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Review Article

Autism Spectrum Disorder: Myths and Realities

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Abstract

Autism spectrum disorder is part of a neurodevelopment disorder in which in 75% of cases we cannot find an etiology. It is characterized by communication and social interaction difficulties and also by restricted and repetitive behaviors The classification is based mainly in the DSM-5. Clinically the symptoms are variable in young and older children.

We need to try to find an etiology for ASD so we can start the early intervention, mainly speech and occupational therapy and physiotherapy, always with the participation of the family.

Keywords: *Autism Spectrum Disorder; Myths; Realities*

Classification

Identification of autism spectrum disorder (ASD) is of great importance for early intervention.

ASD, according to the old definition (DSM-4), includes autistic disorder (autism), Asperger's Syndrome, childhood disintegrative disorder and pervasive developmental disorder without other specific disorders The new classification - DSM-5 in 2013 completely changed the classification.

The DSM-5 - Diagnostic and Statistical Manual of Mental Disorders - is a document created by the American Psychiatric Association (APA) - 5th Edition From the DSM-5, autism is now called autism spectrum disorder, classified as one of the neurodevelopmental disorders, characterized by communication and social interaction difficulties and also by restricted and repetitive behaviors.

The criteria are divided into A, B, C, D and E with some specific points within them:

A. Persistent deficits in communication and social interaction in various contexts such as:

- Limitation in emotional and social reciprocity, with difficulty sharing interests and establishing a conversation;
- Limitation in non-verbal communication behaviors used for social interaction, varying between poorly integrated verbal and non-verbal communication and with difficulty in the use of gestures and facial expressions;
- Limitations in initiating, maintaining and understanding relationships, with variations in the difficulty of adapting behavior to fit in social situations, sharing imaginary games and lack of interest in peers.

B. Repetitive and restricted patterns of behavior, activities, or interests, as manifested by at least two of the following, or past history:

- Motor movements, use of objects or repetitive and stereotyped speech (stereotypies, aligning toys, rotating objects, echolalia);
- Insistence on the same things, inflexible adherence to ritualized patterns and routines of verbal or non-verbal behavior (extreme distress at small changes, difficulty with transitions, need to do the same things every day);
- Interests highly restricted or fixed in intensity, or focus much greater than expected (strong attachment or preoccupation with objects, condomive or excessive interest in specific subjects);
- Hyper- or Hyporeactivity to sensory stimuli or unusual interest in sensory aspects of the environment (apparent indifference to pain/temperatures, averse reaction to specific textures and sounds, visual fascination with movements or lights).

C. Symptoms must be present early in the developmental period, but they may not be fully apparent until there is a social need for these skills to be exercised, or they may be masked by possible strategies throughout life.

D. These symptoms cause clinically significant impairment in social, professional, personal functioning or other important areas of the person.

E. These disorders are not well explained by cognitive and intellectual impairment or global developmental delay.

In ICD-11 (last version of this manual) diagnoses of autism become part of autism spectrum disorder (6A02), which can be identified in the following ways:

- Level 1 - Mild: - 6A02.0 ASD without Intellectual Disability (ID) and with mild or no functional language impairment - 6A02.1: ASD with ID and with mild or no functional language impairment.
- Level 2 - Moderate: - 6A02.2: ASD without ID and impaired functional language - 6A02.3: ASD with ID and impaired functional language.
- Level 3 - Severe: - 6A02.4: ASD without ID and lacking functional language - 6A02.5: ASD with ID and lacking functional language.

The DSM and the ICD are different in their purposes. The first is done by the APA and focuses on describing and classifying mental disorders. The second, made by the WHO, focuses on describing and classifying diseases, injuries and causes of mortality.

Epidemiology

The prevalence of ASD, as currently defined, is at least 76 per 10,000 (1 in 132) and possibly as high as 110 per 10,000 (1 in 91). Boys are more affected than girls with an average ratio of 3:1.

However, this proportion is substantially affected by cognitive impairment; the ratio among individuals without intellectual disability (formerly called mental retardation) is at least 5:1, and the ratio among those with intellectual disability is closer to 2:1.

Etiology

In 75 - 80% of cases the etiology is unknown.

In some cases, patients with ASD may also have mental retardation, epilepsy, structural brain malformations, dysmorphism, microcephaly and macrocephaly why is it so important to find an etiology?

We can initiate treatment (pharmacological and intervention strategies), find out what the prognosis is, carry out prevention and offer prenatal diagnosis and genetic counseling for the couple.

This stimulation will offer a series of stimuli aimed at boosting the child's motor and cognitive development. They can be auditory, visual, motor function, manual, cognitive and social, language and orofacial motricity stimuli. The stimulation, in addition to being carried out by the speech therapist, occupational therapist and physiotherapist, must have the participation of the family.

