as 30 microg mL(-1). Hence, MFNs appear to be a valuable and safe bimodal contrast agent that can be developed for the noninvasive diagnosis of breast cancer.

Corti Stefania, Donadoni Chiara, Ronchi Dario, Bordoni Andreina, Fortunato Francesco, Santoro Domenico, Del Bo Roberto, Lucchini Valeria, Crugnola Veronica, Papadimitriou Dimitra, Salani Sabrina, Moggio Maurizio, Bresolin Nereo, Comi Giacomo Pietro (2009); AMYOTROPHIC LATERAL SCLEROSIS LINKED TO A NOVEL SOD1 MUTATION WITH MUSCLE MITOCHONDRIAL DYSFUNCTION; Journal of the Neurological Sciences, 276(1-2):170-174

I.F. 2008: 2,359

Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative motor neuron disorder. Mutations in Cu,Zn superoxide dismutase (SOD1) cause approximately 20% of familial ALS. One of the possible mechanisms whereby they induce disease is mitochondrial dysfunction in motor neurons. Here we describe a patient with ALS and muscle mitochondrial oxidative defect associated with a novel SOD1 mutation. Direct sequencing of SOD1 gene revealed a heterozygous mutation in codon 22 substituting a highly conserved amino acid, from glutamine to arginine (Q22R). Muscle biopsy showed a neurogenic pattern associated with cytochrome c oxidase (COX) deficiency in several muscle fibers. Western blot analysis demonstrated a reduction in SOD1 content in the cytoplasmic and mitochondrial fractions. These results suggest that a minute quantity of mutant SOD1 protein contributes to a mitochondrial toxicity also in muscle tissue.

Corti Stefania, Nizzardo Monica, Nardini Martina, Donadoni Chiara, Salani Sabrina, Del Bo Roberto, Papadimitriou Dimitra, Locatelli Federica, Mezzina Nicoletta, Gianni Francesca, Bresolin Nereo, Comi Giacomo Pietro (2009); MOTONEURON TRANSPLANTATION RESCUE THE PHENOTYPE OF SMARD1 (SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1); The Journal of Neuroscience, 29(38):11761-11771

I.F. 2008: 7,452

Spinal muscular atrophy with respiratory distress type 1 (SMARD1) is a fatal form of infantile motoneuron disease. There is currently no effective treat-

ment, although motor neuron replacement is a possible therapeutic strategy. We transplanted purified motor neurons into the spinal cord of nmd mice, an animal model of SMARD1. We also administered pharmacological treatment targeting the induction of axonal growth toward skeletal muscle target. At the end stage of the disease, donor-derived motor neurons were detected in the nmd anterior horns, extended axons into the ventral roots, and formed new neuromuscular junctions. These data correlated with improved neuromuscular function and increased life spans. The neuroprotective effect was associated with a reduction in proinflammatory molecules in treated spinal cords. This is the first report that functional restoration of motor units with transplanted motoneurons is feasible in an animal model of a human motoneuron disease, opening up new possibilities for therapeutic intervention.

Crimella Claudia, Arnoldi Alessia, Crippa Francesca, Mostacciuolo Maria Luisa, Boaretto Francesca, Sironi Manuela, D'Angelo Maria Grazia, Manzoni Simona, Piccinini Luigi, Turconi Anna Carla, Toscano Antonio, Musumeci Olimpia, Benedetti Sara, Fazio Raffaella, Bresolin Nereo, Daga Andrea, Martinuzzi Andrea, Bassi Maria Teresa (2009); POINT MUTATIONS AND A LARGE INTRAGENIC DELETION IN SPG11 IN COMPLICATED SPASTIC PARAPLEGIA WITHOUT THIN CORPUS CALLOSUM; Journal of Medical Genetics, 46(5):345-351

I.F. 2008: 5,713

BACKGROUND: Hereditary spastic paraplegia (HSP) with thin corpus callosum (HSP-TCC) is a frequent subtype of complicated HSP clinically characterised by slowly progressive spastic paraparesis with cognitive impairment and thin corpus callosum (TCC). SPG11, the gene associated with the major locus involved, encodes spatacsin, a protein of unknown function. METHODS: Different types of mutations were identified in patients with the complex form of HSP (cHSP) including TCC. We screened a series of 45 index patients with different types of cHSP with (n = 10) and without (n = 35) TCC. RESULTS: Ten mutations, of which five are novel, were detected in seven patients. Of importance, three out of seven mutated patients present with cHSP without TCC. Among the novel mutations identified, we characterised a large intragenic rearrangement deleting 2.6 kb of the SPG11 gene. The rearrangement is due to non-allelic homologous recombination between Alu sequences flanking the breakpoints. CONCLUSIONS: These findings expand the mutation spectrum of SPG11 and suggest that SPG11 mutations may occur more frequently in familial than sporadic forms of cHSP without TCC. This helps to define further clinical and molecular criteria for a correct diagnosis of the SPG11 related form of cHSP. In addition, the intragenic deletion detected

here, and the mechanism involved, both provide clues to address the issue of SPG11 missing mutant alleles previously reported.

D'Angelo Maria Grazia*, Berti Matteo*, Piccinini Luigi, Romei Marianna, Guglieri Michela, Bonato Sara, Degrate Alessandro, Turconi Anna Carla, Bresolin Nereo (2009); GAIT PATTERN IN DUCHENNE MUSCULAR DYSTROPHY; Gait & Posture, 29(1):36-41

I.F. 2008: 2,743

**Autori che hanno contribuito in ugual misura al lavoro*

We investigated the gait pattern of 21 patients with Duchenne muscular dystrophy (DMD), compared to 10 healthy controls through 3D Gait Analysis. An overall observation of gait pattern in our DMD patients when compared to controls confirmed the data previously reported for small dystrophic groups. An excessive anterior tilt of pelvis and abnormal knee pattern in loading response phase were found. Since during the swing phase the DMD foot is too plantarflexed, patients adopt a higher flexion and abduction of the hip in order to advance the swinging limb. Velocity and cadence of DMD patients resulted similar to those calculated for healthy subjects, whereas stride length was reduced and step width was increased. We then divided the DMD patients in to two subgroups (treated with steroids and untreated), and we observed that the only statistically significant differences between the two groups in Gait Analysis parameters were found for the maximum of ankle power. 3D Gait Analysis gives objective and quantitative information about the gait pattern and the deviations due to muscular situation of DMD subjects; being our study a single moment evaluation, it is otherwise unable to unravel changes only detectable through serial analysis during the time course of the disease and, if any, due to the treatment.

De Palma Clara, Di Paola Rosanna, Perrotta Cristiana, Mazzon Emanuela, Cattaneo Dario, Trabucchi Marco, Cuzzocrea Salvatore, Clementi Emilio (2009); IBUPROFEN-ARGININE GENERATES NITRIC OXIDE AND HAS ENHANCED ANTI-INFLAMMATORY EFFECTS; Pharmacological Research, 60(4):221-228

I.F. 2008: 3,287

Ibuprofen, a chiral non-steroidal anti-inflammatory drug chemically related to fenoprofen and naproxen, has moderate but definite anti-inflammatory, analgesic and antipyretic properties, with considerably less gastrointestinal adverse effect than other drugs in the same family. Currently available in the market are preparations in which bioavailability of ibuprofen is increased by salification with various salts. We have investigated the pharmacological

properties of one such salt, ibuprofen-arginine, of biological interest because l-arginine acts as substrate of the nitric oxide (NO) synthesising enzymes. Using epithelial HeLa cells expressing the endothelial NO synthase we show that ibuprofen-arginine releases NO and that this NO protects against the cytotoxic apoptogenic effects of staurosporine. We also found that ibuprofen-arginine is endowed with enhanced anti-inflammatory effects with respect to ibuprofen, as shown by reduced hind paw oedema, neutrophil infiltration and chondrocyte apoptosis in collagen-induced mouse arthritis, a model of chronic inflammation. NO has pleiotropic beneficial effects that may contribute to limit inflammation and anti-inflammatory compounds able to release NO display higher efficacy than the parent drugs in defined clinical settings. Our results open the possibility that NO generation contributes to the enhanced anti-inflammatory effects of ibuprofen-arginine vs. ibuprofen, suggesting co-administration of anti-inflammatory drugs and arginine as an additional way to exploit the beneficial effects of NO.

De Polo Gianni, Pradal Monica, Bortolot Sonia, Buffoni Mara, Martinuzzi Andrea (2009); CHILDREN WITH DISABILITY AT SCHOOL: THE APPLICATION OF ICF-CY IN THE VENETO REGION; Disability and Rehabilitation, 31(S1):S67-S73

I.F. 2008: 1,395

Purpose. To show the feasibility and consequences at 1 year of the use of the International Classification of Functioning, Disability and Health, version for Children and Youth (ICF-CY) in the process of social and scholastic inclusion for students with disability in a district of Northeastern Italy (Treviso province). Methods. We describe the novel procedure for inclusion of students with disability launched by disability and education Services of the Treviso Province. The protocol was organized in four steps and involved health professionals and teachers throughout the whole Province. The implementation was preceded by intensive exposure of the involved professionals to the ICF-CY model and structure and by workshops in which the participants elaborated the actual documents accompanying the process of scholastic inclusion according to the specific Law (104/1992): the notification card, the identification of the student with disabilities, the functional diagnosis, the dynamic functional profile and the individual educational plan.
Results. The results show that the adherence to the new protocol was very satisfactory, as well as the perceived validity and relevance of the new documents elaborated with ICF language. The experimentation in progress provided interesting indications on the way to apply the ICF-CY to the scholastic inclusion processes.

Conclusions. The largest alphabetization effort on ICF attempted in a Public Health System (disability service) and in the school system improve the social and scholastic participation of student with disability and can reduce the barriers in the environment.

Deponti Daniela, Buono Roberta, Catanzaro Giuseppina, De Palma Clara, Longhi Renato, Meneveri Raffaella, Bresolin Nereo, Bassi Maria Teresa, Cossu Giulio, Clementi Emilio, Brunelli Silvia (2009); THE LOW AFFINITY RECEPTOR FOR NEUROTROPHINS P75NTR PLAYS A KEY ROLE FOR SATELLITE CELL FUNCTION IN MUSCLE REPAIR ACTING VIA RHOA; Molecular Biology of the Cell, 20(16):3620-3627

I.F. 2008: 5,558

Regeneration of muscle fibers, lost during pathological muscle degeneration or after injuries, is mediated by the production of new myofibres. This process, sustained by the resident stem cells of the muscle, the satellite cells, is finely regulated by local cues, in particular by cytokines and growth factors. Evidence in the literature suggests that nerve growth factor (NGF) is involved in muscle fiber regeneration; however, its role and mechanism of action were unclear. We have investigated this issue in in vivo mouse models of muscle regeneration and in primary myogenic cells. Our results demonstrate that NGF acts through its low-affinity receptor p75(NTR) in a developmentally regulated signaling pathway necessary to myogenic differentiation and muscle repair in vivo. We also demonstrate that this action of NGF is mediated by the down-regulation of RhoA-GTP signaling in myogenic cells.

Di Fonzo Alessio, Ronchi Dario, Lodi Tiziana, Fassone Elisa, Tigano Marco, Lamperti Costanza, Corti Stefania, Bordoni Andreina, Fortunato Francesco, Nizzardo Monica, Napoli Laura, Donadoni Chiara, Salani Sabrina, Saladino Francesca, Moggio Maurizio, Bresolin Nereo, Ferrero Iliana, Comi Giacomo Pietro (2009); THE MITOCHONDRIAL DISULFIDE RELAY SYSTEM PROTEIN GFER IS MUTATED IN AUTOSOMAL-RECESSIVE MYOPATHY WITH CATARACT AND COMBINED RESPIRATORY-CHAIN DEFICIENCY; American Journal of Human Genetics, 84(5):594-604

I.F. 2008: 10,153

A disulfide relay system (DRS) was recently identified in the yeast mitochondrial intermembrane space (IMS) that consists of two essential components: the sulfhydryl oxidase Erv1 and the redox-regulated import receptor Mia40. The DRS drives the import of cysteine-rich proteins into the IMS via an oxi-

dative folding mechanism. Erv1p is reoxidized within this system, transferring its electrons to molecular oxygen through interactions with cytochrome c and cytochrome c oxidase (COX), thereby linking the DRS to the respiratory chain. The role of the human Erv1 ortholog, GFER, in the DRS has been poorly explored. Using homozygosity mapping, we discovered that a mutation in the GFER gene causes an infantile mitochondrial disorder. Three children born to healthy consanguineous parents presented with progressive myopathy and partial combined respiratory-chain deficiency, congenital cataract, sensorineural hearing loss, and developmental delay. The consequences of the mutation at the level of the patient's muscle tissue and fibroblasts were 1) a reduction in complex I, II, and IV activity; 2) a lower cysteine-rich protein content; 3) abnormal ultrastructural morphology of the mitochondria, with enlargement of the IMS space; and 4) accelerated time-dependent accumulation of multiple mtDNA deletions. Moreover, the Saccharomyces cerevisiae erv1(R182H) mutant strain reproduced the complex IV activity defect and exhibited genetic instability of the mtDNA and mitochondrial morphological defects. These findings shed light on the mechanisms of mitochondrial biogenesis, establish the role of GFER in the human DRS, and promote an understanding of the pathogenesis of a new mitochondrial disease.

Donati Chiara, Cencetti Francesca, De Palma Clara, Rapizzi Elena, Brunelli Silvia, Cossu Giulio, Clementi Emilio, Bruni Paola (2009); TGFBETA PROTECTS MESOANGIOBLASTS FROM APOPTOSIS VIA SPHINGOSINE KINASE-1 REGULATION; Cellular Signalling, 21(2):228-236

I.F. 2008: 4,305

Mesoangioblasts are vessel-derived progenitor cells that can be induced to differentiate into different cell types of the mesoderm such as muscle and bone. Here we examined the role of transforming growth factor-beta (TGF-beta), a pleiotropic cytokine that plays a major role in development and specifically induces smooth muscle differentiation of mesoangioblasts, in the regulation of death and survival of these cells. TGFbeta exerts a marked anti-apoptotic action in mesoangioblasts with a mechanism involving regulation of sphingosine kinase 1 (SphK1), one of the isoforms responsible for S1P formation. Treatment with the cytokine efficaciously protected mesoangioblasts from apoptosis induced by serum starvation or staurosporine treatment assessed by various means such as activation of caspase-3, determination of cytoplasmic histone-associated-DNA-fragments and PE-AnnexinV staining. The protective action of TGFbeta from staurosporine-induced apoptosis was strongly reduced when the SphK activity was inhibited by drugs, when SphK1

but not SphK2 was downregulated by specific siRNA and when a SphK1 dominant negative mutant was overexpressed. Staurosporine treatment induced down-regulation of both SphK isoforms and TGFbeta rescued SphK1 but not SphK2 expression. Interestingly, TGFbeta strongly enhanced SphK activity during staurosporine-induced cell death. Both TGFbeta-induced SphK1 up-regulation and TGFbeta anti-apoptotic action were found to be dependent on p42/44 MAPK activation.

Facchin Paola, Rizzotto Melissa Rosa, Turconi Anna Carla, Pagliano Emanuela, Fazzi Elisa, Stortini Massimo, Fedrizzi Ermellina, GIPCI Study Group (Ancona Vera*, Cazzagon Monica*, Germinasi Chiara, La Gamba Annalisa*, Martinuzzi Andrea, Megliani Chiara, Trabacca Antonio) (2009); MULTI-SITE TRIAL ON EFFICACY OF CONSTRAINT INDUCED MOVEMENT THERAPY IN CHILDREN WITH HEMIPLEGIA: STUDY DESIGN AND METHODOLOGY; American Journal of Physical Medicine and Rehabilitation, 88(3):216-230

I.F. 2008: 1,695

OBJECTIVE: In the past decades, several treatment approaches have been used to improve upper limb function in hemiplegic cerebral palsy. Only recently has constraint-induced movement therapy emerged as a treatment approach for children with hemiplegic cerebral palsy with the aim of reversing the behavioral suppression of movement in the affected upper limb. To date, evidence on this treatment has been very poor and limited, because all currently available trials reveal methodological limitations and a need for additional research to support the application of this treatment technique. This article presents the methodological choices, design, and main characteristics of an ongoing controlled clinical trial on the effectiveness and safety of constraint-induced movement therapy combined with an intensive rehabilitation program and compared with two comparison groups: one treated with an intensive rehabilitation program and the other with standard treatment. METHODS: Twenty-one rehabilitation sites are currently recruiting patients with hemiplegic cerebral palsy, aged between 2 and 8 yrs, who have never undergone constraint therapy. Primary outcome measures include two major domains: upper limb motor ability (Quality of Upper Extremity Skills Test) and hand function assessment evaluating both grip function and spontaneous use of the affected side (Besta scale). Secondary outcome measures concern overall function, behavior, compliance, and satisfaction with treatment program of both child and family. Patients' follow-up is of 12 mos after treatment. RESULTS: Research in children has always been neglected in comparison with adults, because of ethical reasons regarding the use of

children for experimental purposes. The consequence has been the utilization of treatment and assessment tools and techniques that have not always been tested in pediatric patients or evidence is very scarce. CONCLUSION: Discussing and working on pediatric research methods represents an urgent need in rehabilitation research.

Francescutti Carlo, Martinuzzi Andrea, Leonardi Matilde, Kostanjsek Nenad Friedrich Ivan (2009); EIGHT YEARS OF ICF IN ITALY: PRINCIPLES, RESULTS AND FUTURE PERSPECTIVE; Disability and Rehabilitation, 31(S1):S4-S7 - Editoriale

I.F. 2008: 1,395

Purpose. To report on the process of implementation and dissemination of the International Classification of Functioning, Disability and Health (ICF) [1] in Italy.
Methods. The Agenzia Regionale della Sanita` of Friuli Venezia Giulia Region (ARSFVG) is a WHO Collaborating Centre for the Family of International Classifications. It collaborated with Italian research institutions such as the C. Besta Neurological Institute IRCCS Foundation, the Medea Institute, and the National Institute of Statistics in revising, field testing and validating the ICF in Italy and in the preparation of ICF-CY (Children and Youth Version).
Results. The value of ICF profiles in defining personalised programmes of interventions was explored by evaluating the link between ICF items and the UN Convention, which was taken as a criterion of clear ethical and political orientation in the evaluation of the disability condition. The first and main effort of ICF implementation was directed in the field of public health and welfare policies. Two main nationwide projects were launched: ICF and the labour polices in 2003 and ICF and the disability certification reforms in 2006. ICF also received a strong attention by the professional working in the school system, and was used to define the functioning profile of children and to establish personalised educational programmes.
Conclusions. The implementation of ICF in Italy was strongly facilitated by a favourable cultural and scientific context.

Francescutti Carlo, Frattura Lucia, Troiano Raffaella, Gongolo Francesco, Martinuzzi Andrea, Sala Marina, Meucci Paolo, Raggi Alberto, Russo Emanuela, Buffoni Mara, Gorini Giovanna, Conclave Mario, Petrangeli Agostino, Solipaca Alessandro, Leonardi Matilde (2009); TOWARDS A COMMON DISABILITY ASSESSMENT FRAMEWORK: THEORETICAL AND METHODOLOGICAL ISSUES FOR PROVIDING PUBLIC SERVICES AND BENEFITS USING ICF;

Disability and Rehabilitation, 31(S1):S8-S15

I.F. 2008: 1,395

Purpose. To report on the preliminary results of an Italian project on the implementation of an ICF-based protocol for providing public services and benefits for persons with disabilities.
Methods. The UN Convention on the Rights of persons with disabilities (UNC) was mapped to the ICF, and core elements were implemented in an ICF-based evaluation protocol. A person–environment interaction classification (PEIC) tree was also developed for defining evaluation outputs.
Results. The PEIC and the ICF-based protocol are the guideline and the data interpretation source, respectively, for providing public services and benefits. They enable to assign persons to different services, from surveillance and monitoring to facilitator provision or sustain over time, to barrier removal or to the reorganisation of environmental factors provision. A detailed description of the target intervention is made available through the implementation of a protocol, which points out the effect of personal support and other environmental factors.
Conclusions. The detailed description of functioning and disability provided by our methodology can help policy makers and administrators in decision making, on the basis of a description of real needs, and in targeting person-tailored interventions.

Francescutti Carlo, Fusaro Guido, Leonardi Matilde, Martinuzzi Andrea, Sala Marina, Russo Emanuela, Frare Mara, Pradal Monica, Zampogna Daniela, Cosentino Alessandro, Raggi Alberto (2009); ITALIAN ICF TRAINING PROGRAMS: DESCRIBING AND PROMOTING HUMAN FUNCTIONING AND RESEARCH; Disability and Rehabilitation, 31(S1):S46-S49

I.F. 2008: 1,395

Purpose of the article is to report on 5 years of ICF training experiences in Italy aimed at promoting a consistent approach to ICF's field application. More than 7000 persons participated in around 150 training events: almost half were organised by political bodies, at national, regional or local level, directly linked to implementation experiences. Few training events were organised by the school sector, while training commissioned by NGOs represent a relevant area and, in our opinion, constitute the first step towards a full inclusion of persons with disabilities. Central pillars of our training modules are: the inclusion of all ICF components in the description of functional profiles, the need of providing brief theoretical background information before moving to practical aspects and the importance of providing personalised face to face

training modules, in contrast to self-administered learning modules, or web-based protocols. On the basis of our experience, we can conclude that training's objectives are generally reached: trainees improved their knowledge of the ICF and its related tools, and are able to begin practical applications in their contexts.

Frigerio Alessandra, Rucci Paola, Goodman R., Ammaniti Massimo, Carlet Ombretta, Cavolina Pina, De Girolamo Giovanni, Lenti Carlo, Lucarelli Loredana, Mani Elisa, Martinuzzi Andrea, Micali N., Milone Annarita, Morosini Pierluigi, Muratori Filippo, Nardocci Franco, Pastore Valentina, Polidori Gabriella, Tullini Andrea, Vanzin Laura, Villa Laura, Walder Mauro, Zuddas Alessandro, Molteni Massimo (2009); PREVALENCE AND CORRELATES OF MENTAL DISORDERS AMONG ADOLESCENTS IN ITALY: THE PRISMA STUDY; European Child & Adolescent Psychiatry, 18(4):217-226

I.F. 2008: 1,769

BACKGROUND: While in the last 5 years several studies have been conducted in Italy on the prevalence of mental disorders in adults, to date no epidemiological study has been targeted on mental disorders in adolescents. METHOD: A two-phase study was conducted on 3,418 participants using the child behavior checklist/6-18 (CBCL) and the development and well-being assessment (DAWBA), a structured interview with verbatim reports reviewed by clinicians. RESULTS: The prevalence of CBCL caseness and DSM-IV disorders was 9.8% (CI 8.8-10.8%) and 8.2% (CI 4.2-12.3%), respectively. DSM-IV Emotional disorders were more frequently observed (6.5% CI 2.2-10.8%) than externalizing disorders (1.2% CI 0.2-2.3%). In girls, prevalence estimates increased significantly with age; furthermore, living with a single parent, low level of maternal education, and low family income were associated with a higher likelihood of suffering from emotional or behavioral problems. CONCLUSIONS: Approximately one in ten adolescents has psychological problems. Teachers and clinicians should focus on boys and girls living with a single parent and/or in disadvantaged socioeconomic conditions.

Frigerio Alessandra, Ceppi Elisa, Rusconi Marianna, Giorda Roberto, Raggi Maria Elisabetta, Fearon Pasco (2009); THE ROLE PLAYED BY THE INTERACTION BETWEEN GENETIC FACTORS AND ATTACHMENT IN THE STRESS RESPONSE IN INFANCY; Journal of Child Psychology and Psychiatry, 50(12):1513-1522

I.F. 2008: 4,854

BACKGROUND: The importance of understanding which environmental

and biological factors are involved in determining individual differences in physiological response to stress is widely recognized, given the impact that stress has on physical and mental health. METHODS: The child-mother attachment relationship and some genetic polymorphisms (5-HTTLPR, COMT and GABRA6) were tested as predictors of salivary cortisol and alpha amylase concentrations, two biomarkers of hypothalamic-pituitary-adrenocortical (HPA) axis and sympathetic adrenomedullary (SAM) system activity, during the Strange Situation (SS) procedure in a sample of more than 100 healthy infants, aged 12 to 18 months. RESULTS: Individual differences in alpha amylase response to separation were predicted by security of attachment in interaction with 5-HTTLPR and GABRA6 genetic polymorphisms, whereas alpha amylase basal levels were predicted by COMT x attachment interaction. No significant effect of attachment, genetics and their interaction on cortisol activity emerged. CONCLUSIONS: These results help to disentangle the role played by both genetic and environmental factors in determining individual differences in stress response in infancy. The results also shed light on the suggestion that HPA and SAM systems are likely to have different characteristic responses to stress.

Fumagalli Matteo, Cagliani Rachele, Pozzoli Uberto, Riva Stefania, Comi Giacomo Pietro, Menozzi Giorgia, Bresolin Nereo, Sironi Manuela (2009); WIDESPREAD BALANCING SELECTION AND PATHOGEN-DRIVEN SELECTION AT BLOOD GROUP ANTIGEN GENES; Genome Research, 19(2):199-212

I.F. 2008: 10,176

Historically, allelic variations in blood group antigen (BGA) genes have been regarded as possible susceptibility factors for infectious diseases. Since host-pathogen interactions are major determinants in evolution, BGAs can be thought of as selection targets. In order to verify this hypothesis, we obtained an estimate of pathogen richness for geographic locations corresponding to 52 populations distributed worldwide; after correction for multiple tests and for variables different from selective forces, significant correlations with pathogen richness were obtained for multiple variants at 11 BGA loci out of 26. In line with this finding, we demonstrate that three BGA genes, namely CD55, CD151, and SLC14A1, have been subjected to balancing selection, a process, rare outside MHC genes, which maintains variability at a locus. Moreover, we identified a gene region immediately upstream the transcription start site of FUT2 which has undergone non-neutral evolution independently from the coding region. Finally, in the case of BSG, we describe the presence of a highly divergent haplotype clade and the possible reasons for its main-

tenance, including frequency-dependent balancing selection, are discussed. These data indicate that BGAs have been playing a central role in the host-pathogen arms race during human evolutionary history and no other gene category shows similar levels of widespread selection, with the only exception of loci involved in antigen recognition.

Fumagalli Matteo*, Pozzoli Uberto*, Cagliani Rachele, Comi Giacomo Pietro, Riva Stefania, Clerici Mario, Bresolin Nereo, Sironi Manuela (2009); PARASITES REPRESENT A MAJOR SELECTIVE FORCE FOR INTERLEUKIN GENES AND SHAPE THE GENETIC PREDISPOSITION TO AUTOIMMUNE CONDITIONS; The Journal of Experimental Medicine, 206(6):1395-1408

I.F. 2008: 15,463

**Autori che hanno contribuito in ugual misura al lavoro*

Many human genes have adapted to the constant threat of exposure to infectious agents; according to the "hygiene hypothesis," lack of exposure to parasites in modern settings results in immune imbalances, augmenting susceptibility to the development of autoimmune and allergic conditions. Here, by estimating the number of pathogen species/genera in a specific geographic location (pathogen richness) for 52 human populations and analyzing 91 interleukin (IL)/IL receptor genes (IL genes), we show that helminths have been a major selective force on a subset of these genes. A population genetics analysis revealed that five IL genes, including IL7R and IL18RAP, have been a target of balancing selection, a selection process that maintains genetic variability within a population. Previous identification of polymorphisms in some of these loci, and their association with autoimmune conditions, prompted us to investigate the relationship between adaptation and disease. By searching for variants in IL genes identified in genome-wide association studies, we verified that six risk alleles for inflammatory bowel (IBD) or celiac disease are significantly correlated with micropathogen richness. These data support the hygiene hypothesis for IBD and provide a large set of putative targets for susceptibility to helminth infections.

Fumagalli Matteo, Cagliani Rachele, Pozzoli Uberto, Riva Stefania, Comi Giacomo Pietro, Menozzi Giorgia, Bresolin Nereo, Sironi Manuela (2009); A POPULATION GENETICS STUDY OF THE FAMILIAL MEDITERRANEAN FEVER GENE: EVIDENCE OF BALANCING SELECTION UNDER AN OVERDOMINANCE REGIME; Genes and Immunity, 10(8):678-686

I.F. 2008: 4,006

Familial Mediterranean Fever (FMF) is a recessively inherited systemic autoinflammatory disease caused by mutations in the MEFV gene. The frequency of different disease alleles is extremely high in multiple populations from the Mediterranean region, suggesting heterozygote advantage. Here, we characterize the sequence variation and haplotype structure of the MEFV 3' gene region (from exon 5 to the 3' UTR) in seven human populations. In non-African populations, we observed high levels of nucleotide variation, an excess of intermediate-frequency alleles, reduced population differentiation and a genealogy with two common haplotypes separated by deep branches. These features are suggestive of balancing selection having acted on this region to maintain one or more selected alleles. In line with this finding, an excess of heterozygotes was observed in Europeans and Asians, suggesting an overdominance regime. Our data, together with the earlier demonstration that the MEFV exon 10 has been subjected to episodic positive selection over primate evolution, provide evidence for an adaptive role of nucleotide variation in this gene region. Our data suggest that further studies aimed at clarifying the role of MEFV variants might benefit from the integration of molecular evolutionary and functional analyses.

Galbiati Susanna, Recla Monica, Pastore Valentina, Liscio Mariarosaria, Bardoni Alessandra, Castelli Enrico, Strazzer Sandra (2009); ATTENTION REMEDIATION FOLLOWING TRAUMATIC BRAIN INJURY IN CHILDHOOD AND ADOLESCENCE; Neuropsychology, 23(1):40-49

I.F. 2008: 3,201

Traumatic brain injury (TBI) frequently affects both the basic and the superordinate components of attention; deficits vary according to patient age. This study evaluated the efficacy of a specific remediation intervention for attention. Sixty-five TBI patients (aged 6?18 years) with attention deficit were assessed at baseline and at 1-year follow-up: 40 patients received attention-specific neuropsychological training for 6 months, and the control group comprised 25 patients. Cognitive assessment included a Wechsler Intelligence Scale (e.g., A. Orsini, 1993) and the Continuous Performance Test II (CPT II; C. K. Conners, 2000). The Vineland Adaptive Behavior Scales (VABS; S. Sparrow, D. Balla & D. V. Cicchetti, 1984) was administered to assess the treatment's ecological validity. At baseline, all patients presented with a mild intellectual disability and pathological scores on the CPT II. At follow-up, significant differences were found between the 2 groups on the CPT II and VABS: The clinical group improved more than the control group. Specific remediation training for attention, including a combination of a process-specific approach

and metacognitive strategies, significantly improved attention performance. Improvement in attention skills also affected adaptive skills positively. (c) 2009 APA, all rights reserved.

Gandola Lorenza, Massimino Maura, Cefalo Graziella, Solero Carlo L., Spreafico Filippo, Pecori Emilia, Riva Daria, Collini Paola, Pignoli Emanuele, Giangaspero Felice, Luksch Roberto, Berretta Serena, Poggi Geraldina, Biassoni Veronica, Ferrari Andrea, Pollo Bianca, Favre Claudio, Sardi Iacopo, Terenziani Monica, Fossati-Bellani Franca (2009); HYPERFRACTIONATED ACCELERATED RADIOTHERAPY AFTER INTENSIVE NEO-ADJUVANT CHEMOTHERAPY IN THE MILAN STRATEGY FOR METASTATIC MEDULLOBLASTOMA; Journal of Clinical Oncology, 27(4):566-571

I.F. 2008: 17,157

PURPOSE: With a view to improving the prognosis for patients with metastatic medulloblastoma, we tested the efficacy and toxicity of a hyperfractionated accelerated radiotherapy (HART) regimen delivered after intensive sequential chemotherapy. PATIENTS AND METHODS: Between 1998 and 2007, 33 consecutive patients received postoperative methotrexate (8 g/m(2)), etoposide (2.4 g/m(2)), cyclophosphamide (4 g/m(2)), and carboplatin (0.8 g/m(2)) in a 2-month schedule, then HART with a maximal dose to the neuraxis of 39 Gy (1.3 Gy/fraction, 2 fractions/d) and a posterior fossa boost up to 60 Gy (1.5 Gy/fraction,2 fractions/d). Patients with persistent disseminated disease before HART were consolidated with two myeloablative courses and circulating progenitor cell rescue. RESULTS: Patients were classified as having M1 (n = 9), M2 (n = 6), M3 (n = 17), and M4 (n = 1) disease. Seven patients younger than 10 years old who achieved complete response after chemotherapy received a lower dose to the neuraxis (31.2 Gy). Twenty-two of the 32 assessable patients responded to chemotherapy; disease was stable in five patients and progressed in five patients. One septic death occurred before radiotherapy. Eight patients experienced relapse after a median of 12 months. Fourteen of the 33 patients underwent consolidation therapy after HART. With a median 82-month survivor follow-up, the 5-year event-free, progression-free, and overall survival rates were 70%, 72%, and 73%, respectively. No severe clinical complications of HART have emerged so far. CONCLUSION: HART after intensive postoperative chemotherapy, followed by myeloablative chemotherapy in selected cases, proved feasible in children with metastatic medulloblastoma. The results of our treatment compare favorably with other series treated using conventional therapies.

Ghezzi Serena, Del Bo Roberto, Scarlato Marina, Nardini Martina, Carlesi Cecilia, Prelle Alessandro, Corti Stefania, Mancuso Michelangelo, Briani Chiara, Siciliano Gabriele, Murri Luigi, Bresolin Nereo, Comi Giacomo Pietro (2009); IS ERYTHROPOIETIN GENE A MODIFIER FACTOR IN AMYOTROPHIC LATERAL SCLEROSIS?; Neurobiology of Aging, 30(5):842-844

I.F. 2008: 5,959

To investigate the role of erythropoietin (EPO) as genetic determinant in the susceptibility to sporadic amyotrophic lateral sclerosis (SALS). We sequenced a 259-bp region spanning the 3'hypoxia-responsive element of the EPO gene in 222 Italian SALS patients and 204 healthy subjects, matched for age and ethnic origin. No potentially causative variation was detected in SALS subjects; in addition, two polymorphic variants (namely C3434T and G3544T) showed the same genotype and haplotype frequencies in patients and controls. Conversely, a weak but significant association between G3544T and age of disease onset was observed (p=0.04). Overall, our data argue against the hypothesis of EPO as a genetic risk factor for motor neuron dysfunction, at least in Italian population. However, further studies on larger cohort of patients are needed to confirm the evidence of EPO gene as modifier factor.

