Case Report

Plexiform Neurofibromatosis with Abnormal Pachydermatocele: A Rare Case Report

Agarwal AK[1], Gupta Anil Kumar[2], Kumar Dileep[3], Kumar Vijay[4], Yadav Ganesh[5]

Abstract

Plexiform neurofibromatosis, also known as pachydermatocele, is a rare type of neurofibromatosis, caused by excessive growth of the neural tissue in the subcutaneous fat. This condition is rare in Indian subcontinent but when it occurs, it causes cosmetic as well as functional disability. The optimal management of plexiform neurofibroma is not well defined and surgery is often delayed. We report a rare case of neurofibromatosis with pachydermatocele involving the nerves of the left thigh and leg with disfigurement and associated disability.

Keywords: **Plexiform, neurofibromatosis, disability.**

Introduction:

Neurofibromatosis types 1 and 2 (NF1, NF2) are autosomal dominant disorders that primarily affect the development and growth of nerve cell tissues[1]. Plexiform neurofibroma, also known as pachydermatocele, is a type of neurofibromatosis, caused by excessive growth of the neural tissue in the subcutaneous fat. It is also seen in connection with the branches of trigeminal nerve[2]. It has also been reported in retroperitoneal region, paraspinal and mediastinal area and anterior abdominal wall[3,4]. Plexiform neurofibroma is reported to occur in 26.7% of patients with type I neurofibromatosis[5]. The optimal management of plexiform neurofibroma is not well defined and surgery is often delayed until significant disfigurement has occurred. This condition is commonly seen in the west, but rarely found in the Indian subcontinent[6] .We report a rare case of neurofibromatosis with pachydermatocele involving the nerves of the left thigh and leg with disfigurement and associated disability.

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Case Report:

We have admitted an adolescent of 16 years with the chief complaint of hand on knee gait on left side at the Department of Physical Medicine and Rehabilitation, C.S.M. Medical University, Lucknow. He also gave the history of minor injury to left knee region followed by effusion which was aspirated by a local physician followed by POP immobilisation one year back. At the time of the injury, x-ray of the patient was within normal limit. One year back, he had apparently no knee joint problem. On detailed interrogation of this patient, he also complained of hypertrophy of left thigh and upper third of the left leg with very soft, flabby, elastic swelling in the skin fold (Fig 1) since childhood. His parents informed that at the time of birth, he had hyperpigmented patches on anterior abdominal wall with no other skin or skeletal abnormality. His milestones were normal and became ambulatory within 14 months of birth.

On examination of the case, we found apparent lengthening of left lower limb, nearly 15 hyperpigmented patches on whole of the body (Fig 2). The size variation was 1cmx1cm to 3.5cm × 2cm. The less than 1.5cm patches were 8 and more than 1.5cm patches were 10 in number. He has painless, extra fold of skin which is soft,

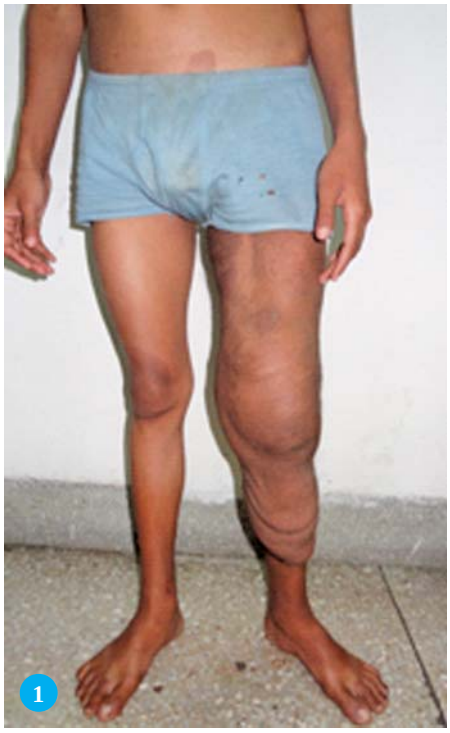
mobile, without resistance and tenderness giving a picture of pachydermatocele. It gives a picture of hypertrophy of left upper thigh and upper leg which infact is not a hypertrophy but additional skin fold on left thigh and leg. He has 1 brother, alive and healthy and parents are apparently normal.

