

Presidenza del Consiglio dei Ministri



**MENTAL DISABILITY IN DEVELOPING AGE AND
AUTISM**

19th of April 2013

General Conclusions

Having completed the analytical examination of the major scientific, bioethical and legal issues related to the "autistic spectrum", the NBC now repropose in a concise and unified way the conclusions reached in the debate, advancing proposals for the further promotion of knowledge of this complex syndrome and improvement of the living conditions of people with autism and their families. Moreover, the phenomenon of the increase in incidence (real or presumed?), probably due to multiple factors, is comprised the improvement of diagnostic capabilities (according to current data until 1:88 or even 1:50, according to some case studies) is an item of data that must be taken into serious consideration.

From the point of view of the contribution of biological and neuroscientific research the most important aspects which seem to emerge are the following:

1. Understanding of the genesis of the autistic disorder has progressed in recent decades, through comparative research between the behaviours of healthy subjects and subjects with autism, observed during the trajectory of child development, adolescence and adulthood.

A number of different factors have been recognised as being involved in determining personal autistic behaviour with obvious repercussions on social behaviour; also the techniques of modern neuroimaging and neuroanatomical investigation [e.g. structural and functional magnetic resonance imaging (MRI and fMRI); PET (Positron Emission Tomography); EEG (electroencephalogram), etc.], applied to both the subject with autism and to the equivalent control subject, have identified various structures that are activated in the course of development in relation to vital and environmental stimuli, at times of a varying degree between normal subjects and subjects with autism.

2. Despite the lack of extensive anatomopathological documentation, research in the last decades has, however, allowed to establish the brain regions and structural brain nuclei most involved in the "autistic spectrum" and to recognise the characteristics of the connections between the different structures (functional circuits), appreciating their efficiency always by means of comparison between autistic and normal subjects. The set of data generally indicates defects in connectivity in the autistic subject, which affects both the systems that govern certain cognitive aspects and affective aspects of the mind.

3. Sufficiently clarified is the epidemiological data, the prevalence of males, frequent family history, the amplitude of personal variations in symptomatology, etc., so that today autism forms part of a "phenotype" present in each human population.

4. The prevalent search for causative factors gives prominence to the genetic factors that regulate early ontogenesis of cerebral personal life: stem cell activity, the function of interneuronal cells, the processes of neuronal migration; those of receptor response to neural stimulation by neurotrophins; the micro-columnar structures which allow for correct function and finally the processes of excitatory and inhibitory specialisation of neurochemical transmission at synapses.

Increasingly consistent research interest has been devoted to the study of the biology of the neuron in autism: features of the axon, myelination, training

and development of dendrites, number and structure of the spine and finally the neurochemistry of synapses, with interesting zonal differentiation.

Even conventional "genetic" research has paid off, despite still being subject to continuous progress in relation to the development of techniques: of the approximately 250 genes considered to be "involved", about thirty have been recognised as having a greater role in the control of altered functions on the neuronal cell level, of synapses or glial structures of connection, even through the analysis of microdeletions, repetitions of triplets and other anomalies, once not suspected as a possible basis of the symptomatologies of autism and those commonly associated with it (mental retardation: RM; epilepsy. states of anxiety, etc.). This research must be pursued by documenting in parallel and with unexceptionable rigour the clinical conditions of the person concerned.

Lastly an interesting investigation has been started into cerebral "zoning" in the expression of genetic functions, of gene transcription and protein metabolism: this study will probably bring about exciting progress in the future.

Finally, benefits are gained - although to a lesser extent to that initially hypothesised - from the symptomatic and behavioural similarities produced experimentally in mice and non-human primates in the context of human autism. This line of research should be pursued for the possible positive effects in the identification of pharmacologically useful molecules for therapeutic purposes.

To conclude on the role of this highly developed area of study, it can be said that - at the stage reached by research - the framework for "essential" autism (i.e. non- symptomatic and forming part of the broader etiologic and diagnostic structure of other diseases), corresponds to the intervention of multiple genetic factors, whose variation from the norm causes different effects on the level of the microstructures of the brain (neurons, synapses, glia, etc.) and related functional consequences (the genesis of abnormal proteins, etc.).

5. There is increasing interest towards consideration of pathogenic factors defined as environmental that act during the course of intrauterine life and with which - be they of maternal origin, or broadly of environmental origin - the baby comes into contact. These factors may act on a designated genetic level and give rise to a cascade of dysfunctional events even some time after birth.

In this regard, not to be forgotten is the close functional correlation between the nervous system (the central one in particular) and the immune system, which the research of the past decades increasingly considers as being "integrated" in the defense and promotion of the "internal medium", contrary to the old theory of isolation provided by the "blood- brain- barrier", a theory which today is no longer sustainable.

Further research in this area must be carried out, even as regards greater accuracy in the collection of clinical-epidemiological factors.

6. In recent years, in parallel with the development of the "functional biology" of the nervous system and investigation into its behaviour in the "autistic spectrum", a more extensive form of neuroscientific reflection has emerged which does not shy away from considering the "classic" issues of the mind, of SELF awareness, of formation of one's own awareness of things as well as that of others even in the mentally disabled during infancy, and the course of pre-adolescent development, which investigates emotion and rationality, responsibility and the sense of good and evil. Laboratory tests, consistent with clinical observation show in the majority of cases, not an

absence of reactions, but rather a slowdown of the genesis and the development of response.

