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Neurofibromatosis – A Guide for Educaters

NF1

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Contents

Professor Green
Director, Centre of Medical Genetics

"I am delighted to support and endorse the new information sheets for people with Neurofibromatosis 1 and their families. The information will be of great help to the many families in Ireland with NF1, and will help those families to understand better the many ways in which Neurofibromatosis 1 can affect people. The National Centre for Medical Genetics is delighted to be associated with the Neurofibromatosis Association of Ireland. The NCMG has a wealth of experience with Neurofibromatosis and sees many families with the condition throughout Ireland. The NCMG holds genetics clinics in Dublin, Cork, Limerick and Galway, and is happy to see families with NF1, with a referral from their own doctor.

Prof. Andrew Green

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INTRODUCTION

A diagnosis of Neurofibromatosis type 1 (NF1) in a child raises many important issues for care, whether in the doctor's office, at home or in school. Children with NF1 can enjoy good health and academic success, but many have special needs – especially in a school environment. Proper attention to these needs can dramatically increase the chances that children with NF1 will do well, both in school and in later life.

Classroom and special education teachers, school psychologists and counsellors, occupational therapists, speech therapists, and school nurses can make a significant difference in the lives of children with NF1 by gaining knowledge about the disorder and providing early intervention when and if needed.

We thank you for your concern and interest in reading this brochure. This is a time of exciting progress in our understanding of NF1.

ACKNOWLEDGEMENT

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ABOUT NEUROFIBROMATOSIS

Neurofibromatosis is a term that encompasses three distinct disorders: NF Type 1 (NF1), NF Type 2 (NF2) and Schwannomatosis. All are genetic conditions that cause tumours to form on nerves. This brochure deals exclusively with NF1, which is commonly associated with learning difficulties in addition to tumours.

NF1 strikes one in every 3,000 births and can lead to a wide range of medical problems. The features of NF1 vary greatly from one person to the next. Some children are quite severely affected, while most have considerably milder cases. Although many children with NF1 generally enjoy good health, the disorder can lead to malignancy, loss of vision, bone and skeletal defects, disfigurement, cardiovascular problems and other serious complications.

Research indicates that approximately 50-60% of all children with NF1 have some form of learning difficulty. Many will require special education services for learning difficulties, speech problems, motor deficits, or psychosocial problems. There is also a higher incidence of attention deficit disorder among children with NF1 compared to the general population. In addition to the potential for cognitive difficulties, some children may have visible manifestations of NF1 that draw attention and cause added stress for them in social situations.

THE CAUSE OF NF1

NF1 is caused by a change, or mutation, in the structure of a gene. Because of this, a child can inherit NF1 from a parent who has the disorder. About half of the time, however, a child with NF1 is the only person in the family who has the disorder.

In such instances, the NF1 gene change occurred as the result of "spontaneous mutation" – a random error in the process of copying genetic information.

NF1 is not the consequence of drug, alcohol or X-ray exposure, or any other factor under the control of the child's parents. NF1 is not contagious. No amount of contact between an affected and unaffected child can transmit the condition.

DIAGNOSING NF1

NF1 is generally diagnosed by a geneticist or paediatric neurologist using defined clinical criteria. A diagnosis of NF1 is often made by the presence of six or more pigmented spots on the skin (called café-au-lait patches) in combination with other hallmark features of the disorder.

A separate brochure ("About Neurofibromatosis1") is available from NFA Ireland providing more specific information on the diagnosis and management of NF1.

MEDICAL COMPLICATIONS OF NF1

Generally, it is the job of medical professionals, not school personnel, to monitor the health of children with NF1. However, it is also helpful for educators to be aware of some of the potential complications and manifestations of NF1 that can affect children.

Tumours: Because NF1 is associated with tumour formation, one of the most feared complications is cancer. Fortunately, the most common tumours associated with NF1 (called Neurofibromas) typically are not malignant. They may, however, require surgery to allay the chance that serious complications will develop. It is important to note that any sudden growth or pain in a neurofibroma can be a sign of

malignancy onset and should be brought to the attention of a medical specialist.

