

## Ask a Researcher: Can a Person with Down Syndrome Also Have Autism?

Autism spectrum disorder (ASD) and Down syndrome (DS) are neurodevelopmental conditions that are both variable in how they present and share some overlapping characteristics. While it was once believed that individuals with DS could not develop ASD, we now know that these conditions can co-occur. Estimating the prevalence of ASD among children and adults with Down syndrome is difficult. In part, this is due to changes in diagnostic criteria and limited documentation of occurrence over the years. Currently, estimates vary between 5 and 39%<sup>1</sup>. Despite this wide range, it is clear that people with DS are more likely to have ASD than the general population.



Down syndrome is the most common genetic disorder in the United States, affecting about 1 in 800-1000 babies<sup>2</sup>. It occurs when a person has an extra copy of chromosome 21. Down syndrome is diagnosed through genetic testing which detects the additional chromosomal material associated with DS. It is possible to screen for DS prenatally and to diagnose DS before birth using prenatal testing through amniocentesis or chorionic villus sampling (CVS), or after birth following an exam and confirmation with a blood test. In contrast, the causes and diagnosis of ASD are much more complex and there is currently no blood test for autism. Autism spectrum disorder is diagnosed by observing and assessing a child's behavior and developmental history. While it's possible for specialists to diagnose ASD in someone as young as 2 years of age, some autistic people don't receive a diagnosis until they're much older. Making the diagnosis in someone with DS can be particularly challenging given the many shared traits between these conditions.

### Early Signs: What to Look For

Understanding the early signs of ASD in children with DS can make a significant difference in accessing appropriate tailored care and support services. The core components of an ASD diagnosis include challenges in social interaction and communication, as well as restricted and repetitive interests and behaviors, such as lining up or spinning objects repeatedly and non-intended play with toys. These symptoms can sometimes be difficult to detect when they coincide with a genetic condition like DS.

While many aspects of ASD appear similarly in children with and without DS, children with DS-ASD may exhibit behaviors distinct from those with DS or ASD alone. For example, they might have fewer social communication impairments compared to children with ASD alone, but more than children with DS alone<sup>3</sup>. Some behaviors that caregivers initially brought to the attention of their healthcare providers are stereotypy, severe communication difficulties, and behavioral challenges<sup>1</sup>. Here are some signs to look for in loved ones who may have ASD along with DS<sup>4,5</sup>. Keep in mind that these signs can sometimes be present in individuals with DS alone, so discussing any concerns you have with a healthcare provider is important:

*Social and communication challenges:*

- Displays little engagement with other people
- Struggles to try to get the attention of others and communicate wants
- Does not use words, point, or gesture
- Seems to make little eye contact with others
- Uses limited gestures and facial expressions
- Shows a loss of communication or social skills

*Restricted behaviors and repetitive interests:*

- Demonstrates repetitive play with toys that is not intended, like lining toys up or playing with a specific part of a toy rather than the whole toy
- Often presents with repetitive movements that are not purposeful and can be disruptive
- Creates unusual or repetitive sounds or makes statements that are not appropriate to the context

*Sensory sensitivities:*

- Displays extra sensitivity to loud noises, physical affection, smells, tastes, textures, and grooming tasks

*Behavioral difficulties:*

- Has tantrums often
- Runs away randomly
- Has self-harming behaviors, like head banging, skin picking or biting
- Struggles extremely with changes to routine

Compared to peers with DS only, a person with DS-ASD may have greater <sup>6,1</sup>:

- Behavioral challenges
- Difficulties with everyday personal care and independent living skills
- Communication impairment
- Mood concerns
- Stereotyped behavior, like hand flapping or head banging
- Perseverative behavior, like staring at ceiling fans or lights
- Repetitive language, like saying the same phrase repeatedly, echoing others, or frequently using scripted phrases from TV, movies, books, etc.
- Overactivity

If a parent or a caregiver is noticing signs of ASD in their child, they should raise their concerns with their pediatrician or healthcare provider. A specialist, such as a developmental and behavioral pediatrician or a clinical neuropsychologist, can do a detailed assessment and determine if there is an underlying ASD diagnosis, or may identify other contributing factors, such as obsessive compulsive disorder (OCD) or anxiety.

