Case Report

Craniofacial Fibrous Dysplasia: A Case Report

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Abstract

***Background**: Fibrous dysplasia (FD) is a rare condition of benign fibro-osseous lesions, which causes considerable deformities and functional disturbances. It is usually seen in the first 3 decades of life, but it is more evident between 5 and 15 years of age. Symptoms at presentation depend on the bone involved. Commonly mandible and maxilla bones. Therefore, Patients with FD may present with facial asymmetry, swelling, proptosis, hearing loss and headaches.*

***Case Description:** A 20-year-old male suffered for one year from painless right forehead swelling that has increased in size, associated with an on/off headache and decrease in vision of the right eye with diplopia on the right gaze, yet, ophthalmology exams were normal. CT brain demonstrated a mixed cystic and solid mass. MRI brain showed significant thickening of the frontal bones. Two years later, the patient came complaining of an increase in the size of the swelling. CT brain repeated and showed new development of four intradiploic bony lytic lesions. The mainstay treatment of CFD is surgical either remodeling or radical excision and reconstruction. As per the patient's request for cosmetic reasons, he underwent bifrontal craniectomy and removal of two aneurysmal cysts, followed by 3D custom-made cranioplasty.*

Conclusion: *It is recommended to monitor the management response, progression and regrowth with serial follow-up, CTs, Serum alkaline phosphatase (ALP), a marker for bone turnover, and urinary hydroxyproline. Despite a wide range of therapeutic options, good outcomes can be achieved with proper understanding, diagnosing, treatment planning and follow-ups.*

Keywords: *Craniofacial, fibrous dysplasia, boney developmental disorder.*

Introduction

Fibrous dysplasia (FD) is a benign bony developmental disorder, as described by Lichtenstein in 1938, characterized by progressive replacement of normal bone elements to disorganized and immature fibrous tissue. **[**1**]** FD results from an activating mutation of the GNAS gene that encodes the α subunit of stimulatory G protein (Gsα) located at 20q13.2-13.3, which results in inhibition of the differentiation and proliferation of osteoblast. **[**2**]** It has an incidence of 1:4000- 1:10,000 and represents about 2.5% of all bone lesions and 5-10% of all benign bone tumors.**[**1,3**]** They are seen in the first 3 decades of life, but they are more evident during the rapid bone growth phase that is considered to be between 5 and 15 years of age.**[**4**]** Craniofacial FD (CFD), one of the 3 subtypes of FD, described the bone lesions that are confined to the craniofacial bones.**[**5**]** CFD is mostly presented as facial asymmetry and swelling, however, in severe cases it can cause functional disturbances of the vision, hearing or breathing depending on the bone involved. **[**6**]** With the recent advances in surgical techniques, complete resection and bone graft becoming an ideal treatment modality.**[**7**]** This is a case report of a 20 years old male with CFD and approach towards management. **[**3**]**

Case Report

A 20-year-old male, with no medical illnesses, presented with a 1year history of painless right forehead swelling that has increased in size. It is associated with an on/off headache and a decrease in vision of the right eye. There was no history of trauma, seizure, neurological deficit, skin pigmentation, swelling elsewhere in the body, sinusitis, or endocrine/pituitary symptoms. Clinically, the patient presented with frontal bone asymmetry and right eye ptosis with the prominence of the right frontal bone area that is firm, not mobile, not tender and no discoloration. The patient had diplopia on the right gaze, nonetheless, the ophthalmology exams were normal. All routine labs were unremarkable. CT brain demonstrated a mixed cystic and solid mass-like lesion noted in the right frontal area anteriorly involving the right frontal sinus with an extraosseous and intracranial extension which seems to be compressing the adjacent CSF space in keeping with fibrous dysplasia with aneurysmal bone cyst [**figure 1**]. These

lesions showed a ring enhancement however it might represent a normal venous flow with no evidence of internal enhancement.

