

# Neurofibromatosis Network Impact Statement

## The Issue

Imagine that you are a parent of a child diagnosed with Neurofibromatosis (NF), a genetic disorder that causes tumors to form on the nerves. Neurofibromatosis (NF) is a set of complex genetic disorders that affects almost every organ system, causing a predisposition for tumors to grow on nerves in the brain and throughout the body. Although many affected persons inherit the disorder, 50% of new cases arise spontaneously through change in an individual's genes.

Imagine you are that parent and are told that there are currently no effective treatments. In some cases surgery can be helpful to alleviate pain and disfiguring tumors.

Imagine that there are no NF specialist in the state where you live and you do not know where to take your child for knowledgeable treatment. Imagine the isolation and fear of the unknown since you have not met any other individuals or families living with NF. Imagine there was no federal funding available for the hope that medical research on NF provides.

## Neurofibromatosis Defined

In most cases, symptoms of NF1 are mild allowing patients to live normal and productive lives. However, it can also be debilitating and in some cases life-threatening. NF can lead to problems within various systems, organs and functions of the body including:

- **DEVELOPMENT:** learning disabilities, poor school performance, hyperactivity/attention deficit
- **BRAIN:** brain tumors, vision loss, seizures, paralysis, headaches
- **NERVES:** nerve tumors, weakness, pain
- **BONE:** scoliosis and bone deformities
- **HEART:** valve defects, abnormalities of the arteries
- **SKIN:** pigmentary abnormalities, particularly café-au-lait spots

Of the two types of NF that have been identified, Type 1 (NF1) is among the world's most common genetic disorders, occurring in about one of every 3,000 births. This makes it more widespread than cystic fibrosis, hereditary muscular dystrophy, Huntington's disease and Tay Sachs combined.

Diagnosed most often in children and young adults, NF1 occurs worldwide and in all races, ethnic groups and both genders. NF1 can appear in any family.

*NF Defined by David Gutmann, MD, PhD., Washington University, Neurofibromatosis Clinic, St. Louis, MO*

- **Eliminate Suffering**
- **Create Hope**
- **Advance NF Research**



## **Our Response**

The NF Network is the national leader in the promotion of federally-funded NF research. We bring the voices of the NF community directly to Congress each year to secure desperately needed funds for NF research.

The NF Network maintains a Clinical Referral Database of physicians with NF expertise, available on the national website, [www.nfnetwork.org](http://www.nfnetwork.org). The Network and its local organizations can provide referrals by phone or email.

The NF Network is introducing the first phase of a new program, Clinical Care Options, for the management of NF1. This service helps bridge the care gap between the local physician and an experienced NF specialist. For the NF1 patient with no access to an NF Clinic, we provide a comprehensive, scientifically-reviewed publication created for the express purpose of improving clinical care. Designed for both the patient and the local physician, whose NF knowledge may be limited, the document includes information on the diagnosis and management of NF1, specific tumor types, and a list of Neurofibromatosis Clinical Trials Consortium clinics. NF Consortium practitioners are up-to-date on available treatments and have agreed to be contacted by clinicians for management advice or patient referral. It is critical that NF be managed by an experienced clinical team. It is our hope that the NF Clinical Care program will help achieve top quality care for all NF patients.

The NF Network produces educational materials for adults and children to help them understand NF. The contents include the cause of NF, issues which may arise at various ages, and where to get help. Also produced are webinars on management whereby national experts share their knowledge with a national audience. Individuals across the country can participate without the need to travel. Archived webinars are available on the Network website [www.nfnetwork.org](http://www.nfnetwork.org).

Three times a year the NF Network produces the Network Edge newsletter, summarizing recently published NF research. The Network Edge highlights research projects funded by the federal NIH and NFRP programs. Using this tool families learn about the latest research on manifestations which affect them. The Network Edge enables researchers and clinicians to keep track of recent findings all in one publication.

The NF Network holds educational meetings around the country, providing access to NF specialists. Attendees have the opportunity to ask questions of the experts and to interact with others affected by NF. One of our most successful programs is, Inspire, the Network's online health community for all forms of NF.

The NF Network plays a vital role on the national NF stage, bringing the voice of the community to researchers, funders, and clinicians, pushing ever closer to the cure.

## **Some Highlights**

Through awareness programs the NF Network organizations have identified more than 14,000 families with NF nationally. Educational materials and referrals to physicians with NF expertise are provided upon request to those contacting the Network.

Since 1996 the NF Network Advocacy Program has worked to secure nearly \$288 million for the Neurofibromatosis Research Program. That money has been matched almost dollar for dollar at the National Institutes of Health.

The Network hosts structured educational meetings in several locations around the country, providing vital information about the management and care of NF to those affected.

The Network also hosts NF Chats, informal gatherings that bring together individuals living with or caring for someone with neurofibromatosis. The chats break down the isolation that living with a genetic condition creates.

Great Steps4NF walks take place in communities nationwide. These inspiring events create bonds that build a supportive NF community while raising awareness and providing funding for NF research.

Annual NF Hope concerts are star-studded events which raise awareness of the disorder, fund NF services and strengthen hope in the NF community.