Living with Neurofibromatosis Type 1: A Guide for Adults

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Neurofibromatosis (NF) is a genetic condition that causes bumps and nodules to grow on nerves. It is a common genetic condition that affects approximately 1 in 4000 persons. There are two types of NF, referred to as NF1 and NF2, that are completely different conditions. Persons with NF1 are not at risk for the medical problems seen in people with NF2.

This booklet is designed to address the questions and concerns that are unique to the adult with NF1. Adults with NF1 have concerns that differ from those in children. They include changes in physical appearance, medical issues, having children, social acceptance, and what to expect during adulthood.

In general, most people with NF1 do well and do not have serious medical problems. They often marry, have children, and have rewarding careers in fields like teaching, business, construction, health care, and computers, to name just a few.

Overall, most of the complications that adults face relating to their NF1 are not life threatening; and for most people who have few complications from their NF1, life expectancy is not reduced. However, for some people, there may be reduced life expectancy because of some of the more significant health problems associated with NF1. These various complications and concerns that adults with NF1 experience are addressed in this booklet.

TUMORS AND CANCER

Tumors are common in adults with NF1; but they are usually benign growths, which means they are not a cancer or a malignancy. The kinds of tumors seen in persons with NF1 include neurofibromas, plexiform neurofibromas, and astrocytomas. Each of these tumor types will be discussed in the following paragraphs.

The biggest change that an adult with NF1 may notice is the gradual and continual development of new *neurofibromas.* Neurofibromas are benign tumors that develop on or

under the skin along a nerve. They may look like small bumps or nodules. Even though they are called "tumors," neurofibromas will never develop into cancer. Neurofibromas may often start developing during times of hormone changes, like adolescence and during pregnancy. Many women also note an increase in the growth of pre-existing tumors during pregnancy. Typically, adults with NF1 will acquire more tumors as they age. Neurofibromas usually develop gradually over time and may continue to grow in size for a period of time. There is no way to determine how many neurofibromas a person with NF1 may develop, when they will start to develop, or where on the body they will grow. These factors vary from person to person. Some people with NF1 may have only one or two neurofibromas while others may develop several. They can grow anywhere there are nerves - on the face, the scalp, the chest, around the nipple, as well as inside the body. Approximately only one in five people with NF1 will have neurofibromas on the face.
There is nothing a person can do to prevent the growth of neurofibromas. Things like certain foods, smoking, caffeine, and heat do not stimulate their growth. Although a formal study has not been done, there is some speculation that the use of oral contraceptives may contribute to their growth. Some people may elect to have a neurofibroma removed for cosmetic reasons, because it itches or stings, or because it is in a place where it is frequently irritated or catches on clothing. Neurofibromas should be removed by a doctor who has experience with NF1. They may be removed by surgery or occasionally by laser. If a neurofibroma is removed, it may or may not grow back. Occasionally, removal of a neurofibroma may cause a thickened scar.

Plexiform neurofibromas are another type of benign tumor that grows along nerves. Unlike the typical skin neurofibroma that grows as a small-defined nodule, the plexiform neurofibroma is a larger growth with less well-defined borders. It can feel like a bunch of cords or knots beneath the skin. Plexiform neurofibromas also differ from the discrete neurofibromas because they usually appear before adulthood. These tumors can be painful if hit or bumped. They also can be disfiguring depending on their size and location. Less commonly, a plexiform neurofibroma tumor can become cancerous. **People with a plexiform neurofibroma should always let their doctors know immediately if they develop a new persistent pain or a change in their usual physical abilities and activity (weakness, numbness, change in bladder or bowel function). They also need to seek medical advice if the tumor grows rapidly, changes in shape or color, or feels harder in texture.**

An *optic glioma* is a tumor on the optic nerve inside the skull. The optic nerve functions to transmit vision to our brains. Because these tumors grow only in children, adults with NF1 do not need to worry about this type of tumor and the visual problems that can be associated with it. An optic glioma diagnosed during childhood rarely progresses during the adult years to cause problems. Individuals who notice a change in their eyesight should consult their ophthalmologists. The most common reason for deterioration in vision in adults result from changes in the eye lens associated with age. This is unrelated to NF1.

