

# Huge occipital myxomatous plexiform neurofibroma in the absence of neurofibromatosis

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## ABSTRACT

A case of huge occipital myxomatous plexiform neurofibroma is presented. The lesion slowly enlarged since childhood and was associated with suboccipital bone hypoplasia. At operation the lesion was clearly originating from the occipital nerve and was totally excised. The case represents a rare occurrence of occipital myxomatous plexiform neurofibroma and suboccipital bone hypoplasia in the absence of evidence of neurofibromatosis.

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Neurofibroma is the most common benign peripheral nerve neoplasm. They may occasionally occur as solitary nodules or more frequently as multiple skin lesions in neurofibromatosis (von Recklinghausen's disease), particularly the plexiform type. Most cases are of the benign type with occasional malignant changes. Occipital subcutaneous neurofibroma in the presence of suboccipital bone hypoplasia has been reported in association with neurofibromatosis. The literature has very limited reports on cases of occipital myxomatous plexiform neurofibroma associated with bone hypoplasia in the absence of evidence of neurofibromatosis.

**Case Report.** A 42-year-old male patient was referred to King Fahd Hospital of the University, Al-Khobar, Kingdom of Saudi Arabia as a case of occipital mass. The patient gave a history of a painless occipital mass since childhood that was very slowly progressing in size. There was no history of trauma, headache or seizure. He gave a history, from his parents, of cauterization on the occipital region just after birth, but could not recall the reason. He presented for medical attention for cosmetic reasons in preparation for his marriage. On examination, he was a fully conscious

and oriented person. Local examination showed a non-tender, non-pulsatile mass located at the left suboccipital region, extending from the level of the third cervical spine to above the level of the occipital protuberance by 6 cm. It is of a mixed consistency with parts fluctuant and parts of hard nodules. There was no bruit. General systemic examination failed to reveal any evidence of neurofibromatosis. Computed tomography (CT) scan of the head and upper cervical spine results showed large heterogeneous density lesion with intense enhancement extending from the occipital bone down to the C3 level. The lesion contained numerous low-density cystic areas. It is predominantly extracranial with small extension through the left jugular foramen. Bone window CT scan of the region revealed loss of bone continuity at the left side of the suboccipital bone and foramen magnum with no evidence of active bone erosion (**Figure 1a**). Magnetic resonance imaging (MRI) of the head and upper cervical spine revealed heterogeneous intensity mass of smooth outline with no local invasion. On T1 weighted images the mass is iso- to hypo-intense with numerous variable sized cystic hypointense regions (**Figure 1b**). Following gadolinium intake, the cystic regions did not enhance and became

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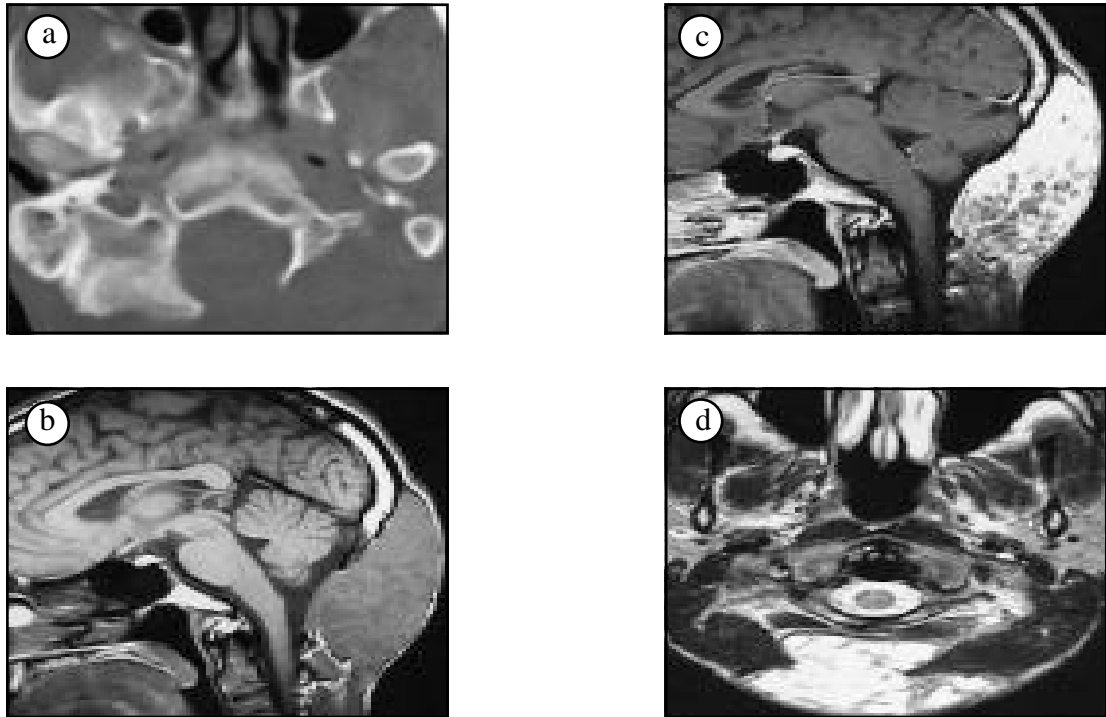


