



Case Report

Neurofibromatosis Type 2 Presenting with Ptosis

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Abstract

Neurofibromatosis type 2 (NF 2), formerly called bilateral Acoustic Neurofibromatosis, is a distinct disease, which must be separated clinically and radiologically from Neurofibromatosis type 1. We presented a case of NF 2 of 20-year-old female presented with a rare symptom, right-sided ptosis due to superior rectus muscle paresis, multiple subcutaneous nodules and hearing impairment.

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Introduction

Type 2 neurofibromatosis (central or bilateral neurofibromatosis) is an autosomal dominant genetic disease characterized by development of bilateral vestibular Schwannoma in second and third decade of life. Neurofibromatosis (NF) is an inherited disorder characterized by the growth of tumors known as hamartomas. These tumors may affect the skin, eyes, and nervous system. Clinical signs of NF may be present in childhood or later in adulthood. Two variants of the syndrome have been identified: NF type 1 (NF1) and NF type 2 (NF2). NF1, also called von Recklinghausen NF or peripheral NF, is the common form of NF (frequency 1:4,000). NF2, also called bilateral acoustic neurofibromatosis (BAN) or central bilateral acoustic NF, is a rare type of NF (frequency 1:50,000)¹ NF2 has an autosomal dominant inheritance pattern of transmission. The gene responsible is located on chromosome 22. NF2 gene has been referred to as “neurofibromin 2” or “merlin” (the latter an acronym for moesin-ezrin-radixin-like protein); the gene product is a cytoskeletal protein although its precise function is unknown^{1,2}. The NF2 gene is highly penetrant, causing the development of acoustic neuromas in almost everyone (90%) carrying the gene. Approximately half the affected population reports the occurrence of other nervous system tumors such as meningioma, gliomas, vestibular schwannoma, ependymoma, and recognizing NF2-related skin tumors are important for early diagnosis, especially in pediatric patients^{3,4,5,6}. Ocular findings are mainly represented by juvenile posterior subcapsular lens opacity.¹

Case report

A 20 year girl was referred to us from an eye specialist with a complaint of right sided ptosis due to paresis of superior rectus muscle, headache with bilateral hearing impairment. The patient had a positive familiar history of NF (his father was suffering from cutaneous and subcutaneous neurofibromas); there is no significant past medical history. About for last three months the patient first developed left-eye superior rectus muscle paresis and some months later, the condition is gradually deteriorating. Best-corrected

visual acuity was 20/20 in both eyes. Pupillary reflexes to light and near vision were normal in both eyes. Intraocular pressure was normal bilaterally. Subcapsular posterior opacity of the lens was present in both eyes, and ophthalmoscopy was negative. The eye motility examination confirmed left-eye superior rectus paresis, with distorted palpebral levator muscle function (Fig. 1). Accentuated vertical diplopia appeared with manual lifting of the left upper lid. Bell’s phenomenon was absent. Visual-field examination revealed no alterations. Neurological examination revealed slight hypokinesia of the left facial nerve. Brain computed tomography (CT scan) and magnetic resonance imaging (MRI) showed bilateral acoustic neuromas and a left superior parietal meningioma. Multiple cutaneous neurofibromas are also found in different sites.

The patient also showed hearing impairment on the both side and increased latencies on audiometric testing.



Fig 1: Patient with left sided ptosis.

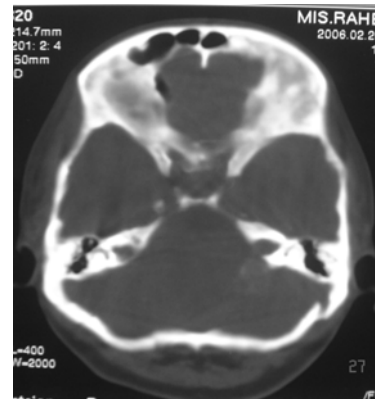


Fig 2: Bone window of CT scan- widening of bilateral internal acoustic meatus

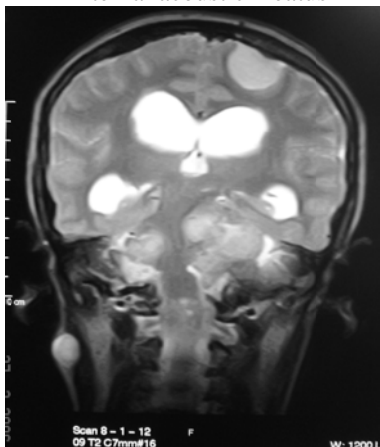


Fig 3: Coronal T2WI- Bilateral acoustic neuromas with meningioma and sub. neurofibroma

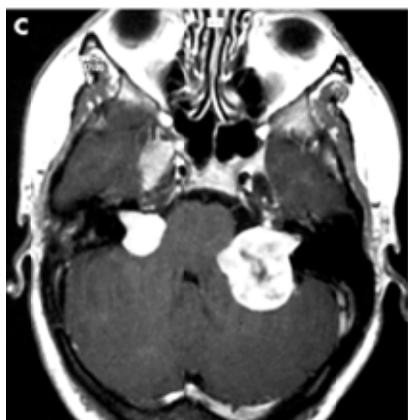


Fig 4: Contrast enhanced MRI showing bilateral acoustic neuroma

Discussion

Neurofibromatosis type 2 also known as bilateral acoustic neurofibromatosis or central neurofibromatosis is an autosomal dominant disorder occurring in about 1 in 50,000 individuals. Eighth nerve tumors are characteristic, but other intracranial and intraspinal tumors are common. The most characteristic ocular features of NF2 are cataract (69–81% of patients), retinal haematomas (8–22%), and motor abnormalities (12%). Other ocular abnormalities more rarely reported include optic disc gliomas, epiretinal membranes, conjunctival neurofibroma, retinal hemangioma, choroidal hamartomas, and optic

nerve sheath meningiomas. Ocular symptoms are the initial manifestation of the disease in a minority of cases, but the mean age of onset of ocular symptoms is younger compared with extraocular symptoms^{7,8,9}. Palpebral ptosis has rarely been reported in patients with NF2 compared with patients with NF1 (mechanical ptosis due to skin neurofibroma). The literature describes isolated monocular oculomotor palsy in five children. Ptosis was originally believed to be idiopathic but subsequently documented to be secondary to a presumed neuroma of the third nerve distal to the mesencephalon. In that report, no patient was described to be affected with NF¹⁰ Egan et al., in 2001, described four of 29 consecutive patients with NF2 who were referred for neuroophthalmic screening and in whom a monocular elevator paresis was discovered. The cause of ophthalmoparesis in two of their cases proved to be a tumor of the ipsilateral third nerve with radiographic features typical of a schwannoma; in the other two cases, third-nerve schwannomas too small to have reached threshold for radiologic detection can also explain the ophthalmoparesis¹² Also in the case here described, the ptosis associated with rectus superior palsy was caused by expansive processes in the temporal region close to the cavernous sinus (neural ptosis). Although third, fourth, and sixth nerve palsies and other forms of strabismus have been described in NF2, monocular elevator paresis had rarely been. Ocular motility should be monitored in patients with NF2; discovery of monocular elevator paresis may be the first manifestation of third-nerve palsy and may indicate the presence of a tumor of the third nerve.

References

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