

REVIEW ΑΝΑΣΚΟΠΗΣΗ

The epidemiology of autism spectrum disorder and factors contributing to the increase in its prevalence

Autism spectrum disorder (ASD) is a multifactorial neurodevelopmental disorder which has attracted the interest of researchers in many scientific fields. The prevalence of a disease or a disorder is important for healthcare and socio-economic reasons. According to the World Health Organization (WHO), currently, 1 in 160 children is born with ASD (0.63%), possibly a conservative estimate, as many studies have reported a prevalence higher than 1%. Even so, the WHO rate is approximately 14 times higher than that reported in the first epidemiological study on ASD, which was conducted in the UK 50 years ago. It is currently estimated that approximately 60 million people worldwide have ASD. Diagnosis and documentation of these individuals is challenging, due to the nature of the disorder and its broad spectrum. Many countries have no epidemiological data on ASD and several epidemiological studies on ASD record significant methodological limitations, but the currently available data indicate that the prevalence of ASD has risen over the years. The interpretation of this trend remains uncertain, but several factors have been documented that may have contributed to this apparent increase, including revision of the diagnostic criteria, overdiagnosis, scientific advances, accessibility to services, increase in social awareness, and improvement in the methodology applied in the epidemiological studies, in addition to environmental and other factors.

1. INTRODUCTION

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that was first described by the psychiatrist Leo Kanner in 1943. Until a few decades ago, ASD was considered a rare disorder, the causes of which were mostly unknown. Today, it is considered a multifactorial disorder. Epidemiological studies on ASD began after the second half of the 20th century and have revealed a continuing and rapid increase in prevalence. Estimation of the prevalence of a disease or a disorder is important for healthcare and socio-economic reasons. Various factors have been identified that have contributed to this increase, but interpretation of this apparent trend remains uncertain.

This is a literature review on the epidemiology of ASD, conducted in the course of a doctoral thesis currently in progress at the Department of Social Work of the Hellenic Mediterranean University (HELMPEA), as part of its theory section. The aim of this article is to highlight the upward trend observed in the prevalence of ASD, and to cite the possible etiological factors that have been reported.

2. AUTISM SPECTRUM DISORDER

ASD is a heterogeneous, lifelong, neurodevelopmental disorder characterized by persistent deficits in social communication and social interaction across multiple contexts, restricted/repetitive patterns of interests/behavior, and sensory dysfunction.¹ These characteristics often present during the child's early development as significant deficits in communication, social behavior and cognitive and adaptive functioning.¹ ASD is often accompanied by comorbid conditions, including intellectual disability (ID), attention deficit hyperactivity disorder (ADHD) and obsessive compulsive disorder (OCD).²⁻⁴ It is a disorder that cannot be easily identified by either the parents or the doctor in the early stages, but atypical behaviors can usually be observed after the age of 6 months.⁵⁻⁷

3. EPIDEMIOLOGY OF AUTISM SPECTRUM DISORDER

The World Health Organization (WHO)⁸ reports that 1 in 160 children (0.63%) is born with ASD, which is possibly

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a conservative estimate.⁹ In terms of gender, the disorder is observed four times more frequently in boys than in girls.^{8,10} According to the US Centers for Disease Control and Prevention (CDC),¹¹ the first epidemiological study on ASD, conducted by Lotter in 1966 in the county of Middlesex in England, reported a 0.045% prevalence (approximately 1 in 2,220 children).¹² Since then, the majority of epidemiological studies on ASD have documented an upward trend in prevalence.¹¹ Although not the most appropriate comparison, it is of note that the current rate reported by the WHO is approximately 14 times higher than in the first study, and the number of individuals with ASD is currently estimated to be more than 60 million globally.^{13,14} Accurate identification of the number of individuals with ASD is a challenging task. The wide spectrum of this disorder makes it difficult to identify the more highly functioning and better socially adjusted individuals with ASD, with less obvious ASD characteristics, which would otherwise lead to evaluation and subsequent diagnosis.

Epidemiological studies have been conducted in an attempt to determine the ASD prevalence in many countries. The heterogeneity of the results over geographic area and time reveals a diversity in terms of societies, cultures, services and accessibility, diagnostic procedures, availability of data, research adequacy and methodology and factors that are perceived to contribute to the appearance of the disorder and its apparent increase. Annually, ASD is diagnosed in more children than childhood diabetes mellitus (DM), childhood cancer and AIDS, combined.¹⁵

The majority of epidemiological studies have been conducted in the United States of America (USA), reflecting the fact that ASD is a major source of concern among health and social scientists in that country.¹⁶ The most recent CDC data (2016)¹⁰ showed a prevalence of 1 in 54 children (1.85%), in comparison with the first US epidemiological study conducted by Treffert in the 1960s, which reported a prevalence of 0.031% (1 in 3,225 children).¹¹ In 2000, the rate had increased to 0.67% (1 in 150 children), followed by 1.47% in 2010 (1 in 68 children), 1.45% in 2012 (1 in 69 children), reaching 1.68% in 2014 (1 in 60 children).¹¹ It is estimated that more than 5 million individuals with ASD live in the USA, considerably more than 1.5% of the population.¹⁷

