The Prevalence of Autism

Article *in* JAMA The Journal of the American Medical Association · February 2003

DOI: 10.1001/jama.289.1.87 · Source: PubMed

1 author:

Eric Fombonne
Oregon Health & Science University
523 PUBLICATIONS **58,156** CITATIONS

SEE PROFILE

The Prevalence of Autism

Eric Fombonne, MD

THE NUMBER OF EPIDEMIOLOGICAL STUDIES OF AUTISM has increased in recent years, including in the United States, where investigators are now catching up in what has traditionally been a weak area of child psychiatric research in North America. In this issue of THE JOURNAL, Yeargin-Allsopp et al[1] report the findings of a survey, which was funded by the Centers for Disease Control and Prevention, that found a rate of 34 per 10 000 for autism spectrum disorders (ASDs) among 3- to 10-year-old children in metropolitan Atlanta.

The strengths of the survey include use of multiple ascertainment sources and large sample size (ie, 987 confirmed ASD cases compared with a median sample size of 50 in 32 previous studies),[2] thereby allowing the authors to have good precision in the estimates and to conduct meaningful subgroup analyses. In addition, this study is the first to derive a robust population-based estimate for the rate of ASD in black children, which is comparable to other racial groups. Other findings are typical of those found in previous surveys with ASD cases, with a strong overrepresentation of boys, cognitive impairments in more than two thirds of cases, and a relatively high rate (8%) of epilepsy. Approximately 18% of the sample did not have a previous diagnosis or were not suspected of having ASD, and children from black, younger, or less educated mothers were more often identified through schools as the only source of case finding. These findings highlight the need to rely on multiple ascertainment sources in epidemiological studies of ASD and caution against findings that are based on single service provider databases.

The prevalence rate of 34 per 10 000 is, however, likely to be an underestimate. First, as the authors point out, children with milder or high-functioning (ie, normal IQ) ASD subtypes are likely to have been missed. Second, the lower prevalence in 3- and 4-year-olds may reflect lower sensitivity of case identification among younger children for developmental disorders that often are diagnosed later. Third, there was an unexpected decrease in prevalence among 9- and 10-year-olds. Although it would be tempting to interpret this age trend as indicative of a secular increase in the rate of ASD (ie, the younger the birth cohort, the higher the prevalence), such an explanation is both unlikely and biologically implausible because rates plateaued for birth cohorts aged 5 through 8. Rather, the authors suggest that these differences might reflect new diagnostic criteria for autism and increased availability of developmental disability services for children with autism in the 1990s. What this means, however, is that the rate of 41 to 45 in 10 000 obtained for the 5- to 8-year-olds might be more accurate. This rate also is more in line with those of 3 recent surveys that yielded prevalence estimates in the range of 60 per 10 000.[3-5]

High prevalence rates from more recent epidemiological surveys have fueled the debate about a possible epidemic of autism. However, 4 separate issues need to be addressed. The first issue concerns the best current estimate for the prevalence of autism and related disorders. Increasing and consistent evidence from recent surveys shows that the prevalence rate for ASDs (including not only autism disorder but also Asperger disorder and pervasive developmental disorder–not otherwise specified) is approximately 60 per 10 000[3-5]; the study results from Yeargin-Allsopp et al concur with this conclusion. This estimate translates to approximately 425 000 children younger than age 18 years with ASDs in the United States, including 114 000 children younger than 5 years.

The second issue is whether the prevalence of ASD has increased over time. Surveys conducted in the 1960s and 1970s only dealt with autism disorder (as opposed to ASD) and with a rather narrow definition of autism, as per Kanner's descriptions,[6] and not accounting for autism occurring in subjects who are not mentally retarded. Thus, comparisons of rates over time generally deal with studies that have used different case definitions, making interpretation of time trends difficult. The closest estimate of ASD prevalence available in the late 1970s was 20 per 10 000 in a survey from the United Kingdom that was limited to the severely impaired children with ASD.[7] Comparing rates for subtypes of ASD provide another avenue for estimation over time especially for autism disorder, but as shown by Yeargin-Allsopp et al[1] and other surveys,[3-5] the breakdown in ASD subtypes is not always reli-

See also p 49.