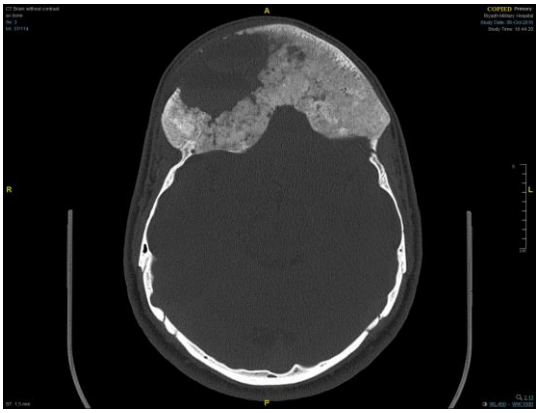

Fig. 1-A

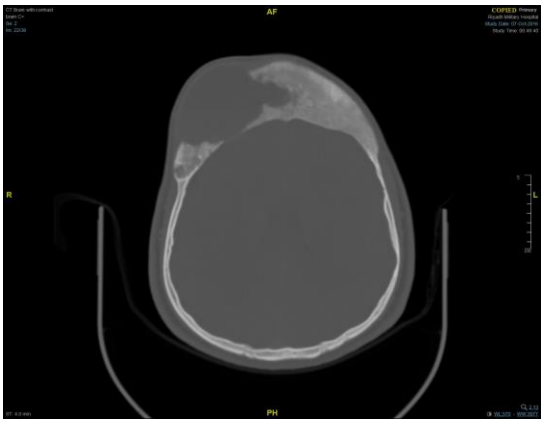

Fig. 1-B

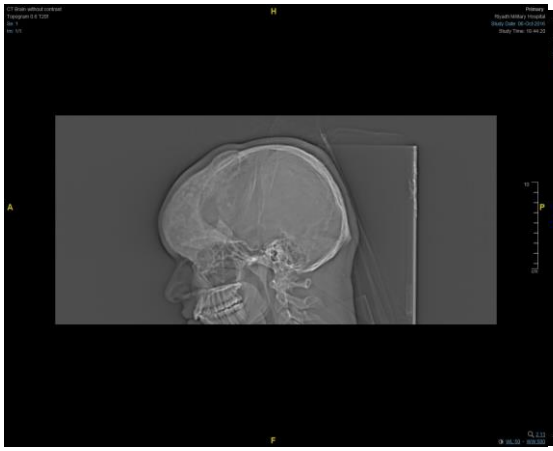

Fig. 1-C

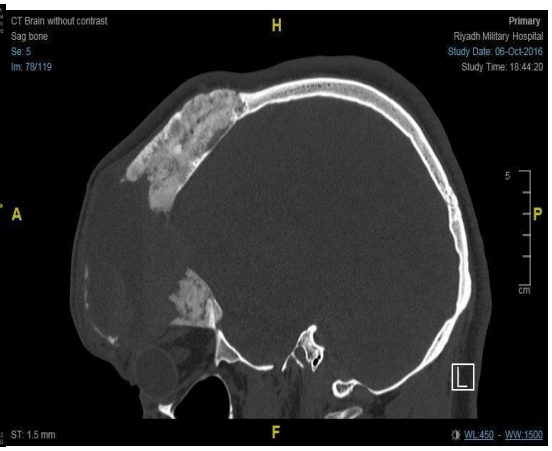

Fig. 1-D

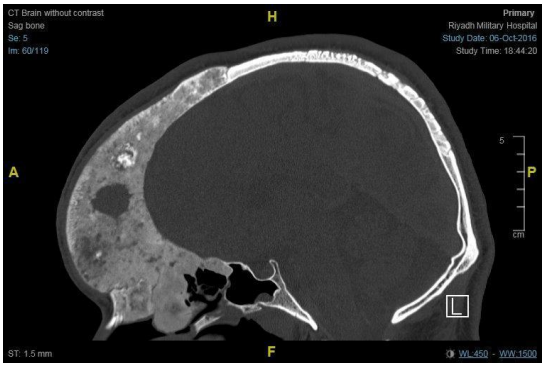

Fig. 1-E

Figure 1-A, 1-B: Axial CT brain show a mixed cystic and solid mass lesion in the right frontal area anteriorly involving the right frontal sinus with an extraosseous and intracranial extension.

Figure 1-C, 1-D, 1-E: Sagittal CT brain demonstrate a mixed cystic and solid mass.

MRI brain showed significant thickening of the frontal bones including the orbital roof, the floor of the anterior cranial fossa, nasal and probably lacrimal bones associated with multiple cysts with the intracystic fluid-fluid level seen at the right side of this enlarged bone, the largest cyst measures 6.0 x 6.0 x 5.8 cm and erode the inner table with subsequent indentation of the dura at the right frontal pole. Erosion of the ipsilateral orbital roof and subsequent deviation of the eye globe inferolateral. Post-contrast images show heterogeneous enhancement of the thickened bone as well as thin rim enhancement of the large cyst [**figure 2**].

