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Fibrous dysplasia of Faciomaxillary region case reports and review of literature

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Abstract:
This article discusses the author's experience in managing fibrous dysplasia of faciomaxillary region. Data was accumulated from 2005 – 2011. All these cases were managed by the author. Commonest bone involved by fibrous dysplasia was maxilla (Literature search did not reveal any female preponderance in monostotic fibrous dysplasia[15])In the author's series all of them were females. All of them were monostotic fibrous dysplasia.

Introduction:
Fibrous dysplasia is a benign slow growing fibro-osseous disease characterised by replacement of normal bone with varying degrees of fibrous tissue / immature woven bone. This was first described by Lichtenstein in 1938[1] who called it "*perverted activity of bone forming mesenchyme"*.

Classification of fibrous dysplasia[2] :

1. Monostotic variety – Involving single bone
2. Polyostotic variety – Involving multiple bones
3. McCune-Albright syndrome – Characterised by polyostotic fibrous dysplasia in assocaition with hyperfunctional endocrinopathies (precocious puberty, hyperthyroidism, or acromegaly) and cafe au lait spots involving the skin.

Epidemiology:

Classic epidemiological features of this disorder include:

1. It accounts for about 10% of all bony tumors[4]
2. Monostotic variety is ten times more prevalent than polyostotic variety
3. Majority of monostotic fibrous dysplasia manifest during the first three decades of life
4. Polyostotic fibrous dysplasia presents early during childhood
5. McCune-Albright syndrome also manifests during early childhood.

6. Among facial bones commonest to be involved is ethmoid, followed by frontal and maxilla[3]

At present involvement of skull base is the most common feature when fibrous dysplasia involves craniofacial skeleton[5].

Pathophysiology:

Intramedullary lesions grossly appear as well cirucumscribed tumors varying greatly in size. Large intramedullary lesions tend to cause bone expansion and distortion.
Microscopic appearance: These lesions appear as irregular trabeculae of woven immature bone. They resemble chinese characters. These lesions are surrounded by normal bone and covered byr cellular fibrous stroma with osteoblast progenitor cells closely resembling fibroblasts.
In fibrous dysplasia bone marrow stromal cell differentiation is arrested. This leads to proliferation of immature cells causing the characteristic fibrous dysplasia.[6]

The exact molecular etiology causing this problem has been identified as somatic missense mutation involving GNAS 1 gene on chromosome 20.[7] This gene is responsible for encoding ά subunit of stimulatory G protein coupled receptor Gs ά. In fibrous dysplasia cells arginine is replaced by either cysteine / histidine. This results in inhibition of intrinsic GTPase activity of Gs ά protein. This causes independent activation and accumulation of Cyclic AMP. In bone this causes an effect similar to that of continuous parathormone stimulation. Growth of fibrous dysplasia is enhanced in the presence of growth stimulating hormones. Growth rate of fibrous dysplasia decreases with age as the growth stimualting hormone levels decrease.[8]

Clinical features of fibrous dysplasia:

Clinical features are largely dependent on compression of adjacent structures.

1. Asymptomatic painless bony enlargement causing deformity and asymmetry[9]
2. Headahce
3. Blurring of vision [10]
4. Visual field defects [10]
5. Diplopia [10]
6. Hearing loss
7. Epiphora
8. Eyelid position abnormalities

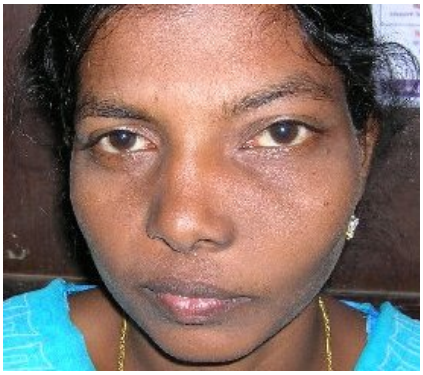


Image showing fibrous dysplasia of right maxilla showing asymmetry of face [11]

Non skeletal manifestations of Fibrous dysplasia:[11]

1. Abnormal cutaneous pigmentation (Jagged “coast of Maine” border
2. Precocious puberty
3. Hyperthyroidism
4. Cushing's disease
5. Hyperparathyroidism
6. Hypophosphatemic rickets

Albright syndrome:

Also known as McCune-Albright syndrome is characterised by:

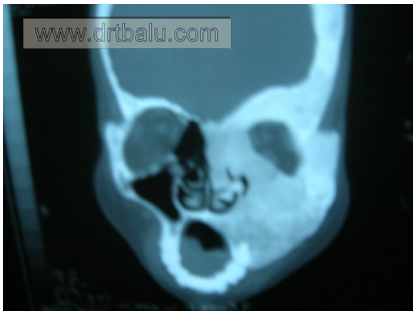
1. Precocious puberty
2. Polyostotic fibrous dysplasia
3. Cutaneous pigmentation

Important feature of this syndrome is that precocious puberty is seen affecting only females where as other endocrine anamolies tend to affect both males and females rather equally. Almost all these disorders have been known to be caused by the same underlying mutation.