Author Affiliation: McGill University and Montreal Children's Hospital, Montreal, Quebec.
Corresponding Author and Reprints: Eric Fombonne, Montreal Children's Hospital, 4018 St Catherine W, Montreal, Quebec, Canada (e-mail: eric.fombonne@mcgill.ca).

able. Nevertheless, rates of autism disorder in recent surveys have consistently been more than 10 per 10 000 whereas previous prevalence estimates ranged from 4 to 5 in 10 000.[2] Therefore, from the available evidence it can be concluded that recent rates for both ASD and autism disorder are 3 to 4 times higher than 30 years ago.

The third issue addresses possible interpretations of this increase in prevalence. That is, does this increase reflect a broadening of the concept of ASD with more inclusive diagnostic criteria and improved methods of case finding in population surveys? It is generally agreed that the definition of autism has been broadened over the last decades, particularly at the less severe end of the spectrum. These major changes occurred in nosology from the *Diagnostic and Statistical Manual of Mental Disorders, Third Edition* (*DSM-III*)[8] in 1980 to the *DSM-Revised Third Edition*[9] in 1987 and the *DSM, Fourth Edition*[10] in 1994. Kanner's infantile autism[6] was replaced in 1980 by the concept of pervasive developmental disorder. Among the pervasive developmental disorders, pervasive developmental disorder–not otherwise specified (or atypical autism) has now become the most widely used ASD diagnosis, and Asperger disorder emerged as a new diagnostic category in the 1990s. Unless comparisons also control rigorously for changing case definitions, interpretation of differences in prevalence rates over time and across surveys will be virtually impossible.

Moreover, there is strong evidence that differences in methods for case finding can account for a huge proportion of the variability of prevalence estimates between surveys. For example, in 4 US and 4 UK studies published recently, 14- and 6-fold variations in prevalence rates were found, respectively.[2] Although these 2 sets of studies were conducted at the same time, in similar age groups, and in the same countries, the lack of consistency in estimates is striking and demonstrates how unique design features within each study can affect the prevalence estimation. In both countries, studies relying on single administrative sources for identifying cases yielded low estimates, whereas investigations using proactive methods for case finding, that is, multiple sources of ascertainment and direct diagnostic procedures, yielded much higher rates. Needless to say, comparisons of population surveys over time are bound to be even more confounded by factors difficult to control.

Referral statistics also have been used to evaluate trends over time, but these data are confounded by changes over time in factors such as referral patterns, availability of services, public and professional awareness, age at diagnosis, and diagnostic concepts and practices. For example, the report from the Department of Developmental Services, Sacramento, Calif,[11] showed an increase in the number of children receiving public services, but it failed to adjust for key factors, such as changes in population size, diagnostic practices, or differential migration.[12] Another widely publicized report on children enrolled in this public service system concluded that "some, if not all, of the observed increase represents a true increase in cases of autism in California."[13(p42)] Yet, the authors stated earlier in this report that "Improved case finding could result in an apparent increase in the number of cases. . . . This study does not examine the extent to which differences in case finding over time have resulted in any changes in the number of autistic children who present to the Regional Centers."[13(p13)]

By contrast, a recent reanalysis of this dataset indicated that during 1987 to 1994, diagnostic substitution occurred; thus, while the prevalence of autism increased from 5.8 to 14.9 per 10 000, the prevalence for mental retardation decreased from 28.8 to 19.5 per 10 000. These trends then cancel each other.[14] According to the authors, new federal legislation (Individuals with Disabilities Education Act[15]) mandating that states provide early intervention programs for toddlers with developmental delays played a role in the increasing use of the diagnosis of autism. Moreover, in the last 15 years evidence has accumulated for the effectiveness of early intensive behavioral interventions for autism,[16] and most families could not support their high costs outside the public service delivery system. Thus, there is good evidence to support that higher prevalence rates reflect changes in diagnostic practices, improved identification and availability of services, and other similar factors.

