

# Neurofibromatosis 1

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### Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

# Neurofibromatosis 1

## At a Glance

Neurofibromatosis 1 (NF1) is a rare genetic disorder that can affect multiple body systems. It is a highly variable condition that varies even within a family. Most people with NF1 have a mild form. Less than 10% have serious and lasting physical problems. Many of those problems are evident at birth or develop before adolescence.

About half of children who have NF1 will also have an affected parent, while the other 50% may be the only family member with NF1. There is a genetic test for NF1, but the diagnosis can be made by a specialist after a physical examination.



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## Common Features of NF1:

- Multiple café-au-lait (light brown) spots - individuals will have at least 6 café-au-lait spots, that may look like large freckles
- Multiple cutaneous neurofibromas benign tumors under the skin.
- Plexiform neurofibromas slow-growing tumors that may cause disfigurement, affect nerve function, or be life threatening.
- Lisch nodules - small, harmless clumps of pigment in the iris of the eye that do not affect vision.
- Optic glioma - tumors affecting the optic nerve; can cause a range of visual problems including blindness in the most severe cases.
- Specific skeletal changes including thinning of the outer parts of bones such as the shin bone and bones around the eye socket.
- Learning disabilities are present in about 50-60% of individuals with NF1.

## In Depth

### Medical and Dietary Considerations

#### What you need to know

The list of possible medical problems in NF1 is quite long. Usually, each person has only some of the problems. The severity of any one of these medical problems varies widely between people. Many people with NF1 have only the skin findings (café-au-lait spots and neurofibromas) and Lisch nodules (colored spots in the iris of the eye). However, some children will have more substantial tumor growth that may significantly affect their appearance, mobility, vision, or even be life threatening. It is important to ask the parents about their child's medical issues.

There is no cure for NF1. No special diet is required for NF1, but a well-balanced diet is important. Children with NF1 should have annual doctor visits and may have specialist appointments to monitor medical symptoms. Each symptom must be addressed individually.

Note: The teen years are a time of change, and this includes changes in the issues of NF1.

- Teens with NF1 may see neurofibromas appear for the first time.
- Neurofibromas that are already present may grow in size.
- Skin freckling may increase.
- Changes in body image may result in an increase in depression, anxiety, and social isolation - Counseling may be helpful.

Also note that pregnancy may cause a rapid increase in the number and size of neurofibromas.

#### What you can do

- Encourage yearly checkup and studies as needed in the child's Medical Home. Obtain any updated care plans.
- Make sure that immunizations are up to date
- Support good hand washing to reduce the spread of viruses.
- Notify parents of any changes in the child's energy level.
- Be aware of changes in behavior or mood that seem unusual. Notify the parents.
- Be aware of any unusual symptoms such as an increase in number or severity of headaches. Notify the parents.

### Education Supports

It is important to have high learning expectations for children who have NF1, utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

## What you need to know

Intelligence is usually within the normal range. However, about 50-60% of children with NF1 experience learning disabilities. A variety of learning problems including visual spatial performance and attention difficulties can be seen. Children with NF1 may require special education for learning, speech, motor, or psychosocial problems.

### Executive Functioning

- These skills needed for purposeful, goal-oriented activity are a common cognitive difficulty in individuals with NF1.
- Working memory, planning, organization, and complex problem solving may be difficult.
- Intellectual development, academic achievement, personality, social skills, relationships, and communication with others may be affected. These difficulties impact all areas of learning, sometimes subtly.
- Planning and organization difficulties may make it hard to decide on a starting point at school/home. Children may be overwhelmed with projects. They might be described as inflexible and concrete thinkers.

### Attention

- One of the most frequent concerns of parents
- Poses significant challenges in academics and a child's achievements.

### Math Difficulties

- Can result from difficulties in reading and languages, visual perceptual problems, confused arrangement of numbers and letters, and difficulty with abstract information.
- Word problems can be an area of weakness. Problems with many steps (e.g., algebra, long division) may place a heavy load on working memory, comprehension, and language.
- Difficulties can lead to math illiteracy later in life. This can affect daily living and vocational skills.