Some children with NF1 develop brain tumours. The most common of these are optic gliomas, which involve the optic nerve that controls vision. Typically, these are asymptomatic and do not require treatment, although they can cause problems with vision or early onset of puberty. In such cases, effective treatments are available. Other types of brain tumours are rare in children with NF1. These may be preceded by headaches, seizures, or changes in behaviour – all of which should be medically evaluated as soon as possible after onset. Most such changes are not indications of brain tumour, but medical evaluation is important.

Migraine: Some children with NF1 have a migraine syndrome that can include headache, abdominal pain, nausea, vomiting, malaise, fatigue or dizziness. Headache may be a minor feature, or may not be present at all. These children may miss many days of school or be sent frequently to the school nurse. A medical evaluation should be initiated if these symptoms are present, in order to rule out, for example, that there is a brain tumour. The good news is that children with NF1 can respond dramatically to appropriate medications for prevention and treatment of migraine.

Scoliosis: Children with NF1 are at higher than average risk for scoliosis (abnormal curvature of the spine) which can appear at a much younger age than is typical in the general population and can be detected by regular screening. Early management is critical to preventing serious complications.

Bone defects: Abnormal bone development occurs in approximately 14% of individuals with NF1. Most bone defects of NF1 will be evident at birth or shortly thereafter (some, such as vertebral defects,

can occur later). They can occur in almost any bone, but are seen most often in the skull and limbs.

High blood pressure: Children with NF1 are also at higher than average risk for high blood pressure, and again this can appear at a much younger age than in the general population. This can be detected by regular screening and appropriately managed.

Speech and motor deficits: These are often associated with NF1. Children exhibiting these problems tend to benefit greatly from early intervention through speech and occupational therapy.

PHYSICAL ACTIVITIES FOR CHILDREN WITH NF1

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, for example those relating to bone defects, scoliosis or tumours that may place them at risk for injury. In these cases, the child's physician will point out any restrictions on physical activity.

COGNITIVE & BEHAVIORAL CONSEQUENCES OF NF1

One of the most important complications for school personnel to be aware of in children with NF1 is to ensure early recognition of cognitive or behavioural problems. It is important to remember that at least half of all children with NF1 have some degree of cognitive or behavioural difficulties. The possibility of such problems should be kept in mind for any child with NF1.

It is believed that cognitive and behavioural problems may be caused by changes in the structure of the

brain due to NF1. No specific profile of cognitive or behavioural impairment seems to be unique to NF1. Rather, problems overlap with those seen in the general population – and children with NF1 respond to the same interventions as are used for children with cognitive or behavioural impairment who do not have NF1.

However, it is equally critical to recognise that roughly half of all children with NF1 have no cognitive or behavioural complications. There can be a danger of "over-diagnosing or over-analysing" the condition and assuming that such problems will be present in a child with NF1. For this and many other reasons it is important for educators and parents to work together in evaluating whether a problem exists.

INTELLIGENCE & NF1

As in the general population, intelligence in children with NF1 spans the entire range, from below average to above average. Studies have shown a tendency for IQ scores in children with NF1 to be "shifted to the left" (i.e. lowered) to the mid-80s. However, any one individual may score below or above this.

There does not seem to be consistent discrepancy between verbal and performance IQ in NF1. Severe problems (classifiable as mental retardation) are rare and these are generally obvious in the first few years of life.

NF1 – ASSOCIATED LEARNING DIFFICULTIES

A learning difficulty is defined as a problem with a specific cognitive function that is necessary for learning in individuals with average or even above average intelligence. As in the general population, there is a wide range in the character and severity of

learning difficulties that can be seen in children with NF1 – including both verbal and non-verbal difficulties.

The learning difficulties seen in NF1 can include dysfunction in visual or auditory perception (not to be confused with visual or auditory acuity); information integration (such as sequencing, abstraction or organisation); memory; language; gross motor, fine motor, or oral motor skills (causing clumsiness, poor handwriting, or poor articulation); and social skills or behaviour (including misperception of social cues, attention deficit, or hyperactivity). They may present problems in reading, spelling, maths, spatial ability, neatness, test taking, speech, ability to make friends, or many other functions.

