

Opinion N° 102

"On the situation of autistic children and adults in France"

Members of the Working Group:

Jean-Claude Ameisen (rapporteur)
Chantal Deschamps
Claude Kordon
Haïm Korsia
Chantal Lebatard
Philippe Rouvillois

Persons heard:

- Dominique Amy, psychologist, psychoanalyst,
- Michel Favre, President , Association Pro Aid Autisme
- Evelyne Friedel, President, Autisme France
- Bernard Golse, Departmental Head, Pædiatric Psychiatry, Hôpital Necker-Enfants-Malades
- Françoise Grémy, Association Sésame Autisme
- Daniel Kipman, psychiatrist

"Proximity is not a state, it is an action. It is my attitude towards others that brings them close to me."¹

On July 10, 2005, several associations of families with autistic relatives referred to CCNE². The associations voiced strong objections to the concepts currently prevailing in France as regards the care provided for autistic children and individuals, in particular the absence or lack of educational programmes, contrary to current European policy on accompanying measures. They also deplore the attitude of a large portion of French society towards the disabled, the unconcern for the major problems besetting autistic individuals and their families on a daily basis and the absence of any functional and general policy for social integration.

Introduction

Autism is a frequent and major disability which, in its more severe forms, leads to a significant deficit in capacity for social interaction and for verbal and nonverbal communication; behavioural disorders with restricted interests and/or stereotyped behaviour and fear of the unexpected or the unknown. Boys are nearly four times more likely to be affected than girls. Autism can and should be diagnosed at about three years of age, but is frequently suspected earlier.

Today, over sixty years after the first description of autism, plural wordings (autistic spectrum disorders) **or "autistic syndromes" or "pervasive developmental disorders" are considered to be more appropriate descriptions of this impairment.**

In fact, despite the existence of common characteristics, autistic syndromes cover a wide spectrum of traits. At one extreme, individuals with severe impairments who are incapable of any verbal communication and, at the other, adults able to enjoy a certain degree of autonomy, sometimes associated with exceptional intellectual skills (see Annex 1).

Progression of the disorder is also highly variable, reflecting the wide disparity between autistics syndromes, the probable variety of their causes, but also variations in the earliness of diagnosis and treatment and the degree of divergence in educational and medical management. Generally, about half the population of children with autistic disorders suffer from severe verbal or nonverbal communication deficits. Most autistic adults are unable to live autonomously, but some 15 or 20% can manage

¹ Simone Weil. (La proximité n'est pas un état mais une action. C'est mon attitude envers l'autre qui en fait mon prochain).

² Associations Asperger Aide; Autistes Sans Frontières; Fondation Autisme, Agir et Vaincre; Pro Aid Autisme.

independently as long as they benefit from adequate support throughout their lifetime.

Epidemiological studies in Europe, America and Japan reveal an increase in the prevalence of autistic syndromes. The cause is disputed. In France, epidemiological studies are lacking, but the most recent data collected in other countries indicates prevalence ranging from 0.6% to 1%, of which approximately a third is represented by the typical forms of autism. Extrapolating the data to **France, there would be some 350,000 to 600,000 people with autistic syndromes in the broadest acceptance of the term (pervasive developmental disorders) and 5,000 to 8,000 newborns every year would be expected to develop the handicap.**

Currently, diagnosis is based exclusively on a detailed analysis of behaviour and communication skills at about three years of age (see Annex 2). **There is no cure available at this point, but an accumulation of data over forty years demonstrates that an early and appropriate programme of attention and care, based on a case-by-case assessment of educational, behavioural and psychological needs, significantly increases relational and social interaction capacities in autistic children, their degree of autonomy and their capacity to acquire language and nonverbal communication skills.**

The possibility of making a reliable diagnosis at the earliest possible time, so that appropriate care can begin at the earliest age is a pressing and justified demand from families of children with pervasive development disorders.

I. The tragic situation in France of autistic individuals and their families. A long tale of hardship.

Compared to many other countries, particularly in the English-speaking world and northern Europe, France is far behind.

Autistic children and adults and their relatives in France still fall prey to serial misdiagnosis, frequently leading to tardy diagnosis. They experience great difficulty in obtaining early and suitable educational training and in accessing suitable care facilities which are in short supply. Families are not given a choice of the way in which their child's treatment is organised. Support to families is insufficient as are accompanying measures, medical care and social integration of adults and elderly individuals with autistic disorders.

Despite a succession of reports, recommendations, circulars and laws over more than ten years — including CCNE's Opinion n° 47 dated January 10, 1996 *On the management of autistic persons in France*, the December 11, 1996 law modifying the June 30, 1975 law, the Parliamentary Report by Jean-François Chossy in October 2003 on the *Situation of autism in France*, the circular published on March 8, 2005, the recommendations made by ANAES (Agence nationale d'accréditation et d'évaluation en santé/National Agency for Health-related Accreditation and Evaluation, now replaced by the Haute Autorité de Santé/ French National Authority for Health) in 2005 and 2006 on practices to screen for autism, the law 2005-102 dated February 11, 2005 on *Equality of rights and opportunity, participation and citizenship of the disabled*, the opening of Departmental Homes for the Handicapped (*Maisons Départementales des personnes handicapées - MDPH*) and the setting up in 2006 of Autism resource centres (*Centres de Ressources Autisme - CRA*) to guide and assist families, — **the management gap is still considerable and for most families in this situation life is still a tragic struggle.**

A statement by the United Nations, supported by the European Commission (Luxembourg, February 2005) insists on the rights of people with autistic spectrum disorders to adequate medical management, appropriate educational training, assistance respectful of their dignity, absence of discrimination, social integration, protection of their fundamental rights, truthful information on the various forms of treatment available and access to such treatment.

In 2004, the Council of Europe condemned France for not fulfilling its obligations regarding access to education for autistic children, but there was no significant change in the educational opportunities granted to them as a result.

The February 11, 2005 law on *Equality of rights and opportunity, participation and citizenship of the disabled*, made registration with a school mandatory for disabled children. However, despite the creation of special classes (*Classe d'intégration scolaire/CLIS*) in primary schools and special pedagogical integration units (*Unités pédagogiques d'intégration/UIP*) in secondary schools, together with the creation of posts for special education assistants in schools (*Auxiliaires de vie scolaire /AVS*), **registration with a school is still only too often the single expression of this fictitious education due to a lack of teachers and assistants with appropriate training. This is an expression of our country's tendency to consider that symbolic recognition of a right is a good enough substitute for actual access to that right.** In European countries as far apart as Sweden and Italy, the right to education of disabled children has become a reality. As an example, in Italy when a child with communication and intellectual difficulties is registered and included in a class, the number of children in

the class is automatically divided by two and a second specialised education teacher comes to assist the regular teacher.