### Motor and Sensory Challenges

- Gross and fine motor skill delays are found in many children with NF1.
- These may impact a child's everyday life. For example, it may be difficult to keep up with their peers on the playground, in sports, or in written tasks.
- This can lead to coordination problems that can persist into adolescence.

### Communication

- Language skills may also be an area of weakness. There can be difficulties with both receptive and expressive language.
- Some expressive language difficulties may include:
  - Coming to a point, organizing speech, or finding the right word
  - Having a conversation

- Recalling or retelling information
  - Completing oral and written assignments
- Some receptive language difficulties may include:
  - Following directions
  - Understanding complex sentence structure
  - Understanding meaning and/or content of speech
  - Discriminating between sounds, understanding word meanings, and understanding lengthy or complex speech
- Children may appear to be ignoring directions. They may not be able to keep up with classmates (academically or socially)

### Reading and Spelling

- Children who have NF1 may have difficulty in reading (literacy), and with reading skills (i.e. letter recognition).
- Many children who have NF1 have a weakness in their ability to sound out words when reading (Developmental Phonological Dyslexia). They may find it hard to learn phonics or rules about which sounds correspond to letters.
  - This can lead to a lack of motivation, confidence, and self-esteem.
- Problems with phonological awareness can lead to problems with hearing sounds properly. This can cause difficulty in separating words into syllables, making individual units of sound, or learning rhymes
- Children with reading difficulties often have spelling problems.
- Those with phonological dyslexia have problems with spelling because of the challenge of sounding out words.

### Visual Perceptual Issues

- Children may have a hard time processing visual information. They may struggle with spatial awareness tasks. These problems often go unnoticed.
- They may have problems coordinating what they see with their motor skills (visual motor integration).
- Visual perceptual problems may lead to problems with comprehension, following task instructions, copying, and handwriting. Copying text is difficult because coordination and holding information in memory for the short term is needed.

## What you can do

### Interventions for Executive Functioning and Attention

- Present information in concrete manner
- Use manipulatives to show concepts
- Simplify verbal information and explain concepts clearly
- Provide visual cues and instructions
- Repeat information and use positive reinforcement
- Ask child to repeat instructions

- Help the child find a starting point, especially on complex tasks
- Select relevant task goals
- Use a calendar to track important events
- Organize a means to solve complex problems such as break down problems into parts
- Monitor and evaluate behavior and emotions
- Help organize everyday needs at school and at home by having a place for all things, using different colored notebooks for different subjects, etc.
- Limit written homework
- Allow longer time to write
- Modify notetaking (e.g., fill in the blank notes)

### Interventions for Learning Math

- Teach the association between numbers and quantities. Playing number board games may help.
- Break down word problems and help with reading. Have the child verbalize each step of the problem and explain their work. Make sure math terms are understood.
- Line up calculations. Graph paper may be useful.
- Group similar problems together.
- Encourage the child to double-check all work and to look at the question with their answer to see if it makes sense.
- Help children apply information they have learned to new situations.
- Begin with concrete examples. Proceed to the abstract once the concrete examples are mastered or understood.
- If individuals have difficulty remembering basic number facts, use concrete aids. Help them make up memory strategies or use mnemonic devices.
- Encourage frequent repetition and practice of math concepts.
- Complex diagrams and graphs may be overwhelming. Use verbal explanations instead.
- Encourage use of calculator or other assistive devices if needed.