Gimelli Stefania, Beri Silvana, Drabkin Harry A., Gambini Claudio, Gregorio Andrea, Fiorio Patrizia, Zuffardi Orsetta, Gemmill Robert A, Giorda Roberto, Gimelli Giorgio (2009); THE TUMOR SUPPRESSOR GENE TRC8/RNF139 IS DISRUPTED BY A CONSTITUTIONAL BALANCED TRANSLOCATION T(8;22)(Q24.13;Q11.21) IN A YOUNG GIRL WITH DYSGERMINOMA; Molecular Cancer, 8(1):52

I.F. 2008: 5,362

BACKGROUND: RNF139/TRC8 is a potential tumor suppressor gene with similarity to PTCH, a tumor suppressor implicated in basal cell carcinomas and glioblastomas. TRC8 has the potential to act in a novel regulatory relationship linking the cholesterol/lipid biosynthetic pathway with cellular growth control and has been identified in families with hereditary renal (RCC) and thyroid cancers. Haploinsufficiency of TRC8 may facilitate development of clear cell-RCC in association with VHL mutations, and may increase risk for other tumor types. We report a paternally inherited balanced translocation t(8;22) in a proposita with dysgerminoma. METHODS: The translocation was characterized by FISH and the breakpoints cloned, sequenced, and compared. DNA isolated from normal and tumor cells was checked for abnormalities by array-CGH. Expression of genes TRC8 and TSN was tested both on dysger-

minoma and in the proposita and her father. RESULTS: The breakpoints of the translocation are located within the LCR-B low copy repeat on chromosome 22q11.21, containing the palindromic AT-rich repeat (PATRR) involved in recurrent and non-recurrent translocations, and in an AT-rich sequence inside intron 1 of the TRC8 tumor-suppressor gene at 8q24.13. TRC8 was strongly underexpressed in the dysgerminoma. Translin is underexpressed in the dysgerminoma compared to normal ovary.TRC8 is a target of Translin (TSN), a posttranscriptional regulator of genes transcribed by the transcription factor CREM-tau in postmeiotic male germ cells. CONCLUSION: A role for TRC8 in dysgerminoma may relate to its interaction with Translin. We propose a model in which one copy of TRC8 is disrupted by a palindrome-mediated translocation followed by complete loss of expression through suppression, possibly mediated by miRNA.

Giorda Roberto*, Bonaglia Maria Clara*, Beri Silvana, Fichera Marco, Novara Francesca, Magini Pamela, Urquhart Jill, Sharkey Freddie H., Zucca Claudio, Grasso Rita, Marelli Susan, Castiglia Lucia, Di Benedetto Daniela, Musumeci Sebastiano, Vitello Girolamo A., Failla Pinella, Reitano Santina, Avola Emanuela, Bisulli Francesca, Tinuper Paolo, Mastrangelo Massimo, Fiocchi Isabella, Spaccini Luigina, Torniero Claudia, Fontana Elena, Lynch Sally Ann, Clayton-Smith Jill, Black Graeme, Jonveaux Philippe, Leheup Bruno, Seri Marco, Romano Corrado, Dalla Bernardina Bernardo, Zuffardi Orsetta (2009); COMPLEX SEGMENTAL DUPLICATIONS MEDIATE A RECURRENT DUP(X)(P11.22-P11.23) ASSOCIATED WITH MENTAL RETARDATION, SPEECH DELAY, AND EEG ANOMALIES IN MALES AND FEMALES; American Journal of Human Genetics, 85(3):394-400

I.F. 2008: 10,153

**Autori che hanno contribuito in ugual misura al lavoro*

Submicroscopic copy-number variations make a considerable contribution to the genetic etiology of human disease. We have analyzed subjects with idiopathic mental retardation (MR) by using whole-genome oligonucleotide-based array comparative genomic hybridization (aCGH) and identified familial and de novo recurrent Xp11.22-p11.23 duplications in males and females with MR, speech delay, and a peculiar electroencephalographic (EEG) pattern in childhood. The size of the duplications ranges from 0.8-9.2 Mb. Most affected females show preferential activation of the duplicated X chromosome. Carriers of the smallest duplication show X-linked recessive inheritance. All other affected individuals present dominant expression and comparable clinical phenotypes irrespective of sex, duplication size, and X-inactivation

pattern. The majority of the rearrangements are mediated by recombination between flanking complex segmental duplications. The identification of common clinical features, including the typical EEG pattern, predisposing genomic structure, and peculiar X-inactivation pattern, suggests that duplication of Xp11.22-p11.23 constitutes a previously undescribed syndrome.

Griffo Giampiero, Leonardi Matilde, Martinuzzi Andrea, Francescutti Carlo, Raggi Alberto, Kosic Vladimir, Barbieri Pietro Vittorio (2009); MOVING TOWARDS ICF USE FOR MONITORING THE UN CONVENTION ON THE RIGHTS OF PERSONS WITH DISABILITIES: THE ITALIAN EXPERIENCE; Disability and Rehabilitation, 31(S1):S74-S77

I.F. 2008: 1,395

Abstract non disponibile

Griseri Paola, Vos Yvonne, Giorda Roberto, Gimelli Stefania, Beri Silvana, Santamaria Giuseppe, Mognato Guendalina, Hofstra Robert MW, Gimelli Giorgio, Ceccherini Isabella (2009); COMPLEX PATHOGENESIS OF HIRSCHSPRUNG'S DISEASE IN A PATIENT WITH HYDROCEPHALUS, VESICO-URETERAL REFLUX AND A BALANCED TRANSLOCATION T(3;17)(P12;Q11); European Journal of Human Genetics, 17(4):483-490

I.F. 2008: 3,925

Hirschsprung's disease (HSCR), a congenital complex disorder of intestinal innervation, is often associated with other inherited syndromes. Identifying genes involved in syndromic HSCR cases will not only help understanding the specific underlying diseases, but it will also give an insight into the development of the most frequent isolated HSCR. The association between hydrocephalus and HSCR is not surprising as a large number of patients have been reported to show the same clinical association, most of them showing mutations in the L1CAM gene, encoding a neural adhesion molecule often involved in isolated X-linked hydrocephalus. L1 defects are believed to be necessary but not sufficient for the occurrence of the intestinal phenotype in syndromic cases. In this paper, we have carried out the molecular characterization of a patient affected with Hirschsprung's disease and X-linked hydrocephalus, with a de novo reciprocal balanced translocation t(3;17)(p12;q21). In particular, we have taken advantage of this chromosomal defect to gain access to the predisposing background possibly leading to Hirschsprung's disease. Detailed analysis of the RET and L1CAM genes, and molecular characterization of MYO18A and TIAF1, the genes involved in the balanced

translocation, allowed us to identify, besides the L1 mutation c.2265delC, different additional factors related to RET-dependent and -independent pathways which may have contributed to the genesis of enteric phenotype in the present patient.

Kanovsky Petr, Bares Martin, Severa Stanislav, Richardson Alan, Dysport Paediatric Limb Spasticity Study Group (Turconi Anna Carla) (2009); LONG-TERM EFFICACY AND TOLERABILITY OF 4-MONTHLY VERSUS YEARLY BOTULINUM TOXIN TYPE A TREATMENT FOR LOWER-LIMB SPASTICITY IN CHILDREN WITH CEREBRAL PALSY; Developmental Medicine and Child Neurology, 51(6):436-445

I.F. 2008: 2,561

In this study, we compared the long-term efficacy and tolerability of two dosage regimens of the potent botulinum toxin type A (BoNT-A; Dysport; Ipsen Ltd, Slough, UK) in children with cerebral palsy (CP) and lower-limb spasticity. Children aged 1 to 8 years with diplegic CP who were able to walk (aided or unaided) were randomized (1:1) to 30 LD(50) units/kg total body weight of BoNT-A (injected into gastrocnemius muscles) every 4 months or once yearly for 2 years in this multicentre, assessor-blinded, parallel-group study. In the 4-monthly group (n=110, 39 males, 71 females), mean age was 3 years 8 months (SD 1 y 6 mo, range 1-8 y). In the yearly group (n=104, 47 males, 57 females), mean age was 4 years 4 months (SD 1 y 6 mo, range 2-8 y). Both treatment groups had similar baseline Gross Motor Function Measure scores. At month 28 (primary endpoint; intention-to-treat group), median maximum passive ankle dorsiflexion was 12.00 degrees in the 4-monthly and 11.00 degrees in the yearly group. Between-group difference of 1.67 degrees was not statistically significant (p=0.055). Other efficacy endpoints showed no significant difference between the regimens. The results of the study do not allow a clear conclusion of the preferred injection regimen.

Kemp Graham J., Tonon Caterina, Malucelli Emil, Testa Claudia, Liava Alexandra, Manners David, Trevisi Enrico, Martinuzzi Andrea, Barbiroli Bruno, Lodi Raffaele (2009); CYTOSOLIC PH BUFFERING DURING EXERCISE AND RECOVERY IN SKELETAL MUSCLE OF PATIENTS WITH MCARDLE'S DISEASE; European Journal of Applied Physiology, 105(5):687-694

I.F. 2008: 1,931

Cellular pH control is important in muscle physiology, and for interpretation of (31)P magnetic resonance spectroscopy (MRS) data. Cellular acidification in exercise results from coupled glycolytic ATP production mitigated by cytosolic buffering, 'consumption' of H(+) by phosphocreatine (PCr) breakdown, and membrane transport processes. Ex vivo methods for cytosolic buffer capacity are vulnerable to artefact, and MRS methods often require assumptions. (31)P MRS of early exercise, when pH increases unopposed by glycolysis, is conceptually simple, but limited in normal muscle by time resolution and signal-to-noise. A therapeutic trial (Martinuzzi A et al. Musc Nerve 37: 350-357, 2007) in McArdle's disease (glycogen phosphorylase deficiency), where pH does not decrease with exercise, offered the opportunity to test (31)P MRS data obtained throughout incremental plantar flexion exercise and recovery in ten McArdle's patients against the simple model of cellular pH control. Changes in pH, [Pi] and [PCr] throughout exercise and recovery were quantitatively consistent with mean +/- SEM buffer capacity of 10 +/- 1 mM/(pH unit), which was not significantly different from the control subjects under the initial-exercise conditions where the comparison could be made. The simple model of cellular acid-base balance therefore gives an adequate account of cellular pH changes during both exercise and recovery in McArdle's disease.

Lanfranconi Silvia, Corti Stefania, Bersano Anna, Costa Antonella, Prelle Alessandro, Sciacco Monica, Bresolin Nereo, Ghione Isabella (2009); APHASIC AND VISUAL AURA WITH INCREASED VASOGENIC LEAKAGE: AN ATYPICAL MIGRAINOSUS STATUS; Journal of the Neurological Sciences, Short Communication 285(1-2):227-229

I.F. 2008: 2,359

The pathogenesis of migraine with aura has not been fully established yet. The release of vasoactive substances and intracranial extracerebral blood vessel vasodilatation are probably related to stimulation of meningeal blood vessels through trigeminal afferents. Here, we report a 67 year old woman developing migraine with aphasia and right hemianopsia, lasting 4 days in duration. After spontaneous recovery, she experienced the same symptoms without migraine lasting for 15 h. MRI, performed during this last episode, revealed increased perfusion and leakage of contrast into subarachnoid space. In migraine with prolonged aura hyperperfusion with vasogenic edema might explain cortical function impairment and contribute to neurological deficits.

Leonardi Matilde, Martinuzzi Andrea (2009); ICF AND ICF-CY FOR AN INNOVATIVE HOLISTIC APPROACH TO PERSONS WITH CHRONIC CONDITIONS; Disability and Rehabilitation, 31(S1):S83-S87

I.F. 2008: 1,395

This introduction presents a brief review of projects carried out in Italy applying ICF model. ICF allows to capture and appropriately describe human functioning and disability in terms of body functions and body structures, life activities, participation in society, modulating contextual factors. ICF framework allows to have a complex profile of persons with different medical conditions. Several clinical studies have been performed using ICF and ICF-CY, in Italy from the publication of ICF in 2001 and ICF-CY in 2007. Most of them highlight the utility and feasibility of using ICF to describe the functional profiles of patients with different health conditions. The clinical applications of ICF clearly show that ICFbased functional profiles are useful in designing better interventions – directed not only to patients' health condition and symptoms but also to the most important activities of their daily living and environmental factors that may act as barriers or facilitators to the patients' recovery, well-being and participation in society.

Locatelli Federica, Bersano Anna, Ballabio Elena, Lanfranconi Silvia, Papadimitriou Dimitra, Strazzer Sandra, Bresolin Nereo, Comi Giacomo Pietro, Corti Stefania (2009); STEM CELL THERAPY IN STROKE; Cellular and Molecular Life Sciences, 66(5):757-772 - Review

I.F. 2008: 5,511

Recent work has focused on cell transplantation as a therapeutic option following ischemic stroke, based on animal studies showing that cells transplanted to the brain not only survive, but also lead to functional improvement. Neural degeneration after ischemia is not selective but involves different neuronal populations, as well as glial and endothelial cell types. In models of stroke, the principal mechanism by which any improvement has been observed, has been attributed to the release of trophic factors, possibly promoting endogenous repair mechanisms, reducing cell death and stimulating neurogenesis and angiogenesis. Initial human studies indicate that stem cell therapy may be technically feasible in stroke patients, however, issues still need to be addressed for use in human subjects.

Lolmede Karine, Campana Lara, Vezzoli Michela*, Bosurgi Lidia, Tonlorenzi Rossana, Clementi Emilio, Bianchi E. Marco, Cossu Giulio, Manfredi Angelo A., Brunelli Silvia, Rovere-Querini Patrizia (2009); INFLAMMATORY AND ALTERNATIVELY ACTIVATED HUMAN MACROPHAGES ATTRACT VESSEL-ASSOCIATED STEM CELLS, RELYING ON SEPARATE HMGB1- AND MMP-9-DEPENDENT

PATHWAYS; Journal of Leukocyte Biology, 85(5):779-787

I.F. 2008: 4,605

Inflammatory macrophages recruited at the site of damaged muscles progressively acquire an alternative activation profile. Inflammatory (M1) and alternatively activated (M2) macrophages exert various and even opposite functions. M1 cells amplify tissue damage, and M2 cells dispose of necrotic fibers and deliver survival signals to myogenic precursors, finally supporting healing. A critical step in muscle healing is the recruitment of myogenic stem cells, including vessel-associated stem cells (mesoangioblasts), which have been demonstrated to home to damaged skeletal muscle selectively and preferentially. Little information is available about the signals involved and the role played by infiltrating macrophages. Here, we report that the polarization of macrophages dramatically skews the secretion of high mobility group box 1 (HMGB1), TNF-alpha, vascular endothelial growth factor, and metalloproteinase 9 (MMP-9), molecules involved in the regulation of cell diapedesis and migration. All polarized macrophage populations were strikingly effective at inducing mesoangioblast migration. By means of specific inhibitors, we verified that the recruitment of mesoangioblasts requires the secretion of HMGB1 and TNF-alpha by M1 cells and of MMP-9 by M2 cells. Together, these data demonstrate a feature, unrecognized previously, of macrophages: their ability to attract stem cells, which is conserved throughout their polarization. Moreover, they open the possibility of novel strategies, aimed at interfering selectively with signals that recruit blood-derived stem cells toward pro- or anti-inflammatory macrophages.

Longatti Pierluigi, Fiorindi Alessandro, Martinuzzi Andrea, Feletti Alberto (2009); PRIMARY OBSTRUCTION OF THE FOURTH VENTRICLE OUTLETS: NEUROENDOSCOPIC APPROACH AND ANATOMIC DESCRIPTION; Neurosurgery, 65(6):1078-1086

I.F. 2008: 3,398

OBJECTIVE: Primary obstruction of the foramina of Magendie and Luschka is an uncommon and still unclear cause of noncommunicating hydrocephalus. The aim of this work is the description, for the first time, of the inner aspect of these velar obstructions of the fourth ventricle outlets and the demonstration of the efficacy of neuroendoscopic treatment. METHODS: Of 240 hydrocephalic patients treated in our institution with endoscopic third ventriculostomy, a subgroup of 10 cases with closure of the fourth ventricular outlets without associated Chiari malformation and syringomyelia was selected. In all of these cases, a transaqueductal endoscopic navigation of the fourth ventricle was performed, and the obstructed outlets were inspected. All of the clinical

data and, in particular, the videotape records of endoscopic operations, as well as the cine-magnetic resonance imaging scans, were reviewed to evaluate their patency status. RESULTS: Various degrees of stenosis were found endoscopically: restriction of the Magendie contour with thick and opaque membrane, transparent spider web-like membrane, and dense membrane with fissures acting as valves. Endoscopic third ventriculostomy was effective in almost all patients, although we noticed an unforeseen high incidence of closure of the stoma. The restored normal cerebrospinal fluid flux after ventriculocisternostomy and magendieplasty was demonstrated by comparative study of cerebrospinal fluid flow measurements by cine-magnetic resonance imaging. CONCLUSION: This report demonstrates the effectiveness of neuroendoscopic third ventriculostomy as well as magendiestomy in cases of tetraventricular hydrocephalus attributable to primary obstruction of the outlets of the fourth ventricle and, for the first time, presents direct images of various types of outlet obstructive pathology.

Losito Luciana, De Rinaldis Marta, Gennaro Leonarda, Priori Silvia G., Bloise Raffaella, Bassi Maria Teresa, Bresolin Nereo, Trabacca Antonio (2009); CHARCOT-MARIE-TOOTH TYPE 1A IN A CHILD WITH LONG QT SYNDROME; European Journal of Paediatric Neurology, 13(5):459-462

I.F. 2008: 1,421

Charcot-Marie-Tooth disease (CMTD) is a hereditary demyelinating peripheral neuropathy clinically presenting with sensory and motor defects, but rarely affecting cardiac function. Long QT syndrome (LQTS) is a congenital or acquired cardiovascular disorder characterized by ventricular depolarization defect. No studies reported CMTD in association with LQTS. We describe a child and his family who had both CMT1A and LQTS.

Massimino Maura, Gandola Lorenza, Spreafico Filippo, Biassoni Veronica, Luksch Roberto, Collini Paola, Solero Carlo L., Simonetti Fabio, Pignoli Emanuele, Cefalo Graziella, Poggi Geraldina, Modena Piergiorgio, Mariani Luigi, Potepan Paolo, Podda Marta, Casanova Michela, Pecori Emilia, Acerno Stefania, Ferrari Andrea, Terenziani Monica, Meazza Cristina, Polastri Daniela, Ravagnani Fernando, Fossati-Bellani Franca (2009); NO SALVAGE USING HIGH-DOSE CHEMOTHERAPY PLUS/MINUS RE-IRRADIATION FOR RELAPSING, PREVIOUSLY IRRADIATED MEDULLOBLASTOMA; International Journal of Radiation Oncology Biology Physics, 73(5):1358-1363

I.F. 2008: 4,639

PURPOSE: Myeloablative regimens were frequently used for medulloblastoma relapsing after craniospinal irradiation (CSI): in 1997-2002, we used repeated surgery, standard-dose and myeloablative chemotherapy, and reirradiation. METHODS AND MATERIALS: In 10 patients, reinduction included sequential high-dose etoposide, high-dose cyclophosphamide/vincristine, and high-dose carboplatin/vincristine, then two myeloablative courses with high-dose thiotepa (+/- carboplatin); 6 other patients received two of four courses of cisplatin/etoposide. Hematopoietic precursor mobilization followed high-dose etoposide or high-dose cyclophosphamide or cisplatin/etoposide therapy. After the overall chemotherapy program, reirradiation was prescribed when possible. RESULTS: Seventeen patients were treated: previous treatment included CSI of 19.5-36 Gy with posterior fossa/tumor boost and chemotherapy in 16 patients. Fifteen patients were in their first and 2 in their second and third relapses, respectively. First progression-free survival had lasted a median of 26 months. Relapse sites included leptomeninges in 9 patients, spine in 4 patients, posterior fossa in 3 patients, and brain in 1 patient. Three patients underwent complete resection of recurrence, and 10 underwent reirradiation. Twelve of 14 patients with assessable tumor had an objective response after reinduction; 2 experienced progression and were not given the myeloablative courses. Remission lasted a median of 16 months. Additional relapses appeared in 13 patients continuing the treatment. Fifteen patients died of progression and 1 died of pneumonia 13 months after relapse. The only survivor at 93 months had a single spinal metastasis that was excised and irradiated. Survival for the series as a whole was 11-93 months, with a median of 41 months. CONCLUSIONS: Despite responses being obtained and ample use of surgery and reirradiation, second-line therapy with myeloablative schedules was not curative, barring a few exceptions. A salvage therapy for medulloblastoma after CSI still needs to be sought.

Maziade Michel, Rouleau Nancie, Gingras Nathalie, Boutin Pierrette, Paradis Marie-Eve, Jomphe Valerie, Letourneau Karine, Gilbert Elsa, Lefebvre Andree-Anne, Dorè Marie-Claire, Marino Cecilia, Battaglia Marco, Merette Chantal, Roy Marc-Andrè (2009); SHARED NEUROCOGNITIVE DYSFUNCTIONS IN YOUNG OFFSPRINGS AT EXTREME RISK FOR SCHIZOPHRENIA OR BIPOLAR DISORDER IN EASTERN QUEBEC MULTIGENERATIONAL FAMILIES; Schizophrenia Bulletin, 35(5):919-930

I.F. 2008: 6,592

BACKGROUND: Adult patients having schizophrenia (SZ) or bipolar disorder (BP) may have in common neurocognitive deficits. Former evidence sug-

gests impairments in several neuropsychological functions in young offspring at genetic risk for SZ or BP. Moreover, a dose-response relation may exist between the degree of familial loading and cognitive impairments. This study examines the cognitive functioning of high-risk (HR) offspring of parents having schizophrenia (HRSZ) and high-risk offspring of parents having bipolar disorder (HRBP) descending from densely affected kindreds. METHODS: The sample consisted of 45 young offspring (mean age of 17.3 years) born to a parent having SZ or BP descending from large multigenerational families of Eastern Québec that are densely affected by SZ or BP and followed up since 1989. The offspring were administered a lifetime best-estimate diagnostic procedure (Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition [DSM-IV]) and an extensive standard neuropsychological battery. Raw scores were compared with age- and gender-matched controls. RESULTS: The offspring displayed differences in memory and executive functions when compared with controls. Moderate to large effect sizes (Cohen d) ranging from 0.65 to 1.25 (for IQ and memory) were observed. Several of the cognitive dysfunctions were present in both HRSZ and HRBP, even when considering DSM-IV clinical status. CONCLUSIONS: HRSZ and HRBP shared several aspects of their cognitive impairment. Our data suggest that the extremely high genetic and familial loading of these HRs may have contributed to a quantitatively increased magnitude of the cognitive impairments in both HR subgroups, especially in memory. These offspring at heightened risk present difficulties in processing information that warrant preventive research.

Montirosso Rosario, Ceppi Elisa, D'Aloisio Chiara, Zucca Claudio, Borgatti Renato (2009); INTERNATIONAL CLASSIFICATION OF FUNCTIONING, DISABILITY AND HEALTH IN SUBJECTS WITH ALTERNATING HEMIPLEGIA OF CHILDHOOD; Disability and Rehabilitation, 31(S1):108-115

I.F. 2008: 1,395

Purpose. To examine the functioning of patients with alternating hemiplegia of childhood (AHC), a very rare clinical condition manifesting with recurrent episodes of hemiplegia lasting from few minutes to several days, paroxysmal occurrence of tonic/dystonic attacks and other autonomic disturbances. Furthermore, patients exhibit chronic disabilities as well as mental retardation, epilepsy and motor disorders that affect the patients' everyday functioning to a considerable extent.
Method. Data about 25 patients with AHC (F¼13) aged 3–34 years were collected with International Classification of Functioning Disability and Health (ICF) questionnaires. Data analysis was carried out on four age groups: 3–6,

7–12, 13–18 and 418 years, using only the ICF questionnaires' cross-age items.
Results. In the body functions component, paying attention, seeing, muscle tones were common problems for all age groups. In the activity and participation component, all participants showed problems in basic interpersonal interactions. Finally, in the environmental factors component, barriers concern the climate and health services. On the other hand, families reported adequate support from social services.
Conclusions. Although these findings must be validated, they appear to be promising and can contribute to understanding the daily functioning features of patients with AHC.

Montirosso Rosario, Morandi Francesco, D'Aloisio Chiara, Berna Anna, Provenzi Livio, Borgatti Renato (2009); INTERNATIONAL CLASSIFICATION OF FUNCTIONING, DISABILITY AND HEALTH IN CHILDREN WITH CONGENITAL CENTRAL HYPOVENTILATION SYNDROME; Disability and Rehabilitation, 31(S1):S144-S152

I.F 2008: 1,395

Purpose. The main aim of this study is to examine the functioning of children with congenital central hypoventilation syndrome (CCHS), a rare disorder of respiratory control associated with physiological and anatomical manifestations of a generalised autonomic nervous system dysfunction, using WHO's International Classification of Functioning, Disability and Health, Children and Youth version (ICF-CY).
Method. The data of 26 children, (F¼17) aged 1.5–17.5 years, were collected. Data were analysed in the following four age groups: 53, 3–6, 7–12 and 13–18 years, using only the ICF-CY questionnaires' cross-age items.
Results. In the body functions, component breathing and paying attention were common problems for four age groups. In the activity and participation component, all children, except adolescents, showed problems with language. Furthermore, problems in social interaction were evident for all age ranges, except the youngest. Finally, in the environmental factors component, parents reported limitations concerning the natural environment and human-made changes to the environment that were common to all ages.
Conclusions. The study supports the usefulness of supplementing diagnostic classifications with functional classifications to obtain complete information on health-related conditions in children with CCHS.

Moro Valentina, Pernigo Simone, Urgesi Cosimo, Zapparoli P., Aglioti Salvatore (2009); FINGER RECOGNITION AND GESTURE

IMITATION IN GERSTMANN'S SYNDROME; Neurocase, 15(1):13-23

I.F. 2008: 0,918

We report the association between finger agnosia and gesture imitation deficits in a right-handed, right-hemisphere damaged patient with Gerstmann's syndrome (GS), a neuropsychological syndrome characterized by finger and toe agnosia, left-right disorientation and dyscalculia. No language deficits were found. The patient showed a gestural imitation deficit that specifically involved finger movements and postures. The association between finger recognition and imitation deficits suggests that both static and dynamic aspects of finger representations are impaired in GS. We suggest that GS is a disorder of body representation that involves hands and fingers, that is, the non-facial body parts most involved in social interactions.

Nery Fabiano G., Chen Hua-Hsuan, Hatch John P., Nicoletti Mark A., Brambilla Paolo, Sassi Roberto B., Mallinger Alan G., Keshavan Matcheri S., Soares Jair C. (2009); ORBITOFRONTAL CORTEX GRAY MATTER VOLUMES IN BIPOLAR DISORDER PATIENTS: A REGION-OF-INTEREST MRI STUDY; Bipolar Disorders, 11(2):145-153

I.F. 2008: 3,959

OBJECTIVES: Functional and postmortem studies suggest that the orbitofrontal cortex (OFC) is involved in the pathophysiology of bipolar disorder (BD). This anatomical magnetic resonance imaging (MRI) study examined whether BD patients have smaller OFC gray matter volumes compared to healthy comparison subjects (HC). METHODS: Twenty-eight BD patients were compared to 28 age- and gender-matched HC. Subjects underwent a 1.5T MRI with 3D spoiled gradient recalled acquisition. Total OFC and medial and lateral subdivisions were manually traced by a blinded examiner. Images were segmented and gray matter volumes were calculated using an automated method. RESULTS: Analysis of covariance, with intracranial volume as covariate, showed that BD patients and HC did not differ in gray matter volumes of total OFC or its subdivisions. However, total OFC gray matter volume was significantly smaller in depressed patients (n = 10) compared to euthymic patients (n = 18). Moreover, total OFC gray matter volumes were inversely correlated with depressive symptom intensity, as assessed by the Hamilton Depression Rating Scale. OFC gray matter volumes were not related to lithium treatment, age at disease onset, number of episodes, or family history of mood disorders. CONCLUSIONS: Our results suggest that abnormal OFC gray matter volumes are not a pervasive characteristic of BD, but may be associated with specific clinical features of the disorder.

Nifosi Francesco, Martinuzzi Andrea, Toffanin Tommaso, Costanzo Raffaella, Vestri Alec, Battaglia Maria Amalia, Bertagnoni Gian E., Lupi Andrea, Amistà Pietro, Carollo Carla, Perini Giulia (2009); HIPPOCAMPAL REMODELLING AFTER MDMA NEUROTOXICITY: A SINGLE CASE STUDY; World Journal of Biological Psychiatry (The), Case Report, 10(4-3):961-968

I.F. 2008: 3,582

Acute ingestion of MDMA (ecstasy) causes a transient marked increase in serotonin and dopamine at central synapses. Recent studies demonstrated that MDMA induces damage of serotonergic nerve terminals and alters hippocampal processing. Pronounced cognitive deficits in MDMA users affect learning and memory abilities. This pattern of predominant and long-lasting memory dysfunction suggests that the functioning of the hippocampus might be affected by the neurotoxic effects of MDMA. We present the case of a 16-year-old girl who developed an acute organic and psychotic syndrome caused by occasional use of low to moderate dose of MDMA. Serial neuroimaging ((18)F-FDG-PET and brain MRI) were correlated with her neurocognitive performance and clinical evolution. The structural and metabolic changes correlated with a severe cognitive impairment. After 16 months of intensive neuropsychological rehabilitation she showed significant improvement in hippocampal-related memory cognitive functions, which correlated with normalization of her (18)F-FDG-PET and remarkable hippocampal remodelling. This case report indicates that even non-chronic MDMA use may cause subacute toxic encephalopathy in which the clinical evolution is paralleled by neuroimaging changes in specific cerebral areas. The most relevant aspect is the reversibility of the volumetric changes, which may be the structural correlate of an ongoing hippocampal remodelling.

Nobile Maria, Rusconi Marianna, Bellina Monica, Marino Cecilia, Giorda Roberto, Carlet Ombretta, Vanzin Laura, Molteni Massimo, Battaglia Marco (2009); THE INFLUENCE OF FAMILY STRUCTURE, THE TPH2 G-703T AND THE 5-HTTLPR SEROTONERGIC GENES UPON AFFECTIVE PROBLEMS IN CHILDREN AGED 10-14 YEARS; Journal of Child Psychology and Psychiatry, 50(3):317-325

I.F. 2008: 4,854

BACKGROUND: Both genetic and psychosocial risk factors influence the risk for depression in development. While the impacts of family structure and of serotonergic polymorphisms upon individual differences for affective problems have been investigated separately, they have never been considered together in a gene-environment interplay perspective. METHODS: We exam-

ined the effects of family structure and two serotonergic polymorphisms (the TPH2 G-703T and the 5-HTTLPR) upon depressive symptoms assessed by the new CBCL/6-18 DSM-oriented Affective Problems scale in a general population sample of 607 Italian children aged 10-14 years. RESULTS: Belonging to 'one-parent' families, the TPH2 G-703T 'G variant', and the 5-HTTLPR 'short' alleles were associated - both alone and in apparent gene-by-environment interaction - with higher Affective Problems scores. As predicted by quantitative genetics theory, both polymorphisms contributed with a small effect size, while 'family structure' had a moderate effect size. CONCLUSIONS: A putative hazard factor impinging on individual risk at the family-wide level, namely family structure, appears to act interactively with two pivotal serotonergic genes in heightening risk for Affective Problems. Although it remains to be demonstrated that belonging to a one- rather than a two-parent family has true environmental causal effects on Affective Problems, these data may contribute to identify/prevent risk for depression in childhood.

Novara Francesca, Beri Silvana, Bernardo Maria Ester, Bellazzi Riccardo, Malovini Alberto, Ciccone Roberto, Cometa Angela Maria, Locatelli Franco, Giorda Roberto, Zuffardi Orsetta (2009); DIFFERENT MOLECULAR MECHANISMS 9P21 DELETIONS IN ACUTE LYMPHOBLASTIC LEUKEMIA OF CHILDHOOD; Human Genetics, 126(4):511-520

I.F. 2008: 4,042

Deletion of chromosome 9p21 is a crucial event for the development of several cancers including acute lymphoblastic leukemia (ALL). Double strand breaks (DSBs) triggering 9p21 deletions in ALL have been reported to occur at a few defined sites by illegitimate action of the V(D)J recombination activating protein complex. We have cloned 23 breakpoint junctions for a total of 46 breakpoints in 17 childhood ALL (9 B- and 8 T-lineages) showing different size deletions at one or both homologous chromosomes 9 to investigate which particular sequences make the region susceptible to interstitial deletion. We found that half of 9p21 deletion breakpoints were mediated by ectopic V(D)J recombination mechanisms whereas the remaining half were associated to repeated sequences, including some with potential for non-B DNA structure formation. Other mechanisms, such as microhomology-mediated repair, that are common in other cancers, play only a very minor role in ALL. Nucleotide insertions at breakpoint junctions and microinversions flanking the breakpoints have been detected at 20/23 and 2/23 breakpoint junctions, respectively, both in the presence of recombination signal sequence (RSS)-like sequences and of other unspecific sequences. The majority of

breakpoints were unique except for two cases, both T-ALL, showing identical deletions. Four of the 46 breakpoints coincide with those reported in other cases, thus confirming the presence of recurrent deletion hotspots. Among the six cases with heterozygous 9p deletions, we found that the remaining CDKN2A and CDKN2B alleles were hypermethylated at CpG islands.

Orso Genny, Pendin Diana, Liu Song, Tosetto Jessica, Moss Tyler J., Faust Joseph E., Micaroni Massimo, Egorova Anastasia, Martinuzzi Andrea, McNew James A., Daga Andrea (2009); HOMOTYPIC FUSION OF ER MEMBRANES REQUIRES THE DYNAMIN-LIKE GTPASE ATLASTIN; Nature, 460(7258):978-983

I.F. 2008: 31,434

Establishment and maintenance of proper architecture is essential for endoplasmic reticulum (ER) function. Homotypic membrane fusion is required for ER biogenesis and maintenance, and has been shown to depend on GTP hydrolysis. Here we demonstrate that Drosophila Atlastin--the fly homologue of the mammalian GTPase atlastin 1 involved in hereditary spastic paraplegia--localizes on ER membranes and that its loss causes ER fragmentation. Drosophila Atlastin embedded in distinct membranes has the ability to form trans-oligomeric complexes and its overexpression induces enlargement of ER profiles, consistent with excessive fusion of ER membranes. In vitro experiments confirm that Atlastin autonomously drives membrane fusion in a GTP-dependent fashion. In contrast, GTPase-deficient Atlastin is inactive, unable to form trans-oligomeric complexes owing to failure to self-associate, and incapable of promoting fusion in vitro. These results demonstrate that Atlastin mediates membrane tethering and fusion and strongly suggest that it is the GTPase activity that is required for ER homotypic fusion.

Oteri Alessandro, Cattaneo Maria Teresa, Filipazzi Virginio, Ferrario Sabrina, Gambaro Anna, Isabella Luigi, Tosca Nicoletta, Clementi Emilio, Radice Sonia, Piazza Elena (2009); A CASE OF BULLOUS DERMATITIS INDUCED BY ERLOTINIB; The Oncologist, 14(12):1201-1204

I.F. 2008: 6,630

Herein, we report a case of bullous dermatitis that occurred in a 61-year-old woman 5 days after beginning therapy with erlotinib for the treatment of stage IV pulmonary adenocarcinoma with metastases at the hypophyseal level. Skin reactions are the most common adverse drug reactions (ADRs) associated with epidermal growth factor receptor tyrosine kinase (EGFR-TK) inhibitors, and acneiform rash is the most frequently reported ADR in pa-

tients treated with erlotinib. To our knowledge, this is the first case of bullous dermatitis induced by erlotinib. This report highlights the need for additional research in the field of skin toxicity of EGFR-TK inhibitors.

