

NEUROFIBROMATOSIS TYPE 1 (NF1)





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 no vo mutations, and all individuals with the mutation will show symptoms by age 5 (100% penetrance).

AD Autosomal dominant

CLINICAL FEATURES



Axillary & inguinal freckling Lisch Nodules Positive family history First-degree relative with a diagnosis of NF1



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S







Café au lait ≥ 6 macules

(> 5 mm in pre-pubertal and > 15 mm in

post-pubertal children) AND SHE

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

Skeletal anomalies

Sphenoid dysplasia Scoliosis Bowing Tibial pseudoarthrosis



Optic

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.



SJ Ball

The book delves into Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood. Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood is an essential topic that needs to be grasped by everyone, ranging from students and scholars to the general public. The book will furnish comprehensive and in-depth insights into Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood, encompassing both the fundamentals and more intricate discussions.

- 1. This book is structured into several chapters, namely:
 - Chapter 1: Introduction to Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood
 - o Chapter 2: Essential Elements of Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood
 - Chapter 3: Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood in Everyday Life
 - Chapter 4: Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood in Specific Contexts
 - Chapter 5: Conclusion
- 2. In chapter 1, the author will provide an overview of Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood. The first chapter will explore what Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood is, why Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood is vital, and how to effectively learn about Neurofibromatosis Type 1 In Childhood.
- 3. In chapter 2, this book will delve into the foundational concepts of Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood. The second chapter will elucidate the essential principles that need to be understood to grasp Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood in its entirety.
- 4. In chapter 3, the author will examine the practical applications of Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood in daily life. This chapter will showcase real-world examples of how Neurofibromatosis Type 1 In Childhood Can be effectively utilized in everyday scenarios.
- 5. In chapter 4, the author will scrutinize the relevance of Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood in specific contexts. This chapter will explore how Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood is applied in specialized fields, such as education, business, and technology.
- 6. In chapter 5, this book will draw a conclusion about Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood. The final chapter will summarize the key points that have been discussed throughout the book. This book is crafted in an easy-to-understand language and is complemented by engaging illustrations. This book is highly recommended for anyone seeking to gain a comprehensive understanding of Neurofibromatosis Type 1 In Childhood Neurofibromatosis Type 1 In Childhood.

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