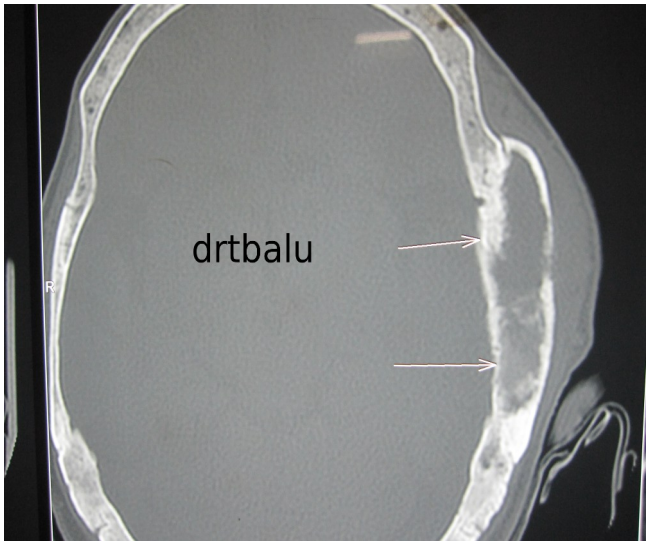
Role of radiology in the diagnosis of fibrous dysplasia:
Majority of fibrous dysplasia are asymptomatic but for the asymmetry caused by the lesion. Since these tumors are very slow growing ones, it could take a long time before the effects of neurovascular compression begin to take place.
Radiological diagnosis alone would suffice for all patients with monostotic fibrous dysplasia. But polyostotic fibrous dysplastic lesions should always be biopsied for histopathological correlation.[12]

CT appearance of fibrous dysplasia:

Commonly the lesion appears like ground glass – 50-60%
Homogenous dense pattern – seen in 20-30%
Cystic pattern – Seen in 10-20% of cases

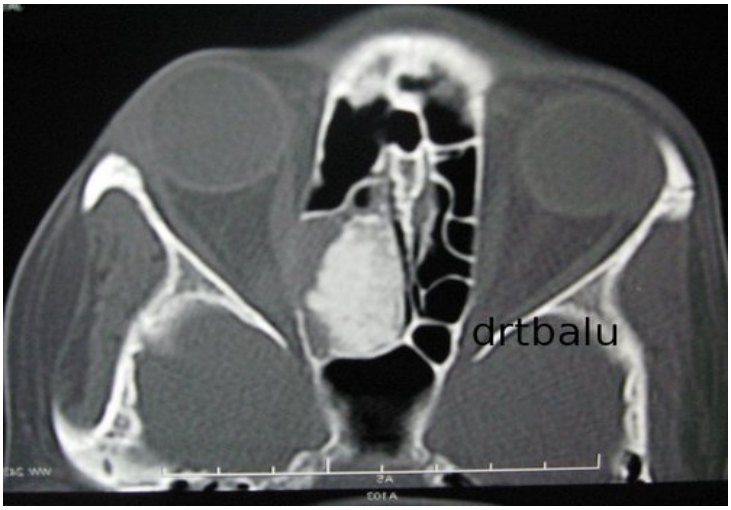


CT coronal cut of nose and paranasal sinuses showing fibrous dysplasia of left maxilla. This is the classic homogenous dense pattern



CT scan showing fibrous dysplasia of zygoma

This lesion is confined to the interior of the maxilla without any soft tissue component. This feature is very useful in distinguishing this condition from malignancy. Malignant lesions involving maxilla is always associated with osteolysis, bone destruction with extension into surrounding soft tissues.[11]
CT scan is very efficient in assessing cranial nerve / optic nerve compression.



