M.I.N.D

AUTISM

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Disorder name

AUTISM (AUTISM SPECTRUM DISORDER)

Introduction

Autism spectrum disorder (ASD) refers to a neurodevelopmental condition defined by a number of behavioral features. According to DSM-5, the core clinical characteristics of ASD include impairments in two areas of functioning (social communication and social interaction), as well as restricted, repetitive patterns of behavior, interests or activities. These symptoms are present in the early developmental period, but may not be fully manifest until social demands exceed the child's limited capacities, or may be masked by learned strategies in later life.The treatments now available can achieve a far better quality of life for those with ASD than was the case just a few years ago.

Overview and facts

Autism was once considered a relatively rare condition. Recent epidemiological data have radically altered this perception. Based on large surveys in the US, the Centers for Disease Control and Prevention (CDC), estimates the prevalence of ASD (including all forms) as 1 in 68 children, occurring in all racial, ethnic and socioeconomic groups, although it is five times more common among boys (1 in 42) that girls (1 in 189).

Symptoms

Qualitative impairments in social interaction and social communication:
Of the core symptom domains that define autistic disorder, impairment in social interaction and communication is central. This includes impairment in non-verbal behaviors used to regulate social interactions, failure to develop peer relationships appropriate to the child's developmental level, and lack of spontaneous seeking to share enjoyment, interests or achievements with others (e.g., by a lack of showing, bringing or pointing objects of interest to the attention of others).

Children with impairments in these areas lack social or emotional reciprocity.Responding to joint attention and initiating joint attention is very important in social learning and is associated with language and cognitive development. Impairment in joint attention is a very important early symptom that can be seen even in very young children with autism.

Another important concept is stimulus over-selectivity: children with ASD exhibit overly selective attention to some particular stimuli, not to the gestalt of what is being seen or heard (like making a puzzle not using the image to be built but paying attention only to the shape of the pieces). This is also not unique to ASD and can be seen in children with intellectual disabilities.

Children with ASD use nonverbal behaviors such as eye contact, gestures, body postures and facial expressions less often than typically developing children.

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One of the most important findings in recent years has been the observation that two-year-olds with autism fail to orient towards biological motion – human bodies in motion – and they do not preferentially look to the eyes of approaching adults.

Even high-functioning individuals with ASD have problems in peer relations. Some persons with ASD do not have any interest at all in relating to peers while others may not be able to play in different sides of a game (e.g., seeking and hiding). Some may want to relate to peers but have problems in interpreting other's actions and responding accordingly. Many subjects with ASD do not comprehend the nature of social relations, that is, the intuitive or deductive hidden norms or meanings that govern our relationships and may fail to develop adequate empathy. All of these difficulties lead to impairments in social relations.

In terms of qualitative impairment in communication, symptoms may include:
• Delay in, or a total lack of development of spoken language, which is not accompanied by compensatory attempts
• Marked impairment in the ability to initiate or sustain conversations
• Stereotyped, repetitive or idiosyncratic language; and
• Lack of varied, spontaneous imitative or make-believe play.

Repetition of another person's words, echolalia, is frequent in ASD. The rate, volume and intonation of speech can be abnormally high, low, fast, slow, jerky, monotonous, etc. Individuals with ASD may invent their own words orphrases and language can be repetitive, may repeat the same phrases even when they are inappropriate to the context. Even high-functioning individuals with ASD can have problems initiating and sustaining a conversation. This includes lack of small-talk, not providing enough information, not asking for information and not building on other people's comments. When combined with restricted interests, conversation with persons with ASD can be very difficult to sustain. Phrases and language can be repetitive, may repeat the same phrases even when they are inappropriate to the context.

Individuals with ASD can focus on a very specific part of the object of their interest, for example, only the number of teeth in dinosaurs. They can have problems in switching to other topics even when other people are clearly not interested in what they are talking about. They keep focusing on the topic when they are supposed to do other tasks and may become distressed or even agitated when they are interrupted. They may show less interest in sharing their hobby in social ways, like joining a club.

Inflexible adherence to specific, non-functional routines or rituals is also a typical symptom of ASDs. Difficulties with minor changes in personal routine and resistance to even small changes in the environment can cause significant problems in their and their families' daily lives (e.g., severe tantrums).

Stereotyped and repetitive motor mannerisms and persistent preoccupation with parts of objects is more evident in younger children and individuals with intellectual disability. These include hand and finger flicking, rocking, toe walking, sniffing and licking non-food objects, spinning, and unusual visual gaze, among others. Persistent preoccupation with parts of objects can be seen, for example spinning wheels, flickering the eyes of dolls.

