

# Epidemiology of Autism Spectrum Disorder

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## Abstract

In this selective review of the literature, we present the most recent prevalence estimates for autism spectrum disorder (ASD) and discuss the limitations and challenges in interpreting changes in prevalence estimates over time. Increases in ASD prevalence estimates cannot currently be attributed to a true increase in the incidence of ASD due to multiple confounding factors. These include broader diagnostic criteria and a greater awareness of ASD. The current average prevalence of ASD is approximately 66/10,000, which translates to approximately 1 in 152 children affected, with males consistently outnumbering females by about 5:1. Several recent studies have reported higher estimates ranging from 147 (one in 68) to 264 (one in 38) per 10,000. This is in sharp contrast to the figures of about 1-5/10,000 quoted in earlier studies that used a narrow definition of autistic disorder and were not inclusive of all disorders falling onto the autism spectrum. It remains to be seen how changes to diagnostic criteria introduced in the DSM-5 will impact estimates of ASD prevalence.

## Introduction

Determining the prevalence of autism spectrum disorder (ASD) and monitoring it over time is important to ensure the training of ASD diagnosticians, improve access to necessary interventions and understand causal mechanisms of ASD. However, because ASD is a behaviorally defined disorder, determining its prevalence is more challenging than for a disorder where clear biological markers exist. The symptoms of ASD vary in severity and may present differently in children with a mixture of cognitive abilities (1). Furthermore, how the data are gathered, analyzed, and interpreted impacts the conclusions made regarding the prevalence of ASD (2). Changes in societal awareness and the public health response also have an impact on prevalence estimates. As a result, there is controversy surrounding the prevalence of ASD, in particular whether the recent rise in reports of the disorder is due to a true increase in incidence or whether there are other factors that could be impacting estimates.

## Prevalence

ASD and pervasive developmental disorders (PDDs) are used interchangeably in various articles but are very similar in meaning. In this review, we use ASD as the term of choice. Based on current best estimates of the prevalence of ASD derived from studies published in English since 2000, the current average prevalence of ASD is approximately 66/10,000, which translates to approximately 1 in 152

children affected, with males consistently outnumbering females by about 5:1 (2). Although this estimate is primarily based on studies conducted in North America and Northern Europe, a recent systematic review that included a wider representation of global prevalence estimates yielded similar figures (3). However, several studies that are notable in terms of the size of the population sampled and their methodological rigor indicate even higher estimates ranging from 147 (one in 68) to 264 (one in 38) per 10,000 (4-6). These figures are markedly higher than the earliest prevalence estimates for autistic disorder using narrow diagnostic criteria that were reported in studies conducted in the 1960s and 1970s, namely 0.7 - 4.8 per 10,000 (7-11). The progressive recognition of the importance and relevance of the milder ASD phenotypes has led to changes in the design of more recent epidemiological surveys and, as discussed below, challenges in interpreting patterns of prevalence estimates over time.

## Interpreting ASD Prevalence Estimates

The series of reports from the US Center for Disease Control and Prevention (CDC) indicates a clear rise in prevalence of ASD in recent years. News reports and some advocacy groups have called this an "Autism Epidemic," with the most recent CDC estimate (based on the 2010 surveillance year) suggesting that one in 68 children 8 years of age have an ASD – the highest to date of all the previous CDC reports (6). When prevalence estimates are considered against year of publication, there is indeed clear support for the claim for a rise in prevalence reported for ASD over time. For example, prevalence estimates from the CDC were 80/10,000 (~one in 125) in 2004 (12), 90/10,000 (~one in 110) in 2006 (12), and 113/10,000 (~one in 88) in 2008 (13), with the 2010 prevalence estimate marking a 30% increase since 2008 and roughly 120% increase since estimates from 2000 and 2002. This pattern has caused much controversy because it has generated confusion as to whether the rise in prevalence can be equated with a rise in incidence. Prevalence is the proportion of individuals in a population who have a defined disorder at any point in time, while incidence is the number of new cases occurring in a population during a certain period of time, excluding individuals already diagnosed or treated for the condition in the same period. As a consequence, causal research can only be investigated through incidence rates, whereas prevalence estimates are useful for estimating needs and planning services.

In the case of ASD, patterns in incidence cannot be implied from patterns in prevalence because they require different methodologies and analyses. Also, there are various problems with interpreting and comparing prevalence estimates that include different definitions, different methodologies, different sources and different time periods across epidemiological surveys. Awareness of autism, advocacy groups and changes in policy may also impact prevalence data, since as more people learn and seek help for autism, prevalence data will report a higher number of cases with ASD over time. These issues are each discussed in turn in the following sections.

## Case Definition

Case definition refers to the diagnostic criteria used in epidemiological studies of ASD, which have changed over time. Earlier diagnostic criteria (14-16) reflected the more qualitatively severe forms of the autism behavioral phenotype, usually associated with severe delays in language and cognitive skills. It was only in the 1980s that less severe forms of autism (i.e., Pervasive Developmental Disorders Not Otherwise Specified; PDD-NOS) were recognized, and Asperger Disorder was acknowledged as a separate diagnostic category in the early 1990s in both ICD-10 (17) and DSM-IV (18).