The fourth issue involves the hypothesis of an increasing trend in the incidence of ASD. Whereas evidence exists that a substantial part of the increase in prevalence is due to methodological factors, the additional possibility of a secular increase in the incidence of autism cannot be ruled out. Unfortunately, most available epidemiological data are derived from prevalence surveys, and the few studies that provide incidence rate estimates have not been adequate to test this hypothesis. In addition, no strong candidate environmental exposures have been identified. Claims of an association with measles-mumps-rubella immunization have not been borne out by recent studies,[17-19] and evidence for causal association with other exposures, such as mercury-containing vaccines, is weak.[20,21]

Extending the already substantial research effort, the Centers for Disease Control and Prevention has recently funded a surveillance network across several states.[22] This and other initiatives should help address more directly hypotheses about secular changes in the incidence of ASDs.

Finally, the current social context seems to exert a stronger influence on the debate than the scientific arguments. Although claims about an epidemic of autism and about its putative causes have the most weak empirical support, the subsequent controversy has put autism on the public agenda. In recent years, children with autism, their families, and professionals involved in their care and in research have seen welcome and legitimate increases in public funding. Yet, ironically, what has triggered substantial social policy changes in autism appears to have little connection with the state of the science. Whether this will continue to be the case in the future remains to be seen, but further consideration should

be given to how and to why the least evidence-based claims have achieved such impressive changes in funding policy.

REFERENCES

1. Yeargin-Allsopp M, Rice C, Karapurkan T, Doernberg N, Boyle C, Murphy C. Prevalence of autism in a US metropolitan area. *JAMA*. 2003;289:49-55.
2. Fombonne E. Epidemiological trends in rates of autism. *Mol Psychiatry*. 2002; 7(suppl 2): S4-S6.
3. Baird G, Chairman T, Baron-Cohen S, et al. A screening instrument for autism at 18 months of age: a 6 year follow-up study. *J Am Acad Child Adolesc Psychiatry*. 2000;39:694-702.
4. Chakrabarti S, Fombonne E. Pervasive developmental disorders in preschool children. *JAMA*. 2001;285:3093-3099.
5. Bertrand J, Mars A, Boyle C, Bove F, Yeargin-Allsopp M, Decoufle P. Prevalence of autism in a United States population: the Brick Township, New Jersey, investigation. *Pediatrics*. 2001;108:1155-1161.
6. Kanner L. Autistic disturbances of affective contact. *Nervous Child*. 1943;2: 217-250.
7. Wing L, Gould J. Severe impairments of social interactions and associated abnormalities in children: epidemiology and classification. *J Autism Dev Disord*. March 9, 1979:11-29.
8. American Psychiatric Association. *Diagnostic and Statistical Manual of Mental Disorders, Third Edition*. Washington, DC: American Psychiatric Association; 1980.
9. American Psychiatric Association. *Diagnostic and Statistical Manual of Mental Disorders, Revised Third Edition*. Washington, DC: American Psychiatric Association; 1987.
10. American Psychiatric Association. *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition*. Washington, DC: American Psychiatric Association; 1994.
11. Department of Developmental Services. Changes in the population of persons with autism and pervasive developmental disorders in California's Developmental Services System: 1987 through 1998. Report to the Legislature March 1, 1999:1-19. Available at: http://www.dds.ca.gov: 1999. Accessed December 11, 2003.
12. Fombonne E. Is there an epidemic of autism? *Pediatrics*. 2001;107:411-413.
13. Report to the Legislature on the Principal Findings from the Epidemiology of Autism in California. *A Comprehensive Pilot Study*. Davis, Ca: M.I.N.D. Institute, University of California, Davis; October 17, 2002.
14. Croen LA, Grether JK, Hoogstrate J, Selvin S. The changing prevalence of autism in California. *J Autism Dev Disord*. 2002;32:207-215.
15. The Education for All Handicapped Children Act of 1975. Pub L No. 94-145, 20 USC 1401 et seq. *Federal Register*. August 23, 1977; 42(163):42474-42518.
16. Rogers S. Empirically supported comprehensive treatments for young children with autism. *J Clin Child Psychol*. 1998;27:168-179.
17. Fombonne E, Chakrabarti S. No evidence for a new variant of measles-mumps-rubella-induced autism. *Pediatrics*. 2001;108:E58.
18. Madsen KM, Hviid A, Vestergaard M, et al. A population-based study of measles, mumps, and rubella vaccination and autism. *N Engl J Med*. 2002;347:1477-1482.
19. Taylor B, Miller E, Lingam R, Andrews N, Simmons A, Stowe J. Measles, mumps, and rubella vaccination and bowel problems or developmental regression in children with autism: population study. *BMJ*. 2002;324:393-396.
20. Stratton K, Gable A, McCormick MC, eds. *Immunization Safety Review Committee: Thimerosal-Containing Vaccines and Neurodevelopmental Disorders*. Washington, DC: National Academies, Institute of Medicine; 2001.
21. Pichichero ME, Cernichiari E, Lopreiato J, Treanor J. Mercury concentrations and metabolism in infants receiving vaccines containing thiomersal: a descriptive study. *Lancet*. 2002;360:1737-1741.
22. Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities, Autism and Developmental Disabilities Monitoring Network. Available at: http://www.cdc.gov/ncbddd/dd/aic/states/default.htm#addm. Accessed December 10, 2002.

