Autism spectrum disorder (ASD): an important issue to be addressed

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Autism spectrum disorder (ASD) occurs in early childhood and represents a neurodevelopmental disorder. Its etiology is multifactorial and remains unknown, although genetic factors stand out. Additionally, it is characterized by a wide range of symptoms. Various types of ASD have been identified, including classic autistic disorder or Kanner's syndrome, Rett syndrome, childhood disintegrative disorder, Asperger's syndrome and pervasive developmental disorder not otherwise specified or atypical autism (1).

In recent years, there has been an increase in the prevalence of ASD. Several scientific articles corroborate this rise, with the indicator increasing from 4–5/10,000 cases in the 1960s to 260/10,000 cases, a trend that has become more visible in the 21st century (2,3).

In many countries, the increased prevalence of ASD may be attributed to the implementation of measures such as the variation and improvement of diagnostic criteria, promotion and encouragement of special education policies, increased availability of services and awareness of ASD among the general public and professionals.

The increase in the number of children diagnosed with ASD in the 1990s led to the parallel establishment of services for managing children with ASD in several countries, causing a chain reaction (3).

Regarding the epidemiological data related to ASD, some Latin American countries, including Peru, lack such data.

The Registro Nacional de Personas con Discapacidad (RNPCD – National Registry of Persons with Disabilities), under the governance of the Consejo Nacional para la Integración de la Persona con Discapacidad (CONADIS – National Council for the Integration of Persons with Disabilities), recorded a total of 219,249 persons with disabilities as of August 31, 2018, out of whom 4,528 (2.06 %) had ASD, with 3,663 (80.9 %) being males and 865 (19.1 %) females. The annual increase in registrations of persons with ASD is evident: 2,248 in 2015, 2,830 in 2016 and 3,709 in 2017.

The prevalence of ASD appears to be increasing worldwide. Better awareness of the problem as well as improved diagnostic criteria and tools are cited as possible explanations for this increase (4).

For instance, Li et al. reported a prevalence rate of 2.38/10,000 for autism in mainland China; however, they admitted that this figure might be underestimated. Zhang and Ji found a higher prevalence of 11/10,000 based on a survey conducted in Tianjin, China. In Taiwan, Chien et al. reported a cumulative prevalence of ASD, increasing from 1.79/10,000 to 28.72/10,000 in 1996. Furthermore, a study based on Taiwan's national survey, using health insurance information, noted a high prevalence rate of 122.8/10,000 in 2007. In Hong Kong, Wong et al. estimated an incidence of ASD at 5.49/10,000, with an average prevalence of 16.1/10,000 between 1986 and 2005. Twelve studies found in Chinese databases reported ASD prevalence ranging from 2.8/10,000 to 29.5/10,000. Although the existing data seem to suggest this, it remains unclear whether there is a true increase in the prevalence of ASD in the ethnic Chinese population (5).

The first documented descriptions of what we now recognize as ASD can be found in the publications of Leo Kanner (1943) and Hans Asperger (1944). However, individuals with these characteristics have likely existed throughout history.

One of the first testimonies dates back to the 16th century, in which Johannes Mathesius, chronicler of the German religious reformer Martin Luther, narrated that an adolescent of approximately 12 years of age had a severe case of autism. However, Luther erroneously thought that the boy was nothing more than a soulless mass of flesh possessed by the devil and suggested that the child should be suffocated to death (6).

The term "autism" was coined by Eugen Bleuler and derived from two Greek words: "autos" meaning "self" and "ismos" referring to the state of being. Bleuler's use of this term led to misunderstandings since it was assigned to patients with schizophrenia.

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Following World War II, two major studies on this psychopathology, which was then called “pathology of autism,” were conducted. Notably, Kanner made the first significant contribution with his article *Autistic Disturbances of Affective Contact* (7), which many specialists consider fundamental in understanding current concepts of autism. The second contribution came from Asperger, who published *Autistic Psychopathy in Childhood* in 1944, a report detailing a study conducted with four youngsters with autism (8). Coincidentally, both Kanner and Asperger used the term “autism” in their work, despite the fact that their work and research were unknown at the time (1,6).

ASD manifests throughout the life of the affected person with a wide variability of symptoms, ranging from mild personality alterations to severe disability.

Clinical manifestations of patients with ASD are categorized into three dimensions: impaired social interaction, impaired verbal and nonverbal communication, and lack of interest in common situations and repetitive and obsessive behavior patterns (e.g., eating the same food over and over again). Additionally, unusual responses to sensory and tactile stimuli, behavioral disturbances and special skills may be observed (9).

On the other hand, clinical manifestations of autism typically emerge between one and a half and two years of age and remain stable during the preschool and school years. In general, parents start to express concerns and ask for help after the child’s first year.

