What is Neurofibromatosis?

The neurofibromatosis is made up of two disorders, NF-1 and NF-2. These are genetic disorders of the nervous system that primarily affect the development and growth of neural (nerve) cell tissues. These disorders allow tumors to grow on nerves and produce other abnormalities such as skin changes and bone deformities. The neurofibromatoses occur in both sexes and in all races and ethnic groups. NF1 is more common than NF-2. NF1 occurs in about 1 in 4,000 individuals in the United States. 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. Previously, NF1 was known as peripheral neurofibromatosis (or von Recklinghausen's neurofibromatosis) because some of the symptoms (skin spots and tumors) seemed to be limited to the outer nerves, or peripheral nervous system, of the affected person. This name is no longer technically accurate because central nervous system tumors are now known to occur in NF1. In most cases, symptoms of both NF-1 and NF-2 are mild and patients live normal and productive lives. In some cases, however, can be severely debilitating. Symptoms and severity of the disorder may vary among members of affected families.

How are these disorders diagnosed?

- NF1: a physician looks for changes in skin appearance, tumors, or bone abnormalities, and/or a parent, sibling, or child with NF1. Symptoms of NF1, particularly those on the skin, are often evident at birth or during infancy and almost always by the time a child is about 10 years old. Neurofibromas may become evident at around 10 to 15 years of age. In diagnosing NF1, a physician looks for two or more of the following:
    - 5 or more light brown skin spots (cafe-au-lait macules) measuring more than 5 millimeters in diameter in patients under the age of puberty or more than 15 millimeters across in adults and children over the age of puberty;
    - 2 or more neurofibromas (tumors that grow on a nerve or nerve tissue, under the skin) or one plexiform neurofibroma (involving many nerves);
    - freckling in the armpit or groin areas;
    - benign growths on the iris of the eye (known as Lisch nodules or iris hamartomas);
    - a tumor on the optic nerve (optic glioma);
    - severe scoliosis (curvature of the spine);
    - enlargement or deformation of certain bones other than the spine; and
    - a parent, sibling, or child with NF1.
- NF2 is less common. NF2 is characterized by bilateral (occurring on both sides of the body) tumors on the eighth cranial nerve. The tumors cause pressure damage to neighboring nerves. To determine whether an individual has NF2, a physician looks for bilateral eighth nerve tumors and similar signs and symptoms in a parent, sibling, or child. Affected individuals may notice hearing loss as early as the teen years. Other early symptoms may include tinnitus (ringing noise in the ear) and poor balance. Headache, facial pain, or facial numbness, caused by pressure from
- Genetic testing is available for families with documented cases of NF1 and NF2. New (spontaneous) mutations cannot be confirmed genetically. Prenatal diagnosis of familial NF1 or NF2 is also possible utilizing amniocentesis or chorionic villus sampling procedures.

Are there any treatments?

- Treatments for both NF1 and NF2 are presently aimed at controlling symptoms.
- NF-1: Surgery can help some bone malformations and remove painful or disfiguring tumors; however, there is a chance that the tumors may grow back and in greater numbers. In rare instances tumors become malignant (3 to 5 percent of all cases), treatment may include surgery, radiation, or chemotherapy.

Are there any treatments?

- 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. Other options include partial removal of tumors, radiation, and if the tumors are not progressing rapidly, the conservative approach of watchful waiting.

What is the problem in NF?

- NF-1: the gene causing NF1 is on chromosome 17 and produces a large and complex protein called neurofibromin. One portion of this protein is similar to a family of proteins called GAP (guanosine triphosphatase-activating protein) which help to prevent tumor develop. However, in NF-1, the protein is abnormal and allows excessive cell growth leading to neurofibromas and possibly development of cancers.
- NF-2: the gene causing NF2 is on chromosome 22. The NF2 gene product is a tumor suppressor protein.

What is the prognosis?

- NF-1: In most cases, symptoms of NF1 are mild, and patients live normal and productive lives. In some cases, however, NF1 can be severely debilitating and includes tumors (gliomas) of the Central Nervous System (usually brain) including the optic nerves that can impair vision. These typically grow slowly and are difficult to treat because surgery or radiation therapy may also damage the nerve which would cause more visual loss. Some neurofibromas grow in areas that compress nerves (spinal column) and cause pain and weakness. Over years, gliomas and neurofibromas may "transform" (change) and develop into malignant cancer.
- NF-2: the damage to nearby vital structures, such as other cranial nerves and the brainstem, can impair function (such as hearing loss) or can rarely become life-threatening.

Where can I get more information?

You can obtain further information on the neurofibromatoses, including information about treatment centers and genetic counseling from the following voluntary health organizations:

Organizations

National Neurofibromatosis Foundation

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16th Floor
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