7. Theory of Mind, Theory of mirror neurons, theory of central coordination and finally Theory of the lack of connectivity can be considered as different angles of interpretation of a reduced profile of some (but not all) of the neurological functions chiefly involved in the social behaviour of the person with autism.

From the perspective of bioethics and bio-law, essential recommendations can be summarised as follows:

- to promote the fostering of research on the development of autism spectrum disorders during the cycle of life and on treatments also appropriate to adulthood in a translational research which bridges the gap that exists today between the advancement of knowledge both biological and neuroscientific in the laboratory and "field research", in which it is necessary to standardise the rules of communication and "counterproof" according to the latest criteria of objectivity;

- to guarantee the right to a conscious choice of care, through the request for informed consent to the parents or to the subject, in the case of this being an adult capable of expressing consent, before each treatment - assuming that any treatment can if there is no hypothesis of reliable effectiveness for that reason be a harmful waste of time and energy - the request must provide thorough information on the different treatments available and the related assumptions of effectiveness;

- to guarantee the right to health care through the verification and maintenance of diagnosis, and to ensure continuation of educational-interventions appropriate to the pathology and to age throughout the whole trajectory of life;

- to resort to pharmacological interventions solely in the interests of the person;

- to ensure the right to development of capacity not limited to providing the adolescent and young adult with autism merely a targeted executive capacity, but also providing a suitable cultural preparation according to the ability demonstrated by the individual;

- to not waste the results of possible improvement of executive capacities, obtainable with some of the habilitation techniques, this can be achieved by increasing the chances of employment through economic and social policies aimed at people with disabilities, organising ad hoc mentoring services and reacting to any form of "stigmatisation".

To this end, it seems appropriate to:

- a. encourage the formation of more specialised support teachers of various types and grades of school education;

- b. encourage the creation of social cooperatives that make provision for a percentage of individuals with disabilities;

- c. favour the establishment of day care centres that promote integration and work activities, according to personalised educational-habilitation programs, synergising with psychiatric services;

- d. promote and monitor at the same time the rise of protected communities for adults with severe autism, overseeing the assurances given about the quality of services, the training of personnel, the organisation of the facility, in order to ensure the best quality of life and greatest possible independence for the persons they are intended for.

Lastly, it is right to take care of the families of persons with autism through *ad hoc* social policies, ensuring their welfare and economic support.

The combination of these measures is justified by the consideration of autism, as in general with physical and mental disabilities, not according to a medical classification in the strictest sense, but according to a complex perspective, of a bio-psycho-social nature.

The -bio-psycho-social approach to disability brings - in turn - acceptance of a perspective of bioethics, which can be construed as the *bioethics of care and solidarity*, which takes charge particularly of the disabled for the additional vulnerability that their condition involves. This bioethics examines not so much the occasional problems of the dilemmas between principles - which also exist - but rather "the problem" which is unacceptable of the discrepancy between the affirmation of universally shared principles - found in the Declarations of International Organisations as well as in our legal system - and the lack or the inadequacy of their actually being implemented.

Bioethical reflection leads, therefore, not to argumentation and affirmations as to "which" ethical principles should prevail, but to "recommendations" on best practices for the implementation of principles that are as shared in theory as they are in practice disregarded or incompletely respected. Ultimately, a bioethics that bends from the theoretical level to the analysis of reality, listening to the voices that come from the world of autism, their repeated complaints about the many rights not realised, to call for greater ethical-political commitment for the "care" of persons with autism.

To sum up what has emerged during auditions from the dutiful "consultation" of experts, but also of those working "in the field" and families (especially parents), now universally recognised as "essential" elements in the habilitative process of childhood and early adolescence of the person with autism, is the ascertainable passion and suffering that characterise these people, as well as their determination to continue their educational role.

At the legislative level, there has been a steady improvement over the last few decades of the general and special "framework" in which to view the help offered by the State - even in relation to a favourable evolution in public "opinion" in many European countries, towards affirmation of the dignity of the autistic and the elimination of discrimination and stigmatisation. The awareness of this undeniable increase in protection and sensitivity has also been reaffirmed in the "auditions", accompanied, however, by a deep disappointment regarding the shortcomings in the implementation of various laws.

The text of the Opinion" contains several comments, evaluations and requests (along with the relative proposer) which emerged during the "auditions" which highlight the great local variability in the organisation of care and support still existing today in Italy. Reference is made to the text for the important observations that were collected.

Moreover, all those intervening showed great awareness of their educational role to which they intend to give the best of their ability, calling however, with dignity for the sympathetic support of the civil Community and the State.

Even the Document: "Guidelines for the promotion and improvement of the quality and appropriateness of care interventions in the field of pervasive developmental disorders (PDD), with particular reference to the appearance of autistic disorders" - published November 22, 2012 - is a solemn attestation of the will to proceed, even in the current budget difficulties, with improving, within

the different local realities, the response to the needs of children and adults with autism, and those of their families.

The NBC hopes that even in the current legislature the same attention will be given to the problems of disability. However, it expresses deep concern regarding the risks of excessive regulatory fragmentation. It would be advisable to favour legislation that is unitary by principles, and which leaves to regulatory and/or administrative processes, its adaptation to specific circumstances and needs.