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## Neurofibroma Arising from Genitofemoral Nerve in Neurofibromatosis Type I— A Rare Entity

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**Abstract:** Neurofibromatosis type I is a hamartomatous disorder with CNS, orbital, musculoskeletal, pulmonary and cutaneous manifestations. Neurofibromas are of three types – localised, diffuse and plexiform. Plexiform neurofibromas (PNF) are exclusively associated with NF1. Plexiform neurofibroma (PNF) grows along nerve roots from the main nerve root to a small distal branch. It is of two main types – internal tumour and superficial tumour. Internal PNF usually extends through multiple tissue planes and usually are not completely resectable. The risk of malignant progression is high in internal PNF. We report a case of 25-year-old female patient with NFM1 presenting with a lump in abdomen on the left side of the midline for past 1 month. Ultrasound revealed a well-defined mass in the retroperitoneum on left side along the course of genitofemoral nerve with cystic degeneration and haemorrhage in the central portion. These findings were confirmed on Computed tomography (CT) and Magnetic resonance imaging (MRI) of abdomen and pelvis and on histopathology. The unique feature of our case is a neurofibroma arising from genitofemoral nerve which is extremely rare.

**Keywords:** Neurofibromatosis, Neurofibroma, Plexiform, Genitofemoral nerve

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### INTRODUCTION

Neurofibromatosis type I is also called Von Recklinghausen disease. It is the most common phacomatosis with multisystem neurocutaneous disorder. Neurofibromatosis type I affects 1 in 3000 individuals. It is inherited as Autosomal dominant condition in half of cases, while it occurs due to new mutations in 50% of cases. Though there is variable expression, 100% penetrance occurs by 5 years of age [1].

Neurofibromatosis type I is a group of genetic disorder that affects the cellular growth of neural tissues. There are 8 forms of Neurofibromatosis of which two main forms exist – Type I (Neurofibromatosis type I) and Type II (Neurofibromatosis type II). Neurofibromatosis type I is also called Von Recklinghausen disease. It was first discovered by pathologist Friedrich Daniel Von Recklinghausen in 1882 and is a neurodermal dysplasia [1]. On histopathological examination, neurofibromas are composed of Schwann cells, perineural cells and endoneural fibroblasts and are not capsulated[1].

Neurofibromatosis type I have CNS, orbital, musculoskeletal, pulmonary and cutaneous manifestations. It is primarily a hamartomatous disorder that involves the ectoderm and mesoderm. Neurofibroma are of three types – Localised neurofibroma (cutaneous) located in the dermis and subcutis; Diffuse neurofibroma (subcutaneous) located in subcutis usually in head and neck region and Plexiform neurofibroma seen in pelvis, neck and extremities[2].

Genitofemoral nerve originates from the upper L1 and L2 segments of the lumbar plexus. It pierces psoas major and emerges from its anterior surface. It courses inferiorly in pelvis along the anterior surface of left psoas and iliacus and divides into genital and femoral branches which courses in inguinal and femoral canal respectively [3].

### CASE REPORT

A 25-year-old female patient presented with a lump in abdomen on the left side of the midline since 1 month. There were no bowel or bladder complaints or history of fever. The patient was a known case of Von

Recklinghausen disease having multiple café au lait spots and cutaneous neurofibroma in abdominal wall (Figure 1). Her elder brother gave a history of similar disease.

USG abdomen and pelvis (Figure 2) showed a well-defined mass measuring approximately 11.6 (CC) x 7.3(AP) x 9.8 (T) cm on the left side of abdomen anterior to left psoas and iliacus in left lower lumbar and iliac region. Its peripheral portion was solid while central portion was anechoic with fine internal echoes and fine septations suggestive of cystic necrosis/degeneration. Doppler (Figure 2C) showed mild vascularity in peripheral solid portion. It was compressing and displacing left iliac vessels medially. Inferiorly, the mass was extending in left inguinal region.

