

Case Report

A rare retroperitoneal schwannoma in a patient with neurofibromatosis Type 2

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Abstract

Neurofibromatosis Type 2 (NF2) is a dominantly inherited tumour-prone disorder, characterized by the development of multiple schwannomas, meningiomas and ependymomas. Its prevalence is around 1:60 000. Vestibular schwannoma (VS) is the hallmark of NF2. Retroperitoneal schwannomas are expected to occur in only 3% of cases. We present the case of a large retroperitoneal schwannoma in a patient with NF2. A well-circumscribed heterogenic mass ($9.5 \times 4 \times 4$ cm) behind and under the left kidney and extending into the left retroperitoneal space was revealed during a lumbar and retroperitoneal space magnetic resonance imaging (MRI). Brain, orbits, cervical, thoracic and lumbar MRI revealed bilateral VS, multiple meningiomas as well as multiple schwannomas and ependymomas in the cervical, thoracic and lumbar spine. The retroperitoneal mass represents a schwannoma probably derived from an intercostal nerve. The patient underwent neurosurgical excision of the VS, and 3 months later, the patient's condition remained stable.

Keywords: meningioma; neurofibromatosis Type 2; retroperitoneal; schwannoma

Introduction

Neurofibromatosis Type 2 (NF2) is a dominantly inherited tumour-prone disorder, characterized by the development of multiple schwannomas, meningiomas and ependymomas. Its prevalence is estimated to be around 1:60 000 [1]. Moreover, retroperitoneal schwannomas are expected to occur in only 3% of the cases [2].

Bilateral schwannoma involvement of the superior vestibular branch of the eighth cranial nerve, known as vestibular schwannoma (VS), is the hallmark of NF2. Hearing loss, tinnitus or imbalance or a combination of the three symptoms are expected to be found in patients with VS. Schwannomas of other cranial, spinal and peripheral nerves; meningiomas, both intracranial (including optic

nerve meningiomas) and intraspinal; and some low-grade central nervous system malignancies (ependymomas and gliomas) are other main tumours found in NF2. Reduced visual activity due to various causes, juvenile cataract (posterior subcapsular opacities), epiretinal membranes, combined pigment epithelial and retinal hamartomas and optic disc gliomas represent the ophthalmologic stigmata of NF2 [1,3]. VS are expected to be found in more than 96% of the patients. Additionally, spinal tumours are almost as frequent in these patients (90%) [4]. NF2 is caused by inactivating mutations of a tumour suppressor gene found on chromosome 22q12 [3]. More than 50% of patients present new mutations, and as many as one-third of patients are mosaic for the underlying disease-causing mutation. The diagnostic approach is based both on clinical and neuroimaging studies. The Manchester [modified National Institutes of Health (NIH)] diagnostic criteria for NF2 are shown in Table 1 [1].

Case presentation

We report a case of a 24-year-old woman referred to us due to a retroperitoneal mass observed adjacent to her left kidney. This was an accidental finding on a magnetic resonance imaging (MRI) prescribed due to lower back pain. A well-circumscribed heterogenic mass posterior and inferior to the left kidney, measuring $9.5 \times 4 \times 4$ cm, was revealed (Figure 1). The tumour presented two compartments; the upper compartment showed both cystic and solid elements, while the lower compartment was divided by septa and presented only cystic elements. From the patient's personal history, learning difficulties had been reported since she was 6 years old. She also reported altered vision at the age of 8. At this time, an epiretinal membrane near the macula in the right eye, strabismus, myopia and astigmatism in the left eye were diagnosed. At the age of 14, the patient started suffering from lower back pain. By the age of 22, she could not walk easily due to muscle weakness and imbalance. During the past year, the patient's hearing declined, and lower back pain worsened, radiating to the



Fig. 1. MRI revealing a well-circumscribed heterogenic mass posterior and inferior to the left kidney, extending into the left retroperitoneal space sized $9.5 \times 4 \times 4$ cm.