Parini Sergio, Maggi Luca, Turconi Anna Carla, Andreoni Giuseppe (2009); A ROBUST AND SELF-PACED BCI SYSTEM BASED ON A FOUR CLASS SSVEP PARADIGM: ALGORITHMS AND PROTOCOLS FOR A HIGH-TRANSFER-RATE DIRECT BRAIN COMMUNICATION; Computational Intelligence and Neuroscience, doi:10.1155/2009/864564

I.F. 2008: 0,000

In this paper, we present, with particular focus on the adopted processing and identification chain and protocol-related solutions, a whole self-paced brain-computer interface system based on a 4-class steady-state visual evoked potentials (SSVEPs) paradigm. The proposed system incorporates an automated spatial filtering technique centred on the common spatial patterns (CSPs) method, an autoscaled and effective signal features extraction which is used for providing an unsupervised biofeedback, and a robust self-paced classifier based on the discriminant analysis theory. The adopted operating protocol is structured in a screening, training, and testing phase aimed at collecting user-specific information regarding best stimulation frequencies, optimal sources identification, and overall system processing chain calibration in only a few minutes. The system, validated on 11 healthy/pathologic subjects, has proven to be reliable in terms of achievable communication speed (up to 70 bit/min) and very robust to false positive identifications.

Pensiero Stefano, Fabbro Franco, Michieletto Paola, Accardo Agostino, Brambilla Paolo (2009); SACCADIC CHARACTERISTICS IN AUTISTIC CHILDREN; Functional Neurology, XXIV(3):153-158

I.F. 2008: 1,133

Some studies suggest that individuals with autism present abnormal saccadic eye movements due to an altered strategy for exploration of the surrounding environment. In this study, potential early abnormalities of saccadic movements were explored in 14 male children with autism (5- to 12-year-olds) and in 20 agematched normal males. Only one patient showed clear abnormalities of the “main sequence”; all the other patients, although showing slight changes in saccadic eye movements, did not present classic deficits. Therefore our results did not confirm the presence of saccadic movement alterations in the early stage of autism. Nonetheless, tracts of saccadic initiation failure, continuous changes in saccadic velocity profiles, and instability of fixation were often observed in the autistic population. These findings could be the expression of an early brainstem impairment in autism.

Perego Paolo, Forti Sara, Crippa Alessandro, Valli Angela, Reni Gianluigi (2009); REACH AND THROW MOVEMENT ANALYSIS WITH SUPPORT VECTOR MACHINES IN EARLY DIAGNOSIS OF AUTISM; Conference Proceedings: ... Annual International Conference IEEE Engineering in Medicine and Biology, 1:2555-2558

I.F. 2009: 0,000

Movement disturbances play an intrinsic part in autism. Upper limb movements like reach-and-throw seem to be helpful in early identification of children affected by autism. Nevertheless few works investigate the application of classifying methods to upper limb movements. In this study we used a machine learning approach Support Vector Machine (SVM) for identifying peculiar features in reach-and-throw movements. 10 pre-scholar age children with autism and 10 control subjects performing the same exercises were analyzed. The SVM algorithm proved to be able to separate the two groups: accuracy of 100% was achieved with a soft margin algorithm, and accuracy of 92.5% with a more conservative one. These results were obtained with a radial basis function kernel, suggesting that a non-linear analysis is possibly required.

Petacchi Elisa, Armellin Maria Teresa, Facchin Dina, Gubernale Marco, Moret Ornella, Buffoni Mara, Salghetti Annamaria, Martinuzzi Andrea (2009); THE DYSTONIC CHILD TREATED WITH DEEP BRAIN STIMULATION: ICF READING OF A HIGH-TECH APPROACH; Disability and Rehabilitation, 31(S1):S159-S169

I.F. 2008: 1,395

Purpose. The available tools used to describe childhood dystonia tend to offer a monodimensional view of the person functioning, which may overlook significant changes induced by treatment. We applied the International Classification of Functioning, Disability and Health (ICF) perspective to the description of the clinical picture of a dystonic child treated with deep brain stimulation (DBS) to get a more global representation of the treatment effect. Method. An 8-year-old child with secondary dystonia was selected within the institutional program for advanced treatment of pediatric dystonia as a candidate for bilateral implantation of electrodes into globus pallidus and chronic stimulation. The International Classification of Functioning, Disability and Health -children and youth (ICF-CY) based project and program format was used by the rehabilitation team to define the clinical picture, rehabilitation

objectives, and to verify the outcome.
Results. The rehabilitation project and program included 39 ICF categories: 14 body functions, two body structures, 18 activities and participation, and five environment. On such basis we defined the individualized specific rehabilitation objectives and we checked for clinical changes after DBS.
Conclusion. The ICF-CY format provides a complete and balanced profile of functioning in secondary dystonia treated with DBS and it could offer a novel perspective for outcome evaluation.

Piccinini Luigi, Cimolin Veronica, Turconi Anna Carla, Galli Manuela (2009); RELATIONSHIP BETWEEN KINEMATIC KNEE DEVIATIONS AND FEMORAL ANTEVERSION IN CHILDREN WITH CEREBRAL PALSY; Hip International, 19(1-S6):S63-S68

I.F. 2008: 0,215

The aim of the study was to determine the possible correlation between the degree of femoral anteversion and the quantitative data obtained by 3D Gait Analysis (GA) and then to investigate the relationship between femoral anteversion and the reduced knee flexion during swing phase in children with Cerebral Palsy. Twenty-seven diplegic children with severe rectus femoris spasticity and 20 healthy children (CG) were considered. Clinical evaluation of femoral anteversion, Duncan Ely test and Gait Analysis were performed in all patients. From Gait Analysis data some indices were identified and calculated and statistical analysis performed. Clinical evaluations made the distinction between patients with excessive femoral anteversion (Group 1) and those with normal value (Group 2). Both groups showed a blunt maximum of knee flexion in swing (KMSw), representative of rectus femoris spasticity, but two different gait strategies were found for the timing of KMSw. Group 1 exhibited a reduced KMSw value with its timing close to normal value and an excessive hip internal rotation (Mean Hip Rotation index), correlated to high femoral anteversion; Group 2 presented a limited KMSw and a significant delay of its timing, with Mean Hip Rotation index close to Control Group. No differences were found for other indices. The results demonstrated that the presence of reduced KMSw only can be directly connected to excessive femoral anteversion; the coexistence of reduced KMSw and its delayed timing reveals that the rectus femoris spasticity may be due to rectus spasticity added to an incorrect motor selective control. The results are clinically crucial for treatment strategies (derotative femoral osteotomy vs rectus transfer).

Poggi Geraldina, Liscio Mariarosaria, Pastore Valentina, Adduci Annarita, Galbiati Susanna, Spreafico Filippo, Gandola Lorenza,

Massimino Maura (2009); PSYCHOLOGICAL INTERVENTION IN YOUNG BRAIN TUMOR SURVIVORS: THE EFFICACY OF THE COGNITIVE BEHAVIOURAL APPROACH; Disability and Rehabilitation, 31(13):1066-1073

I.F. 2008: 1,395

PURPOSE: Cognitive and behavioural therapy (CBT) is often used to treat behavioural and emotional disorders in children, and its efficacy has been described in several studies. As behavioural and emotional disorders are frequent sequelae in brain tumor survivors, the goal of this work is to describe the efficacy of a CBT intervention in the treatment of young brain tumor survivors. METHODS: Forty young patients, aged 4-18 years, were included in the study. The treatment group, composed of 17 patients, received sessions of CBT. The Child Behaviour Checklist 4-18 (CBCL/4-18) and the Vineland Adaptive Behavioural Scales (VABS) were administered to parents at the beginning and at the end of the hospitalisation. The statistical significance of changes for clinical subjects during the CBT administration was estimated. RESULTS: With regard to the CBCL/4-18, the clinical group showed a significant advantage on the withdrawn, somatic complaints, social problems, attention problems, internalising and total problem scales. On the VABS, the treatment group improved to a significantly greater extent in the social skills domain. CONCLUSIONS: These results substantiate our assumption that CBT is an effective intervention for young patients surviving brain tumors and may be particularly helpful to younger individuals in managing cancer-related limitations.

Porcelli Anna Maria, Angelin Alessia, Ghelli Anna, Mariani Elisa, Martinuzzi Andrea, Carelli Valerio, Petronilli Valeria, Bernardi Paolo, Rugolo Michela (2009); RESPIRATORY COMPLEX I DYSFUNCTION DUE TO MITOCHONDRIAL DNA MUTATIONS SHIFTS THE VOLTAGE THRESHOLD FOR OPENING OF THE PERMEABILITY TRANSITION PORE TOWARD RESTING LEVELS; Journal of Biological Chemistry, 284(4):2045-2052

I.F. 2008: 5,520

We have studied mitochondrial bioenergetics in HL180 cells (a cybrid line harboring the T14484C/ND6 and G14279A/ND6 mtDNA mutations of Leber hereditary optic neuropathy, leading to an approximately 50% decrease of ATP synthesis) and XTC.UC1 cells (derived from a thyroid oncocytoma bearing a disruptive frameshift mutation in MT-ND1, which impairs complex I assembly). The addition of rotenone to HL180 cells and of antimycin A to XTC.UC1 cells caused fast mitochondrial membrane depolarization that was

prevented by treatment with cyclosporin A, intracellular Ca2+ chelators, and antioxidant. Both cell lines also displayed an anomalous response to oligomycin, with rapid onset of depolarization that was prevented by cyclosporin A and by overexpression of Bcl-2. These findings indicate that depolarization by respiratory chain inhibitors and oligomycin was due to opening of the mitochondrial permeability transition pore (PTP). A shift of the threshold voltage for PTP opening close to the resting potential may therefore be the underlying cause facilitating cell death in diseases affecting complex I activity. This study provides a unifying reading frame for previous observations on mitochondrial dysfunction, bioenergetic defects, and Ca2+ deregulation in mitochondrial diseases. Therapeutic strategies aimed at normalizing the PTP voltage threshold may be instrumental in ameliorating the course of complex I-dependent mitochondrial diseases.

Restuccia Domenico, Zanini Sergio, Cazzagon Monica, Del Piero Ivana, Martucci Lucia, Della Marca Giacomo (2009); SOMATOSENSORY MISMATCH NEGATIVITY IN HEALTHY CHILDREN; Developmental Medicine and Child Neurology, 51(12):991-998

I.F. 2008: 2,561

AIM: Event-related potentials (ERPs) obtained when focused attention is kept away from the stimulus (unnoticed stimulation) are possibly linked to automatic mismatch-detection mechanisms, and could be a useful tool to investigate sensory discrimination ability. By considering the high impact of impaired somatosensory integration on many neurological disturbances in children, we aimed to verify whether mismatch-related responses to somatosensory stimulation could be obtained in healthy children. METHOD: Eleven healthy participants (age range 6-11y, mean 8y 2mo, SD 1y 7mo; seven males, four females) underwent 'oddball' electrical stimulation of the right hand (80% frequent stimuli delivered to the thumb, 20% deviant stimuli delivered to the fifth finger). Data were compared with those obtained when the frequent stimuli to the thumb were omitted ('standard-omitted' protocol). ERPs were recorded at frontal, central, and parietal scalp locations. Children's overt attention was engaged by a demanding video game. RESULTS: In the oddball protocol, deviant stimulation elicited a left central negativity at about 160ms latency, followed by a left frontal negative response at about 220ms latency. Standard-omitted traces showed only a left parietal negative response spreading to right parietal regions. INTERPRETATION: Mismatch-related somatosensory responses can be reliably obtained in children, providing that appropriate technical contrivances are used. In clinical use, the frontal components,

which are present only during the oddball protocol, could be a reliable and unequivocal neurophysiological marker of the automatic mismatch-detection mechanism.

Revenkova Ekaterina, Focarelli Maria Luisa, Susani Lucia, Paulis Marianna, Bassi Maria Teresa, Mannini Linda, Frattini Annalisa, Delia Domenico, Krantz Ian, Vezzoni Paolo, Jessberger Rolf, Musio Antonio (2009); CORNELIA DE LANGE SYNDROME MUTATIONS IN SMC1A OR SMC3 AFFECT BINDING TO DNA; Human Molecular Genetics, 18(3):418-427

I.F. 2008: 7,249

Cornelia de Lange syndrome (CdLS) is a clinically heterogeneous developmental disorder characterized by facial dysmorphia, upper limb malformations, growth and cognitive retardation. Mutations in the sister chromatid cohesion factor genes NIPBL, SMC1A and SMC3 are present in approximately 65% of CdLS patients. In addition to their canonical roles in chromosome segregation, the cohesin proteins are involved in other biological processes such as regulation of gene expression, DNA repair and maintenance of genome stability. To gain insights into the molecular basis of CdLS, we analyzed the affinity of mutated SMC1A and SMC3 hinge domains for DNA. Mutated hinge dimers bind DNA with higher affinity than wild-type proteins. SMC1A- and SMC3-mutated CdLS cell lines display genomic instability and sensitivity to ionizing radiation and interstrand crosslinking agents. We propose that SMC1A and SMC3 CdLS mutations affect the dynamic association between SMC proteins and DNA, providing new clues to the underlying molecular cause of CdLS.

Riano Elena, Martignoni Monica, Mancuso Giuseppe, Cartelli Daniele, Crippa Francesca, Toldo Irene, Siciliano Gabriele, Di Bella Daniela, Taroni Franco, Bassi Maria Teresa, Cappelletti Graziella, Rugarli Elena I. (2009); PLEIOTROPIC EFFECTS OF SPASTIN ON NEURITE GROWTH DEPENDING ON EXPRESSION LEVELS; Journal of Neurochemistry, 108(5):1277-1288

I.F. 2008: 4,500

Hereditary spastic paraplegia (HSP) is characterized by weakness and spasticity of the lower limbs, owing to degeneration of corticospinal axons. The most common form is due to heterozygous mutations in the SPG4 gene, encoding spastin, a microtubule (MT)-severing protein. Here, we show that neurite growth in immortalized and primary neurons responds in pleiotropic ways to changes in spastin levels. Spastin depletion alters the development of

primary hippocampal neurons leading to abnormal neuron morphology, dystrophic neurites, and axonal growth defects. By live imaging with End-Binding Protein 3-Fluorescent Green Protein (EB3-GFP), a MT plus-end tracking protein, we ascertained that the assembly rate of MTs is reduced when spastin is down-regulated. Spastin over-expression at high levels strongly suppresses neurite maintenance, while slight spastin up-regulation using an endogenous promoter enhances neurite branching and elongation. Spastin severing activity is exerted preferentially on stable acetylated and detyrosinated MTs. We further show that SPG4 nonsense or splice site mutations found in hereditary spastic paraplegia patients result in reduced spastin levels, supporting haploinsufficiency as the molecular cause of the disease. Our study reveals that SPG4 is a dosage-sensitive gene, and broadens the understanding of the role of spastin in neurite growth and MT dynamics.

Sala Gessica, Trombin Federica, Mattavelli Laura, Beretta Simone, Tremolizzo Lucio, Andreoni Simona, Calabrese Elena, Sanvito Lara, Ferrarese Carlo (2009); LACK OF EVIDENCE FOR OXIDATIVE STRESS IN SPORADIC AMYOTROPHIC LATERAL SCLEROSIS FIBROBLASTS; Neurodegenerative Diseases, 6(1-2):9-15

I.F. 2008: 2,989

BACKGROUND: It is conceivable that an early therapeutic intervention in amyotrophic lateral sclerosis (ALS) would lead to better results in terms of disease progression for these patients. One possible strategy to increase the sensitivity of the diagnosis is represented by the use of biological parameters reflecting, for example, oxidative stress alterations associated with ALS. Such biomarkers would be valuable tools both for a better diagnostic evaluation and for studying the impact of therapeutic interventions on the disease course. A special category of experimental models is represented by peripheral cells obtained directly from patients (ex vivo). OBJECTIVE: In this study, primary fibroblasts obtained from 10 sporadic ALS (SALS) patients and 10 healthy matched controls were used to investigate a panel of parameters related to the oxidative status. METHODS: Reactive oxygen species production, protein carbonylation and nitration, susceptibility to hydrogen peroxide exposure, p38-mitogen-activated protein kinase activation and adenosine triphosphate intracellular content were evaluated. RESULTS: No significant difference was observed in all investigated parameters between patient and control cells, and no correlation with the disease severity was found. CONCLUSION: Collectively, our data show no major alterations of the oxidative and bioenergetic status in SALS cultured fibroblasts, suggesting that these cells do not represent a useful model to study the oxidative dysfunction as-

sociated with SALS. Copyright 2008 S. Karger AG, Basel.

Sala Michela, Lazzaretti Matteo, De Vidovich Giulia, Caverzasi Edgardo, Barale Francesco, D'Allio Giorgio, Brambilla Paolo (2009); ELECTROPHYSIOLOGICAL CHANGES OF CARDIAC FUNCTION DURING ANTIDEPRESSANT TREATMENT; Therapeutic Advances in Cardiovascular Disease, 3(1):29-43 - Review

I.F. 2008: 0,000

Some antidepressant agents can cause electrophysiological changes of cardiac function leading to ventricular arrhythmias and sudden death. However, antidepressants have also protective effects on the heart through their capacity to modulate cardiac autonomic-mediated physiological responses. Heart rate variability and QTc length are two strictly linked parameters that allow us to appreciate the effects of different drugs on cardiac physiology. Heart rate variability reflects functioning of the autonomic nervous system and possibly also regulation by the limbic system. Autonomic regulation of cardiac activity influences also cardiac repolarization and QT length, both directly and via its effects on heart rate. In this review we present the methodologies adopted to study the effect of antidepressant drugs on QT length and heart rate variability and we summarize data on electrophysiological changes related to antidepressant treatment. Clinical implications for the choice of different antidepressants in different clinical populations are discussed.

Sala Michela, Caverzasi Edgardo, Marraffini E., De Vidovich Giulia, Lazzaretti Matteo, D'Allio Giorgio, Isola Miriam, Balestrieri Matteo, D'Angelo E., Zappoli Thyrion F., Scagnelli P., Barale Francesco, Brambilla Paolo (2009); COGNITIVE MEMORY CONTROL IN BORDERLINE PERSONALITY DISORDER PATIENTS; Psychological Medicine, 39(5):845-853

I.F. 2008: 4,718

BACKGROUND: It has been demonstrated that the mechanism of cognitive memory control in humans is sustained by the hippocampus and prefrontal cortices, which have been found to be structurally and functionally abnormal in borderline personality disorder (BPD). We investigated whether the memory control mechanism is affected in BPD. METHOD: Nineteen Diagnostic and Statistical Manual of Mental Disorders (DSM)-IV BPD patients and 19 matched healthy controls (HC) performed a specific think/no-think paradigm exploring the capacity of remembering and suppressing pair of words previously learned. After the think-no think phase, the second member of each word pair has to be remembered either when subjects are presented with

the cue word showed at the beginning of the test (Same Probe Test; SPT) or when they are presented with an extra-list categorical word (Independent Probe Test; IPT). We evaluated the effect of suppression and of retrieval activity on later retention of words. RESULTS: Both on the SPT and on the IPT, HC showed the expected improvement of memory retrieval on to-be-remembered words, unlike BPD patients. On the SPT, HC, but not BPD patients, correctly recalled significantly more words among remembered words (RW) than among suppressed words (SW). Similarly to HC, subjects with BPD without a history of childhood abuse showed a significantly higher percentage of correctly recalled words among RW than among SW. CONCLUSIONS: The mechanism of active retrieval of memories and of improvement through repetition is impaired in BPD, particularly in those who experienced traumatic experiences. This impairment might play an important role, possibly resulting in the emergence of unwanted memories and dissociative symptoms.

Salghetti Annamaria, Betto Silvana, Russo Emanuela, Petacchi Elisa, Pradal Monica, Martinuzzi Andrea (2009); PROJECTING AND PROGRAMMING REHABILITATION BASED ON ICF-CY FORMAT IN A NEUROPEDIATRIC HOSPITAL UNIT; Disability and Rehabilitation, 31(S1):S55-S60

I.F. 2008: 1,395

Objective. To follow-up the impact of a format based upon the International Classification of Functioning Disability and Health, version for children and youth (ICF-CY) as a roadmap for in-hospital pediatric neuro-rehabilitation.
Design. Longitudinal study in a single Centre testing with impact assessment.
Subjects/patients. The team members of a tertiary care pediatric neuro-rehabilitation unit approaching 88 consecutive patients with various physical and/or neurological conditions.
Methods. A revised version of the protocol ICF-CY based format for rehabilitation projecting and programming was applied for 24 months. Impact on the rehabilitation team was assessed with an ad hoc questionnaire.
Results. Fifteen questionnaires were returned, all of them reporting strongly positive judgements. Time constraint was no longer considered the main problem, probably because of a learning effect. Ongoing utilization reduced the time needed and the problems perceived in completing the task.
Conclusion. This application confirmed that ICF contribute to improve the quality of interdisciplinary work and to share the rehabilitation process between team members and family. Indeed ICF-CY works efficiently as a roadmap for in-hospital pediatric neuro-rehabilitation. Its implementation results in perceived improvements in the process, ongoing utilization reduces the

time needed and the problems encountered in completing the task.

Sciorati Clara, Touvier Thierry, Buono Roberta, Pessina Patrizia, Francois Stephanie, Perrotta Cristiana, Meneveri Raffaella, Clementi Emilio, Brunelli Silvia (2009); NECTID IS EXPRESSED IN CACHECTIC SKELETAL MUSCLE TO PROTECT FIBERS FROM TUMOR-INDUCED WASTING; Journal of Cell Science, 122(8):1119-1125

I.F. 2008: 6,247

Skeletal muscles of subjects with advanced cancer undergo progressive wasting, referred to as cachexia. Cachexia is an important area for medical research because strategies proposed until now have yielded little benefit. We have recently identified necdin as a key player in fetal and postnatal physiological myogenesis and in muscle regeneration. Here we show that necdin is selectively expressed in muscles of cachetic mice and prove that its expression is causally linked to a protective response of the tissue against tumor-induced wasting, inhibition of myogenic differentiation and fiber regeneration. Necdin carries out this role mainly via interference with TNFalpha signaling at various levels, including regulation of expression of TNFR1 and p53, and regulation of the activity of caspase 3 and caspase 9. These data suggest that inhibition of muscle wasting using necdin is a feasible approach to treat cachexia in neoplastic patients.

Tavano Alessandro, Galbiati Susanna, Recla Monica, Formica Francesca, Giordano Flavio, Genitori Lorenzo, Strazzer Sandra (2009); LANGUAGE AND COGNITION IN A BILINGUAL CHILD AFTER TRAUMATIC BRAIN INJURY IN INFANCY: LONG-TERM PLASTICITY AND VULNERABILITY; Brain Injury, 23(2):167-171

I.F. 2008: 1,116

PRIMARY OBJECTIVE: This study aimed at investigating the long-term effects of the combination of severity of injury and time of injury in a 6-year-old bilingual Arabic-Italian child who sustained a severe left traumatic brain injury at the age of 7 months. METHODS AND PROCEDURES: Standard neurological, cognitive and neuropsychological assessments were administered at 40 days after surgery and again at 18, 31, 62 and 73 months. MAIN OUTCOMES AND RESULTS: The child presented with developmental arrest at 18 and 31 months. Later on, right hemiparetic and oculomotor signs gradually improved to a significant extent, as well as dysexecutive, visuospatial and praxic deficits. At present, persistent language disorders in a fluent speech characterize the child's profile to a similar extent and type in both languages,

suggesting common underlying learning strategies which are ineffective for procedurally acquiring language. CONCLUSIONS: This case confirms that children who sustain severe left hemisphere traumatic brain injury in infancy present with increased vulnerability to linguistic deficits. Left frontotemporal, cortical-subcortical lesions which occur during very early language development may permanently disrupt the procedural language acquisition network required for first language acquisition.

Tomelleri Luisa, Jogia Jigar, Perlini Cinzia, Bellani Marcella, Ferro Adele, Rambaldelli Gianluca, Tansella Michele, Frangou Sophia, Brambilla Paolo, The Neuroimaging Network of the ECNP networks iniziative (2009); BRAIN STRUCTURAL CHANGES ASSOCIATED WITH CHRONICITY AND ANTIPSYCHOTIC TREATMENT IN SCHIZOPHRENIA; European Neuropsychopharmacology, 19(12):835-840

I.F. 2008: 3,661

Accumulating evidence suggest a life-long impact of disease related mechanisms on brain structure in schizophrenia which may be modified by antipsychotic treatment. The aim of the present study was to investigate in a large sample of patients with schizophrenia the effect of illness duration and antipsychotic treatment on brain structure. Seventy-one schizophrenic patients and 79 age and gender matched healthy participants underwent brain magnetic resonance imaging (MRI). All images were processed with voxel based morphometry, using SPM5. Compared to healthy participants, patients showed decrements in gray matter volume in the left medial and left inferior frontal gyrus. In addition, duration of illness was negatively associated with gray matter volume in prefrontal regions bilaterally, in the temporal pole on the left and the caudal superior temporal gyrus on the right. Cumulative exposure to antipsychotics correlated positively with gray matter volumes in the cingulate gyrus for typical agents and in the thalamus for atypical drugs. These findings (a) indicate that structural abnormalities in prefrontal and temporal cortices in schizophrenia are progressive and, (b) suggest that antipsychotic medication has a significant impact on brain morphology.

Trabacca Antonio, Dicuonzo Franca (2009); LIVING WITH ONE HEMISPHERE: A LARGE PORENCEPHALIC CYST; New England Journal of Medicine, Image, 361(16):1584

I.F. 2008: 50,017

Abstract non disponibile

Travaglini Lorena, Brancati Francesco, Attie-Bitach Tania, Audollent Sophie, Bertini Enrico, Kaplan Josseline, Perrault Isabelle, Iannicelli Miriam, Mancuso Brunella, Rigoli Luciana, Rozet Jean-Michel, Swistun Dominika, Tolentino Jerlyn, The International JSRD Study Group (Borgatti Renato, Cazzagon Monica), Dallapiccola Bruno, Gleeson Joseph G., Valente Enza Maria (2009); EXPANDING CEP290 MUTATIONAL SPECTRUM IN CILIOPATHIES; American Journal of Medical Genetics Part A, 149A(10):2173-2180

I.F. 2008: 2,555

Ciliopathies are an expanding group of rare conditions characterized by multiorgan involvement, that are caused by mutations in genes encoding for proteins of the primary cilium or its apparatus. Among these genes, CEP290 bears an intriguing allelic spectrum, being commonly mutated in Joubert syndrome and related disorders (JSRD), Meckel syndrome (MKS), Senior-Loken syndrome and isolated Leber congenital amaurosis (LCA). Although these conditions are recessively inherited, in a subset of patients only one CEP290 mutation could be detected. To assess whether genomic rearrangements involving the CEP290 gene could represent a possible mutational mechanism in these cases, exon dosage analysis on genomic DNA was performed in two groups of CEP290 heterozygous patients, including five JSRD/MKS cases and four LCA, respectively. In one JSRD patient, we identified a large heterozygous deletion encompassing CEP290 C-terminus that resulted in marked reduction of mRNA expression. No copy number alterations were identified in the remaining probands. The present work expands the CEP290 genotypic spectrum to include multiexon deletions. Although this mechanism does not appear to be frequent, screening for genomic rearrangements should be considered in patients in whom a single CEP290 mutated allele was identified.

Vantaggiato Chiara, Redaelli Francesca, Falcone Sestina, Perrotta Cristiana, Tonelli Alessandra, Bondioni Sara, Morbin Michela, Riva Daria, Saletti Veronica, Bonaglia Maria Clara, Giorda Roberto, Bresolin Nereo, Clementi Emilio, Bassi Maria Teresa (2009); A NOVEL CLN8 MUTATION IN LATE-INFANTILE-ONSET NCL REVEALS ASPECTS OF CLN8 NEUROBIOLOGICAL FUNCTION; Human Mutation, 30(7):1104-1116

I.F. 2008: 7,033

The late-infantile-onset forms of neuronal ceroid lipofuscinosis (LINCL) are the most genetically heterogeneous group among the autosomal recessive neuronal ceroid lipofuscinoses (NCLs), with causative mutations found in

CLN1, CLN2, CLN5, CLN6, CLN7 (MFSD8), and CLN8 genes. Homozygous mutations in CLN8 are associated with two distinct phenotypes: progressive epilepsy and mental retardation (EPMR), first identified in Finland; and a variant of late-infantile NCL (v-LINCL) described in a subset of Turkish and Italian patients. The function of the protein encoded by CLN8 is currently unknown. Here we report the identification of an Italian v-LINCL patient with a complete isodisomy of chromosome 8, leading to homozygosity of a maternally-inherited 3-bp deletion in CLN8 gene (c.180_182delGAA, p.Lys61del). Notably, uniparental disomy (UPD) has never been described associated with the NCLs. In addition, we provide evidence of the biological role of CLN8 characterized by expressing in different neuronal cell models the native protein, the protein carrying the mutation identified here, or three additional missense mutations previously described. Our results, validated through a gene silencing approach, indicate that CLN8 plays a role in cell proliferation during neuronal differentiation and in protection against cell death. © 2009 Wiley-Liss, Inc.

Vignoli Aglaia, Canevini Maria Paola, Darra Francesca, La Selva Lorita, Fiorini Elena, Piazzini Ada, Lazzarotto Francesca, Zucca Claudio, Dalla Bernardina Bernardo (2009); RING CHROMOSOME 20 SYNDROME: A LINK BETWEEN EPILEPSY ONSET AND NEUROPSYCHOLOGICAL IMPAIRMENT IN THREE CHILDREN; Epilepsia, 50(11):2420-2429

I.F. 2008: 3,733

PURPOSE: Ring chromosome 20 [r(20)] syndrome is a well-defined chromosomal disorder characterized by epilepsy, mild-to-moderate mental retardation, and lack of recognizable dysmorphic features. Epilepsy is often the most important clinical manifestation of the syndrome, even if its appearance is not constantly precocious. Seizures are frequently drug resistant. METHODS: We describe three children with [r(20)] syndrome in whom the onset of epilepsy (age at onset range: 4 years and 6 months to 9 years and 4 months) determined a kind of epileptic status (age at onset range: 6 years and 10 months to 9 years and 8 months) with dramatic neuropsychological deterioration. This epileptic status lasted for several months because of refractoriness to most antiepileptic drugs (AEDs), but it was treated successfully with a combination of valproate and lamotrigine in two children. RESULTS: As soon as seizures stopped, the children showed prompt recovery with partial restoration of the neuropsychological impairment. CONCLUSION: This clinical picture can be described as abrupt epileptic encephalopathy.

Virgilio Roberta, Ronchi Dario, Bordoni Andreina, Fassone Elisa,

Bonato Sara, Donadoni Chiara, Torgano Giuseppe, Moggio Maurizio, Corti Stefania, Bresolin Nereo, Comi Giacomo Pietro (2009); MITOCHONDRIAL DNA G8363A MUTATION IN THE tRNAlys GENE: CLINICAL, BIOCHEMICAL AND PATHOLOGICAL STUDY; Journal of the Neurological Sciences, 281(1-2):85-92

I.F. 2008: 2,359

The G8363A is a very rare mtDNA tRNA(Lys) gene mutation that has been associated to MERRF-like syndrome, cardiomyopathy or Leigh syndrome. Here, we describe the clinical and molecular features of a new large multigenerational family and we review the literature of cases with this mutation. In our family seven members presented a heterogeneous mitochondrial disease phenotype, from MERRF-like syndrome to isolated psychiatric disorder, associated with the G8363A mutation. The two probands are dizygotic twin sisters affected by mental retardation, neural deafness, myopathy, myoclonic epilepsy and ataxia. Twins' muscle biopsies showed a severe cytochrome c oxidase (COX) deficiency and ragged-red fibers. Their mitochondrial respiratory chain was defective in complexes I and IV in muscle. A severe reduction in complex IV activity was also observed in fibroblasts and myoblasts. Molecular analysis showed a G8363A transition in the mtDNA tRNA(Lys) gene. The mutation was almost homoplasmic (>90%) in muscle and blood of the twins and heteroplasmic (55+/-8%) in blood sample from affected maternal relatives. Based on our family data and the meta-analysis of the literature, we confirm that mutational load directly correlates with severity of the disease (severe vs mild/moderate phenotype; P=0.00168) and with disease onset (P<0.00001). However the presence of several exceptions and overlaps among patients with different clinical severity limits the clinical usefulness of this observation. Although the pathogenicity of the G8363A mutation is well established, counselling is a difficult task for clinicians because of the large phenotypical variability. Our study contributes further data on the clinical spectrum and its relation with the level of G8363A tRNA(Lys) mtDNA mutation.

Zuffardi Orsetta, Bonaglia Maria Clara, Ciccone Roberto, Giorda Roberto (2009); INVERTED DUPLICATIONS DELETIONS: UNDERDIAGNOSED REARRANGEMENTS?; Clinical Genetics, 75(6):505-513

I.F. 2008: 3,206

Molecular techniques led to the discovery that several chromosome rearrangements interpreted as terminal duplications were in fact inverted duplications contiguous to terminal deletions. Inv dup del rearrangements

originate through a symmetric dicentric chromosome that, after asymmetric breakage, generates an inv dup del and a deleted chromosome. In recurrent inverted duplications the dicentric chromosome is formed at meiosis through non-allelic homologous recombination. In non-recurrent inv dup del cases, dicentric intermediates are formed by non-homologous end joining or intrastrand annealing. Some authors hypothesized that in these cases the dicentric may have been formed directly in the zygote. Healing of the broken dicentric chromosomes can occur not only in a telomerase-dependent way but also through telomere capture and circularization thus creating translocated or ring inv dup del chromosomes. In all the cases reported up to now, the duplicated region was always longer than the deleted one, but we can safely assume that there is another group of rearrangements where the deleted region is longer than the duplicated portion. In general, in these cases, the cytogeneticist will suspect the presence of a deletion and confirm it by FISH with a subtelomeric probe, but he/she will almost certainly miss the duplication. It is likely that the conventional analysis techniques used until now have led to a substantial underestimate of the frequency of inv dup del rearrangements and that the widespread use of array-CGH in routine analysis will allow a more realistic estimate. Obviously, the concomitant presence of deletion and duplication has important consequences in genotype/phenotype correlations.