CT abdomen (Figure 3 A-I) showed a large well defined mass measuring approximately 12.6(CC) x 8.6(AP) x 9.1(T) cm in the retroperitoneum in left side of the abdomen in left lumbar and iliac region extending inferiorly up to left inguinal region. It closely abutted underlying left psoas and iliacus muscle, which were flattened with obliteration of intervening fat planes. Anteriorly, it was abutting overlying anterior abdominal wall muscles. Medially, it was causing extrinsic compression on left external and internal iliac vessels. The peripheral portion of the mass was solid (CT value 20-30 HU) on plain study showing heterogeneous enhancement (CT value 45-60 HU). The central portion of the mass appeared hypodense with

lack of enhancement (CT value 15-25 HU) suggestive of central necrosis. No calcification was noted. Mild mass effect was noted on the left lateral wall of urinary bladder. Adjoining portion of left ureter was compressed and displaced medially with no proximal obstructive uropathy.

On MRI (Figure 4 A-D), the mass appeared heterogeneously hypointense on T1WI (Figure 4 A) and heterogeneously hyperintense on T2WI (Figure 4-B,C) suggestive of necrotic areas. Hyperintense foci noted on T1WI within the mass were suggestive of subacute haemorrhage. The peripheral solid portion appeared slightly hyperintense with respect to muscle while the central predominant cystic portion appeared hyperintense on T2WI and STIR with septations (Figure 4 D).

Multiple small, well defined soft tissue density lesions were seen in skin on anterior abdominal wall on the left side in the left paraumbilical region, right lateral abdominal wall and both gluteal regions, left posterior abdominal wall in lumbar region. They showed enhancement on contrast on CT (Figure 3-D). On MRI they appeared hypointense on T1 and hyperintense on T2 suggestive of multiple cutaneous neurofibromas (Figure 4A, B).

FNAC was suggestive of spindle cell tumour with multiple spindle cells with wavy nucleus without pleomorphism, with increased cellularity and arranged in fascicles intermingled with collagen (Figure 5).



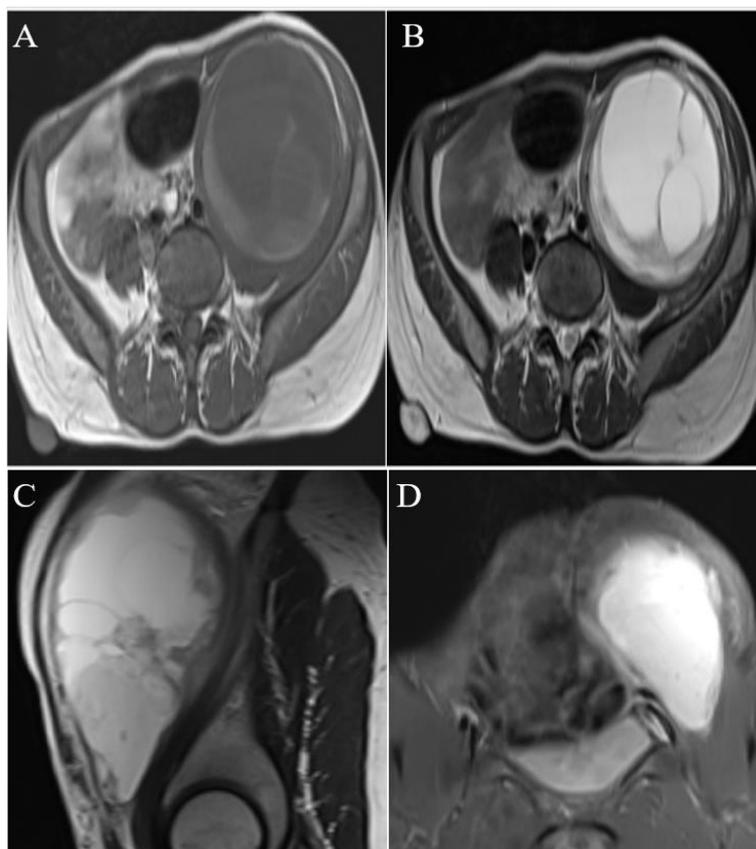
**Fig-1: Photograph of abdomen showing multiple café au lait spots and cutaneous neurofibromas.**



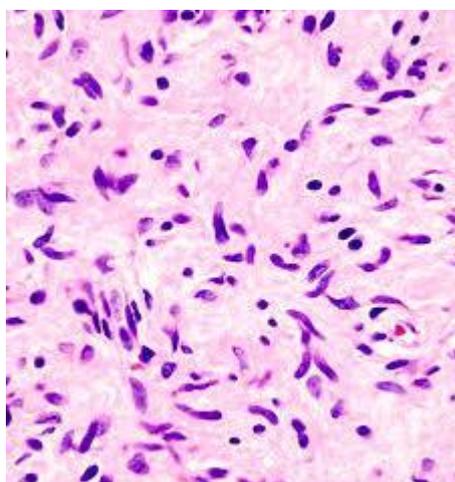
**Fig-2 (A, B):** USG showing a well-defined mass in retroperitoneum anterior to left iliacus, solid in the periphery with central cystic degeneration showing mild peripheral vascularity on doppler(C).



