Case Report

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Large Thoraco-Lumbar Mass of Solitary Plexiform Neurofibroma: In A Patient of Neurofibromatosis Type - 1

Sunil Vitthalrao Jagtap*, Heena P. Shah, Swati Sunil Jagtap***, Atul Beniwal**, Vinay Raje****, Dheeraj Kumar Shukla****

*Professor, **Assistnat Lecturer, Department of Pathology, ***Associate Professor, Department of Physiology, ****Professor, Department of Neurosurgery, Krishna Institute of Medical Sciences, University Karad-415110, India.

Abstract

Plexiform neurofibroma are rare benign tumors of the peripheral nerves and usually considered pathognomic of neurofibromatosis Type -1. Plexiform neurofibromas are a more aggressive subtype of neurofibroma and many times shows infiltration of surrounding tissue making it difficult to remove. We present the case of 16 years old male child having history of gradually enlarging mass over back -D9-L3, since last 10 years associated with mild low back pain, skin pigmentation and history of blurred vision. On examination he was found to be case of Neurofibromatosis Type -1. Mass was resected and histopathologically diagnosed as plexiform neurofibroma-nodular type. We are presenting this case for its clinical, radiological and histopathological features.

Keywords: Neurofibroma; Nerve Tumor; Neurofibromatosis; Soft Tissue Tumors Back.

Introduction

Neurofibromas are benign peripheral nerve tumors that present as cutaneous, subcutaneous, nodular and diffuse plexiform lesions. The Plexiform neurofibromas are described to be highly linked to Neurofibromatosis Type - 1. Plexiform neurofibroma originates from a proliferation of nerve sheath cells, extending across the length of the nerve, often involving multiple nerves fascicles and presents as mass of thickened irregular nerve branches [1].

Case Report

A 16 year old male patient presented to our institute in department of neurosurgery having history of back pain and swelling in back for last 10 years. The mass was situated at lower thoracic spinal to mid lumbar spinal area which was soft-firm irregular diffuse and nodular measuring 16x3x2 cm. History of back pain having mild to moderate severity and gradually increasing was given. Complaint of blurring of vision since 6 months was there. The skin over lesion showed discoloration macules of light brown colour, circumscribed, larger measuring 19 mm in diameter. Kyphosis was present. Neurological examination was normal. No history of trauma at site. Family history was present. No additional neurofibromas were present. Eye examination showed Lisch nodules. On radiological evaluation-CT/MRI showed ill-defined peripherally enhancing multiple round lesions with central non enhancing foci giving target appearance involving posterior spinal subcutaneous soft tissue extending from lower D9 to lower L3 extending within the interspinous soft tissue with malformed spinal processes, lesion suggestive of Plexiform neurofibroma.

Patient Underwent Wide Resection of Mass

Gross

We received single large mass measuring 16x3x2 cm which was elongated, fusiform with small nodular areas (Figure 1). Cut section showed grey white, glistening surface with nodular areas (Figure 2).

On Microscopy

Multiple sections showed proliferation of Schwann

Corresponding Author: Sunil Vitthalrao Jagtap, Professor of Pathology, Krishna Institute of Medical Sciences, University, Karad (Dist.Satara) 415539, Maharashtra, India.
E-mail: drsvjagtap@gmail.com

cells, nerves, fibroblasts and perineural cells. The neoplastic spindle cells were arranged in bundles, clusters, sheets, fascicles and plexiform pattern (Figure 3). In areas myxoid degeneration is noted (Figure 4). There was no evidence of any malignant transformation on multiple section study. On histopathology diagnosis was given as Solitary Plexiform Neurofirboma – mass at back, thoraco-lumbar region.

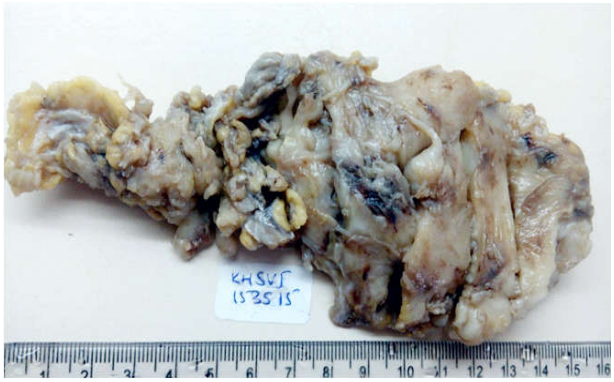

Fig. 1: Showing single large mass measuring 16x3x2 cm which is elongated, fusiform with small nodular areas

