Bilateral Acoustic Neurofibroma: A Further Case Report of the Rare Neurofibromatosis

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The neurofibromatosis consist of two distinct disorders, the genes for which have recently been located on separate chromosomes. Neurofibromatosis 1 [NF-1], also called von Recklinghausen's neurofibromatosis, is a chromosome 17 abnormality[1] and neurofibromatosis 2 [NF-2], is a chromosome 22 abnormality[2]. Bilateral acoustic neuromas are the hallmark of the genetically distinct NF-2 in which they practically always occur before the age of 21 and show a strong (autosomal dominant) heredity. This case is being presented for its rarity.

THE CASE

An 18 year old female presented with ringing in the ears (bilateral high pitched machinery like roaring sound) and vertigo of 1 year duration. She was treated by several physicians and later by psychiatrists for her symptoms without relief. Examination revealed an ill looking young girl with impaired mentation, bilateral deafness, absent corneal reflexes and bilateral ataxia. Detailed physical examination revealed two skin neurofibromas, one in the forearm and the other in the abdomen. There were no lens opacities or cafe-au-lait spots. There was no family history of neurofibromatosis. Her mother, brother and sister were found to be normal on thorough clinical examination. Her father died five years prior to her admission of unrelated illness (myocardial infarction).

Fundus examination showed bilateral papilloedema. CT scan showed bilateral large sized tumours occupying the cerebellopontine angle (Fig 1). Posterior fossa exploration and removal of left cerebellopontine angle tumour was attempted through a suboccipital craniotomy. The capsule of the tumour was adherent to the brain stem and was extremely vascular. Intratumoural bulk reduction was done and 50% tumour removal could be achieved in 8 hours. Profuse bleeding prevented complete removal and a decision to close and operate later was taken. The histopathological appearance is depicted in Fig 2. Relatives of the patient refused further operation and she was discharged against medical advice. She was lost to follow up.

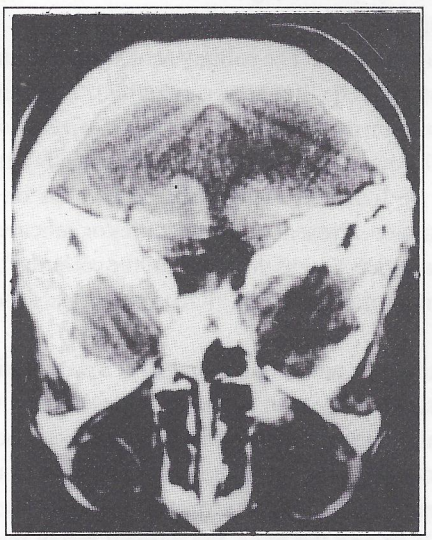

Figure 1: *CT Scan showing bilateral walnut-sized tumours occupying the cerebellopontine angle*

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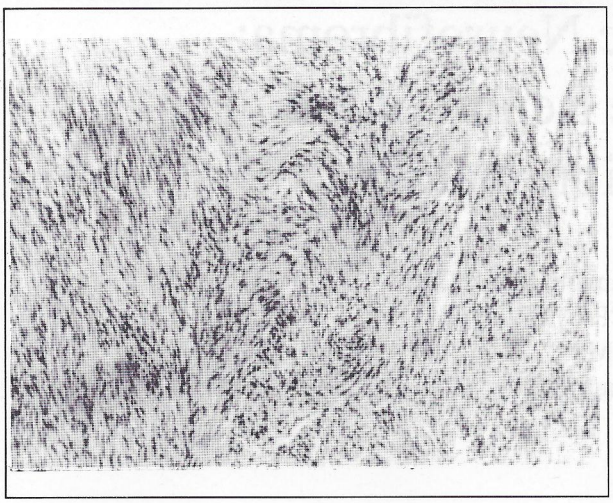

Figure 2: *Microphotograph showing spindle cells with wavy nucleii and myxoid stroma, suggestive of a neurofibroma*

DISCUSSION

Cases of bilateral acoustic neuromas were first reported more than 150 years ago[3]. In 1930, 38 members of one family were found to have acoustic neuromas[4]. Forty years later, the same family had more than 100 affected members[5], which indicates that a single mutant gene is responsible for the disorder which is distinct from the abnormality producing NF-1. It is also important to differentiate between NF-2 and the sporadic unilateral acoustic neuroma. The solitary acoustic neuromas tends to develop later in life, is not inherited, and raises lesser problems in management[6]. Cafe-au-lait-spots and skin neurofibromas, although less common in NF-2 than in NF-1, can be found in many affected persons on thorough examination. Presenile lens opacities or subcapsular cataracts have been found in about half the patients[7,8]. Multiple tumours of meningeal and glial origin may also be present[7].

Acoustic neuromas practically always originate on the vestibular division of the eighth nerve just within the internal auditory canal. It first compresses the cochlear nerve causing ringing in the ear (tinnitus) and ultimately deafness[7]. As the tumour grows into the angle between the cerebellum and the pons, the corneal reflex may be lost, signifying compression of afferent fibers of the trigeminal nerve. Later, other trigeminal motor and sensory functions may also be lost. The facial nerve of ipsilateral cerebellar hemisphere is next to be involved. The brain stem ultimately becomes compressed, causing cortico-spinal tract signs or the aque duct may be narrowed causing hydrocephalus and symptoms of increased intracranial pressure[7].

Audiological and vestibular evaluation include the tuning fork tests, pure tome and speech audiometry, auditory fatigue and recruitment tests, brainstem evoked responses and vestibular tests. Magnetic resonance imaging is more sensitive than contrast-enhanced computerised tomography in detecting small acoustic neuromas, especially intracranial ones[8]. Microsurgical suboccipital transmetal excision is the favoured approach. The translabyrinthine operation is the procedure of choice for removing acoustic neuromas in patients with normal hearing in the contralateral ear[9]. Patients with bilateral acoustic neuromas present a much more difficult set of treatment problems[6]. The treatment decision must be individualised and should be influenced by such factors as the patient's age, neurological condition, level of hearing in each ear, and tumour size and growth rate, as well as the social, psychological and occupational circumstances[6-8].

Antigenic activity of nerve growth factor and glial growth factor-like activity is increased in NF-2[8,10]. The isolation and cloning of the gene for NF-2 will have profound implications in the diagnosis and prevention of the disorder by genetic counselling and prenatal testing.

CONCLUSION

NF-2 is a rare disorder. Clinical features, diagnosis and treatment modalities are discussed.

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