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Craniofacial fibrous dysplasia: A case report

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Abstract
Craniofacial fibrous dysplasia is a rare sub-type of fibrous dysplasia which is a congenital, noninherited, benign intramedullary bone lesion in which the normal bone marrow is replaced by abnormal fibro-osseous tissue of the skull and facial bones. The clinical manifestation of cranial asymmetry, facial deformity, fractures or other abnormalities necessitates a comprehensive approach for accurate diagnosis and optimal management. We present the case of a 12-year-old female who presented with one-year history of progressive facial and skull swellings, visual impairment and headache.
General physical examination revealed swelling of the skull, especially the left frontoparietal region. Laboratory tests were unremarkable except for the elevated levels of alkaline phosphatase and phosphorus. A brain computed tomography showed expansile lesions with ground-glass changes in the left frontal bone, parietal bone and clivus.
A diagnosis of fibrous dysplasia was made which was confirmed by Bone biopsy and histology. This case underscores the importance of collaboration between Radiologists, Neurosurgeons and Pathologists in achieving an accurate diagnosis and satisfactory outcome for craniofacial fibrous dysplasias which are rare tumours.

Keywords: Craniofacial fibrous dysplasia, fibro-osseous tissue, skull and facial bones, computed tomography, bone biopsy

Introduction

Craniofacial fibrous dysplasia is a rare type of fibrous dysplasia and is characterized, as the name suggests, by involvement of the skull and facial bones [1]. Fibrous dysplasia (FD) is a developmental benign medullary fibro-osseous process characterized by the failure to form mature lamellar bone and arrest as woven bone that can be multifocal [2]. It can affect any bone and occurs in a monostotic form involving only one bone or polyostotic form involving multiple bones [2]. The WHO Classification of soft tissue and bone tumors (5th edition) titled it a benign bony neoplasm [3].
Fibrous dysplasia is uncommon but occurs in children and adults. It can affect all age groups and can be classified into several subtypes as follows:

- Monostotic fibrous dysplasia -the lesion involves a single bone
- Polyostotic fibrous dysplasia - involves multiple, but never all bones
- McCune Albright syndrome -a condition where polyostotic fibrous dysplasia associated with cutaneous pigmentation and precocious puberty
- Mazabraud syndrome - polyostotic fibrous dysplasia associated with intramuscular myxomas
- Cherubism - is familial fibrous dysplasia of the jaw. However, there is a controversy as to whether Cherubism is part of fibrous dysplasia spectrum. Some considered it a separate entity.

The craniofacial skeleton can be involved in any of these disorders in that, 25% in monostotic disease, 50% in polyostotic disease [4]. The most commonly affected bones are the ethmoids, followed by the sphenoid, frontal and maxillary bones. The maxilla is affected twice as often as the mandible [5].
The clinical presentation of craniofacial fibrous dysplasia often involves cranial asymmetry, facial deformity, nasal stuffiness, proptosis and visual impairment/unilateral blindness [1].
This case study contributes to the plethora of cases which constitute the body of literature, by creating awareness of the radiological features and complications while emphasizing the differential considerations and multidisciplinary approach which are necessary for satisfactory patient outcomes.

Case Presentation

Miss A.V, a 12-year-old female who presented to the neurosurgical out-patient clinic with a history of progressive facial and skull swelling, visual impairment and headache of one-year duration.
The onset was insidious without any history of trauma or triggering factors. The symptoms and signs gradually intensified over time within a space of one year.
There is no history of systemic illness or pevious surgerry.
General physical examination revealed swelling of the skull, especially the left frontal and parietal regions.
The patient was not in an obvious painful distress at the time of presentation but claimed to have experienced severe headache at the onset of the skull swelling.
In view of the clinical presentation, an urgent computed tomography (CT) scan was requested for and it showed an area of expansile bony lesion in the left frontal and parietal bones with background mixed sclerotic and lytic changes. There are also background ground-glass changes. Similar changes are found in the left orbital roof. There was also expansion of the clivus with ground-glass changes as well.