Fig. 2: Cut section showing grey white, glistening surface with nodular areas

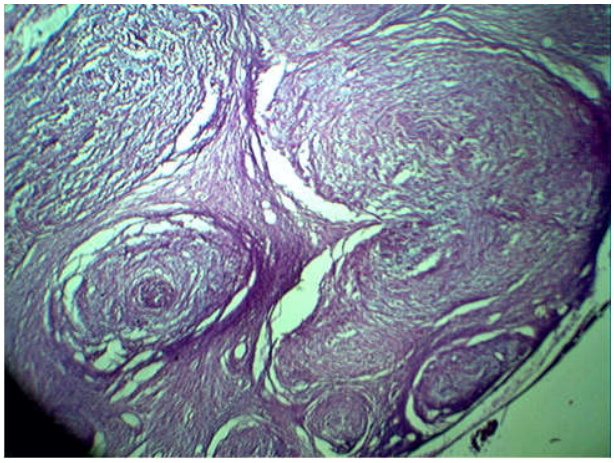

Fig. 3: Photomicrograph showing neoplastic spindle cells arranged in bundles, clusters, sheets, fascicles and plexiform pattern (H&E stain, 100x)

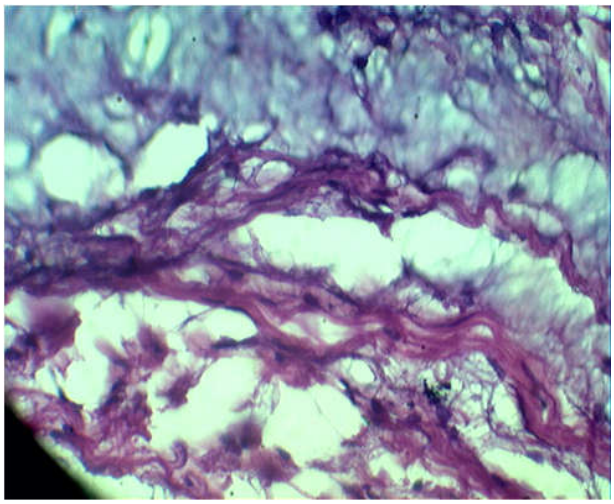

Fig. 4: Photomicrograph showing neurofibroma with areas myxoid degeneration (H&E stain, 400x)

Discussion

Neurofibromatosis Type -1 is an autosomal dominant disorder affecting the RAS proto-oncogene, resulting in growth of neuron and fibrotic tissues [2]. Neurofibromatosis Type-1 also known as Von Recklinghausen's disease has an incidence of 1 in 2600-3000 individuals. It has equal men and women distribution and in one half of the patient's family history is positive for NF-1 [3]. The National Institute of Health (NIH) [4] has given seven point diagnostic criteria of patients with Neurofibromatosis Type -1. When two or more of these listed below are present the diagnosis of Neurofibromatosis Type -1 is established.

1. Six or more café au lait maclues (>0.5cm in children or >1.5 cm in adults)
2. Two or more cutaneous/subcutaneous neurofibromas or one plexiform neurofibroma
3. Axillary or groin freckling
4. Optic pathway glioma
5. Two or more Lisch nodules (iris hamartomas seen on slit lamp examination)
6. Bony dysplasia (sphenoid wing dysplasia, bowing of long bone/pseudarthrosis)
7. First degree relative with Neurofibromatosis Type -1

Our patient had one plexiform neurofibroma, multiple café-au-lait macules, Lisch nodules in eye and history of first degree relative-father with Neurofibromatosis Type -1.

Various subtypes of neurofibromas have been described based on histology and locations. These include the cutaneous, sub-cutaneous, nodular and diffuse plexiform variant [5]. In our case it is plexiform neurofibroma of solitary type.

Plexiform neurofibromas can be seen in either type of neurofibromatosis. Approximately 20-40% of individuals with Neurofibromatosis Type -1 develop plexiform neurofibromas [6]. They are slow growing tumors, diffuse, large, elongated fibromas which may be present at birth or develop later in life. On histopathology they are similar to discrete neurofibromas. They arise in various regions of the body including face, neck, trunk, limbs, abdomen, pelvis [6]. Few cases show massive enlargement of the limbs (elephantiasis neurofibromatosa) [1].

Plexiform neurofibromas on microscopic examination show variety of elements including Schwann cells, collagen, fibroblasts, neuronal and vascular proliferation. Rarely melanin pigmentation is noted. The differential diagnosis of Schwannoma should be carefully evaluated in these lesions.

The plexiform neurofibroma is especially of concern as it has 5 to 20% risk for malignant transformation into malignant peripheral nerve sheath tumors [2,7]. We have taken multiple tissue section from mass, but there was no evidence of any malignant transformation.

The primary treatment continues to be complete surgical resection and this can be challenging as the lesion may involve multiple nerve fascicles, may be of large size, have irregular serpiginous growth, infiltrate to surrounding structures, and have significant vascularity [8]. Indications for resections of plexiform neurofibromas are severe pain, neurological deficit, suspected malignant transformation, cosmoses and progressive enlargement with compressive effects [9]. No specific treatment for plexiform neurofibromas currently exists, aside surgical resection. Various trials on chemotherapeutic agents, antibiotic agents, etc have shown to have variable results.

Our patient received treatment of complete surgical resection and is on regular follow up. Post operative period was uneventful.

Conclusion

As plexiform neurofibromas have the major cause of morbidity, disfigurement, recurrence and risk of malignancy, proper care of the patient is required for better survival.

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