



Neurofibromatosis Type One: A Guide for Educators by Bruce R. Korf, M.D., Ph.D.

The diagnosis of neurofibromatosis (NF) in a child raises many important issues for care—whether at home, at school, or in the doctor's office. Persons with neurofibromatosis can enjoy good health and academic success. Sometimes they have special needs that if given proper attention, increase the chances that they will do well. Some do not require special help, but may have visible manifestations of the disorder that will attract attention.

Families often have mixed feelings about sharing information about neurofibromatosis with school personnel. Some fear that a child known to have NF will be assumed to have medical problems or Learning disabilities, even if he or she does not. They worry that the expectation of learning disabilities will set up a "self-fulfilling prophecy"—that problems will be found even if not present. On the other hand, for anyone involved with a child with NF, knowledge should translate into better care.

The very fact that you have been provided with this brochure may indicate that you are part of a team caring for a child with NF. You will find that much has been written about NF, but not all of it is accurate. Some materials—in spite of good intentions—emphasize severe medical complications, which are rare, but dramatic. This brochure has been prepared to provide educators with up-to-date information on NF and the learning disorders that sometimes accompany the disorder.

WHAT IS NEUROFIBROMATOSIS?

NF is actually a term that encompasses two separate disorders, referred to as NF1 and NF2. Although they share a common name, these are really distinct disorders. Both are genetic conditions that cause tumors to form around nerves, but otherwise there is little resemblance between them. This brochure deals exclusively with NF1. The reason is that NF2 only rarely shows effects in children, and is usually not associated with learning disabilities. In contrast, NF1 generally begins in young children, and is commonly associated with learning problems. A separate brochure is available about NF2 if you are dealing with a child who has this disorder.

NF1 is a complex disorder, in which a number of diverse features may occur. Moreover, one of its hallmarks is diversity of clinical expression from one person to the next—some are quite severely affected. There does not seem to be any way to predict severity or specific complications. No medical test provides such predication, and the condition even varies from person to person in the same family. Overall, we estimate that NF1 causes mild manifestations in about 2/3 of affected individuals. Do not be misled by newspaper or magazine articles, or television programs that depict NF1 as invariably severe. NF1 can produce severe complications, but, fortunately,

these are rare. Unfounded assumptions that a child has a serious medical disorder can do just as much harm as overlooking the medical condition. Please keep in mind that children with NF1 generally enjoy good health.

WHAT ARE THE FEATURES OF NF1?

The signs of NF1 usually begin to appear in childhood or adolescence. If two or more of the following signs are present, a diagnosis of NF1 is confirmed:

- Family history of NF1
- 6 or more light brown (cafe-au-lait) spots on the skin
- Presence of pea-sized bumps (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 shinbone
- Tumor on the optic nerve (optic glioma) that rarely interferes with vision

NF AND CANCER

One of the most feared complications of NF1 is malignancy. Like most other complications, this has gotten a disproportionate amount of attention compared with its frequency. Overall, we estimate a lifetime risk of NF1-related cancer to be around 5%-just slightly higher than the 25% risk of cancer faced by all people—with or without NF1. Sudden painful growth of a neurofibroma can be sign of malignancy that should be brought to medical attention. On the other hand, most neurofibromas that grow are not malignant.

Aside from malignant growths in neurofibromas, some children with NF1 get tumors in the brain. The most common of these involves the optic nerve, and is referred to as an "optic glioma." Most optic gliomas are asymptomatic and do not require treatment. Rarely, they may cause problems with vision or early onset of puberty. In such cases, very effective treatments, using hormone injections, radiation, or chemotherapy, may be used. Other brain tumors are rare in children with NF1. Although these may be heralded by headaches, changes in behavior, or seizures, it is important to mention that most such changes are not indications of brain tumor.

CAN CHILDREN WITH NF1 PARTICIPATE IN PHYSICAL ACTIVITIES?

In general, children with NF1 are not unusually fragile, and do not require special protection. They are capable of participation in a full range of normal activities. The only exception is for those who have specific complications that may place them at risk for injury. The child's physician will point out any restrictions on physical activity.

HOW IS NF1 DIAGNOSED?

NF1 is generally diagnosed by clinical criteria. These include many of the features already mentioned, such as cafe-au-lait spots, neurofibromas, or optic glioma. Other features may not be readily apparent, but will be searched for by a physician. Often, the diagnosis will remain uncertain for many years. This is due to the fact that some features of NF1 are age-dependent. They may not be seen in young children, but only appear as a child gets older. Usually cafe-au-lait spots are the first features to appear. Children with multiple cafe-au-lait spots alone, however, although suggestive of NF1, are not sufficient to make the diagnosis.

At some point, we may have a test for NF1 that will provide a rapid and definitive diagnosis. Until then, clinical follow-up is the general approach. So medical tests, X-

rays, or scans are invariably helpful. The child's physician will decide specific tests on a case-by-case basis.

CAN NF1 BE TREATED?

There is, as yet, no specific medical therapy to prevent or reverse features of NF1. In particular, no medication has yet been found to shrink or prevent neurofibromas. Specific complications, on the other hand, may be treated either medically or surgically depending on the problem. Research may lead to new methods of treatment in the near future. This may involve a child's participation in protocols for experimental treatment. In this case, instructions for school personnel will be provided if necessary.