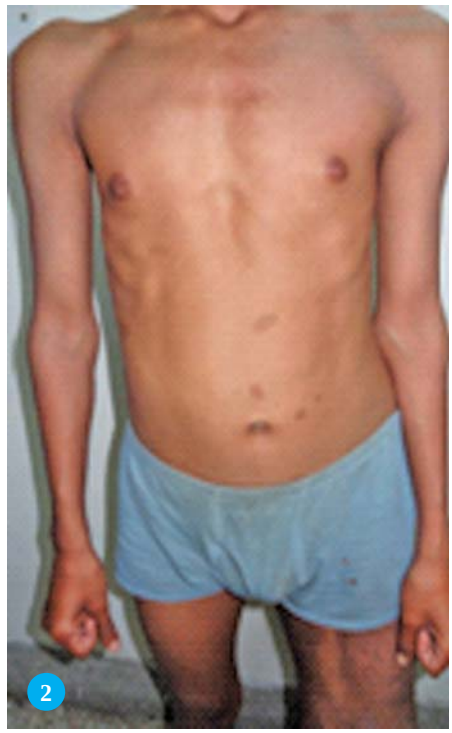
On examination of left hip, the greater trochanter is broad and shifted upwards as compared to right side, giving the picture of upwards subluxation of hip. Apparently, all movements of left hip were normal except wide abduction was not possible. There was no tenderness at knee joint but knee joint was hypermobile with marked recurvatum. Patient was walking with hand on knee gait (Fig 3). Left ankle joint was within normal limits. There was no apparent abnormality in spine and rest of his other joints. During ophthalmologic examination multiple Lisch nodules were present on the outer surface of the iris of both eyes. Neurological examination did not reveal any motor weakness or sensory loss in the lower extremities.

All routine blood investigations were within normal limit. Skiagram of pelvis with bilateral hip joints AP view showed degenerative changes in left hip region, marked abnormalities of head of femur. There was proximal migration of head of femur and picture looks as pseudoarthosis of left hip. Skiagram of left knee (Fig 4) shows marked degenerative changes in the lower end of femur, upper end of tibia with small patella. Skiagram of spine (Fig 5) showed mild scoliosis and anterior scalloping of vertebrae**.** Colour and pulsed Doppler study of vessels of left lower limb did not reveal any significant abnormality in the vascular system, however there was redundancy of skin with prominent great saphenous vein and its tributaries. MRI (Fig 6) of left thigh and calf revealed that there is hypertrophy of left thigh and calf with signal intensity alteration and heterogeneity of the muscles and subcutaneous fat of left thigh, and similar intensity alterations in muscles and subcutaneous soft tissues of left inguinal and gluteal region, suggestive of plexiform neurofibromatosis. Left femoral head, neck, greater trochanter and acetabulum are deformed in shape with irregular surfaces and articular surface of left knee joint are deformed with large osteophytes and patellar spur. Nerve conduction study of all peripheral nerves did not reveal any abnormality.

Patient was started on strengthening exercise programme and left side knee brace (Fig 7) was given followed by mobility training. He was than referred to plastic surgery for debulking surgery of left thigh and leg.

Plastic surgeon performed the unusual debulking procedure and removed excessive loose skin around thigh and leg, subcutaneous fascia including extra fat from left thigh and leg. The cutaneous nerves were thickened and vascularity was much more. Proper care was taken





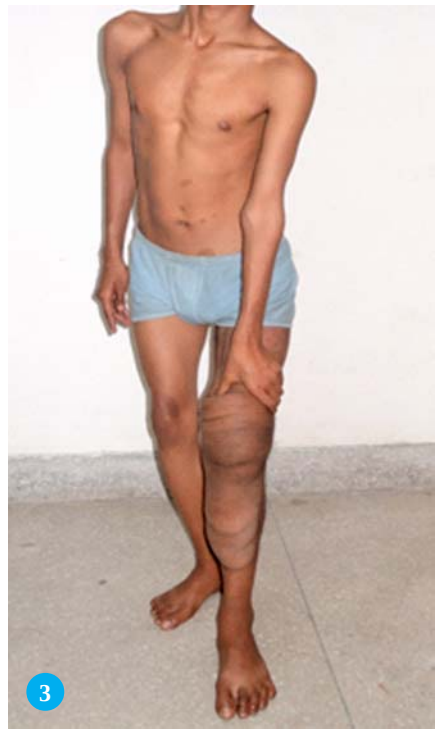


Fig 1- *Hypertrophy of Left Thigh and Leg* **Fig 2-** *Multi Cafe-lu Spot* **Fig 3-** *Hand on Knee Gait during Walking*

to preserve delicate cutaneous nerves during debulking procedure and shape of thigh/leg was restored to near normal. The patient was happy and satisfied by outcome of surgical intervention.