Figure 1 - Pre-operative radiological findings **a)** Bone window CT scan of the posterior fossa revealed loss of bone continuity at left suboccipital bone and foramen magnum with no evidence of active bone erosion. **b)** T1 weighted MRI, sagittal view; the mass is iso- to hypo-intense with numerous variable sized cystic hypointense regions. **c)** T2 weighted MRI, sagittal view; following Gadolinium intake, the cystic regions did not enhance and became more apparent compared to the rest of the mass background that enhanced intensely. **d)** T2 weighted MRI, axial view; the lesion is heterogeneously hyperintense with strands of hypointense character in between the cystic regions.



Figure 2 - Totally excised mass. Inset, the nerve of origin is seen blending into the tumor (arrow).

more apparent compared to the rest of the mass background that enhanced intensely (**Figure 1c**). On T2 weighted images the lesion is heterogeneously hyperintense with strands of hypointense character in between the cystic regions (**Figure 1d**). Selective cerebral angiography showed intense tumor blush on selective injection through the left occipital artery. Computed tomography scan and MRI of the brain did not show any evidence of intracranial neoplastic changes.

**Operative findings.** The tumor was found severely adherent to, but dissectable from, the subcutaneous tissue. The mass was highly vascular and easily bleeds on touch. A large branch from the left occipital artery supplying the mass was ligated and cut. A cutaneous nerve, at the site of the left occipital nerve, was seen blending into the mass. Distally it was running in the subcutaneous tissue and was of normal appearance. At the junction with the tumor, it disappeared into the lesion with no possibility of dissecting them out of each other (**Figure 2**). Two more cutaneous nerves were running superficial to the tumor, not involved in the lesion, and were dissected freely and preserved. Adhesion was seen between the mass and the dura in the lower end. The mass was excised completely in one piece. The lower suboccipital bone was seen to be hypoplastic confirming the preoperative image findings. The suboccipital muscles were inserting off the midline, indicating the chronicity of the lesion dating to childhood. On gross examination the mass consisted of fragment of grayish, slightly firm tissue, measuring 11 x 8 cm. Cutting through it showed areas of cystic spaces with yellowish discoloration. Microscopic examination showed proliferation of spindle cells with elongated nuclei, collagen bundles and increased vascularity in a myxomatous stroma. The tumor forms a prominent plexiform pattern focally.

