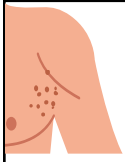
Neurofibromatosis Type 1 (NF1)

Chr 17

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**).

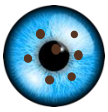
AD
Autosomal dominant

CLINICAL FEATURES

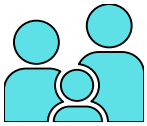


Axillary & inguinal freckling

Eyes
Lisch Nodules



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



C A F E S P O T

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

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

Skeletal anomalies
Sphenoid dysplasia
Scoliosis
Bowing
Tibial pseudoarthrosis

Optic
Optic glioma

Tumor

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 2º 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.

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