Neurofibromatosis 1 - *At a Glance*

The GEMSS website is intended primarily for educators and families of the preschool through elementary school age group. While much of the information may also apply to older children, the authors are aware that there is a lack of information about transition issues that would be helpful for older children and young adults. It is our hope that this information can be added in the future.

Neurofibromatosis 1 (NF1) is a highly variable condition that involves many body systems. It 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 prior to adolescence.

NF1 is one of the most common dominant inherited genetic disorders. It occurs in about 1 in 3,000 births. About half of children who are affected with NF1 will also have an affected parent, while the other 50% may be the only family member with NF1.

It is also known as von Recklinghausen's Disease. NF1 is a separate and distinct condition from NF2. NOTE: Until the mid-1980's there were many reports that NF1 was the condition found in the Elephant Man (Joseph Merrick). This has been shown to NOT be the case.

NF1 is characterized by:

- *Multiple cafe-au-lait (light brown) spots.* Individuals with NF1 will have at least 6 café-au-lait spots, but there may be many more. The number and size of the café-au-lait spots is important for diagnosis. However, the number or size of café-au-lait-spots doesn't affect the severity of NF1.
- *Axillary (underarm) and inguinal (groin) freckles*.
- *Multiple cutaneous neurofibromas.* These are benign tumors under the skin. The total number varies widely among people affected. In an adult, the number may range from a few to hundreds or even thousands. Some people with NF1 never develop any neurofibromas. In general, children do not have many cutaneous neurofibromas. At puberty, the number increases. They may cause chronic itching in some children.
- *Plexiform neurofibromas.* These are slow growing neurofibromas that may cause disfigurement, affect nerve function, or be life threatening. They are usually present at birth, but may not be identified until later in life.
- *Lisch nodules.* These are small, harmless clumps of pigment in the iris of the eye. These do not cause problems with vision. They can often be seen by an ophthalmologist.

- *Optic glioma.* This is a tumor affecting the optic nerve. Optic gliomas can cause a range of visual problems, including blindness in the most severe cases. The majority of gliomas do not need treatment. Gliomas typically develop in the first eight years of life.
- *Specific skeletal changes.* These can include 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.

Other findings in NF1:

- Headaches are common. They can include migraines, with symptoms including: headaches, abdominal pain, nausea, vomiting, tiredness, fatigue, or dizziness.
- Neurofibromas may be painful depending on location. They may be painful if bumped or rubbed against often.
- Scoliosis is common and in some cases may require a brace or surgery
- Vasculopathy, which is a disease of blood vessels
- High blood pressure
- Below average height
- Larger than average head size
- Puberty usually is in normal time frame but can be early or delayed.
- Tumors other than neurofibromas and optic gliomas can also occur in NF1 but are uncommon.
- NF1 can result in disfigurement in a number of ways.
    - Skin neurofibromas may develop on the face or on exposed areas of the arms and legs.
    - Larger and deeper plexiform 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. Specifically, changes in the bones of the skull may occur over time and may affect the eye socket.
    - Pseudoarthrosis (a fracture of the bone that doesn't heal properly) can result in amputation of part of a limb and the use of a brace or prosthesis.
    - Any of these differences can lead to questions and teasing. This could further lead to social isolation, poor self-esteem, anxiety and depression.

- Parents sometimes may not choose to inform their young child of his or her diagnosis of NF1, particularly if the only sign is café-au-lait spots.

Things to Think About

1. Medical / Dietary Needs

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. It is important to ask the parents about their child's medical issues.

Many people with NF1 have only the skin findings (café-au-lait spots and neurofibromas) and Lisch nodules.

School age children with NF1 may have annual doctor and specialist visits to monitor medical

Annual physical Annual eye exam Regular developmental assessment of children Blood pressure monitoring

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.
- Some teens benefit from attending an NF summer camp or joining Youth CONNECT, an NF1 chat room and Facebook and Twitter connection. (http://www.ctf.org/Learn-About-NF/YouthCONNECT.html).

Pregnancy may cause a rapid increase in the number and size of neurofibromas.

No special diet is required for NF1. A well-balanced diet is important.

What you can do

- A yearly checkup and studies as needed should occur in the child's Medical Home.
- Up to date immunizations are important. Most children can receive live virus vaccinations. Recording information about the types of vaccinations given is important.
- 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.
- Be aware of any academic changes. Contact parents when differences are noticed.
- Advocate for sufficient speech and language support so the child can communicate effectively all day. This may include communication augmentation devices in some situations.

2. Education Supports

It is important to have HIGH EXPECTATIONS for learning for children who have Neurofibromatosis 1.

Information comes from the Children's Tumor Foundation www.ctf.org/
Please see the following pamphlets on the website for additional information.

- The Child with NF1
- NF1: About Learning Disabilities
- NF1: A Guide for Educators
- Transitioning to Adulthood

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.

> ***Learn more:***
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> Cognitive issues may include non-verbal and verbal learning disorders. Children may have trouble with visual and auditory perception. Information integration (sequencing, abstraction, or organization) may be a challenge. They may have problems in reading, math, spatial ability, neatness, test taking, speech difficulties, or making friends.

Executive Functioning and memory

Executive function (skills needed for purposeful, goal-oriented activity) is 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 effected.
- 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:

- Attention is one of the most frequent concerns of parents and poses significant challenges in academics and a child's achievements.

