Atypical Presentations in Neurofibromatosis- A Series of Five Cases

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Abstract: Neurofibromas 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-arthrosis 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.

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 (30 mm 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.

**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-arthrosis 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.

**REFERENCES**

5. Crenshaw AH (Ed.); Campbell’s operative orthopedics surgery, 8th edition, p-271,317

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