Axial CT showing fibrous dysplasia involving poterior ethmoid cells (ground glass type) on the right side. Note the lesion is seen compressing optic nerve

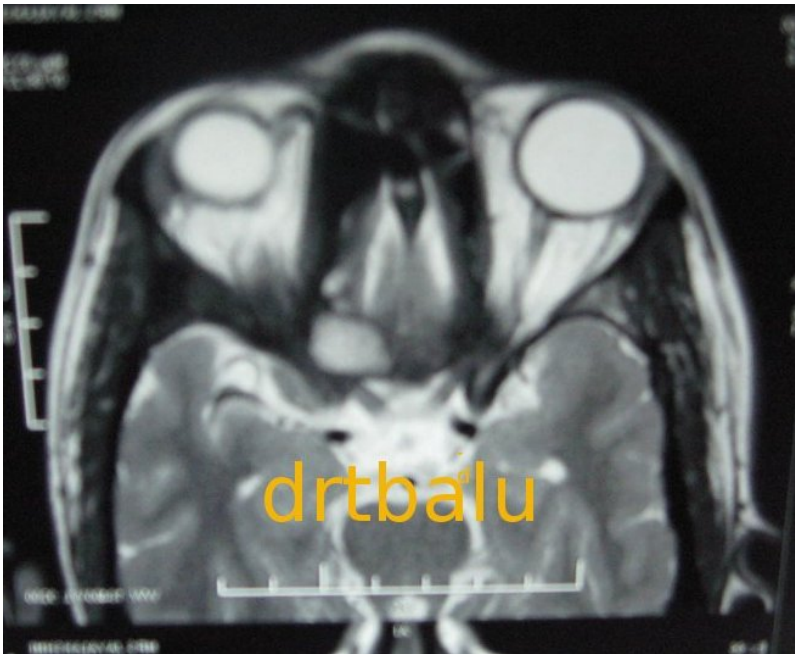
In studies involving skull base three dimensional reconstitution of Helical CT images will help in visualizing fibrous dysplasia involving skull base.

MRI:

In MRI the signal intensity on T1 and T2 weighted images depends on:

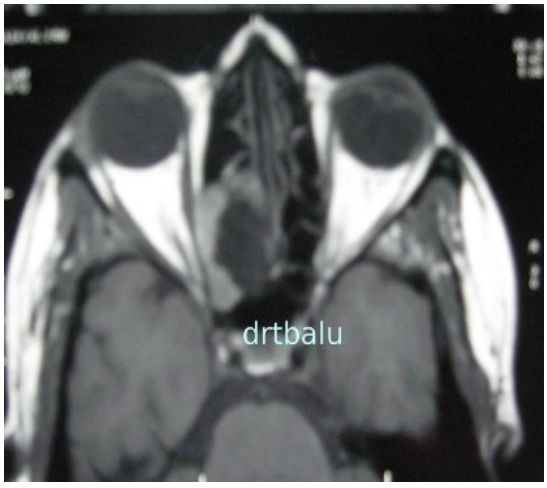
1. Amount of bone trabeculation
2. Degree of cellularity

Characteristically MRI images show low signal intensity with well defined borders both in T1 and T2 weighted images. This reduction in signal intensity is due to the presence of numerous bony trabeculae. Replacement of normal bone marrow tissue by fibrocellular tissue is responsible for signal enhancement in MRI studies[13].

