Neurofibromatosis Type 2 (NF2)

Neurofibromatosis type 2 (NF2-related schwannomatosis) is a genetic condition that causes tumors to grow around nerves, often in the brain and spine. These tumors are usually non-cancerous (benign). But because of where they grow, they can cause serious problems and may need to be treated with surgery or radiation therapy.

What is neurofibromatosis type 2 (NF2)?

NF2, also known as **NF2-related schwannomatosis**, is a genetic condition that causes tumors to grow around nerves. These tumors are often non-cancerous (benign), commonly found in the brain and spine.

Signs and symptoms of neurofibromatosis type 2 (NF2)

NF2 symptoms can vary from person to person. Symptoms of NF2 can include:

- **Vestibular schwannomas (acoustic neuromas)** are present in almost all patients with NF2. These tumors form from Schwann cells and involve the nerves that connect the ear to the brain. It can lead to hearing loss, ringing in the ears (tinnitus),

or balance problems. Vestibular schwannomas typically develop on both hearing nerves.
- **Meningiomas** are tumors that form from meninges, the protective covering of the brain and spinal cord. Most meningiomas are intracranial (located within the skull). People with NF2 are more likely to have multiple meningiomas.
- **Ependymomas** are tumors that form from ependymal cells, which line the ventricles of the brain and the spinal cord. People with NF2 are at a higher risk of developing ependymomas, especially in the spinal cord.
- **Cataracts** cloud the lens in the eyes and cause vision problems. This can happen earlier than normal in patients with NF2.
- **Neuropathies** can occur when a tumor compresses a nerve, and causes weakness or numbness in the arms, legs and or face. This can cause facial paralysis, and speech or swallowing difficulty.

What causes neurofibromatosis type 2 (NF2)?

NF2 is caused by a change (mutation) in a gene called ***NF2***. The *NF2* gene, located on chromosome 22, makes a protein called **merlin** (also known as **schwannomin**). Merlin is a tumor suppressor protein[1] and helps control cell growth. When *NF2*is mutated, it can no longer make normal merlin, which allows tumors to form.

Approximately 50% of NF2 cases are inherited from a parent, while the other 50% of cases arise from spontaneous mutation in the NF2 gene.

How common is neurofibromatosis type 2 (NF2)?

NF2 is rare and affects about 1 in 25,000 people worldwide.

How is neurofibromatosis type 2 (NF2) diagnosed?

NF2 can be diagnosed by looking at symptoms, family history, imaging tests, and genetic testing. These include:

- The presence of **bilateral** (both ears) **vestibular schwannomas** is the main sign of NF2.
- A **family history** of NF2
- The presence of **other tumors,** such as meningiomas or spinal tumors, even if

there are no vestibular schwannomas.
- **Genetic Testing** is commonly done to confirm that diagnosis of NF2.

Does neurofibromatosis type 2 (NF2) increase a person's risk for cancer?

Most tumors in NF2 are non-cancerous (benign), but because of where they grow, they can cause serious problems, such as pain, weakness, and numbness. Rarely, these tumors can become cancerous.

How is neurofibromatosis type 2 (NF2) managed and treated?

There is no cure for NF2, but regular check-ups and treatments can help manage symptoms and complications.

Management

- MRI of the brain and spine to check for tumor growth
- Hearing and vision tests to catch changes early
- Neurological exams to check for problems with nerves

- Hearing aids to help with hearing loss.
- Physical therapy and occupational therapy to improve balance and movement.

Treatment

Treatment depends on the type and location of the tumor:

- **Watchful waiting:** Doctors may monitor (watch) tumors that aren't causing problems.

- **Surgery:** Tumors that are causing hearing loss, pain, or pressure may need to be removed. Surgery can have risks, especially for tumors near important nerves.
  Radiation therapy:

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