The laboratory investigations including inflammatory markers were within normal limits except for alkaline

phosphatase and phosphorus which were elevated. (Table 1).

The histopathological analysis was done by Consultant Pathologists in the Hospital. Histopathological examination of the bone biopsy showed low-cellular dense collageneous matrix with spindle-shaped fibroblasts, pathognomonic for fibrous dysplasia.

Table 1: Showing the laboratory test results with corresponding reference range.

Laboratory tests	Patient's result	Reference range
White blood count	6.9 x 10^3/uL	4.0-11x10^3/uL
Packed cell volume (PCV)	36%	34-44%
Haemoglobin	11.9g/dL	12.0-16.0g/dL
Creatinine	0.7mg/dL	0.5-0.9mg/dL
Blood urea nitrogen	13mg/dL	7.0-20mg/dL
Platelet Count	374 x 103/uL	150-400 x 103/uL
Calcium	8.6mgldL	8.5-10.7mg/Dl
Sodium	140mg/dL	135-145mg/dL
Potassium	3.6mmol/L	3.5-5.0mmol/L
Alkaline phosphatase	150 IU/L	30-120 IU/L
Phosphorus	6 mg/dl	2.5-4.5 mg/dl

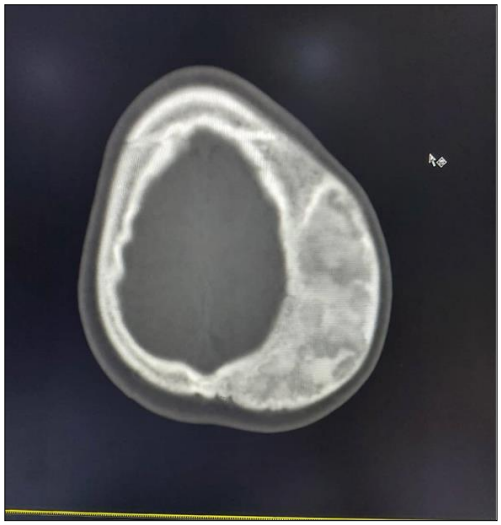

Fig 1: Axial brain Computed tomography (bone window) showing expansion of the left frontal and parietal bones with a ground-glass appearance and background sclerotic and lytic changes

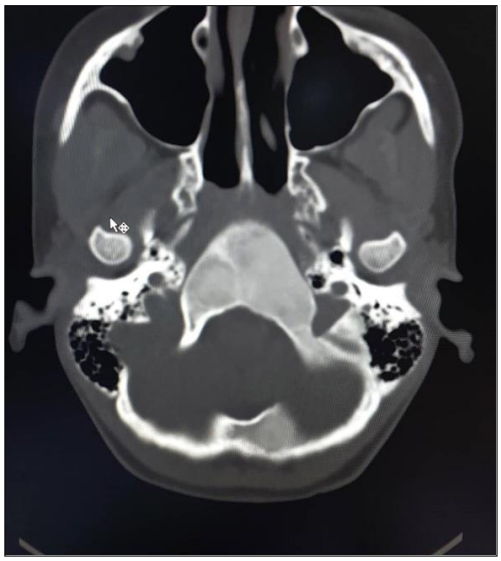

Fig 2: Bone window of axial brain Computed tomography (CT) showing expansion of the clivus with ground-glass changes.

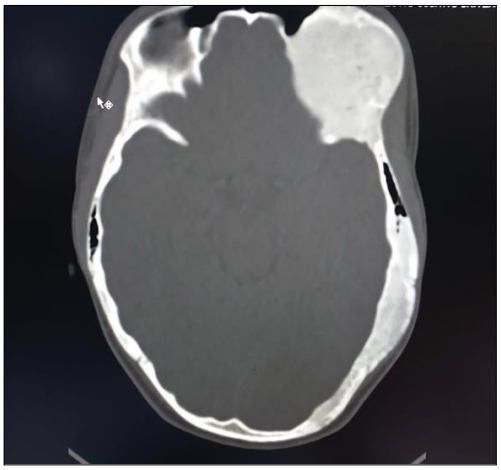

Fig 3: Axial brain Computed tomography (Bone window) showing expansion of the left orbital plate of frontal bone (orbital roof) with background ground-glass changes.