**Discussion.** The scalp is a thick layer of primarily hair-bearing skin and underlying subcutaneous tissue and galea together with peripheral nerves and blood vessels.<sup>1</sup> Neoplastic growth may occur in any of the cell lines of the skin and associated tissues. Neurofibroma is the most common benign peripheral nerve neoplasm. They may occasionally occur as solitary nodules or more frequently as multiple skin lesions in neurofibromatosis (von Recklinghausen's disease), particularly the plexiform type.<sup>2</sup> Neurofibromatosis is inherited in an autosomal dominant pattern, although many cases are the result of spontaneous mutations. Findings of characteristic chromosomal aberrations have been reported in neurofibroma. These chromosomal imbalances are more commonly seen with neurofibromatosis-associated tumors than in sporadic neurofibromas. In the benign form of neurofibroma of both types, the number of losses is higher than the number of gains, suggesting a predominant role of tumor suppressor genes in tumorigenesis.<sup>3</sup> The presence of a café au lait spot, axillary large flat lightly pigmented lesion is considered pathognomonic for the inherited condition. The importance is the risk of occurrence of malignancies, especially intracranially, such as

schwannomas and glial tumors.<sup>2</sup> On T1-weighted MRI neurofibroma appears isointense or slightly hyperintense to muscle. A target pattern with peripheral hyperintense rim and central low intensity is recognized on T2-weighted images and enhanced T1-weighted images, which corresponds histologically to peripheral myxomatous changes and central fibrocollagenous tissue.<sup>4</sup> Histological study reveals wavy fiber bundles in a mucopolysaccharides-rich extracellular matrix.<sup>5</sup> Plexiform neurofibromas have a predominant intrafascicular (endoneurial) growth, resulting in the formation of a convoluted mass. The nerve components are dispersed by intervening spindle cells, wavy collagen bundles and increased intercellular mucinous substance. Cases of plexiform neurofibroma have been reported in the occipital region with occipital - suboccipital bone hypoplasia. These cases have been documented as neurofibromatosis.<sup>6-8</sup> The literature has very limited reports on cases of occipital myxomatous plexiform neurofibroma associated with bone hypoplasia in the absence of evidence of neurofibromatosis.<sup>8,9</sup> The presented case in this report had neither superficial manifestations of neurofibromatosis nor associated neoplastic formation. The patient had the occipital swelling since childhood. The suboccipital bone was found partially hypoplastic in the midline, and suboccipital muscles were attached to the bone off the midline.

In conclusion, occipital myxomatous plexiform neurofibroma associated with bone hypoplasia in the absence of evidence of neurofibromatosis is rarely reported. The presented case details such occurrence. The treatment is curable if total excision is feasible.

## References

1. Arnold PG, Meland NB. Tumours of the scalp. In: Youmans JR, editor. Neurological surgery. Philadelphia (PA): WB Saunders; 1999. p. 3676-3703.
2. Belzberg AJ, Campbell JN. Nerve tumours. In: Wilkins RH, Rengachary SS, editors. Neurosurgery. New York (NY): McGraw Hill Co; 1996. p. 3217-3223.
3. Koga T, Iwasaki H, Ishiguro M, Matsuzaki A, Kikuchi M. Losses in chromosomes 17, 19, and 22q in neurofibromatosis type 1 and sporadic neurofibromas: a comparative genomic hybridization analysis. *Cancer Genet Cytogenet* 2002; 136: 113-120.
4. Varma DG, Mouloupoulos A, Sara AS, Leeds N, Kumar R, Kim EE, et al. MR imaging of extracranial nerve sheath tumors. *J Comput Assist Tomogr* 1992; 16: 448-453.
5. Burger PC, Scheithauer BW, Vogel FS. Nerve sheath neoplasms. In: Burger PC, Scheithauer BW, Vogel FS, editor. Surgical pathology of the nervous system and its coverings. New York (NY): Churchill Livingstone; 1991. p. 676-700.
6. Duchateau J, Lejour M. Neurofibromatosis of the scalp associated with occipital bone aplasia. *Ann Chir Plast Esthet* 1986; 31: 73-78.
7. Nakasu S, Nakasu Y, Matsuda I, Handa J. Giant neurofibroma of the occipital scalp associated with lambda defect-case report. *No To Shinkei* 1981; 33: 181-185.
8. Garcia-Uria J, Sola RG, Carrillo R, Bravo G. Epicranial plexiform neurofibroma. *Surg Neurol* 1979; 11: 390-392.
9. Ohaegbulam SC. Congenital plexiform neurofibroma of the occipital scalp. Case report. *J Neurosurg* 1977; 46: 245-247.