Neurofibromatosis Type 2

Review Guidelines

Current and revised Manchester criteria for NF2:

ANNUAL REVIEW RECOMMENDED	
At time of diagnosis, or possible diagnosis, ALL patients should be seen in a genetics department. Those with significant complications will be followed up as appropriate through the nationally funded Complex NF2 Service. Annual review should be undertaken by a Community/District Paediatrician and GP throughout childhood, and by a GP in adulthood. Patients, paediatricians, and GPs have telephone access to the NF Service in Genetic Medicine for NF-related concerns.	
Diagnosis Definition	
Diagnosis definition 1	• Bilateral Vestibular Schwannomas (<70 years old)
Diagnosis definition 2	• First-degree relative family • history of NF2 with • Unilateral Vestibular • Schwannoma (<70 years old)
Diagnosis definition 3	First-degree relative family history of NF2 paired with any two tumour types such as: • Schwannoma • Neurofibroma • Cerebral calcification • Glioma • Cataract • Meningioma If patient also had >2 no-intradermal schwannomas then LZTR1 testing should be carried out on tumour tissue. However, as mosaicism does not appear as common in LZTR1 affected individuals, blood will still give a useful result and probably exclude a germline schwannomatosis condition.
Diagnosis definition 4	Multiple meningiomas (two or more) and two tumours of the following types: • Unilateral Vestibular Schwannoma • Schwannoma • Neurofibroma • Cerebral calcification • Glioma • Cataract • Meningioma
Diagnosis definition 5	Constitutional or mosaic pathogenic NF2 gene mutation in blood or identical mutations in two distinct tumours.

(Under Revision)

Source:

Smith M. J et al (2017) revisiting Neurofibromatosis Type 2 diagnostic criteria to exclude LZTR1-related Schwannomatosis. Neurology. Jan 3; 88 (1): 87-92.

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