Math learning difficulties

Math challenges 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 may place a heavy load on working memory (algebra, long division), comprehension, and language.
- Difficulties can lead to math illiteracy later in life. This can affect daily living and vocational skills.

Motor and sensory abilities

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. They may find it hard to perform tasks that require skilled control of movements.

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 difficulty:
    - 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

Children who have NF1 may have difficulty in reading (literacy). This may lead to problems in math as well. They may have difficulty with reading skills (i.e. letter recognition). Many children who have NF1 have a weakness in their ability to sound out words when reading. This is known

as Developmental Phonological Dyslexia. They may find it hard to learn phonics or rules about which sounds correspond to letters.

- This can lead to lack of motivation, confidence, and self-esteem.
- Subtle aspects of language, such as phonological awareness, can lead to problems with learning rhymes, or hearing sounds properly. This can cause difficultly in separating words into syllables and making individual units of sound.

Spelling

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

- Children who have visual perceptual problems 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 attention and memory

- Help with organization
- 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. They often have a hard time with multiple step tasks and lose track of what they are doing.

- Select relevant task goals.
- Use a calendar to track important events.
- Organize a means to solve complex problems.
- Monitor and evaluate behavior and emotions.
- Help organize everyday needs at school and at home. For example, have a place for all things, use different colored notebooks for different subjects, etc.

Interventions for learning math

- Foster the development of quantity representations. Teach the association between numbers and quantities. Playing number board games may help.
- Break down word problems and help with reading. Have 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 child to double-check all work. Encourage child 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.
- They may be overwhelmed by complex diagrams and graphs. 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.
- Limit written homework.
- Use manipulate activities (Legos, play dough)
- Practice cutting

- Allow longer time to write
- 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 by using 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 by:
    - Providing ample time for responding
    - Increasing the child's self-confidence by calling on them when they know answer
    - Encouraging a child to repeat the questions before responding
    - Allowing 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.
- Identifying rhyming and non-rhyming words
- Playing 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.

- Practice reading stories and 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

- Modify copying. For example provide a copy of teacher's or other student's notes.

- Provide simple overview or summary before lesson.

- 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 as they may have trouble figuring out where to place written responses on a sheet of paper.

- Allow extra time on 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.

- May have difficulty matching shapes and sizes. Puzzles may be challenging
- May have difficulty reading words in correct order. Mark desk with left and right.

3. Behavior & Sensory Support

What you need to know

Children who have NF1 may have poorer social skills and more 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, 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 is affected and has learning or physical issues themselves.**

Approximately ½ of individuals who have NF1 will have some degree of behavioral difficulties. No specific profile is noted and individuals respond to the same interventions as other children with these same conditions (e.g., individualized attention, positive behavioral supports and sometimes medication).

Behavioral characteristics of NF1 vary and include:

- Anxiety
- Withdrawal
- Depression
- Impulsivity
- Hyperactivity
- Inattention
- social skill deficit
- inflexibility
- ADHD

Psychosocial Considerations:

NF1 can result in disfigurement in a number of 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.

    - Specifically, changes in the bones of the skull may occur over time and may affect the eye socket.

    - Pseudoarthrosis (a fracture of the bone that doesn't heal properly) can result in amputation of part of a limb and the use of a brace or prosthesis.

- Any of these differences can lead to questions and teasing, which may lead to social isolation, poor self-esteem, anxiety or depression.

What You Can Do:

- 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.

4. Physical Activity, Trips, Events

What you need to know

No special accommodations are needed, unless required due to the child's learning and/or behavioral difficulties.

Individuals who have NF1 are not usually limited in their activities. The only exception is for those children with 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.

5. School Absences and Fatigue

What you need to know

School age children who have NF1 may have increased school absences or may need to visit the nurse due to migraines/headache. If there is a change in the frequency of nurse visits for headaches, notify the parents.

- Migraines may also cause fatigue.

6. Emergency Planning

What you need to know

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.

7. Resources

Children's Tumor Foundation, including pamphlets

http://www.ctf.org/

Children's Tumor Foundation includes research updates, latest news and upcoming events. *"Life is full of possibilities for a kid living with NF."*

Please see their pamphlets for more information:

http://www.ctf.org/images/pdf/nf1aboutlearningdisabilities.pdf
NF1: About Learning Disabilities

http://www.ctf.org/images/pdf/nf1guideforeducators.pdf
NF1: A Guide for Educators

http://www.ctf.org/images/pdf/childwithnf1.pdf
NF1: The Child with NF1

http://www.ctf.org/Patient-Information/Patient-Information-Brochures.html
Transitioning to Adulthood (scroll down)

Neurofibromatosis, Inc.

www.nfnetwork.org/

Neurofibromatosis, Inc. includes a description of the condition, help finding a doctor, and educational materials. *"We are the national non-profit network serving families and individuals affected by Neurofibromatosis".*

National Center for Biotechnology Information (NCBI) Bookshelf – NF1

http://www.ncbi.nlm.nih.gov/books/NBK1109/

Learn more about the genetics of NF1

Understanding NF1

http://www.understandingnf1.org/index.html

Understanding NF1 is a medical resource about Neurofibromatosis 1 for parents, patients, and providers. It includes sections on *identifying, explaining, managing and supporting.*

American Academy of Pediatrics (AAP) – Health Supervision Guidelines

http://pediatrics.aappublications.org/content/96/2/368.abstract

The AAP endorses these Health Supervision Guidelines. Families may find these helpful when talking to their pediatricians or family physicians.