

## Neurofibromatosis Type 1 (NF1)

**Condition Description:** [Autosomal dominant inheritance](#). Neurofibromatosis type 1 is a condition characterized by changes in skin coloring (pigmentation) and the growth of tumors along nerves in the skin, brain, and other parts of the body. The signs and symptoms of this condition vary widely among affected people. NF1 is caused by mutations in the [Neurofibromin1](#) gene.

**Diagnostic Criteria:** Two or more of the following<sup>1</sup>:

1. Six or more café-au-lait spots 1.5 cm or larger in post-pubertal individuals, 0.5 cm or larger in pre-pubertal individuals
2. Two or more neurofibromas of any type or one or more [plexiform neurofibroma](#)
3. Freckling in the axilla or groin
4. Optic glioma (tumor of the optic pathway)
5. Two or more Lisch nodules (benign iris hamartomas)
6. A distinctive bony lesion: dysplasia of the sphenoid bone or dysplasia or thinning of long bone cortex
7. A first-degree relative with NF1

**Other associated findings:** High blood pressure (hypertension), short stature, macrocephaly, and skeletal abnormalities such as scoliosis. Usually normal intelligence, but learning disabilities and attention deficit hyperactivity disorder occur frequently in affected individuals.

**Related disorders:** [Neurofibromatosis Type 2](#); [Legius syndrome](#) (multiple café-au-lait spots)

**Action required:** Refer for [clinical genetics evaluation](#).  
Follow management recommendations.<sup>2</sup>

**Caveats:**

- Genetic testing is not routinely indicated without genetics evaluation

**Reference/Resources**

1. [Neurofibromatosis. Conference statement](#). National Institutes of Health Consensus Development Conference. Arch Neur 1988 45:575-8.  
(<http://consensus.nih.gov/1987/1987Neurofibromatosis064html.htm>)
2. AAP Committee on Genetics. [Health Supervision for Children with Neurofibromatosis](#) Pediatrics 2008 121:633-642.  
(<http://aappolicy.aappublications.org/cgi/content/full/pediatrics;121/3/633>)
3. Genetic Home Reference - [Neurofibromatosis](#) (<http://ghr.nlm.nih.gov/condition/nf1>)
4. Gene Review – [NF1](#) - (<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=nf1>)

## Patient Resources:

- [Neurofibromatosis, Inc.](http://www.nfinc.org/) <http://www.nfinc.org/>
- [The NF Consortium \(clinical trials\)](http://www.nfconsortium.org) <http://www.nfconsortium.org>
  - **NB The University of Utah is a member of the consortium**  
<http://www.nfconsortium.org/Utah.html>