In rare instances a person with NF1 can develop a brain tumor called an *astrocytoma.* Adults with NF1 who experience persistent new headaches or a change in their usual physical abilities or activity (like weakness, numbness, or a change in personality or memory) should promptly notify their doctors. Further evaluation may be needed.

HIGH BLOOD PRESSURE, PAIN, HEADACHES, & ITCHING
High blood pressure, also called hypertension, can be seen in persons with NF1. Thus, it is important that adults have their blood pressure monitored yearly by their doctors.

Sometimes the high blood pressure is not specifically related to NF1, inasmuch as high blood pressure is common among people who don't have NF1. Regardless of the cause, high blood pressure is treatable. Rarely, individuals with NF1 can have high blood pressure caused by pheochromocytomas, tumors that are usually benign and secrete certain hormones. In these cases, blood pressure is high only on some occasions; and headaches, sweating, and palpitations may accompany the hypertension. Pheochromocytomas can be treated with drugs and removed surgically.

Some adults complain of chronic *pain.* Back pain is sometimes related to neurofibromas involving the spine that are not easily removed with surgery. For the adult with severe and chronic pain that has been fully investigated, a referral to a doctor who specializes in pain management can sometimes be helpful in reducing the degree of pain experienced. Chronic pain from neurofibromas can often be alleviated by a combination of pain-relief medicines.

Headaches can be a problem for some individuals. Doctors specializing in neurology and/or pain management may have ideas to reduce the frequency and/or intensity of the headaches.

Although it is not understood why *itching* occurs, it is a frequent complaint of persons with NF1. Some people benefit from medications like antihistamines that reduce the itching.

PREGNANCY AND CHILDREN

Women with NF1 may wonder about the effects of their NF1 on pregnancy as well as the effects of pregnancy on their NF1. The answers to these questions are not completely known. We do know that fertility is not reduced in women who have NF1 or in the wives of men with NF1. Anecdotal evidence suggests an increase in the number of neurofibromas with the use of birth control pills; however, this has not been proven to be true. During pregnancy, about half of women report an increase in the number of neurofibromas and/or enlargement of already existing neurofibromas. Although there are no specific pregnancy concerns, women with NF1 tend to have a higher Caesarian section rate than women without NF1; but women with NF1 usually have regular deliveries that do not require a Caesarian section. Some doctors originally reported problems with high blood pressure and preterm labor in pregnant women with NF1; however, more recent studies have not found such correlations. Regardless, women should let their obstetricians know that they have NF1 and also inform their NF1 doctors when they become pregnant.

When the man or woman in a couple has NF1, the decision about whether or not to have children can sometimes be a difficult one and must be decided by the couple themselves based upon what the couple thinks will be best for their family. What one couple decides may not be what is best for another couple and family. We know that when someone with NF1 has children, each child will have a 50-50 chance of having NF1. Someone's personal experience with NF1 may also affect his or her decision about having children. It is important to remember that, because NF1 is so variable from person to person, a child may or may not have the same medical problems as his or her parent. Talking with a genetic counselor before conception is recommended to help persons with NF1 better understand the chances of having a child born with NF1, the spectrum of medical concerns associated with NF1, and genetic testing, as well as alternatives such as adoption and artificial insemination.

Genetic counselors also help by providing additional information, but they will not tell a couple what they should do. Should a couple decide to have children, each child should

be evaluated for features of NF1 by a doctor specializing in NF1 in the first few months after birth and by an ophthalmologist beginning at about 1 year of age.

OTHER MEDICAL CONCERNS

Most adults with NF1 will have *Lisch nodules.* These are the small raised nodules that develop on the colored part of the eye (the iris) as someone ages. Lisch nodules do not affect vision and do not cause any problems but simply serve as a feature helpful in diagnosing NF1.

Some persons with NF1 may have been diagnosed with a *learning disorder* as a child. These learning disabilities are not progressive and will not worsen over time as an adult. Some adults find difficulty with reading, writing, concentration, coordination and finding their way around. They may experience problems in filling in application forms for jobs or for social services. Individuals should consult their doctors, for adult literacy programs and support networks are available to help with these difficulties.

Some adults with NF1 may have had *speech* problems in childhood which may continue in adulthood. There is no evidence that speech problems are progressive in adulthood, and they are usually mild. Individuals may have difficulty in adjusting the rate, volume and pitch (tone) of their speech, and sometimes their speech has a nasal quality. We do not know whether speech therapy iNF1uences these problems.

