Case Report

www.ijrhs.com
ISSN (o):2321–7251

Plexiform neurofibroma of the scalp a rare entity – a case report and review

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Submission Date: 23-04-2014, Acceptance Date: 25-04-2014, Publication Date: 30-04-2014

How to cite this article:

Vancouver/ICMJE Style

Pukar MM, Gadhavi RH, Mewada SG. Plexiform neurofibroma of the scalp a rare entity – a case report and review. Int J Res Health Sci [Internet]. 2014 Apr 30;2(2):711-4. Available from http://www.ijrhs.com/issues.php?val=Volume2&iss=Issue2

Harvard style

Pukar, M.M., Gadhavi, R.H., Mewada, S.G. Plexiform neurofibroma of the scalp a rare entity – a case report and review. *Int J Res Health Sci*. [Online] 2(2). p.711-4 Available from: http://www.ijrhs.com/issues.php?val=Volume2&iss=Issue2

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Abstract

Neurofibromas (NF) are benign tumors of neural origin, of which roughly 90% appear as solitary lesions. They are classified into cutaneous, subcutaneous, and plexiform subtypes. Plexiform neurofibromas are the least common variant and usually are pathognomonic for NF I. Here, we present a case of isolated plexiform neurofibroma with a painless enlarging mass of the skull involving temporal, parital and occipital region in a 16 year old boy. The present case is a diffuse isolated plexiform neurofibroma of the skull. Neurofibromatosis (NF) is a genetically inherited, autosomal dominant disease, characterized by multiple cafe au lait spots, cutaneous neurofibromas and "Lisch nodules." Neurofibromatosis can develop from a neural source at any age. These entities consist of multiple, twisted masses that grow along the axis of a large nerve, infiltrating and separating normal nerve fascicles. Malignant transformation is the main associated complication. Organisation (WHO) has subdivided neurofibromas into two categories, dermal and plexiform. Dermal neurofibroma arises from single peripheral nerve while plexiform neurofibroma is associated with a multiple nerve bundle.

Key words: Plexiform neurofibromas; Lisch nodules

Introduction

Plexiform neurofibroma is the term applied to a diffuse neurofibromatosis of nerve trunks, which is often associated with an overgrowth of the skin and subcutaneous tissues. It is a distinct type of neurofibroma that expands a nerve into a large tortuous mass of fibers that has a “bag of worms” Appearance [1]. Neurofibromatosis is classified into cutaneous, subcutaneous, and plexiform subtypes. Plexiform neurofibromas are the least common variant and usually are pathognomonic for NF I. The most commonly involved sites are the temple, upper lid, back of the neck, and the tongue. This systemic process, originally described by von Recklinghausen,

is an autosomal dominant disorder caused by a defect on chromosome 17. Patients usually present in childhood with numerous cutaneous or subcutaneous neurofibromas, melanotic lesions in the iris called Lisch nodules, café au lait spots, and optic gliomas.[2] Plexiform neurofibromas occur in only 17% of cases of NF I. When they occur in the setting of NF I, 91% are solitary lesions, and most of the lesions occur in the trunk, extremities, and head neck region [2]. These lesions are often locally invasive and may undergo malignant transformation. The estimated prevalence of transformation to a malignant peripheral nerve sheath tumour (MPNST) is approximately 5%. The incidence of NF1 has been estimated to be between 1 and 3000–4000 [3] and affects male and female subjects equally in all races. Plexiform neurofibromas (PNFs) rarely grow to be larger than 5 cm; however, neurofibromas can undergo continuous enlargement and eventually become giant lesions. Furthermore, PNFs have a potential for transformation into highly malignant peripheral nerve sheath tumors, which occur in approximately 5% of patients [4]. PNFs are difficult to manage surgically as they are extensively infiltrative, highly vascularized and tend to recur. Surgical treatment must be decided judiciously and individualized for each patient. Major complications of neurofibromas include malignant differentiation [5] and potentially life-threatening hemorrhage fortunately, these are quite rare. On the other hand, minor complications such as local infections or wound problems are very common.

