A SERIOUS AND MISUNDERSTOOD DISORDER

Although neurofibromatosis has been known to the medical community since 1882, there has been widespread misinformation and a lack of precise medical management guidelines available to both physicians and patients until recently.

Pittsburgh's NF Clinic serves as a beacon of information and medical expertise for thousands of families in the region. At its founding, in 1989, access to medical specialists and diagnostic testing and technology was sporadic, at best. In the 15 years since, physicians, social workers, geneticists, diagnostic technicians, nurses and other service providers have focussed their clinical efforts and research interests, resulting in the highest possible care and guidance for NF patients and their families.

The NFCA (Neurofibromatosis Clinics Association, Inc.) acts as an information resource for health care professionals, educators, patients and the general public throughout southwestern Pennsylvania and surrounding regions.

nfca

Neurofibromatosis Clinics Association

Growing awareness. Seeking hope.

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SKELETAL

NEUROFIBROMATOSIS:

RESPIRATORY

A DISTURBING

CIRCULATORY

AND ELUSIVE DISEASE

ENDOCRINE

What is **Neurofibromatosis?**

A short course on a disturbing and elusive disease

Q. What is neurofibromatosis?

A. The neurofibromatoses (NF1 and NF2) are two separate genetic disorders which cause tumors (mostly benign) to grow on all types of nerves, including those under the skin and elsewhere, potentially affecting the brain, eye, ear, throat and spinal cord. NF can also affect the development of non-nervous tissues such as bones and skin.

Q. Does NF have a treatment or cure?

A. At the present time, there is no way to prevent or cure NF. Some symptoms can be improved with surgery or other interventions. Many patients successfully manage the disease and lead productive lives – but only with the help of a well-informed and dedicated medical and social service team.

Q. Who gets NF?

A. Approximately one in every 3,000-4,000 babies born has NF. Approximately 100,000 people in the United States have NF. NF occurs in equal numbers of men and women, and in every racial and ethnic group.

Q. What causes NF?

A. NF can be inherited from a parent (autosomal dominant) *or* caused by a genetic change (spontaneous mutation), meaning any child can be born with NF. There is nothing a parent can do or not do to cause this change. Those with a spontaneous genetic mutation did not inherit the disease, but can pass it on to their children.

Q and A

NF1

If two or more of the following signs are present, a diagnosis of NF1 is likely:

- Family history of NF1
- 6 or more light brown ("café-au-lait") spots on the skin
- Presence of pea-sized or larger bumps (2 or more neurofibromas) on the skin
- Larger areas on the skin that look enlarged (plexiform neurofibromas)
- Freckling under the arms or in the groin area
- Pigmented bumps on the eye's iris (Lisch nodules)
- Skeletal abnormalities such as bowing of the legs, curvature of the spine (scoliosis), or thinning of the shin bone
- Tumor on the optic nerve that can interfere with vision

NF2

NF2 (Central Bilateral Acoustic NF) is a rarer disorder characterized by multiple tumors on the head and spinal nerves. The hallmark of NF2 is tumors on both of the auditory nerves. If a combination of the following signs are present, a diagnosis of NF2 is likely:

- Family history of NF2
- Tumors on both of the auditory nerves which may cause deafness, ringing in the ears, or balance problems
- Tumors on the brain, spinal cord or meninges
- Cataracts at a young age

Q. What makes NF so difficult and expensive to manage?

A. No two NF patients, including those from the same family, report the same severity or type of symptoms. The unpredictable nature of the disease requires close coordination between medical specialists and frequent follow-up examinations and testing. Patients, even those with mild symptoms, carry the concern of unforeseen developments, especially during adolescence and pregnancy. These symptoms can also increase with age.

Q. Is there a test for NF?

A. In the past, testing to determine whether an individual has the changed gene that causes NF1 or NF2 has required that at least two family members with NF and multiple family members without NF provide blood samples. This type of testing is called **linkage analysis**. This *cannot* be used to diagnose individuals who have no family history of NF.

Recently **direct gene testing** has become available. This means an individual can be tested without blood samples from other relatives. However, it is not 100% accurate. Further research is necessary to make testing for both NF1 and NF2 more accurate. For more information on gene testing consult the geneticist at the NF Clinic.

Q. What should someone do if they think they have NF?

A. Children's Hospital of Pittsburgh operates an NF Clinic each month. A team of neurologists, genetic physicians, genetic counselors and other medical and support staff evaluate children and adults at the clinic, guide patients to follow-up testing and offer individual and family support services.

Q. What is the role of the NFCA in supporting the NF Clinic?

A. NFCA volunteers and facilitated the opening of an NF Clinic through Children's Hospital and UPMC in 1983. Since then, thousands of people throughout Western Pennsylvania and the tri-state area have been screened for NF.

To schedule an appointment at the NF Clinic, call 412.692.5520. Or to contact a nurse practitioner or physician, call 412.692.3413. The NFCA also provides funding for two social workers to assist families with a variety of special needs and services, including advocacy for NF children with learning disabilities. **To contact a social worker at the NF Clinic, call 412.692.6544.**

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