Zuliani Riccardo, Moorhead T. William J., Job Dominic, McKirdy James, Sussmann Jessika E.D., Johnstone Eve C., Lawrie Stephen M., Brambilla Paolo, Hall Jeremy, McIntosh Andrew M. (2009); GENETIC VARIATION IN THE G72 (DAOA) GENE AFFECTS TEMPORAL LOBE AND AMYGDALA STRUCTURE IN SUBJECTS AFFECTED BY BIPOLAR DISORDER; Bipolar Disorders, 11(6):621-627

I.F. 2008: 3,959

BACKGROUND: Variation in the G72 (DAOA) gene is understood to convey susceptibility for bipolar disorder through an uncertain mechanism. Little is known about the structural brain phenotypes associated with this gene. We hypothesised that reductions in temporal lobe and amygdala gray matter would be associated with variation at two loci in the gene for which evidence of genetic linkage has been repeatedly demonstrated. METHODS: We examined the temporal lobe and amygdala gray matter associations of the risk variants M23 and M24 at the 5' end of the gene encoding G72 in 81 controls and 38 people with bipolar disorder. RESULTS: Genetic variation at both the M23 and M24 loci in G72 were associated with decreased gray matter density within the left temporal pole in people with bipolar disorder. M23 was also associated with reductions in right amygdala gray matter density. The genetic imaging associations were found only in patients with bipolar disorder. CONCLUSIONS: Genetic variation at single nucleotide polymorphisms in the G72 gene previously associated with bipolar disorder is related to reductions in temporal pole and amygdala gray matter structure in people with bipolar disorder.

LAVORI PER ESTESO PUBBLICATI SU RIVISTE RECENSITE - ANNO 2009

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LAVORI PER ESTESO PUBBLICATI SU RIVISTE RECENSITE Anno 2010

Airoldi Giovanni, Guidarelli Andrea, Cantoni Orazio, Panzeri Chris, Vantaggiato Chiara, Bonato Sara, D'Angelo Maria Grazia, Falcone Sestina, De Palma Clara, Tonelli Alessandra, Crimella Claudia, Bondioni Sara, Bresolin Nereo, Clementi Emilio, Bassi Maria Teresa (2010); CHARACTERIZATION OF TWO NOVEL SETX MUTATIONS IN AOA2 PATIENTS REVEALS ASPECTS OF THE PATHOPHYSIOLOGICAL ROLE OF SENATAXIN; Neurogenetics, 11(1):91-100

I.F. 2009: 3,486

Ataxia with oculomotor apraxia (AOA) type 2 (AOA2 MIM 606002) is a recessive subtype of AOA characterized by cerebellar atrophy, oculomotor apraxia, early loss of reflexes, and peripheral neuropathy. Various mutations either in homozygous or compound heterozygous condition were so far identified in the associated gene SETX (MIM 608465). SETX encodes a large protein called senataxin with a DNA-RNA helicase domain and a putative N-terminus protein interaction domain. Here, we report the identification of two novel homozygous mutations in SETX gene, c.340_342delCTT (p.L114Del) and c.1669C > T (p.R557X), in two AOA2 families. The characterization of the mutant lymphoblastoid cell lines for sensitivity to oxidative DNA-damaging agents indicates that the p.L114Del deletion confers an increased sensitivity to H2O2, camptothecin, and mitomycin C, previously found to induce death in lymphoblasts harbouring other SETX mutations; the cells carrying the nonsense mutation display instead values within the normal range. Further analysis of a neuronal cell model SKNBE, transfected with the mutant senataxin proteins, reveals increased sensitivity also to staurosporine and excitotoxicity associated with the p.L114Del mutant only. We also demonstrate that the sensitizing effect of p.L114Del on apoptosis can be reversed by senataxin silencing. The ability of a single amino acid deletion to sensitize cells to death by different agents, compared to the lack of effect of a whole protein deletion, seems to exclude a protective role played by the native protein while suggesting that a specific mutation confers to the protein the ability to enhance the toxic effect of various cell damaging agents.

Ascenzi Paolo, Di Masi Alessandra, Sciorati Clara, Clementi Emilio

(2010); PEROXYNITRITE - AN UGLY BIOFACTOR?; Biofactors, 36(4):264-273

I.F. 2009: 0,912

Cellular damage occurring under oxidative conditions has been ascribed mainly to the formation of peroxynitrite (ONOOH/ONOO(-)) that originates from the reaction of NO(*) with O(2) (*-). The detrimental effects of peroxynitrite are exacerbated by the reaction with CO(2) that leads to ONOOC(O)O(-), which further decays to the strong oxidant radicals NO(2) (*) and CO(3) (*-). The reaction with CO(2), however, may redirect peroxynitrite specificity. An excessive formation of peroxynitrite represents an important mechanism contributing to the DNA damage, the inactivation of metabolic enzymes, ionic pumps, and structural proteins, and the disruption of cell membranes. Because of its ability to oxidize biomolecules, peroxynitrite is implicated in an increasing list of diseases, including neurodegenerative and cardiovascular disorders, inflammation, pain, autoimmunity, cancer, and aging. However, peroxynitrite displays also protective activities: (i) at high concentrations, it shows anti-viral, anti-microbial, and anti-parasitic actions; and (ii) at low concentrations, it stimulates protective mechanisms in the cardiovascular, nervous, and respiratory systems. The detrimental effects of peroxynitrite and related reactive species are impaired by (pseudo-) enzymatic systems, mainly represented by heme-proteins (e.g., hemoglobin and myoglobin). Here, we report biochemical aspects of peroxynitrite actions being at the root of its biomedical effects.

Battaglia Marco, Zanoni Annalisa, Ogliari Anna, Crevani Federica, Falzone Lidia, Bertoletti Eleonora, Di Serio Clelia (2010); IDENTIFICATION OF GRADUALLY CHANGING EMOTIONAL EXPRESSIONS IN SCHOOLCHILDREN: THE INFLUENCE OF THE TYPE OF STIMULI AND OF SPECIFIC SYMPTOMS OF ANXIETY; Cognition & Emotion, 24(6):1070-1079

I.F. 2009: 1,901

We investigated in two hundred twenty-eight schoolchildren aged 8-11 years whether: (a) the ease of identification of gradually changing emotional expressions varies across different types of expressions; (b) “accurate” and “inaccurate” identifications imply different compromises between speed and accuracy; (c) different forms of anxiety (generalised, separation, somatic/panic, social) affect expressions’ identification. In all ten trials, presented in the form of a videogame, a neutral face gradually morphed through fourteen steps into one of the five basic facial expressions of joy, anger, fear, disgust and surprise. Analysis of variance showed that the number of mistakes in

categorising the anger expression was significantly higher than for any other expression. Survival analyses showed that “anger” was associated with delayed identification, and that accurate and fast performances coincided. Cox’s survival function showed that social anxiety was the only anxiety dimension to predict delayed identification of anger. Social anxiety/phobia predicts biased decoding of signs of interpersonal hostility/rejection.

Bellani Marcella, Marzi Carlo Alberto, Savazzi Silvia, Perlini Cinzia, Cerruti Stefania, Ferro Adele, Marinelli Veronica, Sponda Silvia, Rambaldelli Gianluca, Tansella Michele, Brambilla Paolo (2010); LATERALITY EFFECTS IN SCHIZOPHRENIA AND BIPOLAR DISORDER; Experimental Brain Research, 201(2):339-344

I.F. 2009: 2,256

There are numerous reports in the literature of lateralised structural cerebral abnormalities and alterations of the corpus callosum in the major psychoses. In the light of these findings the purpose of this study was to directly compare hemispheric differences and callosal interhemispheric transmission (IT) in schizophrenia and bipolar disorder. To do that we tested schizophrenic (SCZ), bipolar disorder (BD) patients and controls in a simple manual reaction time (RT) task with lateralised visual stimuli (Poffenberger paradigm) which enables one to test both laterality effects and IT time. We found an overall slowing of responses with the right hand in schizophrenics but not in bipolar patients, who, like controls, showed no hand differences. This selective slowing down of the right hand is likely to be related to abnormalities of intrahemispheric cortico-cortical connections in the left hemisphere. In contrast, IT time was similar in SCZ and BD patients and did not differ with respect to controls. Two are the novel findings of the present study: first both SZC and BD share a normal IT of visuomotor information despite the presence of callosal abnormalities. Second, an impairment of intrahemispheric left hemispheric processing is present only in SCZ patients. This represents a potentially important clue to a further understanding of the pathogenetic differences between the two major psychoses.

Bellani Marcella, Cerruti Stefania, Brambilla Paolo (2010); ORBITOFRONTAL CORTEX ABNORMALITIES IN SCHIZOPHRENIA; Epidemiologia e Psichiatria Sociale, 19(1):23-25

I.F. 2009: 1,860

The magnetic resonance imaging studies investigating the volumes of the orbitofrontal cortex in patients suffering from schizophrenia are here presented, trying to elucidate its role for the pathophysiology and for the cognition of the

disease.

Bellani Marcella, Dusi Nicola, Brambilla Paolo (2010); LONGITUDINAL IMAGING STUDIES IN SCHIZOPHRENIA: THE RELATIONSHIP BETWEEN BRAIN MORPHOLOGY AND OUTCOME MEASURES; Epidemiologia e Psichiatria Sociale, 19(3):207-210

I.F. 2009: 1,860

Imaging studies have tried to identify morphological outcome measures of schizophrenia in the last two decades. In particular, longitudinal studies have reported a correlation between larger ventricles, decreased prefrontal volumes and worse outcome.
This would potentially allow to isolate subtypes of schizophrenia patients with a worse prognosis and more evident biological impairments, ultimately helping in designing specific rehabilitation interventions.

Bellani Marcella, Ferro Adele, Brambilla Paolo (2010); THE POTENTIAL ROLE OF THE PARIETAL LOBE IN SCHIZOPHRENIA; Epidemiologia e Psichiatria Sociale, 19(2):118-119

I.F. 2009: 1,860

Although the anatomy of the parietal lobe has been under-investigated in schizophrenia, some magnetic resonance imaging studies have shown decreased volumes, suggesting its possible implication for the pathophysiology of the disease.

Bersano Anna, Ballabio Elena, Lanfranconi Silvia, Boncoraglio Giorgio B., Corti Stefania, Locatelli Federica, Baron Pierluigi, Bresolin Nereo, Parati Eugenio A., Candelise Livia (2010); CLINICAL STUDIES IN STEM CELLS TRANSPLANTATION FOR STROKE: A REVIEW; Current Vascular Pharmacology, 8(1):29-34

I.F. 2009: 2,970

Stroke is a significant cause of long-term disability. Currently, once damage from a stroke is established little can be done to recover lost function. Cell transplantation emerged as possible alternative therapy, on the basis of animal studies showing that cells transplanted into the brain not only survive, but also lead to functional improvement in different neurodegenerative diseases. Stem cells have been tested in stroke patients as a possible treatment option. While initially stem cells seemed to work by a 'cell replacement' mechanism, it is emerging that cell therapy works mostly by providing trophic support to the injured tissue and brain, fostering both neurogenesis and angiogenesis.

This review summarizes clinical studies on stem cell transplantation in stroke patients to evaluate the safety, feasibility of administration and tolerability of this experimental treatment. At present there is little evidence to assess the applicability of this treatment in stroke patients and well designed clinical trials are necessary to evaluate safety and toxicity as well as optimal cell type, route and time of delivery.

Boaretto Francesca, Vettori A., Casarin A., Vazza Giovanni, Muglia M., Rossetto Maria Giovanna, Cavallaro T., Rizzuto N., Carelli Valerio, Salviati L., Mostacciuolo Maria Luisa, Martinuzzi Andrea (2010); SEVERE CMT TYPE 2 WITH FATAL ENCEPHALOPATHY ASSOCIATED WITH A NOVEL MFN2 SPLICING MUTATION; Neurology, 74(23):1919-1921

I.F. 2009: 8,172

Mutations in the MFN2 gene, encoding mitofusin2, cause autosomal dominant axonal Charcot-Marie-Tooth type 2 (CMT2A, MIM: 608507).1 MFN2 mutations are also found in CMT2 subjects with optic atrophy2 or cognitive impairment.3 The sibship we studied comprised 3 affected and 3 apparently healthy individuals (figure e-1 on the Neurology® Web site at www.neurology.org).

Bonaglia Maria Clara, Marelli Susan, Novara Francesca, Commodaro Simona, Borgatti Renato, Minardo Grazia, Meno Luigi, Mangold Elisabeth, Beri Silvana, Zucca Claudio, Brambilla Daniele, Molteni Massimo, Giorda Roberto, Weber Ruthild G., Zuffardi Orsetta (2010); GENOTYPE-PHENOTYPE RELATIONSHIP IN THREE CASES WITH OVERLAPPING 19p13.12 MICRODELETIONS; European Journal of Human Genetics, in press

I.F. 2009: 3,925

We describe the detailed clinical and molecular characterization of three patients (aged 7, 8(4/12) and 31 years) with overlapping microdeletions in 19p13.12, extending to 19p13.13 in two cases. The patients share the following clinical features with a recently reported 10-year-old girl with a 19p13.12 microdeletion: mental retardation (MR), psychomotor and language delay, hearing impairment, brachycephaly, anteverted nares and ear malformations. All patients share a 359-kb deleted region in 19p13.12 harboring six genes (LPHN1, DDX39, CD97, PKN1, PTGER1 and GIPC1), several of which may be MR candidates because of their function and expression pattern. LPHN1 and PKN1 are the most appealing; LPHN1 for its interaction with Shank family proteins, and PKN1 because it is involved in a variety of functions in neurons,

including cytoskeletal organization. Haploinsufficiency of GIPC1 may contribute to hearing impairment for its interaction with myosin VI. A behavioral phenotype was observed in all three patients; it was characterized by overactive disorder associated with MR and stereotyped movements (ICD10) in one patient and hyperactivity in the other two. As Ptger1-null mice show behavioral inhibition and impulsive aggression with defective social interaction, PTGER1 haploinsufficiency may be responsible for the behavioral traits observed in these patients.European Journal of Human Genetics advance online publication, 21 July 2010; doi:10.1038/ejhg.2010.115.

Brambilla Paolo, Bellani Marcella (2010); LIMITED EVIDENCE THAT ANTIPSYCHOTIC DRUG TREATMENT IS ASSOCIATED WITH REDUCED BRAIN VOLUME; Evid Based Mental Health, 13(2):64 – Commentary

I.F. 2009: 0,000

Abstract non disponibile

Brighina Laura, Prigione Alessandro, Begni Barbara, Galbussera Alessio, Andreoni Simona, Piolti Roberto, Ferrarese Carlo (2010); LYMPHOMONOCYTE ALPHA-SYNUCLEIN LEVELS IN AGING AND IN PARKINSON DISEASE; Neurobiology of Aging, 31(5):884-885 – Negative Results

I.F. 2009: 5,937

In this study we employed an ELISA assay to measure alpha-synuclein protein in lymphomonocytes from 78 PD patients and 78 controls. We correlated protein levels with demographic and clinical characteristics and with the chymotryptic and tryptic activities of the 20S proteasome. Alpha-synuclein levels were not significantly different between patients and controls. In control subjects, alpha-synuclein protein levels increased significantly with age and were significantly higher in men compared to women. Proteasome activity was not significantly different between cases and controls. In control group, the 20S chymotryptic activity tended to decrease significantly with increasing age, though it was not correlated to alpha-synuclein levels. The 20S tryptic activity was not significantly correlated to age, but was inversely correlated to alpha-synuclein levels. Our findings suggest that alpha-synuclein levels in lymphomonocytes are affected by age, gender, and by the 20S proteasome activity in control subjects, but they are not useful as a diagnostic biomarker for PD.

Brunetti-Pierri Nicola, Paciorkowski Alex R., Ciccone Roberto, Della

Mina Erika, Bonaglia Maria Clara, Borgatti Renato, Schaaf Christian P., Sutton V. Reid, Xia Zhilian, Jelluma Naftha, Ruivenkkamp Claudia, Bertrand Mary, De Ravel Thomy JL, Jayakar Parul, Belli Serena, Rocchetti Katia, Pantaleoni Chiara, D'Arrigo Stefano, Hughes Jeff, Cheung Sau Wai, Zuffardi Orsetta, Stankiewicz Pawel (2010); DUPLICATIONS OF FOXG1 IN 14Q12 ARE ASSOCIATED WITH DEVELOPMENTAL EPILEPSY, MENTAL RETARDATION, AND SEVERE SPEECH IMPAIRMENT; European Journal of Human Genetics, in press

I.F. 2009: 3,564

Genome-wide high-resolution array analysis is rapidly becoming a reliable method of diagnostic investigation in individuals with mental retardation and congenital anomalies, leading to the identification of several novel microdeletion and microduplication syndromes. We have identified seven individuals with duplication on chromosome 14q11.2q13.1, who exhibited idiopathic developmental delay and cognitive impairment, severe speech delay, and developmental epilepsy. Among these cases, the minimal common duplicated region on chromosome 14q11.2q13.1 includes only three genes, FOXG1, C14orf23, and PRKD1. We propose that increased dosage of Forkhead Box G1 (FOXG1) is the best candidate to explain the abnormal neurodevelopmental phenotypes observed in our patients. Deletions and inactivating mutations of FOXG1 have been associated with a Rett-like syndrome characterized by hypotonia, irritability, developmental delay, hand stereotypies, and deceleration of head growth. FOXG1, encoding a brain-specific transcription factor, has an important role in the developing brain. In fact, in vivo studies in chicken brain demonstrated that overexpression of FOXG1 results in thickening of the neuroepithelium and outgrowth of the telencephalon and mesencephalum, secondary to a reduction in neuroepithelial cell apoptosis. European Journal of Human Genetics advance online publication, 25 August 2010; doi:10.1038/ejhg.2010.142.

Cagliani Rachele, Fumagalli Matteo, Riva Stefania, Pozzoli Uberto, Comi Giacomo Pietro, Bresolin Nereo, Sironi Manuela (2010); GENETIC VARIABILITY IN THE ACE GENE REGION SURROUNDING THE ALU I/D POLYMORPHISM IS MAINTAINED BY BALANCING SELECTION IN HUMAN POPULATIONS; Pharmacogenetics and Genomics, 20(2):131-134 – Rapid Communication

I.F. 2009: 3,991

OBJECTIVE: Angiotensin-converting enzyme plays a critical role in the maintenance of cardiovascular homeostasis. Extensive research has aimed at

identifying ACE genetic variants responsible for variation in enzyme plasma concentrations and associated with human diseases. These efforts have been hampered by the extensive linkage disequilibrium across the gene and the identity or location of the functional polymorphism(s) is at presently unknown. The aim of our study was to verify whether the Alu insertion/deletion (Alu I/D) polymorphism or any linked variant has been maintained by natural selection in human populations. METHODS: We resequenced a gene region surrounding the Alu I/D polymorphism in four human populations; we applied population neutrality tests and performed haplotype analysis for this region. RESULTS: We observed high levels of nucleotide diversity, an excess of intermediate frequency alleles and, at least in African populations, a higher level of within-species diversity compared with interspecific divergence. Analysis of haplotype genealogy indicated the presence of two major clades separated by deep branches with a coalescence time older than 1.5 million years. All these features strongly suggest the action of balancing selection and we verified that the selection signature is restricted to the gene region surrounding the Alu I/D. CONCLUSION: Our data imply the presence of a functional polymorphism in the Alu I/D region and illustrate the contribution of evolutionary models to classic single nucleotide polymorphism-phenotype association approaches by providing information about the localization of candidate functional variants.

Cagliani Rachele, Fumagalli Matteo, Riva Stefania, Pozzoli Uberto, Fracassetti Marco, Bresolin Nereo, Comi Giacomo Pietro, Sironi Manuela (2010); POLYMORPHISMS IN THE CPB2 GENE ARE MAINTAINED BY BALANCING SELECTION AND RESULT IN HAPLOTYPE-PREFERENTIAL SPLICING OF EXON 7; Molecular Biology and Evolution, 27(8):1945-1954

I.F. 2009: 9,872

The CPB2 gene encodes thrombin-activatable fibrinolysis inhibitor (TAFI), a hepatically secreted zymogen acting as a molecular link among coagulation, fibrinolysis and inflammation. Variants in CPB2 have been associated with several human conditions. We resequenced and analyzed the two regions carrying previously known nonsynonimous SNPs (Ala147Thr and Ile325Thr) and variants affecting transcript stability. Our data indicate that while the gene portion extending from exon 9 to the 3'UTR fits a model of neutral evolution, variants in the region encompassing exons 6-7 have been maintained by balancing selection. Indeed, we verified that the region displays high nucleotide diversity, many intermediate frequency variants and an excess of polymorphism compared to interspecific divergence. Consistently, haplotype

analysis indicated the presence of two major haplotype clades separated by deep branches. Transcript analysis revealed that in both HepG2 cells and human liver samples CPB2 exon 7 undergoes haplotype-preferential skipping. Therefore, we indicate that balancing selection has been maintaining functional variants that promote alternative exon 7 splicing. Although transcripts lacking exon 7 represent a minority of total CPB2 products, the effect on antifibrinolytic activity might be much greater as the intrinsic instability of TAFI is a major determinant of its antifibrinolytic potential. These data highlight the contribution of population genetics approaches to the analysis of functional genetic variation and may orient further biochemical and genetics studies on the pathophysiolgic role of CPB2 gene products.

Cagliani Rachele, Fumagalli Matteo, Biasin Mara, Piacentini Luca, Riva Stefania, Pozzoli Uberto, Bonaglia Maria Clara, Bresolin Nereo, Clerici Mario, Sironi Manuela (2010); LONG-TERM BALANCING SELECTION MAINTAINS TRANS-SPECIFIC POLYMORPHISMS IN THE HUMAN TRIM5 GENE; Human Genetics, in press

I.F. 2009: 4,523

The human TRIM5 genes encodes a retroviral restriction factor (TRIM5alpha). Evolutionary analyses of this gene in mammals have revealed a complex and multifaceted scenario, suggesting that TRIM5 has been the target of exceptionally strong selective pressures, possibly exerted by recurrent waves of retroviral infections. TRIM5 displays inter-individual expression variability in humans and high levels of TRIM5 mRNA have been associated with a reduced risk of HIV-1 infection. We resequenced TRIM5 in chimpanzees and identified two polymorphisms in intron 1 that are shared with humans. Analysis of the gene region encompassing the two trans-specific variants in human populations identified exceptional nucleotide diversity levels and an excess of polymorphism compared to fixed divergence. Most tests rejected the null hypothesis of neutral evolution for this region and haplotype analysis revealed the presence of two deeply separated clades. Calculation of the time to the most recent common ancestor (TMRCA) for TRIM5 haplotypes yielded estimates ranging between 4 and 7 million years. Overall, these data indicate that long-term balancing selection, an extremely rare process outside MHC genes, has maintained trans-specific polymorphisms in the first intron of TRIM5. Bioinformatic analyses indicated that variants in intron 1 may affect transcription factor-binding sites and, therefore, TRIM5 transcriptional activity. Data herein confirm an extremely complex evolutionary history of TRIM5 genes in primates and open the possibility that regulatory variants in the gene modulate the susceptibility to HIV-1.

Cagliani Rachele, Riva Stefania, Biasin Mara, Fumagalli Matteo, Pozzoli Uberto, Lo Caputo Sergio, Mazzotta Francesco, Piacentini Luca, Bresolin Nereo, Clerici Mario*, Sironi Manuela* (2010); GENETIC DIVERSITY AT ENDOPLASMIC RETICULUM AMINOPEPTIDASES IS MAINTAINED BY BALANCING SELECTION AND IS ASSOCIATED WITH NATURAL RESISTANCE TO HIV-1 INFECTION; Human Molecular Genetics, in press

I.F. 2009: 7,386

**Autori che hanno contribuito in ugual misura al lavoro*

Human ERAP1 and ERAP2 encode two endoplasmic reticulum aminopeptidases. These enzymes trim peptides to optimal size for loading onto MHC class I molecules and shape the antigenic repertoire presented to CD8(+) T cells. Therefore, ERAP1 and ERAP2 may be considered as potential selection targets and modulators of infection susceptibility. We resequenced two genic regions in ERAP1 and ERAP2 in three HapMap populations. In both cases we observed high levels of nucleotide variation, an excess of intermediate-frequency alleles, and reduced population genetic differentiation. The genealogy of ERAP1 and ERAP2 haplotypes was split into two major branches with deep coalescence times. These features suggest that long-standing balancing selection has acted on these genes. Analysis of the Lys528Arg (rs30187 in ERAP1) and Asn392Lys (rs2549782 in ERAP2) variants in an Italian population of HIV-1 exposed seronegative (ESN) individuals and a larger number of Italian controls indicated that rs2549782 significantly deviates from Hardy-Weinberg equilibrium (HWE) in ESN but not in controls. Technical errors were excluded and a goodness-of-fit test indicated that a recessive model with only genetic effects adequately explains HWE deviation. The genotype distribution of rs2549782 is significantly different in the two cohorts (p= 0.004), mainly as the result of an over-representation of Lys/Lys genotypes in the ESN sample (p for a recessive model= 0.00097). Our data suggest that genetic diversity in ERAP1 and ERAP2 has been maintained by balancing selection and that variants in ERAP2 confer resistance to HIV-1 infection possibly via the presentation of a distinctive peptide repertoire to CD8(+) T cells.

Calvo-Merino Beatriz, Urgesi Cosimo, Orgs G., Aglioti Salvatore, Haggard Patrick (2010); EXTRASTRIATE BODY AREA UNDERLIES AESTHETIC EVALUTATION OF BODY STIMULI; Experimental Brain Research, 204(3):447-456

I.F. 2009: 2,256

Humans appear to be the only animals to have developed the practice and culture of art. This practice presumably relies on special processing circuits within the human brain associated with a distinct subjective experience, termed aesthetic experience, and preferentially evoked by artistic stimuli. We assume that positive or negative aesthetic judgments are an important function of neuroaesthetic circuits. The localisation of these circuits in the brain remains unclear, though neuroimaging studies have suggested several possible neural correlates of aesthetic preference. We applied repetitive transcranial magnetic stimulation (rTMS) over candidate brain areas to disrupt aesthetic processing while healthy volunteers made aesthetic preference judgments between pairs of dance postures, or control non-body stimuli. Based on evidence from visual body perception studies, we targeted the ventral premotor cortex (vPMC) and extrastriate body area (EBA), in the left and right hemispheres. rTMS over EBA reduced aesthetic sensitivity for body stimuli relative to rTMS over vPMC, while no such difference was found for non-body stimuli. We interpret our results within the framework of dual routes for visual body processing. rTMS over either EBA or vPMC reduced the contributions of the stimulated area to body processing, leaving processing more reliant on the unaffected route. Disruption of EBA reduces the local processing of the stimuli and reduced observers' aesthetic sensitivity. Conversely, disruption of the global route via vPMC increased the relative contribution of the local route via EBA and thus increased aesthetic sensitivity. In this way, we suggest a complementary contribution of both local and global routes to aesthetic processing.

Cantiani Chiara, Lorusso Maria Luisa, Valnegri Camilla, Molteni Massimo (2010); PERCEPTION OF NON-VERBAL AUDITORY STIMULI IN ITALIAN DYSLEXIC CHILDREN; Developmental Neuropsychology, 35(1):115-123

I.F. 2009: 2,321

Auditory temporal processing deficits have been proposed as the underlying cause of phonological difficulties in Developmental Dyslexia. The hypothesis was tested in a sample of 20 Italian dyslexic children aged 8-14, and 20 matched control children. Three tasks of auditory processing of non-verbal stimuli, involving discrimination and reproduction of sequences of rapidly presented short sounds were expressly created. Dyslexic subjects performed more poorly than control children, suggesting the presence of a deficit only partially influenced by the duration of the stimuli and of inter-stimulus intervals (ISIs).

Cattaneo Dario, Baldelli Sara, Conti Francesca, Cozzi Valeria, Clementi Emilio (2010); DETERMINATION OF LINEZOLID IN HUMAN PLASMA BY HIGH-PERFORMANCE LIQUID CHROMATOGRAPHY WITH ULTRAVIOLET DETECTION; Therapeutic Drug Monitoring, Short Communication, 32(4):520-524

I.F. 2009: 2,429

A high-performance liquid chromatographic method for the determination of linezolid in human plasma was developed and validated. After precipitation of plasma proteins with perchloric acid, the protein-free supernatant was separated by isocratic reverse-phase chromatography on a X Bridge C18 column. The mobile phase consisted of a mixture of phosphoric acid 0.05%: acetonitrile (75:25, v/v) with a flow rate of 1 mL/min. The column elute was monitored at 254 nm. The method was linear from 0.2 to 48 mg/L (mean r = 0.9996, n = 10). The observed intra- and inter-day assay imprecision ranged from 2.83% to 8.16% (18.80% at the lower limit of quantification); inaccuracy varied between -0.33% and 8.18%. Mean drug recovery was 99.8% for linezolid and 90.0% for the internal standard (para-toluic acid). The method was found to be precise and accurate and suitable for therapeutic drug monitoring of linezolid in routine clinical practice.

Chiesa Alberto, Brambilla Paolo, Serretti Alessandro (2010); FUNCTIONAL NEURAL CORRELATES OF MINDFULNESS MEDITATIONS IN COMPARISON WITH PSYCHOTHERAPY, PHARMACOTERAPY AND PLACEBO EFFECT. IS THERE A LINK?; Acta Neuropsychiatrica, 22(3):104-117

I.F. 2009: 0,944

Objective: Mindfulness meditations (MM) are a group of meditation practices which are increasingly receiving attention. The aim of the present work is to review current findings about the neural correlates of MM and compare such findings with other specific and non-specific treatments.
Methods: A literature search was undertaken using MEDLINE, ISI web of knowledge, the Cochrane database and references of retrieved articles.
Studies which focused on the functional neural correlates of MM, psychotherapy, pharmacotherapy and placebo published up to August 2009 were screened in order to be considered for the inclusion.
Results: Main findings suggest that long-term MM practice allows a more flexible emotional regulation by engaging frontal cortical structures to dampen automatic amygdala activation. A large overlap exists between cerebral areas activated during MM, psychotherapy, pharmacotherapy and those activated by placebo. However, while MM, psychotherapy and placebo seem

to act through a top-down regulation, antidepressants seem to act through a bottom-up process.
Conclusion: MM seem to target specific brain areas related to emotions and emotional regulation. Similar mechanisms have been observed also in other interventions, particularly psychotherapy.

Cimolin Veronica, Piccinini Luigi, Turconi Anna Carla, Crivellini Marcello, Galli Manuela (2010); ARE KNEE KINEMATIC ANOMALIES IN SWING DUE TO RECTUS FEMORIS SPASTICITY DIFFERENT FROM THOSE DUE TO FEMORAL ANTEVERSION IN CHILDREN WITH CEREBRAL PALSY? A QUANTITATIVE EVALUATION USING 3D GAIT ANALYSIS; Journal of Pediatric Orthopaedics - Part B, 19(3):221-225

I.F. 2009: 0,660

Quantitative comparison of gait strategy between stiff knee gait caused by rectus femoris spasticity versus that caused by femoral anteversion was the objective of this study. Twenty-three diplegic were divided into group 1 (excessive femoral anteversion without rectus femoris spasticity) and group 2 (normal femoral anteversion and rectus femoris spasticity). Both groups showed low knee flexion during swing (KMSw), but although group 1 exhibited normal KMSw timing and high hip intrarotation, group 2 presented delayed KMSw timing, with normal hip rotation. Reduced KMSw may be because of two different conditions: excessive femoral anteversion, leading only to KMSw reduction, and rectus femoris spasticity, inducing coexistence of reduced KMSw and its delayed timing.

Conti Elisa, Galimberti Gloria, Piazza Fabrizio, Raggi Maria Elisabetta, Ferrarese Carlo (2010); INCREASED SOLUBLE APPalpha, ABETA 1-42 AND ANTI ABETA 1-42 ANTIBODIES IN PLASMA FROM DOWN SYNDROME PATIENTS; Alzheimer Disease and Associated Disorders, 24(1):96-100

I.F. 2009: 2,875

Down syndrome (DS) is the most common genetic disorder characterized by an extra copy of chromosome 21. DS subjects show signs of progressive cognitive decline, and most of them develop Alzheimer's type dementia at the age of 50 to 55 years. The aim of this study was to evaluate amyloid precursor protein (APP) metabolites and anti-Abeta 1-42 antibodies plasma levels in DS as possible biomarkers of Abeta accumulation potentially leading to neurodegeneration. We investigated plasma levels of sAPPalpha, Abeta 1-42, and anti-Abeta 1-42 antibodies by enzyme-linked immunosorbent as-

say in 24 DS subjects, 10 non-DS mentally retarded subjects and 18 age-matched controls. We found that sAPPalpha levels were about 1.5-fold higher and Abeta 1-42 levels were about 6-fold higher in DS respect to mentally retarded patients and to controls. DS patients showed Abeta 1-42 antibodies levels 4-fold higher than non-DS mentally retarded group and 2-fold higher than controls. Moreover, anti-Abeta 1-42 antibodies levels were inversely correlated with age in DS subjects. Our results suggested sAPPalpha as a possible peripheral marker for the alteration in APP metabolism in DS and highlighted an alteration in anti-abeta antibodies, for the first time evaluated in plasma from DS subjects.

Corti Stefania, Nizzardo Monica, Nardini Martina, Donadoni Chiara, Salani Sabrina, Ronchi Dario, Simone Chiara, Falcone Marianna, Papadimitriou Dimitra, Locatelli Federica, Mezzina Nicoletta, Gianni Francesca, Bresolin Nereo, Comi Giacomo Pietro (2010); EMBRYONIC STEM CELL-DERIVED NEURAL STEM CELLS IMPROVE SPINAL MUSCULAR ATROPHY PHENOTYPE IN MICE; Brain, 133(Pt2):465-481

I.F. 2009: 9,490

Spinal muscular atrophy, characterized by selective loss of lower motor neurons, is an incurable genetic neurological disease leading to infant mortality. We previously showed that primary neural stem cells derived from spinal cord can ameliorate the spinal muscular atrophy phenotype in mice, but this primary source has limited translational value. Here, we illustrate that pluripotent stem cells from embryonic stem cells show the same potential therapeutic effects as those derived from spinal cord and offer great promise as an unlimited source of neural stem cells for transplantation. We found that embryonic stem cell-derived neural stem cells can differentiate into motor neurons in vitro and in vivo. In addition, following their intrathecal transplantation into spinal muscular atrophy mice, the neural stem cells, like those derived from spinal cord, survived and migrated to appropriate areas, ameliorated behavioural endpoints and lifespan, and exhibited neuroprotective capability. Neural stem cells obtained using a drug-selectable embryonic stem cell line yielded the greatest improvements. As with cells originating from primary tissue, the embryonic stem cell-derived neural stem cells integrated appropriately into the parenchyma, expressing neuron- and motor neuron-specific markers. Our results suggest translational potential for the use of pluripotent cells in neural stem cell-mediated therapies and highlight potential safety improvements and benefits of drug selection for neuroepithelial cells.