Children with NF1-associated learning difficulties can benefit greatly from evaluating areas of strength and weakness, and from an individualised education program tailored to the unique needs of the child. With early and appropriate intervention, these children can indeed succeed in school.

A separate brochure ("NF1: About Learning Difficulties") is available from NFA Ireland which outlines specific learning difficulties and practical suggestions for classroom modifications that may be helpful.

BEHAVIOURAL PROBLEMS

Behavioural problems associated with NF1 include attention deficit/hyperactivity disorder (ADHD), as occurs in the general population. Although this problem is a direct result of this complex genetic condition, children with NF1 who have behavioural problems respond to the same interventions as any child: individualised attention, behavioural modification and, in some cases, medication.

PSYCHOSOCIAL CONCERNS

Children with learning difficulties often find themselves bullied by fellow students. The rare disfiguring complications of NF1 in childhood are often subject to questions or teasing from classmates, requiring a high level of sensitivity to both the child with NF1 and the other students. This behaviour often results from ignorance among their peer group and can be counteracted by providing accurate, age-appropriate information.

Children with NF1 often suffer form social isolation, poor self-esteem, anxiety or depression. For those coping with the combined burden of medical, learning and social issues associated with NF1, the typical school day can be psychologically exhausting.

MAINSTREAM CLASSES OR SPECIAL EDUCATION?

Many children with NF1 benefit from special education services and this should be sought as early as possible. Children with learning difficulties resulting from NF1 may learn academic skills at a slower rate than their peers; but research informs us that they will, in time, learn the necessary material – just as their classmates do. They often struggle with rote foundation skills despite having adequate comprehension abilities. Educators and parents should maintain high expectations for the child, but not demand mastery to the point of frustration. Children enrolled in special education classrooms should be evaluated regularly to assess whether special services continue to be needed, or if the child is increasingly capable of succeeding in mainstream education classes.

FINDING HELP

Some children with learning deficits may not meet eligibility criteria for receiving special services based solely on evaluation within the school system. However, children with NF1 are eligible for special education as verified by a physician. Speech and occupational therapy may be provided as if needed. Assistive technology services, such as access to laptop computers if handwriting is a problem, are also available and helpful for many children.

COMMUNICATION AMONG PARENTS, CHILDREN & EDUCATORS

Because of the variability of features in NF1, some children are obviously affected by the disorder whereas others are not. It is important for educators to discuss with a child's parents what the child knows about his or her condition. Many parents wait to tell their child about NF until they feel the child is able to handle the information emotionally. How much and what to tell a child must be individualised to his or her particular complications, age, maturity, and level of understanding.

Parents often have mixed feelings about sharing their child's diagnosis of NF1 with school personnel. Some parents fear that a child known to have NF1 will be assumed to have medical problems or learning difficulties, even if he or she does not. They worry that an assumption of learning difficulties may create a "self-fulfilling prophesy" that can lower a teacher's expectations of the child – and, in turn, lower a child's self-esteem and performance – even when no learning problems are present. However, for anyone involved with a child who has NF1, knowledge typically translates to better care.

Thank you for taking the time to learn about this common, yet under-recognised neurological disorder.

CHILDREN'S TUMOR FOUNDATION

The American Children's Tumor Foundation supports persons with NF and their families by providing thorough and accurate information. It also offers a YouthCONNECT program which includes an online youth chatroom. **www.ctf.org**

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NF1 BROCHURES

- OVERVIEW OF NEUROFIBROMATOSIS TYPE 1
- A GUIDE FOR EDUCATORS
- LEARNING & COGNITIVE DIFFICULTIES
- NEUROFIBROMATOSIS TYPE 1 FOR TEENS
- THE CHILD WITH NEUROFIBROMATOSIS TYPE 1
- TALKING TO YOUR CHILD
- INFORMATION HELP & SUPPORT FOR THOSE WHO NEED IT