Epilepsy (seizures) is seen a little more frequently in people with NF1 than in people who do not have NF1. The seizures are usually easily controlled by drugs. Individuals with this complication should consult a neurologist for advice and treatment.

Physical Appearance: Some adults worry about becoming disfigured as a result of their NF1. Although most adults will develop neurofibromas on their skin, severe disfigurement is not common. NF1 is not the same as "Elephant Man's disease", which is a rare and entirely separate condition. Curvature of the spine, called scoliosis, can be seen in growing children, but it is not a feature that starts in adulthood. A scoliosis diagnosed in childhood will persist in adulthood and can occasionally cause some problems. Plexiform neurofibromas can sometimes be disfiguring depending upon their size and where on the body they are located.

Some people with NF1 may be embarrassed by the appearance of the neurofibromas on their skin; and they may be especially self-conscious with regard to the opposite sex. Sometimes it helps to talk about one's feelings about NF1 with friends, family, others who have NF1, or a professional counselor.

The Children’s Tumor Foundation in the United States and the Neurofibromatosis Association in Britain offer support group meetings with discussions to foster mutual support. They help individuals with NF1 in regard to concerns about such things as their appearance, feelings of loneliness, anxiety over possible medical problems, and what to tell others.

SCREENING RECOMMENDATIONS

It is recommended that adults with NF1 have evaluations once a year by a doctor who knows about NF1. The doctor will check blood pressure and perform a detailed physical and neurologic exam. MRI exams are not part of the usual recommendations unless there is a specific concern or problem. Brain MRIs in persons without medical problems are often normal. They may show UBOs (unidentified bright objects). UBOs are bright spots

on the MRI that are often seen in persons with NF1. They are of no clinical significance, do not correlate with other medical problems or cancer, and do not require any follow-up.

ORGAN AND BLOOD DONATION
People with NF1 can donate their blood and organs to other people who are in need of blood or an organ transplant. The person who receives their blood and/or organs will not develop NF1 as a result of the blood/organ donation.

INSURANCE
Occasionally people have had difficulty with their insurance. These individuals should contact their NF specialists who can try to help resolve these issues.

CLINICAL TRIALS
A clinical trial is a research study in which a person participates to help doctors learn more about a particular treatment or way of looking at a medical problem. In a clinical trial, the treatment/evaluation has not yet been proven to work, but the trial is an important step in finding out whether it will be beneficial in the treatment of NF1-related medical problems. The doctor directing the clinical trial will review the potential benefits and risks before a person agrees to participate. Everyone has the right to decline participation. Clinical trials for NF1 are listed on the Children's Tumor Foundation website www.nf.org

FINDING A DOCTOR
It is important that people with NF1 see a doctor who can answer their questions and who knows about NF1. To find a doctor, consult the Children's Tumor Foundation in the United States, or in the United Kingdom, the Neurofibromatosis Association. Websites for both organizations have information on nearby doctors, medical centers, clinics, and NF support coordinators (UK) that advise and treat people with NF1.

SUPPORT GROUPS FOR NEUROFIBROMATOSIS
The Children's Tumor Foundation and the Neurofibromatosis Association are national voluntary non-profit health organizations designed to help people with NF. Their goals are:

- to promote and support research to find the cause(s) and cure for NF
- to provide information to those who are affected by NF and to promote public awareness
- to serve as a resource on NF for medical and health professionals

In the United States, many states have local support groups and chapters of the Children's Tumor Foundation where people can learn more about NF1, meet others with NF1, gain support, and build friendships. To locate a chapter or support group near you, call the Children's Tumor Foundation at (800) 323-7938 or consult the Foundation's website (www.ctf.org).

In the United Kingdom, the Neurofibromatosis Association has a network of family support coordinators who are able to advise on NF specialists in their local areas of the UK. To find the coordinator near you, call the Neurofibromatosis Association at 0208 547 1636 or view its website.

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ADDITIONAL INFORMATION

The Children's Tumor Foundation can be a source of educational materials and support. They can be reached at:

The Children's Tumor Foundation
95 Pine Street, 16th Floor
New York, NY 10005
212-344-6633 or 1-800-323-7938
Email: Info@ctf.org
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