Epidemiological data from countries in Europe, Asia and Oceania also indicate an increasing trend of the disorder, with most of the recent studies reporting rates higher or significantly higher than 1%. The interpretation of this trend remains uncertain.¹⁸ No data on the prevalence of ASD are currently available for Africa or for Central and South America.¹⁹

4. FACTORS ASSOCIATED WITH AN INCREASE IN THE PREVALENCE OF AUTISM SPECTRUM DISORDER

4.1. Revision of diagnostic criteria

Modification of the diagnostic criteria of a disorder is considered a major reason for a change in its epidemiology.^{19,20–24} Until 1980, when the Diagnostic and Statistical Manual of Mental Disorders, third edition (DSM-III) was issued, there had been no official diagnosis for ASD. The first diagnostic criteria in the DSM-III included characteristics that were similar to “classic autism” which had been described by Kanner.²⁵ In the subsequent 1987 revised version, DSM-III-R,²⁶ it was noted that, in order for an individual to be diagnosed with autism, at least eight of the 16 characteristics should apply, allowing for a more heterogeneous group.²⁵ In the following revision, DSM-IV, in 1994, the criteria were changed further, but difficulties were encountered during the diagnostic procedure in differentiating between the different types of autism. Because of this, in the most recent revision in 2013, DSM-V, the diagnoses of “classic autism”, Asperger syndrome and pervasive developmental disorders not otherwise specified, were classified in a single diagnostic category under the international term “Autism spectrum disorder – ASD”.¹ Differentiation in ASD is now based on clinical descriptors, such as the extent of dysfunctionality.^{1,25} According to a US epidemiological study conducted in seven states, the prevalence of ASD was 20% higher when assessed according to DSM-IV-TR than with DSM-V.⁶

4.2. Overdiagnosis

It is hypothesized that the changes in diagnostic criteria, especially those introduced since the 1990s, have led to broader consequences, making diagnostic identification and calculation of ASD prevalence more challenging.²¹ The changes in the diagnostic procedure and the concurrent increase in the prevalence of ASD revealed by epidemiological studies, can be considered to have attributed to increasing clinical awareness and diagnosis of the disorder.²¹

4.3. Scientific advances

Scientific advances and a multidisciplinary scientific approach have led to improvement in the knowledge about ASD symptomatology and diagnostic procedures, and the design of new tools.^{14,21,23,24} As a result of these advances, health professionals have more and better options available for the diagnosis of ASD.^{5,6,20,27} In addition, the current enhanced reproductive methods, including

specific intervention for the treatment of infertility, imaging during pregnancy and other perinatal factors have been proposed as factors contributing to the increase in diagnosis of ASD.⁹

4.4. Accessibility to services

The global increase in the numbers of people diagnosed with ASD has given rise to a need to establish specialized services in ever more communities, which has contributed to easier and faster recognition of the disorder.^{6,14,23,28} On the other hand, social exclusion, poverty and the cultural and organizational level of various countries and regions impair the capabilities for recognition of the disorder and may deprive sufferers of the benefit from services.²⁹

4.5. Methodology in epidemiological studies

Another probable cause for the apparent rise in ASD prevalence is the variety of methodological approaches in research. Sources of data, small samples (mostly in the earlier years), the changing diagnostic criteria of autism, the number of resources available to researchers, and other limitations affecting epidemiological studies over time, are all factors that affect prevalence evaluation.^{19,20,30} Some epidemiological studies have used data from the data base of a single service, while others have obtained their data from several services.¹⁹ Some have studied only one geographical region, while others have studied more regions in a country, or were conducted at a national level.¹⁹ Epidemiological studies that elicit their data from interviews with parents, teachers or health professionals have been reported to over-estimate the prevalence of ASD.¹⁹ Population-based studies indicate somewhat higher rates of prevalence than studies which derive their data from databases, although this difference is not considerable.¹⁹ Population-based epidemiological studies are evaluated as being of a higher research standard than studies based on selected samples or those that use databases, because they derive their sample from the entire population, in accordance with the criteria determined by each study.¹⁹ Methodological differences cannot fully account for the considerable variations between epidemiological studies in estimates of ASD prevalence, which implies the need to take more factors into consideration.¹⁹

4.6. Social awareness

Increasing numbers of people have an individual with ASD in their family, in their neighborhood or in their broader

social environment. In addition, public information campaigns on the disorder have steadily increased in recent years. On December 18th 2007, the United Nations (UN)³⁷ adopted resolution 62/139 declaring April 2nd as World Autism Awareness Day. There are now more references to ASD in the media, and in films and theatre performances. The internet, websites and social media provide information and regarding services and or specialists, and motivate people to participate. Social awareness is acknowledged to be one of the major factors that has contributed to the recorded increase in prevalence of ASD.^{21–24} Increasingly more parents, and also individuals who have discerned difficulties in themselves, are becoming more aware of the disorder and consult the relevant services for evaluation.³²