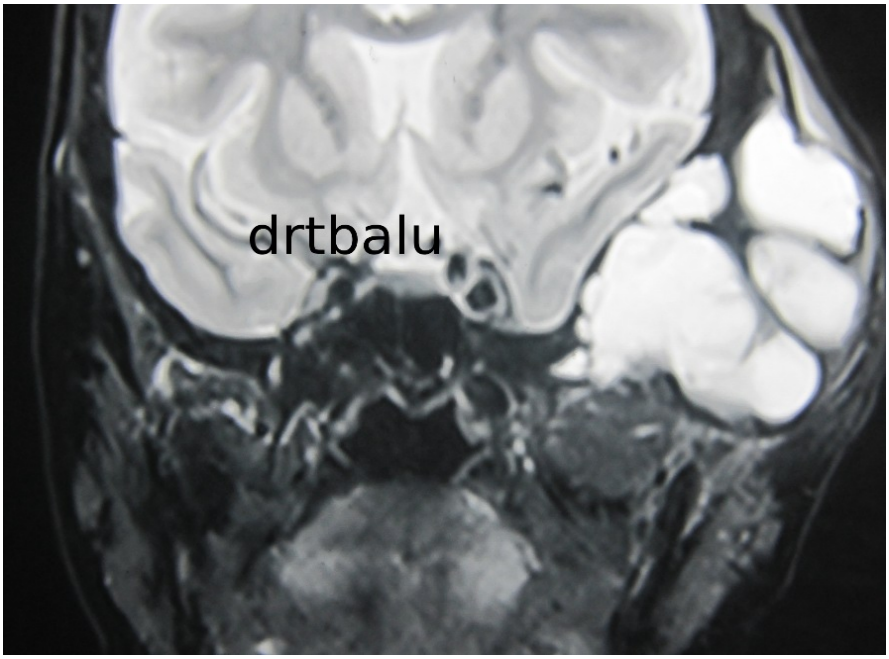


MRI T1 weighted scan showing intense lesion involving posterior ethmoid cells impining on right optic nerve

The best imaging modality which can detect the boundaries of fibrous dysplasia involvementis single photon emission CT scan [14].



MRI T1 weighted showing fibrous dysplasia involving posterior ethmoid on the right side. The mass appears to have low signal intensity in the middle surrounded by peripheral enhancement. This enhancement has been attributed to the presence of fibrous tissue which has replaced the normal bone architecture in this patient.



MRI T1 weighted image showing hyperintense mass involving left zygoma. (Fibrous dysplasia of zygoma)

Inclusion criteria:
All cases of fibrous dysplasia diagnosed radiologically / Histopathologically during the period 2005 – 2010 were included in this study.

Total number of cases – 15

Maxilla – 10
Mandible – 3
Posterior ethmoid / sphenoid – 1
Zygoma – 1

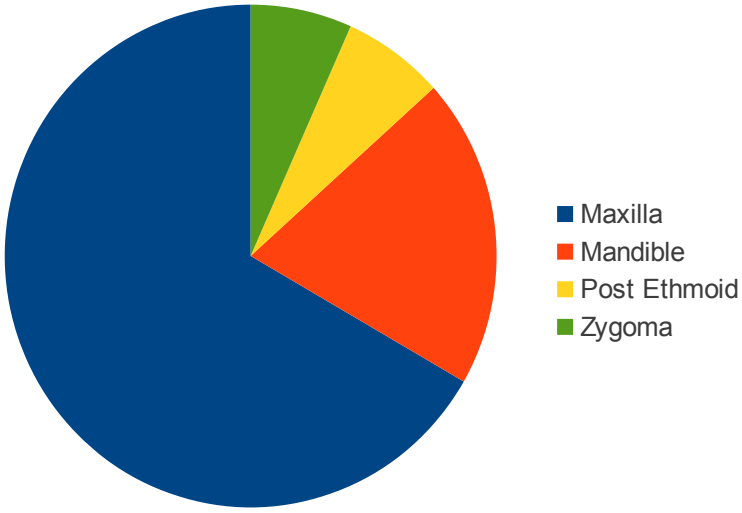
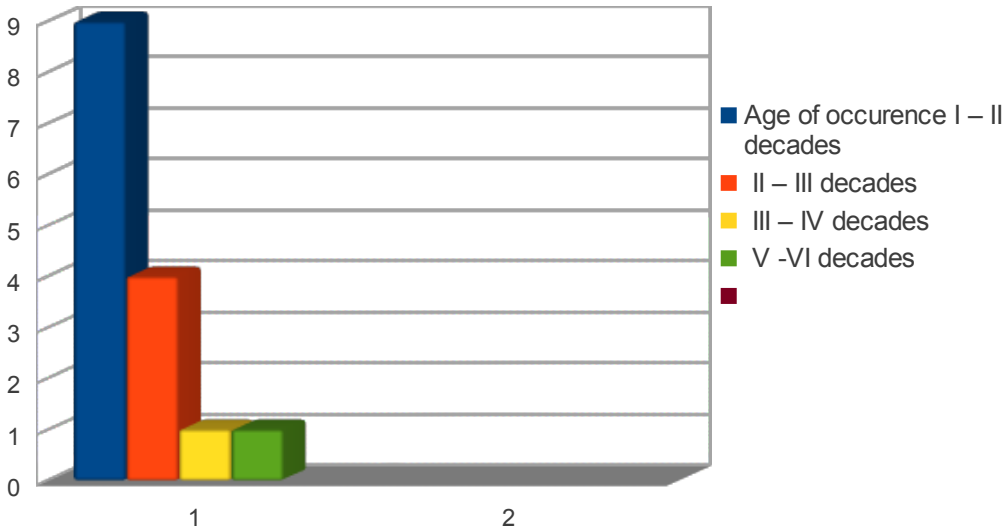


Chart showing the distribution pattern of fibrous dysplasia in various craniofacial bones among 15 cases taken up for study.

