



NF1 is caused by a mutation in the NF1 tumor suppressor gene on chromosome 17. It is the most common autosomal dominant condition, affecting 1 in 3,000 people. Half of cases arise from *de novo* mutations, and all individuals with the mutation will show symptoms by age 5 (100% penetrance).

AD
Autosomal
dominant

CLINICAL FEATURES



Axillary &
Inguinal
freckling

Eyes
Lisch
Nodules



Positive family history
First-degree relative
with a diagnosis of NF1



C

Café au lait
≥ 6 macules
(> 5 mm in
pre-pubertal
and
> 15 mm in
post-pubertal
children)

A



F

Fibromas
≥ 2
neurofibromas
(any type) or
1 plexiform
neurofibroma

E

Skeletal anomalies
Sphenoid dysplasia
Scoliosis
Bowling
Tibial pseudoarthrosis



S

P

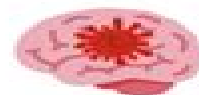
O

Optic

T

Tumor

Optic glioma



ASSOCIATED FEATURES

- Macrocephaly (50%)
- Learning disabilities (50%)
- Short stature (33%)
- ADHD, ASD
- Headaches/migraines
- Seizures
- Congenital cardiac anomalies, vascular stenosis, aneurisms
- Higher incidence of Moya Moya

DIAGNOSIS

- **Clinical diagnosis:** a diagnosis of NF1 can be made in a child with 2 or more of the above criteria
- **Genetic Testing:** mutation in the NF1 tumor suppressor gene on chromosome 17

MANAGEMENT

Developmental assessments:

- Monitor for learning delays.

Cancer Surveillance:

- **CNS tumors:** regular MRI/CT for brain and spinal cord tumors.
- Screen for malignant peripheral nerve sheath tumors, breast cancer, leukemia, colorectal cancer, and soft tissue cancers.

Tumor Management:

- **Surgery:** for symptomatic tumors (e.g., large neurofibromas, optic gliomas).
- **Radiation is avoided** due to risk of 2° malignancies.

Complications:

- **Eye exams:** for optic gliomas and cataracts.
- **Orthopedic care:** monitor for scoliosis and bone deformities.

Genetic Counseling:

- Important for family planning and inheritance risk.

Supportive Care:

- **Psychological support:** address emotional and social impacts.
- **Educational support:** implement IEPs for learning difficulties.



February 2025

Dr. Katharine V. Jensen (Pediatric Resident, University of Alberta) and Dr. Karen Forbes (Professor of Pediatrics, University of Alberta) for www.pedscases.com

Neurofibromatosis Type 1 In Childhood

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**Audrey M. Walker, Cynthia R.
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