MEDICAL SCIENCE MONITOR

REVIEW ARTICLES

e-ISSN 1643-3750
© Med Sci Monit, 2024; 30: e944161
DOI: 10.12659/MSM.944161

Received: 2024.02.17
Accepted: 2024.03.25
Available online: 2024.04.19
Published: 2024.06.04

Autism Spectrum Disorders: Etiology, Epidemiology, and Challenges for Public Health

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Statistical Analysis C
Data Interpretation D
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Financial support: None declared
Conflict of interest: None declared

Autism spectrum disorder (ASD) is a growing public health problem. American psychiatrist Leo Kanner is considered the "father of autism."

ASD encompasses a range of neurodevelopmental disorders that last throughout life. Symptoms of ASD include impairments in social skills, including specific repetitive behaviors, as well as abnormal sensory responses. The clinical symptoms of ASD vary among patients. Their severity also differs, both in the area of social communication and cognitive functioning. The etiology of ASD is still unclear, although a role is attributed to both genetic and environmental factors. According to the World Health Organization, 1/100 children have ASD, but these estimates vary depending on the methodology used. Nevertheless, early detection of ASD and initiation of appropriate therapy may be essential in the continued functioning of patients and their families.

The purpose of this article is to provide an overview of current knowledge about autism spectrum disorders. We discuss factors associated with autism and the prevalence of ASD in various parts of the world, and identify the most common diseases comorbid with ASD, pointing to limitations in the quality of life of patients with ASD and their families.

Keywords: **Mental Disorders Diagnosed in Childhood • Autism Spectrum Disorder • Quality of Life • Public Health**

Abbreviations: **ADHD** – attention deficit hyperactivity disorder; **AI** – artificial intelligence; **ASD** – autism spectrum disorders; **ID** – intellectual deficit; **DALY** – disability-adjusted life years; **ML** – machine learning; **MRI** – magnetic resonance imaging; **SDI** – social development index; **WHO** – World Health Organization

Full-text PDF: https://www.medscimonit.com/abstract/index/idArt/944161

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Introduction

Autism spectrum disorder (ASD) is a significant public health problem. The term autism was coined by the Swiss psychiatrist Eugen Bleuler in the early 20th century, but the first descriptions of the disorder were made by the American psychiatrist Leo Kanner in his work Disturbances of Affective Contact, published in 1943. In 1944, further cases were described by the Austrian pediatrician Hans Asperger [1]. ASD is a lifelong neurobiological disorder characterized by 2 main symptoms: (1) disturbances in social skills, including deficits in social communication, and engaging in repetitive, restricted behavior patterns; and (2) abnormal sensory reactions [2,3].

The clinical symptoms of ASD vary among patients. Their severity also differs, both in the area of social communication and cognitive functioning [4]. The most characteristic symptoms of ASD are deficits in social interactions, including empathy deficits, limited vocabulary, inability to adhere to accepted norms and behaviors, and difficulties maintaining eye contact [5,6]. The diversity and varying degrees of symptom severity contribute to significant differences in the independence levels of individuals with ASD. While some can lead independent lives, others need lifelong support, often having substantial levels of disability [7].

ASD is a group of genetically diverse disorders. Research so far indicates that it may be caused by both hereditary and new gene changes. More and more de novo genes are being identified that cause deficits in communication, behavior, and social functioning. It is estimated that these genes are responsible for 10-20% of ASD cases. Additionally, patients with similar genes may be diagnosed at different levels of the spectrum [8,9].

ASD manifests identifiable symptoms in children as early as 18-24 months of age [2,3], with deviations from typical developmental trajectories. According to data from the World Health Organization (WHO), 1/100 children have autism spectrum disorders [7]. However, ASD is usually detected much later. The average age of ASD diagnosis is 4 years 3 months and, despite many years of activities to increase the awareness of the public and health care professionals, this has not changed significantly, in part because behavioral tools for diagnosing ASD are not fully appropriate for young children. Many social communication skills are developed by children later in life, and some of the behaviors typical of people with ASD are normal in early childhood [10].