Corti Stefania, Nizzardo Monica, Nardini Martina, Donadoni Chiara, Salani Sabrina, Simone Chiara, Falcone Marianna, Riboldi Giulietta, Govoni Alessandra, Bresolin Nereo, Comi Giacomo Pietro (2010); SYSTEMIC TRANSPLANTATION OF C-KIT+ CELLS EXERTS A THERAPEUTIC EFFECT IN A MODEL OF AMYOTROPHIC LATERAL SCLEROSIS; Human Molecular Genetics, 19(19):3782-3796

I.F. 2009: 7,386

Amyotrophic lateral sclerosis (ALS) is a progressive, fatal neurodegenerative disease characterized by the loss of motor neurons. Motor neuron degeneration is likely both a cell autonomous and a non-autonomous event. Therefore, manipulating the diseased microenvironment via non-neural cell replacement could be a therapeutic strategy. We investigated a cell therapy approach using intravascular injection to transplant a specific population of c-kit-positive stem/progenitor cells from bone marrow into the SOD1G93A mouse model of ALS. Transplanted cells engrafted within the host spinal cord. Cell transplantation significantly prolonged disease duration and lifespan in SOD1 mice, promoted the survival of motor neurons, and improved neuromuscular function. Neuroprotection was mediated by multiple effects, in particular by the expression of primary astrocyte glutamate transporter GLT1 and by the non-mutant genome. These findings suggest that this type of somatic cell transplantation strategy merits further investigation as a possible effective therapy for ALS and other neurodegenerative diseases.

Crimella Claudia, Tonelli Alessandra, Airoldi Giovanni, Baschirotto Cinzia, D'Angelo Maria Grazia, Bonato Sara, Losito Luciana, Trabacca Antonio, Bresolin Nereo, Bassi Maria Teresa (2010); THE GST DOMAIN OF GDAP1 IS A FREQUENT TARGET OF MUTATIONS IN THE DOMINANT FORM OF AXONAL CHARCHOT MARIE TOOTH TYPE 2K; Journal of Medical Genetics, in press

I.F. 2009: 5,751

Background Mutations in GDAP1 associate with demyelinating (CMT4A) and axonal (CMT2K) forms of CMT. While CMT4A shows recessive inheritance, CMT2K can present with either recessive (AR-CMT2K) or dominant segregation pattern (AD-CMT2K), the latter being characterised by milder phenotypes and later onset. The majority of the GDAP1 mutations are associated with CMT4A and AR-CMT2K, with only four heterozygous mutations identified in AD-CMT2K. Methods We screened GDAP1 gene in a series of 43 index patients, 39 with CMT2 and 4 with intermediate CMT, with sporadic and familial occurrence of the disease. Results Three novel mutations were identified in three families with dominant segregation of the disease: two mis-

sense changes, p.Arg226Ser and p.Ser34Cys, affecting the GST domain of the GDAP1 protein and a novel deletion (c.23delAG) leading to early truncation of the protein upstream the GST domain. Wide variability in clinical presentation is shared by all three families mostly in terms of age at onset and disease severity. A rare variant p.Gly269Arg, located within the GST domain, apparently acts as phenotype modulator in the family carrying the deletion. Conclusion The results obtained reveal a GDAP1 mutation frequency of 27% in the dominant families analysed, a figure still unreported for this gene, thus suggesting that GDAP1 involvement in dominant CMT2 might be higher than expected.

De Palma Clara, Falcone Sestina, Pisoni Serena, Cipolat S., Panzeri Chris, Pambianco S., Pisconti Addolorata, Allevi Raffaele, Bassi Maria Teresa, Cossu Giulio, Pozzan T., Moncada Salvador, Scorrano L., Brunelli S., Clementi E. (2010); NITRIC OXIDE INHIBITION ID DRP1-MEDIATED MITOCHONDRIAL FISSION IS CRITICAL FOR MYOGENIC DIFFERENTIATON; Cell Death and Differentation, in press

I.F. 2009: 8,240

During myogenic differentiation the short mitochondria of myoblasts change into the extensively elongated network observed in myotubes. The functional relevance and the molecular mechanisms driving the formation of this mitochondrial network are unknown. We now show that mitochondrial elongation is required for myogenesis to occur and that this event depends on the cellular generation of nitric oxide (NO). Inhibition of NO synthesis in myogenic precursor cells leads to inhibition of mitochondrial elongation and of myogenic differentiation. This is due to the enhanced activity, translocation and docking of the pro-fission GTPase dynamin-related protein-1 (Drp1) to mitochondria, leading also to a latent mitochondrial dysfunction that increased sensitivity to apoptotic stimuli. These effects of NO inhibition were not observed in myogenic precursor cells containing a dominant-negative form of Drp1. Both NO-dependent repression of Drp1 action and maintenance of mitochondrial integrity and function were mediated through the soluble guanylate cyclase. These data uncover a novel level of regulation of differentiation linking mitochondrial morphology and function to myogenic differentiation. Cell Death and Differentiation advance online publication, 14 May 2010; doi:10.1038/cdd.2010.48.

De Rinaldis Marta, Losito Luciana, Gennaro Leonarda, Trabacca Antonio (2010); LONG-TERM ORAL BACLOFEN TREATMENT IN A

CHILD WITH CEREBRAL PALSY: EEG CHANGES AND CLINICAL ADVERSE EFFECTS; Journal of Child Neurology, in press

I.F. 2009: 1,592

Baclofen is widely used to control spasticity in children with cerebral palsy. Several publications described clinical adverse effects of baclofen oral treatment, but the effect of baclofen on seizure potentiation is still controversial. We describe a 10-year-old female patient with cerebral palsy, epilepsy, and mental retardation who developed clinical adverse effects (confusion, agitated state, insomnia, diffuse hypotonia, and hyporeflexia) and electroencephalographic (EEG) changes (quasiperiodic, generalized burst of sharp waves that take up >50% of standard EEG) during long-term oral baclofen treatment, after gradually increasing the dosage but still within the therapeutic dose. Our case showed clearly that the EEG changes in our patient, with a history of epilepsy in good control, have been induced by the baclofen increase, and we describe the possible mechanisms that could explain proconvulsive effect of baclofen.

Facoetti Andrea, Trussardi Anna Noemi, Ruffino Milena, Lorusso Maria Luisa, Cattaneo Carmen, Galli Raffaella, Molteni Massimo, Zorzi Marco (2010); MULTI-SENSORY SPATIAL ATTENTION DEFICITS ARE PREDICTIVE OF PHONOLOGICAL DECODING SKILLS IN DEVELOPMENTAL DYSLEXIA; Journal of Cognitive Neuroscience, 22(5):1011-1025

I.F. 2009: 5,382

Although the dominant approach posits that developmental dyslexia arises from deficits in systems that are exclusively linguistic in nature (i.e., phonological deficit theory), dyslexics show a variety of lower level deficits in sensory and attentional processing. Although their link to the reading disorder remains contentious, recent empirical and computational studies suggest that spatial attention plays an important role in phonological decoding. The present behavioral study investigated exogenous spatial attention in dyslexic children and matched controls by measuring RTs to visual and auditory stimuli in cued-detection tasks. Dyslexics with poor nonword decoding accuracy showed a slower time course of visual and auditory (multisensory) spatial attention compared with both chronological age and reading level controls as well as compared with dyslexics with slow but accurate nonword decoding. Individual differences in the time course of multisensory spatial attention accounted for 31% of unique variance in the nonword reading performance of the entire dyslexic sample after controlling for age, IQ, and phonological skills. The present study suggests that multisensory “sluggish attention

shifting"-related to a temporoparietal dysfunction-selectively impairs the sublexical mechanisms that are critical for reading development. These findings may offer a new approach for early identification and remediation of developmental dyslexia.

Facoetti Andrea, Corradi Nicola, Ruffino Milena, Gori Simone, Zorzi Marco (2010); VISUAL SPATIAL ATTENTION AND SPEECH SEGMENTATION ARE BOTH IMPAIRED IN PRESCHOOLERS AT FAMILIAL RISK FOR DEVELOPMENTAL DYSLEXIA; Dyslexia, 16(3):226-239

I.F. 2009: 1,176

Phonological skills are foundational of reading acquisition and impaired phonological processing is widely assumed to characterize dyslexic individuals. However, reading by phonological decoding also requires rapid selection of sublexical orthographic units through serial attentional orienting, and recent studies have shown that visual spatial attention is impaired in dyslexic children. Our study investigated these different neurocognitive dysfunctions, before reading acquisition, in a sample of preschoolers including children with (N=20) and without (N=67) familial risk for developmental dyslexia. Children were tested on phonological skills, rapid automatized naming, and visual spatial attention. At-risk children presented deficits in both visual spatial attention and syllabic segmentation at the group level. Moreover, the combination of visual spatial attention and syllabic segmentation scores was more reliable than either single measure for the identification of at-risk children. These findings suggest that both visuo-attentional and perisylvian-auditory dysfunctions might adversely affect reading acquisition, and may offer a new approach for early identification and remediation of developmental dyslexia.

Fagiolari Gigliola, Cappellini Anna, Cagliani Rachele, Prelle Alessandro, Lucchini Valeria, Fortunato Francesco, Locatelli Federica, Crugnola Veronica, Comi Giacomo Pietro, Bresolin Nereo, Moggio Maurizio, Lamperti Costanza (2010); MUSCULAR DYSTROPHY: CENTRAL NERVOUS SYSTEM ALPHA-DYSTROGLYCAN GLYCOSYLATION DEFECTS AND BRAIN MALFORMATION; Journal of Child Neurology, 25(3):312-320

I.F. 2009: 1,592

The authors describe the case of a patient affected with congenital muscular dystrophy with lack of muscle alpha-dystroglycan. Brain gross anatomy showed lissencephaly and pachygyria. Light microscopy showed heterotopias in white matter. The brain stem and cerebellum were normal. They

found no expression of alpha-dystroglycan either in the frontal cortex or in the heterotopic nuclei, while a normal expression was found in the cerebellum. These results suggest that alpha-dystroglycan glycosylation defects may account for both the muscle disease and the brain supratentorial malformation in our patient. The authors did not identify any mutations in the genes most frequently related to these syndromes. Therefore, this case suggests that a new gene may be associated with congenital muscular dystrophy with alpha-dystroglycan glycosylation defects, cortical migration defects, and sparing of the cerebellum.

Fumagalli Matteo, Pozzoli Uberto, Cagliani Rachele, Comi Giacomo Pietro, Bresolin Nereo, Clerici Mario, Sironi Manuela (2010); GENOME-WIDE IDENTIFICATION OF SUSCEPTIBILITY ALLELES FOR VIRAL INFECTIONS THROUGH A POPULATION GENETICS APPROACH; Plos Genetics, 6(2):e1000849

I.F. 2009: 9,532

Viruses have exerted a constant and potent selective pressure on human genes throughout evolution. We utilized the marks left by selection on allele frequency to identify viral infection-associated allelic variants. Virus diversity (the number of different viruses in a geographic region) was used to measure virus-driven selective pressure. Results showed an excess of variants correlated with virus diversity in genes involved in immune response and in the biosynthesis of glycan structures functioning as viral receptors; a significantly higher than expected number of variants was also seen in genes encoding proteins that directly interact with viral components. Genome-wide analyses identified 441 variants significantly associated with virus-diversity; these are more frequently located within gene regions than expected, and they map to 139 human genes. Analysis of functional relationships among genes subjected to virus-driven selective pressure identified a complex network enriched in viral products-interacting proteins. The novel approach to the study of infectious disease epidemiology presented herein may represent an alternative to classic genome-wide association studies and provides a large set of candidate susceptibility variants for viral infections.

Fumagalli Matteo, Cagliani Rachele, Riva Stefania, Pozzoli Uberto, Biasin Mara, Piacentini Luca, Comi Giacomo Pietro, Bresolin Nereo, Clerici Mario, Sironi Manuela (2010); POPULATION GENETICS OF IFIH1: ANCIENT POPULATION STRUCTURE, LOCAL SELECTION AND IMPLICATIONS FOR SUSCEPTIBILITY TO TYPE 1 DIABETES; Molecular Biology and Evolution, in press

I.F. 2009: 9,872

The human IFIH1 gene encodes a sensor of double strand RNA involved in innate immunity against viruses, indicating that this gene is a likely target of virus-driven selective pressure. Notably, IFIH1 also plays a role in autoimmunity as common and rare polymorphisms in this gene have been associated with type 1 diabetes (T1D). We analysed the evolutionary history of IFIH1 in human populations. Results herein suggest that two major IFIH1 haplotype clades originated from ancestral population structure (or balancing selection) in the African continent and that local selective pressures have acted on the gene. Specifically, directional selection in Europe and Asia resulted in the spread of a common IFIH1 haplotype carrying a derived His460 allele. This variant changes a highly conserved arginine residue in the helicase domain, possibly conferring altered specificity in viral recognition. An alternative common haplotype has swept to high frequency in South Americans as a result of recent positive selection. Previous studies suggested that a portion of risk alleles for autoimmune diseases could have been maintained in humans as they conferred a selective advantage against infections. This is not the case for IFIH1 as population genetic differentiation and haplotype analyses indicated that the T1D susceptibility alleles behaved as neutral or nearly neutral polymorphisms. Our findings suggest that variants in IFIH1 confer different susceptibility to diverse viral infections and provide insight into the relationship between adaptation to past infection and predisposition to autoimmunity in modern populations.

Fumagalli Matteo*, Pozzoli Uberto*, Cagliani Rachele, Comi Giacomo Pietro, Bresolin Nereo, Clerici Mario, Sironi Manuela (2010); THE LANDSCAPE OF HUMAN GENES THE IMMUNE RESPONSE TO PARASITIC WORMS; BMC Evolutionary Biology, 10(1):264

I.F. 2009: 4,294

**Autori che hanno contribuito in ugual misura al lavoro*

Background - More than 2 billion individuals worldwide suffer from helminth infections. The highest parasite burdens occur in children and helminth infection during pregnancy is a risk factor for preterm delivery and reduced birth weight. Therefore, helminth infections can be regarded as a strong selective pressure.

Results - Here we propose that candidate susceptibility genes for parasitic worm infections can be identified by searching for SNPs that display a strong correlation with the diversity of helminth species/genera transmitted in different geographic areas. By a genome-wide search we identified 3478 variants

that correlate with helminth diversity. These SNPs map to 810 distinct human genes including loci involved in regulatory T cell function and in macrophage activation, as well as leukocyte integrins and co-inhibitory molecules. Analysis of functional relationships among these genes identified complex interaction networks centred around Th2 cytokines. Finally, several genes carrying candidate targets for helminth-driven selective pressure also harbour susceptibility alleles for asthma/allergy or are involved in airway hyper-responsiveness, therefore expanding the known
parallelism between these conditions and parasitic infections.

Conclusions - Our data provide a landscape of human genes that modulate susceptibility to helminths and indicate parasitic worms as one of the major selective forces in humans.

Gennaro Leonarda, Russo Luigi, Losito Luciana, Zaccaria Alessia, De Rinaldis Marta, Trabacca Antonio (2010); MOVEMENT DISORDERS IN A TWINS PAIR: A CASUAL EXPRESSION OR GENETIC DETERMINATION?; Research in Developmental Disabilities, 31(3):692-697

I.F. 2009: 4,410

A twin study is an excellent means of assessing the contribution of heritability to motor behaviour. We present a movement video-analysis of a monozygotic twins pair with a motor repertoire which is almost totally constituted by persistent and subcontinuous motor stereotypies. PURPOSE: The specific aim of this study is to verify the heritable quantum of motor behaviour and to determine which among the motor patterns we analysed are more likely to be conditioned by inheritance. METHODS: Stereotyped movements were videotaped in two standardized sessions: at rest and in relation to preordained sensory stimulations. We estimated the concordance index (CI) between the observers to evaluate the reliability of the observations. The validity was accepted as being CI>0.80. RESULTS: The results showed a very high concordance rate (>90%) for all the stereotypies analysed. An almost superimposable trend of the stereotyped movements was found both at rest and in relation to the sensory stimulations. CONCLUSIONS: Such strong data suggest that genetic factors have a primary influence on all the movement disorders analysed. This study contributes to a better understanding of the complex relationships between genes and functions.

Iannicelli Miriam, Brancati Francesco, Mougou-Zerelli Soumaya, Mazzotta Annalisa, Thomas Sophie, Elkhartoufi Nadia, Travaglini

Lorena, Gomes Celine, Ardissino Gian Luigi, Bertini Enrico, Boltshauser Eugen, Castorina Pierangela, D'Arrigo Stefano, Fischetto Rita, Leroy Brigitte, Loget Philippe, Bonniere Maryse, Starck Lena, Tantau Julia, Gentilin Barbara, Majore Silvia, Swistun Dominika, Flori Elizabeth, Lalatta Faustina, Pantaleoni Chiara, Penzien Johannes, Grammatico Paola, The International JSRD Study Group (Borgatti Renato, Cazzagon Monica), Dallapiccola Bruno, Gleeson Joseph G., Attie-Bitach Tania, Valente Enza Maria (2010); NOVEL TMEM67 MUTATIONS AND GENOTYPE-PHENOTYPE CORRELATES IN MECKELIN-RELATED CILIOPATHIES; Human Mutation, 31(3):E1319-E1331

I.F. 2009: 6,887

Human ciliopathies are hereditary conditions caused by defects of proteins expressed at the primary cilium. Among ciliopathies, Joubert syndrome and related disorders (JSRD), Meckel syndrome (MKS) and nephronophthisis (NPH) present clinical and genetic overlap, being allelic at several loci. One of the most interesting gene is TMEM67, encoding the transmembrane protein meckelin. We performed mutation analysis of TMEM67 in 341 probands, including 265 JSRD representative of all clinical subgroups and 76 MKS fetuses. We identified 33 distinct mutations, of which 20 were novel, in 8/10 (80%) JS with liver involvement (COACH phenotype) and 12/76 (16%) MKS fetuses. No mutations were found in other JSRD subtypes, confirming the strong association between TMEM67 mutations and liver involvement. Literature review of all published TMEM67 mutated cases was performed to delineate genotype-phenotype correlates. In particular, comparison of the types of mutations and their distribution along the gene in lethal versus non lethal phenotypes showed in MKS patients a significant enrichment of missense mutations falling in TMEM67 exons 8 to 15, especially when in combination with a truncating mutation. These exons encode for a region of unknown function in the extracellular domain of meckelin. ©2010 Wiley-Liss, Inc.

Kalyva Efrosini, Pellizzoni Sandra, Tavano Alessandro, Iannello Paola, Siegal Michael (2010); CONTAMINATION SENSITIVITY IN AUTISM, DOWN SYNDROME, AND TYPICAL DEVELOPMENT; Research in Autism Spectrum Disorders, 4(1):43-50

I.F. 2009: 2,267

Although typically developing children are attuned early to others' communicative signals, one of the very first noticeable impairments in children with autism is in attending to voices and speech.
Yet it is through conversations with others that children are made aware that

apparently edible substances may in reality be contaminated. In two experiments, we examined contamination sensitivity in children with autism, typically developing children, and a group of children with Down syndrome. In Experiment 1, many children with autism who ranged in age from 4 to 10 years
were prepared to drink liquids that had been contaminated by insects. There was evidence for a developmental delay as contamination sensitivity in autism was associated with increasing age. In Experiment 2, children with autism were prepared to drink liquids that had been contaminated by human hair or had insects in close proximity. By contrast, in both experiments, both typically developing children and children with Down syndrome demonstrated strong contamination sensitivity. We discuss the results in terms of constraints on the early learning of the edible–inedible distinction. © 2010 Elsevier Ltd. All rights reserved.

Lanfranconi Silvia, Locatelli Federica, Corti Stefania, Candelise Livia, Comi Giacomo Pietro, Baron Pierluigi, Strazzer Sandra, Bresolin Nereo, Bersano Anna (2010); GROWTH FACTORS IN ISCHEMIC STROKE; Journal of Cellular and Molecular Medicine, in press

I.F. 2009: 5,228

Data from preclinical and clinical studies provide evidence that colony stimulating factors (CSFs) and other growth factors can improve stroke outcome by reducing stroke damage through their anti-apoptotic and anti-inflammatory effects, and by promoting angiogenesis and neurogenesis. This review provides a critical and up-to-date literature review on CSF use in stroke. We searched for experimental and clinical studies on haemopoietic growth factors such as granulocyte colony stimulating factor (G-CSF), erythropoietin (EPO), granulocyte-macrophage colony stimulating factor (GM-CSF), stem cell factor (SCF), vascular endothelial growth factor (VEGF), stromal cell-derived factor-1 alpha (SDF-1alpha), and stem cell factor (SCF) in ischemic stroke. We also considered studies on insulin-like growth factor-1 (IGF-1) and neurotrophins. Despite promising results from animal models, the lack of data in humans hampers efficacy assessments of growth factors on stroke outcome. We provide a comprehensive and critical view of the present knowledge about growth factors and stroke, and an overview of ongoing and future prospects.

Lo Mauro Antonella, D'Angelo Maria Grazia, Romei Marianna, Motta Francesca, Colombo Daniele, Comi Giacomo Pietro, Pedotti

Antonio, Marchi Eraldo, Turconi Anna Carla, Bresolin Nereo, Aliverti Andrea (2010); ABDOMINAL VOLUME CONTRIBUTION TO TIDAL VOLUME AS AN EARLY INDICATOR OF RESPIRATORY IMPAIRMENT IN DUCHENNE MUSCULAR DYSTROPHY; European Respiratory Journal, 35(5):1118-1125

I.F. 2009: 5,527

Duchenne muscular dystrophy (DMD) is characterised by progressive loss of muscular strength that leads to an increasingly restrictive pulmonary syndrome. However, it is still not clear whether this determines alterations in the breathing pattern. We studied: 66 DMD patients at different stages of the disease (mean+/- sem age 12.6+/-0.6 yrs, range 5-22 yrs of age), subdivided into four groups according to age; and 21 age-matched healthy male controls. Spirometry, lung volumes and nocturnal oxygen saturation were measured in all DMD patients. Ventilatory pattern and chest wall volume variations were assessed by optoelectronic plethysmography during spontaneous breathing both in seated and supine positions. Whilst in a seated position, no significant differences were found between patients and controls or between different age groups. In the supine position, the average contribution of abdominal volume change (DeltaV(AB)) to tidal volume progressively decreased with age ($p<0.001$). The patients who showed nocturnal hypoxaemia showed significantly lower Delta V(AB). In conclusion, chest wall motion during spontaneous breathing in awake conditions and in supine position is an important indicator of the degree of respiratory muscle impairment in DMD. DeltaV(AB) is not only an important marker of the progression of the disease but is also an early indicator of nocturnal hypoxaemia.

Marini Andrea, Martelli Sara, Gagliardi Chiara, Fabbro Franco, Borgatti Renato (2010); NARRATIVE LANGUAGE IN WILLIAMS SYNDROME AND ITS NEUROPSYCHOLOGICAL CORRELATES; Journal of Neurolinguistics, 23(2):97-111

I.F. 2009: 1,660

The cognitive profile of individuals with Williams' Syndrome (WS) shows peaks and troughs, with fairly good linguistic performance and a well described weakness in visual-spatial abilities. This study aims to describe in detail the narrative abilities of a group of 9 WS participants who underwent careful cognitive evaluation to assess their visual-spatial abilities, sustained attention, phonological short-term memory and lexical as well as grammatical skills in tests of expressive and receptive language. Furthermore, they performed a picture-description task in order to elicit more fluid and communicative speech samples, which were then compared with those provided

by a group of 29 children with typically developing language (TD) matched for mental age. The WS participants showed visual-spatial deficits but scored within the normal range, according to their mental age, in the linguistic assessment. For the narrative task, they showed good phonological, lexical and syntactic skills, but their story descriptions were less effective than those produced by the TD group on measures assessing global coherence and lexical informativeness, showing dissociation between macro and microlinguistic abilities. These impairments were not correlated to the visual-spatial disturbances.
These data suggest that the domain of discourse processing is a relative weakness in WS individuals.

Martinuzzi Andrea, Salghetti Annamaria, Betto Silvana, Russo Emanuela, Leonardi Matilde, Raggi Alberto, Francescutti Carlo (2010); THE INTERNATIONAL CLASSIFICATION OF FUNCTIONING DISABILITY AND HEALTH, VERSION FOR CHILDREN AND YOUTH AS A ROAD-MAP FOR PROJECTING AND PROGRAMMING REHABILITATION IN A NEUROPAEDIATRIC HOSPITAL UNIT; Journal of Rehabilitation Medicine, 42(1):49-55

I.F. 2009: 1,882

OBJECTIVE: To test the impact of introducing a format based on the International Classification of Functioning Disability and Health, version for children and youth (ICF-CY) as a road-map for in-hospital paediatric neuro-rehabilitation on target definition, intra-team communication and workload as perceived by involved professionals. DESIGN: Single-centre pilot testing with impact assessment. PATIENTS: Team members of a tertiary care paediatric neurorehabilitation unit included 15 consecutive patients with severe neurological conditions. METHODS: An ICF-CY based format for rehabilitation projection and programming was constructed and tested for 12 months. The format comprises 3 sections: project, programme, and follow-up. Impact on the rehabilitation team was assessed with a questionnaire. RESULTS: All cases were described according to their specific needs with appropriate ICF-CY codes, and the interventions were linked to needs and targets. ICF-CY was judged an efficient tool in providing a road-map for rehabilitation in this setting, although concern was voiced about timing and workload. CONCLUSION: ICF-CY may work as a road-map for in-hospital paediatric neuro-rehabilitation. Its implementation results in perceived improvements in the process. Training requirements and accurate evaluation of timing, workload and organizational context are critical issues that should be addressed before results from the present experience are generalized.

Massimino Maura, Spreafico Filippo, Riva Daria, Biassoni Veronica, Poggi Geraldina, Solero Carlo L., Gandola Lorenza, Genitori Lorenzo, Modena Piergiorgio, Simonetti Fabio, Potepan Paolo, Casanova Michela, Meazza Cristina, Clerici Carlo Alfredo, Catania Serena, Sardi Iacopo, Giangaspero Felice (2010); A LOWER-DOSE, LOWER-TOXICITY CISPLATIN-ETOPOSIDE REGIMEN FOR CHIDLHOOD PROGRESSIVE LOW-GRADE GLIOMA; Journal of Neuro-Oncology, in press

I.F. 2009: 2,752

After successfully using cisplatin (30 mg/m(2)/day) and etoposide (150 mg/m(2)/day) in ten three-day courses for progressive low-grade gliomas, a subsequent protocol reduced the daily doses of cisplatin (to 25 mg) and etoposide (to 100 mg), with the objective of achieving the same response and three-year PFS rates with lower neurotoxicity and myelotoxicity. We treated 37 patients (median age 6 years); 23 had optochiasmatic tumours and nine were metastatic cases. Diagnoses were clinical in 13 cases and histological in 24, and comprised: pilocytic astrocytoma (17), ganglioglioma (3), pilomyxoid astrocytoma (2), and fibrillary astrocytoma (2). Treatment was prompted by radiological evidence of progression and/or clinical deterioration a median 18 months after the first diagnosis. After initial MRI staging, neurological and clinical examinations were performed before each chemotherapy cycle, with MRI after the first three courses and every three months thereafter. After a median 48 months, a volume reduction was appreciable in 24 cases (65%) and response was maximum 12 months after starting treatment. The three-year EFS and OS rates were 65 and 97%, respectively. Clinical, neurological, or functional improvements were seen in 26/37 cases. No children had a WBC nadir below 2,000/mm(3). Audiological toxicity caused damage in 4/34 cases. The previous protocol had achieved volume reductions in 70% of cases, causing audiological damage (data updated) in 11/31 (P = 0.023), with three-year PFS and OS rates of 70 and 100%, respectively. Lower doses of cisplatin/etoposide are still effective in progressive low-grade glioma, with less acute and persistent morbidity.

Massimino Maura, Giangaspero Felice, Garrè Maria Luisa, Gandola Lorenza, Poggi Geraldina, Biassoni Veronica, Gatta Gemma, Rukowski Stefan (2010); CHILDHOOD MEDULLOBLASTOMA; Critical Reviews in Oncology/Hematology, in press

I.F. 2009: 5,269

Abstract non disponibile

Maziade Michel, Rouleau Nancie, Merette Chantal, Cellard Caroline, Battaglia Marco, Marino Cecilia, Jomphe Valérie, Gilbert Elsa, Achim Amélie, Bouchard Roch-Hugo, Paccalet Thomas, Paradis Marie-Eve, Roy Marc-André (2010); VERBAL AND VISUAL MEMORY IMPAIRMENTS AMONGST YOUNG OFFSPRING AND HEALTHY ADULT RELATIVES OF PATIENTS WITH SCHIZOPHRENIA AND BIPOLAR-DISORDER: SELECTIVE GENERATIONAL PATTERNS INDICATE DIFFERENT DEVELOPMENTAL TRAJECTORIES; Schizophrenia Bulletin, in press

I.F. 2009: 7,467

Objective: Memory deficits have been shown in patients affected by schizophrenia (SZ) and bipolar (BP)/mood disorder. We recently reported that young high-risk offspring of an affected parent were impaired in both verbal episodic memory (VEM) and visual episodic memory (VisEM). Understanding better the trajectory of memory impairments from childhood to adult clinical status in risk populations is crucial for early detection and prevention. In multigenerational families densely affected by SZ or BP, our aim was to compare the memory impairments observed in young nonaffected offspring with memory functioning in nonaffected adult relatives and patients. Methods: For 20 years, we followed up numerous kindreds in the Eastern Québec population. After having characterized the Diagnostic and Statistical Manual of Mental Disorders phenotypes, we assessed cognition (N = 381) in 3 subsamples in these kindreds and in controls: 60 young offspring of a parent affected by SZ or BP, and in the adult generations, 92 nonaffected adult relatives and 40 patients affected by SZ or BP. VEM was assessed with the California Verbal Learning Test and VisEM with the Rey figures. Results: The VEM deficits observed in the offspring were also found in adult relatives and patients. In contrast, the VisEM impairments observed in the young offspring were present only in patients, not in the adult relatives. Conclusion: Implications for prevention and genetic mechanisms can be drawn from the observation that VEM and VisEM would show distinct generational trajectories and that the trajectory associated with VisEM may offer a better potential than VEM to predict future risk of developing the disease.

Mazzucchelli Serena, Colombo Miriam, De Palma Clara, Salvadè Agnese, Verderio Paolo, Coghi Maria D., Clementi Emilio, Tortora Paolo, Corsi Fabio, Prosperi Davide (2010); SINGLE-DOMAIN PROTEIN A-ENGINEERED MAGNETIC NANOPARTICLES: TOWARD A UNIVERSAL STRATEGY TO SITE-SPECIFIC LABELING OF

ANTIBODIES FOR TARGETED DETECTION OF TUMOR CELLS; ACS Nano, in press

I.F. 2009: 7,493

Highly monodisperse magnetite nanocrystals (MNC) were synthesized in organic media and transferred to the water phase by ultrasound-assisted ligand exchange with an iminodiacetic phosphonate. The resulting biocompatible magnetic nanoparticles were characterized by transmission electron microscopy, dynamic light scattering, and magnetorelaxometry, indicating that this method allowed us to obtain stable particle dispersions with narrow size distribution and unusually high magnetic resonance T(2) contrast power. These nanoparticles were conjugated to a newly designed recombinant monodomain protein A variant, which exhibited a convincingly strong affinity for human and rabbit IgG molecules. Owing to the nature of antibody-protein A binding, tight antibody immobilization occurred through the Fc fragment thus taking full advantage of the targeting potential of bound IgGs. If necessary, monoclonal antibodies could be removed under controlled conditions regenerating the original IgG-conjugatable MNC. As a proof of concept of the utility of our paramagnetic labeling system of human IgGs for biomedical applications, anti-HER-2 monoclonal antibody trastuzumab was immobilized on hybrid MNC (TMNC). TMNC were assessed by immunoprecipitation assay and confocal microscopy effected on HER-2-overexpressing MCF-7 breast cancer cells, demonstrating excellent recognition capability and selectivity for the target membrane receptor.

Menghini D., Finzi A., Benassi M., Bolzani R., Facoetti Andrea, Giovagnoli S., Ruffino Milena, Vicari Stefano (2010); DIFFERENT UNDERLYING NEUROCOGNITIVE DEFICITS IN DEVELOPMENTAL DYSLEXIA: A COMPARATIVE STUDY; Neuropsychologia, 48(4):863-872

I.F. 2009: 4,345

The aim of this study was to investigate the role of several specific neurocognitive functions in developmental dyslexia (DD). The performances of 60 dyslexic children and 65 age-matched normally reading children were compared on tests of phonological abilities, visual processing, selective and sustained attention, implicit learning, and executive functions. Results documented deficits in dyslexics on both phonological and non-phonological tasks. More stringently, in dyslexic children individual differences in non-phonological abilities accounted for 23.3% of unique variance in word reading and for 19.3% in non-word reading after controlling for age, IQ and phonological skills. These findings are in accordance with the hypothesis that DD is a multifactorial deficit and suggest that neurocognitive developmental dysfunctions in DD may not be limited to the linguistic brain area, but may involve a more multifocal cortical system. Copyright © 2009 Elsevier Ltd. All rights reserved.

Milani Anna, Lorusso Maria Luisa, Molteni Massimo (2010); THE EFFECTS OF AUDIOBOOKS ON THE PSYCHOSOCIAL ADJUSTEMENT OF PRE-ADOLESCENTS AND ADOLESCENTS WITH DYSLEXIA; Dyslexia, 16(1):87-97

I.F. 2009: 1,176

The objective of the present research study was to understand what benefits the use of audiobooks (both school-books and books of various genres, recorded on digital media) could bring to preadolescents and adolescents with developmental dyslexia. Two groups, each consisting of 20 adolescents, were compared. The experimental group used the audiobooks, while the control group continued to use normal books. After 5 months of experimental training, the experimental group showed a significant improvement in reading accuracy, with reduced unease and emotional-behavioural disorders, as well as an improvement in school performance and a greater motivation and involvement in school activities.

Molteni Massimo (2010); BEHAVIOR, HUMAN; Clinica Terapeutica, 161(1):91-92

I.F. 2009: 0,000

Human behavior is the collection of actions or reactions exhibited by human beings in relation to the environment, and it can be categorized as either innate or learned. In psychology, behavior became an important construct with the advent of behaviorism, a theoretical framework that required the study of only observable facts or events which can be seen or manipulated, in response to external or internal stimuli. More recently, the Relational Frame Theory (RFT) suggests that also some psychological events such as thoughts and emotions can be explained as learned responses. In TACT project, behavior is the voluntary movement of reaching and grasping in a fixed condition.

Molteni Massimo (2010); NEUROLOGY, CHILD; Clinica Terapeutica, 161(2):195-196

I.F. 2009: 0,000

Child Neurology concerns the study of the human nervous system during its development, as it is affected by conductivity, embryology and neuro-immunological factors in developmental brain disorders, coagulation disorders

and the metabolism of nervous tissue. During a neurological examination the differential diagnosis depends on the localization of the symptoms within vision, strength, coordination, refl exes and sensation. Neurological disorders of the central nervous system may represent the cause of some mental illnesses, making hard to discriminate between the fi eld of application proper of neurology and the one of psychiatry. As suggested by Jeste et al. (3) child neurologists may provide a substantial contribution to the investigation of neuro-behavioral disorders such as autism, in creation of neurologically based endophenotypes; the detection of early behavioral markers that precede a formal diagnosis and in the comprehension of disorders evolution through the life span.