In a broader context, beyond the specific issue of autism, **this referral calls upon us to face up to a major cultural and social problem in this country:** giving assistance, access to education and social integration to children and adults with a communication impairment, or more generally to the most vulnerable, seems to be a particularly difficult achievement and even the cause of some reluctance.

II. From the "empty fortress" theory to the concept of "pervasive developmental disorder": an endless dispute in France.

A. 1940-1960: a period during which a scientific theory aiming at an understanding of the sufferings of children inflicted more suffering on both parents and children.

The tragedy of autism is a particularly harrowing example of the consequences that theories on the causes of a handicap or a disease can have in terms of human suffering and respect for individuals. The psychoanalytical theories on autism — the psychodynamic theories, including the "Empty Fortress" concept³ — proposed in the 1950s a description and explanation of the inner world of autistic children, and led to incriminating the behaviour of parents, mothers in particular, for their child's autism (see Annex 3). They were described as "refrigerator mothers" or "mortiferous mothers". Blaming the mother for her child's condition, severing the links between mother and child, waiting for a child to express a wish for contact with the therapist (although these children are in fact panic stricken by their environment), are a demonstration of the violence of the attitude, the distress it caused and the impasse to which it led as regards assistance, treatment and social integration.

B. A revolution in the 80s: emergence of the "pervasive developmental disorder" concept.

In the 1970s, a new organic and neurobiological concept of autism emerged, describing it as a "pervasive developmental disorder" which led, in particular in the English-speaking world and in northern European countries, to the development of radically new systems for assistance, social integration, de-institutionalising and early educational, psychological and therapeutic management of autistic children in full cooperation with parents and families. It also drew

³ Bruno Bettelheim. *The Empty Fortress : Infantile Autism and the Birth of the Self*. Free Press, 1967

attention to the distress of families, led to providing them with assistance and thereby to some extent relieving their sufferings.

In the 1980s, the international classification of autistics disorders under the name of "pervasive developmental disorders" **led to dropping the psychodynamic theory of autism and the notion of "autistic psychosis" in almost every country with the exception of France** and certain countries in Latin America where psychoanalytic culture exerts a particularly strong influence on psychiatric practice.

The long-established and marked opposition in this country of most psychiatrists to essentially psychoanalytic approaches **gradually, too rarely**, — at the pressing demand of parents' groups for access to educational and therapeutic methods developed abroad — **began to give way to interesting forms of participation by psychoanalysts in multidisciplinary assistance and management procedures based on educational methods recommended internationally. It can only be hoped that such change will be continued and amplified.**

In any event, **the earliest possible diagnosis is needed so that parents can secure appropriate educational training for their child as quickly as possible and, as a result, improve their understanding of the causes of their child's distress and provide the best possible care.**

III. The inner life of autistic people and their social integration.

- *"This experience of life that life itself experiences as it lives"⁴.*

Poignant books have been written by some autistic people, who upon reaching adulthood, became capable of writing their stories⁵.

"In 1986", wrote the neurologist and author Oliver Sacks, "a quite extraordinary, unprecedented and, in a way, unthinkable book was published⁶. Unprecedented because there had never before been an 'inside narrative' of autism, unthinkable because it had been medical dogma for forty years or more that there *was* no 'inside', no inner life, in the autistic... extraordinary because of its extreme (and strange) directness and clarity. (This) voice came from a place which had never had a voice... and she spoke not only for herself, but for thousands of other...autistic adults..."

These accounts change our attitude to autistic people and pave the way for a less dehumanising view. The situation bears some similarity with the

⁴ Jorge Semprun. *L'écriture ou la vie*. Gallimard, 1994.

⁵ Temple Grandin. *Thinking in pictures : And Other Reports from My Life with Autism*. Doubleday. 1995.

⁶ Temple Grandin, Margaret Scariano. *Emergence: Labeled Autistic*. Arena Press. 1986

reports written by patients with locked-in syndrome who were thought to have no inner life because they could neither move nor speak⁷.

The writings of autistic authors reveal that their extreme difficulty with communication and their repetitive, aggressive or self-injurious behaviours are not an expression of mental retardation or of an impoverished inner life, but a particular and agonising way of existing and of experiencing relationships with others. It is important to remember that educational, psychological and therapeutic assistance primarily allows the personality and the inner life of autistic people to be revealed when they acquire the capacity of entering into contact with others, in a manner which is comprehensible to others — that is to ourselves.

It seems fairly obvious that early and appropriate care, particularly educational training, can have a favourable impact on the progression of the impairment:

- relational, cognitive and behavioural neuroplasticity are all the greater when a child is young;

- in any event and for any child, total or partial absence of appropriate relational stimulation in communication and learning skills can only compromise development.

Children affected by this severe disability and who are deprived of early diagnosis, of access to education and socialisation, who are not given the chance of early and appropriate management of their condition, also suffer a loss of opportunity which is tantamount to maltreatment by neglect.

IV. Advancing research to identify the causes of the autistic disorders.

"[...] metamorphosis, brought about by neurological chance, but metamorphosis into alternative states of being, other forms of life, no less human for being so different⁸

A. Neurobiological research

At the end of the 1970s, a theory on the existence of singularities in the secretions of certain neurotransmitters and later a theory on the existence of particular kinds of connections between different cerebral areas strongly

⁷ Jean-Dominique Bauby *Le Scaphandre et le papillon*. Robert Laffont. 1999. (The Diving Bell and the Butterfly).

⁸ Oliver Sacks. Oliver Sacks. *An anthropologist on Mars*. Vintage Books. 1995 (*Un anthropologue sur Mars*. Editions du Seuil, 1996)

contributed to the notion that autistic syndromes were due to early developmental disorders, with probable onset before birth.

Research in the neurosciences has advanced significantly in the last decade, in particular cerebral imagery, so that many neurobiological characteristics were correlated with autistic spectrum disorders: impaired synchronisation between different cerebral regions, dysfunctions in specific areas of the brain, damaged "theory of mind" ability and "mirror neuron" dysfunction (see Annex 4).

Although these results indicate the existence of very early neurobiological development difficulties in children with autistic spectrum disorders, it is still difficult at this point to determine whether the observed anomalies are some of the actual neurobiological causes of the condition or are the consequences of social interaction problems for which the biological origins, as the brain develops, arise even earlier and are still largely unknown.

In other words, it is hardly surprising to find neurobiological correlates to pronounced behavioural and social interaction variations. However, the existence of these neurobiological correlates is difficult to interpret in terms of causality. The power of evocation of brain imagery — the imagery's "visible evidence" — reinforces the notion that cerebral imagery must reveal the cause of the impairment. But neglecting the possibility that the severe and early modifications of social interaction which are characteristic of autistic disorders could leave traceable imprints on cerebral activity, independently of the biological mechanisms initially involved in the early emergence of brain development problems, would reveal a lack of understanding of the importance of neuroplasticity in response to interaction with the environment.

