

Introduzione Prof Garaci

Convegno internazionale “AUTISM FROM RESEARCH TO POLICY”

Roma, Aula Pocchiari, 11-12 ottobre 2010

I am very happy to introduce this international conference entitled “**Autism from research to policy**”. This is in fact the second conference held in ISS on this important health issue. In 2008, the panel of experts on autism and related disorders coordinated by the Ministry of Health recommended that the Istituto Superiore di Sanità should cooperate with the regional health authorities in establishing guidelines for diagnosis and treatment, in planning services organization, and in promoting basic, clinical and epidemiological research on autism and related disorders. On these bases, a multidisciplinary Working Group on Autism was established at the ISS, including basic researchers in neuro and psychobiology, epidemiologists, psychiatrists and pediatricians, and supported by the Data Management and Documentation service of our Institute.

Autism is a pervasive developmental disorder of the brain functions which profoundly impairs social interactions and communication capacities. Autism is a serious life-long disability, and although it was once believed to be rare, it has now been demonstrated that it can affect up to 1% of children, a very high prevalence when compared to other pediatric conditions (such as diabetes, cancer and HIV). Autism must thus be recognized as a major health problem, deserving international efforts and cooperation to promote research, surveillance and advanced health policies.

We are glad to host today some of the major world experts of autism who will illustrate the latest research advances in the most debated aspects of these disorders.

While there is a general agreement that autism prevalence had an impressive increase, even more than twenty fold in the last decade, the interpretation of such phenomenon is still controversial. Recent epidemiological studies found that improvement in diagnostic criteria and increased clinical awareness for these disorders can only partially account for prevalence increase, and many questions, mainly concerning autism etiology, still remain unanswered.

As a matter of fact the causes of autism are unknown. It is a complex disorder that likely has many variants and various etiologies. We know that autism has a strong inherited basis, although the genetics are complex. It is becoming evident that autism may result from multigene interactions or from spontaneous mutations in genes with major effects, but the interaction between genetic and environmental factors is an area requiring much more intensive research. Among the risk factors that have been proposed to contribute to autism etiology are neurotoxins and environmental pollutants, viral and infective agents, and exposure to hormones or maternal antibodies in the intrauterine life. Studies in large epidemiological cohorts, conducted mainly in US, have attempted to clarify the impact of these factors in increasing risk of developing autism in individuals with genetic vulnerability, but more efforts are needed in this direction.

More information of the complex interaction between genes and the environment in autism may come for basic research. One compelling approach to test hypotheses about the many candidate genes for autism and their interaction with environmental factors is to generate analogous mutations in the mouse genome and evaluate the mutant line for phenotypes analogous to the symptoms of autism. Mouse models with construct and face validity provide ideal translational tools for understanding mechanisms and assessing effectiveness of treatments.

One of the major obstacles to understanding the causes of and finding effective treatments for autism is that it has diverse outcomes. Some individuals with autism may have severe developmental delays, but others have normal cognitive capacities. This heterogeneity raises the possibility that there are several types of autism, with a variety of causes. The application of brain imaging techniques to autism has provided important clues, as they have described aberrant trajectories of brain growth during early childhood which may account for the different neurobehavioral phenotypes. This approach may help elucidate what brain regions are implicated in autism and contribute to understand its complex neurobiological bases. In addition, identification of functional and structural brain changes in autistic individuals might be useful for early detection of infants at high risk.

Altogether, through the integrated contribution of basic, clinical and epidemiological research we hope to clarify the causes of autism.

So far, there is no known cure for autism, but there are behavioral treatment and education approaches that may help in reducing some of the symptoms and allowing for greater

independence. But to be more effective, treatment must begin as early as possible. There is the need of new methods for detecting autism well before the appearance of manifest symptoms, and of treatments tailored to very young children.

Together with early behavioral interventions, that may possibly guide brain and behavioral development back toward a “physiological” pathway, the use of pharmacological treatments is debated. Although no medicine has been shown yet to really improve the core difficulties of autism, they can help with some of the very problematic symptoms associated with autism. Benefit must be weighed carefully against side effects of drugs. The possible integration of behavioral and drug treatments represents an important area of future work.

Even today, due to lack of proper diagnosis and services, and of variety of treatments offered to the families not always supported by scientific evidence, many people with autism do not receive adequate care, especially when they become adult.

What is clear is the importance of guidelines giving evidence-based and good practice recommendations on diagnosis, clinical interventions and health service organization, such as the SIGN guidelines. This issue will be also addressed in the second day of the conference, focused on the Italian situation. The ISS is presently involved in preparation of national guidelines on treatment which will be presented in a preliminary form. The problem of translating guidelines into clinic practice will be also discussed. The Round Table at the end of the conference will bring together representatives of the Minister of Health, of Regional health authorities, Scientific Societies involved in Autism (Child and Adolescent Neuropsychiatry, Psychiatry and Pediatrics) and last but not least the Family Associations. We aim to create a stable link among all these entities, to promote policy and practice-level coordination among national and regional administrations, to provide integrated and comprehensive community-based supports and services as to improve the quality of life for people with ASD and their families.

To the foreign and Italian speakers and to all the participants, I wish a profitable conference with the hope it will become a regular appointment.