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CASE REPORT

Abdominal schwannoma in a case of neurofibromatosis type 2: A report of a rare combination

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ABSTRACT

Neurofibromatosis-2 (NF2) is an autosomal-dominant disease, which is characterized by vestibular schwannomas (VSs) (acoustic neurinoma) as well as tumours of the peripheral and central nervous system, demonstrating a variety of expression. A 12-year-old girl presented to us with headache and ataxia for four months. We examined and found a lump in the right side of her abdomen. On magnetic resonance imaging (MRI) of brain, a bilateral VS at the cerebellopontine (CP) angle was detected, and on computerized tomography (CT) scan and ultrasonography of her abdomen a large retroperitoneal schwannoma was revealed in the right side of her abdomen. At first, the right-sided CP angle tumour and two months later, the left-sided lesion was operated. After some days, she became mute and incontinent, and was found to have hydrocephalus on CT scan. We introduced a ventriculoperitoneal shunt. Then we operated the abdominal lump, which was histologically proven as schwannoma. The association of these three tumours is rare and untiring surgical approaches made her better. The patient recovered well except bilateral mild facial and vestibulocochlear deficit.

Key words: Schwannoma, neurofibromatosis type 2, abdominal schwannoma

Introduction

Neurofibromatosis type 2 (NF2) is an autosomal, dominantly inherited genetic disorder characterized by neoplastic and dysplastic lesions of Schwann cells (schwannomas and schwannosis), meningeal cells (meningiomas and meningioangiomatosis), and glial cells (gliomas and glial microhamartomas).[1] Its prevalence is estimated to be about 1:60,000.[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 intra-cranial (including optic nerve meningiomas) and intra-spinal; and some low-grade central nervous system malignancies (ependymomas and gliomas) are other main tumours found in NF2. VSs are expected to be found in more than 96% of patients. Additionally, spinal tumours are almost as frequent in these patients (90%).[3] Here we report a case of NF2, where there were bilateral VS and large retroperitoneal schwannoma.

Case Report

A 12-year-old girl presented to us with headache and ataxia for two months. Her headache was moderate to severe in intensity, more in the morning, and associated with nausea and vomiting. She had no other complaints then. On a general examination, she had no deformity or abnormal pigmentation. On neurological examination, she had normal higher psychic function with intact cranial nerves except bilateral papilloedema and balance problem. Her motor and sensory examination revealed no other abnormality. But her gait was ataxic and Romberg's test was positive. She had neither any cerebellar signs nor any spasticity.

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Examination of other systems revealed no abnormality except a fixed mass in the right side of her abdomen. The mass was palpable, 5 cm away from the costal margin at the anterior axillary line, and it was about 10 cm long along its long axis. It was firm in consistency with an ill-defined margin.

MRI was performed and it revealed a bilateral VS [Figure 1]. Pure-tone audiometry (PTA) revealed right-sided mild hearing loss but left-sided normal hearing. Ultrasonography revealed a large retroperitoneal mass in the right side of her abdomen. CT scan of abdomen revealed a retroperitoneal mass just in front of the right kidney [Figure 2]. At first, we operated on the right cerebello-pontine angle through posterior fossa craniectomy in the sitting position. Near-total removal of tumour was accomplished. It was confirmed as schwannoma by histopathology [Figure 3], where the sections show a capsulated spindle cell neoplasm composed of Antoni A and Antoni B areas.

Her recovery was normal except right-sided mild facial palsy and mild hearing loss confirmed by PTA. Two months later, we operated on the left-sided tumour [Figure 4]. It was also near totally removed and confirmed as before. Then she also had mild facial palsy and mild hearing loss in her left side.

She recovered well post-operatively, but after 2 weeks she became drowsy. One month after the operation, she became mute and developed urinary incontinence. She was again investigated by CT scan of the head, which revealed communicating hydrocephalus. She was treated with a ventriculoperitoneal shunt inserted into the right side. After 5 days, the shunt was found to be non-functional. The shunt was re-inserted into the left side of the abdomen, keeping the cranial end *in situ*. After this, she started to gain her consciousness gradually. She was then discharged.

