VIEWPOINT

Population-based studies key for assessing autism prevalence

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Studies of developmental disorders, including autism, have been plagued withinconsistent findings and inadequate replications. This problem affects a broad spectrum of research in autism, ranging from **prevalence** to genetics and environmental risk studies, and is extremely frustrating for both the scientific and lay communities.

For example, over the past four decades, researchers have conducted more than **50 studies of autism prevalence**, in many countries and on every inhabited continent, with the exception of **Africa**. Studies conducted since 1985 have reported progressively higher autism prevalence estimates, ranging from as few as 7 to as many as 157 affected individuals per 10,000[1].

Large variations in reported prevalence exist even within the same country. For example, four studies conducted over the past ten years in the U.K. reported autism prevalence estimates as 0.1 percent using administrative records, such as registries of special-needs children[2], 0.6 percent when researchers screened and assessed preschool-aged children from one town in the U.K.[3], 1.2 percent when researchers screened and examined 9- to 10-year-old children who were highly likely to have autism, including those with special needs, developmental delays or who had used psychiatric services[4] and 1.6 percent when researchers screened children from both mainstream and special-education schools[1].

It is surprising, if not shocking, that autism prevalence in the U.K. alone has risen by 16-fold over only ten years. How is this possible?

Puzzling pieces:

Researchers have attributed variations and inconsistencies in prevalence estimates to many causes, including differences in how autism is diagnosed, diagnosis of younger children and varying degrees of public awareness. However, they have not yet examined one very important source of variation: the differences among target populations used for the various studies.

As previously noted for the U.K. studies, prevalence estimates from administrative datasets, which include children with autism previously identified in clinical or educational settings, are consistently lower than those from studies that include an in-person assessment. Autism prevalence estimates are also lower in studies that include only those individuals at high risk of having developmental disorders, such as people who use psychiatric or educational services, compared with studies that include children in the general population.

For the sake of saving both cost and time, almost all autism research to date has relied upon clinical samples. However, this convenience comes at a price in sampling errors. Clinical samples are an important part of the disease profile, but they do not represent the entire spectrum of the disorder, especially in the case of autism. In other words, clinical samples are a small part of the larger population.

This makes extrapolating autism's prevalence from just clinical samples exceedingly difficult, not unlike putting together a puzzle without knowing the entire picture or context from which the puzzle pieces came. We will surely arrive at some findings, but there is also a good chance that what we find will be imprecise, inaccurate or even invalid.

We learned the importance of choosing a representative epidemiological sample in our **Korean study**, which we conducted in response to concern over reported increases in autism prevalence. These increases had persisted even when the diagnostic criteria remained stable and in areas where public awareness had been consistently high. However, no one had examined whether differences among the study populations in the previous studies could be the source of these increases.

To address this, we used a total population approach. We included a clinical population of children with autism who had previously been identified by service systems, and what we dubbed a 'non-clinical population' of children who had never received services.

We screened about 25,000 children in the Ilsan district of Goyang City in South Korea using parent or teacher reports on the Autism Spectrum Screening Questionnaire. We then confirmed the diagnosis using the Autism Diagnostic Observation Schedule, Autism Diagnostic Interview-Revised and cognitive testing to directly assess the children who screened positive.

Hidden group:

In a paper published last year in *The American Journal of Psychiatry*, we reported a 2.64 percent estimate for autism prevalence[5]. But close examination of our data shows that the prevalence in the clinical sample is 0.8 percent, which is comparable to prior studies in the U.S. and Europe that also relied on clinical populations.

We reported a 1.8 percent prevalence in the non-clinical sample. In short, this means that the 'new

increased prevalence' in our study came from the 1.8 percent, or previously unrecognized and untreated children with autism in the non-clinical population — a study population that had largely been neglected in previous studies.

Our study methods and findings have also allowed us to identify distinct differences between individuals with autism in the clinical group and those in the non-clinical group.

The majority of children with autism in the clinical sample have autistic disorder with severe symptoms. They have a mean intelligence quotient (IQ) of 75 compared with 100 for the general population, and 60 percent have intellectual disability. They also have a mean adaptability level of 38, as measured by the parental report of the Behavior Assessment System for Children (BASC), compared with the population average of 50. BASC assesses a child's ability to adapt to various situations. In this group, roughly five times as many boys are affected by the disorder as girls**,** which is in line with the **four-to-one gender ratio** reported by most studies.

In striking contrast, in the children in the non-clinical sample are diagnosed with types of autism other than autistic disorder, such as **Asperger syndrome** and pervasive developmental disorder–not otherwise specified, and have lower levels of severity. Their mean performance IQ is 98, with only 16 percent showing a mild cognitive deficit, a BASC mean adaptability level of 43 and a gender ratio of 2.5 boys to girls.

Despite the fact that they appear to be generally higher-functioning, children in the non-clinical sample are still less able than the general population to adapt to different situations, especially in a social context, according to the BASC adaptability scale.

Our data suggest that previous studies missed a major proportion of individuals with autism who have a distinct profile. This is likely to have important implications for analyses of the underlying causes of the disorder.

In summary, researchers must carefully consider the choice of study subjects to understand etiology, symptoms, natural history and interventions. They must start out with a study group that is representative of a source population and of the entire distribution of autism.

Without a rigorous epidemiological study of the disorder, it is nearly impossible to place individual cases, specific symptom characteristics or disease mechanisms in proper perspective. The population-based epidemiological sample should include both clinical and non-clinical samples of autism to allow for valid inferences and to permit generalization to other and larger populations. Finally, researchers should replicate results with multiple, independent studies.

This is an exciting time for autism research: As long as we are methodologically rigorous and careful, we will see rapid advances, with the potential to identify causes, treatments and even ways to prevent the disorder.

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References:

1: Baron-Cohen S. *et al. Br. J. Psychiatry* **194**, 500-509 (2009) **PubMed**

2: Taylor B. *et al. Lancet* **353**, 2026-2029 (1999) **PubMed**

3: Chakrabarti S. and E. Fombonne *Am. J. Psychiatry* **162**, 1133-1141 (2005) **PubMed**

4: Baird G. *et al. Lancet* **368**, 210-215 (2006) **PubMed**

5: Kim Y.S. *et al. Am. J. Psychiatry* ***168****, 904-912* (2011) **PubMed**