Diagnosis of ASD among children is carried out by specialized psychological and pedagogical clinics or private diagnostic and therapeutic centers specializing in the field of autism spectrum. The diagnosis consists of an interview with the parents, observation of the child, analysis of additional materials (eg, the educator's opinion), and sometimes also includes additional tests. A specialist team is involved in the diagnosis process, which includes a psychiatrist, psychologist, and sometimes other specialists, such as a pedagogue, audiologist, and speech therapist. Based on the team's opinion, the psychiatrist issues an appropriate diagnosis [11].

Importantly, autism spectrum disorders are frequently associated with other psychological problems, which may additionally reduce the quality of life of patients and their families [12]. Early detection facilitates the implementation of timely and tailored interventions, including therapeutic measures, which can significantly enhance the daily functioning of individuals with ASD [7].

The purpose of this article is to provide an overview of current knowledge about autism spectrum disorders. The content covers the possible causes of this type of disorders, prevalence in various parts of the world, co-occurring diseases, and limitations in quality of life, as well as challenges in providing appropriate care for people with ASD.

Factors Associated with the Development of ASD

ASD is a neurodevelopmental disorder influenced by a complex interplay of genetic and environmental factors. Despite extensive research, potential etiological mechanisms of ASD remain unknown. Although research confirms that genetics plays an important role in the etiology of ASD, genetic risk, responsible for up to 60% of cases [13], is known to be modulated by prenatal, perinatal, and postnatal environmental factors [14].

The increase in the incidence of ASD in recent years or decades is attributed, in part, to heightened exposure to environmental risk factors [2]. Notably, empirical evidence establishes that prenatal exposure to thalidomide and valproic acid increases the risk of ASD occurrence in a newborn. Advanced parental age and childbirth within the context of infertility treatments also emerge as contributing factors [15]. Moreover, maternal autoimmune conditions, such as thyroid disease, diabetes, or psoriasis, as well as various infections during pregnancy, may play a role in a development of ASD in a child [16].

Some studies indicated a relationship between the intervals between pregnancies and the occurrence of ASD, with both shorter and longer intervals being important [17]. ASD is more common in prematurely-born children, but also in those born by cesarean section, who have low birth weight or a low Apgar score [16,18,19].

Prevalence of Autism Spectrum Disorders

Due to the multitude and varying degrees of severity of symptoms, diagnosing ASD is often very difficult. Diagnosis requires

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high awareness of parents, educators, and caregivers, who are usually the first to notice typical symptoms and contact specialists to conduct a multi-stage diagnosis and confirm or refute assumptions [2].

There is considerable variability in ASD prevalence estimates across different regions of the world, which may stem from differences in awareness levels of primary caregivers and methodological distinctions inherent in various studies [20]. Methodological factors include diverse detection methods, varying study populations (typically focused on children, with limited data on the African or Middle-Eastern populations), and divergent case definitions. It is also important to emphasize the evolving nature of the clinical definition of autism and related disorders, which also influences the estimates [21].

Globally, the prevalence of ASD is estimated to range from 0.02% to 3.66% of the population [6]. Systematic reviews reveal varying prevalence rates, with ASD affecting approximately 0.38% to 3.13% of the European population, 0.11% to 1.53% in the Middle-East, 0.08% to 9.3% in Asia, 0.87% to 2.21% in North America, around 1% in Africa, and 1.7% in Australia [2,6,22]. In Europe, the highest incidence of ASD is observed in Sweden (115/10 000). In Poland, the average is estimated at 35/10 000 children and is approximately 4 times higher in men (however, the data do not cover all regions of Poland) [23].

Data collected by the ASDEU (Autism Spectrum Disorders in the European Union) project indicate an average ASD incidence of 12.2 per 1000 children aged 7-9 years, or 1 in 89 [24]. Over the past few decades, there has been a steady increase in the incidence of ASD in many regions of the world, including Europe and the United States. The incidence is higher in boys/men and in White children [3,25]. However, attention should be paid to the decreasing ratio of men to women with ASD, which may be attributed to increased awareness of ASD in women [26].

