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# IMAGING FINDINGS OF AN INTERESTING CASE OF RETRO-PERITONEAL MASS TURNED OUT TO BE VON RECKLINGHAUSEN DISEASE (MULTIPLE PLEXIFORM NEUROFIBROMAS)

Radiology

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## ABSTRACT

Neurofibromatosis is not a single entity but a group of genetically and clinically distinct disorders with few overlapping features. Although they are the most common CNS tumor predisposition syndromes, neurofibromas are multisystem disorders with both neoplastic and non- neoplastic manifestations.

## KEYWORDS

NF-1, Plexiform neurofibroma, MRI.

**INTRODUCTION:**

Neurofibromatosis (Type-1) was formerly known as Von Recklinghausen disease or peripheral neurofibromatosis[1]. Extreme form of NF-1 can be highly disfiguring and is noted as elephantiasis neuromatosa / elephant man disease[2]. NF-1 is an autosomal – dominant disorder. Plexiform neurofibromas develop in individuals with NF-1 when there is widespread involvement along a nerve segment and its branches producing dumb- bell shaped configuration / bag of worms appearance, are largely pathognomonic for neurofibromatosis type 1 (NF1)[3].Cause of death in patients with NF-1 are due to malignant transformation[4]. In this literature we report an interesting case of retroperitoneal mass tuned out to be plexiform neurofibroma with skin and bone lesions.

**Case Report:**

A Thirty-one year old male , smoker , alcoholic with no co-morbidities referred from surgery , opinion regarding USG and CT Abdomen films taken elsewhere. Past history reveals subcutaneous swelling and skin patches all over the body which are asymptomatic and increase in size and number were reported. Similar family history revealed his father had multiple subcutaneous swelling in the back with skin pigmentation. No history of medical or surgical treatment documented.

General examination of the patient shows, Dolichocephaly (Figure: 1), hyper pigmented macules (Café au lait spots) of varying sizes noted all over the body, predominantly in the anterior chest wall and back (Figure: 2), multiple subcutaneous swelling seen in neck (Figure: 3) and axial freckles (Figure: 4). Ocular examination shows Lisch nodule (Figure: 5) with normal vision. Motor and sensory examination appears normal.

NCCT-abdomen shows liver cyst (Figure: 6), Bilateral multi-lobulated hypo dense lesions posterior to psoas major muscle probably originating from bilateral neural foramina (Figure: 7) these multi-lobulated mass seen extending into the pelvis giving conglomerate appearance with widening of sacral foramina (Figure: 8).Bilateral lobulated para vertebral lesions with neural foramina widening and scalloping of posterior margin of dorsal and lumbar vertebra (Figure: 9), After complete CT evaluations we are convinced lesions are not lymphadenopathy and patient is further evaluated with X-ray and MRI.

CXR-PA view (Figure: 10) shows mild scoliosis with convexity towards right with mediastinal widening more superiorly. Ribbon shaped ribs with subtle superior rib notching. Skull x-ray lateral and AP view (Figure: 11) shows Dolicocephaly with sella widening (J shaped sella).

MRI imaging ( Figure :12- 17) reveals multiple lobulated T2 hyper intense mass of varying sizes seen involving the exiting nerve roots of all levels, largest seen involving at the level of C6 causing spinal canal widening. These masses also seen involving the peripheral nerve roots mainly involving left sided radial nerve, bilateral sciatic nerves and intercostal nerves. Smooth tubular symmetric T2 hyper intense paraspinal mass with enlargement of neural foramina giving dumb-bell shape configuration. These masses had extension out of neural foramina creating a large conglomerate pelvic mass. No significant abnormality detected in the MRI brain study.