After 2 months, she came for removal of the abdominal mass. We accessed her abdomen through a right transverse incision for the lump and debulked the tumour gradually and removed it completely. The patient eventually recovered well except bilateral mild seventh and eighth nerve palsy.

Discussion

NF2 was first described in 1820 by the Scottish surgeon JH Wishart.[4] The diagnostic criteria for NF2 included bilateral VSs or a family history of NF2 plus: (1) Unilateral VS or (2) any two of the following: Meningioma, glioma, neurofibroma, schwannoma, and posterior sub-capsular lenticular opacities. Additional criteria

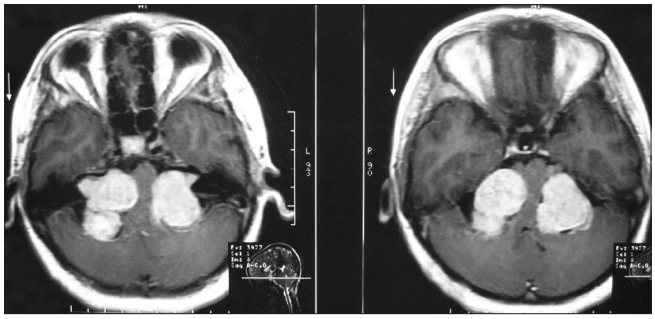

Figure 1: Contrast-enhanced MRI showing bilateral VS

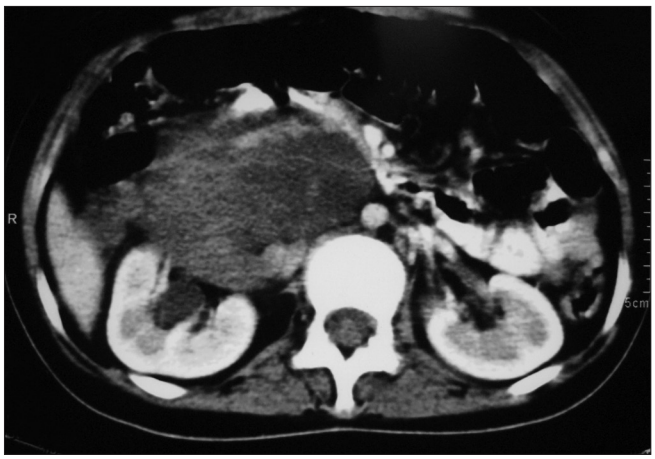

Figure 2: Contrast-enhanced CT scan of the abdomen showing a huge right-sided retroperitoneal mass in front of the right kidney

