

When Autistic Behavior Suggests a Disease Other than Classic Autism



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KEYWORDS

- Autism spectrum disorder • Specific language impairment
- Social (pragmatic) communication disorder • Anxiety disorder • Selective mutism

KEY POINTS

- Most neurodevelopmental disorders are defined by their clinical symptoms and many disorders share common features.
- In recent years, there has been an increase in the number of children diagnosed with autism spectrum disorder, although concerns have been raised about the accuracy of the reported prevalence rates.
- This article reviews the essential features of autism spectrum disorder and describes other conditions that may include similar symptoms that may be misdiagnosed as autism spectrum disorder (primary communication disorders, anxiety disorders, attachment disorders, intellectual disability, vision and hearing impairment, and normal variations).
- An approach to differential diagnosis is discussed with particular attention to evaluation of young children.

There has been a significant increase during the past decade in the number of children diagnosed with autism spectrum disorders (ASD). A disorder once thought to be exceedingly rare is now estimated to affect 1 in 45 (2.2%) US children. Public awareness campaigns by advocacy groups, testimonials by celebrities with affected children, and increased funding for treatment programs and research have also contributed to parents' and professionals' better recognition of children with this condition. However, concerns have been raised about the accuracy of the prevalence figures and the dramatic increase in autism diagnoses over time.¹⁻⁴

Most neurodevelopmental and behavior disorders are defined by their clinical symptoms and many disorders share common features. The key to accurate

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differential diagnosis is identifying the core, or essential, symptoms that help distinguish one condition from others that have overlapping symptom manifestations.⁵ No single behavior or category of behavior is characteristic (pathognomonic) of ASD. To the contrary, there is evidence that some of the central features of ASD are fractionable (ie, they occur individually at fairly high frequency in the normal population even to the extent seen in individuals with ASD).⁶ A further complication arises with the use of the term “spectrum.” The American Psychiatric Association Diagnostic and Statistical Manual of Mental Disorders-5 (DSM-5) clearly intended use of this term as a way to account for differences in symptom severity between individuals, and changing manifestations related to developmental level and chronologic age. Thus, it is often the unique clustering of symptoms that defines a specific disorder.

This article provides information about ASD and other disorders that share common symptoms that should be considered as part of the differential diagnosis of a child with a developmental disorder (**Box 1**). It is not sufficient when screening children with early signs of ASD to simply rule in or rule out an ASD diagnosis. The screening must encompass consideration of which diagnoses could best account for the child’s symptoms and what interventions or treatments will be most effective.

AUTISM SPECTRUM DISORDER

The most recent revision of the DSM-5 identifies two core domain deficits in ASD. The first involves a persistent impairment in social communication and social interaction across multiple contexts. Individuals with ASD have significant difficulty in three areas of social functioning: (1) social-emotional reciprocity; (2) using nonverbal communicative behaviors for social interaction; and (3) developing, maintaining, and understanding relationships. DSM-5 specifies that deficits must be present in all three areas. The second domain involves restricted, repetitive patterns of behavior, interests, or activities (RRBIs) manifesting in at least two of the following categories of behavior: stereotyped or repetitive motor movements, use of objects, or speech; insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior; highly restricted, fixated interest that are abnormal in intensity or focus; and hyperactivity or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment. RRBIs must be present in the early

Box 1

Conditions commonly misdiagnosed as autism spectrum disorder

- Primary communication disorder
 - Specific language impairment
 - Social (pragmatic) communication disorder
- Anxiety disorder
 - Selective mutism
- Reactive attachment disorder
 - Postinstitutional autistic syndrome
- Cognitive impairment
- Visual impairment
- Hearing impairment
- Normal behavioral variations

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