

# Accuracy in Screening for Autism Spectrum Disorder in Children with Down Syndrome

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## Abstract

There is a high variance in prevalence of Autism Spectrum Disorder in individuals with Down syndrome because of the absence in accurate screening measures for this specific population. Many professionals mistake a child's deficits in social, language, linguistic, and adaptive skills as symptoms of an intellectual disability, which is a common characteristic found in individuals with DS, rather than as characteristics of co-occurring ASD. Due to this, appropriate interventions may be hindered or the child may be placed in an inappropriate educational placement, which could lead parents and/or caregivers to unnecessary hardship.

Learner outcomes: Participants will be able to understand:

- Why there is a high variance in prevalence amongst this population
- What is needed in order to accurately diagnose
- Why there is a need for these modifications
- The symptomology of individuals with ASD and DS
- What screening measurements are most commonly used by professionals and their psychometric properties
- What is recommended as the most effective form of assessment for this population.

## Introduction

**Autism Spectrum Disorder (ASD):** a lifelong developmental disability defined by diagnostic criteria that include deficits in social communication and social interaction and restricted, repetitive patterns of behavior, interests, or activities.<sup>[3]</sup>

**Down syndrome (DS):** the set of physical, mental, and functional abnormalities that result from Trisomy 21, the presence in the genome of three rather than the typical two chromosomes 21<sup>[8]</sup>; most commonly recognized genetic cause of intellectual disability (ID).<sup>[2]</sup>

### Prevalence:

- In 1999, this comorbidity was considered rare<sup>[9]</sup>; however, estimates now range from 5-39%.<sup>[11]</sup>
- ASD is more commonly found in individuals who present with a genetic syndrome than compared to the general population or idiopathic ASD.<sup>[12]</sup>
- ASD has been diagnosed late than compared to children without DS.<sup>[10]</sup>

**This high variation in prevalence is due to the difficulty in screening for and diagnosing comorbid ASD.<sup>[5]</sup>**

- Currently, all available measures are not able to accurately capture and separate symptoms from individuals with comorbid ASD and individuals with significant ID<sup>[5]</sup>, also known as diagnostic overshadowing.<sup>[11]</sup>

## Methodology

**What is needed in this realm of research?**

- An ASD screener with population-based norms specific to Down syndrome.<sup>[5]</sup>
- Early identification.

**Why is this research needed?**

- Early detection of ASD is essential for appropriate intervention; however, the characteristics associated with DS may influence the recognition of ASD and hinder the implementation of appropriate interventions.<sup>[10]</sup>
- Inaccurate diagnosis can lead to inappropriate educational placement and unnecessary emotional hardship for the parents.<sup>[4]</sup>