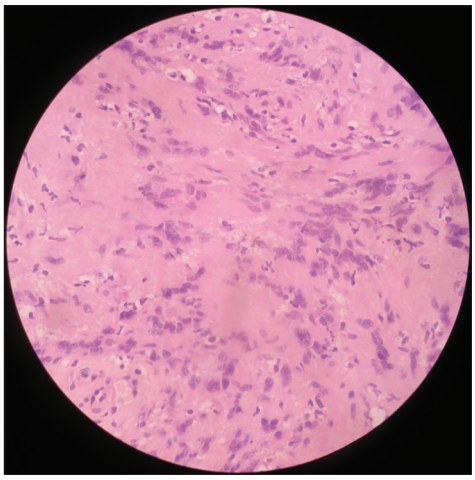

Figure 3: A histopathological picture of the retroperitoneal schwannoma

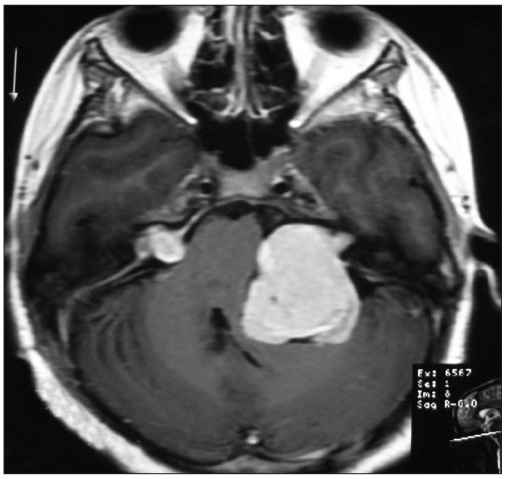

Figure 4: Contrast-enhanced MRI after the first operation showing a left-sided VS

were as follows: (1) Unilateral VSs + any two of the following: Meningioma, glioma, neurofibroma, schwannoma, and posterior sub-capsular opacities; or (2) multiple meningioma (two or more) + unilateral VS or any two of the following: Glioma, neurofibroma, schwannoma, and cataract.[5]

Approximately half of the patients seen are the first case in their families. There is increasing evidence that a small but significant proportion of these cases arise through somatic mutation of NF2 genes.[6] Our case is also the first case in her family. Patients with NF2 also develop other cranial, spinal, and peripheral tumours (meningiomas, gliomas, neuromas).[7]

The clinical presentation of NF2 varies, but approximately 45% of patients are first diagnosed because of symptoms resulting from Seventh Cranial Nerve (CN VIII) schwannomas, such as hearing loss, tinnitus, balance impairment, and weakness in CN VII distribution. This is because CN VIII schwannomas are symptomatic at a relatively small size.[8]

Evans *et al*. reported in a series that only 26 of 61 patients presented initially with features consistent with VSs (hearing loss, tinnitus, and facial palsy), whereas 19 presented with symptoms of a meningioma, seven with a spinal tumour, and fivewith a cutaneous tumour. Six children presented with a unilateral facial palsy. Facial paralysis is an uncommon presentation of VSs and because the initial diagnosis was as long as 15 years ago, it is possible that the palsy was the result of mononeuropathy in some cases. Two cases presented with a foot drop that did not fully recover. In one of these children the spinal magnetic resonance imaging (MRI) scan was normal. Twenty were diagnosed initially with an isolated non-VS feature of NF2, over half of whom had no previous family history of the disease.[5]

In NF2, different to sporadic schwannomas, cure from a specific tumour cannot be achieved, as even in radical resection, due to the nature of the disease, a recurrence can occur at any site of a preserved nerve and at much higher rate and speed. Especially young patients below 20 and even 30 years of age present with much faster growth patterns and a far more active tumour biology.[9]

According to Halliday *et al*., schwannomas are the most associated nerve sheath tumours with NF2.[10] As the condition has an insidious course and VSs may reach large dimensions before the onset of audiological, vestibular, and facial symptoms, early detection of NF2 is very important for the preservation of hearing and prevention of other neurological sequelae.[11]

Retroperitoneal schwannomas are expected to occur in only 3% of the cases of NF2.[12] 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. Retroperitoneal schwannomas predominate in women.[11] The tumour's origin is difficult to define, and a pre-operative diagnosis is achieved with difficulty due to lack of specific clinical symptoms or laboratory data.[13] Patrinou *et al*. reported a case of NF2 in which they found a large retroperitoneal schwannoma, in which MRI of the brain, orbits, cervical, thoracic, and lumbar revealed bilateral VS, multiple meningiomas, as well as multiple schwannomas and ependymomas in the cervical, thoracic, and lumbar spine.[14] Nevertheless, surgical removal of symptomatic cranial and spinal tumours is the mainstay of management.

Conclusion

Though this is a rare combination of tumours, our case suggests that VS in a child requires rapid and extensive investigation as a case of NF2 and thorough search for other tumour should be borne in mind. Early diagnosis, continuous follow-up, and unwearied approach of treatment, which may need multiple operations, should be accomplished to optimize care.

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Conflicts of interest
There are no conflicts of interest.

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