

## CONGRESS REPORT

*Brussels, 20.10.2010*

The IX International Congress of Autism-Europe “A Future for Autism”, held in Catania, Italy, 8-10 October 2010, provided a unique opportunity for updating and sharing knowledge on a wide range of issues concerning Autism, from genetic features and neuro-biological findings to social and cognitive development, from diagnosis and classification to intervention and policies.

Some 1200 delegates, researchers, professionals, parents and self-advocates coming from everywhere in the world, enjoyed this unique opportunity to discuss with the best known scientists in the field of ASD their concerns and hopes for significant advances in terms of knowledge, provisions and proper support to persons with ASD in the world .

18 keynote speakers, 70 selected speakers and 185 selected poster exhibitors and other relevant personalities in the field of EU policies contributed to its success through the high quality and variety of their presentations, providing a comprehensive picture of the substantial gains in knowledge during the last 3 years and their implications for intervention.

Progress in scientific knowledge has come particularly from a greater use of experimental designs, the application of eye-tracking methodology, the use of functional brain imaging, the ‘baby-sibling’ prospective studies.

Greater gains in the fields of prevention and intervention can be expected in the years ahead. A better Future for Autism, persons affected and their families is close.

*The following report, while not exhaustive, summarises the more relevant issues which have been presented at the Congress.*

### **GENETICS FINDING AND BRAIN DEVELOPMENT**

The “autism genome project” (AGP) on > 1000 patients with ASD, confirmed rare pathogenic gene mutations affecting molecules (neuroligins, neurexin and SHANK 3), which are involved in the synaptic cell adhesion, and detected new synaptic genes associated with ASD, suggesting that ASD could be the consequence of an alteration in the homeostasis of the synaptic currents in specific regions of the brain. In general, the genes that have been reported to be associated with ASD are all involved in coding for neuronal organization and neuronal migration. Errors in these genes are likely to be responsible for the dysregulation of brain growth seen in autism in the first few years of life. Neural overgrowth and/or malfunctioning neuronal pruning are reflected in the increase in brain size specifically during preschool years ,

which has been demonstrated by head circumference measurements and by structural brain imaging. This increase seems peculiar to autism.

Though twin and family studies have been consistent in indicating that autism has a very high heritability (circa 90%), it has been proved difficult to find the specific genes responsible for ASD. In addition to the likelihood of genetic heterogeneity and of very small effects of individual genes, the explanation may lie in synergistic effects among genes or in gene-environment correlations and interactions.

## **ENVIRONMENTAL RISK FACTORS FOR ASD**

The raising number of persons diagnosed with an ASD across the world can have several different reasons (enlarged diagnostic criteria, earlier and more extensive diagnosis, trend to make a diagnosis which is likely to ensure more support). While actually the occurrence of an epidemic of Autism has not been proved, the evidence of the multi-factorial nature of autism spectrum disorders (ASD) means that some environmental factors are likely to be implicated in causation.

Historical studies on prenatal exposure to congenital rubella and phenylketonuria, as well as the more recent studies on prenatal exposures to valproic acid, thalidomide and pesticides, and new studies on circulating maternal antibodies, have suggested a risk association with ASD in offspring.

Abnormalities of the immune system in a subset of mothers of children with autism have been found. Circulating antibodies anti-foetal brain cells have been found in a proportion of mothers of children with autism. The inoculation of these human antibodies in pregnant monkeys has been demonstrated to lead to a nonhuman primate model of autism in offspring.

Current findings related to environmental risk factors for autism point to pre-natal exposure to a range of environmental agents. These findings could help to identify biologic diagnostic criteria and early prevention in a sub-population of children at risk of developing ASD. On the contrary, though all the environmental agents potentially enhancing the risk for ASD have not been identified. Maternal immune factors seem to be involved in enhancing the risk for developing autism in the offspring. However, restrictive diets or food supplements administered to children with ASD are unlikely to have some efficacy in curing ASD, as the condition develops in the pre-natal life.

Prospective longitudinal studies of very large samples starting during pregnancy, and including good biological measures, are needed to test the environmental factors exposures which are likely to enhance the risk of developing ASD. The MARBLE study is one of such investigation.

## **MMR AND THIMEROSAL**

Claims have been made that either the measles, mumps, rubella (MMR) vaccine or thimerosal (a mercury preservative used in some vaccines), or both, were responsible for an epidemic of autism. Research findings using a range of strategies have been consistently negative regarding both these possibilities.

## **REGRESSION**

Sibling prospective studies found that 86.4 percent of the infants who later develop autism show a clear decline in social communication after 12 months. This differs from previous reports, based on which the researchers expected to see regression in only a small proportion of the children, and suggests regression occurs in the majority, if not all, children with ASD.

Epidemiological research suggests that regression is much more common in autism than in other developmental disorders. This process is peculiar of ASD and is likely to reflect some neurological process to be investigated in the future.

## **EPILEPSY**

Follow-up studies have shown that autism is associated with epilepsy that is particularly likely to have an onset in adolescence or early adult life. Once again, the unusual age of onset is peculiar to autism, and must have a neuropathological meaning to be investigated.

## **MENTAL RETARDATION**

Family studies have shown no association of ASD with mental retardation in families. Mental retardation is currently recognised to be an expression of the severity of the core deficits of ASD, i.e. a consequence of autism, which is likely to be prevented by addressing social and communication symptoms. In fact, early intensive treatments focusing on the core symptoms of autism have shown to impact intellectual disability in children with ASD.

## **SOCIAL /COGNITIVE FUNCTIONING**

There are well replicated findings on impairments in 'theory of mind', joint attention, imitation and action understanding. Progress has come particularly from a greater use of experimental designs, the application of eye-tracking methodology, the use of functional brain imaging, and the 'baby-sibling' prospective studies.

