NF MIDWEST

Information for Medical Professionals

I've been diagnosed...

Neurofibromatosis Type 1 (NF1) is an autosomal dominant condition with a high degree of variability between individuals. NF1 occurs in approximately 1 in 3000 individuals and 50% of individuals have an affected parent. It affects all ethnicities and occurs equally in males and females. NF1 can be associated with most systems in the body including the cutaneous, neurologic, ophthalmologic, musculoskeletal and cardiovascular systems.

Anticipatory guidance and care is essential to monitor for manifestations that may not be obvious. This information sheet is designed for you to provide basic facts about NF1 and also outline issues that may come up later on. Together, we can ensure that I get the best care possible.

Diagnosis

Two or more of the following signs:

- Six or more café-au-lait macules >5 mm in prepubertal individuals or >15 mm after puberty
- Two or more neurofibromas of any type or one or more plexiform neurofibroma
- Freckling under the arms or in the area of the groin
- Tumor of the optic nerve pathway
- Two or more Lisch nodules (iris hamartomas)
- Distinctive osseous lesion (sphenoid wing dysplasia or long-bone bowing)
- First degree relative with NF by the above criteria

Using clinical evaluation, a diagnosis can be made in 95% of affected individuals by age 11. However, the diagnostic criteria cannot ascertain the severity of the disorder or the prognosis. This can provide significant distress for those newly diagnosed with NF1.

Clinical Manifestations

The clinical picture for any one individual with NF1 is variable and difficult to predict. It is unlikely that an individual will experience all of the listed symptoms but surveillance is required. Clinical manifestations and available frequencies are as follows:

Cutaneous

- Multiple café-au-lait spots (95% or less)
- Freckling under the arm or in the area of the groin (65-85%)
- Dermal neurofibromas (65-85%)
- Xanthogranulomas (2-5%)
- Hemangiomas (5-10%)

Ophthalmologic

- Optic nerve pathway tumor/optic glioma (15%)
- Lisch nodules (65-85%)
- Glaucoma (rare)

Musculoskeletal

- Sphenoid wing dysplasia (1%)
- Long-bone bowing/pseudarthrosis (2-5%)
- Scoliosis (20%-50%)
- Short stature (25-35%)
- Relative macrocephaly (45%)

Cardiovascular

- Hypertension (2-5%)
- Congenital heart defect (2%)

Neurological

- Hydrocephalus (5%)
- Seizures (11%)
- Educational difficulty (40-60%)
- Sensorineural hearing loss (5%)
- Precocious puberty (2-5%)

Tumors

- Plexiform neurofibromas (50%)
- Malignant peripheral nerve sheath tumors/MPNST (5-10%)
- Central nervous system glioma (2%)
- Pheochromocytoma, rhabdomyoma, neuroblastoma (1%)
- Mylogenous leukemia (rare)

Information for

Medical Professionals

Anticipatory Guidance

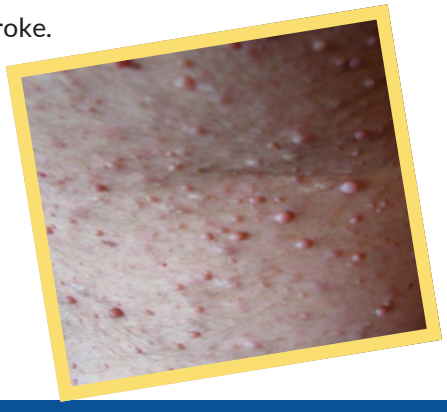
Signs and symptoms are diverse and present in an age-specific manner. Close monitoring is essential.

Newborn to 2 years	*2-10 years*	*10 years to adulthood*	*Adult*
• Café-au-lait spots • Long-bone bowing/ pseudarthrosis • Plexiform neurofibromas • Optic pathway tumors/optic gliomas • Developmental delay assessment	• Optic pathway tumors/optic gliomas • Plexiform neurofibromas • Scoliosis • Hypertension • Freckling patterns • Learning problems • Precocious Puberty	• Onset of dermal neurofibromas • Learning problems • Self-esteem • Scoliosis • Plexiform neurofibromas • Reproductive decisions • Hypertension	• Offspring • Progression of dermal neurofibromas • Malignant peripheral nerve sheath tumors (MPNST) • Hypertension • Plexiform neurofibromas • Increased risk of cancer, including breast cancer • Depression

Management

Anticipatory guidance is vital for monitoring for manifestations that may not otherwise be apparent. An example of this is optic nerve pathway tumors.

Early detection allows for timely treatment and improved outcomes. Other age-related manifestations such as hypertension are easily treated and contribute towards better overall health as well as modifying risk factors for heart disease and stroke.



Treatment for NF1 is largely aimed at controlling symptoms and may involve various medical disciplines including neurology; vascular, plastic and orthopedic surgery. As a general practitioner, your role may be to integrate my care, working with an NF clinic if possible. Together, we can ensure that my health is at its optimum.

Resources:

NF Clinical Trials Consortium
www.uab.edu/nfconsortium
(205) 934-9411

Find a NF Specialist in the Midwest
www.nfmidwest.org/findadoctor

For more information contact:

Neurofibromatosis Midwest
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NF Midwest serves the NF community in Illinois, Indiana, Iowa, Kentucky, Wisconsin, and the east half of Missouri.