An incision was made along the border of lesion. Dissection through the superficial tissues necessitated some dissection through tumor tissue, which was highly vascular with large friable vessels. Once deep to the lesion, it was possible to encompass it and dissect it off the deep structures through a normal fatty plane, tying off the main feeding vessels. The huge skull mass was removed completely. Good hemostasis achived and closure of the skin done. Although some features of NF1, such as café-au-lait spots and Lisch nodules, are clinically silent, neurofibromas cause a significant degree of morbidity, mortality, and cosmetic disfigurement. Childhood through early adulthood is a vulnerable period for the growth of these lesions. Of all the manifestations of neurofibromatosis, plexiform neurofibroma is one of the most difficult to treat. Genitourinary neurofibroma is rare with fewer than 40 pediatric cases of genitourinary neurofibromatosis reported in the literature, most of them associated with generalised neurofibromatosis, with boys being affected nearly twice as often as girls.

Neurofibromas have extensive vascularity and tend to bleed during surgery. Therefore, excessive bleeding should be kept in mind while attempting surgical removal.

Case report

A 16-year-old, male presented with a painless swelling over skull involving temporal, parietal, and occipital region/ Mass is hanging over left ear. Physical examination showed a firm non-tender, swelling over the skull which is nodular. Occipital lesions are globular. Overlying skin is smooth and normal swelling around 15 x 14 cm on temporoparietal region which is hanging over ;eft ear. Occipital region there are two masses globular in shape of the size of 8 cm in diameter.There were no palpable lymph nodes. Excision of swelling done good hemostasis achieved. Suturing was done with ethilon. Post operatively patient was uneventful and he was discharged from the hospital on eighth day. The operative site healed by primary intention. Follow up for 12 months was uneventful without any complication and recurrence. Microscopic examination – Microscopy revealed, hypertrophic nerves that considered mainly of spindle-shaped fibroblast. The nerves were surrounded by a myxoid stroma that extended into the papillary dermis. The findings were compatible with the diagnosis of Plexiform neurofibroma with no malignant transformation. Immunohistochemistry for S -100 protein was positive.

Discussion

Neurofibromas are benign tumors of neural origin, of which roughly 90% appear as solitary lesions. Solitary plexiform neurofibromas arising outside the context of NF I, as illustrated in the patient presented here, are quite rare, with only scattered cases reported in the literature till date. Although, the disease became widely recognized as a pathological in the late 19th century: it was only recently, that its two subsets have been defined [6]. Plexiform neurofibroma is a specific variant of neurofibroma exhibiting a bizarre histopathologic picture. It is poorly circumscribed, locally invasive and leads to great deal of deformity.

In the literature, there are only few reports of macroglossia caused by plexiform neurofibroma, and the cases are almost always associated with neurofibromatosis.[7] Hence, isolated plexiform neurofibroma is even rarer. We could not find any other signs or symptoms other than isolated

