Progressive Craniofacial Abnormality in a Case of Congenital Neurofibromatosis

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ABSTRACT

Introduction: Neurofibromatosis is a generalized form of benign tumour of peripheral nerves, from fibrous element of nerve sheath. There are two types of neurofibromatosis, both being autosomal dominant. Type 1 also known as VON RECKLINGHAUSEN disease. We present a rare case of 24 years female with congenital neurofibromatosis type 1 and correlate with clinical, radiological and histomorphological features.

Clinical case: Radiological CT, MRI images, histomorphological features correlated with clinical data of a 24 years female presented with facial asymmetry, diminished vision on right side due to orbito facial swelling since 3 months of life which gradually increased in size. Clinically the swelling was suspected as malignancy. Radiologically the patient had right sided sphenoid dysplasia with orbital encephalocele (figure 3) and associated thickening and nodularity of orbit frontotemporal region. Progressive craniofacial abnormalities in a case of congenital neurofibromatosis confirmed histopathologically and correlated with radiological and clinical profile of the patient.

Conclusion: Early diagnosis with the available modalities can have a better prognostic outcome in a case of congenital neurofibromatosis and associated craniofacial abnormalities. They require intensive neurosurgical and ophthalmological workup for improving patient's outcome.

Keywords: Neurofibroma, Congenital, Craniofacial, Neurofibromatosis.

INTRODUCTION

Neurofibromatosis (NF) is a benign tumour of the nerves. There are two forms chiefly neurofibromatosis type 1 (NF-1), better known as Von Recklinghausen disease which occurs due to mutation on Chromosome 17. It is more common form. The 2nd form is associated with a mutation on chromosome 22. Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder caused by mutations of the neurofibromin gene located on the long arm of chromosome17. Neurofibromin is a tumour suppressor gene, whose mutation leads to development of this disease early in life.[1] It is neuroectodermal in origin with varying involvement of the skin, subcutaneous tissue, and bone.[2] Von Recklinghausen Neurofibromatosis is a form of the disease mostly associated with craniofacial manifestation. It usually presents with numerous clinical features manifested in the soft tissue and skeletal structures.

Soft tissue manifestation

The soft tissue manifestation involves the scalp, periorbital and intraorbital tissues. The commonest cutaneous presentation is the presence of café-au-lait spots. There are also multiple nodular outgrowths over the scalp and face described as a "bag of worm.[3,4]

Skeletal and cranial manifestations

Sphenoid dysplasia is an important feature of Neurofibromatosis.[1,2] Surgical explorations of patients with Neurofibromatosis 1 with skeletal defects didn't o reveal a tumour and a congenital neuroectodermal and mesodermal maldevelopment hypothesis was propounded to explain these bone changes.[3]

Orbital features

The orbital changes in neurofibromatosis type 1 are due to sphenoidal dysplasia, orbital invasion and proliferation of the plexiform neurofibromatosis (PNF). The combination of anterior displacement of the globe due to encroachment by the middle cranial fossa and downward displacement results in enlargement of the orbital rim to produce the classic egg-shaped orbital deformity as described in literature.[3,4]

CASE REPORT

We present a case of a 24 year old female complained of swelling of right facial area since 3 months of age, associated with decreased vision since childhood. The swelling was initially small in size which gradually increased in size, involving the right eye and obscuring the vision along with disfigurement of face (figure 1). Cranial examination revealed right 3rd, 4th and 6th cranial nerve involvement led to restricted eye movement. Right facial nerve was also involved resulted in disfigurement of face.Ophthalmological examination revealed inferior diplopia and exposure keratopathy. MRI Scan was performed which revealed right sided sphenoid dysplasia (figure 2A) with temporal lobe encephalomalacia with meningo-encephalocele.(figure 2B),midline shift (figure 2C) and gliosis (figure 2D). An ill defined T2 flair iso to hyperintense with intense post contrast enhanacement involving the right premaxillary region, frontal, temporal, masticator space along with mass effects s/o plexiform neurofibroma. Histomorphological features

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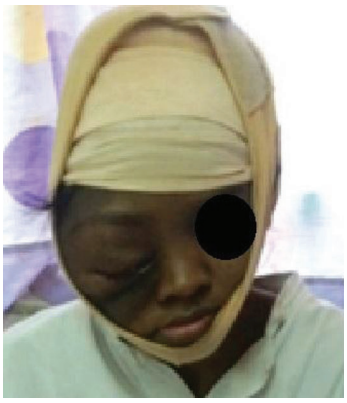

Figure-1: Swelling involving the right eye and obscuring the vision along with disfigurement of face.

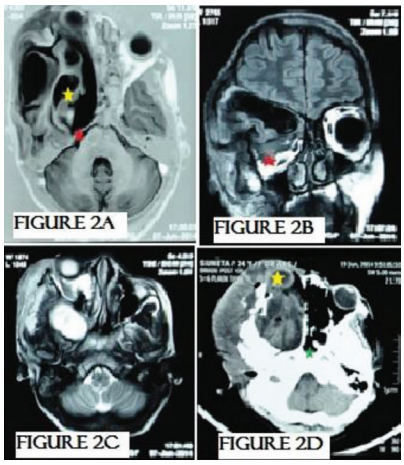

Figure-2A: Hypoplasia of right half of sphenoid, bone, temporal and roof of orbit. **Figure 2B:** Orbital encephalocoele. **Figure 2C:** Midline shift. **Figure 2D:** Gliosis

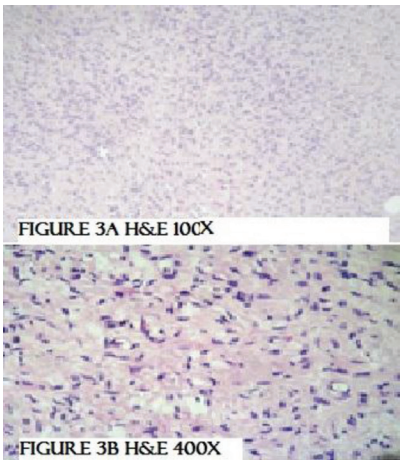

Figure-3 A and B: non encapsulated tumor composed of interlacing bundles of elongated cells with wavy nuclei, fibromyxoid background. Occasional mast cells seen

suggest non encapsulated tumor composed of interlacing bundles of elongated cells with wavy nuclei, fibromyxoid background. Occasional mast cells seen. (Figure 3 A and B)

DISCUSSION

Current neuroimaging has dramatically improved the ability to resolve the soft-tissue and bone changes due to neurofibroma.[4] Radiologic and clinical reports have shown that neurofibromas induce the craniofacial manifestations which might be caused by contiguous tumour as decribed in literature[5,6] Macfarlane et al[7] studied young patient of neurofibroma with craniofacial manifestations and proposed that abnormalities of sphenoid dysplasia associated with NF1 either occur due to abnormal bone formation or secondary to the presence of adjacent tumour.[8] Redistribution of the forces and abnormal CSF flows might lead to development of craniofacial manifestation in patients with NF1 without contiguous tumor. Anteroposterior middle cranial fossa enlargement and anterior displacement of the greater sphenoid wing are central to the concept of sphenoid dysplasia.

CONCLUSION

Neurofibromas present early in life. It is believed that they interact with the skull, orbital structures, and globe while they are still in the developmental stage. The tumours affect nearby bone and invade the orbital structures. Therefore, the term secondary dysplasia is suitable for describing the craniofacial manifestations. Early diagnosis can have a better prognostic outcome. Intensive neurosurgical and ophthalmological workup for improving patient's outcome.

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