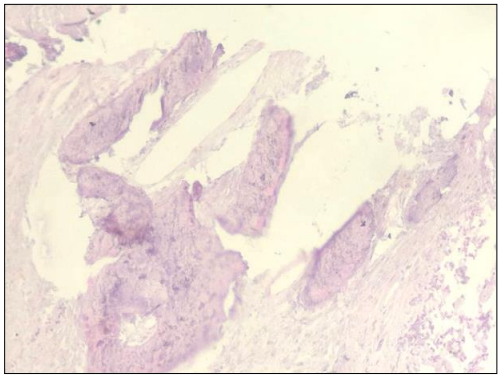

Fig 4: Histopathological image of the bone biopsy showing low-cellular dense collageneous matrix with spindle-shaped fibroblasts.

Management of craniofacial fibrous dysplasia was aimed at establishing the extent of the disease and the maintenance of bone quality via dietary measures and exercise which the patient was encouraged to engage in subsequently. The surgical team was also invited to evaluate the patient and the surgeons expressed satisfaction with the on-going management in the absence of any complication. The outcome was favourable evidenced by reduction in size of the skull swelling and disappearance of headache without any obvious complication. No further treatment was required and the patient was discharged.

Discussion

This case of craniofacial fibrous dysplasia involving the left frontal and parietal bones as well as the left orbital roof and the clivus is actually a rare form of Fibrous dysplasia. Abdelkarim *et al* [5], who stated in their study, that the most commonly affected bones are the ethmoids, followed by the sphenoid, frontal and maxillary bones. They also stated that the maxillary bone is affected twice as often as the mandible.

In making accurate diagnosis pathologically, fibrous dysplasia lesions are well-circumscribed, intra-medullary lesions of varying size. Large lesions can distort and expand the affected bone. The lesion is composed of trabeculae of woven bone surrounded by a moderately cellular fibroblastic proliferation. All the components of normal bone are present, but local developmental arrest prevents differentiation into mature structures [6].

In this case, a 64-slice Computed tomography machine played a crucial role is demonstrating the anatomical extent of the bony lesion that did not warrant further characterisation with MRI. Fibrous dysplasia is composed mainly of fibrous tissues and bone, hence, T1 images (MRI) typically have a low-intensity signal while the T2 images are mixed. Most commonly T2-weighted imaging have a higher intensity signal that is not as bright as fluid, however, some lesions can have diffuse low T2 signal. T1 post-contrast images typically show heterogeneous contrast enhancement in the affected areas [7].

Technetium 99m-methyle diphosphonate bone scanning may be used to gauge the extent of the disease at initial presentation. Active fibrous dysplasia lesions in younger patients have greatly increased radiotracer uptake. The uptake becomes less intense as the lesions mature [7].

Differential diagnoses include various pathologies which include cemento-ossifying fibroma, intraosseous meningioma, pagets disease and sclerotic metastases [1]. Differentiation from these pathologies require a comprehensive combination of radiological and pathological investigations.

Conclusion

The index case of craniofacial fibrous dysplasia is actually a rare form of fibrous dysplasia in our environment, which usually occurs predominantly in children. The multidisciplinary approach that was engaged comprised of detailed clinical examination and state of the art radiological imaging underscores the importance of a comprehensive protocol in addressing such rare pathologies. This case, without any doubt, contributes to the growing body of knowledge on fibrous dysplasia, buttressing the importance for ongoing research to enhance diagnostic precision and early management of such cases so as to ensure favourable outcomes.

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