Montirosso Rosario, Borgatti Renato, Trojan Sabina, Zanini Rinaldo, Tronick Edward (2010); A COMPARISON OF DYADIC INTERACTIONS AND COPING WITH THE STILL-FACE IN HEALTHY PRETERM AND FULL TERM INFANTS; British Journal of Developmental Psychology, 28(2):347-368

I.F. 2009: 1,418

Pre-term birth has a significant impact on infants' social and emotional competence, however, little is known about regulatory processes in pre-term mother–infant dyads during normal or stressful interactions. The primary goals of this study were to investigate the differences in infant and caregiver interactive behaviour and dyadic coordination of clinically healthy pre-term compared to full-term infant–mother dyads and to examine pre-term infants' capacity for coping with stress using the face-to-face still-face paradigm (FFSF). Fifty mother–infant dyads, including 25 pre-term infants and 25 full-term infants were videotaped during the FFSF. All infants were 6–9 months of age (corrected for gestational age in the pre-term group). Infant and maternal socioemotional expressivity and self-regulatory behaviours were coded and measures of dyadic coordination (Matching, Reparation Rate, and Synchrony) were calculated. There were no significant differences in infant and caregiver socio-emotional behaviours between the two groups and both groups demonstrated the still-face (SF) effect and the reunion effect. There was a difference in self-regulatory behaviour. Pre-term infants were more likely than full-term infants to use distancing (e.g., by turning away, twisting, or arching) from their mothers during the FFSF. Additionally, during the Reunion episode of the FFSF pre-term infants showed more social monitoring compared to full-term infants. Regardless of the birth status, the dyads showed less coordination and a slower rate of reparation during the Reunion episode than during the Play episode. The higher proportion of distancing in the pre-term

group and the increase in social monitoring suggest that even in normal interactions pre-term infants may experience a higher level of stress and have less capacity for self-regulation compared to the full-terms and that pre-term infants appear to use a compensatory strategy of increased social monitoring to cope with the stress of renegotiating the interaction during Reunion. The findings suggest that pre-term infants have different regulatory and interactive capacities than full-term infants.

Montirosso Rosario, Peverelli Milena, Frigerio Elisa, Crespi Monica, Borgatti Renato (2010); THE DEVELOPMENT OF DYNAMIC FACIAL EXPRESSION RECOGNITION AT DIFFERENT INTENSITIES IN 4- TO 18-YEAR-OLDS; Social Development, 19(1):71-92

I.F. 2009: 1,723

The primary purpose of this study was to examine the effect of the intensity of emotion expression on children's developing ability to label emotion during a dynamic presentation of five facial expressions (anger, disgust, fear, happiness, and sadness). A computerized task (AFFECT—animated full facial expression comprehension test) was used to display facial emotion expressions as animations with four levels of intensity (35, 50, 75, and 100 percent). In this study, which employed a cross-sectional design, 240 participants from 4 to 18 years completed the AFFECT. Results indicated that recognition ability developed for each of the emotions, with the exception of disgust, over the age range tested. Girls were more accurate than boys, especially for anger and disgust expressions. Recognition accuracy was found to increase as a function of the intensity of emotional expressions.

Montirosso Rosario, Cozzi Patrizia, Putnam Samuel P., Gartstein Maria A., Borgatti Renato (2010); STUDYING CROSS-CULTURAL DIFFERENCES IN TEMPERAMENT IN THE FIRST YEAR OF LIFE: UNITED STATES OF AMERICA (US) AND ITALY; International Journal of Behavioral Development, in press

I.F. 2009: 1,416

An Italian translation of the Infant Behavior Questionnaire-Revised (IBQ-R) was developed and evaluated with 110 infants, demonstrating satisfactory internal consistency, discriminant validity, and construct validity in the form of gender and age differences, as well as factorial integrity. Cross-cultural differences were subsequently evaluated for matched samples of Italian and United States (US) (N ¼ 110) 3–12-month-olds. Across infancy, parents of US infants reported higher levels of activity, high and low intensity pleasure, and vocal reactivity, whereas Italian infants, particularly males, were rated

higher on cuddliness. In early infancy only, US infants were viewed as higher on high intensity pleasure and perceptual sensitivity.

Montirosso Rosario, Riccardi Bruno, Molteni Erika, Borgatti Renato, Reni Gianluigi (2010); INFANT'S EMOTIONAL VARIABILITY ASSOCIATED TO INTERACTIVE STRESSFUL SITUATION: A NOVEL ANALYSIS APPROACH WITH SAMPLE ENTROPY AND LEMPEL-ZIV COMPLEXITY; Infant Behavior & Development, 33(3):346-356

I.F. 2009: 1,341

This study examined to which extent the lack of the mother's communicative input is associated to the variability of the infant's behavioral and emotional states at a microtemporal level. Two novel non-linear signal-processing metrics were used as regularity indexes during both normal and stressful mother-infant interactions (Face-to-Face Still-Face paradigm): (1) Sample Entropy estimates the presence of epochs of similar states in a data-series, according to a moment-to-moment analysis; (2) Lempel-Ziv Complexity evaluates the occurrence and recurrence of the patterns of analogous states along the data sequence. Fourteen mothers and their healthy full-term 7-month-old infants were videotaped and the infants' socio-emotional behaviors were micro-analytically coded off-line using a .20s time sampling method. During the maternal still-face episodes, when infants were confronted with the perturbation of their caregiver remaining unresponsive, both regularity indexes were lower than in normal interactions. Evidence is provided that non-linear techniques are suitable to detect variability in the infant's states.

Moruzzi Sara, Pesenti-Gritti Paola, Brescianini Sonia, Salemi Miriam, Battaglia Marco, Ogliari Anna (2010); CLUMSINESS AND PSYCHOPATHOLOGY: CAUSATION OR SHARED ETIOLOGY? A TWIN STUDY WITH THE CBCL 6-18 QUESTIONNAIRE IN A GENERAL SCHOOL-AGE POPULATION SAMPLE; Human Movement Science, 29(2):326-338

I.F. 2009: 2,148

In a sample of 398 twin pairs aged 8-17 belonging to the Italian Twin Registry we explored the extent to which physical clumsiness/motor problems covary with a broad spectrum of behavioral problems identified by the Child Behavior Checklist 6-18/DSM oriented scales, and the causes of such covariation. Only Anxiety and Attention Deficit Hyperactivity (ADH) Problems maintained significant correlation with Clumsiness after partialling out the effects of the other problem scales. By the co-twin control method we found no indication

of clear, direct causal effect of Clumsiness upon Anxiety or ADH Problems, or vice versa. Twin bivariate analyses showed that the co-occurrence of motor problems and Anxiety/ADH Problems is best explained by genetic factors shared between Clumsiness and the behavioral problems phenotypes.

Najt Pablo, Fusar-Poly Paolo, Brambilla Paolo (2010); CO-OCCURRING MENTAL AND SUBSTANCE ABUSE DISORDERS: A REVIEW ON THE POTENTIAL PREDICTORS AND CLINICAL OUTCOMES; Psychiatry Research, in press

I.F. 2009: 2,373

This article reviews the literature on co-occurring mental disorders and substance use disorders. The co-occurrence of mental disorders with substance use disorders presents a major challenge to those who provide psychiatric services. Despite the clinical and social burdens caused by this complex problem, research in this area is still insufficient. We found 18 studies showing potential predictors of co-occurring disorders (COD). Poor outcomes have been associated with: (i) COD compared to single disorders and (ii) COD with prior mental disorder compared to COD with prior substance use disorders. Poorer outcomes were reported for substance use disorder patients with comorbid major depressive disorder, and patients with substance use disorder and post-traumatic stress disorder. Furthermore, more negative outcomes were related to COD patients with temporally prior onset of mood disorders. Comorbidity between major depressive disorder or post-traumatic stress disorder and substance use disorder is suggested in the literature as a potential predictor of COD problems.

Nobile Maria, Rusconi Marianna, Bellina Monica, Marino Cecilia, Giorda Roberto, Carlet Ombretta, Vanzin Laura, Molteni Massimo, Battaglia Marco (2010); COMT VAL158MET POLYMORPHISM AND SOCIECONOMIC STATUS INTERACT TO PREDICT ATTENTION DEFICIT/HYPERACTIVITY PROBLEMS IN CHILDREN AGED 10-14; European Child & Adolescent Psychiatry,19(7):549-557

I.F. 2009: 1,651

The functional Val158Met COMT polymorphism appears to affect a host of behaviours mediated by the pre-frontal cortex, and has been found associated to the risk for disruptive behaviours including ADHD. Parental socioeconomic status (SES) has also been reported as a predictor for the same childhood disorders. In a general population sample of 575 Italian pre-adolescents aged 10-14, we examined the association of the functional Val158Met COMT polymorphism and SES-both as linear and interactive effects-with oppositional

defiant problems, conduct problems, and attention deficit/hyperactivity problems, as defined by the newly established Child Behaviour Check-List/6-18 DSM oriented scales. Multivariate- and subsequent univariate-analysis of covariance showed a significant association of COMT x SES interaction with CBCL 6/18 DOS attention deficit/hyperactivity problems ($p = 0.004$), and revealed higher scores among those children with Val/Val COMT genotype who belonged to low-SES families. We also found a significant association of SES with attention deficit/hyperactivity problems and conduct problems DOS ($p = 0.04$ and 0.01, respectively). Our data are consistent with a bulk of recent literature suggesting a role of environmental factors in moderating the contribution of specific genetic polymorphisms to human variability in ADHD. While future investigations will refine and better clarify which specific environmental and genetic mechanisms are at work in influencing the individual risk to ADHD in pre-adolescence, these data may contribute to identify/prevent the risk for ADHD problems in childhood.

Nobile Maria, Perego Paolo, Piccinini Luigi, Mani Elisa, Rossi Agnese, Bellina Monica, Molteni Massimo (2010); FURTHER EVIDENCE OF COMPLEX MOTOR DYSFUNCTION IN DRUG NAIVE CHILDREN WITH AUTISM USING AUTOMATIC MOTION ANALYSIS OF GAIT; Autism, in press

I.F. 2009: 2,534

Abstract non disponibile

Novara Francesca, Beri Silvana, Giorda Roberto, Ortibus Els, Nageshappa S., Darra Francesca, Dalla Bernardina Bernardo, Zuffardi Orsetta, Van Esch Hilde (2010); REFINING THE PHENOTYPE ASSOCIATED WITH MEF2C HAPLOINSUFFICIENCY; Clinical Genetics, Short Report in press

I.F. 2009: 3,304

Recently, submicroscopic deletions of the 5q14.3 region have been described in patients with severe mental retardation (MR), stereotypic movements, epilepsy and cerebral malformations. Further delineation of a critical region of overlap in these patients pointed to MEF2C as the responsible gene. This finding was further reinforced by the identification of a nonsense mutation in a patient with a similar phenotype. In brain, MEF2C is essential for early neurogenesis, neuronal migration and differentiation. Here we present two additional patients with severe MR, autism spectrum disorder and epilepsy, carrying a very small deletion encompassing the MEF2C gene. This finding strengthens the role of this gene in severe MR, and enables further

delineation of the clinical phenotype.

Ogliari Anna, Spatola Chiara A.M., Pesenti-Gritti Paola, Medda Emanuela, Penna Luana, Stazi Maria Antonietta, Battaglia Marco, Fagnani Corrado (2010); THE ROLE OF GENES AND ENVIRONMENT IN SHAPING CO-OCCURENCE OF DSM-IV DEFINED ANXIETY DIMENSIONS AMONG ITALIAN TWINS AGED 8-17; Journal of Anxiety Disorders, 24(4):433-439

I.F. 2009: 2,682

This study investigated the ultimate causes of co-variation between symptoms of four common DSM-IV anxiety dimensions - Generalized Anxiety, Panic, Social Phobia and Separation Anxiety disorder - assessed with the Italian version of the Screen for Child Anxiety-Related Emotional Disorders questionnaire in a sample of 378 twin pairs aged 8-17 from the population-based Italian Twin Register. Genetic and environmental proportions of covariance between the targeted anxiety dimensions were estimated by multivariate twin analyses. Genetic influences (explaining from 58% to 99% of covariance) and unique environmental factors were the sole sources of co-variation for all phenotypes under study. Genetic influences associated with different anxiety dimensions coincide remarkably, as indicated by genetic correlations ranging from 0.40 to 0.61, while unique environmental overlap is less substantial. Thus, while additive genetic effects are important in explaining why children report symptoms from multiple anxiety disorders, environmental idiosyncratic factors seem to play a marginal role in shaping the co-occurrence of different anxiety dimensions in childhood.

Ogliari Anna, Tambs Kristian, Harris Jennifer, Scaini Simona, Maffei Cesare, Reichborn-Kjennerud Ted, Battaglia Marco (2010); THE RELATIONSHIPS BETWEEN ADVERSE EVENTS, EARLY ANTECEDENTS, AND CARBON DIOXIDE REACTIVITY AS AN INTERMEDIATE PHENOTYPE OF PANIC DISORDER; Psychotherapy and Psychosomatics, 79(1):48-55

I.F. 2009: 5,368

BACKGROUND: Although adverse events have been consistently described to precede and potentially precipitate the onset of panic disorder, there is no information about their ability to alter the individual reactivity to inhaled carbon dioxide, a putative intermediate phenotype of susceptibility to panic disorder. METHOD: Seven-hundred twelve subjects belonging to the general population-based Norwegian Institute of Public Health Twin Panel under-

went a 35% CO(2)/65% O(2) inhalation challenge test and interview-based lifetime assessments of DSM-IV panic disorder, separation anxiety disorder, childhood parental separation/loss, major life events, adverse events of suffocative nature and common stressful life events. Regression models were applied to predict global subjective anxiety and DSM-IV panic symptoms after 35% CO(2)/65% O(2) inhalation. RESULTS: The responses to the challenge measured as semicontinuous variables were predicted by symptoms of childhood separation anxiety, childhood parental loss, common stressful events, major life events, suffocative events and the female gender. The role of most of these predictors was confirmed and held true after the exclusion of subjects with lifetime panic attacks/disorder from the analyses. CONCLUSIONS: Several factors which have been reported by previous clinical studies to influence the individual susceptibility to develop panic disorder seem to affect the individual reactivity to inhaled carbon dioxide in people from the general population. Some elements of risk may impact simultaneously upon the individual liability to panic and exaggerated sensitivity to hypercapnia.

Optale Gabriele, Urgesi Cosimo, Busato Valentina, Marin Silvia, Piron Lamberto, Priftis Konstantinos, Gamberini Luciano, Capodieci Salvatore, Bordin Adalberto (2010); CONTROLLING MEMORY IMPAIRMENT IN ELDERLY ADULTS USING VIRTUAL REALITY MEMORY TRAINING: A RANDOMIZED CONTROLLED PILOT STUDY; Neurorehabilitation and Neural Repair, 24(4):348-357

I.F. 2009: 5,398

BACKGROUND: Memory decline is a prevalent aspect of aging but may also be the first sign of cognitive pathology. Virtual reality (VR) using immersion and interaction may provide new approaches to the treatment of memory deficits in elderly individuals. OBJECTIVE: The authors implemented a VR training intervention to try to lessen cognitive decline and improve memory functions. METHODS: The authors randomly assigned 36 elderly residents of a rest care facility (median age 80 years) who were impaired on the Verbal Story Recall Test either to the experimental group (EG) or the control group (CG). The EG underwent 6 months of VR memory training (VRMT) that involved auditory stimulation and VR experiences in path finding. The initial training phase lasted 3 months (3 auditory and 3 VR sessions every 2 weeks), and there was a booster training phase during the following 3 months (1 auditory and 1 VR session per week). The CG underwent equivalent face-to-face training sessions using music therapy. Both groups participated in social and creative and assisted-mobility activities. Neuropsychological and functional evaluations were performed at baseline, after the initial training phase, and

after the booster training phase. RESULTS: The EG showed significant improvements in memory tests, especially in long-term recall with an effect size of 0.7 and in several other aspects of cognition. In contrast, the CG showed progressive decline. CONCLUSIONS: The authors suggest that VRMT may improve memory function in elderly adults by enhancing focused attention.

Pagnamenta Alistair T., Khan Hameed, Walker Susan, Gerrelli Dianne, Wing Kirsty, Bonaglia Maria Clara, Giorda Roberto, Berney Tom, Mani Elisa, Molteni Massimo, Pinto Dalila, Le Couteur Ann, Hallmayer Joachim, Sutcliffe James S., Szatmari Peter, Paterson Andrew D., Scherer Stephen W., Vieland Veronica J., Monaco Anthony P. (2010); RARE FAMILIAL 16Q21 MICRODELETIONS UNDER A LINKAGE PEAK IMPLICATE CADHERIN 8 (CDH8) IN SUSCEPTIBILITY TO AUTISM AND LEARNING DISABILITY; Journal of Medical Genetics, in press

I.F. 2009: 5,751

Abstract non disponibile

Pastore Valentina, Colombo Katia, Liscio Mariarosaria, Galbiati Susanna, Adduci Annarita, Villa Federica, Strazzer Sandra (2010); EFFICACY OF COGNITIVE BEHAVIOURAL THERAPY FOR CHILDREN AND ADOLESCENTS WITH TRAUMATIC BRAIN INJURY; Disability and Rehabilitation, in press

I.F. 2009: 1,555

Purpose. Behavioural and psychological disorders after traumatic brain injury (TBI) are very common. The purposes of this study were to estimate the frequency of these problems in our sample, to evaluate the effectiveness of cognitive behavioural therapy (CBT) and to assess the predictive value of important clinical variables for the treatment outcome. Method. Forty patients aged 4-18 years were included in this study. Twenty-eight patients received CBT at our Institute (clinical group), while 12 patients did not receive any treatment at all (control group). The CBCL/4-18 and the VABS were administered to parents at the beginning of the study and after 12 months. Results. A high frequency of psychological and behavioural problems was found in both groups of patients. After CBT, the clinical group showed a significant advantage on several CBCL scales and a greater increase in adaptive behaviour on the VABS Socialisation domain. The Glasgow Coma Scale score, days of unconsciousness and age at injury were not predictors of the severity of psychological problems at the follow-up for the patients of the clinical group. Conclusions. Our results suggest that CBT is an effective intervention

for young patients with psychological problems after TBI.

Perrotta Cristiana, Clementi Emilio (2010); BIOLOGICAL ROLES OF ACID AND NEUTRAL SPHINGOMYELINASES AND THEIR REGULATION BY NITRIC OXIDE; Physiology, 25(2):64-71

I.F. 2009: 6,945

Generation of the pleiotropic sphingolipid mediator ceramide by acid and neutral sphingomyelinases is a key event in many cellular pathophysiological processes including survival, death, proliferation, and differentiation, in which also the short-lived gaseous messenger nitric oxide plays a crucial role. This review describes how the outcome of these key cellular processes is finely tuned by surprising and complex interplays among nitric oxide, ceramide, and their effectors.

Piazza Manuela, Facoetti Andrea, Trussardi Anna Noemi, Berteletti Ilaria, Conte Stefano, Lucangeli Daniela, Dehaene Stanislas, Zorzi Marco (2010); DEVELOPMENTAL TRAJECTORY OF NUMBER ACUITY REVEALS A SEVERE IMPAIRMENT IN DEVELOPMENTAL DYSCALCULIA; Cognition, 116(1):33-41

I.F. 2009: 3,562

Developmental dyscalculia is a learning disability that affects the acquisition of knowledge about numbers and arithmetic. It is widely assumed that numeracy is rooted on the “number sense”, a core ability to grasp numerical quantities that humans share with other animals and deploy spontaneously at birth. To probe the links between number sense and dyscalculia, we used a psychophysical test to measure the Weber fraction for the numerosity of sets of dots, hereafter called number acuity. We show that number acuity improves with age in typically developing children. In dyscalculics, numerical acuity is severely impaired, with 10-year-old dyscalculics scoring at the level of 5-year-old normally developing children. Moreover, the severity of the number acuity impairment predicts the defective performance on tasks involving the manipulation of symbolic numbers. These results establish for the first time a clear association between dyscalculia and impaired “number sense”, and they may open up new horizons for the early diagnosis and rehabilitation of mathematical learning deficits. 2010 Elsevier B.V. All rights reserved.

Piccinelli Paolo, Beghi Ettore, Borgatti Renato, Ferri Matteo, Giordano Laura, Romeo Antonino, Termine Cristiano, Viri Maurizio, Zucca Claudio, Balottin Umberto (2010); NEUROPSYCHOLOGICAL AND BEHAVIOURAL ASPECTS IN CHILDREN AND ADOLESCENTS

WITH IDIOPATHIC EPILEPSY AT DIAGNOSIS AND AFTER 12 MONTHS OF TREATMENT; Seizure - European Journal of Epilepsy, in press

I.F. 2009: 2,233

Purpose: To study neuropsychological functions in children with idiopathic epilepsy at onset of treatment and after 1 year of therapy and to identify factors associated with cognitive impairment.
Methods: 43 Subjects aged 5.2–16.9 years with newly diagnosed idiopathic epilepsy were enrolled and started treatment with valproate or carbamazepine. At admission and after 12 months, all patients underwent clinical examinations, the Child Behavioural Checklist, EEG and a neuropsychological test battery. The results of each test were correlated to demographic, clinical, electrophysiological and therapeutic variables.
Results: Except for attention, all neuropsychological functions were normal at admission and after 12 months. An improvement with time was noted for memory ($p < 0.05$) and logical-executive functions ($p < 0.01$). Attentive deficit was worse at 12 months (53.5% vs. 32.6%). Low socio-economic level and emotional and behavioural disturbances were the only factors negatively correlated to intelligence, memory and attention. Compared to valproate, carbamazepine was most commonly implicated.
Discussion: Idiopathic epilepsy can affect attention, even before starting treatment. Emotional and behavioural difficulties and a low socio-economical status are associated with cognitive impairment.

Piccinini Luigi, Cimolin Veronica, D’Angelo Maria Grazia, Turconi Anna Carla, Crivellini Marcello, Galli Manuela (2010); 3D GAIT ANALYSIS IN PATIENTS WITH HEREDITARY SPASTIC PARAPARESIS AND SPASTIC DIPLEGIA: A KINEMATIC, KINETIC AND EMG COMPARISON; European Journal of Paediatric Neurology, in press

I.F. 2009: 2,007

The predominant clinical feature of patients with Hereditary Spastic Paraparesis (HSP) is gait disturbance owing to spasticity and weakness of the lower limbs; the spasticity in early-onset disease (infancy or childhood) often cannot be distinguished from mild form of spastic diplegia (SD). The aim of this study was to quantify the gait strategy in HSP and SD children, focusing on the differences between groups as concerns functional limitation during gait. 9 HSP and 16 SD children were evaluated using Gait Analysis; kinematic and kinetic parameters and EMG pattern during walking were identified and

calculated to compare the two gait strategies. The results revealed that these two pathologies are characterised by different gait strategies. In particular we found that knee joint, in terms of kinematics and kinetics, and rectus femoris pattern represent discriminatory aspects in order to compare and differentiate gait patterns of HSP and SD children. The findings strongly support the issue that HSP and SD patients need individualised therapeutical program, either neurosurgical or pharmacological treatment, based on the quantification of gait deficiencies and in order to address the peculiarity of their motor limitations and to prevent the onset of compensatory strategies.

Pittaccio Simone, Viscuso Stefano, Beretta Elena, Turconi Anna Carla, Strazzer Sandra (2010); PILOT STUDIES SUGGESTING NEW APPLICATIONS OF NITI IN DYNAMIC ORTHOSES FOR ANKLE JOINT; Prosthetics and Orthotics International, 34(3):305-318

I.F. 2009: 0,563

NiTi is a metal alloy with unconventional functional characteristics: Shape memory and pseudoelasticity. Its use in the field of rehabilitation is very innovative. This work presents applications in lower limb orthotics. Three different devices were assembled and tested: An equinus gait dynamic splint, a compliant ankle positioning brace, and a dual-mode haptic/active exerciser for the dorsiflexors. Results are derived from technical and preclinical trials. The gait splint improves several walking parameters even better than a traditional flexible ankle-foot orthoses (AFO). In particular, it supports mid-stance and propulsion biomechanics and affects physiological activation of tibialis anterior during swing much less than posterior leaf AFO. The haptic/active exerciser, able to provide dorsiflexion through a suitable articular range, could be controlled on the basis of minimal surface electromyographic (sEMG) signals, suggesting its use as an aid for early active workouts as soon as patients start to recover voluntary control of tibialis anterior. Further evidence must be sought in future to confirm for the ankle joint the promising results obtained in repositioning applications in prior upper limb studies. The work done so far on the tested prototypes is encouraging: Material characteristics and dimensioning will be optimized so that customized NiTi devices can be prescribed to best meet individual patients' requirements.

Porcelli Anna Maria, Ghelli Anna, Ceccarelli Claudio, Lang Martin, Cenacchi Giovanna, Capristo Mariantonietta, Pennisi Lucia Fiammetta, Morra Isabella, Ciccarelli Enrica, Melcarne Antonio, Bartoletti-Stella Anna, Salfi Nunzio, Tallini Giovanni, Martinuzzi Andrea, Carelli Valerio, Attimonelli Marcella, Rugolo Michela,

Romeo Giovanni, Gasparre Giuseppe (2010); THE GENETIC AND METABOLIC SIGNATURE OF ONCOCYTIC TRANSFORMATION IMPLICATES HIF1alpha DESTABILITAZION; Human Molecular Genetics, 19(6):1019-1032

I.F. 2009: 7,386

We previously showed that disruptive complex I mutations in mitochondrial DNA are the main genetic hallmark of oncocytic tumors of the thyroid and kidney. We here report a high frequency of homoplasmic disruptive mutations in a large panel of oncocytic pituitary and head-and-neck tumors. The presence of such mutations implicates disassembly of respiratory complex I in vivo which in turn contributes to the inability of oncocytic tumors to stabilize HIF1alpha and to display pseudo-hypoxia. By utilizing transmitochondrial cytoplasmic hybrids (cybrids), we induced the shift to homoplasmy of a truncating mutation in the mitochondria-coded MTND1 gene. Such shift is associated with a profound metabolic impairment leading to the imbalance of alpha-ketoglutarate and succinate, the Krebs cycle metabolites which are the main responsible for HIF1alpha stabilization. We conclude that the main hallmarks of oncocytic transformation, namely the occurrence of homoplasmic disruptive mutations and complex I disassembly, may explain the benign nature of oncocytic neoplasms through lack of HIF1alpha stabilization.

Pozzoli Uberto, Fumagalli Matteo, Cagliani Rachele, Comi Giacomo Pietro, Bresolin Nereo, Clerici Mario, Sironi Manuela (2010); THE ROLE OF PROTOZOA-DRIVEN SELECTION IN SHAPING HUMAN GENETIC VARIABILITY; Trends in Genetics, 26(3):95-99

I.F. 2009: 8,689

Protozoa exert a strong selective pressure in humans. The selection signatures left by these pathogens can be exploited to identify genetic modulators of infection susceptibility. We show that protozoa diversity in different geographic locations is a good measure of protozoa-driven selective pressure; protozoa diversity captured selection signatures at known malaria resistance loci and identified several selected single nucleotide polymorphisms in immune and hemolytic anemia genes. A genome-wide search enabled us to identify 5180 variants mapping to 1145 genes that are subjected to protozoa-driven selective pressure. We provide a genome-wide estimate of protozoa-driven selective pressure and identify candidate susceptibility genes for protozoa-borne diseases.

Prigione Alessandro, Piazza Fabrizio, Brighina Laura, Begni Barbara, Galbussera Alessio, Di Francesco Jacopo, Andreoni

Simona, Piolti Roberto, Ferrarese Carlo (2010); ALPHA-SYNUCLEIN NITRATION AND AUTOPHAGY RESPONSE ARE INDUCED IN PERIPHERAL BLOOD CELLS FROM PATIENTS WITH PARKINSON DISEASE; Neuroscience Letters, 477(1):6-10

I.F. 2009: 1,925

Several lines of evidence implicate a central role for alpha-synuclein (aSN) in the pathogenesis of Parkinson's disease (PD). Besides rare genetic mutations, post-translational mechanisms, such as oxidative stress-related nitration, may alter the protein properties in terms of propensity to aggregate or be degraded. Our group previously described increased reactive oxygen species (ROS) production within easily accessible peripheral blood mononuclear cells (PBMCs) in PD patients compared to healthy elderly subjects. In the present work, we demonstrated a significant induction of nitrotyrosine (NT)-modifications of aSN within PBMCs derived from individuals with idiopathic PD compared to controls, while aSN protein appeared similarly expressed in the two populations. The amount of NT-modified aSN within PBMCs was positively correlated with intracellular ROS concentration and inversely related to daily dosage of levodopa, making its measurement potentially relevant for disease-intervention studies. Neither aSN expression nor its NT-modifications showed any correlation to specific REP1 genotypes, polymorphic variants within aSN gene promoter whose association to PD susceptibility may occur through the modulation of aSN protein expression. Moreover, although NT-modified aSN has been linked to enhanced propensity to aggregate, we failed to detect an increased presence of insoluble aSN aggregates in PBMCs from PD subjects relative to controls, despite a lack of changes in the ubiquitin-proteasome expression or activity. Nonetheless, a significant activation of the autophagy response was identified within PBMCs from PD individuals, which could represent a protective mechanism against abnormal protein accumulation and may explain the lack of aSN aggregation. We discuss the relevance of these findings with respect to PD pathogenesis and biomarker development. 2010 Elsevier Ireland Ltd. All rights reserved.

Resta Nicoletta*, Giorda Roberto*, Bagnulo Rosanna, Beri Silvana, Della Mina Erika, Stella Alessandro, Piglionica Marilidia, Susca Francesco Claudio, Guanti Ginevra, Zuffardi Orsetta, Ciccone Roberto (2010); BREAKPOINT DETERMINATION OF 15 LARGE DELETIONS IN PEUTZ-JEGHERS SUBJECTS; Human Genetics, 128(4):373-382

I.F. 2009: 4,523

**Autori che hanno contribuito in ugual misura al lavoro*

The Peutz-Jeghers Syndrome (PJS) is an autosomal dominant polyposis disorder with increased risk of multiple cancers. STK11/LKB1 (hereafter named STK11) germline mutations account for the large majority of PJS cases whereas large deletions account for about 30% of the cases. We report here the first thorough molecular characterization of 15 large deletions identified in a cohort of 51 clinically well-characterized PJS patients. The deletions were identified by MLPA analysis and characterized by custom CGH-array and quantitative PCR to define their boundaries. The deletions, ranging from 2.9 to 180 kb, removed one or more loci contiguous to the STK11 gene in six patients, while partial STK11 gene deletions were present in the remaining nine cases. By means of DNA sequencing, we were able to precisely characterize the breakpoints in each case. Of the 30 breakpoints, 16 were located in Alu elements, revealing non-allelic homologous recombination (NAHR) as the putative mechanism for the deletions of the STK11 gene, which lays in a region with high Alu density. In the remaining cases, other mechanisms could be hypothesized, such as microhomology-mediated end-joining (MMEJ) or non-homologous end-joining (NHEJ). In conclusion we here demonstrated the non-random occurrence of large deletions associated with PJS. All our patients had a classical PJS phenotype, which shows that haploinsufficiency for SBNO2, C19orf26, ATP5D, MIDN, C19orf23, CIRBP, C19orf24,and EFNA2, does not apparently affect their clinical phenotype.

Restuccia Domenico, Del Piero Ivana, Martucci Lucia, Zanini Sergio (2010); HIGH-FREQUENCY OSCILLATIONS AFTER MEDIAN-NERVE STIMULATION DO NOT UNDERGO HABITUATION: A NEW INSIGHT ON THEIR FUNCTIONAL MEANING?; Clinical Neurophysiology, in press

I.F. 2009: 3,122

OBJECTIVE: Amplitude decrease of cortical responses after repeated stimuli ('habituation') is a well-known phenomenon, the functional meaning of which is to prevent sensory overflow and to save resources for meaningful and novel stimuli. It is known that the primary low-frequency N20 somatosensory evoked potential (SEP) undergoes habituation in healthy subjects. By contrast, the presence of this phenomenon has never been tested in High Frequency Oscillations (HFOs), which probably reflect the activity of a somatosensory arousal system. METHODS: We recorded SEPs after right median nerve stimulation in 19 healthy volunteers. Six consecutive series of 500 sweeps were collected and averaged at a repetition rate of 5Hz. SEPs were recorded by means of Erb'point-to-Fz, Cv6-to-AC and P3-to-F3 arrays. P3-to-F3 recording further underwent narrow-bandpass (400-800Hz) digital

filtering to selectively analyse high-frequency components. RESULTS: Statistical analysis revealed a significant amplitude decrease of the primary N20 LF-SEP between the first and sixth block of stimuli. By contrast, HFO amplitudes remained substantially unchanged throughout the whole procedure. CONCLUSIONS: Differently from the N20 LF-SEP, scalp-recorded HFOs do not undergo habituation. SIGNIFICANCE: Our findings reinforce the view that HFOs reflect the activity of an arousal somatosensory system, which is able to signal novel stimuli, the relevance of which points out high synaptic efficacy.

Rizzotto Melissa Rosa, Visonà Dalla Pozza L., Turconi Anna Carla, Tornetta L., Andreucci Elena, Zambonin Fabio, Fedrizzi Ermellina, Facchin Paola (2010); THE PERCEPTION OF INVOLVED PROFESSIONALS TOWARDS RESEARCH FEASIBILITY AND USEFULNESS: LESSONS FROM THE MULTI-SITE TRIAL ON EFFICACY OF CONSTRAINT INDUCED MOVEMENT THERAPY IN CHILDREN WITH HEMIPLEGIA; European Journal of Physical and Rehabilitation Medicine (continues Europa Medicophysica), in press

I.F. 2009: 0,000

BACKGROUND: In the last decades, the world of rehabilitation has been more and more calling for clear evidence to support intervention and numerous research programs have been developed. At stake, relatively little research on opinions and attitude of rehabilitation personnel involved in research conducted in real clinical settings has been carried out. AIM: To explore the opinion of professionals involved in a national clinical trial on research. DESIGN: Multicentre cross-sectional study. SETTING: 19 rehabilitation centres/services (4 research institutes, 15 local rehabilitation services). POPULATION: All professional participating to a multi-centre clinical trial on the effects of Constraint Induced Movement Therapy on children with hemiplegic cerebral palsy. METHODS: A 15-questions questionnaire inquiring feasibility, usefulness, products, costs, judgement and perceptions about clinical research in rehabilitation was admistered. RESULTS: Among those working in one of the 19 rehabilitation centres part of the multicentric study, 76 professionals were asked to fill in the questionnarie. 68 professionals answered (89.4% of response rate). More than 75% of the sample thinks that its rehabilitation centre is suited to develop clinical research. Research results useful for the development of their daily activities (new tools for the assessment of children, to demonstrate the efficacy of a new treatment option and to learn a new way of working, and to strengthen the ties within the working team). Research is

costly in terms of personal time and effort, but it can modify the rehabilitation praxis (assessment tools, the relationship with colleagues/patients). 98% of the interviewees declared the willingness to participate to other research projects. CONCLUSION AND CLINICAL REHABILITATION IMPACT: This survey highlights the importance of conducting research in local rehabilitation services, not only in terms of generation of new evidences, but also in terms of building networks, sharing experiences and knowledge, connecting with centers of excellence and providing a specific training for research conduction.