LEAFLETS

- NF1 REVIEW CHECKLIST FOR CHILDREN & ADULTS
- SCHWANNOMATOSIS

INFORMATION FOR HEALTHCARE PROFESSIONALS

- CLINICAL GUIDELINES FOR THE MANAGEMENT OF INDIVIDUALS WITH NEUROFIBROMATOSIS TYPE1

NEUROFIBROMATOSIS TYPE 2 BROCHURES

- FOR FAMILIES
- FOR HEALTH PROFESSIONALS

Neurofibromatosis is a Little Known Genetic Condition and Can Manifest Itself in a Whole Lot of Different Ways

The care of persons with NF is made complex by the wide range of expression of the disorder. It is difficult to predict the specific problems that will occur in a particular individual. Diagnosis is made if an individual has two or more of the following features.

The diagnosis is based on the following clinical criteria:

1. **Six or more (café au lait)** coffee coloured patches sized 5mm or over in pubertal individuals and over 15mm in size in post pubertal individuals.

2. **Freckling** under the **arm** or in the **groin** area.

3. **Two or more Neurofibromas** of any type (growth of tumours on nerve tissue anywhere on the body) usually first seen on the skin.

4. **Plexiform Neurofibromas –** large bundle of nerves are thickened and appear as a soft tissue mass under the skin, these growths often large, can change the normal shape of the body.

5. **Optic Glioma –** thickening of the optic nerve.

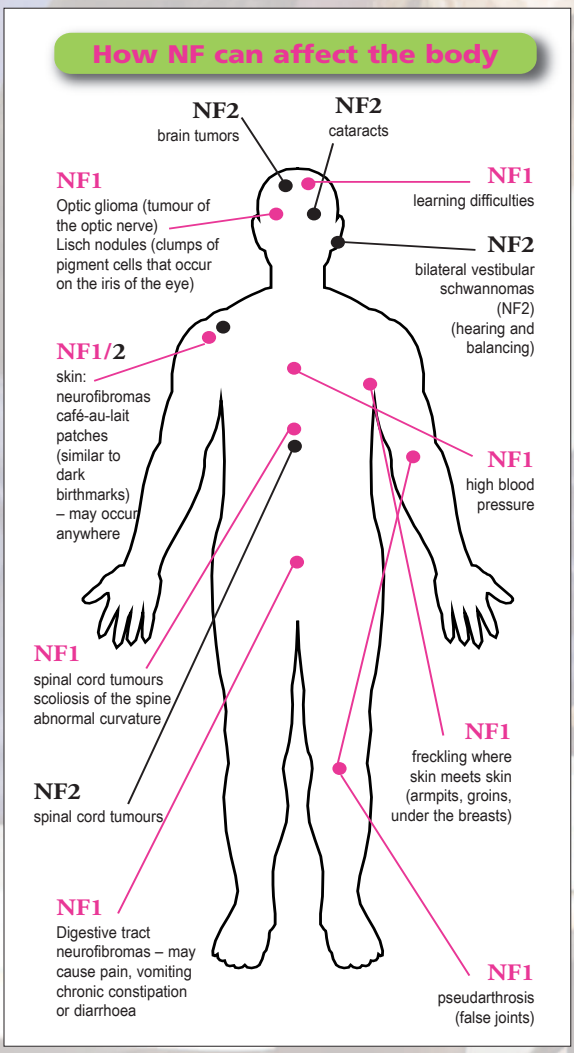
6. **Lisch Nodules –** clumps of pigment cells that occur on the iris of the eye.

7. **Orthopaedic** problems include **scoliosis** (curvature of the spine) **abnormal bone development**, such as overgrowth in long bones causing bowing and deformity that result in fractures, which fail to heal.

8. **First-degree relative with NF** e.g. parent, sibling, offspring.

Neurofibromatosis Type 2

Another rarer type of Neurofibromatosis and distinct in its clinical feature is **NF2.** The gene for **NF2** is located on chromosome 22, Features include: **Vestibular schwannomas** (tumour on hearing nerve). **Schwannoma** (type of tumour of the substance that covers nerve fibres). **Meningiomas** (tumour of the covering of the brain). **Cataract.**



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