Improving Reports of Studies of Diagnostic Tests

The STARD Initiative

Drummond Rennie, MD

AUTHORS PREPARING REPORTS FOR THE JOURNAL ON the accuracy of diagnostic tests will notice a change in our Instructions for Authors.[1] They will be referred to a checklist and flow diagram for a study of diagnostic accuracy on the *JAMA* Web site (http://www.jama.com) and to a statement about this checklist and flow diagram,[2] as well as a background document,[3] both of which are being published simultaneously in *Clinical Chemistry* and are available without charge at http://www.clinchem.org/cgi/content/full/49/1/1 and http://www.clinchem.org/cgi/content/full/49/1/7. A statement will also be published in other journals in laboratory medicine, as well as in *Radiology*, another journal that often publishes evaluations of tests, and it is free from copyright restrictions. Those desiring to do so may publish and use it freely.

In 1999, *JAMA* published a report by Lijmer et al[4] that drew attention to the poor state of such reporting, many such reports lacked essential information on their design, conduct, and analysis, thus rendering them excessively positive about the test, and frequently invalid. The Cochrane Diagnostic and Screening Test Methods Working Group noted the poor quality of the methods and reporting of evaluations of diagnostic tests evaluations at the 1999 Cochrane Colloquium in Rome, Italy.

As a result, Jeroen Lijmer, and subsequently Patrick Bossuyt, both of the University of Amsterdam, organized an attempt to produce standards for reporting, and the STARD (Standards for Reporting of Diagnostic Accuracy) group was formed. It consists of 25 members, who are an international ensemble of researchers, methodologists, statisticians, journal editors, and representatives of professional organizations. A 2-day consensus conference was held, and the STARD documents were subsequently developed after extensive discussion and modification.

Readers will be reminded of the CONSORT statement, which describes those essential features the absence of which has been shown to materially affect the quality of reporting

Author Affiliations: Dr Rennie is Deputy Editor, *JAMA*.
Corresponding Author and Reprints: Drummond Rennie, MD, *JAMA*, 515 N State St, Chicago, IL 60610 (e-mail: Drummond_Rennie@ama-assn.org).