Removed specimen was sent for histopathological examination. Three specimen were sent, I piece was 39 × 9 × 1cm, 2nd piece was 20 × 5 × 11cm and 3rd piece was 27 × 7 × 1cm. Outer surface showed skin with prominent hair follicles. On microscopic examination, there was proliferation of monomorphic spindle cells, which have fine chromatin with inconspicuous nuclei and abundant cytoplasm with out any atypical cell. The spindle tumour cell infiltrating in adenexal structural and deep within adipose tissue. The above findings are suggestive of diffuse neurofibroma.

Discussion:

Neurofibromatosis-1 is a hamartomatous disorder, with the genetic defect localised to the long arm of chromosome 17q11.2[7]. The National Institutes of Health (NIH) in 1987 established diagnostic criteria of patients with NF-1. These well-recognised diagnostic criteria are neurofibroma (two or more simple, or one plexiform neurofibroma), cafe-au-lait spots (six or more, >5mm in greatest diameter in children and >15mm in adults), Lisch hamartomas in iris (two or more), axillary or inguinal freckling, skeletal abnormalities (sphenoid dysplasias or cortical thinning, with or without pseudo-arthrosis), optic glioma and first-degree relative with NF-1. Presence of two or more of these seven criteria establishes the diagnosis of NF-1.[8] In our patient, three of the seven above mentioned diagnostic features were present.

Plexiform neurofibroma, found in up to 26.7% of patients with NF-1 is considered an uncommon skin tumour[5], usually presenting at birth or during the first several years of life. They are non-encapsulated, poorly circumscribed tumours that diffusely infiltrate the nerve and the adjacent fat and muscle. As a result, neurofibromas are usually unresectable tumours, where tumour resection is impossible without sacrificing the nerve tissue. Fusiform enlargement of multiple nerve fascicles and branches are characteristic. Plexiform neurofibromas contain a mixture of Schwann cells, fibroblasts, reticulin and collagen fibres and a loose mucoid matrix interspersed between the axons of the parent nerve. They typically affect the trunk and extremities, but may also involve the head-neck and bladder. Associated bone dysplasia is often encountered secondary to chronic hyperaemia or

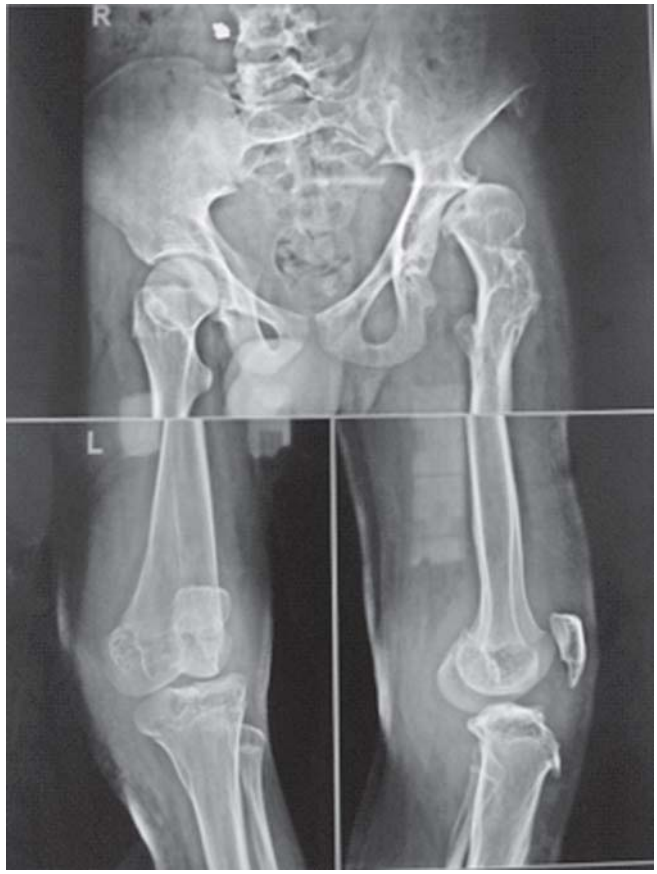


Fig 4- *Showing Proximal Migration of Femoral Head and Deformation of Acetabulum, Head of Femur and Distal Femur*