### Interventions for Motor and Sensory development

- Occupational and physical therapy may be helpful
- Visual instruction may work better than verbal
- Use manipulative activities (Legos, play dough)
- Practice cutting
- Write on every other line
- Allow tracing

### Interventions for Communication Challenges

- Individuals with speech and motor difficulties often benefit from speech and occupational therapy
- Promote language understanding
  - Use simple short sentences
  - Visual prompts and pictures may help

- Use a child's experiences and interests to engage child in learning
- Allow extra time
- Repeat directions
- Provide lesson summaries
- Record lesson so child can listen again
- Promote language development
  - Provide ample time for responding
  - Increase the child's self-confidence by calling on them when they know the answer
  - Encourage a child to repeat the questions before responding
  - Allow a child time to rehearse and respond

### Interventions for Reading

- Children with phonological dyslexia (difficulty sounding out words when reading) can benefit from explicit instructions in learning concepts like phonological awareness (the ability to perceive and manipulate the sounds of language).
- Individuals must be taught that words can be simplified into smaller units of sounds and that letters represent those sounds.
- Identify rhyming and non-rhyming words
- Play games like "I spy"
- Computer-based phonics training programs may help.
- Avoid having a child read aloud to class. Save this for one-on-one teaching or small group. Allow the child time to practice ahead of time.
- Provide extra reading time.
- When needed, offer more than multiple choice or oral based tests. Offer a separate or quiet space for tests.
- If word recognition is a problem, consider flash card reading or the use of mnemonic devices.

### Interventions for Spelling

- Teach common irregular words (words that don't follow normal spelling or sound rules).
- Encourage the child to keep a file of frequently misspelled words when writing.
- Encourage proof reading. Underline misspelled words and allow correcting before turning it in.
- Encourage activities that involve building printed words with letter tiles or other manipulatives.

### Interventions for Visual Perceptual Difficulties

- Modified notetaking (e.g., fill in the blank notes).
- Provide simple overview or summary before lessons.
- Provide clear tests that are as simple as possible with only a few problems on a page.
- Graph paper may help especially in math problems.
- Use lined paper to facilitate figuring out where to place written responses.

- Allow extra time to work.
- Practice tracing shapes and copying pictures.
- Provide feedback. Individuals may not be aware of mistakes.
- Use verbal descriptions to reinforce the visual.
- Practice folding and cutting with scissors.
- Remember that there may be difficulty matching shapes and sizes. Puzzles may be challenging
- Students may have difficulty reading words in correct order. Mark desk with left and right.

## Behavior & Sensory Support

### What you need to know

Children who have NF1 may have poor social skills and difficulty with peer interactions. They may have personality, behavioral, and quality of life differences. This may be due to learning difficulties, ADHD, low academic achievement, expressive and receptive difficulties, or visual perceptual problems (they may not perceive and interpret social cues). There may also be increased challenges for the child if one of their parents has similar issues themselves.

Approximately half of individuals who have NF1 have some degree of behavioral difficulties. No specific behavioral profile has been identified. Individuals with NF1 respond to the same interventions as other children with those same conditions (e.g., individualized attention, positive behavioral supports and sometimes medication). Some of the more commonly observed behavioral characteristics seen in NF1 include the following:

- Anxiety
- Withdrawal
- Depression
- Impulsivity
- Hyperactivity
- Inattention
- Social Skill Deficits
- Inflexibility
- ADHD

Be aware that there can be psychosocial implications from the physical changes occurring with NF1. NF1 can result in disfigurement in several ways.

- Skin neurofibromas may develop on the face or on exposed areas of the arms and legs.
- Larger and deeper plexiform (slow growing) neurofibromas can lead to overgrowth of a particular area of the body.
- Some of the rare bony complications of NF1 can lead to physical differences.
- Any of these differences can lead to questions and teasing, which may lead to social isolation, poor self-esteem, anxiety and/or depression.



## What you can do

- Consult with a counselor or psychologist to obtain diagnosis and treatment plan for suspected mental health or behavioral disorders as with any child.
- Provide information to peers. Discuss similarities and challenges with them.
- Help develop confidence.
- Focus on strengths.
- Provide positive reinforcement.
- Be consistent with directions, rules, and discipline.
- Teach appropriate social behaviors (role model, friend groups).
- Teach how to recognize facial expressions, body language, and moods in self and others.