WHAT CAUSES NF1?

NF1 is not contagious—no amount of contact between an affected and unaffected child can transmit the condition. In fact, NF1 is known to be caused by a change in the structure of a gene. Because of this, NF1 can be transmitted from generation to generation. An affected individual has a 50% chance of passing the condition on to any child.

About 50% of the time, however, an affected child appears to be the only person in the family with the disorder. In such instances, the NF1 gene change has arisen for the first time in the family in the sperm or egg cell that formed the child. These mutations do not seem to arise from any particular cause, but represent random errors in the process of copying genetic information when sperm or egg cells form. It is *not* the consequence of drug or X-ray exposure, or any other factor under the control of the child's parents.

BEHAVIOR AND COGNITIVE CONSEQUENCES OF NF1

One of the most important reasons for school personnel to be aware of NF1 is to insure early recognition of cognitive or behavior problems. It is estimated that about half of individuals with NF1 have some degree of cognitive or behavioral involvement. This means that half of those with the disorder have no cognitive or behavioral effects. It also means, however, that the possibility of such problems should be kept in mind for any child with NF1.

Although we have much left to learn about cognitive and behavior effects of NF1, a few points are well established and are important to recognize.

- Not all children with NF1 have cognitive or behavioral problems.
- Severe problems classifiable as mental retardation are rare and are generally obvious in the first few years of life.
- Cognitive and behavioral manifestations of NF1 are not progressive—that is they do not get worse with time.
- No specific profile of cognitive or behavioral impairment seems to be unique to NF1. Types of learning disorders overlap with those seen in the general population.
- Cognitive and behavioral disorders in children with NF1 respond to the same approaches as are used in children who do not have NF1.

INTELLIGENCE

Intelligence in individuals with NF1 may span the entire range, from below average to well above average. Although studies have shown a tendency for IQ scores in children with NF1 to be "shifted to the left" (to the mid 80s), an individual may be below this or above this. There does not seem to be consistent discrepancy between verbal and performance IQ.

LEARNING DISABILITY

A learning disability (LD) may be defined as a problem with a specific cognitive function necessary for learning in spite of adequate overall intelligence. Wide ranges of learning disabilities are seen in children with NF1, just as in the general population. This range involves both the character of the LD and its severity. There may be problems with reading, math, visuo-spatial recognition, or other functions. Children with NF1 and learning disabilities respond to the same interventions as any child with LD: evaluation of areas of strength and weakness, and providing an educational program tailored to the needs of the child.

BEHAVIORAL PROBLEMS

Behavioral problems associated with NF1 may include similar problems related to attention deficit disorder or hyperactivity as occur in the general population. They also respond to similar interventions: individualized attention, behavioral modification, and, in some cases, medication. Medication should never be used as a sole approach, but can be helpful in some children with NF1, just as in the general population.

Another form of behavioral problems sometimes associated with NF1 should be mentioned. Some children have a migraine syndrome that includes headache, malaise, stomachache and dizziness. These children may feel fatigue and even appear ill. Many will miss many days of school, or be sent frequently to the school nurse. Headache may be a minor feature, and may not be present at all. A medical evaluation should be initiated if these symptoms are present. Children with NF1 and migraine can respond dramatically to appropriate medications.

ADJUSTMENT OF THE CHILD AND FAMILY

Because of the variability of features, some children with NF1 are obviously affected, whereas others are not. It is important to discuss with a child's parents what the child understands about his or her condition. How much and what to tell a child must be individualized to the child's condition, maturity, and level of understanding. If there is any rule of thumb it is that a child old enough to ask questions is old enough to be given honest answers at a level appropriate to maturity.

Children with the rare disfiguring complications of NF1 may be subject to questions or teasing from classmates. This requires a high level of sensitivity, both to the child and to the rest of the class.

Individuals with NF1 face a lifetime of experiencing the unfairness that results from ignorance. It is never too soon to remedy that with age-appropriate, but accurate information.

WHAT TO WATCH FOR

Generally, it is the job of medical professionals, not school officials, to monitor the health of children with NF1. There is a danger of "over-medicalization" of the condition, which is almost as bad as missing complications. On the other hand, it is tragic when a child with NF1 experiences a cycle of school failure and poor self-esteem that results from behavioral or cognitive problems that go undetected. Many physicians suggest that all children with NF1 be formally evaluated for cognitive function. This depends, in part, on the availability of programs for such evaluation in the community. It is difficult to make a rule about this, but at the least, teachers and other school personnel should be aware of the potential for cognitive or behavioral manifestations, without assuming that these will be present in all children with NF1.

WHAT IS REALLY GOING ON?

The past several years have seen an explosion of knowledge about NF1. The landmark accomplishment was the identification of the gene responsible for NF1 on chromosome 17. This has led to new ideas about therapy that will be tested over the next few years. It is believed that cognitive and behavioral effects are due to changes in the structure of the brain due to NF1, but this has not been established with certainty.

The pace of research is increasing rapidly. A reliable diagnostic test is likely to be developed soon. Further clinical research studies including the cognitive/behavior manifestations are much needed.

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
Internet: www.ctf.org