Romaniello Romina, Zucca Claudio, Tonelli Alessandra, Bonato Sara, Baschirotto Cinzia, Zanotta Nicoletta, Epifanio Roberta, Righini Andrea, Bresolin Nereo, Bassi Maria Teresa, Borgatti Renato (2010); A WIDE SPECTRUM OF CLINICAL, NEUROPHYSIOLOGICAL AND NEURORADIOLOGICAL ABNORMALITIES IN A FAMILY WITH A NOVEL CACNA1A MUTATION; Journal of Neurology, Neurosurgery and Psychiatry, 81(8):840-843

I.F. 2009: 4,869

Background Mutations in the calcium channel voltage dependent P/Q-type alpha-1A subunit (CACNA1A) can cause different neurological disorders which share a wide range of symptoms, including episodic ataxia type 2 (EA2), familial hemiplegic migraine (FHM1) and progressive spinocerebellar ataxia (SCA6). Objective To describe a three generations family in which a spectrum of different phenotypes, ranging from SCA6 (proband), to EA2 (proband's mother) to FHM1 (proband's mother and proband's aunt) was found. All of the family members carried a novel CACNA1A missense mutation. Patients and methods A clinical, molecular, neuroradiological and neurophysiological study was carried out in all subjects. Results A single heterozygous base change in exon 9, c1213G-->A, leading to the amino acid substitution pAla405Thr was found to segregate within the family. Brain MRI showed cerebellar and cerebral atrophy signs in all but one mutation carriers. Neurophysiological findings (electroencephalography and evoked potentials) confirmed possible cerebral cortex and white matter involvement regardless of the clinical symptoms displayed. Conclusions This novel CACNA1A mutation adds to the number of mutations associated with a heterogeneous clinical picture in family members. This mutation might affect the interaction between the intracellular loops and the beta subunit, leading to a relatively rapid cell death. In order to explain the wide phenotypic variability observed in this family, it is hypothesised that additional genetic and environmental (hormonal) factors play a role in the pathophysiology of the disease.

Romei Marianna, Lo Mauro Antonella, D'Angelo Maria Grazia, Turconi Anna Carla, Bresolin Nereo, Pedotti Antonio, Aliverti Andrea (2010); EFFECTS OF GENDER AND POSTURE ON THORACO-ABDOMINAL KINEMATICS DURING QUIET BREATHING IN HEALTHY ADULTS; Respiratory Physiology & Neurobiology, 172(3):184-191

I.F. 2009: 2,135

To investigate the effects of posture and gender on thoraco-abdominal motion and breathing pattern, 34 healthy men and women were studied by Opto-Electronic Plethysmography during quiet breathing in five different postures from seated (with and without back support) to supine position. Chest wall kinematics and breathing pattern were significantly influenced by position and gender. The progressively increased inclination of the trunk determined a progressive reduction of rib cage displacement, tidal volume, and minute ventilation and a progressive increase of abdominal contribution to tidal volume. Female subjects were characterized by smaller dimensions of the rib cage compartment and during quiet breathing by lower tidal volume, minute ventilation and abdominal contribution to tidal volume than males. The effect of posture on abdominal kinematics was significant only in women. The presence of a back support in seated position determined differences in breathing pattern. In conclusion, posture and gender have a strong influence on breathing and on chest wall kinematics. Copyright 2010 Elsevier B.V. All rights reserved.

Ronchi Dario, Virgilio Roberta, Bordoni Andreina, Fassone Elisa, Sciacco Monica, Ciscato Patrizia, Moggio Maurizio, Govoni Alessandra, Corti Stefania, Bresolin Nereo, Comi Giacomo Pietro (2010); THE M.12316G>A MUTATION IN THE MITOCHONDRIAL tRNALEU(CUN) GENE IS ASSOCIATED WITH MITOCHONDRIAL MYOPATHY AND RESPIRATORY IMPAIRMENT; Journal of the Neurological Sciences, 292(1-2):107-110

I.F. 2009: 2,324

Mitochondrial disorders are often associated with mutations in mitochondrial tRNA. Independent observation of the same molecular defect in unrelated subjects is a generally required proof of pathogenicity. A sporadic case of chronic external ophthalmoplegia (cPEO) with ragged red fibres (RRFs) has been previously related to an m.12316G>A substitution in tRNA(Leu(CUN)). Sequencing muscle-derived mtDNA, we found the m.12316G>A substitution in an adult woman with mitochondrial myopathy and respiratory impairment. Her muscle biopsy presented several cytochrome c oxidase-negative (COX-) fibres, and RRFs as signs of mitochondrial proliferation. Restriction-fragment length polymorphism (RFLP) analysis of the mutation in isolated muscle fibres showed a threshold of at least 60% of mutated mtDNA to determine a COX deficiency phenotype. This second report of the m.12316G>A mutation in a sporadic patient consolidates its pathogenic nature and provides further elements for genetic counselling. Copyright 2010 Elsevier B.V. All rights reserved.

Ruffino Milena, Trussardi Anna Noemi, Gori Simone, Finzi Alessandra, Giovagnoli Sara, Menghini Deny, Benassi Mariagrazia, Molteni Massimo, Bolzani Roberto, Vicari Stefano, Facoetti Andrea (2010); ATTENTIONAL ENGAGEMENT DEFICITS IN DYSLEXIC CHILDREN; Neuropsychologia, in press

I.F. 2009: 4,345

Reading acquisition requires, in addition to appropriate phonological abilities, accurate and rapid selection of sublexical orthographic units by attentional letter string parsing. Spatio-temporal distribution of attentional engagement onto 3-pseudoletter strings was studied in 28 dyslexic and 55 normally reading children by measuring attentional masking (AM). AM refers to an impaired identification of the first of two sequentially presented masked objects (O1 and O2). In the present study, O1 was always centrally displayed, whereas the location of O2 (central or lateral) and the O1-O2 interval were manipulated. Dyslexic children showed a larger AM at the shortest O1-O2 interval and a sluggish AM recovery at the longest O1-O2 interval, as well as an abnormal lateral AM. More importantly, these spatio-temporal deficits of attentional engagement were selectively present in dyslexics with poor phonological decoding skills. Our results suggest that an inefficient spatio-temporal distribution of attentional engagement - probably linked to a parietal lobule dysfunction - might selectively impair the letter string parsing mechanism during phonological decoding.

Sala Gessica, Brighina Laura, Saracchi Enrico, Fermi Silvia, Riva Chiara, Carrozza Veronica, Pirovano Marta, Ferrarese Carlo (2010); VESICULAR MONOAMINE TRANSPORTER 2 MRNA LEVELS ARE REDUCED IN PLATELETS FROM PATIENTS WITH PARKINSON'S DISEASE; Journal of Neural Transmission, 117(9):1093-1098

I.F. 2009: 2,259

Despite advances in neuroimaging, the diagnosis of idiopathic Parkinson's disease (PD) remains clinical. The identification of biological markers for an early diagnosis is of great interest to start a neuroprotective therapy aimed

at slowing, blocking or reversing the disease progression. Vesicular monoamine transporter 2 (VMAT2) sequesters cytoplasmic dopamine into synaptic vesicles for storage and release. Thus, VMAT2 impairment can regulate intra- and extracellular dopamine levels, influencing oxidative stress and neuronal death. Because in vivo imaging studies have demonstrated a VMAT2 reduction in PD patients greater than would be explained by neuronal loss alone, as an exploratory study we assessed VMAT2 mRNA and protein levels in platelets from 39 PD patients, 39 healthy subjects and 10 patients with vascular parkinsonism (VP) to identify a possible peripheral biomarker for PD. A significant reduction ($p < 0.05$) of VMAT2 mRNA levels was demonstrated in PD patients versus healthy controls. Patients with VP showed VMAT2 mRNA levels similar to controls. No difference in VMAT2 mRNA levels was found in untreated versus treated patients. No correlation was observed between mRNA levels and demographic or clinical characteristics. Furthermore, eight SNPs tagging the VMAT2 gene did not show effects on VMAT2 mRNA levels. Western blot analysis did not allow the quantification of VMAT2 protein expression in blood platelets. Although further studies in a greater number of cases are needed to confirm our data, the reduction in VMAT2 mRNA in platelets from PD patients suggests the existence of a systemic impairment of this transporter possibly contributing to PD pathology.

Sciorati Clara, Buono Roberta, Azzoni Emanuele, Casati Silvana, Ciuffreda Pierangela, D'Angelo Maria Grazia, Cattaneo Dario, Brunelli Silvia, Clementi Emilio (2010); CO-ADMINISTRATION OF IBUPROFEN AND NITRIC OXIDE IS AN EFFECTIVE EXPERIMENTAL THERAPY FOR MUSCULAR DYSTROPHY, WITH IMMEDIATE APPLICABLITY TO HUMANS; British Journal of Pharmacology, 160(6):1550-1560

I.F. 2009: 5,204

Background and purpose: Current therapies for muscular dystrophy are based on corticosteroids. Significant side effects associated with these therapies have prompted several studies aimed at identifying possible alternative strategies. As inflammation and defects of nitric oxide (NO) generation are key pathogenic events in muscular dystrophies, we have studied the effects of combining the NO donor isosorbide dinitrate (ISDN) and the non-steroidal anti-inflammatory drug ibuprofen. Experimental approach: alpha-Sarcoglycan-null mice were treated for up to 8 months with ISDN (30 mg.kg(-1)) plus ibuprofen (50 mg.kg(-1)) administered daily in the diet. Effects of ISDN and ibuprofen alone were assessed in parallel. Drug effects on animal motility and muscle function, muscle damage, inflammatory infiltrates and cytokine

levels, as well as muscle regeneration including assessment of endogenous stem cell pool, were measured at selected time points. Key results: Combination of ibuprofen and ISDN stimulated regeneration capacity, of myogenic precursor cells, reduced muscle necrotic damage and inflammation. Muscle function in terms of free voluntary movement and resistance to exercise was maintained throughout the time window analysed. The effects of ISDN and ibuprofen administered separately were transient and significantly lower than those induced by their combination. Conclusions and implications: Co-administration of NO and ibuprofen provided synergistic beneficial effects in a mouse model of muscular dystrophy, leading to an effective therapy. Our results open the possibility of immediate clinical testing of a combination of ISDN and ibuprofen in dystrophic patients, as both components are approved for use in humans, with a good safety profile.

Sciruicchio Vittorio, Sardaro Michele, Gagliardi Delio, Trabacca Antonio, Galeone Dante, De Tommaso Marina (2010); A CASE OF EARLY-ONSET AND MONOPHASIC TRIGEMINAL AUTONOMIC CEPHALALGIA: COULD IT BE A SUNCT?; Journal of Headache and Pain (The), 11(4):363-365

I.F. 2009: 2,137

A 2-year-old female came to the Neurological Emergency Room of "Giovanni XXIII" Hospital in Bari, 6 h after the onset of severe facial pain, which occurred soon after awakening. Stabbing pain affected the right frontal and periorbital area, with ipsilateral conjunctival injection, swelling of the eyelids and tearing. Except the duration, from 5 to 30 s., the attacks were stereotyped including the occurrence and features of autonomic signs. Based on the typical clinical findings and the normal magnetic resonance imaging (MRI), we diagnosed short-lasting unilateral neuralgiform headache attacks with conjunctival injection and tearing syndrome (SUNCT). The spontaneous remission within a few hours made prophylactic therapy unnecessary. At the last follow-up, after 3 months, the patient was still symptom free. In our case, after an active period lasting 2 days the disease disappeared completely. However the typical features of the disease (unilateral pain, short duration and high frequency of the attacks, autonomic signs ipsilateral to pain, numbers of attacks) were all present. While the diagnostic criteria of the International Headache Society classification for SUNCT did not include the duration of disease, it is likely that the active period lasting 2 days could be an expression of the clinical variability of the disease.

Siri Chiara, Cilia Roberto, De Gaspari Danilo, Villa Federica,

Goldwurm Stefano, Catalano Marco, Pezzoli Gianni, Antonini Angelo (2010); PSYCHIATRIC SYMPTOMS IN PARKINSON'S DISEASE ASSESSED WITH THE SCL-90R SELF-REPORTED QUESTIONNAIRE; Neurological Sciences, 31(1):35-40

I.F. 2009: 1,120

The frequency of psychopathological symptoms in patients with Parkinson's disease (PD) is often underestimated because of the lack of comprehensive evaluation tools. A total of 486 consecutive non-demented PD patients completed the Symptom Checklist 90 Revised (SCL-90R) self-reported questionnaire, a validated tool for the assessment of psychopathological symptoms on nine dimensions. Somatization, depression, anxiety and obsessive-compulsive behaviors were reported by nearly half of the PD patients. They were more likely to occur in females. Disease-related factors such as duration, severity and daily dosages, but not type of dopaminergic medications, were associated with the occurrence of these symptoms. Psychopathological features are frequent in PD and their occurrence is underlined by disease-related factors.

Sironi Manuela, Clerici Mario (2010); THE HYGIENE HYPOTHESIS: AN EVOLUTIONARY PERSPECTIVE; Microbes and Infection, 12(6):421-427

I.F. 2009: 2,757

The hygiene hypothesis relies on the assumption that humans have adapted to a pathogen-rich environment that no longer exists in industrialized societies. Recent advances in molecular immunology and population genetics allow deeper insight into the evolution and co-evolution of host-pathogen interactions and, therefore, into the foundations of the hygiene hypothesis.

Spatola Chiara A.M., Rende Richard, Battaglia Marco (2010); GENETIC AND ENVIRONMENTAL INFLUENCES UPON THE CBCL/6-18 DSM-ORIENTED SCALES: SIMILARITIES AND DIFFERENCES ACROSS THREE DIFFERENT COMPUTATIONAL APPROACHES AND TWO AGE RANGERS; European Child & Adolescent Psychiatry, 19(8):647-658

I.F. 2009: 1,651

Inasmuch as the newly established DSM-oriented CBCL/6-18 scales are to be increasingly employed to assess clinical/high-risk populations, it becomes important to explore their aetiology both within the normal- and the

extreme range of variation in general population samples and to compare the results obtained in different age groups. We investigated by the Quantitative Maximum Likelihood, the De Fries-Fulker, and the Ordinal Maximum Likelihood methods the genetic and environmental influences upon the five DSM-oriented CBCL/6-18 scales in 796 twins aged 8-17 years belonging to the general population-based Italian Twin Registry. When children were analysed together regardless of age, most best-fitting solutions yielded genetic and non-shared environmental factors as the sole influences for DSM-oriented CBCL/6-18 behaviours, both for the normal and the extreme variations. When analyses were conducted separately for two age groups, shared environmental influences emerged consistently for Affective and Anxiety Problems in children aged 8-11. Oppositional-Defiant, Attention Deficit/Hyperactivity, and Conduct Problems appeared-with few exceptions-influenced only by genetic and non-shared environmental factors in both age groups, according to all three computational approaches. The De Fries-Fulker method appeared to be more sensitive in detecting shared environmental effects. Analysing the same set of data with different analytic approaches leads to better-balanced views on the aetiology of psychopathological behaviours in the developmental years.

Tavano Alessandro, Borgatti Renato (2010); EVIDENCE FOR A LINK AMONG COGNITION, LANGUAGE AND EMOTION IN CEREBELLAR MALFORMATIONS; Cortex, 46(7):907-918

I.F. 2009: 4,058

We compared the neurobehavioral profiles of children with Joubert syndrome (JS participants), a rare autosomal recessive condition characterized on magnetic resonance imaging (MRI) by hypoplasia of the cerebellar vermis and midbrain-hindbrain malformations, and children with malformations confined to the cerebellar vermis and one or both hemispheres (Cerebellar malformations--CM participants). We aimed at investigating the influence of anatomo-clinical similarities (vermian malformation) and differences (intact cerebellar hemispheres vs sparing of the pons, respectively) with respect to cognitive, linguistic and emotional development, assuming as a reference framework the Cerebellar Cognitive Affective Syndrome (CCAS). Results show that severe to moderate mental retardation is infrequent in JS children, while it is present in more than half the sample of CM children. Affect development was generally preserved in JS, in high-functioning CM individuals and also in some of the CM children with moderate mental retardation, which raised questions as to the role of a cerebellar vermis lesion in determining affect disorders. Further, cognitive and linguistic profiles on both intellectual

and neuropsychological evaluations provided evidence for distinct patterns of peaks and valleys in the two groups, with JS children being significantly more impaired in language and verbal working memory and CM individuals showing a significant impairment of executive functions and emotional development. The overall evidence provides support for an important role of cerebellar structures per se in shaping emotional, cognitive and linguistic development, when vermian lesions are associated to cerebellar hemispheric lesions. Cerebellar vermis and brainstem lesions instead appear to have a major impact on motor-related skills, including oro-motor abilities and verbal working memory.

Tavano Alessandro, Gagliardi Chiara, Martelli Sara, Borgatti Renato (2010); NEUROLOGICAL SOFT SIGNS FEATURE A DOUBLE DISSOCIATION WITHIN THE LANGUAGE SYSTEM IN WILLIAMS SYNDROME; Neuropsychologia, 48(11):3298-3304

I.F. 2009: 4,345

The neurocognitive profile of Williams-Beuren syndrome (WBS) is characterized by visuospatial deficits, apparently fluent language, motor soft signs, and hypersociability. We investigated the association between neuromotor soft signs and visuospatial, executive-attentive, mnestic and linguistic functions in a group of 26 children and young adults with WBS. We hypothesized that neurological soft signs could be an index of subtle neurofunctional deficits and thus provide a behavioural window into the processes underlying neurocognition in Williams-Beuren syndrome. Dysmetria and dystonic movements were selected as grouping neurological variables, indexing cerebellar and basal ganglia dysfunction, respectively. No detrimental effects on visuospatial/visuoconstructive skills were evident following the presence of either neurological variable. As for language skills, participants with dysmetria showed markedly reduced expressive syntactic and lexico-semantic skills as compared to non-affected individuals, while no difference in chronological age was evident. Participants with dystonic movements showed reduced receptive syntax and increased lexical comprehension skills as compared to non-affected individuals, the age factor being significant. In both instances, the effect size was greater for syntactic measures. We take these novel findings as suggestive of a double dissociation between expressive and receptive skills at sentence level within the WBS linguistic phenotype. The investigation of neuromotor soft signs and neuropsychological functions may provide a key to new non-cortico-centric genotype/phenotype relationships.

Tomasino Barbara, Bellani Marcella, Perlini Cinzia, Rambaldelli Gianluca, Cerini Roberto, Isola Miriam, Balestrieri Matteo, Calì S., Versace Amelia, Pozzi Mucelli Roberto, Gasparini Anna, Tansella Michele, Brambilla Paolo (2010); ALTERED MICROSTRUCTURE INTEGRITY OF THE AMYGDALA IN SCHIZOPHRENIA: A BIMODAL MRI AND DWI STUDY; Psychological Medicine, in press

I.F. 2009: 5,012

BACKGROUND: The amygdala plays a central role in the fronto-limbic network involved in the processing of emotions. Structural and functional abnormalities of the amygdala have recently been found in schizophrenia, although there are still contradictory results about its reduced or preserved volumes. METHOD: In order to address these contradictory findings and to further elucidate the possibly underlying pathophysiological process of the amygdala, we employed structural magnetic resonance imaging (MRI) and diffusion weighted imaging (DWI), exploring amygdalar volume and microstructural changes in 69 patients with schizophrenia and 72 matched healthy subjects, relating these indices to psychopathological measures. RESULTS: Measuring water diffusivity, the apparent diffusion coefficients (ADCs) for the right amygdala were found to be significantly greater in patients with schizophrenia compared with healthy controls, with a trend for abnormally reduced volumes. Also, significant correlations between mood symptoms and amygdalar volumes were found in schizophrenia. CONCLUSIONS: We therefore provide evidence that schizophrenia is associated with disrupted tissue organization of the right amygdala, despite partially preserved size, which may ultimately lead to abnormal emotional processing in schizophrenia. This result confirms the major role of the amygdala in the pathophysiology of schizophrenia and is discussed with respect to amygdalar structural and functional abnormalities found in patients suffering from this illness.

Tomasino Barbara, Skrap Miran, Rumiati Raffaella Ida (2010); CAUSAL ROLE OF THE SENSORIMOTOR CORTEX IN ACTION SIMULATION: NEUROPSYCHOLOGICAL EVIDENCE; Journal of Cognitive Neuroscience, in press

I.F. 2009: 5,382

Interest in sensorimotor cortex involvement in higher cognitive functions has recently been revived, although whether the cortex actually contributes to the simulation of body part movements has not yet been established. Neurosurgical patients with selective lesions to the hand sensorimotor representation offer a unique opportunity to demonstrate that the sensorimotor cortex plays a causal role in hand action simulations. Patients with damage to hand

representation showed a selective deficit in simulating hand movements compared with object movements (Experiment 1). This deficit extended to objects when the patients imagined moving them with their own hands while maintaining the ability to visualize them rotating in space (Experiment 2). The data provide conclusive evidence for a causal role of the sensorimotor cortex in the continuous update of sensorimotor representations while individuals mentally simulate motor acts.

Tonelli Alessandra, Romaniello Romina, Grasso Rita, Cavallini Anna, Righini Andrea, Bresolin Nereo, Borgatti Renato, Bassi Maria Teresa (2010); NOVEL SPLICE SITE MUTATIONS AND A LARGE INTRAGENIC DELETION IN PLA2G6 ASSOCIATED WITH A SEVERE AND RAPIDLY PROGRESSIVE FORM OF INFANTILE NEUROAXONAL DYSTROPHY; Clinical Genetics, in press

I.F. 2009: 3,304

Infantile neuroaxonal dystrophy, INAD, is a severe progressive psychomotor disorder with infantile onset and characterized by the presence of axonal spheroids throughout the central and peripheral nervous systems. A subset of INAD patients shows also brain iron accumulation which represents instead the distinctive feature of the idiopathic neurodegeneration with brain iron accumulation, NBIA. These diseases share the same causative gene, PLA2G6, encoding iPLA2-VIA, a calcium-independent phospholipase. Mutations that lead to a complete absence of protein are associated with a severe INAD profile, while compound heterozygous mutations with possibly a residual protein activity are instead associated with the less severe NBIA phenotype. Here we describe two INAD patients both with an unusually rapid disease progression and a peculiar neuroradiological presentation in one of them. Compound heterozygosity for a large intragenic deletion and a nonsense mutation was found in one of them while the other is carrying two novel splice-site mutations. Breakpoint-sequence analysis suggests a nonallelic-homologous-recombination (NAHR) event, probably underlying the rearrangement. These findings, while supporting the genotype-phenotype correlation already observed in INAD patients, provide the first sequence characterization of a genomic rearrangement in PLA2G6 gene, thus orienting the search for missing mutant alleles in PLA2G6 related diseases.

Torri Federica, Akelai Anna, Lupoli Sara, Sironi Manuela, Amann-Zalcenstein Daniela, Fumagalli Matteo, Dal Fiume Chiara, Ben-Asher Edna, Kanyas Kyra, Cagliani Rachele, Cozzi Paolo, Trombetti Gabriele, Strik Lievers Luisa, Salvi Erika, Orro Alessandro,

Beckmann Jacques S., Lancet Doron, Kohn Yoav, Milanesi Luciano, Ebstein Richard B., Lerer Bernard, Macciardi Fabio (2010); FINE MAPPING OF AHI1 AS A SCHIZOPHRENIA SUSCEPTIBILITY GENE: FROM ASSOCIATION TO EVOLUTIONARY EVIDENCE; The FASEB Journal, 24(8):3066-3082

I.F. 2009: 6,401

In previous studies, we identified a locus for schizophrenia on 6q23.3 and proposed the Abelson helper integration site 1 (AHI1) as the candidate gene. AHI1 is expressed in the brain and plays a key role in neurodevelopment, is involved in Joubert syndrome, and has been recently associated with autism. The neurodevelopmental role of AHI1 fits with etiological hypotheses of schizophrenia. To definitively confirm our hypothesis, we searched for associations using a dense map of the region. Our strongest findings lay within the AHI1 gene: single-nucleotide polymorphisms rs11154801 and rs7759971 showed significant associations (P=6.23E-06; P=0.84E-06) and haplotypes gave P values in the 10E-8 to 10E-10 range. The second highest significant region maps close to AHI1 and includes the intergenic region between BC040979 and PDE7B (rs2038549 at P=9.70E-06 and rs1475069 at P=6.97E-06), and PDE7B and MAP7. Using a sample of Palestinian Arab families to confirm these findings, we found isolated signals. While these results did not retain their significance after correction for multiple testing, the joint analysis across the 2 samples supports the role of AHI1, despite the presence of heterogeneity. Given the hypothesis of positive selection of schizophrenia genes, we resequenced a 11 kb region within AHI1 in ethnically defined populations and found evidence for a selective sweep. Network analysis indicates 2 haplotype clades, with schizophrenia-susceptibility haplotypes clustering within the major clade. In conclusion, our data support the role of AHI1 as a susceptibility gene for schizophrenia and confirm it has been subjected to positive selection, also shedding light on new possible candidate genes, MAP7 and PDE7B.

Trabacca Antonio, Losito Luciana, De Rinaldis Marta, Gennaro Leonarda (2010); CONGENITAL HYPOTONIA IN A CHILD WITH A DE NOVO 22Q13 MONOSOMY AND 2PTER DUPLICATION: A CLINICAL AND MOLECULAR GENETIC STUDY; Journal of Child Neurology, in press

I.F. 2009: 1,592

The authors describe a 5-year-old girl with a neurological phenotype of 22q13 deletion sindrome (neonatal and persisting hypotonia, developmental delay, absence of language, decreased perception of pain) and minor

dysmophisms. Subtelomeric fluorescent in situ hybridization tests revealed de novo 22q13 monosomy and 2pter duplication. Numerous genetic and neurologic disorders of childhood are characterized by congenital hypotonia. This muscle tone disorder is often one of the symptoms that a neurologist is asked to evaluate. Recent advances in genetic testing can help provide a specific diagnosis for children with this symptom. Subtelomeric deletions are a category of disorders of which hypotonia can be a prominent feature. Deletions of chromosome 22q13 are some of the most commonly observed terminal deletions in humans, whereas duplications of chromosome 2p25.2 are very rare, and little is known about the phenotypic effect of these duplications. To the best of the author's knowledge, this association has never been described before.

Turati Chiara, Montirosso Rosario, Brenna Viola, Ferrara Veronica, Borgatti Renato (2010); A SMILE ANHANCES 3-MONTH-OLDS' RECOGNITION OF AN INDIVIDUAL FACE; Infancy, in press

I.F. 2009: 1,377

Abstract non disponibile.

Urgesi Cosimo, Maieron Marta, Avenanti Alessio, Tidoni Emmanuele, Fabbro Franco, Aglioti Salvatore (2010); SIMULATING THE FUTURE OF ACTIONS IN THE HUMAN CORTICOSPINAL SYSTEM; Cerebral Cortex, in press

I.F. 2009: 6,979

Perception of the final position of a moving object or creature is distorted forward along its actual or implied motion path, thus enabling anticipation of its forthcoming position. In a previous research, we demonstrated that viewing static snapshots that imply body actions activates the human motor system. What remains unknown, however, is whether extrapolation of dynamic information and motor activation are higher for upcoming than past action phases. By using single-pulse transcranial magnetic stimulation, we found that observation of start and middle phases of grasp and flick actions engendered a significantly higher motor facilitation than observing their final postures. Differential motor facilitation during start and end postures was independent of finger configuration at the different hand apertures. Subjective ratings showed that modulation of motor facilitation was not due to the amount of implied motion per se but to the forward direction of the motion path toward upcoming phases. Thus, motor facilitation proved maximal for the snapshots evoking ongoing but incomplete actions. The results provide compelling evidence that the frontal component of the observation-execution

matching system is preferentially activated by the anticipatory simulation of future action phases and thus plays an important role in the predictive coding of others' motor behaviors.

Urgesi Cosimo, Aglioti Salvatore, Skrap Miran, Fabbro Franco (2010); THE SPIRITUAL BRAIN: SELECTIVE CORTICAL LESIONS MODULATE HUMAN SELF TRANSCENDENCE; Neuron, 65(3):309-319

I.F. 2009: 13,260

The predisposition of human beings toward spiritual feeling, thinking, and behaviors is measured by a supposedly stable personality trait called self-transcendence. Although a few neuroimaging studies suggest that neural activation of a large fronto-parieto-temporal network may underpin a variety of spiritual experiences, information on the causative link between such a network and spirituality is lacking. Combining pre- and post-neurosurgery personality assessment with advanced brain-lesion mapping techniques, we found that selective damage to left and right inferior posterior parietal regions induced a specific increase of self-transcendence. Therefore, modifications of neural activity in temporoparietal areas may induce unusually fast modulations of a stable personality trait related to transcendental self-referential awareness. These results hint at the active, crucial role of left and right parietal systems in determining self-transcendence and cast new light on the neurobiological bases of altered spiritual and religious attitudes and behaviors in neurological and mental disorders.

Urgesi Cosimo, Fornasari Livia, De Faccio Sara, Perini Laura, Mattiussi Elisa, Ciano Rossana, Balestrieri Matteo, Fabbro Franco, Brambilla Paolo (2010); BODY SCHEMA AND SELF-REPRESENTATION IN PATIENTS WITH BULIMIA NERVOSA; International Journal of Eating Disorders, in press

I.F. 2009: 2,797

OBJECTIVE: Neuroimaging evidences in eating disorder (ED) patients document dysfunctional neural activity of the posterior parietal cortex, which is engaged in the representation of body schema. Yet a full neuropsychological investigation of body schema representation in ED patients is lacking. We examined mental imagery and body schema representation in patients with bulimia nervosa (BN) and binge eating disorder (BED). METHOD: Consecutive samples of 15 BN patients and 15 BED patients were compared with two groups of 15 age-matched controls in tasks requiring body or object

mental transformation. RESULTS: BN, but not BED patients, were selectively impaired in the mental transformation of their own body, although this deficit was not correlated with measures of body dissatisfaction. In contrast, no patient group was impaired in the mental transformation of external objects. DISCUSSION: Results showed altered self-body representation in BN, but not BED patients, as the neuropsychological consequences of posterior parietal cortex dysfunctions. © 2010 by Wiley Periodicals, Inc. Int J Eat Disord 2010.

Villa Susanna, Micheli Enrico, Villa Laura, Pastore Valentina, Crippa Alessandro, Molteni Massimo (2010); FURTHER EMPIRICAL DATA ON THE PSYCHOEDUCATIONAL PROFILE-REVISED (PEP-R): RELIABILITY AND VALIDATION WITH THE VINELAND ADAPTIVE BEHAVIOR SCALES; Journal of Autism and Developmental Disorders, 40(3):334-341

I.F. 2009: 3,063

The PEP-R (psychoeducational profile revised) is an instrument that has been used in many countries to assess abilities and formulate treatment programs for children with autism and related developmental disorders. To the end to provide further information on the PEP-R's psychometric properties, a large sample (N = 137) of children presenting Autistic Disorder symptoms under the age of 12 years, including low-functioning individuals, was examined. Results yielded data of interest especially in terms of: Cronbach's alpha, interrater reliability, and validation with the Vineland Adaptive Behavior Scales. These findings help complete the instrument's statistical description and augment its usefulness, not only in designing treatment programs for these individuals, but also as an instrument for verifying the efficacy of intervention.

Zanini Sergio, Tavano Alessandro, Fabbro Franco (2010); SPONTANEOUS LANGUAGE PRODUCTION IN BILINGUAL PARKINSON'S DISEASE PATIENTS: EVIDENCE OF GREATER PHONOLOGICAL, MORPHOLOGICAL AND SYNTACTIC IMPAIRMENTS IN NATIVE LANGUAGE; Brain and Language, 113(2):84-89

I.F. 2009: 2,973

Nine early non-demented bilingual (L1 - Friulian, L2 - Italian) patients with Parkinson's disease and nine normal controls matched for age, sex and years of education were studied on a spontaneous language production task. All subjects had acquired L1 from birth in a home environment and L2 at the age of six at school formally. Patients with PD evidenced more phonological,

morphological and syntactic errors in L1 than in L2. The opposite pattern was observed in normal controls as far as grammar was concerned. These findings suggest that implicit language processing is more impaired than explicit language processing in Parkinson's disease. Copyright 2010 Elsevier Inc. All rights reserved.

