Journal of Medical Imaging & Case Reports

https://doi.org/10.17756/micr.2019-031

Clinical Image

Open Access

Extensive Plexiform Neurofibromatosis

Surekha Dabla[1*], Vinay Goyal[2] and Vishal Goyal[3]

[1]*Department of Neurology, Pt BDS PGIMS, India*
[2]*Department of Neurology, All India Institute of Medical Sciences, India*
[3]*Department of Neuroradiology, All India Institute of Medical Sciences, India*

[*]Correspondence to:
Dr. Surekha Dabla, D.M. Neurology
Senior Professor
Department of Neurology
Pt BDS PGIMS, Rohtak
Haryana 124001, India
Tel: 0126282072
E-mail: surekhadabla@yahoo.co.in

Received: February 15, 2019
Accepted: October 10, 2019
Published: October 11, 2019

Citation: Dabla S, Goyal V, Vishal G. 2019. Extensive Plexiform Neurofibromatosis. *J Med Imaging Case Rep* 3(2): 34-35.

Published by United Scientific Group

Abstract

Plexiform neurofibroma is uncommon variant of neurofibromatosis with tendency to malignant transformation. We present a case of 17-year-old girl with neurofibromatosis type 1 who presented with an extensive plexiform neurofibroma.

Keywords

Plexiform neurofibroma, Neurofibromatosis

Case History

A 17-year-old girl, normal birth and developmental history, without any positive family history of similar illness, was presented with gradually progressive swelling over right arm. At the time of birth there was only black discoloration of right upper limb, back of neck and right shoulder. Since last 2-3 years she had black brown pigmented spots on her face and pedunculated swelling on right arm, forearm, neck and right side of chest anteroposteriorly. Since, last one year she had started difficulty in lifting her right arm above shoulder although she could perceive sensation normally all over. Examination revealed a diffuse, nontender, hyperpigmented, freely mobile, soft tissue swelling over right arm extending up to right side nape of neck with multiple cafe –au- lait macules (Figure 1). DTR were elicitable (+1) in all four limbs. Her sensory examination was essentially normal pain, touch and temperature all modalities. Her power was 4+/5 in all four limbs (she only had difficulty in raising her right upper limb because of the large pedunculated mass). Ophthalmological examination was normal. There was no other obvious neurofibromas on any other part of body. Her MRI for brain, cervical spine were done and are presented in figure 2. Axial T2W images of the brain at the level of midbrain and thalami shows round to oval well defined discrete hyper intense signal intensity areas suggestive of FASI (focal area of signal intensity) characteristic of neurofibromatosis. MRI cervical spine sagittal and coronal T2W images shows a small lobulated well defined extramedullary intradural lesion at C4-C5 level on right side compressing the cervical spinal cord & extending in the extradural location through the right C4-C5 neural foramina. In addition, small neurofibroma along the right C6 nerve root in the extradural location through right C5-C6 neural foramina and a large 55 mm x 40 mm sized lesion of neurofibroma in right axillary region in the lower part of brachial plexus was also present.

Discussion

Neurofibromatosis type 1 is amongst most common autosomal dominant

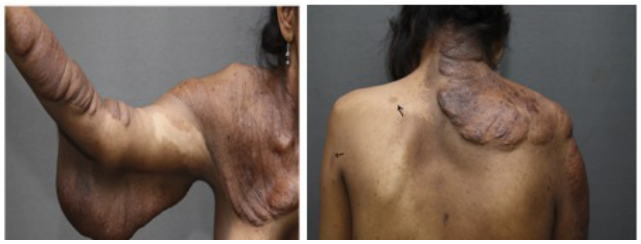

Figure 1: Plexiform neurofibromatosis (right) with hyperpigmentation and cafe -au -lait macules.