While approximately 30 % of affected children experience normal development between their first and second year, some may regress progressively or unexpectedly, leading to a delay in their development. In some cases, symptoms of ASD may not become apparent to parents and teachers until patients reach four or six years of age. They may show profound mental retardation or normal or above-average scores on IQ tests. Those with normal intellectual abilities typically maintain cognitive and language skills, but they may struggle to understand non-verbal messages and experience difficulties in interpersonal relationships. Especially in group settings, they may exhibit poor in-hand manipulation skills, maintain an expressionless face, avoid smiling and may not make eye contact. Those who speak may do so with an unusual tone and engage in repetitive conversations that are of interest only to them (1).

Early diagnosis and intervention are key to optimizing the quality of life and prognosis for individuals with ASD. Timely interventions can significantly improve social relationships, language skills and adaptive behavior, increasing the likelihood to achieve successful inclusion in educational and social settings while reducing family stress and social costs.

Early detection and diagnosis are not free of major difficulties. The presentation and progression of symptoms, along with limited dissemination of diagnostic strategies and tools as well as insufficient experience and training of health professionals to recognize ASD symptoms, make it difficult to establish early detection procedures. Such challenges are exacerbated when screening is performed before 36 months of age, in addition to the high costs and time requirements of diagnostic tests. These barriers need to be addressed at various levels of care.

Additionally, appointment times are often insufficient, which is another aspect that affects the diagnostic process at early ages across all levels of care. Early diagnostic stability remains a significant concern for clinicians faced with the challenge of identifying ASD in young children.

A meta-analysis may show some stability in diagnosis. However, 73 % to 100 % of children aged 3 to 5 years with an ASD diagnosis remain stable.

Clinical judgments based on the Diagnostic and Statistical Manual of Mental Disorders, 4th edition, Text Revision (DSM-IV-TR) and DSM-5 are effective for diagnosis and assessing stability over time (10).

Management of individuals with ASD is primarily educational. It consists in modifying inappropriate behaviors and, in some particular cases, employing specific pharmacological treatments. Such undesirable behaviors could be modified with a particular treatment, reinforcing those that are more appropriate according to their abilities. The objective is to teach and develop daily living skills, foster independence, curb repetitive and obsessive behaviors, optimize adaptive behaviors, and achieve receptive and expressive language acquisition. This language can be used in future work activities.

The effectiveness of the treatment will depend on the characteristics of each child. The most commonly used intervention is Applied Behavioral Analysis, which examines the causes and consequences of inappropriate behaviors and designs strategies to avoid them, so that they are redirected toward desirable and alternative behaviors. This therapy can be administered by an expert or by parents after training (11).

The closest relatives of children with ASD show certain feelings and attitudes toward the diagnosis (guilt, frustration, sadness, stress and uncertainty), which are associated with the need of professional care and adequate information regarding ASD prognosis, management, etiology, etc. Specialized professional help is crucial to deal with the complicated situations that they perceive and must learn to manage and control (12).

ASD is a neurodevelopmental disorder gaining attention due to the increasing health indicators as well as the incidence and prevalence rates reported by developed countries. Such increase is likely explained by a larger number of diagnoses and diagnostic tools, which may not always be scientifically validated. Therefore, it is suggested the involvement of other epigenetic factors that may increase these indicators. The etiology of this condition is not yet fully defined.

In our country, there are neither statistics nor research studies to assess the real behavior of ASD; only an increase is observed in cases reported by families to CONADIS, which is reflected in the Encuesta Demográfica y de Salud Familiar (ENDES – Demographic and Family Health Survey) reports. Furthermore, we lack trained professionals or the experience for early diagnoses that would lead to reduce the disability caused by late diagnosis due to the variability of ASD symptoms, which causes a delayed intervention and poor prognosis.

It is well known that proper training to acquire the necessary competencies to practice as primary care physicians and specialists for early diagnosis and management, as well as conduction of research on the epidemiology, etiology and associated risk factors of ASD in our national context, are important tasks that need to be addressed in the short term. The academia has a significant role in fulfilling its social responsibility and enhancing the effectiveness of care provided by health systems to children and families affected by ASD in our country.

The Ministry of Education (MINEDU), in coordination with the Ministry of Health (MINSA), is implementing the Plan Nacional para las Personas con Trastorno del Espectro Autista (National Plan for Patients with Autism Spectrum Disorder) 2019–2021 and is making efforts to improve their care.

Law No. 30150, i.e., Ley de Protección de las Personas con Trastorno del Espectro Autista (Law on Persons with Autism Spectrum Disorder), enacted in 2014 pursuant to the Constitución Pública del Perú (Public Constitution of Peru) and Law No. 29973, Ley General de las Personas con Discapacidad (General Law on Persons with Disabilities), establish provisions for early detection, early diagnosis, early intervention, comprehensive education, vocational training, and labor and social inclusion (4).

Finally, improving care for patients with ASD requires the participation of multiple stakeholders playing different roles. The academia is crucial in laying the scientific foundations for planning and approaching strategies to achieve pending objectives: to improve the quality of life of individuals affected by ASD.

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