Table 1. Diagnostic criteria for NF2 (including the NIH criteria with additional criteria)

| Main criteria | Additional criteria |
|---|--|
| Bilateral VS <i>or</i> family history of NF2 <i>plus</i> | Unilateral VS <i>plus</i> any two of: meningioma, glioma, neurofibroma, schwannoma and posterior subcapsular opacities |
| 1. Unilateral VS <i>or</i> | <i>or</i> |
| 2. Any two of: meningioma, glioma, neurofibroma, schwannoma and posterior subcapsular opacities | Multiple meningioma (two or more) <i>plus</i> unilateral <i>or</i> any two of: glioma, neurofibroma, schwannoma and cataract |

NF2, neurofibromatosis Type 2; NIH, National Institutes of Health; VS, vestibular schwannomas.

right lower extremity. There were no other findings from her personal and family history.

Clinical assessment revealed a positive Lasègue sign at the right side, left side nystagmus and a left side positive Romberg test. She also presented cerebellar gait. Slit lamp examination revealed optic papilla oedema.

The routine laboratory study was within the normal range. Brain, orbits, cervical, thoracic and lumbar MRI showed bilateral VS, multiple meningiomas and schwannomas and ependymomas in the cervical, thoracic and lumbar spinal cord. The right VS was 1×2 cm, whereas the left one, which was larger, at 4.7×3.5 cm, was compressing and displacing the brainstem and the fourth cilia (Figure 2). Schwannomas derived from the right and left trigeminal nerve were of 1.5 and 2 cm in diameter, respectively. Schwannomas seen in the left cavernous sinus in the right optic foramen were 1.5 and 1 cm in diameter. Multiple meningiomas were detected in the falx cerebri.



Fig. 2. MRI revealing bilateral vestibular schwannomas.

Spinal cord MRI also detected large neurinomas of 3.3 cm diameter at the C5–C6 level and smaller ones at the L4–L5, C5–C6 and C6–C7 levels. Ependymomas at the C7–T1, T3, T7–T9 and T11 levels were also present. A meningioma at the T8 level was revealed as well. Finally, another neurinoma was present at the right axilla. The retroperitoneal mass, which represented a schwannoma, probably derived from an intercostal nerve. These findings correspond to NF2.

The patient was referred to a neurosurgeon for further treatment. Surgical excision of the VS was performed and, 3 months later, the patient's condition was found to be stable.

Discussion

Retroperitoneal schwannomas, particularly the large ones, represent extremely rare tumours. When a schwannoma arises retroperitoneally, it usually reaches a large size, which might raise suspicion for malignancy [5,6]. Retroperitoneal schwannomas predominate in women [5,7]. The tumour's origin is difficult to define, and a preoperative diagnosis is achieved with difficulty due to lack of specific clinical symptoms or laboratory data [5]. Nevertheless, surgical removal of symptomatic cranial and spinal tumours is the mainstay of management. Radiation therapy may prove to be an effective alternative [1]. Treatment choices in NF2 often rely on weighing the possible consequences of progressive brainstem compression and gradual cranial nerve dysfunction against the potential for devastating combinations of cranial nerve deficits from surgical or radiation treatment [8]. Surgical removal almost always leads to total deafness with loss of the cochlear nerve. Complete surgical excision may suffice in limited disease [9]. Treatment of a small series of NF2 using erlotinib and bevacizumab

has recently been reported, showing apparent benefit in some cases: improved hearing in some, but not all patients, and a reduction in the size of most growing VS [8,10]. NF2 remains a life-limiting and life-devastating condition. Prognosis is adversely affected by early age at onset, a higher number of meningiomas and the presence of a truncating mutation [1]. Five-, 10- and 20-year survival rates in patients with symptom onset at an age younger than 25 years are 80, 60 and 28%, respectively, whereas in patients with onset at an age of 25 years or older, the rates are 100, 87 and 62%, respectively. Patients with smaller VS at diagnosis (<2 cm in diameter) have better survival rates [11].

In conclusion, NF2 presents an extremely rare condition. Unfortunately, presenting signs and symptoms frequently appear in early adolescence and are difficult to discriminate. Nevertheless, NF2 should be considered in the differential diagnosis of a retroperitoneal mass with generalized symptoms.

Conflict of interest statement. None declared.

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