On the condition that no hasty conclusions are drawn leading to confusion between correlation and causality, **research in the neurosciences, in particular involving younger children, would probably be the best way to arrive finally at a clearer understanding of pervasive developmental disorders and to arrive at new diagnostic and treatment applications. It is essential that such research should be continued.**

B. Genetic research.

Three major categories of arguments (high correlation in identical twins, increased incidence in families with another affected child, association with known Mendelian genetic disorders) suggest a strong possibility that genetic factors play a role in the development of autism spectrum disorders (see Annex 5).

In recent years, the presence of specific genetic sequences involving almost twenty different genes, was correlated with the existence of autistic syndromes. However, most of these studies reported the involvement of different genes in different families.

Most of the work, including the most recent (see Annex 6), suggests the existence of genetic factors contributing to the development of autistic syndromes. But these factors seem to be complex and, in the overwhelming majority of cases, do not seem to be sufficient in themselves to trigger the disability. The most likely hypothesis is that these multiple genetic factors contribute to the appearance, at some point during development, of more vulnerable networks of nerve connections in response to certain modifications in the environment. Some of these genes could be used by nerve cells to manufacture the proteins which play a role, at various levels, in cerebral function, in particular in the construction of synapses, i.e. the connections between nerve cells. This data would suggest that modifications, at various levels, in the development of different connection networks could have the same possible consequence: the development of different forms of autistic syndromes sharing the same common characteristics and producing a continuum between the most severe and the mildest forms of the condition.

At this point, rather than making any contribution to reaching a diagnosis, since there is no indication that it could be significantly improved by genetic testing, **the most valuable aspect of this research is to contribute to an understanding of the mechanisms involved in pervasive developmental disorders, which could in the future, lead to the development of new therapeutic approaches. It is essential that such research should be continued.**

Understandably, ranging from genetic determinism to factors of vulnerability to one or several unknown environmental factors, physiopathological hypotheses abound.

C. Increased prevalence: an increase in the number of affected people or improved diagnosis?

The prevalence of autistic syndromes has increased by a factor of ten in the last twenty-five years. There are several possible explanations for this increased prevalence, and an unknown quantity: is there or is there not increased incidence? In other words, is the increase due to an increase in the number of cases, to changes in diagnostic criteria and/or improved diagnosis because doctors receive better training and parents are paying more attention?

For pervasive developmental disorders taken as a whole, a collective expertise report by INSERM⁹ in 2002 made an inventory of all publications worldwide on this subject. They concluded that the prevalence of pervasive development disorders was 0.2% to 0.3%, but more recent studies show a

⁹ Collective expertise report by INSERM. *Troubles mentaux, dépistage et prévention chez l'enfant et l'adolescent.* (Mental disorders: screening and prevention in children and adolescents.) Les éditions Inserm, pp 9-16, and 525-559, 2002.

prevalence of 1%, of which one third approximately are classical forms of autism¹⁰. The modifications in clinical criteria for the diagnosis of "autism" as a "pervasive developmental disorder" could suffice to explain the considerable increase in prevalence in the last 15 years (see Annex 2). But the increase is perhaps not simply due to a change in classification: recently — between 1999 and 2002 — prevalence has doubled in the State of California. The increased prevalence is thought by some to be a result of improved diagnosis and by others to be due to increased incidence, attributed to environmental risk factors.

D. Ongoing research on environmental risk factors.

Among the many environmental risk factors that have been put forward, some have provoked a great deal of media attention but have turned out to be, so far, devoid of any scientific validity.

Such is the case for the involvement of several factors connected to vaccines (see Annex 7). The implication of the measles, mumps, rubella vaccine is a typical example of public communication campaigns which have frightened and distressed the general public for almost ten years. In 1998, a scientific article reported correlation in children between the development of autistic syndromes and the presence of persistent viral infection due to the attenuated measles virus present in the MMR vaccine. The effects of this article and the ensuing media campaigns were noteworthy: MMR coverage in the United Kingdom, which was 92% in 1995, fell to 82% in 2002-3, leading to several limited outbreaks of measles. All this took place despite denial of the existence of any such correlation in several scientific articles, the initial article being retracted by all its authors except one and no confirmation of the correlation by any other scientific article.

This example, one of many, underlines the need for independent bodies capable of disseminating entirely objective reports so that information which could raise scientifically unfounded fears or hopes may be properly assessed¹¹.

Other environmental risk factors are now being researched (see Annex 8). The United States Congress requested the NIH (National Institutes of Health) in 2006, whose expenditure on autistic syndrome research is currently over 100 million dollars per annum, to increase resources on research in three areas: early diagnosis, therapy and scientific studies to determine if certain environmental factors might or might not be involved.

¹⁰ See for example G Baird *et al.* Prevalence of disorders of the autism spectrum in a population cohort of children in south Thames : the special needs and autism project (SNAP). *Lancet* 368 : 210-215, 2006.

¹¹ INSERM Ethics Committee. *Avis concernant l'annonce de la commercialisation prochaine d'un test génétique de diagnostic précoce de l'autisme*. Inserm Actualités n°199, May 2006. <http://www.inserm-actualites.com/index.php?id=506>

E. From controversy to conflict.

Controversy on the possible causes of autism between parents, autistic support groups and the public health authorities is ongoing in the United States, the United Kingdom and in France.

But these controversies differ radically from one country to another.

In the United Kingdom and the United States, the dispute does not focus on the importance of early diagnosis, nor with access to suitable educational programmes, nor with the need for social integration for the children, nor on whether family participation in the care provided for children is desirable. There is a consensus on all of these issues. Differences of opinion bear on the possibility of prenatal biological effects on the brain (pesticides, foodstuffs, etc.) or postnatal effects (pesticides, foodstuffs, but also vaccines) by an environmental factor. For instance, several thousand parents in the United States are currently claiming for compensation for their child's handicap which they attribute to the adverse effect of a vaccine.

Such controversies are connected to those regarding the significance of the increased prevalence figures: are we observing increased incidence or simply the broadening of diagnostic criteria and improved screening?

In France, controversies between parent support groups and healthcare authorities do not involve so much the possible nature of the neurobiological cause as the persistent debate on whether the cause is neurobiological or psychological. The matter at stake in this debate is the recognition of the importance of early diagnosis of autism, the need for children to access suitable educational programmes — thus providing them with every chance of better social integration — and the active participation of families in caring for their children. Unlike the situation in the United States and in England, legal proceedings instituted by French associations of parents bear not on the possible cause of the impairment, but on the lack of access for their children to educational services. The Autisme-France, Sésame Autisme and Pro Aid Autisme associations, all three members of Autism-Europe, filed a complaint with the Council of Europe against France regarding failure to provide mandatory educational services and obtained a favourable decision in 2004. For the same reason, the Léa pour Samy association, registered a complaint in France against persons unknown.