What tests are recommended for a child with ASD criteria?

Mainly, cytogenetic tests, (array and fragile X as first line test) metabolic study, exome, genome, neuroimaging and electroencephalogram (EEG).

In an one year review in a genetic consultation, we found 22 patients with ASD and in seven of the them we found an etiology (around 30%) There are diagnosis at 2 years of age and in one case only at 15 years old (Table 1).

Cases	Ages of diagnosis of ASD (years)	Etiology	Comorbidities
1	7	No	No
2	7	No	No
3	5	No	No
4	2	Hunter	No
5	4	No	Sick sinus syndrome
6	2	No	No
7	4	No	No
8	3	No	No
9	15	Trisomy 8	No
10	13	No	No
11	3	Rett	Microcephaly
12	2	No	No
13	9	6p21.3	No
14	2	No	No
15	6	X fragile premutation	No
16	2	No	No
17	12	45X0	Hypothyroidism
18	8	No	No
19	2	X fragile premutation	No
20	3	No	No
21	3	Angelman	No
22	5	Del 2p13	Defect of corpus callosum and cerebellum

Table 1: *ASD-one year review cases [1].*

It is important clinicians send patients with neurodevelopment disorders as soon as possible to a genetic consultation to avoid late diagnosis and a proper genetic counseling for the couple.

Risk of recurrence

Genetic syndromes, mutations, and de novo copy number variants (microdeletions and microduplications) account for 10% to 20% of ASD cases for a couple with one affected child, the recurrence risk in the next child is approximately 5% to 10%, whereas the recurrence risk for monozygotic twins (70% to 90%) are substantially higher than for dizygotic twins (0% -10%).

Test with collaboration of the family

There are many test for the screening of ASD (Table 2), but M-CHAT is the most frequent used (Table 3).

Modified checklist for autism in toddlers (M-CHAT)	16-48 months	5-10 minutes
Pervasive Developmental Disorders Test-II Primary Care Screener (PDDST- II PCS)	18-48 months	10-15 minutes
Childhood Autism Spectrum Test (CAST)	4-11 years	10 minutes
Social Communication Questionnaire (SCQ)	4-18 years	10-15 minutes
Social Responsiveness Scale (SRS)	4-18 years	15-20 minutes
Autism Spectrum Screening Questionnaire	6-17 years	10 minutes

***Table 2**: Type of test for ASD.*

1	If you point across to something the room does, does your child look at it? (Point a toy or an animal)	Yes	No
2	Have you ever wonder your child is deaf?	Yes	No
3	Does your child pretend or make-believe? (Pretend to drink from an empty cup, pretend to talk on a phone or pretend to feed a doll or stuffed animal)	Yes	No
4	Does your child climb on things, such as upstairs, playground equipment or furniture?	Yes	No
5	Does your child look at things you are looking at?	Yes	No
6	Does your child ever use one finger to point, to ask for something or get help? (Pointing a snack or toy)	Yes	No
7	Does your child ever uses one finger to show you something interesting? (Pointing to an airplane in the sky or big truck in the road)	Yes	No
8	Does your child interest in another children? (Watch other children, smile at them or go to them)	Yes	No
9	Does your child show you things by bringing them to you or holding them up for you to see- not to get help, but just to share (Showing you a flower, a stuffed animal or a toy truck)	Yes	No
10	Does your child respond to his/her name when you call? (Look up, talk or babble, stop what he/her is doing when you call)	Yes	No
11	Does your child smile in response to your smile	Yes	No
12	Does your child get upset by everyday noises? (Scream or cry to noise such as vacuum cleaner or loud music)	Yes	No
13	Does your child walk?	Yes	No
14	Does your child look to your eyes when you are talking, playing with her/him or dressing her/him?	Yes	No

15	Does your child copy what you do (Bye-bye, clap, make a funny noise)	Yes	No
16	If you turn your head to look to something, does your child look around to see what you are look at?	Yes	No
17	Does your child try to get you to watch him or her? (Look at you for praise, or say look or watch me)	Yes	No
18	Does your child understand when you tell him or her to do something? (If you don't point, can your child understand put the book on the chair or bring me the blanket)	Yes	No
19	If something new happens, does your child look at your face to see how your feel about it? (If she or he hears a funny or strange noise or sees a new toy, would she or he looks at your face?)	Yes	No
20	Does your child like movement activities? (Being swung or bounced in your knees)	Yes	No

Table 3: *MCHAT-R - 2009 Diane Robins, Deborah Fein and Marianne Barton.*

M-CHAT quotation

For all items except 2, 5, and 12, answering «no» indicates risk for ASD; for items 2, 5, and 12, «yes» indicates high risk of ASD. The following algorithm maximizes the psychometric properties of the M-CHAT:

