Neurofibromatosis Type 2

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KEYWORDS

• Neurofibromatosis type 2 • Vestibular schwannoma • Acoustic neuroma

KEY POINTS

- Care of patients with neurofibromatosis type 2 (NF2) requires knowledge of all tumors and symptoms involved with the disorder.
- It is recommended that patients receive care in a center with expertise in NF2.
- The role of the neuro-otologist in this care is determined by the specialty center.

INTRODUCTION

Neurofibromatosis (NF) is a rare syndrome characterized by bilateral vestibular schwannomas, multiple meningiomas, cranial nerve (CN) tumors, spinal tumors, and eye abnormalities. Neurofibromatosis type 2 (NF2) presents unique challenges to the otologist because hearing loss may be the presenting complaint leading to the diagnosis of the disorder. NF2 is invasive, requiring a multispecialist team approach for the evaluation and treatment of the disorder. The primary impairment is hearing loss resulting from bilateral vestibular schwannomas. NF2 must be differentiated from neurofibromatosis type 1 (NF1); although the names are linked, the disease entities are distinctly different. This article reviews the clinical characteristics of NF2 and current recommendations for evaluation and treatment.

NEUROFIBROMATOSIS TYPE 2 DIFFERENTIATED FROM NEUROFIBROMATOSIS TYPE 1

NF1 has distinctly different characteristics from NF2. NF1 and NF2 have been distinguished as completely different genetic diseases based on the chromosome responsible for the disease. NF1 has been localized to chromosome 17, and NF2 has been localized to chromosome 22. NF1 is a multisystem disorder in which some features may be present at birth and others are age-related manifestations. The National Institutes of Health (NIH) Consensus Development Conference identified the following 7

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features of the disease, of which 2 or more are required to establish the diagnosis of NF1:

1. Six café au lait spots equal to or greater than 5 mm in longest diameter in prepubertal patients and 15 mm in longest diameter in postpubertal patients
2. Two or more neurofibromas of any type or 1 plexiform neurofibroma
3. Freckling in the axilla or inguinal regions
4. Optic glioma (optic pathway glioma)
5. Two or more Lisch nodules (iris, hamartoma)
6. Distinct osseous lesions, such as sphenoid wing dysplasia or cortical thinning of the cortex of long bones with or without pseudoarthrosis
7. First-degree relative (parent, sibling, or child) with NF1 according to the criteria listed earlier

Some patients also manifest learning disabilities or language disorders. A careful examination and a detailed history of the patient's symptoms help distinguish NF1 and NF2.

CLINICAL CHARACTERISTICS OF NEUROFIBROMATOSIS TYPE 2

Definition

The NIH Consensus Development Conference also developed guidelines for the diagnosis of NF2. NF2 is distinguished by bilateral vestibular schwannomas with multiple meningiomas, CN tumors, optic gliomas, and spinal tumors. A definite diagnosis is made from the presence of bilateral vestibular schwannomas or developing a unilateral vestibular schwannoma by age 30 years and a first-degree blood relative with NF2, or the presence of a unilateral vestibular schwannoma and developing at least 2 of the following conditions known to be associated with NF2: meningioma, glioma, schwannoma, or juvenile posterior subcapsular lenticular opacity/juvenile cortical cataract (**Box 1**).[1]

Box 1
NF2 diagnostic criteria

Confirmed (definite) NF2

Bilateral VS or family history of NF2 (first-degree family relative) plus

1. Unilateral VS less than 30 years, or
2. Any 2 of the following: meningioma, glioma, schwannoma, juvenile posterior subcapsular lenticular opacities/juvenile cortical cataract

Presumptive or probable NF2: should evaluate for NF2

Unilateral VS less than 30 years plus at least 1 of the following:

Meningioma, glioma, schwannoma, juvenile posterior subcapsular lenticular opacities/juvenile cortical cataract

Multiple meningiomas ($\geq$2) plus unilateral VS less than 30 years or one of the following:

Glioma, schwannoma, juvenile posterior subcapsular lenticular opacities/juvenile cortical cataract

Abbreviation: VS, vestibular schwannoma.

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