It is our collective capacity to incorporate a rigorous interpretation of the developments of scientific research into a respectful approach to human dignity, seeking to alleviate suffering rather than create more suffering, which should be our safeguard against the risk of scientific and ethical aberration as exemplified by the sad history of autism. The current paucity of epidemiological studies on the prevalence of autistic syndromes in France, in particular in adults, raises a major ethical problem because this absence of data is a contributing factor in the denial of the need for adequate support and care.

V. An international consensus for appropriate care.

« A handicapped child represents a qualitatively different, unique type of development...This uniqueness transforms the minus of the handicap into the plus of compensation¹²»

A. Appropriate care requires:

- **a reliable diagnosis, as early as possible;**
- **educational, behavioural and psychological support, which must be early, appropriate and individualised.** It must include orthophonic training, in school whenever possible. **Families should be involved to the highest degree possible. Emphasis should be on learning relational and communication skills, autonomy and appropriation of the environment;**
- **among the learning methods, the oldest, the most widespread and the most evaluated, is the TEACCH programme** (Treatment and Education of Autistic and related Communication Handicapped Children). Other widely used methods have or are being evaluated such as the ABA (Applied Behavior Analysis) and the PECS (Picture Exchange Communication System) often used as a complement to TEACCH or ABA. Learning programmes relying on over 40 hours of intensive work a week (this is the case with ABA) could be deemed excessive or hard to bear for certain children, very young children in particular. The learning method must be adjusted to the child's needs;
- **a transitory and personalised course of psychotropic medical treatment in about 20% of cases,** to attenuate aggressive or self-injurious behaviours;
- **psychological and social support for families.**

Independent sources of information, as accessible, open and objective as possible, are essential to help families care for their children.

B. Education, including the learning of verbal and nonverbal communication skills, are essential.

Early and appropriate education is the core need in caring for children and for their future welfare. Suitable methods for learning, communicating and appropriating the environment, focusing on both nonverbal and, whenever possible, on verbal means of communication, must be provided so as to fully develop a child's potential.

¹² LS Vigotsky, quoted by Oliver Sacks, opus cit.

To forgo all attempts at education with the excuse that a child is too severely impaired is a major ethical fault: a denial of the fundamental right of all children to learn to live with and amid others.

C. Integrating children in an appropriate environment is a priority.

Today, families encounter serious difficulties, or even find it impossible to access this much-needed education because their children are rejected by the usual structures and cannot find a vacancy in any specialised small-sized structure. Between the extremes of staying with their family, who are not given any specialised assistance, and hospitalisation, **there has to be some room for educational structures capable of laying down the foundations of a future social environment.** It is also important for so-called "normal" children to heighten their awareness of the need to respect those who are different.

D. Doing away with the model of large-scale institutions.

France has always favoured structures to accommodate large numbers of children and adults with autistic syndromes or other disabilities having an impact on behaviour and communication. The underlying principle is that the considerable amount of care required for their specific needs could only be made available in large-scale collective institutions. This theory is contradicted by recent experience abroad. The exclusion of these children from ordinary society is still a major problem. **Large institutions, generally in a remote location and sometimes very far away from where families may be living,** give society a clear conscience that it is providing appropriate management without disturbing the social peace of the majority, **but in fact they represent isolation and exclusion** for children who, as a result, will experience even greater difficulty later on in life when attempts are made to re-enter society.

E. Participation by, and support for, the family are a priority.

This is essential. Not only should families be spared stigma and blame, they must also be actively helped and supported in their efforts to care for their children. Places where children can be cared for and educated must be made available closer to family homes. **Families must be helped to overcome their own pain and the serious difficulties that beset them.** **The situation of mothers** is frequently aggravated by the fact that they are left alone to care for the child as many fathers are less able than their spouses to cope with disability and some leave home after the birth of an autistic child. The attention and effort that mothers must devote, in sometimes extremely arduous conditions, to their autistic child or children, the disturbed lifestyle of the family because of anxiety, lack of sleep and the absence of calm interludes which are essential to provide respite, to which is added the frequent stigmatisation of the sick child or children, all have a traumatic effect on siblings, on their relationship with schoolmates and on

their success at school. Brothers and sisters may also worry about their affected sibling, or about their own future children, generating anxiety about the future in general.

For these reasons, **support must be given to the whole family, not just parents but also to other children.** Home helpers must be made available to take over for a while so that mothers and families can have an occasional "therapeutic holiday", gain some respite from exhaustion and hopelessness and find renewed energy to go on coping with their lives. Families driven to despair by the community's indifference to their suffering and the fate of their child can only aggravate the condition of the sick person in their midst. Inversely, any help provided to the family has a positive effect on that person.

F. The various forms of education and treatment must be validated by research.

Education is a priority. The choice of educational methods and possible treatment associated with those methods must be given to families. Furthermore, **these methods must be subject to research and evaluation so that families may make a choice based on the most reliable information available.** It should be possible to demand and obtain that the analysis of behaviour and other associated disorders be sufficiently precise to permit at least minimal consensus on the efficacy of the various methods. There is no call for stigmatising psychoactive drugs or praising them to the heavens. In certain very specific circumstances, they should be used for the real alleviation of a child's suffering which they can provide. Psychotropic drugs do not exist to help healthcarers and society, but to help the child. The great diversity of the autistic spectrum disorders also makes it difficult to choose the best therapeutic approach. The "pervasive development disorder" concept has probably multiplied tenfold the diagnosis of various forms of autism and would probably deserve more research and significantly more precise definition so as to adapt and evaluate medical management to the best possible effect.

G. Developing the availability of apartments and small supervised homes: a priority for autistic adults.

"Indeed, in a strange way, most people speak only of autistic children and never of autistic adults, as if the children somehow just vanished from the earth¹³".

Autistic children become adolescents and adults. These changes often give rise to a sudden breakdown in existing care arrangements. Excessively strict age-related criteria force a move into structures which do not differentiate between individual specificities (family income, geographic location, severity of the handicap, etc.) and, **more often than not, families who do not benefit from adequate support are left to bear the burden alone, or else these adult or elderly people are institutionalised in**

¹³ Oliver Sacks, opus cit.

unsuitable structures where they are left entirely without care. The Caisse Nationale de Solidarité (National Solidarity Agency) offers an important alternative by re-focusing the assistance provided by the authorities on the specific needs of a person and his or her family. Such efforts, still in the early stages, should be encouraged. **With this in mind, developing the availability of apartments and small supervised homes, close to the family, is probably the best solution.** The Swedish example should serve as a reference.

VI. Social integration and civic rights.

"A person's humanity is revealed when a relationship is established with another human being"¹⁴.

"Included in Society" is a Swedish example launched in 1995. At the time, **Sweden prohibited and eliminated institutions for people affected by mental or intellectual disabilities or impaired communication and behavioural capacities.** Residential schools for children with these handicaps were replaced by special classes in regular schools. Homes for children and adolescents were closed down. Parents were given personal and financial assistance, the right to free pre-school day-care, given by local experts' groups. In cases where parents cannot care for their child in their own home, the child is taken into a home for four children within the community, open at all times including week ends. In this way, children, youngsters, adults and elderly people with disabilities, including autistic syndromes, are integrated into society.