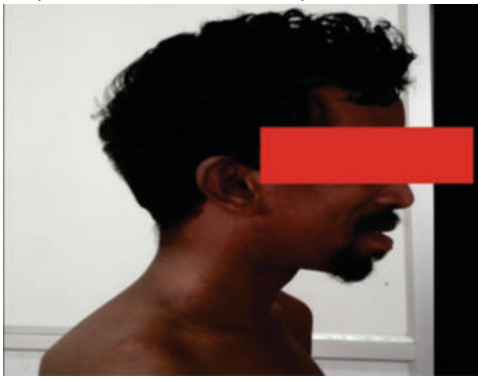

**Figure: 1** Dolicocephaly

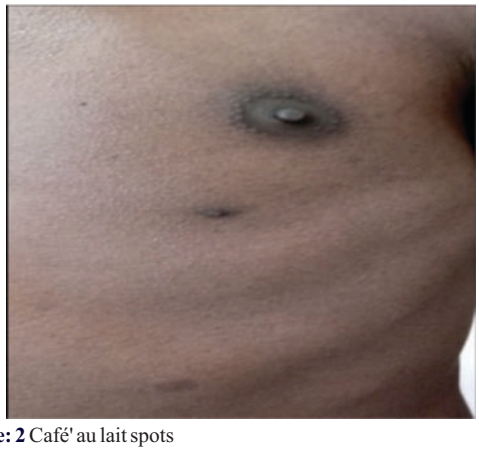

**Figure: 2** Café' au lait spots

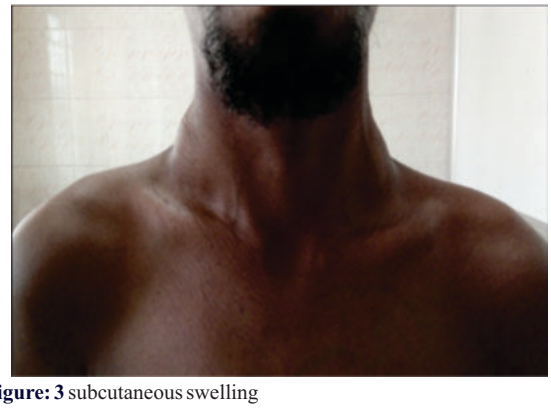

**Figure: 3** subcutaneous swelling

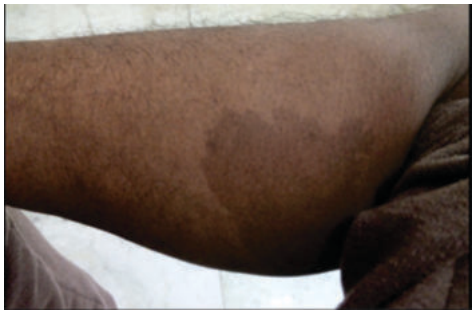

**Figure: 4** Freckles

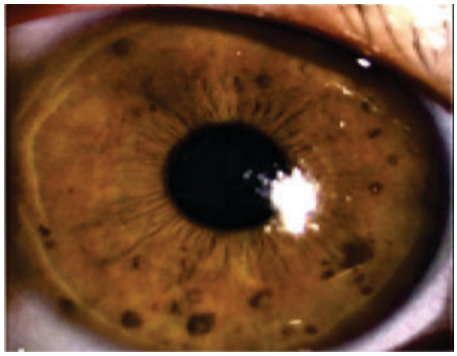

**Figure: 5** Lisch nodule

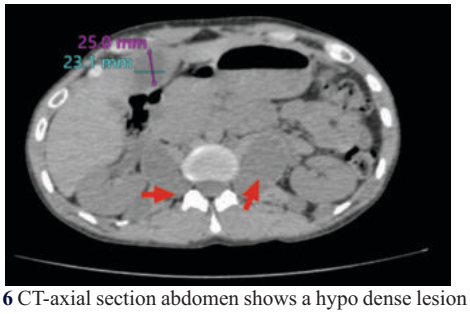


**Figure: 6** CT-axial section abdomen shows a hypo dense lesion in the right lobe of liver – simple cyst.

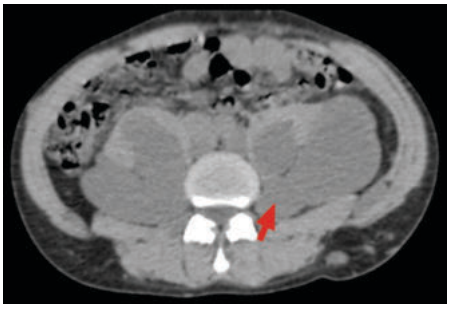

**Figure:7** CT axial section abdomen shows bulky psoas muscle with multilobulated hypo dense lesion posterior to psoas muscle.

