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Review Article

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A New Direction for Autism Research

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Abstract

At first glance, autism prevalence appears to have increased from 0.62 percent in 2012 to 1.85% in 2020. However, during this same 8-year period there has been no reported increase in prevalence for any of the other disorders listed in the DSM-5. This threefold increase in autism diagnoses coincides with the 2013 creation of the autism spectrum disorder (ASD) umbrella category under which are now included a range of confusingly dissimilar conditions. It thus appears that two-thirds of those who have been diagnosed with ASD may not be autistic. If so, then recent research into the genetic causes and risks for autism may be questionable. There is no way of knowing if the correlations researchers are finding are to autism or to other conditions.

Introduction

In 2020, the Centers for Disease Control (CDC) reported that 1 in 54 children is diagnosed with an autism spectrum disorder, for a prevalence rate of 1.85% of the population [1]. A 2012 review of global prevalence estimates of autism spectrum disorders found 62 cases per 10,000 people, for a prevalence rate of 0.62 percent [2]. This apparent 300 percent increase in autism prevalence is in stark contrast to all other disorders listed in the Diagnostic and Statistical Manual of Mental Disorders (DSM-5), for which there has been no reported increase in prevalence over this same eight-year period.

A 10-year Swedish study in 2015 concluded that although the prevalence of the autism phenotype has remained stable, clinically diagnosed autism spectrum disorder has increased substantially [3]. A 2016 study reported that many children originally diagnosed with autism spectrum disorder were later found not to be autistic [4]. A comprehensive 2019 study in JAMA Psychiatry indicates that autism is being significantly over diagnosed [5].

If the increase in prevalence of autism from 2012 to 2020 is due entirely to misdiagnoses, this suggests that two-thirds of those diagnosed with autism spectrum disorder (ASD) are not in fact autistic. The implications for autism research are profound.

Overly Broad Definition of Autism

The epidemic of autism diagnoses began in 2013, the year that the American Psychological Association created the autism spectrum disorder (ASD) umbrella category under which are included a range of conditions of uncertain similarity. Professionals diagnose by ticking off subjective symptoms on a checklist, without questioning the possible causes of said symptoms. This spectrum idea falsely implies that there can be different kinds or varying degrees of autism. Recent neurophysiological research clearly establishes that this is not so. There is only one autism and it is 100 percent. A person is either autistic or s/he is not [6, 7].

Autism is neither neurodevelopmental nor a disorder. It is simply an inherent neurophysiological difference in how the brain processes information [6-8]. Because autism is inheritable, there must be a genetic component that is responsible for this neurophysiological idiosyncrasy. Unfortunately, research into genetic identifiers for autism cannot succeed if it falsely assumes that autism is a heterogeneous mixture of complex neurodevelopmental factors. Four studies between 2016 and 2019attempting to pinpoint a specific DNA marker end up confusing rather than resolving the issue [9-12]. Only if researchers isolate cases of true autism from the misleading spectrum can any headway be made in this direction.

A 2020 study claims to have found in blood samples unique metabolic signatures in 53 percent of children on the autism spectrum [13, 14]. These researchers departed from DNA investigation and instead linked 34 biochemical markers to children who had been diagnosed according to overly broad ASD criteria. This research suffers from three fatal flaws: (1) autism is an entirely neurophysiological phenomenon that is completely unrelated to biochemistry; (2) no control group without ASD was examined for the prevalence of these same biomarkers; and (3) a 53 percent correlation is statistically insignificant. A coin toss yields a 50 percent correlation.

Redefining Autism

The unique and defining characteristic of autism is hyperfocus, the perpetual and unrelenting state of intense single-minded concentration fixated on one thing at a time, to the exclusion of everything else, including one's own feelings. The suspected cause of hyperfocus is dysfunction of the cingulate gyrus (CG), that part of the brain which focuses attention [6, 7].

Conclusion

Research needs to focus on isolated cases of true autism. All subjects being studied must be documented to function with perpetual hyperfocus, the defining characteristic of autism. Then and only then can a meaningful correlation be found between autism and its genetic markers. Perhaps this research may also discover the nature of the dysfunction of the CG that causes autistic hyperfocus.

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