For adults, residential arrangements were created to allow the disabled to live like normal citizens, including those with very invalidating handicaps. Flats to house a few handicapped adults, assisted by appropriately trained "sponsors" to accompany them on shopping expeditions in their own home town or village or take them to the swimming pool, have progressively accustomed other citizens in the area to their presence. Disabled people have become more independent and there is less discrimination against them. Concerning the economic cost of such a policy, studies in Sweden and the United Kingdom show that providing assistance to permit inclusion in society costs no more to organise than institutionalisation. In this way, contrary to what many people tend to believe, the **non-respect of human dignity of people with autistic syndromes in France cannot be justified by economic misgivings; reluctance to accept change is more cultural than economic in nature.**

In Sweden, the absence of social integration is seen as "mistreatment" and an infringement of civic rights. Instead of considering, as is all too often the case in this country, that **because children and adults have difficulty in achieving social interaction,** the first step must be to exclude them from society and isolate them in institutions before providing any other assistance, in Sweden the approach is that **they should be given access to**

¹⁴ Martin Buber. *Je et Tu*. 1923 (Editions Aubier, 1969)

what they are missing and to what everyone has a right to claim: the possibility of living as full a life as possible with and among others.

Despite considerable efforts in the last ten years or so in France, the situation is as disastrous as ever.

VII. For a calling into question

A. For a calling into question of the divides brought about by dualism.

"Autism is part of who I am"¹⁵.

1. Divides between "innate" and "acquired", "body" and "mind", "biology" and "psychology".

Such divides are always reductive and harmful to autistic people and their families. **The attitude to others is what makes others an object.** An indifferent society's reductive and dehumanising attitude to children or adults with impaired communication and behavioural capacities can only aggravate their pain and their condition so that they close up or become aggressive. Inversely, an open, generous attitude, respectful of the singularity of these children and adults, can create a climate of trust, alleviate their distress and encourage a more evolved social behaviour. Thinking based on purely neurobiological or genetic causes in opposition to the purely psychological, the purely innate standpoint in contrast to the purely acquired, ignore the constant interaction between the environment and the person from which emerges and is modified that person's singularity. Surely, by now we should be aware of the errors generated by ideological, reductive, caricatural and antagonistic visions of the world adopted by extremists in favour of the innate such as Galton and his disciples, or in favour of what is acquired such as Lysenko and his disciples. **Although the psychoanalytical view of the cause of autism has been, and all too frequently, still is in this country a cause of suffering for children and their families, certain neurobiological reductive views on children can also be harmful¹⁶. The problem is not therefore so much whether these theories are scientifically valid, but more that we need to be always aware of the risk of causing suffering instead of alleviating it. Respect for people must be a constant consideration. A person never ceases to be in the process of creation. A person always represents more than just a handicap and is more complex than the supposed cause of that handicap.**

2. The divide between handicap and disease.

¹⁵ Temple Grandin quoted by Oliver Sacks, opus cit.

¹⁶ See CCNE's Opinion n°95. *Ethical Issues Raised by Prediction based on Detection of Early Behavioural Disorders in Children..* 6/2/2007.

In France, diseases and handicaps are viewed as two separate entities which are dealt with by two separate healthcare systems. Two different administrations, operating separately, provide care based on different conceptual prerequisites. Budgets are separated into purely medical and medico-social categories, **as though a stringent borderline could be established** between a handicap and chronic disease, **between needs for education, social integration and medical management.**

3. The divide between biomedical research and research on the best methods to assist and support individuals.

There is an essential need to reinforce the development of research in for example genetics, epigenetics and the neurosciences, so that we can hope to understand the mechanisms causing autistic syndromes, discover new therapeutic strategies and explore the possibility that certain biological signs could orient or disprove their diagnosis early and reliably. But focusing all research exclusively on physiopathological mechanisms could prolong the absence of early and appropriate support and assistance which is, in ethical terms, the most serious problem as regards autistic syndromes in France. For this reason, **it is essential that we develop policies which could lead to an evaluation, in our own country and by comparison with other countries, of the educational and psychological support, the therapeutic management and the social integration of children and adults affected by this handicap, so that further research could improve on the current situation. It is also essential to develop research in the social and human sciences**, so that the support given to families, the attitude of healthcarers and educators and more generally of society as a whole can be evaluated and improved. In other words, although efforts to improve quality must be supported, intensified and be allowed to develop entirely without constraint, **scientific, medical and ethical considerations, focused on those who are sick or disabled, must always prevail when possible applications of such research to autistic people is under evaluation.**

4. The divide between specialised support and social integration.

Here again, not only is there no opposition; there is convergence. Social integration must be the objective. **This being clear, support must adapt to all the constraints of integration, and not the other way around. Support in a closed and isolated environment to the detriment of social integration is pointless.**

B. For calling into question of conducts which heap suffering onto suffering.

We should reject once and for all the *a priori* link between research or discovery of a handicap or a disease and the automatic quest for a "culprit", with as a result, stigmatisation. This is an ethical imperative. Neither the gene, nor ancestry, nor society are the "culprit". There are simply human beings who are suffering and who must be supported and accepted as full

members of our community, who must be approached without formulating groundless accusations or insults, and **whose fundamental rights must be recognised — the right to education, to solidarity, to life within the community — all the more because their extreme vulnerability prevents them from claiming those rights.**

Once and for all, we must refuse to exclude the most vulnerable. We must reject the notion that specialised support for a vulnerable person automatically leads to his or her separation or isolation from the rest of society. **The social integration of a disabled or sick child or adult who has achieved the capacity to communicate should be the very paradigm of the most conclusive assistance given to the particularly vulnerable and of the fight against exclusion.**

C. For a calling into question of the denial of a child's right to education.

Providing care for autistic children, if the central and absolute priority is not education, makes no sense at all. In Belgium, the Minister for Education is responsible for the management of autistic children. Doctors, to the same extent as other specialists, provide a complement to that management. In France, psychiatrists are in charge of the management of autistic children and access to education is often seen as a secondary consideration. Most schools reject the children who have registered with them because of the lack of trained educators, but also because parents often refuse to have an autistic child accepted in the class their own child is attending. In some cases even, petitions were made for rejection. There is also a dearth of special educational support structures. **The symbolic recognition of a right to education considered as a substitute for actual access to that right is in itself a major ethical issue. Education must be the absolute objective of any assistance and any treatment.**

D. For a calling into question of the lack of appropriate vocational training in educational support.

The lack of access to appropriate training for educators — teachers in primary and secondary schools, school and social carers, etc. — whose task it is to support autistic children and adults is one of the major deficiencies in France and one of the principal obstacles in the way of an effective and appropriate social integration and support policy.