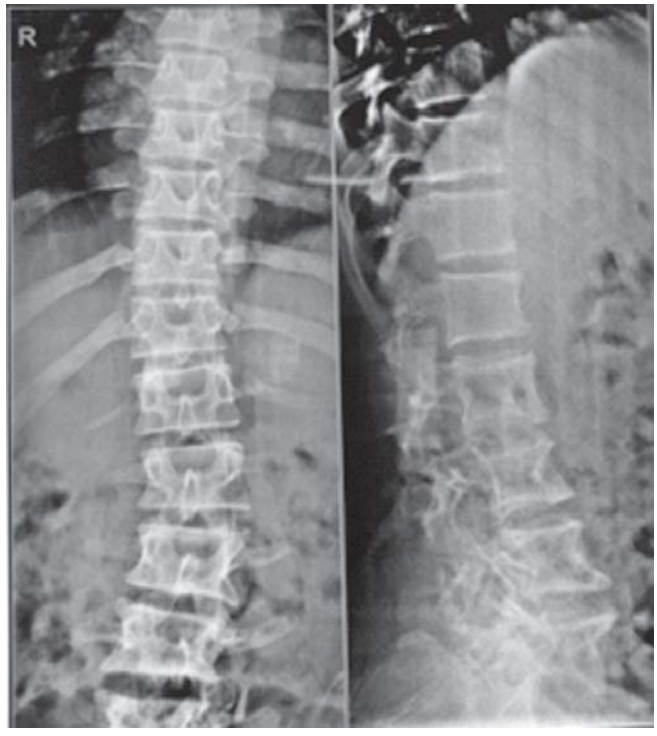


Fig 5- *Anterior Scalloping of Vertebrae with Mild Scoliosis*

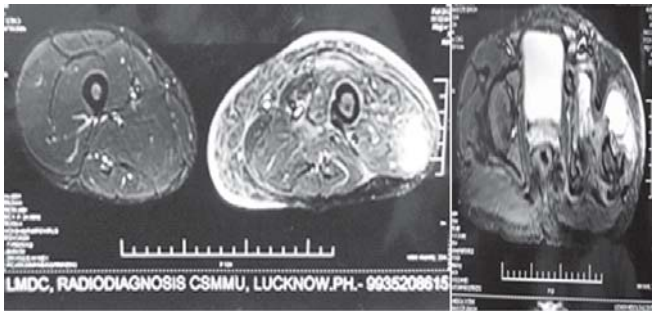


Fig 6- *MRI Left Thigh-hypertrophy of Left Thigh and Calf with Signal Intensity Alteration and Heterogeneity of the Muscles and Subcutaneous Fat of Left Thigh*

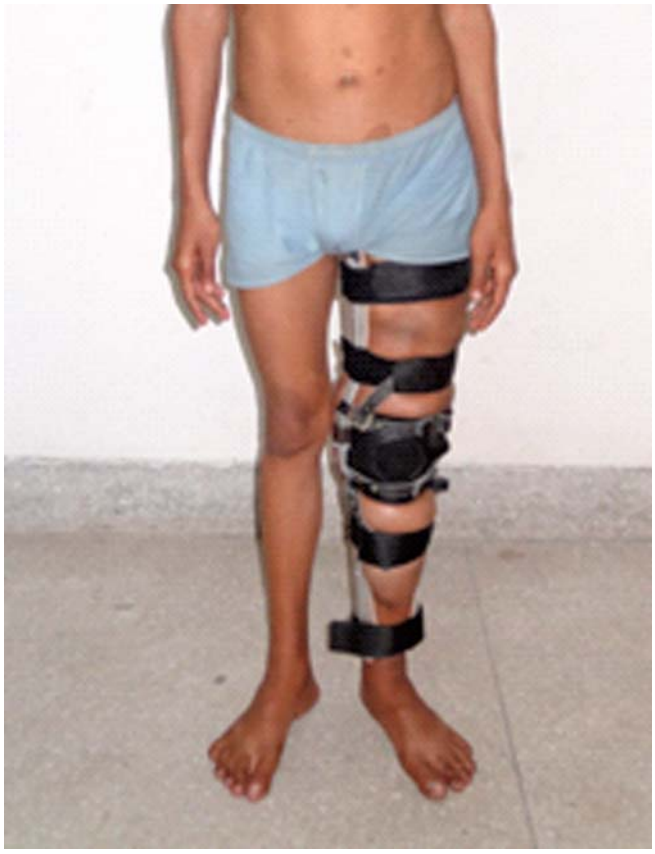


Fig 7- *Patient with Knee Orthosis*

as part of the mesodermal dysplasia. Such tumours give rise to a variety of problems, including disfigurement and functional impairment.[9]

Summary:

We describe a rare case of plexiform neurofibromatosis, presenting as progressive hypertrophy of left thigh and leg, its typical clinical, microscopical, and imaging features diagnostic of NF1. We also propose the proper and meticulous clinical assessment in such cases for early detection of NF1 followed by proper debulking surgical procedure by an experienced plastic surgeon.

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