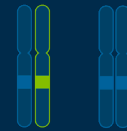
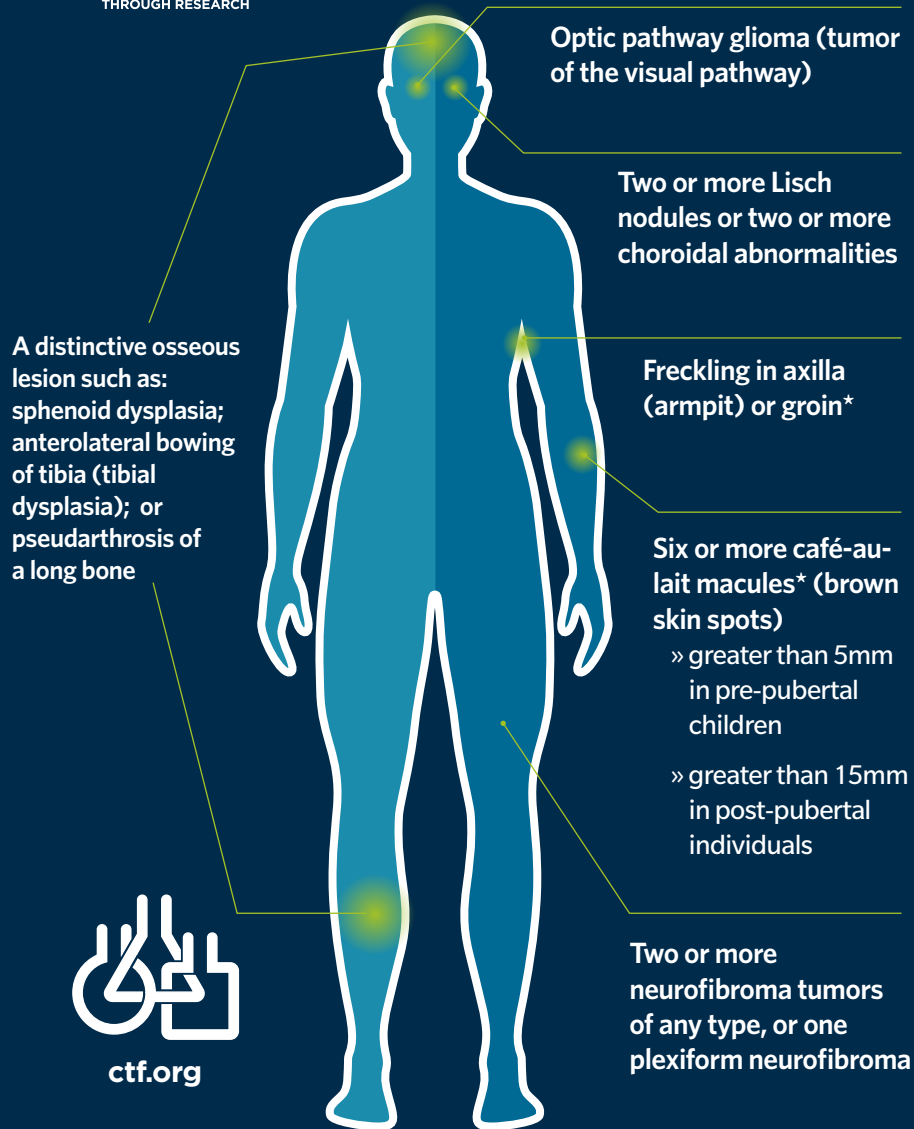


DIAGNOSTIC CRITERIA FOR NEUROFIBROMATOSIS TYPE 1 (NF1)

2021 UPDATE

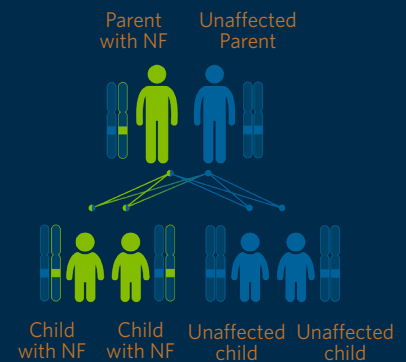
A diagnosis of NF1 can be given if an individual has two or more of the following manifestations.



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.



A parent with NF1 based on diagnostic criteria

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

GLOSSARY

Anterolateral bowing of tibia is 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](https://www.ctf.org/NF1criteriaupdate)

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

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