E. For a calling into question of the denial of the right of families to free and informed decision.

Free and informed consent has now become one of the cornerstones of medical ethics. But for families with autistic children, rights to the most objective and accessible information on the various educational and

therapeutic approaches and to free choice of educational and therapeutic approaches, are not always respected.

There are several reasons for this state of affairs. The first is that the number of available vacancies in structures "with a mental handicap orientation", of whatever kind (medico-social centres, day hospitals, medico-educational institutions) capable of accommodating autistic children is disastrously inadequate. In 2004, the total number of available vacancies was evaluated at some 7,000 and time-to-admission was 2 to 3 years. The prevalence of autistic syndromes was 0.6% to 1%, so that some 60,000 to 100,000 autistic children aged 3 to 15 live in France, of which probably some 20,000 to 30,000 are severely speech-impaired. These figures suggest that **only a tiny minority of autistic children could find a vacancy in these special structures in 2004. In 2006, the *Plan Autisme 2005-2006* was to create 750 new vacancies for children. The vacancy shortfall therefore cannot possibly be filled since the newly created vacancies would cover only a small proportion of children born every year who will later develop a pervasive developmental disorder.**

The *Centres de Ressources Autismes* (CRA) and the Departmental homes for the handicapped (Maisons départementales des personnes handicapées - MDPH) currently guide families by giving them a list of the centres in their region, but the particulars of management, the delay before gaining admission and criteria for admission are not generally provided and there are no vacancies immediately available. With such restricted access, the exercise of free and informed consent is an illusion. **Acceptance, after a long wait, by a sometimes inappropriate structure, several hundred kilometres away from where the family lives, is simply a take-it-or-leave-it offer**, with no other alternative than care provided entirely by the parent or parents in their own home.

A second reason for the absence of access to free and informed consent is that most of the special centres do not offer any choice. They offer methods corresponding to their own experience which may be based on exclusively psychoanalytic inspired approaches, or on the preponderant use of psychotropic drugs. Frequently, their methods have no educational content. **The desperate shortage of specialised centres and the frequent absence of choice regarding the care provided, in particular the possibility of specialised education, lead *de facto* to pure and simple denial to families of their access to free and informed consent.**

F. For a calling into question of the "exile" policy for autistic children and adults.

Some 3,500 children and adults affected by autistic syndromes and behavioural or mental disorders are currently cared for by medico-pedagogical institutes in Wallonia, Belgium. Care for children is financed by the French medical insurance system and for adults by the French Département they came from¹⁷. These children and adults come from the north of France, but also from the east, the greater Paris area and the

¹⁷ JP Stroubant, C Prieur. La Belgique, refuge des autistes français. (Belgium, a refuge for French autists). *Le Monde* 19 avril 2007.

Touraine and Alpine regions. This would seem to have originated about ten years ago, at a time when the French Ministry of Health directed the national health authorities to enter into agreements with Belgian establishments to compensate for the lack of appropriate structures in France.

The shortfall in France of vacancies in appropriate structures for autistic adults means that in fact children sent to Belgium usually stay there all their lives. **This "exile" or lifelong "banishment" of disabled people from their country**, although it may have seemed at some point to be the least unsatisfactory solution to an urgent problem created by the sad lack of appropriate establishments in France, **would become deeply worrying if it were to persist**. It would mean that our society is not ashamed to be sending an implicit message expressing extreme violence against its most vulnerable members and their loved ones. The message would be: the best way of achieving social integration for children and adults with autistic spectrum disorders is to send them to other European countries, where their right to social integration is recognised.

VIII. The tragic situation in which people with autistic spectrum disorders find themselves is emblematic of a major cultural and human problem in our country.

"If the misery of our poor be caused not by the laws of nature, but by our institutions, great is our sin"¹⁸.

Isolation and exclusion are in themselves the source of suffering and sickness. Yet, we are often inclined to isolate or abandon the most vulnerable among us rather than allow them to be integrated in their own home towns and villages: the elderly whose destitution was brutally revealed by the heat wave; old people arriving in Accident and Emergency and sent off immediately to geriatric hospitals far from their homes, where their relatives find it extremely difficult to visit and comfort them; the mentally handicapped or elderly Alzheimer patients, isolated in institutions or cared for by their exhausted families without any outside help; people suffering from serious mental disorders incarcerated in our prisons¹⁹ or living on the streets²⁰; people with a physical or sensorial disability whose lives are confined by their lack of access to public facilities and transport, lack of schooling and access to the workplace; the two million children living under the poverty threshold in our country, the tens of thousands homeless²¹ people that *Médecins du Monde* and later *Les Enfants de Don Quichotte* suddenly brought out of obscurity by giving them tents in mid winter in our

¹⁸ Charles Darwin. *Journal of Researches into the Geology and Natural History of the Various Countries Visited by HMS Beagle*. 1839 (*The voyage of the Beagle*, National Geographic Society, 2004).

¹⁹ CCNE Opinion n° 94. *Health and Medicine in Prison*. December 13, 2006.

²⁰ Cour des Comptes. *Rapport public thématique sur les personnes sans domicile*. (Public thematic report on the homeless). 6/3/2007

²¹ Cour des Comptes, *ibid*.

city centres; the 500,000 people every year reaching life's end and dying alone, with no one to alleviate their suffering or administer palliative care... In all these apparently unconnected situations, one recurrent and inexorable point in common emerges: the absence of any real support, of any real effort to achieve social integration and the lack of true respect for the human dignity of the most vulnerable among us. Is this a form of indifference or fear expressed by a refusal to see what is before us? Or a form of resignation in the face of suffering that we see as unacceptable but find impossible to assist, to share or to alleviate? There is no shortage of generous response in this country, in the form of asserting rights and demanding their inclusion in legislation, but the essential point is forgotten, that of their inclusion in reality, what Amartya Sen, the economist, called "capabilities"²², since without access there is no such thing as a right. Nor does our country suffer from a lack of generous commitments, of far-reaching initiatives, involving the large-scale mobilisation of extremely diverse resources, but all focused and targeted at a single form of vulnerability: the *Téléthon* for children and adults with a physical disability; the *Plan Canicule* for the elderly in summer heat waves; the successive *Plans Autisme*, over the last ten years, for the 350,000 to 600,000 autistic children and adults; the three *Plans Alzheimer* over the last six years, the latest of which has just been proclaimed, for the 600,000 elderly patients affected by that disease and their families, etc.

However laudable and essential, all these initiatives betray a missing component: a global vision, a global approach and a global will to support the most vulnerable among us and integrate them in the midst of our community.

The Swedish experiment, referred to above, is exemplary in this respect. It began with fostering a cultural change, a global approach, centred on a respect for the fundamental rights and dignity of every individual, of all children and adults suffering from a condition affecting their ability to communicate, be it due to mental, intellectual or autistic syndrome disabilities. Building on this, they were able to organise assistance specific to each disability.