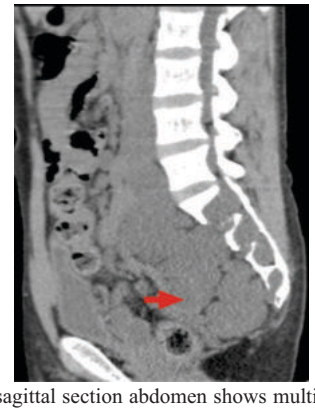

**Figure:8** CT- sagittal section abdomen shows multi- lobulated hypo dense conglomerate mass seen extending into the pelvis with widening of sacral foramina.

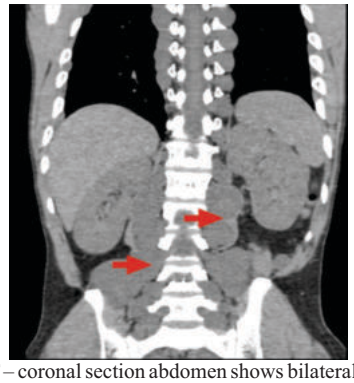

**Figure:9** CT – coronal section abdomen shows bilateral lobulated para vertebral lesions with neural foramina widening and scalloping of posterior margin of dorsal and lumbar vertebra

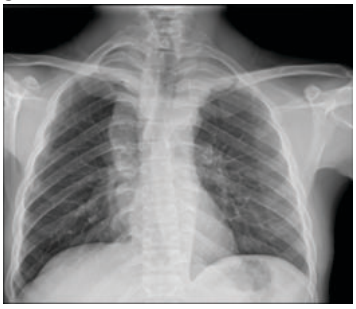

**Figure:10** Chest x-ray PA view showing scoliosis.

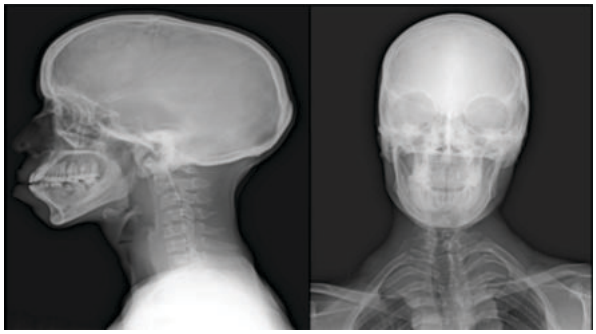

**Figure:11** Skull x-ray lateral and AP view showing dolicocephaly , J shaped sella. Orbit appears normal.

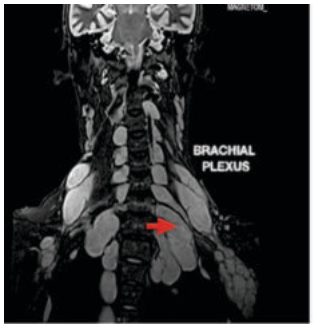


**Figure: 12** Coronal FS T2WI

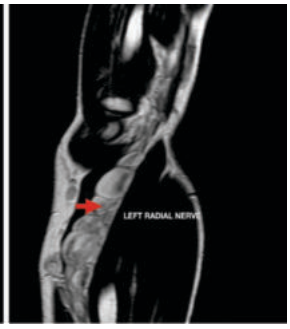


**Figure:13** Saggital T2WI upper limb

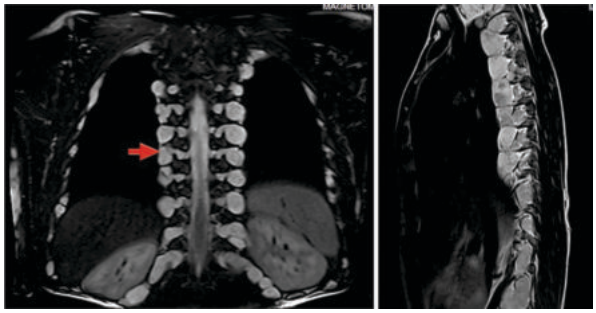

**Figure: 14** Coronal and saggital T2WI sshowing Dumb- bell configuration.

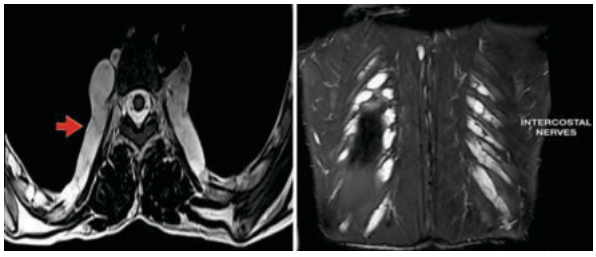


**Figure: 15** Axial and coronal T2WI showing invovlement of intercostal nerve roots.

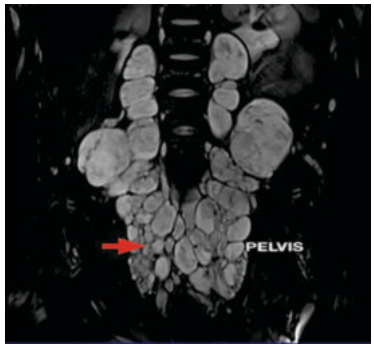


**Figure:16** Coronal T2WI shows lesions extend into pelvis creating a large conglomerate pelvis mass.

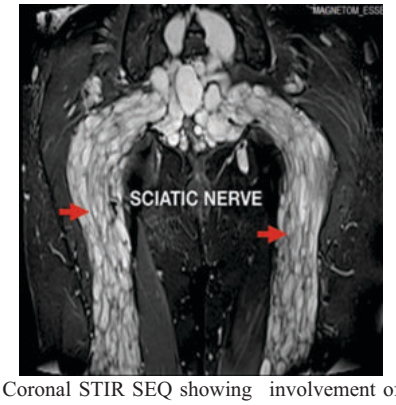


**Figure:17** Coronal STIR SEQ showing involvement of peripheral nerve plexus ( Sciatic nerve roots).

## DISCUSSION:

Plexiform neurofibromas developing into MPNSTs is the main cause of death in NF1 patients[5]. Individuals with NF1 have a five-year survival rate for MPNSTs compared to individuals without NF1[6]. Early diagnosis helps in the proper management that prolong the survival rate; however diagnosing malignant form from NF-1 is challenging due to clinical overlap between benign and malignant tumours[7]. Rapid growth and neurologic impairments are the warning symptoms that malignant peripheral nerve sheath tumor shows most frequently, while pregnancy and puberty can also cause rapid growth of the tumor[8].