The Most Common Comorbidities Associated with ASD

ASD is frequently associated with various comorbidities, predominantly involving mental disorders. It is estimated that autism coexists with intellectual disability in approximately 33% of cases [3], with attention deficit hyperactivity disorder (ADHD) being the most prevalent among mental disorders [27]. The combined prevalence of ASD and ADHD is estimated at 28%, while anxiety disorders and sleep-wakefulness disturbances are reported at 20% and 13%, respectively. Destructive disorders, impulse control, and behavior issues account for 12%, depression for 11%, obsessive-compulsive disorder for 9%, bipolar disorder for 5%, and schizophrenia for 4% [28].

Assessment of the incidence of additional mental disorders depends on the research method used. Notably, the coexistence of depression appears more pronounced in studies utilizing standardized interviews, particularly self-assessment tools, to assess depression. Interestingly, these associations are more prevalent among individuals with higher intelligence levels, and notably among women with ASD [29].

Even greater heterogeneity of disease co-occurrence is observed in children with ASD. The prevalence of at least 1 mental disorder in children with ASD is estimated at 54.8-94%, with the most common comorbidities being ADHD, anxiety, depressive disorders, and sleep disturbances. Research indicates that ADHD occurs in 0-86% of children with ASD, anxiety states occur in 0-82%, depressive disorders occur in 0-74%, sleep disorders occur in 2.08-72.5%, and intellectual deficit (ID) occurs in 0-91.7%. Gender emerges as an influential factor in the manifestation of comorbidities among children with ASD. For instance, the co-occurrence of ADHD is more prevalent in boys, while epilepsy/seizures are more common in girls.

Additionally, age has a significant impact – as it increases, a higher incidence of ADHD and anxiety is observed, but sleep disorders are more common in younger children [2].

Importantly, individuals with ASD are also more likely to have other comorbidities, such as epilepsy/seizures, vision or hearing disorders, or gastrointestinal disorders. These conditions can contribute to fluctuations in ASD-related behaviors, potentially leading to delayed diagnosis or misdiagnosis [30].

Quality of Life Among People with ASD

Research on the quality of life of individuals with ASD indicates it is lower than in typically developing individuals, as assessed using tools designed for the general population [12]. The deterioration in quality of life is associated with behavioral problems in individuals with ASD, the severity of their symptoms, social skills, and co-existing mental health issues or educational factors [31].

Studies reveal that individuals with ASD score lower across all quality-of-life domains. Adults with autism spectrum disorders report elevated levels of anxiety, increased feelings of loneliness, and challenges with sensory processing when compared to typically developing individuals. These factors notably impact their perceived levels of physical and mental health, as well as the quality of their social relationships [32]. The research underscores the significant role of anxiety, influencing both competencies and social interactions [33].

Additionally, the coexistence of other diseases poses a substantial negative impact on both the quality of life and economic

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well-being of individuals affected by ASD [2,34,35]. Despite the lack of precise estimates of disability among individuals with ASD, this aspect should also be taken into account. Insights from the Global Burden of Diseases, Injuries and Risk Factors Study show increasing disability-adjusted life years (DALYs) for those with ASD residing in countries with a high social development index (SDI) score [26].

Furthermore, it is crucial to underscore the profound impact of ASD and its comorbidities on the quality of life experienced by parents of children with ASD, who endure heightened stress levels in comparison to parents of typically developing children.

Parents of children with ASD report greater maladjustment and more personal and family challenges, which translates into a lower quality of life for entire families [36].

Public Awareness About ASD

Due to the increasing number of ASD diagnoses, an ongoing discourse persists regarding whether the apparent rise is attributable to increased exposure to risk factors or is a result of heightened societal awareness, including the awareness of parents and guardians of children and adults with ASD, but also the awareness of medical staff, enhanced availability of diagnoses, and greater accuracy of diagnostic criteria [25,26].

The growing awareness of ASD is underscored in numerous publications, and is notably evident in World Health Organization (WHO) documents. The WHO has embraced initiatives such as the Comprehensive Mental Health Action Plan, advocating for equitable access to high-quality health and social care precisely tailored to the needs of the patient. WHO promotes a multifaceted approach to the patient and strengthening everyday functioning of people with mental disorders [7].

