



Oncogenetic testing for persons with Neurofibromatosis type 2

Neurofibromatosis type 2 is a multiple neoplasia syndrome that results from a mutation in the NF2 tumour suppressor gene. The genotype occurs in one in 25 000 live births and is inherited as an autosomal dominant trait. It has wide phenotypic variability. The penetrance is nearly 100% by 60 years of age.

Improvements in diagnosis and treatment have led to a rise in the diagnostic prevalence to one in 100 000 people.¹

Diagnostic criteria

Diagnostic criteria for NF2 were developed based on consensus, commonly referred to as the '**Manchester criteria**' that are an expansion (additional criteria) of and include the **NIH criteria**:

- Bilateral vestibular schwannomas (VS) *OR*
- Family history of NF2 *PLUS*
- Unilateral vestibular schwannoma (VS) *OR*
- Any two of: meningioma, glioma, neurofibroma, schwannoma, posterior subcapsular lenticular opacities

Additional criteria:

- Unilateral VS plus any two of: meningioma, glioma, neurofibroma, schwannoma, and posterior subcapsular opacities
- Or
- Multiple meningioma (two or more) plus unilateral VS or any two of: glioma, neurofibroma, schwannoma, and cataract

Clinical recommendations

- Patients suspected with NF2 should be referred to a centre for **genetic counselling and testing**.
- Decision to test for NF2 should be based on clinical grounds. Manchester criteria (see above) can provide a guidance but clinical judgment is needed especially with early manifestations, as the sensitivity of the Manchester criteria is low.



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Follow-up of NF2 patients should take place at a specialised multidisciplinary NF clinic:

- Ophthalmological examinations are recommended to begin at birth.
- Audiological examinations are suggested to start in early childhood.
- An annual full neurological examination is advised.
- Gadolinium-enhanced magnetic resonance imaging (MRI) of the head and full spine, starting around age 10–12 years, is recommended for all patients, as tumour growth may occur without symptoms.
- It may be sufficient to perform MRIs every other year up to age 20 and every 3 years thereafter for asymptomatic at-risk individuals without tumours.
- If tumours are present, MRIs should be conducted at least annually until the rates of tumour growth are established.
- Prenatal preimplantation diagnosis should be discussed with the patient.

Reference

1. Asthagiri AR, Parry DM, Butman JA, Kim HJ, Tsilou ET, Zhuang Z, et al. Neurofibromatosis type 2. *Lancet*. 2009;373(9679):1974-86.

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