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

- 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
- Optic pathway glioma (tumor of the visual pathway)
- A distinctive osseous lesion such as: sphenoid dysplasia; anterolateral bowing of tibia (tibial dysplasia); or pseudarthrosis of a long bone
- Two or more neurofibroma tumors of any type, or one plexiform neurofibroma
- A pathogenic NF1 gene variant**

*At least one of the two pigmentary findings (café-au-lait macules or freckling) should be bilateral.

**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.

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

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

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