Causes and risk factors

Genetic factors

Evidence for the importance of genetic factors in the etiology of autism comes from many sources, including twin and family studies. Autism is, for example, 50 to 200 times more prevalent in siblings of autistic probands than in the general population. Among probands' relatives who do not have autism, there is also an increased prevalence of milder forms of developmental difficulties related to communication and social skills (broad phenotype). Concordance rates for autism range from 36% to 96% in monozygotic twins but only 0% to 27% in dizygotic twins.

Neuroanatomic and neuroimaging findings

Neuroanatomic and neuroimaging findings, though not diagnostic, have consistently revealed increased cerebral volume that affects both grey and white matter, as well as enlarged ventricles. Neuroimaging findings also include abnormalities in brain chemistry, serotonin synthesis, and brain electrophysiology.

The autism spectrum is now understood to be neurodevelopmental, meaning that there are differences in the pattern of brain development. For example, early brain overgrowth has been documented in the first two years of life and, in later development; there are clear differences in the function and structure of the 'empathy circuit' of the brain.

Risk factors

The NICE (2011) guideline Autism: Recognition, Referral and Diagnosis of Children and Young People on the Autism Spectrum, while stressing the low quality of evidence found, lists the risk factors for ASDs that are clinically and statistically important as:

- A sibling with autism
- Parental history of schizophrenia-like psychosis
- Parental history of affective disorder
- Parental history of another mental or behavioral disorder
- Maternal age older than 40 years
- Paternal age older than 40 years
- Birth weight less than 2500 g
- Prematurity (under 35 weeks)
- Admission to a neonatal intensive care unit
- Presence of birth defects
- Male gender
- Threatened abortion at less than 20 weeks
- Residing in a capital city
- Residing in suburb of a capital city.

In summary, although heritability of autism has been estimated as extremely high, the challenges faced in understanding the etiology of autism lie in the observation that genetic factors are heterogeneous, complex, and the interaction between genes and environment are poorly understood. There are on-going and ambitious individual and familial longitudinal studies that promise to give us useful data in this regard.

Tests and diagnosis

It is acknowledged that early detection constitutes a major advance in that it enables prompt intervention that may improve prognosis in a significant proportion of children with ASD, but also because it clarifies the doubts and anguish of parents and allows adequate planning for future school placements and community support.

It has been known for some time that there is a higher incidence of ASD among siblings of already identified cases; this observation led to a more detailed examination of newborn siblings and follow up during their first years of life. Trying to identify early developmental signs that precede a diagnosis of ASD in siblings that eventually develop the disorder has been a fruitful area of investigation.

Not all children with ASD show all and each of the symptoms all the time.
Screening instruments for ASD

Among the many instruments available, there are currently two that merit special mention since they are free to use, deal with different age groups (one younger children and the other older ones), have undergone cross-cultural adaptation and translation to many languages, and have been researched in various countries.These are the Modified Checklist for Autism in Toddlers (M-CHAT) and the Childhood Autism Spectrum Disorders Test (CAST) (formerly known as Childhood Asperger Syndrome Test).

Treatment

Treatment of ASDs depends on so many factors that it makes description of 'the treatment' difficult. Differences in age, degree of impairment, comorbid disorders, family and social situation, level of resources and community development, provision of education (or lack of it), health and welfare assistance, opportunities for sheltered employment, and availability of inclusive living in the community in adult life will make a huge difference in the outcome and quality of life. If there are three words that summarize what should be done for people with ASDs, they are to personalize, to contextualize and to empower.

Despite accepting these common sense ideas, many families and clinicians search for a cure for ASD, as if there was a single cause, a unique mechanism, and a single condition underlying the syndrome that, if identified, would lead to a cure for all ASDs.

While there is no cure for ASDs, there is strong evidence that appropriate, lifelong educational approaches, support for families and professionals, and provision of high quality community services can dramatically improve the lives of persons with ASD and their families.

The use of psychotropic medication in this population calls for caution and sound knowledge. In summary, medication is currently justified in the management of challenging behaviors (e.g., aggression, self-harm) that do not respond to other approaches and to treat comorbid conditions e.g. ADHD.

The main treatment for ASD is multidisciplinary, combining behavioural approaches, parent training, special education and other supportive services.

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Sources and Links

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