The changes now occurring in the DSM with the new fifth edition (19) may also impact prevalence estimates in the future. DSM-5 proposes a single new category of Autism Spectrum Disorder, conceptually equivalent to the previous diagnostic class of PDDs. However, fewer diagnostic criteria have been retained that are combined in two clusters of social communication deficits and restricted patterns of behaviors and interests. The removal of the loosely defined PDD-NOS that was in DSM-IV-TR (20) will be likely to increase the specificity of the ASD diagnostic category and the removal of Asperger Disorder as a separate category is consistent with research that has generally failed to provide evidence for the discriminant validity of this diagnostic concept vis-à-vis forms of autistic disorder that are not associated with severe language impairments or intellectual deficits.

The impact of the DSM-5 changes remain to be fully assessed in the context of epidemiological surveys. Two studies have addressed this issue. In a re-analysis of the 2008 CDC data (21), 81.2% of children classified as having ASD according to DSM-IV-TR (20) also met DSM-5 criteria (19), resulting in a DSM-5 based prevalence of 100/10,000. Kim and colleagues (22) reported that 92% of children with ASD according to DSM-IV-TR also met DSM-5 criteria. However, when DSM-5 ASD and Social Communication Disorder (SCD; a new diagnostic category in DSM-5) were considered together, there was no significant change in the prevalence estimate (22). In summary, the evolution of the case definition of autism into a spectrum disorder has created a challenge in interpreting prevalence rates across studies, especially over time (23).

## **Diagnostic Substitution and Accretion**

The modifications in the diagnostic criteria may also impact more recent prevalence estimates because it is possible that some of the cases that meet current ASD diagnostic criteria may not have received a diagnosis previously, using past diagnostic criteria. More specifically, diagnostic substitution- when a case receives one diagnosis at one point and then later receives a different diagnosis- may be playing a role (24). For example, some cases may have received a diagnosis of mental retardation when they were younger and then later received an ASD diagnosis because of the change in diagnostic criteria. It is also possible that some cases diagnosed with one disorder earlier in time may later acquire a comorbid diagnosis that includes ASD, called diagnostic accretion. For example, some cases may have received a diagnosis of mental retardation when they were younger and then later received a comorbid diagnosis including ASD.

## **Variability in Case Ascertainment Methods**

Different strategies have been also been employed to find and identify individuals meeting the case definition set for each survey. Some studies identify cases from pre-existing databases, such as service provider databases, special education databases or national registries. Utilizing these databases to estimate prevalence excludes individuals who have the disorder but are not in contact with the agency maintaining the database, resulting in an underestimation of the true prevalence. In addition, factors such as heightened public awareness, decreasing age at diagnosis, and changes in diagnostic criteria and practices may also contribute to increasing numbers of individuals in these databases, creating an illusion of an increase in ASD prevalence over time.

Other studies employ a multistage approach to identify cases in underlying populations. This approach can result in identifying individuals not previously diagnosed with ASD, enhancing the sensitivity of case identification. The goal of the screening stage is to identify an exhaustive list of cases possibly affected with an ASD, leaving the comprehensive diagnostic assessment to a second stage. The number of data sources, the screening measure selected and the response rate within the population all influence the number of cases identified in the screening stage, which then impacts the number of cases confirmed in the second stage. To confirm case status in the second stage, a combination of data from different informants (parents, teachers, doctors) and other data sources (medical or education records) is often used to evaluate diagnoses, with an in-person assessment of affected individuals in some studies. Even when participants are directly examined, assessments are conducted using various diagnostic instruments, ranging from an unstructured examination by a clinician to the use of a full battery of standardized measures such as the Autism Diagnostic Observation Schedule (ADOS) (25) or the Autism Diagnostic Interview-Revised (ADI) (26). For these reasons, all ASD prevalence estimates must be understood as underestimates of “true” prevalence rates, with the magnitude of this underestimation unknown in each survey.

## **The Public Health Response, Public Awareness, and Policy Changes**

Monitoring prevalence trends over time has become a national priority in a few countries where such estimates are used in service planning and development. In the US, research funding for ASD as well as the number of ASD research grants has increased steadily over the past decade (27). There is no doubt

that the increase in public awareness and access to services, and improved identification of ASD in primary healthcare has contributed toward the increase in prevalence and may account for regional variation within the US (6). Increasing ASD prevalence estimates have provided the impetus to focus on increasing both access to services and the quality of service, which, regardless of the nature of the increase in ASD, has been beneficial to the families and individuals affected by it.

## Conclusions and Implications

The current estimate for the prevalence of ASD is 66/10,000. Although prevalence estimates have steadily increased over time, whether this increase is the sign of a true rise in incidence ("ASD epidemic") is subject to debate. The reasons for the increase in prevalence of ASD are not well understood. Many factors such as the evolving diagnostic criteria for ASD, diagnostic substitution and accretion, variability in case ascertainment methods, and greater public awareness and services, can explain at least some of the increase in prevalence estimates of ASD. It appears as though increasing prevalence of ASD can be explained by factors associated with the collection of data, and not necessarily by an actual increase in incidence. However, the possibility that a true change in the underlying incidence of ASD has contributed to higher prevalence figures remains to be adequately tested.

## GP Comment.

### What have I learned from this paper?

The prevalence rates reported in this paper are surprisingly high. The paper highlights an important issue of diagnosis. I think as primary care providers at the front line of health care, GPs need to take note of this growing trend and the difficulties with diagnosis. Education is needed at a number of levels to raise awareness on recognizing early signs and the appropriate services which families can be signposted too.

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