A diagnosis of NF1 can be given if an individual has two or more of the following manifestations.

- Optic pathway glioma (tumor of the visual pathway)
- Two or more Lisch nodules or two or more choroidal abnormalities
- Freckling in axilla (armpit) or groin*
- Six or more café-au-lait macules* (brown skin spots)
  - greater than 5mm in pre-pubertal children
  - greater than 15mm in post-pubertal individuals
- Two or more neurofibroma tumors of any type, or one plexiform neurofibroma

A pathogenic NF1 gene variant**

**Additional Genetic Criteria Updates:
- The term “mutation” is no longer accepted; pathogenic variant is now the preferred term.
- Genetic analysis is not REQUIRED for diagnosis but may allow for an earlier diagnosis.
- Genetic analysis ALONE is not sufficient to diagnose NF1 - diagnosis requires a second diagnostic feature of NF1.

Glossary

- Anterolateral bowing of tibia: a curving of a bone in the leg
- Café-au-lait macules: also called café-au-lait spots, are flat darkened areas on the skin
- Choroidal abnormalities: are problems with the vascular layer of the eye (the choroid)
- Dysplasia: means abnormal growth
- Lisch nodules: are small tan or brown bumps on the surface of the iris in the eye
- Neurofibroma: is a tumor that forms on a nerve cell sheath
- Osseous lesion: is a problem with a bone that is not due to injury
- Optic pathway glioma: is a tumor of the visual pathway
- Pseudarthrosis: is a condition that occurs when the bones in the leg do not heal correctly
- Sphenoid: is a bone near the base of the cranium behind the eye

A link to the NF1 diagnostic criteria update publication can be found at: [ctf.org/NF1criteriaupdate](http://ctf.org/NF1criteriaupdate)

Learn more about all types of NF on the Children’s Tumor Foundation website at: [ctf.org](http://ctf.org)

Contact us at 1-800-323-7938
212-344-6633 | info@ctf.org