Studies on mirror neurons have detected some possible mechanism leading to the social deficit in ASD. Mirror neurons become active both when the individual executes a specific motor act and when it observes another individual doing a similar act, but only if the observed motor act is part of a specific motor chain (e.g. grasping-to-eating). This mechanism allows the observer to understand the individual's intention.

Children with autism fail to recognize the why of an action. This impairment, which is reflected in a lower activation of mirror neurons, make children with autism to lack experiential understanding of others, relying in their behavior on external factors.

According to the E-S (Empathizing-Systemizing) theory, autism entails below average empathy alongside intact or even superior systemizing. Empathy is the drive to identify another person's mental state. Systemizing is the drive to analyse or build a system. Psychological theories can give rise to practical interventions, for example, in teaching emotion recognition to improve empathy.

Recent studies suggest 2- and 4-year-old children with ASD to show a restricted scanning pattern in response to novel static faces, which appears to hamper their ability to effectively encode facial identity information. Moreover, attention to key facial features appears to decline from 2 to 4 years, which suggests that face processing, might become more abnormal with age. This deficit has been shown to affect learning. A dissociation between sensitivity to referential cues and emotional cues and a causal relationship between difficulties in face processing and action understanding in this population have been demonstrated.

## **DIAGNOSTIC CRITERIA**

Leo Kanner's observations of the particular deficits in affect and social relations as well as the maintenance of sameness in a small group of children 50 years ago have remained the central components of diagnostic criteria for autism but priorities and additional characteristics have varied over the years.

DSM-V, the new forthcoming classification by APA (American Psychiatry Association) will focus on defining appropriate criteria across developmental, chronological and language levels that are easily interpretable by clinicians and that accurately reflect the similarities within the spectrum while differentiating it from other disorders.

There is a consensus that the distinction between impaired social reciprocity and impaired communication is artificial, although uncertainty then arises on abnormal language features (such as pronominal reversal or delayed echolalia).

The evidence suggests that repetitive stereotyped behaviours are separate; although uncertainty remains on whether there should be a split between 'insistence on sameness' and 'repetitive sensorimotor behaviours'

The concept of Asperger syndrome was useful in indicating that autism can develop in individuals who gain speech at a normal age. Nevertheless, the sub classification of autism spectrum disorders has proved problematic and lacking in adequate empirical validation. The DSM-V working parties have argued that it is time to dispense with all the sub classification and have just one diagnosis of autism spectrum disorder.

There will need to be some means of identifying disintegrative disorders - not because we are sure that they are different but, rather, so that the possibility can be investigated.

Both epidemiological and twin study findings have shown that autism extends well beyond the traditional highly disabling diagnostic concept to include a mixture of social and communicative impairments that are similar to, but milder than, those found in autism. These have been termed 'a broader phenotype'. The phenomenon raises the query of how the broader phenotype becomes transformed into autism.

### **PRODROMAL FEATURES IN INFANCY**

Many studies have shown that the siblings of a child with autism have a much increased risk of developing autism. Awareness that this is so has led to multiple international 'baby sibling' studies in which siblings are studied prospectively from early in life to identify and delineate precursors of autism. Diagnostic markers which differentiate children with ASD from their typically-developing peers during the first 6 months of life have been explored. One path toward possible early diagnosis of autism is to use eye-tracking studies to see and measure social engagement. How often, for example, does an infant look at the face of an interviewer? Some children have abnormalities in visual scanning in normal, social settings. Such studies could reveal vulnerabilities for autism in the first months of life, a year or two before the disease begins to become apparent. They might also help predict the degree of impairment and social disengagement that at-risk children can expect.

### **LACK OF MARKED RESPONSE TO MEDICATION**

Numerous studies have documented that psychotropic medication is of some value for associated problems, but Autism stands out from almost all other psychiatric disorders in showing no marked benefits of medication on core symptoms (such as impaired social reciprocity and social communication). Why? One possible implication is that the basic deficit does not involve neurotransmitters; if not, what does it involve?

### **ADULT FUNCTIONING**

Long-term follow-up studies have shown the varied outcomes of autism, with a surprisingly high proportion of individuals with a performance IQ in the normal range remaining severely impaired in their psychosocial functioning. Though the same dramatic effects of early treatments cannot be expected in adults, the demonstration of the brain plasticity and impact of learning on brain development strengthen the need for providing active support and proper services to all persons with ASD, in order to foster the best individual development and to prevent exclusion and dependency, in a rights-based perspective of ASD and regardless the age and severity of the affected person.

## AGEING AND AUTISM

None of the studies into adult life in ASD, to date, involve individuals much beyond the age of 30 or 40 years. This is despite the fact that the percentage of adults in the general population who are over 60 now outstrips the proportion under 16 for the first time, and this is, in fact, the fastest growing age group.

The challenges of an aging population are slowly being recognized by governments, but the potential problems in relation to people with developmental disorders, such as autism, has hardly even been considered.

## PSYCHOLOGICAL TREATMENTS

Recent efficacy studies suggest that very intense, very early behavioural treatment can lead to 'recovery'.

That such treatment can bring worthwhile benefits is not in doubt.

Whether or not there is complete recovery and whether this recovery maintains in the long term is much less certain.

## EVIDENCE BASED TREATMENTS

Research findings on autism and related conditions have implications for intervention programs. Scientific evidence regarding behavioural, pharmacological, complementary and alternative treatments should inform parents and teachers decisions about treatments,

Scientific knowledge should be applied to intervention programmes which are practically helpful while remaining scientifically based.

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