Imaging modalities helpful in diagnosing asymptomatic cases[9]. As in this case patient is asymptomatic with all positive clinical features presented to us with imaging finding of a restroperitoneal mass with further evaluation diagnosis of Plexiform neurofibroma is made with imaging findings and diagnostic criteria namely , major criteria includes > 6 café spots , 2 cutaneous neurofibromas, > 2 lisch nodules . axillary freckling , First degree realtive eith NF-1 and supportive criteria includes scoliosis[10,11].

For improved outcomes and early detection of malignant transformation, NF1 patients must be educated about MPNST clinical characteristics[12]. Additionally annual clinical assessment are advised for NF1 patients to look for the malginant transformation and tratment follow-up.

## CONCLUSION:

Neural origin tumors should be considered as one of the differential diagnosis when a mass is seen in the retro-peritoneal region posterior to psoas muscle.

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## Authors' contribution

Lekha priya M: Writing, Investigation, Analysis, Review and Editing All authors have read and agreed to submit the manuscript. Informed consent, proper written consent obtained from the patient.

G. Murugan: Conceptualization, Supervision, Methodology, Resources, Data Collection, Writing and Formal analysis.

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## Conflict of interest

The authors declare that there is no conflict of interests.

## Data And Materials Availability

All data sets collected during this study are available upon reasonable request from the corresponding author.

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