On the other hand, **a multiplicity of specific projects**, each focusing on some particular vulnerability could be reduced in scope by general cultural reticence or could fail to include innumerable similar conditions which do not meet the named and defined criteria.

A meaningful response to the disastrous situation in which autistic people and their families find themselves in France can only be arrived at **by creating the conditions required for a radical change in our behaviour and our thinking as regards support, access to fundamental rights and social integration of the most vulnerable.**

Recommendations

²² Amartya Sen. *Development as Freedom*. Alfred Knopf. 1999.

- 1) Access to **reliable and early diagnosis** of the autistic syndrome is **essential**. It requires training of doctors and intensifying efforts in pædiatrics and pædopsychiatry. There must be an increase in the number of diagnosis centres, currently very inadequate, to shorten the considerable and all too frequent delays before diagnosis in France, compared to other European countries.
- 2) Once a diagnosis is made, the first **priority is giving children early and specialised educational training**, in close cooperation with their families, so that they can appropriate their environment and develop their relational capabilities. The centres for autism resources (*Centres de Ressources Autisme - CRA*) must be designed and organised specifically to respond to this priority. In addition, support and therapeutic options must also be geared to this educational priority. **Schooling must cease to be no more than a fictional registration: it must be provided in a school close to a child's home, or if the child's condition requires it, in an appropriate special-education structure close to home.** These essential educational programmes will require **major efforts to train** educators and school assistants working both inside and outside establishments run by the public education authorities. **Care provided by people who have not received appropriate training is inadequate and is frequently the source of ill-treatment.**
- 3) Support and assistance to families is an essential priority. **The family must be understood to include not only parents but also siblings who are greatly in need of attention.** Unaffected children are frequently the most vulnerable victims of the distress in a family caused by the lack of appropriate management of an autistic child and by the community's stigmatisation of that child. **If a true change of heart is to be achieved, far-reaching action programmes are needed to overcome the reluctance or refusal of parents and teachers to accept integration for children "with a difference". Clearly, a joint effort on the part of all those concerned is of the utmost importance:** parents, support groups, teachers, educators, school and home assistants, volunteer workers, and administrative and political decision makers.
- 4) **Access to the various kinds of educational management**, as well as access to the various associated therapeutic options, **must be the result of a proper process of free and informed decision by families.** In view of the current diversity of ways in which management is organised and the often conflicting "convictions" of the numerous groups exercising in this field, the Committee considers that it is **only through mediation provided by independent** and adequately trained persons that families **can be given** an overview of the various educational methods and the complementary therapeutic options, **so that families are able to exercise a truly free and informed decision.** *The Centres de Ressources Autisme* should encourage a sharing of experience

between various practitioners so as to promote essential cooperation around an educational project between various forms of expertise and disciplines focusing on the needs of children and their families.

- 5) **Specialist structures can only take in a small minority of the 60,000 to 100,000 autistic children. The shortage of vacancies** puts families in a disastrous situation and is the cause of **ill-treatment by default** for almost this entire population of children. **There is an essential need to promote and support all efforts with the purpose of creating really appropriate structures**, of manageable size, on a human scale, close to the family's home, **focusing on educational projects**, where educators, healthcare providers, orthophonists, psychologists, parents and properly trained volunteers can all operate.
- 6) **It is unrealistic to promote integration in regular schools and create new structures for support to children and adults if the professionals providing the education** (educators, primary and secondary school teachers, school and home assistants, etc.) are not given **appropriate training** along the lines prevailing in other European countries. **Unless professionals are given this training, autistic individuals, their families and the professionals themselves are condemned to failure and exhaustion.**
- 7) **Educational methods and associated therapeutic approaches must be the subject of meticulous research and evaluation through comparative scientific investigation** based on previously discussed criteria, so that professionals and families may in future obtain the most reliable information. **Information on evaluation undertaken in other countries must be communicated to families objectively. Developing research is a priority, both in the biomedical sciences and in the human and social sciences. Possible applications of this research for the disabled must always be considered in the context of scientific, medical and ethical concern for the disabled individual.**
- 8) **An autistic child becomes an adolescent, an adult and finally begins to age. Such transitions must not cause a break or put a stop to care;** support, learning, housing, social and professional integration must be adapted to the various stages of a life. **The scarcity of vacancies in appropriate structures is even more dramatic for the 300,000 to 500,000 autistic adults** in this country. **The creation of appropriate residential units, close to families, and the training of carers are essential to bring about social integration.**
- 9) **"Exile" to neighbouring countries must cease to be an alternative to neglect** for autistic individuals and those suffering from other disabilities affecting communication and behaviour.
- 10) Although measures and projects specifically directed at a particular disability or disease are very useful, the Committee considers **that**

it is only through the clear and expressly stated will to modify significantly society's attitude to its most vulnerable members — those whose autonomy and capacity to communicate is impaired by disability or a mental disorder — that they will be given, apart from the specific assistance made necessary by their condition, **the recognition of their right to live with and among others, without stigmatisation or discrimination.** All children have a right to appropriate education and to care, in agreement and close cooperation with their families. All families in distress have a right to solidarity. All vulnerable adults and elderly people have a right to recognition and social integration.

A society which is incapable of recognising the dignity and pain of those who are most vulnerable and most in need, be they children, adolescents, or adults, and which cuts them off from the community, because of that extreme vulnerability, is a society which is losing its humanity.

November 8, 2007

Annex 1

Autistic syndromes are extremely wide-ranging. At one extreme, the disability may be very profound, with a total absence of verbal communication, while at the other extreme, a certain degree of autonomy on reaching adulthood may be associated with outstanding intellectual abilities ("Asperger's syndrome"). In a third of affected children — but not the others — the autistic syndrome leads in adolescence to the onset of epileptic seizures, which are difficult to treat. In 5 to 10% of affected children — but not the others — the autistic syndrome is associated with certain genetic diseases (monogenetic, Mendelian transmission and highly penetrant diseases such as "the fragile X syndrome"). Boys are much more frequently affected than girls, but a rare form of autistic syndrome, of genetic origin, develops almost exclusively in girls (Rett's syndrome). In a majority of cases, onset is early, but in approximately a quarter of cases, children seem to develop entirely normally up to one and a half or two years of age, after which the first signs of the autistic syndrome appear and progressively worsen (syndrome described by the term "disintegrative disorder").

Annex 2

Currently, the autistic syndrome is diagnosed by observation of behaviour, based in most countries on the international classification of diseases, the DSM-IV (Diagnostic and Statistical Manual published by the American Psychiatric Association, 1994) and the ICD-10 (WHO international classification of diseases, 1994).

The criteria for "pervasive developmental disorders", as described in DSM-IV are based on the identification of impairments in social integration, communication difficulties and the existence of stereotyped behaviour. Diagnosis must include six symptoms relating to these three categories with at least two symptoms indicating a social interaction impairment and one symptom in the communication disorder or the stereotyped behaviour categories. Onset of symptoms occurs before the age of three.