**What are the challenges of obtaining a dual diagnosis?**

Getting a dual diagnosis of DS-ASD can be challenging because some symptoms of ASD overlap with typical behaviors seen in DS. For example, stereotyped behaviors are common in both intellectual disability and ASD, which can complicate the diagnostic process<sup>7</sup>. The distinct

presentation of ASD in individuals with DS compared to those with ASD alone can also create challenges. Due to these complexities, caregivers often experience a larger gap between initial concerns and receiving an official ASD diagnosis for children with DS<sup>1</sup>. It is not unusual for multiple types of providers to be consulted before a diagnosis is made.

### **Why is it important to receive a diagnosis of ASD for a child with Down syndrome?**

Research highlights the benefits of early diagnosis and intervention for ASD in overall development<sup>8</sup>. Early identification of ASD in a child with DS can open doors to supports and services that better address the child's unique needs. For instance, applied behavioral analysis (ABA) therapy, a common intervention for children with ASD, is more effective when started early. Regardless of how the diagnosis impacts care, understanding a child's behaviors, emotions, strengths, and challenges through a DS-ASD diagnosis can lead to positive outcomes overall.

### **Why are individuals with Down syndrome more likely to have ASD than the general population?**

It is not fully understood why individuals with DS have a greater likelihood of being diagnosed with ASD. The presence of the extra chromosome in DS affects brain development and function, which may increase the susceptibility to ASD. The genes on chromosome 21 may also interact with other genetic factors that contribute to ASD. Additional genetic variants have been shown to play a role in the development of ASD in individuals with DS, but this relationship remains unclear and more research is needed<sup>9</sup>. In addition, individuals with DS often have unique neurological and structural brain differences that can overlap with those seen in individuals with ASD<sup>10</sup>. These differences can influence behaviors and developmental patterns associated with ASD. Furthermore, increased awareness and improved diagnostic criteria have led to better recognition of ASD in individuals with DS. In the past, behaviors attributed solely to DS might have masked an underlying ASD diagnosis. As healthcare providers become more adept at distinguishing between these conditions, the prevalence rates reflect a more accurate picture.

It's important to note that while these factors contribute to a higher likelihood of ASD in individuals with DS, each person is unique, and the presence of one condition does not automatically imply the presence of the other. Comprehensive evaluations by healthcare professionals are essential for accurate diagnosis and appropriate support.

### **Additional resources:**

- [The Down Syndrome Program](#) at Massachusetts General Hospital
- [Down Syndrome – Autism Connection](#)
- *When Down Syndrome and Autism Connect: A Guide to DS-ASD for Parents and Professionals* by Margaret Froehlke and Robin Zaborek

## **About the Authors**

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## **Related ongoing research at MGH:**

### **Developing a Pediatric Health Survey at Massachusetts General Hospital (MGH)**

#### **What's the study about?**

Primary caregivers are asked to participate in a survey about the health of individuals with autism spectrum disorder (ASD). The survey collects information on the social, emotional, and physical health of the individual with ASD. This information will be used to create a standardized health instrument for doctors everywhere to better assess the health of their pediatric patients. We need to accurately measure the health of all individuals, and there is a lack of population-specific tools available. Creating such an instrument will provide a barometer of the current state of health for specific pediatric populations and hold use in future research.

#### **Who can participate?**

- You must be the primary caregiver of an individual with ASD (and the individual must be <22 years of age and not have a diagnosis of Down syndrome)
- You, the caregiver, must be ≥ 18 years of age, be fluent in written and spoken English, and be able to read and provide informed consent.
- Participation is completely voluntary, and you have the right to withdraw at any time.

#### **What will participants be doing?**

Primary caregivers will complete a survey about the health of the individual with ASD. The survey is virtual and will take around 45 minutes to complete. You will be reimbursed for your time with a \$25 gift card.

**WHERE?** <https://tinyurl.com/29nskzxd>

## References

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