

Neurofibromatosis

Other Common Names

- Recklinghausen's Disease
- Von Recklinghausen's Disease

Definition/Description

The neurofibromatoses (NF) are genetic disorders of the nervous system, which cause tumors to form on the covering of the nerves anywhere in the body at any time. Three distinct forms of NF have been identified

- neurofibromatosis type 1 (NF1)
- neurofibromatosis type 2 (NF2)
- schwannomatosis

Causes/Etiology

The neurofibromatoses are genetic disorders. Although many affected persons inherit the disorder, between 30 and 50 percent of new cases arise spontaneously through mutation (change) in an individual's genes. Once this change has taken place, the mutant gene can be passed on to succeeding generations.

Incidence

- Neurofibromatosis affects both sexes and all ethnic groups.
- NF1 has an incidence rate of 1 in 4,000.
- Both NF2 and Schwannomatosis have incidence rates of 1 in 40,000.

Characteristics/Classifications

NF1 is characterized by changes in skin appearance, tumors, or bone abnormalities.

Symptoms of NF1, which may be evident at birth and nearly always by the time the child is 10 years old, may include light brown spots on the skin ("cafe-au-lait" spots), two or more growths on the iris of the eye, a tumor on the optic nerve, a larger than normal head circumference, and abnormal development of the spine, a skull bone, or the tibia.

NF2 is characterized by slow-growing tumors on the eighth cranial nerves. The tumors cause pressure damage to neighboring nerves. To determine whether an individual has NF2, a physician looks for eighth nerve tumors, cataracts at an early age or changes in the retina that may affect vision, other nervous system tumors and similar signs and symptoms in a parent, sibling, or child. Additionally NF2 causes hearing loss, ringing in the ears, and poor balance. (It often starts in the teen years)

Schwannomatosis is characterized by the development of multiple schwannomas (tumors made up of certain cells) everywhere in the body except on the vestibular branch of the 8th cranial

nerve. The dominant symptom is pain, which develops as a schwannoma enlarges or compresses nerves or adjacent tissue. Some people may develop numbness, tingling, or weakness in the fingers and toes.

Diagnostic Category and IDEA Category

The International Statistical Classification of Diseases and Related Health Problems-9th Revision-Clinical Modification (ICD-9-CM) categorizes neurofibromatosis under the 237 code. More specifically, the 237.70 code refers to neurofibromatosis unspecified, 237.71 refers to NF1, and 237.72 refers to NF2.

Under the Individuals with Disabilities Education Act (IDEA), neurofibromatosis would most likely be classified under the category of Other Health Impairment given that it is a chronic medical condition that has the potential to adversely affect a child's educational performance. However certain symptoms of neurofibromatosis may fall under other categories as defined by IDEA. For example, a student with NF2 who has hearing loss may meet the criteria for a Hearing Impairment whereas a student with NF1 may have an Orthopedic Impairment. It is most likely given the complex symptoms of neurofibromatosis that a child could be found eligible for special education services under the Multiple Disabilities category.

Deficits

In children diagnosed with NF1, deficits in many body systems may be present. Those include

- Visual impairments or blindness
- Seizures, headaches, brain tumors, learning disabilities, and/or mental retardation
- Speech impairments
- Orthopedic concerns such as scoliosis and bone deformities
- Developmental delays in walking or talking and poor school performance
- Early or delayed puberty
- Digestive tract difficulties including pain, vomiting, constipation, or diarrhea
- Skin abnormalities including café-au-lait spots

In children diagnosed with NF2 the following deficits may occur:

- Cataracts at a young age that can affect vision
- Brain tumors (such as meningiomas) and spinal tumors that affect balance, cause hearing loss or deafness, spinal compression, facial paralysis, and swallowing difficulties
- General muscle weakness
- Skin tumors

Given that schwannomatosis is not diagnosed until after the age of 30 and symptoms first appear in adulthood, children in the schools will not present with symptoms of this form of neurofibromatosis.

Prognosis/ Long Term Developmental Outcome

The prognosis for individuals with NF1 is typically quite good given that the degree of symptoms is typically mild to moderate. These individuals usually lead relatively normal and productive lives. In more rare cases NF1 can cause more severe and debilitating symptoms that cause both significant psychological and cosmetic difficulties. Additionally, individuals with NF1 are up to five times more likely to develop learning disabilities compared to the general population.

The prognosis and long term developmental outcomes for individuals with NF2 are more variable than those diagnosed with NF1. Outcomes depend on the location and growth of the tumors. Because the tumors associated with NF2 can damage vital structures such as the brain, this damage can be life threatening. Most all individuals with NF2 will require surgery at some point in their life to remove tumors. Given the close vicinity of tumors in the brain and along the spine, neurological damage can occur as a result of the tumor removal.

Individuals diagnosed later in life with schwannomatosis live with chronic pain which in some cases can be severe and debilitating. Pain medications are used to alleviate symptoms but the understanding of underlying mechanisms that cause pain are poorly understood and therefore medications are not always effective in pain management.

Assessment Approaches

The actual diagnosis of neurofibromatosis would be made by a medical doctor through the use of findings from an individual's medical history, clinical examination, family history, MRI findings, and/or genetic testing.

The school psychologist may want to interview the parents to obtain an updated medical record and continue to be in touch with the family regarding ongoing medical interventions and/or surgeries. An interview with the child would also be appropriate to gain insight into academic, social, and psychological impacts they face as a result of neurofibromatosis.

The selection of assessment tools would need to be tailored to the student's specific limitations, for example given an orthopedic impairment, hearing impairment, or cognitive impairment. Rating scales from the student, parents, and teachers would provide insight into the child's functioning in various settings and any mood concerns. Assessing the student for mood disorders such as anxiety or depression is particularly important if he or she exhibits deformities that cause social isolation.

Interventions and Treatments

Neurofibromatosis is not curable. Surgery is often recommended to remove the tumors. Some NF1 tumors may become cancerous, and treatment may include surgery, radiation, or chemotherapy. Surgery, radiation, and chemotherapy also may be used to control or reduce the size of optic nerve tumors when vision is threatened. Some bone malformations can be corrected surgically.

For NF2, improved diagnostic technologies, such as MRI, can reveal tumors as small as a few millimeters in diameter, thus allowing early treatment. Surgery to remove tumors completely is one option but may result in hearing loss. Surgery also can correct cataracts and retinal abnormalities.

There is no currently accepted medical treatment or drug for schwannomatosis, but surgical management is often effective. Pain usually subsides when tumors are removed completely. Genetic testing is available for families with documented cases of NF1 and NF2 but such testing for schwannomatosis currently does not exist.

Contributions of the School Psychologist

A school psychologist should be aware of the physical and emotional implications that these children may experience. Classmates should receive information about the condition to minimize teasing and social isolation. Additionally school staff should plan for the possibility of the student missing school for medical procedures. This disorder may manifest through hearing difficulties and is associated with other learning difficulties. Interventions may need to be put into place to help develop both academic and social aspects of these student's lives.

Helpful Resources:

Your Genes Your Health

<http://www.yourgenesyourhealth.org/nf/>

The Children's Tumor Foundation: Ending Neurofibromatosis through Research

<http://www.ctf.org/>

Neurofibromatosis, Inc

<http://www.nfinc.org>

NCBI Genes of Disease

<http://www.ncbi.nlm.nih.gov/disease/Neurofib.html>

National Institute of Neurological Disorders and Stroke

<http://www.ninds.nih.gov/disorders/neurofibromatosis/neurofibromatosis.htm>