4.7. Environmental factors

The fetal brain is a very complex and sensitive organ which develops in a limited and specific time frame during pregnancy.^{7,33} This makes it extremely vulnerable to exposure to toxic agents.^{7,33,34} The fetal blood-brain barrier is not fully formed until the sixth month of pregnancy, leaving the brain unprotected against a variety of toxic substances, including heavy metals,³³ which can enter the fetus from the maternal blood.³³ Even after birth, the brain is unable to detoxify exogenous and lipophilic substances that can be transmitted via maternal breast milk.³³ According to the literature, mercury has been widely associated with ASD manifestation.^{7,35,36} Environmental factors should always be taken into consideration as a possible cause of the disorder, with the environmental changes that have occurred in the last 50 years resulting in human exposure to hundreds of unresearched chemical substances.^{7,36,37}

4.8. Other factors

ASD is a disorder of development that can start during pregnancy and present in the early stages of a child's life.⁹ Its genetic etiology is predominant, but environmental factors interact significantly with the genes.^{9,18} Factors that have been studied for their possible contribution to ASD, as reported in literature, are heredity, parental age, medical conditions during pregnancy (e.g., hypothyroidism), modern lifestyle, diet, obesity, alcohol, smoking, difficulties in pregnancy (e.g., premature birth, infections, medication) and others.^{9,36} In previous decades, vaccination (especially the measles-mumps-rubella [MMR] vaccine) had been suspected to be a major cause of ASD,^{9,36} but subsequent research has proved that vaccination is not associated with ASD risk.⁹

5. CONCLUSIONS

Epidemiological studies conducted thus far have documented an increase in the prevalence of ASD. Research has been conducted into the causal factors of ASD, but also into those factors that possibly contribute to its reported increase. The need for further research on this disorder is globally acknowledged, and a variety of systems for

evaluation, prevalence and conceptualization of the ASD is required. With more information available, the scientific community will be better able to understand and evaluate the needs of individuals with ASD, in order to design more effective, scientifically substantiated, interventions. Comprehensive databases and sound educational and health services are necessary for providing support to the millions of individuals with ASD and their families.

ΠΕΡΙΛΗΨΗ

Η επιδημιολογία της διαταραχής αυτιστικού φάσματος και παράγοντες που συμβάλλουν στην αύξηση του επιπολασμού της

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Η διαταραχή αυτιστικού φάσματος (ΔΑΦ) είναι μια πολυπαραγοντική νευροαναπτυξιακή διαταραχή, που έχει προσελκύσει το ερευνητικό ενδιαφέρον επιστημόνων από πολλά επιστημονικά πεδία. Ο επιπολασμός μιας νόσου ή μιας διαταραχής είναι σημαντικός για υγειονομικούς και κοινωνικοοικονομικούς παράγοντες. Σύμφωνα με τον Παγκόσμιο Οργανισμό Υγείας (ΠΟΥ) 1 στα 160 παιδιά γεννιέται με ΔΑΦ (0,63%), εκτίμηση μάλλον συντηρητική καθώς πολλές μελέτες αναφέρουν επιπολασμό για τη διαταραχή μεγαλύτερο του 1%. Ακόμη και έτσι, το ποσοστό που αναφέρει ο ΠΟΥ είναι περίπου 14 φορές υψηλότερο από την πρώτη επιδημιολογική μελέτη που διεξήχθη για τη διαταραχή, περίπου πριν από 50 έτη στο Ηνωμένο Βασίλειο. Σήμερα, εκτιμάται ότι περίπου 60 εκατομμύρια άνθρωποι σε όλον τον κόσμο έχουν ΔΑΦ. Λόγω της φύσης της διαταραχής και του μεγάλου της φάσματος, η διάγνωση και η καταγραφή των εν λόγω ατόμων έχει δυσκολίες. Πολλές χώρες του κόσμου δεν διαθέτουν επιδημιολογικά στοιχεία για τη ΔΑΦ, και αρκετές επιδημιολογικές μελέτες αναφέρουν σημαντικούς μεθοδολογικούς περιορισμούς. Ωστόσο, τα διαθέσιμα στοιχεία δείχνουν αύξηση του επιπολασμού της διαταραχής με την πάροδο των ετών. Η ερμηνεία αυτής της αυξητικής τάσης παραμένει αβέβαιη. Στη βιβλιογραφία καταγράφονται παράγοντες που πιθανόν έχουν συμβάλει στη συγκεκριμένη αύξηση, όπως είναι η αναθεώρηση των διαγνωστικών κριτηρίων, η υπερδιάγνωση της διαταραχής, η εξέλιξη των επιστημών, η προσβασιμότητα σε υπηρεσίες, η αύξηση της κοινωνικής ευαισθητοποίησης, η βελτίωση της μεθοδολογίας των επιδημιολογικών ερευνών, περιβαλλοντικοί και άλλοι παράγοντες.

Λέξεις ευρητήριο: Αυτισμός, Διαταραχή αυτιστικού φάσματος (ΔΑΦ), Επιδημιολογία ΔΑΦ, Επιπολασμός ΔΑΦ

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