Curiously all the patients presented were females. Literature search reveals female preponderance only to polyostotic fibrous dysplasia. As far as monostotic fibrous dysplasia no demonstrable female preponderance was reported in any literature[1].



Graph showing age at which these patients presented. Majority of patients i.e. 9 presented during first decade of life. Only one patient presented during the 6th decade of life [16].

Signs & symptoms these patients presented with include:

1. Facial asymmetry – 15
2. Mass lesion – 15
3. Loosening of teeth – 4
4. Pain – 10
5. Tenderness – 12
6. Visual disturbances – 1: This patient had a total loss of vision. Vison loss is commonly caused by progressive compression of venous drainage from the optic nerve causing reduced retinal perfusion [19].
7. Proptosis – 8
8. Epiphora – 1

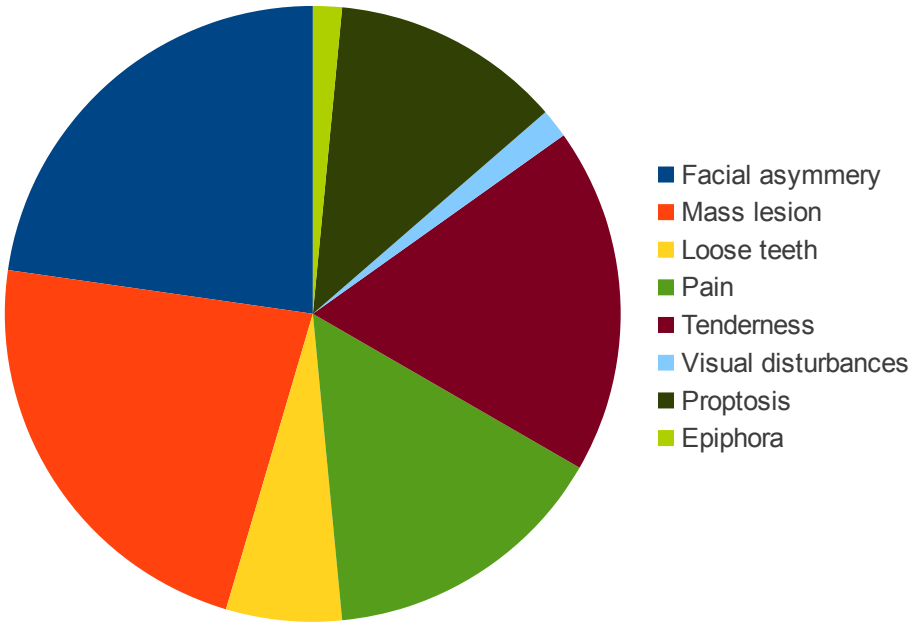
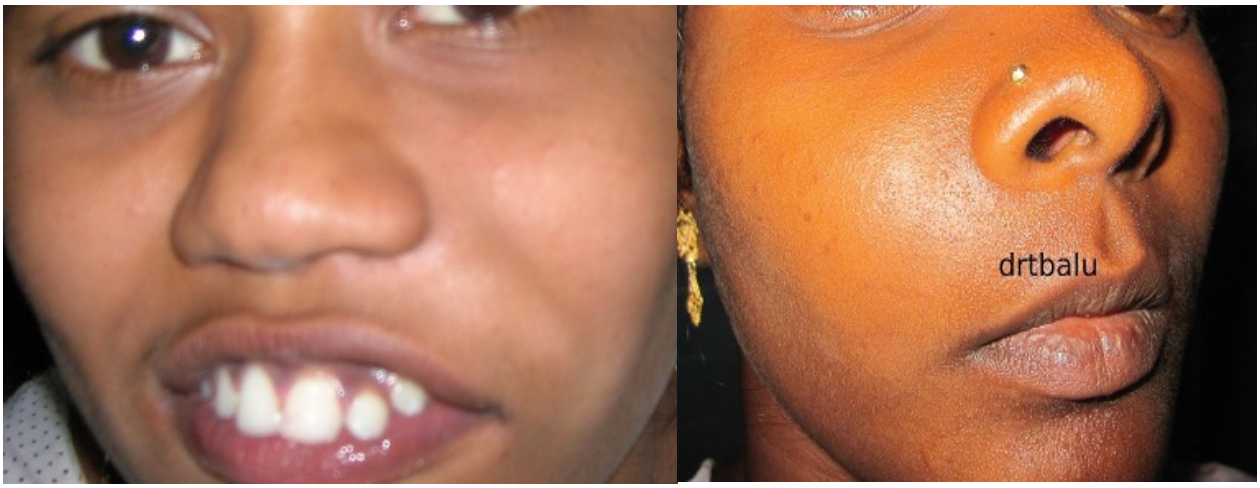


