Review Article

Congenital extra calvarial plexiform neurofibroma in occipito-cervical region with Occipital bone defect with neurofibromatosis type 1 and segmental neurofibromatosis: Case report and review of literature

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ABSTRACT

Plexiform neurofibroma (PNF) of the scalp is an extremely rare lesion reported in association with neurofibromatosis (NF). Occipital location of PNF is even more infrequent; we reported one pediatric case of PNF in occipito-cervical region with multiple small occipital bone defects and associated with NF-1.

Key words: Neurofibromatosis, plexiform neurofibroma, segmental neurofibromatosis

Introduction

Neurofibromatosis type 1 (NF-1) is an autosomal dominant disorder affecting 1 in 2500–3500 individuals.² It has a penetrance of almost 100%.³ The fact that it is a progressive disorder and that almost 50% of cases are sporadic mutations multiplies its clinical magnitude several folds.³ Kurimoto et al.³ discuss a report in which they described and classified seven different types of NF. Of these, NF-1 and NF-2 are distinct forms and the rest represent variants. Segmental neurofibromatosis (SNF) corresponds to the NF-5 of the original classification. It is a distinct relative of NF-1 but is 10%–20% rarer, accounting for approximately one case in 36,000–80,000 individuals.¹,³ We present a case; there was NF-1 with an extra calvarial plexiform neurofibroma (PNF) in occipito-cervical region with intracranial extension. A large portion of the visible swelling consisted of a neurofibroma and multiple café au lait spots on the left upper and lower trunk [Figure 1]. Swelling was excised completely.

Materials and Methods

This 5-year-old boy presented with history of a gradually progressive painless swelling in the occipito-cervical region since birth. On examination, he had a large (10 cm × 10 cm × 5 cm) occipito-cervical swelling, that was soft and nonfluctuant, and transillumination was negative. The swelling had a red telangiectatic hue, but no bruit was auscultated. The boy had no neurological deficits and no...
signs or family history of NF-1. In neuroimaging studies, the lesion was seen as an isointense on T1-weighted and heterogeneously hyperintense mass on the T2-weighted image and heterogeneous enhancement mass on postcontrast study [Figure 2]. There were multiple small underlying bone defect in the right occipital region [Figure 3]. The excision was total, and cerebrospinal fluid (CSF) leak was seen on bone defect site, so periosteum was buried on bone defect site for preventing the CSF leak. Postoperatively, there was no CSF leak. Histopathological findings in hematoxylin and eosin staining were showing focal myxoid generation and spindle cells, with serpentine nuclei and wispy cytoplasmic border suggestive of PNF.

**Discussion**

Previously, Scott[4] in 1906 Helmholtz and Cushing first reported PNF of the scalp. After that PNF of scalp reported by Ohaegbulam.[5] In general, PNFs are found in association with NF (in 26.7% of patients).[6] PNF is usually found along the course of a major nerve trunk, the ophthalmic division in the face.[7] It is an unencapsulated lesion and infiltrates the surrounding soft tissue to produce a fusiform appearance.[7] PNFs have been described very rarely in the literature. SNF is a mosaic form of NF.[1,3] It usually involves one large segment of the body and may involve both sides of the body either symmetrically or asymmetrically.[1] SNF has been very rarely described in the scalp.[1,3] The diagnosis of SNF is based entirely on clinical signs.[9] The patient in this report had a PNF with NF and with telangiectatic discoloration of the overlying swelling, with multiple café au lait spots on the left upper and lower trunk. Review of the literature on SNF revealed only a few recent reports.

Only nine reports were found in the literature, of these nine, one case had nonoperative management, and two had only a biopsy. In the initial description by Helmholtz and Cushing, as detailed by Scott,[4] the location of the PNF was at the forehead and temporal region. It was suggested then that there was a propensity for the forehead and the temporal region, especially the area of the scalp innervated by the trigeminal nerve. Although the forehead and the distribution along the course of the trigeminal nerve are the most common location, there have been a few reports of the PNF occurring at the occiput[2-10] detail given in Table 1; PNF occurs with slight male preponderance (male/female 7:5). Most reported cases have been congenital, but presented later in life due to apprehensions of a social or psychosocial nature, mostly previous reported cases, there was only thinning of the occipital bone with no true dysplasia.[4,9] One patient had multiple areas of bone loss.[10]

NF is generally considered to be a neurocutaneous disorder of neural crest origin with very little emphasis on osseous abnormalities, although osseous dysplasia is one of the seven criteria for diagnosing NF-1.[1] Most of the osseous lesions are thought to be secondary to the altered functioning of the NF-1 gene. Besides true dysplasias, the secondary involvement of the bones may be due to compression of the malignant tumors seen in association with this disorder, such as malignant peripheral nerve sheath tumors and rhabdomyosarcomas. Osseous manifestations in NF-1 are relatively common and occur in up to 50% of patients with NF-1.[2,4,6] Cranial osteolysis in NF-1 is very rare.[2,3,5,7,9,10] There are only a few cases reported, and in most, it is difficult to ascertain whether the osteolysis was a result of the erosion secondary to the giant tumor or was merely associated as part of the disease syndrome. The indications for surgery are pain, cosmetic disfigurement, and neurological involvement.[11]
Surgery for PNF has been described as being difficult because these tumors are usually very vascular, and torrential hemorrhage has been reported.[1,2,9] Several authors have advocated only biopsy of the lesion. Tumor excision in our case was total. There have been earlier reports of tumor firmly adhering to a dominant transverse sinus precluding complete excision.[5,9] Even in patients in whom there was no involvement of the sinuses, some authors have avoided reconstruction due to the theoretical risks of CSF fistulas, the risk of malignancies in NF-1 is approximately 2.7% more than in the general population.[1] There is approximately a 10% chance of malignant transformation reported in an existing case of NF.[2]

Conclusions

PNF associated with NF and SNF is extremely rare. Early surgical management should be the treatment of choice, not only just for cosmetic considerations but also for the theoretical risk of malignant transformation. However, the occurrence of osteolysis may also be prevented if it is indeed secondary to the pressure effects of the lesion, which cannot be disproven conclusively. The outcome is generally good.

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Conflicts of interest
There are no conflicts of interest.

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