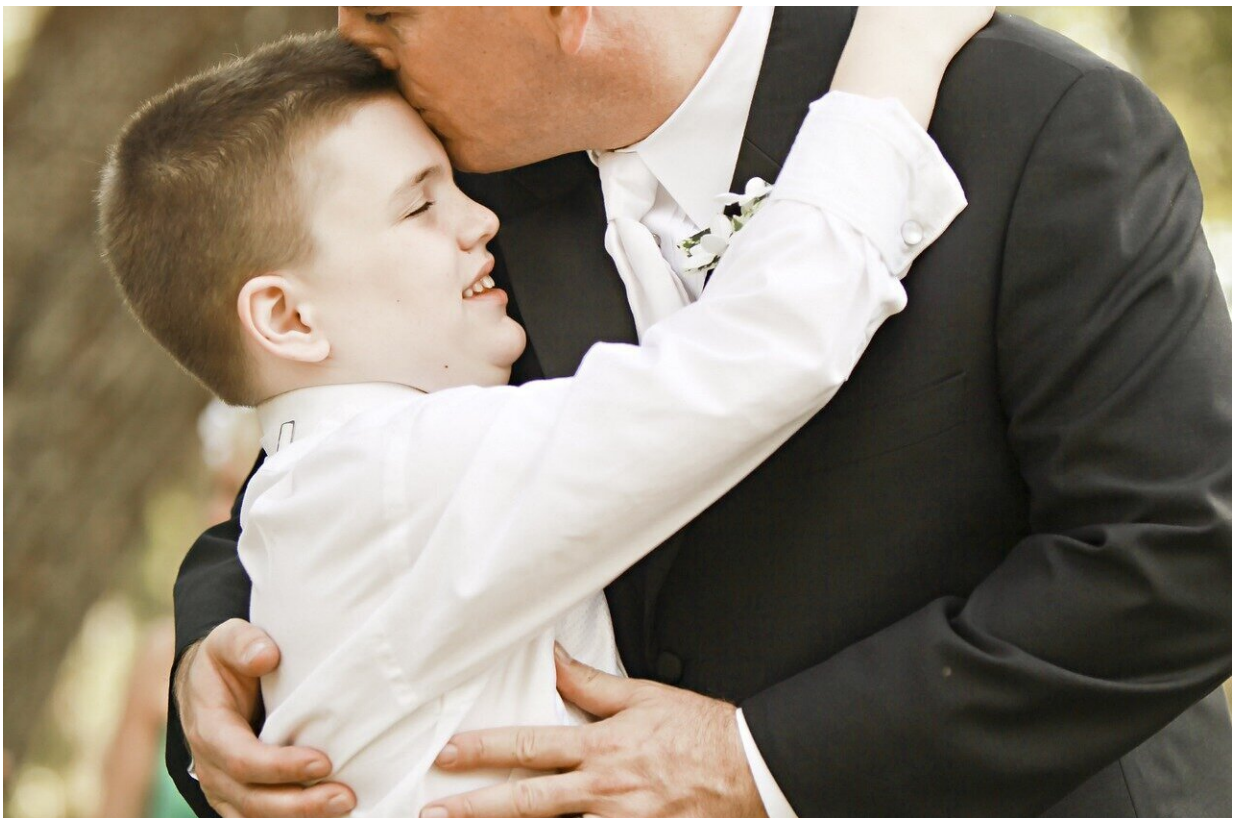


# Underdiagnosis of autism in children with fragile X syndrome reveals need for better education, early screening

August 29 2023, by Erin Bluvas

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Autism spectrum disorder is a much more prevalent co-occurring condition in individuals with fragile X than previously realized,

according to new research. It is also underdiagnosed in community settings.

Published in *Research on Child and Adolescent Psychopathology*, principal investigator Jessica Klusek and her team conducted comprehensive, best-practice clinical autism diagnostic assessments in [young males](#) (ages 15-24) with fragile X syndrome (i.e., the most common single-gene cause of autism spectrum disorder and other intellectual disabilities). They found that, although 75 percent of participants met criteria for autism through the research protocol, only 31 percent had been identified as having autism in the community.

## Underdiagnosed autism

"These findings demonstrate a gross under-identification among male youth with fragile X syndrome," says Klusek, an associate professor of communication sciences and disorders who studies the genetic mutation's communication and cognitive effects on those who have it and their mothers who carry the FMR1 premutation that causes it.

"Individuals with both fragile X syndrome and autism spectrum disorder are at risk for poorer education, medical, employment and independent living outcomes, so [early diagnosis](#) is essential to providing access to services that improve their success and quality of life in these areas."

One in every 36 children has been identified with autism spectrum disorder, according to the Centers for Disease Control and Prevention, but this condition is even more prevalent in children with certain [genetic syndromes](#), such as fragile X syndrome which affects one in 7,000 boys and one in 11,000 girls. Klusek's research shows that about 75 percent of boys with fragile X meet criteria for co-occurring autism.

However, without a genetic or other lab test for autism, clinical and

educational providers rely on their own assessments to identify the disorder, which can pose challenges. The features of fragile X syndrome and autism spectrum disorder are often similar, leading to misattribution of observed symptoms by parents, teachers and clinicians.

In Klusek's study, the researchers found that of the participants who met the clinical standards for autism spectrum disorder, 60 percent had not been previously diagnosed within their communities. Forty percent of the participants had never been evaluated for autism at all.

## Recognizing the signs

Delays in diagnosing [autism](#) spectrum disorder—especially when other conditions such as fragile X syndrome are present—present major barriers for these children and the families who care for them, particularly mothers who may be facing their own challenges as carriers of the FMR1 premutation.

"With 75 percent of our participants experiencing both fragile X syndrome and [autism spectrum disorder](#), it's clear that co-occurrence is common," Klusek says. "Education about these distinct yet overlapping conditions is key to early diagnoses that can then lead to the [support services](#) that are essential to improved outcomes and quality of life."

**More information:** Jessica Klusek et al, Predictors, Parental Views, and Concordance Across Diagnostic Sources of Autism in Male Youth with Fragile X Syndrome: Clinical Best Estimate and Community Diagnoses, *Research on Child and Adolescent Psychopathology* (2023). [DOI: 10.1007/s10802-023-01044-1](https://doi.org/10.1007/s10802-023-01044-1)

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