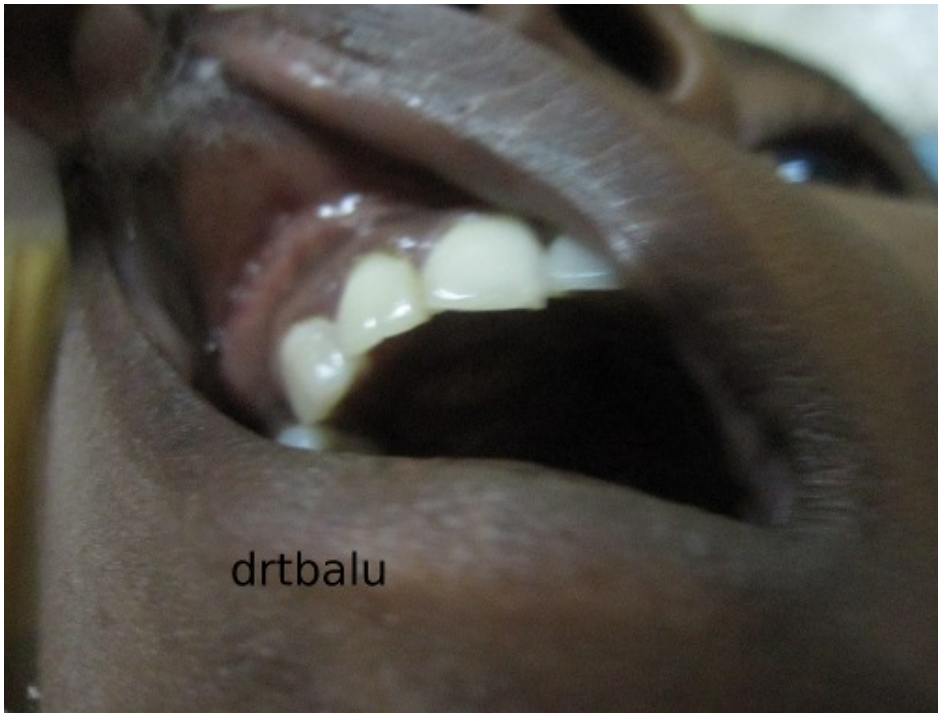
Chart showing the various symptoms patients with craniofacial fibrous dysplasia presented with.

None of these patients were submitted for irradiation because of the fear of sarcomatous malignant transformation [17].



Clinical photographs of patients with fibrous dysplasia of maxilla showing the extent of facial deformity.

Clinical photograph of a patient with fibrous dysplasia of left maxilla with proptosis and upward displacement of left eye.



Fullness and obliteration of bucco gingival sulcus seen in a patient with fibrous dysplasia of maxilla

In this study author was able to observe two histological types of fibrous dysplasia:

1. With increased vascular component. Cyst formation with hemorrhagic spaces lined by fibroblasts and multinucleated giant cells were seen.
2. With reduced vascular component. In these patients there was demonstrable fibroblastic proliferation with mature irregular bone formation.

Patients belonging to category I had brisk bleeding on the table while patients of the second

category provided virtually bloodless field during surgery. Patients with increased vascular components needed bone wax to stop bleeding [19].

Two patients in the authors study belonged to the histologic category I. These patients had brisk bleeding on the table during surgery which needed bone wax occlusion.

Chen & Noordoff classification [20] of craniofacial fibrous dysplasia:

1. Zone I : Fibrous dysplasia involving fronto orbital, zygomatic and upper portions of maxilla
2. Zone II: Fibrous dysplasia involving cranium (in its hair bearing area)
3. Zone III: Fibrous dysplasia involving the central portion of skull base
4. Zone IV: Fibrous dysplasia involving teeth bearing regions of maxilla and mandible.

