

Neurofibromatosis

Hajra Fayyaz

1st year MBBS, Islamabad Medical and Dental College, Islamabad, Pakistan

Key points:

- Introduction
- Types
- Symptoms
- Causes
- Treatment and Medication

Introduction

Neurofibromatosis is a group of genetic disorders that cause tumors to form on nerve tissues. These tumors are usually benign but sometimes can become malignant. The condition affects the development and growth of nerve cell tissues, leading to skin changes, bone deformities, and other complications.

Types

There are three types of neurofibromatosis:

- I. Neurofibromatosis type 1
- II. Neurofibromatosis type 2
- III. Schwannomatosis

Symptoms

NF₁:

- Café-au-lait spots
- Freckling
- Neurofibromas
- Lisch nodule
- Optic glioma
- Learning disabilities

NF₂:

- Hearing and balance issues
- Spinal and cranial nerve tumors
- Eye problems
- Muscle weakness

Schwannomatosis:

- Chronic pain
- Numbness
- Muscle weakness
- Masses under skin

Causes

Neurofibromatosis type 1:

It is most prevalent, accounting for 96% of all cases and characterized by neurofibromas (peripheral nerve tumors) that induces skin changes and bone deformation¹.

- Mutation in NF gene that is located on chromosome 17 which produces a protein neurofibromin.

- It is autosomal dominant inherited disease it means that if one parent has NF 1 child has 50% chances of having NF 1.

- 50% occurs due to spontaneous mutation.

1. Neurofibromatosis type 2:

- NF2 gene mutation on 22q12 gene which produces merlin protein².
- Chromosomal instability
- Epigenetic factor

2. Schwannomatosis:

- Mutation in SMARCB1 gene³
- LZTR1 mutation
- Sporadic cases

Treatment and management

There is no specific treatment for neurofibromatosis. However preventive measures can help to live comparatively healthy life.

- Regular check-ups
- Educational support

Medication

- Selumetinib
- NSAIDs

Radiation therapies are also preferred in treatment.

References

1. Tamura R. Current understanding of neurofibromatosis type 1, 2, and schwannomatosis. International journal of molecular sciences. 2021 May 29;22(11):5850.
2. Friedman JM. Neurofibromatosis 1.
3. Schraepen C, Donkersloot P, Duyvendak W, Plazier M, Put E, Roosen G, Vanvolsem S, Wissels M, Bamps S. What to know about schwannomatosis: a literature review. British journal of neurosurgery. 2022 Mar 4;36(2):171-4.