plexiform neurofibroma of the skull to make a diagnosis of neurofibromatosis, and thus it was concluded to be a case of isolated plexiform neurofibroma of the skull. When these ill-defined tumors grow in the head and neck, they are commonly symptomatic and disfiguring, causing upper airway obstruction, swallowing or mastication difficulties, or cosmetic distortion of the face. Plexiform neurofibromas mostly appear within first 2 years of life. There are two types of plexiform neurofibromas, nodular and diffuse. Diffuse plexiform neurofibroma, is also known as elephantiasis neurofibromatosa, which shows overgrowth of epidermal and subcutaneous tissue along with a wrinkled and pendulous appearance. The major cause of mortality in NF1 patients is transformation of plexiform neurofibromas into MPNSTs [8]. The five-year survival for MPNSTs is 16% for NF1 patients compared with 53% for non-NF1 patients. Ultrasound, CT, and MRI can be useful in the noninvasive diagnosis and characterization of nerve sheath tumours. On ultrasound, most peripheral nerve sheath tumours (PNSTs) are hypoechoic with posterior acoustic enhancement, sometimes mimicking cystic lesions; however, peripheral nerve continuity is diagnostic. The target appearance may be seen with a hyperechoic center and hypoechoic periphery, corresponding to a fibrocollagenous region centrally and a myxomatous region peripherally. Sonography, however, cannot reliably distinguish benign from malignant lesions. MRI is the most useful imaging modality to characterize tumour extent and suggest neurogenic origin [6] due to its high contrast resolution and multiplanar capabilities. Clinical management for the PNF requires a multidisciplinary approach. However, current treatment options for PNF are limited to surgical intervention. Resection is performed when the tumor is severely disfiguring or severely compromises functionality. Complete resection is often difficult because of the extensive and infiltrative nature of these lesions. It is postulated that these hemorrhages are caused by rupture of friable vasculature secondary to arterial dysplasia or vascular invasion by the neurofibroma. Therefore, the most immediate challenge for surgical management is hemostasis, Diathermy is of limited use as the tissue is very friable. A number of authors have reported significant blood loss during surgery requiring high volume transfusion. Pre-operative coil embolization was used to reduce the blood flow to the tumor and to limit intraoperative blood loss. In our case we had done excision of the Plexiform neurofibroma without pre operative embolisation and we had less blood loss. Visceral (extracutaneous) involvement is seen in less than 1% of patients with neurofibromatosis. The gastrointestinal tract and the urinary bladder are the most frequently cited locations of visceral involvement. However, rare reports exist describing neurofibromas, either solitary or diffuse, involving various sites in the female genital tract, the vulva, being the most common. Diffuse plexiform neurofibroma involving the genitourinary system (GUS) is uncommon. Pessin and Bodian theorized that the plexiform neurofibroma involving the bladder is derived from the vesicoprostatic plexus in males and the urethrovaginal plexus in females.The differential diagnosis of genitourinary neurofibroma includes teratoma, meningocele, and benign vascular tumours (e.g. lymphagiomas). Typically, sacro-coccygeal teratoma are seen as a visible mass at birth, making the diagnosis obvious. Lesion with a large intrapelvic component may cause urinary obstruction. Teratoma of vulva and perianal teratoma have been reported. The prognosis is generally good in patients with von Recklinghausen disease, but the course is occasionally complicated by pulmonary hypertension. All the four patients reported by Stewart et al. died of either respiratory failure or presumed cardiac failure. Pulmonary hypertension is therefore a major determinant of the morbidity and mortality in patients with von Recklinghausen disease. Other benign oral peripheral nerve tumours include schwannoma, nerve sheath myxoma, mucosa neuroma, palisade encapsulated neuroma, traumatic neuroma and granular cell tumor.

According to the available literature , among the 66 neurofibromas in the facial region, the following distribution has been found: tongue – 12; palate - 12; mandibular ridge/vestibule-15; maxillary ridge/vestibule-9; buccal mucosa -10; lip-4; mandibular intrabony – 2; gingiva-1 [9]. Thus, the tongue is the most common intra–oral site and the occurrence of neurofibroma on the gingiva as seen in this case is rare. Prognosis is excellent and recurrence is rare.

Conclusion

We report a Plexiform neurofibroma of the scalp which is very rare condition. And with the best of our knowledge only one case of Plexiform neurofibroma of scalp has been reported in literature in 2013. This is the second case which we are reporting.

Source of Funding: Nil

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Figure 1

Competing interest

The authors declare that they have no competing interests.

Figure 2:

Figure 3:

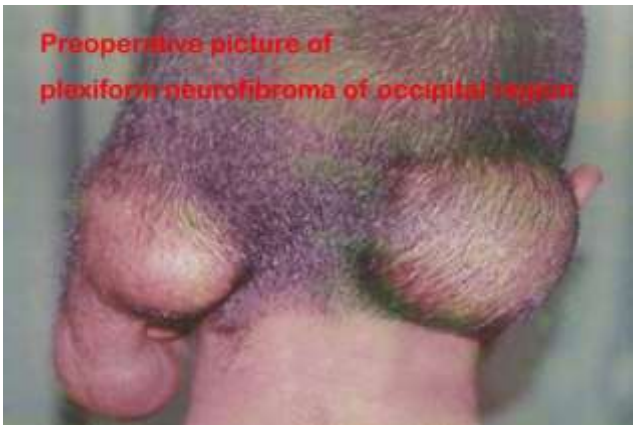

Figure 4:

Acknowledgment

Authors acknowledge the immense help received from the scholars whose articles are cited and included in references of this manuscript. The authors are also grateful to authors/editors/publishers of all those articles, journals and books from where the literature for this article has been reviewed and discussed.