This classification helps in deciding the management modality. Optimal management modalities suggested by Chen & Noordoff according to their classification include:

Zone I – Total excision
Zones II, III and IV – Conservative excision

In this study 12 cases belonged to zone IV category.
One case belonged to zone III.
Two cases belonged to Zone I category

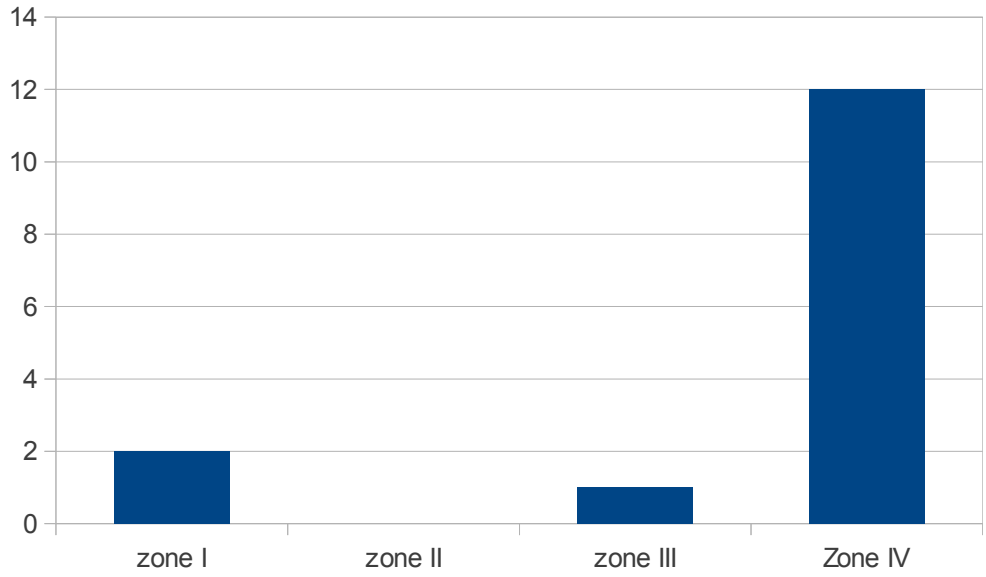
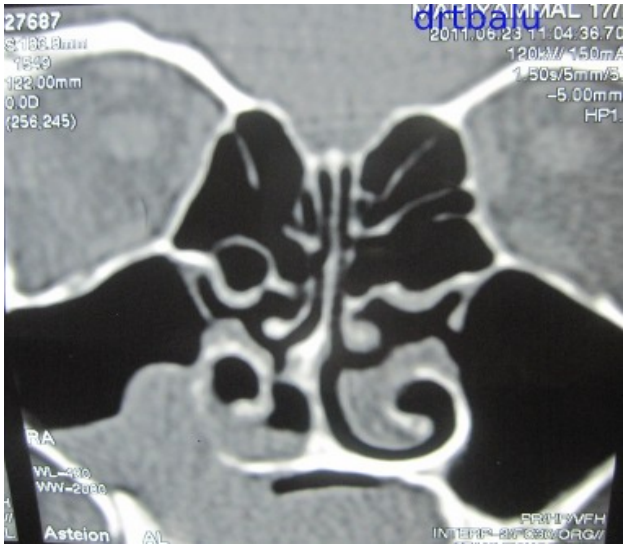
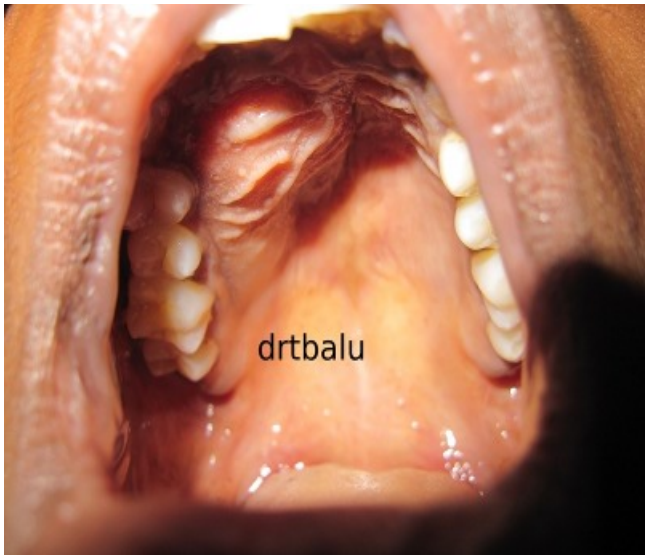


Chart showing the number of patients belonging to various Chen & Noordoff categories





Clinical photograph and CT scan image from a patient with zone IV fibrous dysplasia.

