An interesting case of neurofibromatosis-2

A 24-year-old lady presented with gradually progressive hearing loss and tinnitus.

Imaging Findings:

After detailed clinical evaluation, she was referred to us for MR imaging of brain and the inner ear (7th - 8th nerve complex). On MR imaging, she was found to have large infratentorial extra-axial masses in each of the cerebellopontine angle cistern (Fig. 1). These lesions demonstrated avid enhancement on post contrast MR scan and were extending into the internal auditory canal causing widening of the internal acoustic meatus (Fig. 2). These MRI findings of bilateral vestibular schwannomas were sufficient for a definite diagnosis of neurofibromatosis type 2 (NF-2), according to the National Institutes of Health, Manchester, or National Neurofibromatosis Foundation diagnostic criteria. These posterior fossa schwannomas were associated with multiple supratentorial extra-axial dural based masses suggestive of intracranial meningiomas (Fig. 3). She also had a well circumscribed homogeneously enhancing intracranial mass in the right orbit, which was encasing the optic nerve. On contrast enhanced MRI, the optic nerve appeared as a negative defect in relation to the surrounding thickened enhancing optic-nerve-sheath on either side representing the tram-track sign of optic nerve sheath meningioma (Fig. 4). She also had an intra-parotid well defined mass on the right side (Fig. 5) possibly representing a neurogenic tumour of the facial nerve (neurofibroma/ schwannoma). MR imaging of the spine failed to reveal any spinal tumour.

Discussion:

We present the case of a 24 year-old patient with Neurofibromatosis type 2 with bilateral acoustic schwannomas, multiple intracranial meningioma, optic nerve sheath (orbital) meningioma and a intraparotid neuroma.

Neurofibromatosis 2 (NF-2) is an inherited autosomal dominant neurocutaneous syndrome (phakomatosis). It is characterised by development of multiple central nervous system tumours such as multiple schwannomas, meningiomas, and ependymomas. The NF-2 gene is a tumour-suppressor gene which has been mapped to chromosome 22. Impaired function of this gene is thought to be responsible for the development of clinical symptoms. NF-2 is an autosomal dominant genetic trait i.e. it affects both genders equally and that each child of an affected parent has a 50% likelihood of inheriting the gene. Typically, these patients present in the second or third decade of life. According to the National Institutes of Health, Manchester, or National Neurofibromatosis Foundation diagnostic criteria of NF-2 include: [1] Bilateral acoustic schwannoma seen with appropriate imaging techniques (e.g. computed tomography or magnetic resonance imaging; or [2] A first-degree relative with NF2 and either a unilateral acoustic schwannoma or 2 of the following: neurofibroma/ meningioma/ glioma/ schwannoma/ juvenile
posterior subcapsular lenticular opacity. Acoustic or vestibular schwannomas are usually the most common cause of presentation. These tumours manifest with progressive hearing loss, tinnitus, headache or vestibular symptoms. In spine, majority of intramedullary spinal tumours are ependymomas and arise in either the upper cervical cord or the conus medullaris. Spinal meningiomas present as intradural extramedullary neoplasms most commonly involving the dorsal spine and often are multiple in number.

**Differential Diagnosis List:** Head & neck manifestations in a case of NF-2

**Final Diagnosis:** Head & neck manifestations in a case of NF-2

**References:**


Description: Axial post contrast MR image reveals bilateral acoustic schwannoma and intraconal tumour in the right orbit. Origin:
Description: Well circumscribed extra-axial masses are seen in bilateral cerebello-pontine cisterns. These show avid post contrast enhancement. Origin:
Description: Icecream cone appearance is seen bilaterally due to intracanalicular extension of the lesion. Origin:
Description: A well circumscribed homogeneously enhancing intraconal mass lesion is seen in the right orbit. Origin:
Description: The lesion seems to be encasing the optic nerve. Tram track appearance of the optic nerve sheath can be appreciated. Origin:
Description: Tram track sign is well visualised. The optic nerve appears as a negative defect in relation to the surrounding thickened and enhancing optic nerve sheath on either side. Origin:
Description: Dural based meningiomas are seen bilaterally. The larger one projects towards left side while the smaller one is located cranially and is projecting on to the right. Origin:
Description: Axial image shows well the parasagittal meningioma on the left side. Origin:
**Description:** A well circumscribed intensely enhancing mass lesion is seen in the posterior aspect of right parotid. It is extending into the stylomandibular tunnel. **Origin:**
Description: Coronal image confirms the homogeneously enhancing parotid mass. It mainly involves the deep lobe of right parotid gland. Origin: