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LARGE PLEXIFORM NEUROFIBROMA OF THE SACRO-GLUTEAL REGION – A RARE CASE REPORT

Plastic Surgery

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ABSTRACT

Plexiform neurofibroma is a rare benign nerve sheath tumor that develops in the perineurium and is often considered pathognomonic of neurofibromatosis type 1 (NF1 or von Recklinghausen disease). They occur most frequently in the craniomaxillofacial region, rarely on back and extremities. These lesions are highly vascular and there is 15-20% potential for malignant transformation. Here, we present a 26-year-old female with neurofibromatosis all over her body, but with a large plexiform neurofibroma in the sacral region which was causing difficulty in sitting and lying supine as well as disfigurement of the gluteal region. Surgical excision with primary closure of the swelling was done. Histopathology findings were consistent with neurofibromatosis.

KEYWORDS

Neurofibroma, Plexiform, Rare, Sacral, Surgical Excision

INTRODUCTION

Neurofibromatosis was first described as a multisystem disease by Von Recklinghausen in 1882 as an autosomal dominant inheritance affecting the skin, nervous system, bones, eyes and other organs. [1,2] There are three main types of neurofibromas: localized (most common), diffuse, and plexiform. Neurofibromatosis type 1 presents with cutaneous manifestations such as café-au-lait spots, multiple neurofibromas, ephelides in the skin fold areas, and hamartomatous lesions in the eyes, bones, glands and the central nervous system. [1,3] Plexiform neurofibroma (PN) is an uncommon variant of NF-1 where neurofibromas arise from multiple nerves that may also engage connective tissue and skin folds and are clinically described as "bags of worms".[4] The term plexiform does not imply involvement of a nerve plexus, such as the lumbar or brachial plexus, but a network-like growth of neurofibroma involving multiple fascicles of a nerve and may include multiple branches of a large nerve; leading to a diffuse mass of thickened nerves.5 The majority of neurofibromas occur sporadically and have an extremely low risk of malignant potential, the plexiform type is pathognomonic for NF 1 and carries an increased risk of malignant transformation to neurofibrosarcoma or malignant peripheral nerve sheath tumours.[6-9]. Surgery is the main stay for treatment. And is usually done in case of functional compromise, aesthetic deformity or pain.[10]

Case Report

26 year old female presented to us with multiple swellings all over the body with a large swelling in the sacral and gluteal regions since 5 years. These swellings started spontaneously and gradually progressed to the present size. Her primary complaint was the sacral swelling which was interfering with her sitting and lying supine. There was no history of pain, fever, bleeding or ulceration of the swelling. She had no history of any co-morbid illnesses. On examination, she had multiple neurofibromas all over her body with a swelling in the sacral and right gluteal region measuring 25 x 18 x 5cm, non-tender, soft in consistency, not fixed to the underlying structures with the skin not being pinchable. **(Fig. 1)** She also had multiple café-au-lait spots and Lisch nodules in the iris. A clinical diagnosis of Neurofibromatosis type 1 with plexiform neurofibroma of the sacral region was made. We planned for surgical resection of the plexiform neurofibroma. Under general anesthesia and in prone position, incision was made around the base of the lesion and excised in toto and sent for histopathology. **(Fig. 2,3)** The defect was closed primarily in layers with 2-0 polyglactin and 2-0 nylon sutures over a closed suction drain. The post-operative period was uneventful. **(Fig. 4)** Sutures were removed on the 14th post-operative day. Histopathological examination confirmed neurofibroma with no evidence of any malignancy.

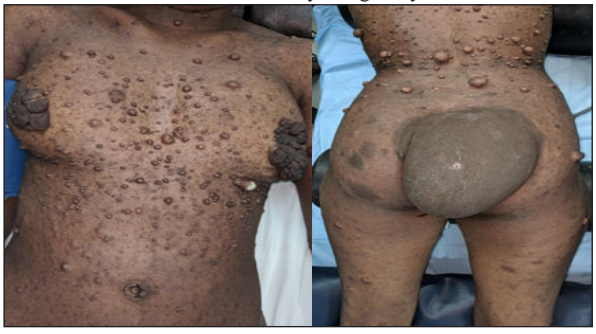

Fig. 1 – Clinical Photograph Showing Multiple Neurofibromas With Plexiform Lesion In The Sacral Region