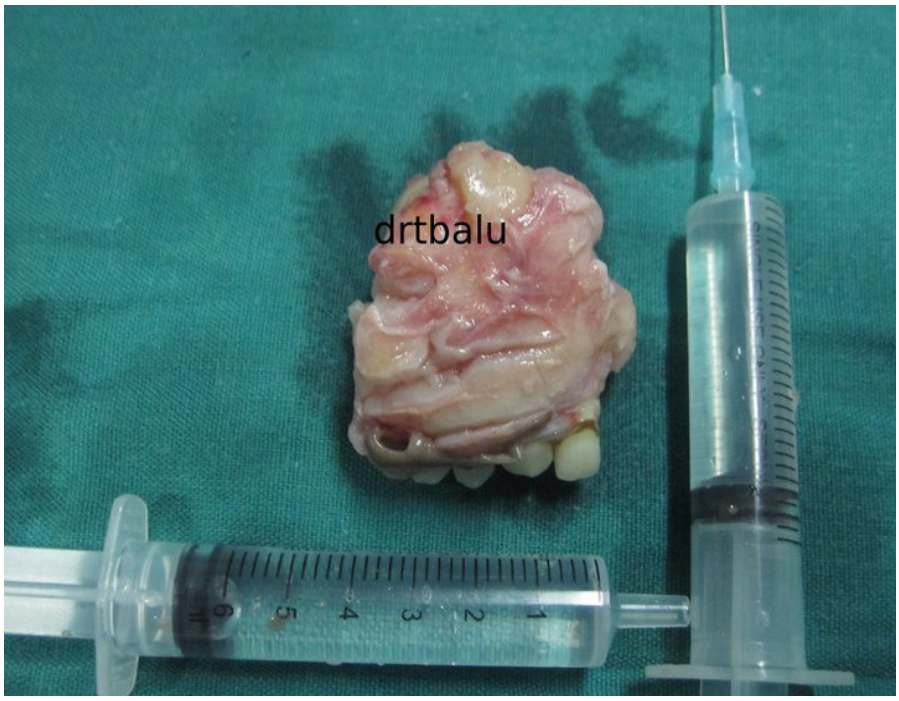
Management:

Review of literature shows a distinct lack of level I evidence regarding medical management of this condition. Attempts at using biphosphonates [21] which could inhibit osteoclastic bone resorption have been successful in varying degrees. Intravenous administration of pamidronate[22] which is a nitrogen containing biphosphonate has been found to be successful when used for a period of more than two years. It reduced pain to a large extent. This drug was initially introduced as a prophylaxis against osteoporosis. It is usually mixed with mannitol and administered intravenously in doses of 90 mg/month. In children this drug can evoke abnormal increase in the bone mass index.
Role of steroids [23] in management of these patients are highly debatable. Current opionion suggests that it can be used in patients with deterioration of vision in order to buy time before definitive decompression surgery could be performed.

Surgery:

Ideally speaking surgery should be deferred till the lesion stabilizes / till the patient grows out of their teens. This wait will ensure less recurrence.

In this study two cases (ZONE I) under went complete resection, and all the other patients underwent curettage. All these patients are under regular follow up since recurrence rate is rather high in them.



Photograph of gross specimen from patient with complete enbloc removal of fibrous dysplasia of maxilla

Discussion:

After review of published literature on this subject in the absence of Level I evidence regarding the optimal treatment modality it has to be said that one has to go by one's own experience. All the patients who reported to this author were initially managed medically with biphosphonates. Two patients showed rapid increase in the bone mass index and hence drugs had to be stopped. Surgery had to be resorted to in all of them because of pain / tenderness / cosmetic disfigurement. All the patients in this study were females and hence pressure on cosmesis was more. One patient with fibrous dysplasia of posterior ethmoids presented with blindness. Steroids was administered in that patient but vision did not improve. One patient in this study presented during 6th decade of her life with fibrous dysplasia of zygoma.

Levels of serum alkaline phosphatase was elevated in 14 out of 15 patients. Serum levels of calcium was found to be raised in 3 patients. They were actually in their early teens.

Conclusion:

This is a genetic non hereditary condition with a benign course in majority of patients. Depending on its location rarely it can cause blindness. Review of literature published reveal that there is no correlation between age and course of the disease. In the absence of Level I evidence regarding the optimal management modality a surgeon is at a dilemma whether to operate or not. Invariably he has to call on his experience to make a decision.

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