

## **Neurofibromatosis 1 (NF1)**

### ***For Healthcare Providers***

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

#### **Physical characteristics and/or symptoms**

*Note: not all people with NF1 will have all of these features.*

- Six or more cafe-au-lait spots.
- Axillary and inguinal freckling
- Multiple cutaneous neurofibromas
- Plexiform neurofibromas
- Lisch nodules.
- Optic glioma
- Distinctive osseous lesion such as sphenoid dysplasia or tibial pseudarthrosis
- Learning disabilities are present in about 50-60% of individuals with NF1.

#### **Other findings in NF1:**

- Headaches
- Scoliosis
- Vasculopathy
- High blood pressure
- Short stature
- Macrocephaly
- Delayed puberty

#### **Recommended Routine Surveillance**

- Annual physical exams by a doctor familiar with NF1
- Annual ophthalmologic exams to include visual field testing when feasible by age
- Regular blood pressure monitoring
- MRI for follow up of clinically suspected intracranial tumors and other internal tumors

#### **Emergency Protocols**

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:

[https://www.aap.org/en-us/Documents/Seizure\\_Action\\_Plan\\_for%20School.pdf](https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf)

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Valvar pulmonic stenosis
  - Vasculopathy
  - Hypertension
- Developmental evaluation:
  - Speech therapy
  - Physical therapy
  - Occupational therapy
- Geneticist / Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Neurologist/Neurosurgeon
  - Seizures
  - Headache/migraines
  - Malignant peripheral nerve sheath tumors
  - Brain stem and cerebellar gliomas
  - Treatment of optic gliomas
- Oncologist
  - Treatment of malignancies or tumors
- Orthopedist
  - Scoliosis
  - Generalized osteopenia
  - Dysplasia of long bones
  - Hypertension
- Ophthalmology
  - Optic gliomas
  - Check for visual field deficits
- Plastic Surgeon/Surgeon

- Plexiform neurofibromas may cause pain, disfigurement and overgrowth of adjacent tissues.

## Sample Forms

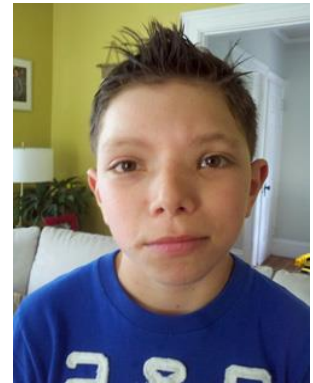
- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with Neurofibromatosis 1 (NF1). NF1 is characterized by abnormalities of the skin, brain, eye, and developmental delays. Medical complications with NF1 syndrome may include management of blood pressure, seizures, brain tumors, plexiform neurofibromas, and headaches. Because of these, \_\_\_\_\_ needs the following accommodations:*

## Seven Important Aspects of School Life

“[NF1 at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

**Gene Reviews- Neurofibromatosis Type 1**

<https://www.ncbi.nlm.nih.gov/books/NBK1109/>

**American Academy of Pediatrics (AAP) – Health Supervision Guidelines**

<http://pediatrics.aappublications.org/content/96/2/368.abstract>

**Children’s Tumor Foundation, including pamphlets**

<http://www.ctf.org/>