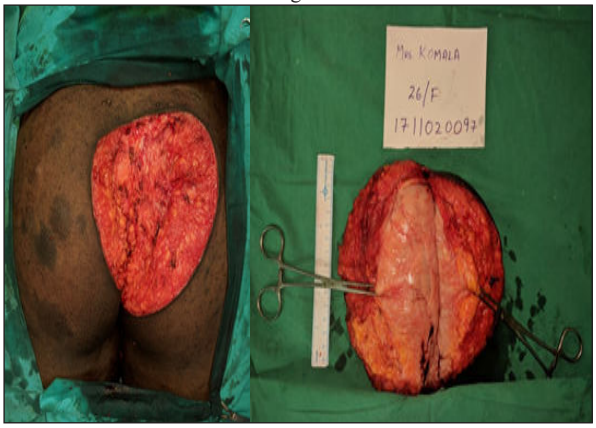

Fig. 2 – Defect After Surgical Excision

Fig. 3 – Specimen After Excision

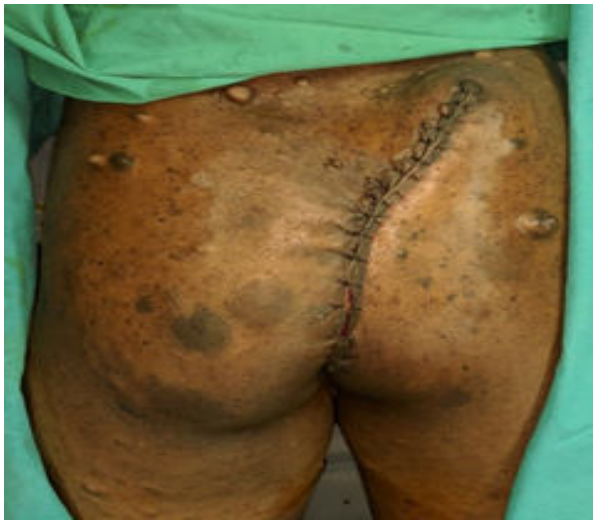

Fig. 4 – Photograph After Primary Closure

DISCUSSION

A neurofibroma is a benign nerve sheath tumor consisting of fibroblasts, Schwann cells, and neural elements that expand and diffusely infiltrate the nerve. Neurofibromas have been subdivided into two broad categories which are dermal and plexiform. Dermal neurofibromas are associated with a single peripheral nerve, while PNs are associated with multiple nerve bundles forming interdigitating network of finger-like projections of tumor.[11] The National Institutes of Health diagnostics criteria for NF1 requires two or more of the following clinical features must be present: six or more café-au-lait macules (>5 mm in greatest diameter in prepubertal individuals or <15mm in post-pubertal individuals), two or more neurofibromas of any type or one plexiform neurofibroma, freckling in the axillary or ingiunal regions, optic glioma, two or more iris hamartomas (Lisch nodules), distinctive bony lesion such as sphenoid dysplasia, or thinning of the long bone cortex with or without pseudoarthrosis and a first-degree relative (parent, sibling or offspring) with NF 1 based on the above criteria.[12] The hallmark of NF1 is the hyperpigmented cutaneous lesions. Neurofibromatosis appears in two different neurocutaneous autosomal dominant genetic forms, types 1 and 2 affecting the nerve cell tissue development and growth.[13] PNs occur in 26.7% of patients with NF1, usually present at birth as an isolated lesion, but often appear between the ages of 2 and 5.[14,15] It is frequently located on the head and neck due to the rich innervations of the area but have also been described in the extremities, where they follow the axis of a nerve track, appearing in a serpentine form.[16,17] Some authors describe as 'giant' for tumors weighing more than 20 kg, even though there is no clear consensus.[18] MRI and CT are used to determine the site and the extent of the plexiform neurofibroma but these are not reliable in differentiating between a PN and those that have progressed into malignant peripheral nerve sheath tumors.[19] Therefore, surgery is not only a therapeutic but also a diagnostic tool. PN is usually a clinical diagnosis, however, the gold standard to identify malignant change is histology. Pain, aesthetic issues and neurological involvements are known potential complications of plexiform neurofibromatosis over the certain areas of body. Malignant transformation is the leading complication of plexiform neurofibroma. The features which favour malignant change are an enlarging mass, size greater than 5 cm, ill-defined margins, perilesional edema, peripheral enhancement pattern, lack of central hypointense 'target' on T2 sequences, and heterogeneity with central necrosis.[20] Surgical resection is the mainstay of treatment, aiming at improving functional and aesthetic effect.[21-23] These tumours are very vascular making excision difficult and if the nerve roots of neurofibromas are from major nerves, surgical resection may result in functional impairment. Needle et al, had the largest series of surgically managed PN (10 year of follow up) found 54% recurrence rate especially in the head and neck region.[24] In our patient with the large plexiform neurofibroma of the lower back, resection of the lesion was done and the defect was primarily closed. Postoperative was uneventful and the patient had excellent recovery with no recurrence after one year of follow up.

CONCLUSION

Neurofibromatosis type-1 is a disease with the involvement of different organs and systems, which can be both limited and diffuse. The diagnosis is almost always clinical confirmed with histology. Imaging tools helps to identify extent of disease. Surgical excision is the mainstay of treatment with regular periodic follow-up as there is a chance for malignant transformation.

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