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Case report

Uniocular Proptosis: A Presenting Feature of Fibrous Dysplasia

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ABSTRACT

Aim – To present a rare case of craniofacial fibrous dysplasia presenting with proptosis and loss of vision. Methods -- A 27 year old male patient presented with uniocular proptosis of the left eye of 5 months duration with progressive deterioration of vision. On examination of left eye there was 3mm of non-axial proptosis. Pupils showed an afferent pupillary defect. Visual acuity was reduced to hand movements close to face. Extraocular movements showed moderate restriction of adduction. Fundus examination showed optic atrophy. Examination of right eye was normal. CT and MRI scans were suggestive of polyostotic craniofacial dysplasia involving sphenoid and ethmoid bones. This diagnosis was confirmed by histopathological examination. Results – The patient was taken up for surgery which consisted of craniotomy, tumor decompression, excision of dysplastic bone and anterior cranial fossa base repair. Following surgery patient had significant reduction of proptosis. Conclusion – Even though rare, craniofacial fibrous dysplasia should always be considered as an important differential diagnosis of unilateral proptosis in young adults. Early diagnosis and appropriate management is very important due to the significant ocular morbidity associated with this condition.

1. Introduction

Fibrous dysplasia was first reliably recognized in 1891 by von Reckling-hausen who described it as a disorder of bone characterized by fibrotic changes and deformity. They used the term "Osteitis fibrosa generalisata" to denote this condition. Later in 1938 Lichtenstein and Jaffe proposed the term "fibrous dysplasia" as the preferred nomenclature for this characteristic disorder of bone [1].

Fibrous dysplasia is a benign, slowly progressive disorder of bone, where normal cancellous bone is replaced by fibrous tissue and immature woven bone. It is a relatively uncommon, nonfamilial congenital disorder of bone that is usually manifested before the third decade of life. There is no sex preference [2]. Fibrous dysplasia comprises approximately 2.5% of all bone tumors and nearly 7.5% of benign bone neoplasms [1]. Clinically FD is divided into three groups (a) Monostotic: single bone involvement, which is most common (70%) (b) Polyostotic: multiple bone involvement, a less common form (30%)

(c) McCune-Albright syndrome (MAS), a rare variant of mono/polyostotic disease in which FD is associated with café-au-lait macules and/or endocrinopathies [3]. In 50% to 100% of patients with the polyostotic form and in 10% of monostotic variant significant craniofacial involvement is present [2]. There is a predilection for involvement of the frontal, sphenoid, ethmoid, and maxillary bone complexes [4].

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The aetiology remains obscure, but the condition is widely believed to be an anomaly of bone forming mesenchyme [5]. It appears to arise from a perturbation in the mesenchymal precursor of bone, producing a defect in osteoblastic differentiation and subsequent maturation of bone i.e, a defect in cells of the osteogenic lineage. Histologically, fibrous dysplasia appears as a non-neoplastic tissue mass in which multiple small and irregular spicules of immature bone are abundant i.e. "woven bone". The woven bone is superimposed on a background of moderately cellular fibrous connective tissue.

Fibrous dysplasia usually presents with bone pains, deformities, recurrent fractures of the affected site and is sometimes associated with endocrine hyperfunction [3].

Ocular complications have been classified into primary and secondary processes. Primary complications include involvement of the frontal bone with proptosis; the skull base with extraocular muscle palsies and trigeminal neuralgia; the optic canal with visual loss and optic atrophy; the sphenoid bones with chiasmal compression and the maxillary bone with epiphora. Visual impairments may present as perturbances in color vision, central and/or peripheral field defects and afferent pupillary defects. Secondary complications comprise malignant changes, ossifying fibroma formation and development of a mucocele. Malignant transformation most commonly takes the form of sarcomatous change occurring spontaneously in 0.5% of cases [5].

2.Case Report

A 27 year old male patient presented with uniocular proptosis of the left eye of 5 months duration with progressive deterioration of vision. The proptosis was sudden in onset, gradually increasing in size and painless in nature. There was no history of diplopia, cranial nerve deficits, thyroid dysfunction, seizures, headache, and sensory/motor deficits.

On examination of the left eye there was 3mm of proptosis, which was non-axial (inferotemporally) non-reducible, non-pulsatile, non-tender and had no thrill and no bruit. Pupils showed an afferent pupillary defect. Rest of anterior segment was normal. Visual acuity was reduced to hand movements close to face. Fundus examination showed secondary optic atrophy. Extra ocular movements showed moderate restriction of adduction.

Ocular examination of the right eye was normal with visual acuity of 6/6 with normal colour vision and full fields. General physical examination was normal; in particular, there was no clinical evidence of thyroid disease, nor any abnormal skin pigmentation or bony deformity. A clinical diagnosis of unilateral proptosis with optic atrophy due to a space occupying lesion was made and further radiological investigation was advised.

Radiological investigations (CT and MRI) showed well defined hyperdense lesion with ground glass matrix involving sphenoid and posterior ethmoid sinus and causing expansion of sphenoid bone with intracranial and left intraorbital extension. The lesion was causing bowing of medial wall of left orbit and mass effect over left medial rectus and left optic nerve [Figure1,2]. Above findings were suggestive of Polyostotic fibrous dysplasia.

The patient underwent a planned surgical procedure consisting of pterional craniotomy, tumour decompression, excision of dysplastic bone and anterior cranial fossa base repair. Histopathological examination confirmed the diagnosis.

After surgery, reduction in the degree of proptosis was noted. A 4 months follow up CT showed minimal residual tumour without progression. There was no significant improvement in visual acuity.