LETTER TO THE EDITOR PUBBLICATE SU RIVISTE RECENSITE

ANNO 2009

LETTER TO THE EDITOR PUBBLICATE SU RIVISTE RECENSITE Anno 2009

Baiano Monica, Fabbro Franco, Balestrieri Matteo, Pera Valentina, Cremaschi Silvana, Novello Mario, Gon Tiziana, Bonn Renzo, Bertoni Angela, Pascolo Michela, Brambilla Paolo (2009); PRECEDENTI NEUROPSICHIATRICI INFANTILI IN PAZIENTI AFFERENTI AI CSM DI UDINE E DELL'ALTO FRIULI (CHILD PREMORBID IMPAIRMENTS IN ADULT PSYCHIATRIC PATIENTS ATTENDING CENTRES FOR MENTAL HEALTH OF UDINE AND ALTO FRIULI, ITALY); Epidemiologia e Psichiatria Sociale, 18(1):59-64 – Letter to the Editor

Abstract non disponibile

Borgatti Renato, Marelli Susan, Bernardini Laura, Novelli Antonio, Cavallini Anna, Tonelli Alessandra, Bassi Maria Teresa, Dallapiccola Bruno (2009); BILATERAL FRONTOPARIETAL POLYMICROGYRIA (BFPP) SYNDROME SECONDARY TO A 16Q12.1-Q21 CHROMOSOME DELETION INVOLVING GPR56 GENE; Clinical Genetics, 76(6):573-576 – Letter to the Editor

I.F. 2008: 3,206

Abstract non disponibile

Gelosa Giorgio, Di Francesco Jacopo, Tremolizzo Lucio, Lanzani F., Rota S., Colombo M., Perego P., Massimini D., Marzorati L., Curtò N.A., Ferrarese Carlo (2009); AUTOIMMUNE ENCEPHALOPATHY IN GRAVES' DISEASE: REMISSION AFTER TOTAL THYROIDECTOMY; Journal of Neurology, Neurosurgery and Psychiatry, 80(6):698-699 – Letter to the Editor

I.F. 2008: 4,622

Abstract non disponibile

Prigione Alessandro, Isaias Ioannis U., Galbussera Alessio, Brighina Laura, Begni Barbara, Pezzoli Gianni, Ferrarese Carlo (2009); INCREASED OXIDATIVE STRESS IN LYMPHOCYTES FROM UNTREATED PARKINSON'S DISEASE PATIENTS; Parkinsonism and Related Disorders, 15(4):327-328 – Letter to the Editor

I.F. 2008: 1,907

Abstract non disponibile

Tonelli Alessandra, Lanfranconi Silvia, Bersano Anna, Corti Stefania, Bassi Maria Teresa, Bresolin Nereo (2009); ABERRANT SPLICING DUE TO A SILENT NUCLEOTIDE CHANGE IN CCM2 GENE IN A FAMILY WITH CEREBRAL CAVERNOUS MALFORMATION; Clinical Genetics, 75:494-497 – Letter to the Editor

I.F. 2008: 3,206

Abstract non disponibile

LETTER TO THE EDITOR PUBBLICATE SU RIVISTE RECENSITE

ANNO 2010

LETTER TO THE EDITOR PUBBLICATE SU RIVISTE RECENSITE Anno 2010

Fumagalli Matteo, Cagliani Rachele, Pozzoli Uberto, Sironi Manuela (2010); RESPONSE TO WILSON ET AL.; American Journal of Human Genetics, 86(3):493-495 – Letter to the Editor

I.F. 2008: 12,303

Abstract non disponibile

Gagliardi Chiara, Maghini Cristina, Germiniasi Chiara, Stefanoni Giuseppe, Molteni Francesca, Burt Michael D., Turconi Anna Carla (2010); THE EFFECT OF FREQUENCY OF CEREBRAL PALSY TREATMENT: A MATCHED-PAIR PILOR STUDY - AUTHOR REPLY; Pediatric Neurology, 42(5):382 – Letter to the Editor

I.F. 2009: 1,497

Abstract non disponibile

LAVORI PER ESTESO PUBBLICATI SU RIVISTE NON RECENSITE

ANNO 2009

LAVORI PER ESTESO PUBBLICATI SU RIVISTE NON RECENSITE Anno 2009

Barone Lavinia, Frigerio Alessandra (2009); QUALITÀ DELLA DISORGANIZZAZIONE NELLE RAPPRESENTAZIONI MENTALI D'ATTACCAMENTO DELLE MADRI: UNO STUDIO PILOTA NELL'AMBITO DEL MALTRATTAMENTO; Maltrattamento e Abuso all'Infanzia, 11(3):39-50

Obiettivo: lo studio prende in considerazione l'analisi delle rappresentazioni mentali d'attaccamento disorganizzato quale strumento per indagare i fattori di rischio della funzione genitoriale in un gruppo di madri "maltrattanti". Metodo: la Adult Attachment Interview è stata somministrata a due campioni di madri appaiati per età e livello socioculturale (N = 10 gruppo sperimentale, N = 10 gruppo di controllo) e i protocolli dell'intervista sono stati codificati usando due metodi complementari per l'analisi della mancata risoluzione dell'esperienza traumatica: il metodo tradizionale di Main e colleghi, e il più recente metodo di Lyons-Ruth e colleghi. Risultati e conclusioni: i dati preliminari indicano una presenza significativa di rappresentazioni mentali di attaccamento disorganizzato in base al nuovo sistema di classificazione ostile-impotente. In particolare, il sottotipo ostile prevale sugli altri indicatori di disorganizzazione evidenziando potenziali implicazioni legate all'esercizio della funzione genitoriale di tipo abusivo.

Bearden Carrie E., Thompson Paul M., Avedissian Christina, Klunder Andrea D., Nicoletti Mark A., Dierschke Nicole, Brambilla Paolo, Soares Jair C. (2009); ALTERED HIPPOCAMPAL MORPHOLOGY IN UNMEDICATED PATIENTS WITH MAJOR DEPRESSIVE ILLNESS; ASN Neuro, 1(4).pii:e00020

Despite converging evidence that major depressive illness is associated with both memory impairment and hippocampal pathology, findings vary widely across studies and it is not known whether these changes are regionally specific. In the present study we acquired brain MRIs (magnetic resonance images) from 31 unmedicated patients with MDD (major depressive disorder; mean age 39.2i11.9 years; 77% female) and 31 demographically comparable controls. Three-dimensional parametric mesh models were created to examine localized alterations of hippocampal morphology. Although global volumes did not differ between groups, statistical mapping results revealed

that in MDD patients, more severe depressive symptoms were associated with greater left hippocampal atrophy, particularly in CA1 (cornu ammonis 1) subfields and the subiculum. However, previous treatment with atypical antipsychotics was associated with a trend towards lager left hippocampal volume. Our findings suggest effects of illness severity on hippocampal size, as well as a possible effect of past history of atypical antipsychotic treatment, which may reflect prolonged neuroprotective effects. This possibility awaits confirmation in longitudinal studies.

Brambilla Daniele, Csillaghy Annalisa (2009); LA SORDITÀ INFANTILE; ARIS Sanità, 3:29-32

Abstract non disponibile

Cannao Milena (2009); COMUNICARE LA DIAGNOSI DI MINORAZIONE CONGENITA; Child Development & Disabilities - Saggi, XXXV(1):98-111

Vengono prese in esame le reazioni dei genitori alla diagnosi di minorazione congenita del figlio, con specifico riferimento alla minorazione visiva (cecità o ipovisione).
Di tali reazioni, inevitabilmente connotate da grande sofferenza, è necessario tener conto quando si comunica la diagnosi, in modo da aiutare i genitori a superare il trauma e riattivare in loro la fiducia e la speranza. Affinché ciò sia possibile la comunicazione deve essere concepita come tappa iniziale di un processo di aiuto da prolungare per tutto il periodo di degenza ospedaliera della madre e del neonato e anche dopo la dimissione. I protagonisti di questo processo sono sia il medico (al quale spetta il compito dell'informazione clinica) sia tutti gli altri operatori sanitari (che devono fornire un costante sostegno), sia i genitori stessi.
Una diagnosi comunicata con rispetto, partecipazione, solidarietà, competenza, prepara il terreno per la costruzione del legame di attaccamento con il figlio minorato e risparmia ai genitori l'ulteriore sofferenza di non sentirsi compresi e accolti in una delle fasi più drammatiche della loro vita.

Cavallini Anna, Giammari Aldè Giuseppina, Sala Maddalena, Marinoni Cinzia, Pozzoni Elisa, Borgatti Renato (2009); LA RIABILITAZIONE DEL BAMBINO IPOVEDENTE; Child Development & Disabilities - Saggi, XXXV(1):48-66

La riabilitazione-abilitazione della funzione visiva è un insieme di interventi, stimolazioni, esercizi volti a condurre il bambino ad usare il residuo visivo al meglio delle proprie possibilità. L'intervento riabilitativo si propone di adattare reciprocamente l'individuo e l'ambiente per portare il soggetto al maggior grado di autonomia, integrazione e indipendenza per lui possibili. L'intervento riabilitativo deve essere quanto più precoce possibile perché la disabilità visiva, come ogni disabilità in età evolutiva, può interferire con lo sviluppo globale del bambino. Punto di partenza di un corretto intervento riabilitativo è la valutazione clinica a cui segue una approfondita valutazione funzionale sia visiva che neuropsicomotoria. Le modalità dell'intervento variano a seconda dell'età, delle caratteristiche funzionali dell'incompetenza visiva e del quadro clinico complessivo. La presa in carico riabilitativa del bambino affetto da ipovisus comprende sia interventi sull'ambiente che interventi con il bambino. I genitori e gli operatori devono essere consapevoli di come vede il bambino per poter adeguare i propri comportamenti e gli ambienti alle sue potenzialità sensoriali. La scuola deve ricevere tutte le opportune informazioni sulla situazione visiva dell'alunno per poter adottare le modalità didattiche più idonee. La finalità del trattamento con il bambino è quella di favorire una migliore conoscenza della realtà conducendolo ad usare in modo ottimale ed integrato tutti i canali sensoriali, favorire, nei più piccoli, l'acquisizione di adeguate competenze psicomotorie e condurre il bambino verso la conquista dell'autonomia.
L'approccio al bambino ipovedente richiede il lavoro di un team multidisciplinare costituito da diverse figure professionali (oftalmologi, neuropsichiatri, psicologi, terapisti della riabilitazione, pedagogisti, ortottisti) che si articolano nelle diverse fasi del follow-up.

Forti Sara, Valli Angela, Perego Paolo, Nobile Maria, Crippa Alessandro, Molteni Massimo (2009); MOVIMENTO FINALIZZATO E DEFICIT DI PROGRAMMAZIONE MOTORIA NELL'AUTISMO INFANTILE; Psichiatria dell'Infanzia e dell'Adolescenza, 76:55-66

In this study it's been measured the kinematics of upper limb goal-directed movements in children with autism aged 32-54 months, compared with a group of healthy children matched by gender and mental age. We aimed to validate the hypothesis that motor deficits in autism may be linked to an incorrect motor planning. In order to do so, we chose a "reaching and throwing" task in which the correct performance was strictly dependant on motor planning skills, and, in the lack of these, late corrective movements would have been recorded. These adjustment movements were indeed recorded by children with autism in the 90,9% of trials compared to only the 25,6% of trials recorded by healthy children. Moreover, in these movements, children with autism showed: a movement duration twice as long the one of healthy children, a higher number of movement units, and an anticipation of the maxi-

mum movement unit in the movement. As a consequence, children with autism may suffer from a dysfunction in motor planning, and may be capable of storing or putting in place motor schemas of limited capacity, therefore being able the initial phases of the movement only.

Frigerio Alessandra, Ceppi Elisa, Colasanto Michela, Molteni Massimo (2009); ATTACCAMENTO E PROBLEMI EMOTIVO-COMPORTAMENTALI IN UN CAMPIONE INFANTILE DI BAMBINI MALTRATTATI; Child Development & Disabilities – Saggi, XXXV(3):117-133

L'importanza di valutare, sin dalla prima infanzia, l'associazione tra una carente e inadeguata qualità delle cure materne e la manifestazione di quadri psicopatologici è ampiamente riconosciuta, alla luce dell'impatto che il maltrattamento esercita sullo sviluppo sia psichico sia biologico del bambino. Il presente studio ha investigato la presenza di problemi emotivo comportamentali e lo stile di attaccamento in un campione di 28 bambini di un anno circa di età, inseriti insieme alle loro madri in comunità di accoglienza, e in un campione (N = 28) di controllo. Le madri compilavano un questionario che misura i problemi comportamentali e le competenze dei bambini (Infant Toddler Social and Emotional Assessment) e partecipavano insieme ai loro figli alla procedura Strange Situation. I bambini inseriti in comunità, rispetto ai coetanei, sono stati descritti come più problematici rispetto ai comportamenti internalizzanti, meno abili sul fronte delle competenze socio-emotive e si è visto che formavano più frequentemente un attaccamento di tipo disorganizzato. Tuttavia, lo stile di attaccamento non è risultato un fattore capace di moderare l'associazione tra maltrattamento e problemi emotivo-comportamentali. In conclusione, lo studio fornisce un contributo alla comprensione del fenomeno del maltrattamento in un fascia poco investigata, verso cui promuovere tempestivamente interventi mirati a migliorare la qualità della relazione madre-bambino.

Guerreschi Massimo (2009); IL BAMBINO IPOVEDENTE NELLA SCUOLA MATERNA. ASPETTI EDUCATIVI E DIDATTICI; Child Development & Disabilities - Saggi, XXXV(1):67-73

L'ipovisione è una condizione molto più diffusa della cecità, ma è ancora ampiamente misconosciuta, sia in ambito clinico-riabilitativo, sia in quello educativo.
Nell'articolo vengono precisate le relazioni tra intervento riabilitativo ed educativo e poi quelle che riguardano educazione e didattica. Vengono individuate le principali risposte richieste alla scuola dell'infanzia in funzione delle

esigenze specifiche dei bambini con ipovisione.

Liscio Mariarosaria, Adduci Annarita, Galbiati Susanna, Poggi Geraldina, Pastore Valentina, Colombo Katia, Bottini Maria Pia, Castelli Enrico, Strazzer Sandra (2009); BEHAVIOURAL MODIFICATION AND WORK PERFORMANCE: AN EXPERIMENTAL STUDY OF A GROUP OF YOUNG PEOPLE WITH MENTAL RETARDATION IN A GUIDED WORK CENTRE; Advances in Clinical and Cognitive Neurosciences, 1

Although the efficacy of behavioural techniques with mentally disabled subjects has been widely demonstrated, their application in the work adjustment training sector appears to be limited to the context of psychiatry or physical disability. This study postulates the hypothesis that such methods are particularly suitable, alongside traditional work adjustment training, in the professional training of young people with mental retardation.
The objective of this study is to verify the efficacy of behavioural modification on working ability, on the motivation to work and on the personal autonomy of subjects with mild-moderate mental retardation who attend a Guided Work Centre, by identifying the psychological, behavioural and performance changes which occur as a consequence of this treatment.
The study, which involved 20 young people divided into an experimental group and a control group, included an evaluation phase, a treatment phase and a re-evaluation phase.
The results show that applying behavioural methodology within traditional work adjustment training procedures determines an improvement in performance as well as facilitates the internalisation of a different attitude towards work and the acquisition of a greater ability to control onès conduct, thus influencing the work performance positively.

Montirosso Rosario, Del Prete Antonio, Cavallini Anna, Cozzi Patrizia, Gruppo di Studio NEO-ACQUA (2009); PROFILO NEUROCOMPORTAMENTALE IN UN GRUPPO DI BAMBINI PRETERMINE SANI. APPLICAZIONE DELLA NICU NETWORK NEUROBEHAVIORAL SCALE (NNNS); Child Development & Disabilities – Saggi, XXXV(3):96-116

Il presente studio è parte di un progetto di ricerca longitudinale multicentrico, denominato NEO-ACQUA (NEONATAL ADEQUATE CARE for QUALITY of LIFE), il cui principale obiettivo è la valutazione della qualità di vita di bambini nati pretermine, ma considerati "sani" alla dimissione per l'assenza di patologie conclamate. In quest'articolo sono riportati i risultati relativi alla va-

lutazione neurocomportamentale. Lo scopo primario è indagare possibili differenze nel profilo neurologico e comportamentale rispetto a bambini nati a termine. Hanno preso parte allo studio 69 bambini nati molto pretermine (età gestazionale < 30a settimana e/o peso alla nascita < 1500 gr) e 33 bambini nati a termine. I pretermine sono stati valutati al raggiungimento dell'età postmestruale a termine (≥ 37 settimane), i nati a termine tra la seconda e la terza giornata di vita. La valutazione neurocomportamentale è stata eseguita tramite la Neonatal Intensive Care Unit Network Neurobehavioral Scale (NNNS). In confronto ai nati a termine, i pretermine presentavano un maggior numero di riflessi non ottimali e una scarsa qualità del movimento. Sul piano comportamentale manifestavano una minore capacità di attenzione e partecipazione allo scambio con l'ambiente. Inoltre risultavano meno abili nella regolazione del distress. Infine, presentavano marcati livelli di stress. I risultati rilevano che, anche in assenza di documentate complicazioni cliniche, i bambini fortemente pretermine presentano un'alterazione del profilo neurocomportamentale. Queste evidenze sono discusse alla luce del possibile utilizzo della NNS in programmi di intervento precoce a favore dei bambini pretermine e di sostegno ai loro genitori.

Montirosso Rosario, Murray Lynne, Ghezzi Perego Guenda, Brusati Roberto, Morandi Francesco, Borgatti Renato (2009); MODALITÀ INTERATTIVE NELLA RELAZIONE PRECOCE TRA MADRE E BAMBINO AFFETTO DA LABIO-PALATO-SCHISI. STUDIO OSSERVATIVO SU UN CAMPIONE ITALIANO; Child Development & Disabilities – Saggi, XXXV(3):134-152

I bambini affetti da labio-palato-schisi (LPS) possono presentare difficoltà nell'interazione socio-emozionale con la madre. L'obiettivo dello studio è analizzare la qualità degli scambi affettivi in una fase precoce dello sviluppo. Hanno preso parte alla ricerca due gruppi (clinico e di controllo) composti entrambi da 16 diadi madre-bambino. Un'interazione di cinque minuti viso-a-viso è stata video-registrata quando il bambino aveva 2 mesi di vita. I comportamenti e lo stile interattivo della madre e del bambino sono stati codificati mediante il sistema GRS – Global Rating Scales (29). È stato inoltre somministrato il questionario BDI – Beck Depression Inventory – compilato dalle madri per valutare la sintomatologia depressiva. I risultati evidenziano che rispetto ai bambini del gruppo di controllo i bambini con LPS manifestano una ridotta partecipazione allo scambio relazionale con la madre. Le madri del gruppo clinico appaiono meno sensibili rispetto alle madri del gruppo di controllo. Tra i due gruppi di madri non emergono differenze ai punteggi ottenuti al questionario sulla sintomatologia depressiva. Tuttavia, nel corso

dell'interazione con il loro bambino le madri del gruppo clinico manifestavano segni di natura depressiva. Globalmente le interazioni madre-bambino affetto da LPS risultano meno fluide e con un minor numero di scambi comunicativi positivi. In conclusione, la presenza di LPS nel bambino interferisce in modo rilevante sulla qualità dell'interazione precoce madre bambino. Questi risultati suggeriscono l'importanza di pianificare interventi precoci indirizzati a facilitare la relazione tra la madre e il bambino affetto da LPS.

Nobile Maria, Molteni Massimo (2009); GENETICA DELL'AUTISMO: RECENTI PROGRESSI E NUOVE SFIDE; Psichiatria dell'Infanzia e dell'Adolescenza, 76:497-512

Despite the relative short history of genetic work in the area of Autism and Autism Spectrum Disorder (ASD), the road has been characterized by extreme optimistic phase and subsequent bitter disillusion. Despite compelling evidence from twin ad family studies suggesting strong evidence of genetic involvement in the etiology of Autism and ASD, the specific transcripts contributing to these disorders have been quite difficult to characterize. The purpose of this brief review is to critically evaluate recent advances in autism genetics, with attention focused on new techniques and analytic approaches. We address what is known about genetic basis and genetic architecture of ASD, and evaluate common methodologies employed in complex disorders, including linkage, cytogenetic and association studies, with special attention for new analytic approaches, namely genome wide association studies and assessment of microstructural variation (Copy Number Variation). Finally, we consider problematic issues and new challenges for autism genetic research, posed by emerging new technology.

Salati Roberto, Cavallini Anna, Giammari Aldè Giuseppina, Borgatti Renato (2009); ALTERAZIONI DELLA MOTILITÀ OCULARE IN 128 SOGGETTI CON CEREBRAL VISUAL IMPAIRMENT; Child Development & Disabilities - Saggi, XXXV(1):20-32

Gli Autori hanno sottoposto ad accurato studio delle caratteristiche oculomotorie un campione di 128 soggetti con diagnosi di Cerebral Visual Impairment (CVI). È stato applicato un protocollo d'indagine che prevedeva l'analisi di otto parametri dell'oculomozione: lo scanning dell'ambiente, il mantenimento della fissazione, i movimenti saccadici intenzionali, i movimenti di inseguimento lento (smooth pursuit), le deviazioni parossistiche dello sguardo (paroxismal ocular deviations), lo strabismo e il nistagmo. I risultati sono interessanti e dimostrano come la motilità oculare sia pesantemente interessata nel CVI (94% dei casi). Questi danni sono specifici e non consistono sem-

plicemente in uno strabismo o nel nistagmo, ma colpiscono il sistema oculomotorio in alcune delle sue funzioni basilari, come l'esecuzione di saccadi volontarie, l'inseguimento, l'esplorazione visiva. Riteniamo che tali alterazioni non consentano di sfruttare appieno le potenzialità visive e più in generale percettive del soggetto e ostacolino l'uso del canale visivo nelle attività cognitive.

Turconi Anna Carla, Stefanoni Giuseppe (2009); STRUMENTI E AMBITI DELLA MISURAZIONE; Child Development & Disabilities - Saggi, XXXV(2):9-13

Nel capitolo vengono descritte le caratteristiche generali degli strumenti di misura utilizzati in riabilitazione con particolare riferimento a finalità d'uso, livelli di misura e proprietà psicometriche.
In base all'area esplorata le scale di misura sono suddivisibili e raggruppabili in misure di funzione e struttura, di attività e di partecipazione secondo i criteri indicati nella "Classificazione Internazionale del Funzionamento della Salute e della Disabilità" (ICF, 2001).

Turconi Anna Carla, Stefanoni Giuseppe (2009); MISURE DI FUNZIONE E STRUTTURA; Child Development & Disabilities - Saggi, XXXV(2):14-27

Nel presente lavoro vengono descritte misure di funzione e struttura che valutano aspetti specifici della motricità non esclusivi delle PCI, ma riguardanti anche altre turbe dovute a lesione del Sistema Nervoso Centrale. Si tratta di misure relative a forza e lunghezza muscolare, ampiezza del movimento articolare, clono, spasticità e distonia.

Turconi Anna Carla, Stefanoni Giuseppe (2009); MISURE DI ATTIVITÀ; Child Development & Disabilities - Saggi, XXXV(2):28-50

In questo articolo vengono descritte misure di Attività validate espressamente per soggetti affetti da Paralisi Cerebrale Infantile.
Attraverso questi strumenti di misura vengono quantificate e valutate competenze posturo-cinetiche generali e attività specifiche come la presa, la manipolazione e il cammino.

Turconi Anna Carla, Stefanoni Giuseppe (2009); MISURA DI PARTECIPAZIONE; Child Development & Disabilities - Saggi, XXXV(2):51-65

Nel presente lavoro vengono descritte le scale di misura della partecipazione che documentano l'autonomia e il grado di indipendenza funzionale del

soggetto.
Sono inoltre presentati alcuni strumenti di misura di soddisfazione e qualità della vita del soggetto e della sua famiglia.

LAVORI PER ESTESO PUBBLICATI SU RIVISTE NON RECENSITE

ANNO 2010

LAVORI PER ESTESO PUBBLICATI SU RIVISTE NON RECENSITE Anno 2010

Betto Silvana, Pasqualotti Sabrina (2010); ICF-CY: ELEMENTO RILEVANTE DEL PROGETTO RIABILITATIVO; Il Fisioterapista, 16(2):31-41

La complessità di bisogni del bambino con pluridisabilità richiede un intervento multidisciplinare e quindi il coinvolgimento di specializzazioni cliniche diverse. La centralità dei bisogni dell'individuo attiene alla responsabilità dell'équipe che lo ha in carico, al fine di definire l'intervento riabilitativo più appropriato. Nell'ambito delle patologie neuromotorie complesse dell'età evolutiva vi è la necessità di avere una visione d'insieme del soggetto e nello stesso tempo di identificare gli ambiti specifici dell'intervento riabilitativo. È inoltre fondamentale organizzare il processo riabilitativo nelle sue diverse componenti: individuazione dei problemi e delle risorse, definizione degli obiettivi riabilitativi, scelta delle procedure terapeutiche e degli strumenti di verifica. Come guida alla definizione dell'intervento riabilitativo l'utilizzo dell'ICF permette di avere una visione di insieme delle problematiche del paziente e di codificare i diversi elementi che compongono valutazioni complesse, quali quelle utilizzate nelle discipline cliniche-riabilitative.

Bonaglia Maria Clara, Giorda Roberto, Ciccone Roberto, Zuffardi Orsetta (2010); CHROMOSOME 22Q13 REARRANGEMENTS CAUSING GLOBAL DEVELOPMENTAL DELAY AND AUTISTIC SPECTRUM DISORDER; Monographs in Human Genetics, 18(Chapter 12):137-150

The constitutional deletion of 22q13 is an example of a new microdeletion syndrome, known as the 22q13.3 deletion syndrome, telomeric 22q13 monosomy syndrome, or Phelan-McDermid syndrome (OMIM #606232). It was identified by the detection of a cytogenetic rearrangement in advance of its clinical definition. The clinical definition came following the introduction of new methods aimed at checking telomere integrity which confirmed the previously observed phenotype consisting of global developmental delay, generalized hypotonia, absent or delayed speech, and normal to advanced growth. Since then, continued improvements in molecular cytogenetic techniques have increased the diagnostic yield of 22q13 deletions further leading to improved definition of the clinical phenotype, although the incidence of the

syndrome has still to be estimated. The deletion occurs with equal frequency in males and females and it has been reported in mosaic and non-mosaic forms; it may occur de novo or be inherited and be associated with ring chromosome 22 and in rare cases with proximal inverted duplications. Rare terminal duplications of 22q13 have also been found. Haploinsufficiency of the SHANK3/ProSAP2 gene, less than 200 kb proximal to the chromosome 22q telomere, is very likely the cause of the major neurological features associated with 22q13 deletion, since the gene is always found disrupted or deleted in patients with the syndrome and a recurrent breakpoint within SHANK3, mediated by a repeated non-B DNA-forming sequence, has been identified in several patients. Owing to its emerging role in neuropsychiatric disorders and the overlap of phenotypes between autism and 22q13.3 deletion syndrome, SHANK3 became eligible for mutation screening in patients with autistic spectrum disorders (ASD) and several studies have discovered de novo mutations in such patients. In this chapter, we review the current knowledge regarding pathological copy number variants of 22q13.3 in developmental delay and ASD. Copyright © 2010 S. Karger AG, Basel

Brenna Viola, Ferrara Veronica, Proietti Valentina, Montirosso Rosario, Turati Chiara (2010); L'EFFETTO DI UN'ESPRESSIONE FELICE O DI PAURA SULL'ABILITÀ DI RICONOSCERE UN VOLTO A TRE MESI DI VITA; Giornale Italiano di Psicologia, XXXVII(3):739-745

Il presente studio si propone di indagare la relazione tra il riconoscimento dell'identità di un volto e l'elaborazione di un'espressione emotiva nei primi mesi di vita. Recenti studi hanno dimostrato una reciproca interazione tra questi processi negli adulti (Bate, Haslam, e Hodgson, 2009). Utilizzando il paradigma della familiarizzazione, si è indagato se a 3 mesi di vita il riconoscimento dell'identità di un volto sia influenzato dall'espressione emotiva positiva (felicità) o negativa (paura) che il volto esprime. I risultati mostrano che i bambini riconoscono l'identità di un volto che nella fase di familiarizzazione esprimeva felicità, ma non sono in grado di riconoscere un volto che nella fase di familiarizzazione esprimeva paura.
Questi dati permettono di supportare l'ipotesi che, anche nella prima infanzia, l'elaborazione di un'espressione emotiva eserciti un'influenza sui processi di riconoscimento dell'identità di un volto.

COMUNICAZIONI SCIENTIFICHE PUBBLICATE SU RIVISTE RECENSITE

ANNO 2009

COMUNICAZIONI SCIENTIFICHE PUBBLICATE SU RIVISTE RECENSITE

Anno 2009

Adduci Annarita, Poggi Geraldina, Sironi Elena, Massimino Maura, Strazzer Sandra, Clerici Carlo Alfredo, Locatelli Federica, Avantaggiato Paolo, Liscio Mariarosaria (2009); COGNITIVE AND EMOTIONAL ROLE TAKING AND RELATIONAL SKILLS IN CHILDREN SURVIVING BRAIN TUMORS; Abstract P2.6.10 of the IPOS 11th World Congress of Psycho-Oncology, Vienna (Austria), Giugno 2009; Psycho-Oncology, 18(S2):S236

I.F. 2008: 3,150

Bardoni Alessandra, Tavano Alessandro, Galbiati Susanna, Recla Monica, Dominici Chiara, Galbiati Sara, Torri Clerici Valentina, Beretta Elena, Strazzer Sandra (2009); DIFFERENCES IN EARLY SPASTICITY PATTERNS IN CHILDREN AND ADULTS AFTER SEVERE TBI; Abstract 08-3 8th Congress of the European Paediatric Neurology Society, Harrogate, United Kingdom, 30.09-03.10.2009; European Journal of Paediatric Neurology, 13(S1):S20

I.F. 2008: 1,421

Bardoni Alessandra, Avantaggiato Paolo, Zanotta Nicoletta, Epifanio Roberta, Formica Francesca, Poggi Geraldina, Locatelli Federica, Zucca Claudio, Strazzer Sandra (2009); MANAGEMENT OF PROPHYLAXIS FOR POST-TRAUMATIC EPILEPSY IN CHILDREN AND YOUNG PATIENTS; Abstract P145 8th Congress of the European Paediatric Neurology Society, Harrogate, United Kingdom, 30.09-03.10.2009; European Journal of Paediatric Neurology, 13(S1):S65

I.F. 2008: 1,421

Bellù Roberto, Montirosso Rosario, Borgatti Renato, Calciolari Guido, Del Prete Antonio, Agosti M., Cavallo C., Zanini Rinaldo (2009); DEPRESSIVE SCORE AND SUPPORT FROM THE STAFF IN MOTHERS OF PRETERM INFANTS; Abstract 010 of the 50th Annual Meeting of the European Society for Paediatric Research, Hamburg,

Germany, 9-12.10.2009; Acta Pediatrica, 98(S460):5

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Beretta Elena, Cimolin Veronica, Piccinini Luigi, Turconi Anna Carla, Galbiati Sara, Crivellini Marcello, Galli Manuela, Strazzer Sandra (2009); ASSESSMENT OF GAIT RECOVERY IN CHILDREN AFTER TRAUMATIC BRAIN INJURY; Abstract P318 Nineteenth Meeting of the European Neurological Society, Milano, 20-24.06.2009; Journal of Neurology, 256(S2):S112

I.F. 2008: 2,536

Bernardi Paolo, Angelin Alessia, Porcelli Anna Maria, Ghelli Anna, Mariani Elisa, Martinuzzi Andrea, Carelli Valerio, Petronilli Valeria, Rugolo Michela (2009); ROLE OF THE PERMEABILITY TRANSITION PORE IN THE PATHOGENESIS OF COMPLEX I DYSFUNCTION DUE TO mtDNA MUTATIONS; Abstract RS A-49-5 of the International Congress of Physiolocial Sciences (IUPS2009), Kyoto, Japan, 27.07-01.08.2009; The Journal of Physiological Sciences, 59(S1):105

I.F. 2008: 0,973

Bizzozero Laura, Milani Greta, Clementi Emilio, Perrotta Cristiana (2009); TUMOUR BIOLOGY OF MELANOMA: A NOVEL ROLE OF ACID SPHINGOMYELINASE; Abstract 7th International Meeting of the Sphingolipid Club - Leiden, The Netherlands, 14-16.22.2008; Naunyn-Schmiedebergs Archives of Pharmacology, 380(4):361-362

I.F. 2008: 2,830

Bonato Sara, D'Angelo Maria Grazia, Gandossini Sandra, Airoldi Giovanni, Turconi Anna Carla, Bassi Maria Teresa, Bresolin Nereo (2009); CLINICAL AND GENETIC STUDY IN ATAXIA-OCULOMOTOR APRAXIA TYPE 2; Abstract P441 Nineteenth Meeting of the European Neurological Society, Milano, 20-24.06.2009; Journal of Neurology, 256(S2):S155-S156

I.F. 2008: 2,536

Colombo Irene, Servida Maura, Bordoni Andreina, Ciscato Patrizia, Prelle Alessandro, Comi Giacomo Pietro, Bassi Maria Teresa,

Bassotti Alessandra, Bresolin Nereo, Moggio Maurizio, Sciacco Monica (2009); MITOCHONDRIAL, EHLERS-DANLOS AND CADASIL FEATURES IN THE SAME FAMILY: A GENETIC PUZZLE OR A NEW DISEASE ENTITY?; Abstract P02.155 American Academy of Neurology 61st Annual Meeting, Seattle (USA), 25.04-02.05.2009; Neurology, 72(11,S3):A87

I.F. 2008: 7,043

Corti Stefania, Nizzardo Monica, Nardini Martina, Donadoni Chiara, Del Bo Roberto, Papadimitriou Dimitra, Locatelli Federica, Mezzina Nicoletta, Bresolin Nereo, Comi Giacomo Pietro (2009); MOTONEURONS TRANSPLANTATION WITH PHARMACOLOGICAL THERAPY TO PROMOTE AXONAL ELONGATION IMPROVES THE PHENOTYPE OF A MODEL OF SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (SMARD1); Abstract S35.002 American Academy of Neurology 61st Annual Meeting, Seattle (USA), 25.04-02-5.2009; Neurology, 72(11,S3):A276

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D'Angelo Maria Grazia, Romei Marianna, Gandossini Sandra, Bonato Sara, Comi Giacomo Pietro, Colombo Daniele, Turconi Anna Carla, Bresolin Nereo, Aliverti Andrea (2009); CLINICAL EVALUATION OF MUSCULAR DYSTROPHIES: NEW TOOLS FROM BIOENGINEERING; Abstract O70 Nineteenth Meeting of the European Neurological Society, Milano, 20-2.06.2009; Journal of Neurology, 256(S2):S26-S27

I.F. 2008: 2,536

D'Angelo Maria Grazia, Romei Marianna, Gandossini Sandra, Bonato Sara, Colombo Daniele, Marchi Eraldo, Lo Mauro Antonella, Turconi Anna Carla, Comi Giacomo Pietro, Aliverti Andrea, Bresolin Nereo (2009); BIOENGINEERING AND MUSCULAR DYSTROPHY; ABSTRACT M.P.2.04 14th International Congress of the World Muscle Society, Geneva, Switzerland, 09-12.09.2009; Neuromuscular Disorders, 19(8-9):572

I.F. 2008: 2,932

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Leonarda, Petrara Lucia, Trabacca Antonio (2009); OUTCOME AND PROGNOSTIC FACTORS OF EPILEPTIC SPASMS IN CHILDREN WITH CEREBRAL PALSY; Abstract P1323 13th Congress of the European Federation of Neurological Societies (EFNS), Firenze, 12-15.09.2009; European Journal of Neurology, 16(S3):156

I.F. 2008: 2,732

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BREVETTI

ANNO 2009

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Titolare: IRCCS “Eugenio Medea” – Associazione La Nostra Famiglia

Inventori: Cavalleri Matteo, Fiorani Federico, Reni Gianluigi

Titolo: SISTEMA PER SCANSIONI TRIDIMENSIONALI

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Breve descrizione

L’invenzione descrive un sistema per scansioni tridimensionali di soggetti che comprende un dispositivo di proiezione atto a proiettare uno o più fasci di luce verso detto soggetto da scansionare, il fascio essendo atto a generare uno o più profili luminosi sul soggetto quando lo colpisce, un dispositivo di rilevamento, atto a riprodurre il soggetto secondo un segnale videocomposito analogico rappresentativo di una o più righe di rilevamento, il dispositivo di proiezione e il dispositivo di rilevamento essendo reciprocamente posizionati in modo tale che le righe di rilevamento sono orientate trasversalmente rispetto all’uno o più profili luminosi generati, il sistema comprendendo inoltre un’unità di elaborazione atta a ricevere in ingresso il segnale videocomposito analogico ed a ricostruire in uscita un segnale d’uscita rappresentativo di una ricostruzione tridimensionale del soggetto scansionato.