

## Autism Spectrum Disorder Symptomatology in Children with NF-1

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### What is an autism spectrum disorder?

- Autism spectrum disorder (ASD; DSM-5; APA, 2013) is a neurodevelopmental disorder characterized by:
  - Persistent deficits in social communication and social interaction
  - Restricted, repetitive patterns of behavior, interests or activities
- There is great variability in how ASD is shown – children show a very broad range of symptoms and levels of ASD-related symptomatology

### How is ASD diagnosed?

- Autism spectrum disorders are typically diagnosed by a Clinical Psychologist or a Psychiatrist; other professionals may also have expertise in diagnosis, including Developmental Pediatricians
- Gold standard diagnostic approaches include:
  - Developmental testing to get a sense of level of cognitive functioning and strengths and weaknesses
  - Information gathered from parents and caregivers
  - Direct observation of the child's behavior
- Assessment and diagnosis of ASD in children with genetic conditions (such as NF1) can be more challenging than assessment of children without genetic conditions in the following ways:
  - Many genetic conditions present with social, behavioral, and communication difficulties that overlap with ASD symptoms; however, the presence of these difficulties themselves does not necessarily warrant a diagnosis of ASD.
  - The diagnostician needs to take into account that some of the difficulties observed may be related to NF1 rather than to ASD
  - At the same time, the diagnostician needs to make sure to avoid “diagnostic overshadowing” which is when difficulties are considered to be related to NF1 when really they do indeed indicate the presence of ASD

### What do we know about ASD in children with NF1?

- Some studies have pointed to high rates of ASD in children with NF1, as high as 1 in 4; estimates generally range between 11% and 29% (Walsh et al., 2013; Garg et al., 2013a; Garg et al., 2013b; Tinker et al., 2014; Adviento et al., 2013; Plasschaert et al., 2014).
- More children are identified as showing ASD symptomatology by parent questionnaires (e.g., Morris et al., 2016) than when a comprehensive evaluation is conducted.
- Children with NF1 have a number of difficulties that overlap with ASD and may increase the likelihood that an ASD diagnosis will be considered. These include speech and language difficulties, inattention and/or impulsivity, and communication difficulties.
- There has been very little research about the development of ASD-related symptoms in children with NF1. There is some research that suggests that ASD symptoms in NF1 increase over time,

which is not a pattern generally seen for children with ASD who do not have NF1 (Plasschaert et al., 2014).

- There is very little research about young children. The one published study with toddlers indicated very low rates of ASD-related symptomatology on an ASD screening measure (Tinker et al., 2014). We need more research about ASD symptomatology for children with NF1 and its developmental pattern so that we can best make diagnostic judgments

### **Why are there such variable rates of ASD reported for children with NF1?**

- Different studies use different approaches to:
  - Recruiting participants
    - If participants have more difficulties overall, they are more likely to also show an Autism Spectrum Disorder
  - Assessing participants
    - Screening measures generally identify more children with NF1 with possible ASD than more comprehensive evaluation
    - Expertise in both ASD and NF1 is important to have a sense of how the ASD-related symptoms fit in with the NF1 condition
  - Specific criteria for diagnosis
    - Most of the research to date uses DSM-IV criteria rather than the current DSM-5 criteria. DSM-5 places more emphasis on restricted and repetitive behaviors and interests, which do not appear to be quite as common as communication difficulties for children with NF1.
    - Some of the published research used research criteria that are somewhat more lax than clinical diagnostic criteria.
- Some of the difficulties seen for children with NF1 are also seen for children with ASD, which makes diagnosis more challenging:
  - Language delays
  - Difficulty with peer relationships

### **What approach should parents take?**

- Watch for signs of a possible ASD (see [http://www.autismnavigator.com/wp-content/uploads/2015/03/What\\_is\\_ASD.pdf](http://www.autismnavigator.com/wp-content/uploads/2015/03/What_is_ASD.pdf)) in early childhood. These include:
  - Difficulty with eye contact
  - Language delay
  - Little use of gestures (e.g., pointing, showing, waving, clapping)
  - Child does not respond when their name is called
  - Does not draw others' attention to things
  - Strong interest in particular objects or in repetitive play with objects
  - Sensitivity to sensory input
- If you have concerns about a possible ASD, have your child evaluated by someone with expertise in ASD assessment. The evaluation should include: a) developmental testing; b) parent

questionnaires and interview about ASD symptomatology; c) direct observation of your child's behavior, ideally using the Autism Diagnostic Observation Schedule – 2 (ADOS-2).

- Children with NF1 who also show social communication and repetitive behavior difficulties that meet criteria for a diagnosis of ASD should be referred for evidence-based autism intervention. Applied behavior analysis (ABA) approaches have a strong evidence base and are also likely to be helpful for children with NF1 who also have ASD.
- Be aware that even if your child does not have difficulties that meet criteria for a diagnosis of ASD, many children with NF1 have social challenges that would benefit from intervention. Many children with NF1 would benefit from participating in a social skills training group.

**We need for more research about:**

- ASD symptomatology in NF1 over the course of development
- Most effective methods to screen for ASD in children with NF1
- Effectiveness of ASD-related interventions with children with NF1

**Resources**

[http://www.autismnavigator.com/wp-content/uploads/2015/03/What\\_is\\_ASD.pdf](http://www.autismnavigator.com/wp-content/uploads/2015/03/What_is_ASD.pdf)

<https://www.autismspeaks.org/what-autism>

<https://www.cdc.gov/ncbddd/autism/signs.html>

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