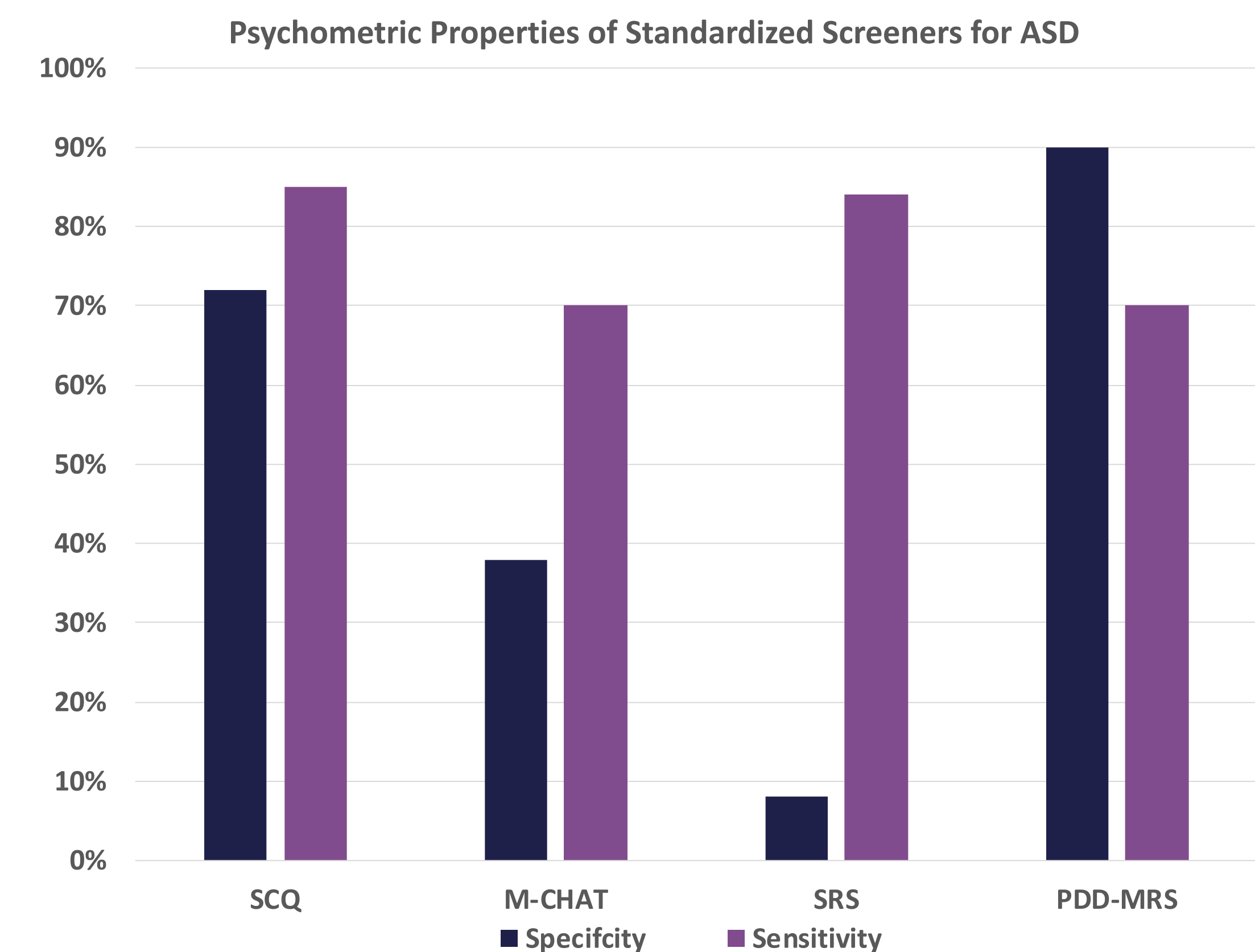
**Symptomology of children with comorbid ASD and DS:**

- Lower IQ scores, poorer expressive and receptive language skills, fewer adaptive behavior skills, higher levels of disruptive behavior, and stereotypy than compared to children with only DS.<sup>[5]</sup>
- Relatively milder social difficulties than compared to children with idiopathic ASD<sup>[13]</sup>. Individuals with comorbid ASD and DS are able to engage in reciprocal social interaction<sup>[13]</sup>; however, they have slightly lower rates of emotional and peer-related problems.<sup>[13]</sup>
- More stereotyped and repetitive speech forms (i.e. if speech is present), limited to no gesturing for communication purposes, limited interest in peers, increased probability of aggression in response to social approaches by peers, the absence of pretend and imaginative play, and the absence of functional play.<sup>[11]</sup>

**The most commonly used ASD screening measures for children with cognitive impairment:**

- [Social Communication Questionnaire \(SCQ\).](#)
- [Modified Checklist for Autism in Toddlers \(M-CHAT\).](#)
- [Social Responsiveness Scale \(SRS\).](#)
- [Pervasive Developmental Disorder in Mental Retardation Scale \(PDD-MRS\).](#)

## Results



## Conclusion

Recommendations for appropriate and accurate screening individuals with comorbid ASD and DS:

- Developmental approach: social or communication function must be both qualitatively different and more impaired than general cognitive function for an additional ASD diagnosis.<sup>[7]</sup>
- [PDD-MRS](#): this screening measurement was created specifically for children and adults with ID by evaluating behaviors that are consistent with an ASD diagnosis.<sup>[11]</sup>
- If specificity of a screener is not as robust, additional screeners are necessary, such as a parent or caregiver interview.<sup>[11]</sup>

## Acknowledgements



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