



Autism Spectrum Disorder In Neurofibromatosis Type 1

WHAT IS NEUROFIBROMATOSIS TYPE 1?

Neurofibromatosis Type 1 (NF1) is a common medical condition that affects nearly one in every 3,000 individuals worldwide.

Typically, children with NF1 are identified by the presence of birthmarks (café-au-lait macules) and freckling in the armpits and groin. However, children with NF1 are also at risk for developing brain and nerve tumors, which often become major concerns for parents and health care providers.

In addition, children with NF1 may also have problems with attention, learning, executive function, sensory processing, and developing peer relationships (friendships). These types of problems can be caused by Autism Spectrum Disorder (ASD), and frequently go unrecognized during childhood. Since these problems make it difficult for children with ASD to make friends, relate to children their same age, and communicate their thoughts and feelings to others, it is important to identify ASD early and obtain appropriate resources to help your children reach their full potential.



WHAT IS AUTISM SPECTRUM DISORDER (ASD)?

Autism Spectrum Disorder (ASD) is a developmental disorder which results in difficulties with social interaction (e.g., difficulty starting and continuing conversations), impairments in verbal and nonverbal communication (e.g., picking up on social cues or facial expressions), repetitive behaviors (e.g., hand-flapping), and/or executive function issues (e.g., difficulty changing routines or understanding the order between steps of a task).

Although ASD is not usually diagnosed until 2 years of age (and sometimes later in the setting of NF1), children as young as 12 months of age can display early signs (e.g., lack of smiles or other facial expressions, no babbling or pointing to objects). Unfortunately, children do not “outgrow” their ASD, and these problems typically persist throughout life.

ASD is common in children with NF1, where approximately 25% of children demonstrate mild-to-moderate ASD symptoms, and nearly 15% exhibit severe ASD symptoms. Although children with mild-to-moderate ASD may not be diagnosed with ASD, symptoms may still be significant enough to affect their daily lives.

In the general population, boys are four times more likely to have ASD than girls. However, in children with NF1, boys are only slightly more likely to have ASD than girls.





EVERY CHILD WITH NF1 IS UNIQUE, AND THERE ARE MANY WAYS IN WHICH ASD MAY AFFECT A CHILD, INCLUDING:

- Exhibits facial expressions that do not match what they are saying
- Difficulty with social interaction
- Avoids eye contact or demonstrates abnormal eye contact (e.g., looking out of the side of their eyes, fleeting peripheral glances)
- Provides unrelated answers to questions
- May think or talk about the same special interest repeatedly
- Easily upset by hectic or busy situations
- Has difficulty making friends, even when trying their best
- Demonstrates repetitive and odd behaviors (e.g., hand-flapping, spinning)
- Lack of awareness of personal space
- Difficulty understanding how events relate to one another (cause and effect)

ASD is a relatively new term, and was previously known by various other names, such as autism, childhood disintegrative disorder, pervasive developmental disorder, and Asperger syndrome.

WHAT CAUSES AUTISM SPECTRUM DISORDER (ASD) IN NF1?

ASD is thought to affect early brain development and, although experts are still uncertain about the causes of ASD, there is a genetic component. In NF1, those with first-degree relatives (a parent-child or sibling pair) who both have NF1, tend to be affected by ASD very similarly. This suggests that specific changes in the NF1 gene may predict how severely an individual with NF1 is affected. Additionally, individuals with NF1 can have other genetic changes that increase their risk for developing ASD. Currently, there is no reliable way to predict who will develop ASD or to what degree; however, research focused on these important risk factors is currently ongoing at the Washington University Neurofibromatosis (NF) Center.

WHAT CAN I DO IF I SUSPECT THAT MY CHILD HAS AUTISM SPECTRUM DISORDER (ASD)?

If you think that your child is displaying signs concerning for ASD at any age, please talk to your child's NF doctor.

During their appointment, your child will be examined by their NF team to look for other associated features, including a unique facial appearance, severe developmental delay or cognitive impairment, difficulties in motor coordination and attention, and problems with sleep. You may be asked to complete questionnaires designed to measure the severity of your child's ASD symptoms. If your child scores within the moderate-to-severe range for ASD symptoms, your child may be referred to a local Autism Center for definitive diagnostic testing and initiation of therapy services.

You may also be asked to complete questionnaires designed to assess your child's attention, as many children with NF1 who have ASD also have Attention-Deficit/Hyperactivity Disorder (ADD/ADHD). If your child is diagnosed with ADD or ADHD, your child's NF team may recommend starting a medication to improve both attentional and ASD-related difficulties.

In certain circumstances, your child's NF team may also recommend further genetic testing to look for other causes of ASD, or may suggest additional academic and cognitive (neuropsychological) testing.

ARE THERE INTERVENTIONS THAT SUPPORT PEOPLE WHO HAVE AUTISM SPECTRUM DISORDER (ASD)?

Currently, there is no cure for ASD; however, there are a few treatment/intervention options that are often provided together.

Behavioral therapy - Applied Behavioral Analysis (ABA) is the most effective intervention for ASD. Ideally, it should be started before the age of two. The goal of ABA therapy is to encourage desired behaviors and discourage undesired ones.

Developmental therapy - Focuses on improving developmental skill sets of children with ASD. Specialists may include speech and language therapists, occupational therapists, and/or physical therapists.

Medications - Medications are available to help improve symptoms associated with ASD, such as anxiety, inability to focus, hyperactivity, and self-harming behavior.

It is important to speak with your child's NF specialist to determine the best plan of care for your child.

WHERE CAN I GET MORE INFORMATION?

Additional information about ASD can be found in the following resources:

Washington University Neurofibromatosis Center — nfcenter.wustl.edu/

Washington University Autism Clinical Center — childpsychiatry.wustl.edu

St. Louis Children's Hospital — stlouischildrens.org

Children's Tumor Foundation — ctf.org

Autism Speaks — autismspeaks.org

Missouri Office of Autism Services — dmh.mo.gov/dd/autism/



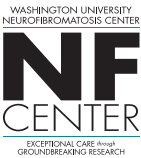


Washington University Neurofibromatosis (NF) Center

Located inside St. Louis Children's Hospital
One Children's Place, Suite 2130
St. Louis, Missouri 63110

314.454.6120

NFClinic@wustl.edu



St. Louis Children's Hospital and Washington University Physicians comply with applicable federal civil rights laws and do not discriminate on the basis of race, color, national origin, age, disability or sex.

Atención: hay servicios de asistencia de idiomas disponibles a su disposición sin costo.
Lláme al 314-747-5682 (TTY: 1-800-735-2966).

注意: 免费提供语言协助服务, 如有需要敬请致电 314-747-5682 (TTY: 1-800-735-2966)。