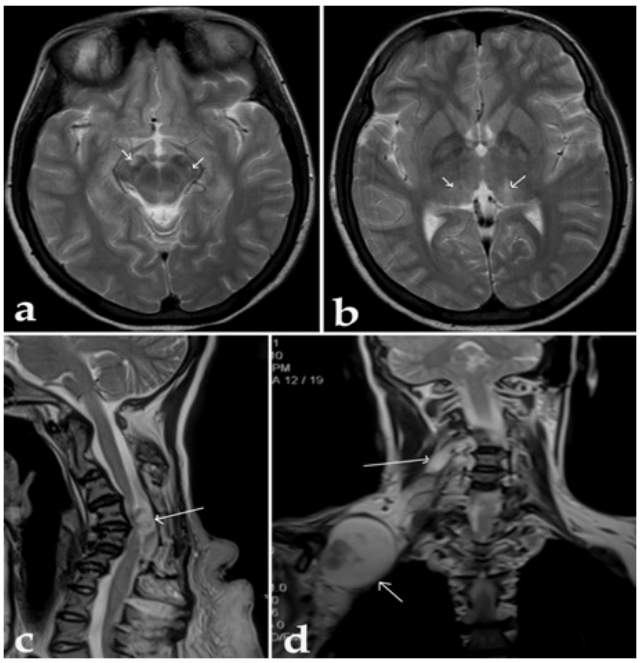

Figure 2: MRI brain and cervical spine T2 weighted images.

disorders, clinically characterized by cafe´-au-lait macules and axillary freckling, lisch nodules, optic gliomas and neurofibromas. Four types of neurofibromas are recognized. The first type of neurofibroma is characterized by discrete, blue-tinged cutaneous neurofibromas in the epidermis and dermis. The second type of neurofibroma is distinguished by subcutaneous neurofibromas occurring deep in the dermis, along the course of peripheral nerves. Both of these types of nodules feel firm and rounded, and the skin moves over them. The third type of neurofibroma consists of localized nodular plexiform neurofibromas that interdigitate in normal tissues. The fourth type of neurofibroma is characterized by diffuse plexiform neurofibromas that infiltrate widely and diffusely into surrounding tissues. Classically, they feel like a "bag of worms" when palpated. Localized and diffuse plexiform neurofibromas are the most active.

About 30% of cases of NF-1 may have plexiform neurofibromas, they manifest early in life, with a very high predisposition to malignancy and later they can transform to malignant peripheral nerve sheath tumors (MPNST) which carries extremely poor prognosis [1]. Although, previously there were no definite clinical, genetic and pathological markers of identifying or predicting which NFs will progress to MPNST, but recently few researchers identified markers like IGFBP 1 (insulin-like growth factor binding protein 1) and RANTES (regulated upon activation, normal T-cell expressed and secreted) in patients suffering with both NF1 and MPNST in comparison to those without MPNST [2]. Treatment of plexiform neurofibromas is surgical, however risk of excessive bleeding during surgical excision is very high because of presence of abnormal vessels in a subset of patients. A preoperative angiography followed by embolization of feeding vessels and complete surgical excision may reduce the risk of bleeding [3]. However, chances of recurrence are 20% despite of best efforts [4].

Conflict of Interest

The authors declared no conflict of interest.

References

1. Sabatini C, Milani D, Menni F, Tadini G, Esposito S. 2015. Treatment of neurofibromatosis type 1. *Curr Treat Options Neurol* 17(6): 355. https://doi.org/10.1007/s11940-015-0355-4
2. Park SJ, Sawitzki B, Kluwe L, Mautner VF, Holtkamp N, et al. 2013. Serum biomarkers for neurofibromatosis type 1 and early detection of malignant peripheral nerve-sheath tumors. *BMC Medicine* 11: 109. https://doi.org/10.1186/1741-7015-11-109
3. Littlewood AH, Stilwell JH. 1983. The vascular features of plexiform neurofibroma with some observations on the importance of pre-operative angiography and the value of pre-operative intra-arterial embolisation. *Br J Plast Surg* 36(4): 501-506. https://doi.org/10.1016/0007-1226(83)90140-6
4. Needle MN, Cnaan A, Dattilo J, Chatten J, Phillips PC, et al. 1997. Prognostic signs in the surgical management of plexiform neurofibroma: the children's hospital of Philadelphia experience, 1974-1994. *J Pediatr* 131(5): 678-682. https://doi.org/10.1016/s0022-3476(97)70092-1