**Fig-3:** CT abdomen and pelvic plain (A-C) and contrast study (D-I) showing a well-defined mass in retroperitoneum anterior to left psoas and iliacus, extending inferiorly in left inguinal region with peripheral solid portion showing heterogeneous enhancement and central cystic degeneration with cutaneous neurofibroma in right gluteal region.



**Fig- 4:** MRI abdomen and pelvic (A: Axial T1WI , B: Axial T2WI, C: Sagittal T2WI, D: Coronal STIR) showing a well-defined mass in retroperitoneum anterior to left psoas and iliacus, extending inferiorly in left inguinal region with peripheral solid portion and central cystic degeneration with areas of haemorrhage appearing hyperintense on T1WI with cutaneous neurofibroma in right gluteal region.



**Fig-5:** HPE (Haematoxylin and eosin stain) showing spindle cells with wavy nuclei without pleomorphism with collagen and mast cells.

#### DISCUSSION

Neurofibromas are benign tumours of peripheral nerves. They can be dermal or plexiform. Dermal neurofibromas are well defined solid cutaneous tumours of limited size. Plexiform neurofibromas (PNF) arise from subcutaneous or visceral peripheral nerves and involve multiple fascicles extending along a variable length of the nerve. These vary in size and can be large. Plexiform neurofibromas (PNF) occur in 30%

in NF1 patients. Plexiform neurofibromas (PNF) are exclusively associated with NF1 while dermal neurofibroma can occur in non NF1 patients. Plexiform neurofibroma (PNF) usually shows growth along nerve roots extending from the main nerve root to a small distal branch. It is of two main types – internal tumour and superficial tumour. Superficial PNF does not cross tissue planes and usually can be completely resected. Internal PNF usually extends through multiple tissue

planes and usually are not completely resectable without damaging adjoining tissues and organs. They appear as a paraspinal mass involving multiple spinal levels in chest, abdomen and pelvis. The risk of malignant progression is high in internal PNF. On histology, they show Schwann cells with wavy contours with ovoid to elongated nucleus with fine dense heterochromatin, intermingled with fibroblast and perineural cells and varying amounts of collagen fibres [4].

Neurofibromatosis type I with abdomen and pelvis involvement can arise in paraspinal, retroperitoneal and mesenteric regions. It may involve numerous ganglia within the abdomen and somatic and autonomic nervous tissue innervating the abdomen and pelvis organs. Multiple neurofibromas arising from nerves traversing retroperitoneum and mesentery can mimic lymphadenopathy (tuberculosis, Whipple's disease, metastatic retroperitoneal lymphadenopathy from seminoma, Mycobacterium avium complex). They usually demonstrate low attenuation in 73% of cases due to cystic degeneration, lipid rich Schwann cells, confluent areas of hypocellularity, entrapped perineural adipose tissue. Peripheral high attenuation is due to diffuse cellular components and collagen rich regions. Peripheral portion shows enhancement due to cellular and fibrous element while the central portion shows lack of enhancement as it is hypovascular. Sarcomatous degeneration of peripheral neurofibroma occurs in 5-15% of patients of neurofibromatosis type I [5].

Degenerative schwannoma usually exhibits cystic degeneration, haemorrhage, calcification and hyalinisation. Till now only 1 case of neurofibroma arising from genitofemoral nerve has been published [6].

## CONCLUSION

Internal plexiform neurofibroma arising from genitofemoral nerve is rare. It mimics the retroperitoneal mass or lymphadenopathy with central necrosis. Presence of cutaneous neurofibromas and family history of NFM1 can give clues to the diagnosis. Degenerative neurofibroma shows cystic degeneration and haemorrhage in its central portion. Sarcomatous degeneration may rarely occur. Complete resection is difficult due to involvement of adjoining structures with chances of recurrence.

## Conflict of interest

None

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