Despite the growing availability of systemic resources that facilitate more comprehensive and accessible support for individuals with ASD, there are reports indicating that some specialists remain unaware of the prevalence of these disorders and the common co-occurrence of ASD with other mental health disorders [24]. Consequently, it is crucial to expand the knowledge of medical professionals, particularly those caring for children, who may notice symptoms of the disorder during routine medical visits, prompting the need for thorough diagnostics [37].

Public Health Challenges in Supporting Individuals with ASD

Various support methods exist that can significantly enhance the daily functioning of individuals with ASD. Early and individually tailored interventions, coupled with support from the immediate environment, can significantly influence the development of people with ASD.

Research indicates that approximately 80% of teenagers and over 60% of adults with ASD benefit from support in the form of mental health services, educational assistance, or interventions that improve sensory/motor functions. Nevertheless, more than 80% of individuals report encountering barriers in accessing such services, particularly those with low income and those residing outside major urban centers [38].

Individuals with ASD have diverse needs for accessing services, particularly in education. Despite notable strides towards ensuring optimal education for individuals with ASD, there remain areas requiring improvement, particularly in Central and Eastern European countries [39].

Significant efforts have been undertaken in certain regions, such as Poland, to address these challenges. For instance, amidst the COVID-19 pandemic, Poland adapted the PEERS® (Social Skills Training) curriculum for young people on the autism spectrum, delivering it in a hybrid, online format. This program demonstrated good effectiveness in fostering social skills, with the positive effects persisting over the subsequent 6-month observation period [40,41].

Expanding public awareness regarding ASD and the specific needs of people with ASD is crucial. Creating an inclusive school environment tailored to the diverse requirements of individuals with autism, including addressing sensory issues, is imperative. This encompasses responding to a spectrum of educational needs by offering diverse activities, nurturing interests, fostering communication and social skills, and safeguarding the rights of individuals with ASD, including the right to safety and protection against discrimination.

Research confirms that early diagnosis and initiation of quick, multiple interventions in people with ASD result in better functioning and better quality of life, not only for people on the autism spectrum, but also for entire families [11].

Future Directions

ASD is a multi-faceted disorder and therefore requires a diverse, often individualized therapeutic approach [11,42]. Long-term management of ASD is essential to maximize functioning and quality of life. It is important to minimize deficits in social skills and communication, facilitate development and learning, and to provide social education (including medical staff) and support for families.

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Given the diagnostic difficulties, more thorough screening is also needed. It is necessary to educate primary health care staff so that they can identify children who may have ASD as quickly as possible. It is also important to know the behaviors and co-occurring conditions that affect the quality of life of people with ASD.

Growing evidence confirms the effects of behavioral interventions, especially those undertaken at the earliest possible age, on the functioning of people with ASD. Help and treatment for people with ASD should be individualized according to age and development level, and should include a variety of interventions.

Despite many reports on the incomplete effectiveness of behavioral observations, more and more research teams are working on developing screening and diagnosing tests for ASD using artificial intelligence (AI) and machine learning (ML), such as using structural magnetic resonance imaging (MRI), functional MRI, and hybrid imaging techniques. The development of this type of diagnostic tool would enable more accurate detection of ASD, including diagnosis in the youngest possible groups of patients, to be able to start early development support interventions [43,44].

Conclusions

Epidemiological data indicate an increasing number of people living with ASD across diverse global regions. This trend can be partially attributed to the increasing public awareness of ASD, facilitating quicker diagnoses for individuals with these disorders. This is particularly important in providing prompt and appropriate support to those in need, including health, social, and educational services. Current medical understanding underscores the necessity for a diverse range of long-term support services for those with ASD and for their families and caregivers.

Numerous studies underscore the effectiveness of diverse therapeutic methods for children, adolescents, and adults with ASD. Therefore, it is crucial for public health initiatives to prioritize the early identification of individuals with ASD, including the diagnosis of symptoms and individualized needs. Equally vital is the awareness and recognition of co-occurring disorders, such as depression and anxiety, in individuals with ASD. Comprehensive interventions initiated at an early stage could significantly enhance social functioning and improving the quality of life of people with ASD and their families.

Department and Institution Where Work Was Done

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