Annex 3

The psychoanalytical theories concerning autism had a positive effect at first in that they identified it as a specific disease, so that some autistic people were taken out of the psychiatric "asylums" where they were being held so that they could be treated in specific institutions. But these same theories simultaneously led to stigmatisation and blaming parents who were made to hand over their children into care while they themselves felt rejected and left to fend without help. The theory was that consciously or not, the mother's

behaviour was involved in the emergence of an "autistic psychosis" in the child. The emphasis placed on the mother's responsibility and her stigmatisation led to separating children from their families. Parents, who were held responsible, were excluded from any real communication with their child for long periods of time, or even from communication with psychiatrists who considered they had little to contribute in response to parents' questions. The only important point was the possible expression by the child of a need as a pointer to therapeutic procedure.

Annex 4

Research in the neurosciences has shown that there is a frequent but non specific increase, a short time after birth, of the growth in volume of the brain, involving mainly the white substance (corresponding to the nerve fibres at the brain's surface which participate in the connections between several areas of the brain) associated with an increase in the volume of the cranium. This increase in the volume of white substance suggests the existence of difficulty with the synchronisation of communications between different cerebral areas. Other studies using cerebral imagery techniques suggest the existence of function disorders in certain particular areas of the brain.

Twenty years ago, neuroscience theorists and philosophers proposed the "theory of mind", the idea that understanding the behaviour and intentions of others implies a capacity for each of us to feel them within ourselves, to translate them into the language of our own emotions. From this theory sprang the notion that our social relatedness could depend on our capacity to simulate within ourselves the emotions felt by others and that certain major social interaction impairments, such as those experienced by people with autistic disorders, could be linked to a modification of the mechanisms underlying the "theory of mind". Over fifteen years ago, the discovery in the brain of "mirror" neuron systems, activated both by making a movement oneself and by observing someone else making that same movement, evidenced a biological correlation with the "theory of mind". These "mirror" neuron systems seem to be involved in the internalisation of numerous behaviours besides simple body movements. Five years ago, the hypothesis was put forward that certain "mirror" neuron dysfunctions could perhaps explain the communication impairment in the autistic spectrum disorders. Since then, scientific publications have reported data which fits in with this hypothesis²³.

Annex 5

²³ See for example, M Dapretto *et al.* Understanding emotions in others : mirror neuron dysfunction in children with autism spectrum disorders. *Nature neuroscience* 9 : 28-30, 2006

Arguments in favour of the existence of genetic factors fall into three major categories.

The first is the similarity in the development of autistic syndromes in genetically identical twins, which has been reported to be between 70 and 90%, whereas similarity between fraternal twins — who are not genetically identical — is low. Nevertheless, it must be remembered that although two genetically identical children have a greater probability of developing a disease than two non genetically identical children, this does not necessarily mean that any specific genetic sequences or "autism genes" as such, are the cause of the disease. Certain particular gene sequences which play a role at various critical phases of cerebral development can make these phases more or less sensitive to modifications in the environment during gestation. If twins are genetically identical, the same modification to their environment will be more likely to trigger a similar modification in cerebral development which could contribute to the development of an autistic syndrome.

The second argument suggesting the existence of a hereditary risk factor (but which again does not in itself indicate the genetic nature of this risk factor) is the much higher probability of a child developing an autistic disorder if parents already have a child with this impairment. If heritability is indeed present, it is high compared to the population in general, but still relatively moderate in families with an affected child. It could be linked to particular, rare combinations of frequent genetic sequences, or to particular combinations of rare genetic sequences, but it could also be linked to epigenetic factors, to environmental factors, in particular during embryonic development, or again a particular sensitivity to certain factors in the environment.

The third argument suggesting the presence of genetic factors, is the existence of autistic syndromes in combination with Mendelian-transmission, highly penetrant, genetic diseases, such as the fragile X syndrome, Bourneville's syndrome (tuberous sclerosis), Rett's syndrome, Timothy syndrome and certain disorders caused by deletion or duplication of certain chromosomal areas. But only a small proportion of autistic people are also affected by one of these genetic disease. And all the people who are affected by these genetic diseases do not develop an autistic syndrome. The fact that certain genetic diseases can be associated with an autistic syndrome does not mean that all the autistic syndromes share this genetic origin. Alzheimer's disease is a good example: in 1 to 2% of patients with Alzheimer's, for whom onset of the disease is very early, the disease is linked to particular Mendelian-transmission, highly penetrant, genetic sequences. But in almost the entire population of Alzheimer patients, these genetic sequences are not present.

Taken together, these three sets of data (high correlation in identical twins, greater incidence in families with another affected child, association with known Mendelian genetic diseases) strongly suggest the existence of genetic factors contributing to the development of autistic syndromes.

Annex 6

In 2007, the *Autism Genome Project Consortium* published the most widely-based study ever produced, involving an analysis of the genome of over 1,000 families with at least two autistic members²⁴. The study reveals that in some 10% of cases various familial genetic particularities were associated with the impairment in various families. Another study published in 2007 reported correlation with some genetic non-inherited (emerging spontaneously at conception) characteristics in a certain number of autistic people²⁵.

Some of these genes could be used by nerve cells in the production of proteins with an effect on cerebral function at various levels, in particular in the construction of synapses (the connections between nerve cells). One of the more highly probable theories is that these multiple genetic factors contribute to the creation, during development, of more fragile synapses in response to certain environmental modifications²⁶.

Annex 7

Among other incriminated factors, linked to childhood vaccinations, were the vaccine against pertussis (whooping cough) and thimerosal, an organomercury compound, used as a preservative in vaccines until 2001. This suspicion led to proposing treatment by heavy metal chelators for children with autistic syndromes although the potential risk to health of such a course is high. Over 10,000 children are receiving this treatment in the United States, but no scientific study has evidenced any correlation between thimerosal and the autistic syndrome and several scientific studies, the latest of which was completed in 2007, have invalidated the existence of any such correlation.

Annex 8

Intolerance to gluten and casein in food has also been incriminated and this hypothesis is currently under examination in certain clinical trials. So far, there are no clear results establishing either validation or invalidation. The theory has prompted diets which are sometimes so restrictive that they could be a health hazard.

Organophosphates (pesticides) were also incriminated and scientific research is in progress to validate or invalidate any possible involvement.

²⁴ The Autism Genome Project Consortium. Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics* 39 : 319-328, 2007.

²⁵ J Sebat *et al.* Strong association of de novo copy number mutations with autism. *Science* 316 : 445-449, 2007.

²⁶ AM Persico, T Bourgeron. Searching for ways out of the autism maze: genetic, epigenetic and environmental clues. *Trends in Neurosciences* 29 : 349-358, 2006.

Finally, some data suggests the existence of risk factors connected to the foetal environment, such as the correlation between particularly severe autistic syndromes and the existence of a complication during pregnancy, leading either to very premature birth or to problems during delivery.