Figure 1: Contrast MRI scan (axial section) of the brain showing lesion arising from sphenoid and ethmoidal sinuses involving the left orbit causing mass effect over the optic nerve.

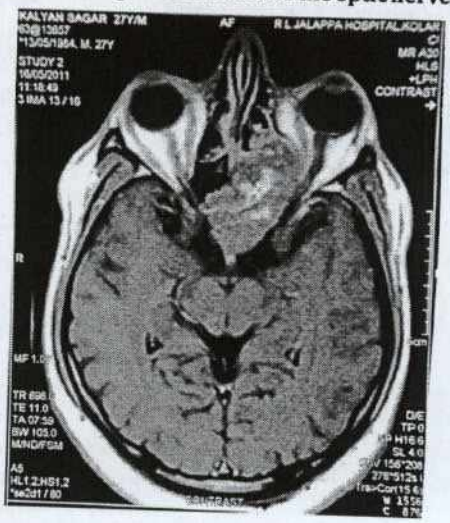
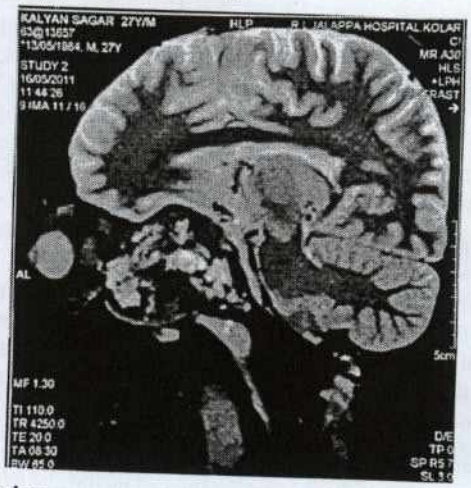


Figure 2: Sagittal section of the lesion showing lesion in the retrobulbar space of the left eye



3.Discussion

Fibrous dysplasia results from a defect in osteoblastic differentiation affecting the final maturation of the bone [6]. Although described as a non-familial, congenital disorder of the bone, it usually manifests before the 3rd decade of life [7]. Our case falls within the age group described in the literature. However there have been reports of presentation as late as 5th decade of life [8]. In a study of 12 cases age of presentation ranged between 5 - 45years of age with a mean age of 18years [5].

Fibrous dysplasia patients typically present with slowly progressive painless proptosis variably associated with nonaxial globe displacement which was similar to our case. There have also been case reports about atypical presentation where patient was aged 41 years and presented with a short history of rapidly increasing proptosis, a mass lesion, globe displacement and pain typical of an infiltrative process [8].

Although visual compromise is less common, both acute and chronic visual loss are well documented. Acute visual loss has been reported in association with mucoceles, hemorrhage and hemorrhagic cysts as well as with fibrous dysplasia alone when it involves the region of the anterior skull base and optic canal. For chronic visual loss several mechanisms have been proposed. Most commonly, this phenomenon is thought to occur secondary to compression of the optic nerve within a stenotic optic foramen. Another mechanism that has been suggested is displacement or distortion of the globe with traction on the optic nerve. External compression by a cystic lesion, separate from stenosis of the optic canal, has also been recognized. Finally, rare vascular events have also been reported to result in blindness [9].

The rapid worsening of visual acuity as described in this case could have been be as a result of cyst formation within the tumour, with resultant compression of the optic nerve. This was supported by the MRI findings.

Although it's a recognized fact that fibrous dysplasia may involve the bones of the optic canal, producing optic canal stenosis, the relationship of this stenosis to visual loss has not been proven. Currently high-resolution CT scans allow a more precise evaluation of the optic canal anatomy and the cause for visual loss in these cases [9].

Establishing the diagnosis of fibrous dysplasia requires close cooperation between a clinician, radiologist and pathologist. Orbital osteoma which is the most common benign tumour of the paranasal sinuses may at times present a diagnostic challenge [10].

Management depends on the course of tumour and the development of complications. This could range from mere observation with serial radiological follow-up to medical therapy with systemic corticosteroids and surgical intervention. A patient with preserved vision and acceptable or minimal cosmetic deformity may be a candidate for serial clinical and radiological observation. Medical therapy has not been clearly proven to cure or impede the progression of fibrous dysplasia. Medications such as bisphosphonates, calcitonin and mithramycin targeted at reducing bone resorption/osteoclastic activity have been prescribed based on the premise that increased osteoclastic activity is seen with fibrous dysplasia. Preliminary data appear promising although inconclusive. Surgical intervention may be indicated when unacceptable cosmetic deformity is present or significant, acute and/or progressive visual impairment is documented by serial examinations. The basic tenets of operative intervention include the preservation of neurological and visual function and correction of cosmetic deformity. Radiotherapy in fibrous dysplasia has not proven to be an effective treatment modality as there is a 44% incidence of malignant transformation reported following radiotherapy [1].

Complete resection of the lesion was not possible in this case because the supero-medial orbit was filled with the lesion and tumor approachability was restricted due to anatomical challenges. We concentrated on curettage to provide enough room for repositioning of the globe using step-wise craniotomy along with anterior cranial fossa base repair for tumor decompression. This approach was associated with less morbidity and quick recovery.

Conservative surgery was previously regarded as the treatment of choice. However, the recurrence rate was 50%-90% as documented in various studies [5].

4.Conclusion

Fibrous dysplasia of the orbit causing neuro-ophthalmic complications associated with compressive mass effect should be considered as a differential diagnosis of slowly progressive proptosis in young adults. Even though fibrous dysplasia is a benign condition an early diagnosis and appropriate management is of utmost importance due to the significant ocular morbidity associated with this condition. The principles of management should include preservation of visual acuity and neurological function along with correction of the cosmetic deformity.

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