

# New findings validate the accuracy of autism diagnosis in children with Down syndrome

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New findings from a 16-year study confirm that the Diagnostic and Statistic Manual of Mental Disorders (DSM), the gold-standard for the classification of mental health conditions, can be used to accurately identify autism spectrum disorders (ASD) in children with Down syndrome, according to research from Kennedy Krieger Institute.

The DSM is used by a wide range of health professionals across clinical and research settings. Previously, the diagnosis of [autism](#) in children with [Down syndrome](#) has been questioned because of the presence of cognitive impairments in these individuals, despite estimates that 10 to 15 percent of children with Down syndrome are affected by both disorders. Autism-like behaviors are often difficult to differentiate from [repetitive behaviors](#), communication difficulties and other cognitive delays associated with intellectual disability. Because of these challenges, physicians often hesitate to diagnose ASD in children with Down syndrome, leaving them unable to receive important therapy and educational services.

"Based on our findings, I encourage parents of children with Down syndrome who display difficulty with [social interaction](#) to ask their providers about the presence of autism or other [intellectual disabilities](#)," says Dr. Walter E. Kaufmann, senior study author and director of Kennedy Krieger's Center for Genetic Disorders of Cognition and Behavior. "Our results will significantly help clinicians in categorizing co-morbidities and support them in developing more targeted educational and intervention services for children with Down syndrome."

Published in the *Journal of Intellectual Disability Research* [Epub ahead of print], the study confirmed the unbiased validity of the DSM for identifying autism in children with Down syndrome by comparing diagnoses based on its criteria with scores from a separate tool, the Aberrant Behavior Checklist – Community (ABC-C).

The ABC-C is one the most commonly used tools for evaluating [behavioral problems](#) in individuals with intellectual disabilities and has no relation to the DSM. The 58-item, parent-completed, clinician-interpreted checklist assesses the severity of non-productive behaviors.

"The behavioral diagnosis of autism will probably always be more complicated than a simple blood test," says Dr. Kaufmann. "However, with this study, we're able to use objective analytical methods to support the accurate diagnosis of autism in children with Down syndrome so that they can receive proper treatment and care."

Researchers used the ABC-C scores to confirm or deny initial DSM-based autism diagnoses in 293 patients from the Institute's Down Syndrome Clinic. The researchers were able to accurately distinguish three diagnostic groups:

- Down syndrome (DS): children with DS, but no significant behavioral problems that would warrant the diagnosis of a coexisting mental health condition
- DS + ASD: children with DS and [autism spectrum disorders](#)
- DS + Disruptive behavior disorder (DBD): children with DS and DBD, characterized by temper tantrums, physical aggression, excessive argumentativeness, stealing and other forms of defiance or resistance to authority

"The most telling result of our study is that we were not only able to

clearly identify patients with Down syndrome and ASD, but also distinguish two other behavioral categories – those without major behavioral problems and those with disruptive behavior disorder (DBD), a widely recognized group among individuals with Down syndrome," says Dr. George Capone, study author and Director of the Down Syndrome Clinic at Kennedy Krieger.

Capone initiated the study in 1992 when few biomedical researchers were interested in studying the connection between the two disorders. Remarkably, the parents of patients in Kennedy Krieger's Down Syndrome Clinic joined Capone in his effort by funding the study with their individual gifts. Capone recalls, "Parents would travel from around the country and the world to be seen here because few other institutions were committed to understanding this dual-diagnosis. Once we gave these families validation that their child was indeed different and required different support strategies than a typical child with Down syndrome, they wanted to help other families access this knowledge."

The findings suggest that a strategy similar to what researchers employed in this study could be used for confirming the diagnosis of ASD in other genetic disorders. Kaufmann says that the research also demonstrates that the ABC-C can be used as a supportive resource for making the diagnosis of ASD and other behavioral problems in Down syndrome. His hope is that this study provides the evidence clinicians need to confidently use the DSM to diagnose ASD in Down syndrome, ensuring better outcomes for affected [children](#) and families.

Researchers also note an unexpected finding: Down syndrome patients with severe autism behaviors were much more likely to have experienced a later onset of ASD. More studies are needed to determine the cause of this phenomenon, which may provide researchers a glimpse into the genetic associations behind autism and Down syndrome.

Provided by Kennedy Krieger Institute

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