

Atypical Presentations in Neurofibromatosis- A Series of Five Cases**Dr. Bijaya Kumar Sadangi¹, Dr. Prangya Panda², Dr. Sanjeeb Patro³, Dr. Sarita Panda⁴, Dr P.C Malla⁵**¹Associate Professor, Dept. of Orthopedics²Associate Professor, Dept. of Ophthalmology³Assistant Professor, Dept. of Anaesthesiology⁴Assistant Professor, Dept. of Ophthalmology⁵Senior Resident, Dept. of Orthopedics

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Abstract: Neurofibromatosis is the most common phacomatosis having a frequency of approximately 1 case per 3500 persons in general population. The syndrome of neurofibromatosis consists of two distinct genetic diseases with considerable phenotype overlap. We present here five cases of neurofibromatosis with atypical presentations. Their clinical presentations and management are different.**Keywords:** neurofibromatosis, phacomatosis, neuroectoderm, phenotype, café au lait spot, axillary freckles, plexiform neurofibromatosis, hamartoma, pseudo-arthritis tibia.**INTRODUCTION**

Neurofibromatosis is the most common phacomatosis having a frequency of approximately 1 case per 3500 persons in general population. The syndrome of neurofibromatosis consists of two distinct genetic diseases with considerable phenotype overlap. These two forms are neurofibromatosis type 1 and neurofibromatosis type 2. Both are characterized by neuroectodermal tumors that arise within multiple organs and have autosomal dominant inheritance. Many features of these syndromes don't appear until late childhood or early adulthood. The severity of the syndrome varies markedly from patient to patient[1,2].

Here we report few atypical presentations of neurofibromatosis specially in eye & bone.

CASE REPORTS.**Case no. 1**

Following trivial injury in a 2 year old child the patient developed hyphaema, proptosis and loss of vision. There was digitally raised intraocular pressure and large cornea. Treatment with antiglaucoma medication(timolol e/d, 2 times daily) and steroid e/d(prednisolone e/d, 4 times daily for 7 days, then tapering doses) caused reduction in intraocular pressure and improvement in hyphema.

**Fig-1: Axillary Freckles****Fig-2: Café Au Lait Spots**

On examination the patient had axillary freckles and café-au-lait spots over the trunk. Under general anaesthesia trabeculectomy was done. The

hyphema and intraocular pressure was controlled. Visual acuity was improved.

Case no. 2

29 year old female presented with red eye and loss of vision. There was uveitis and raised intraocular pressure(30mm hg). On examination there was fibroma molluscum all over body including face. Lisch nodules were present over iris. Café au lait spots were present(8 in number over trunk). Medical treatment with antiglaucoma drugs(timolol 0.5%, 2 times daily) and steroid e/d(prednisolone acetate e/d) 5 times daily for 7 days with tapering doses. The uveitis and glaucoma was controlled medically. Intraocular pressure was improved to 15 mm of hg.



Fig-3: Inside of Plexiform Neurofibromatosis

Case no-3

One 8 year male child presented with progressive bending of right leg, noticed by parents when the child was 1 year old, and there after treated by bone setters by repeated plaster. X-ray of the part revealed bone gap, rounding of the ends & medullary obliteration. On surgical exploration there was intervening synovial joint like cavity with thick synovial fluid inside. The defect was very much resistant to conventional treatment like excision of the ends, bone grafting, DCP plating etc. Histopathological study of excised tissue confirmed neurofibroma. Finally result was excellent only after bypass grafting as recommended by Mc Ferland.

Case no-4

One female child 4 year old was diagnosed pseudo-arthritis tibia (Rt) clinically & radiologically. Biopsy confirmed neurofibroma. Bypass fibula grafting from opposite leg, without disturbing the pseudo arthritis site was undertaken, along with external splintage. The patient is being followed up for last 3 years and results are favorable.

Case no-5

A girl 14 years presented with dull-aching pain proximal tibia since 2 years, had consulted several surgeons. Conventional X-ray did not show any abnormality. Digital X-ray with megaview showed a lytic lesion with ill defined calcification surrounding it.

Cortical window and excision of the lesion with accurate localization under C-arm alleviated the symptoms. Histopathology suggested neurofibroma.

DISCUSSION

From the aforesaid observations it is self evident that diagnosis of neurofibromatosis or Von Reckling Hausens disease is not difficult as the multiple nodular soft swellings of varied size present more or less all over the body surface is diagnostic. However one should have in mind about the unusual sites of presentation and its subsequent complications. Literatures are clear about scoliosis and compressive neuropathy caused by neurofibroma, but we could not find any case from previous records verified by us. Nor there was any record stating malignant transformation of a pre-existing neurofibroma[1, 2]. Ocular features of neurofibromatosis may be optic nerve glioma, other tumours like neurilemmoma, plexiform neurofibromatosis, meningioma, spino-orbital encephalocele[5]. Eyelid plexiform neurofibromatosis when involves upper lid can give rise to mechanical ptosis. Anterior segment changes include lisch nodule, prominent corneal nerves, congenital ectropion uvae. There may be glaucoma, choroidal hamartoma.[1, 6].

CONCLUSIONS

Lisch nodules present on iris in patients with multiple neurofibromatosis can present with raised intraocular pressure, uveitis, hyphaema and give rise various complications leading to loss of vision[3]. Similarly pseudo-arthritis of tibia with resistance to conventional modalities of management is another mode of presentation of neurofibroma. There is paucity of literature about intraosseous neurofibroma which is confused with other dreaded tumorous conditions like osteosarcoma, steoid osteoma, giant cell tumour etc[1,4].

However this is a small series, more and more research and reporting can be more exploarative in these morbiditis.

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