- Low risk: Total rating is 0-2; if the child is younger than 24 months, repeat the M-CHAT-R at 24 months. No further measures are required unless surveillance indicates risk for ASD.
- Moderate risk: Total score is 3-7; conduct a Follow-Up Interview (second step of the M-CHAT-R/F) to obtain additional information on risk responses. If the M-CHAT-R/F score continues to be equal to or greater than 2, the child scores positive in spite. Necessary measures: refer the child for diagnostic evaluation and possible signal for early intervention. If the Follow-Up Interview score is 0-1, the child scores negative. No further measures are required unless surveillance indicates risk for ASD. The child will have to do the screening again later.
- High risk: Total quote is 8-20; it is acceptable to skip the Follow-up Interview and refer the child for diagnostic evaluation and possible signal for early intervention [1,2].

Features in younger children or with more severe disabilities

They showed motor stereotyped type: lapping, flicking fingers, swaying, spinning, skipping or jumping on tiptoes, full body twisting, fascination with objects that move or rotate, atypical use of peripheral vision or side gaze, attachment to unusual objects - often hard rather than soft, the category may be more important than the specific object, behavioral rigidity, difficulty accepting unexpected changes, insistence on the same object or theme, repetitive and stereotyped self-injurious behavior, extreme dietary electivity.

Features in older children

May not have any meaningful speech, higher proportion of syllables with atypical phonation (e.g. squeaks, growls, and screams), delayed receptive and expressive language milestones, increase in idiosyncratic or inappropriate media, self-injurious behavior, aggression, tantrums, echolalia: immediate (e.g. in response to questions), delayed (scripted verses, reciting memorized dialogue), pronoun inversion (often «you» to «I» or «you» to «me»), can label objects or actions, but does not use these words to make requests or answer questions, delayed, diminished, or absent use of gestures (e.g. showing, nodding, pointing, nodding, describing actions), mainly primitive motor gestures to communicate (e.g. leading contact gesture or pulling another person's hand), pronoun inversion (often «you» to «I» or «you» to «me»),can label objects or actions, but does not use these words to make requests or answer questions, delayed, diminished, or absent

use of gestures (e.g. showing, nodding, pointing, nodding, describing actions),mainly primitive motor gestures to communicate (e.g. leading contact gesture or pulling another person's hand) [2-5].

Clinical features

Approximately 20% to 35% of children with ASD start to speak but later experience language regression.

Regression is defined as loss of significant spoken language after acquisition of at least 3 to 5 words, often accompanied by loss of gestural communication, decreased use of gaze to regulate social interaction, social withdrawal, and loss of spontaneous imitation and symbolic game skills.

Regression most often occurs between the ages of 15 and 24 months, can be gradual or sudden, and can occur after typical development or overlap with pre-existing atypical development.

Myths and realities

There is evidence that in utero exposure to drugs such as valproate, misoprostol and thalidomide, and other aspects of the prenatal environment such as testosterone level, alcohol exposure and infections (e.g. rubella, cytomegalovirus) may be associated with a risk increased ASD, but epidemiological data for its confirmation are lacking.

Perinatal factors such as prematurity, low birth weight, intrapartum hypoxia, and neonatal encephalopathy may also be associated with an increased risk of ASD.

Claims that the toxic effects of the measles-mumps-rubella vaccine or a combination of thimerosal-containing vaccines are responsible for the increase in ASD prevalence have been firmly refuted by epidemiological studies, and the weight of all scientific evidence overwhelmingly favors rejection. hypothesis that there is a causal association between vaccines and ASD.

Some research suggests that gastrointestinal (GI) problems such as chronic constipation and diarrhea are more common in children with ASD, but the relationship remains unclear because most studies have not compared representative groups of children with ASD with appropriate controls.

A retrospective study using complete medical records showed that there was no difference in the cumulative incidence of GI problems between a well-characterized population cohort of 121 individuals with autism and 242 controls.

Treatments without scientific evidence

Amino acids, antibiotics, antifungals, antivirals, antioxidants, auditory integration training, chelating agents, chiropractic manipulation, craniosacral therapy, digestive enzymes, folic acid, folinic acid, gluten-free/casein-free diet, hyperbaric oxygen, intravenous or oral immunoglobulin, mineral supplements, music therapy, yoga, massages, omega-3 fatty acids, transcranial magnetic stimulation, trimethylglycine, vitamin A, vitamin B6, magnesium, vitamin B12 (intramuscular or oral) and Vitamin C [2-4].

Conclusion

ASD is a neurodevelopment disease where in about 75% of cases an etiology is not found. There is a variability of clinical symptoms. It is important to screen possible causes of ASD so that the child is treated early and genetic counseling and prenatal diagnosis are performed. The possible causes (e.g. vaccines) and some treatments of ASD did not show clinical evidence in the studies carried out.

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