## Physical Activity, Trips, Events

Individuals who have NF1 are not usually limited in their activities unless there is a specific complication. This might include a bone defect, scoliosis, or tumors that place individuals at risk for injury. A medical doctor will point out if there are any restrictions. If the child had learning and/or behavioral difficulties, accommodations can be outline in the child's IEP.

## School Absences and Fatigue

School age children who have NF1 may have increased school absences, may need to visit the nurse frequently, and may experience fatigue depending on their specific diseased presentation. Notify the parents if there is a change in the frequency of nurse visits, headaches, or other changes in their symptoms.

## Emergency Planning

Emergency plans will be very individually determined, based on child's behaviors and medical issues. It is important to mention any new signs or symptoms and/or pain to the child's parents.

## Resources

### **Medline Plus**

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.

<https://medlineplus.gov/genetics/condition/neurofibromatosis-type-1/>

### **NF Network**

NF Network acts as an umbrella organization for state and regional NF organizations in the US. Their website provides information about NF and ways to connect with researchers, healthcare providers, and the NF community. <https://www.nfnetwork.org/>

### **Children's Tumor Foundation**

The Children's Tumor Foundation is a global nonprofit organization that raises awareness and supports research for NF. Their website provides information for patients and providers. Click on "Resources" and use their search tool to view the numerous options. <https://www.ctf.org/>  
Make sure to check out *NF1: A Guide for Educators* available at this link

[https://www.ctf.org/wp-content/uploads/2023/11/CTF-NF1\\_For\\_Educators\\_Brochure.pdf](https://www.ctf.org/wp-content/uploads/2023/11/CTF-NF1_For_Educators_Brochure.pdf)

## Info for School Nurse and Primary Care Staff

Neurofibromatosis 1 is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

*Gene Reviews* is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for Neurofibromatosis 1 provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. <http://www.ncbi.nlm.nih.gov/books/NBK1109/>

### HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

### Example letter for physician to provide to school

"My patient \_\_\_\_\_ has been diagnosed with Neurofibromatosis 1. This is a rare genetic condition that is characterized by areas of skin discoloration, small tumors in the skin, and the possibility of more destructive tumors in other tissues. Patients with NF1 often experience learning disability or behavior problems. This condition necessitates an Individualized Education Program (IEP) or 504 plan."

## Meet a Child with NF1 – *Alex's Story about School*

Alex is in 4th grade and is now in a school in his town that can offer him the opportunity to be included with peers both with and without disabilities. He loves his new teacher and one-on-one paraprofessional! A good day at school means he has been using his Alpha Smart and can breeze through his math problems. He also has a smoother day when he has used the computer for writing and math, two things that are made easier with the support of this welcome technology.

He is mild mannered and tends to keep a distance if he does not know other children well, preferring to observe from the side than to jump into the fray. It is a bit difficult to mix in since he is new at his school. He has some very good friends, one from his old school. He enjoys playing his tablet, learning new games, internet surfing, and becoming privy to the world of technology.



At home, Alex is working on being more independent with his homework, although his mom or grandmother or teens in the family are there to help him understand and break-down the instructions. Once he understands the instructions, he can do the work! At school, sometimes he might pretend to know the instructions so he can seem to “fit in” with his typical peers but may not quite get the meaning. For example, he came home one day with a question about the word “incentive” and he needed help to understand that word.

Alex was diagnosed with NF when he was 4 years old. He doesn't talk about NF at school but will talk about it at home where he is much more expressive.

His mother hopes to help others with their story. Her strongest piece of advice is not to wait too long if you think a school setting is too restrictive or does not provide adequate support. In hindsight, she wishes she had helped Alex have the opportunity to be included in typical classes earlier so that he might possibly have gained more skills socially and further develop his vocabulary.

*Thank you so much to Alex and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.*