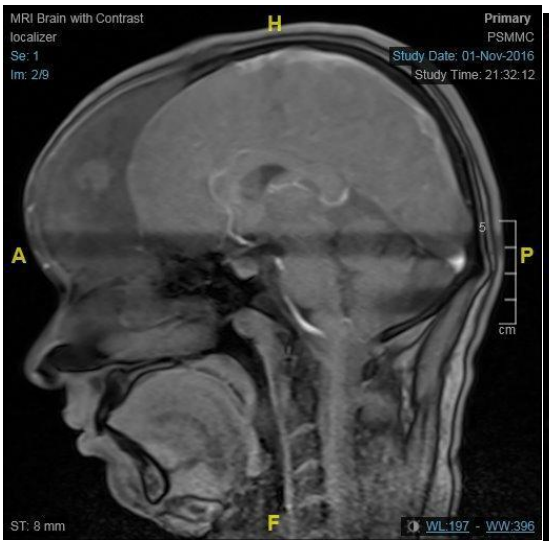

Fig. 2-A

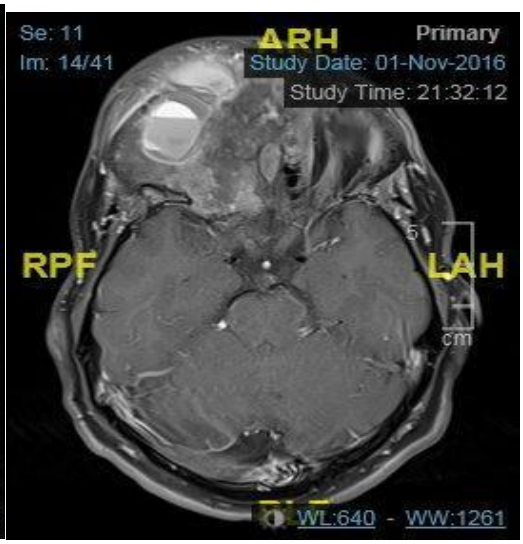

Fig. 2-B

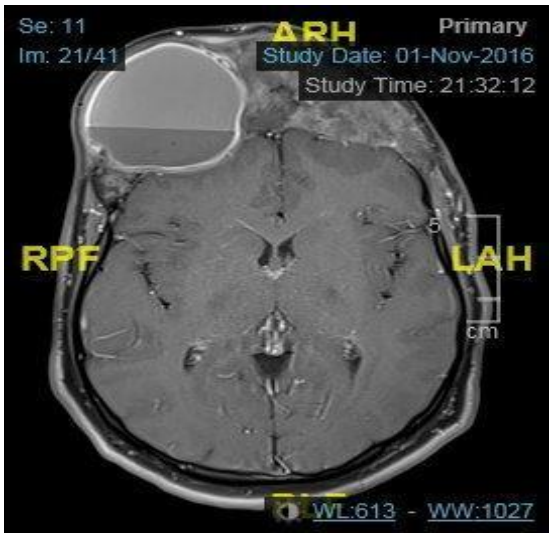

Fig. 2-C

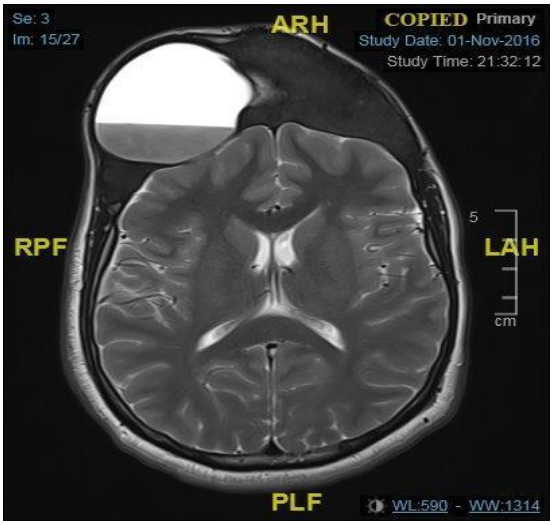

Fig. 2-D

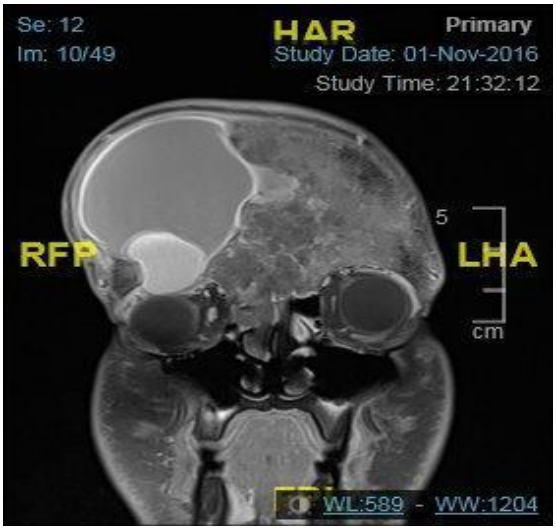

Fig. 2-E

Figure 2-A: Sagittal MRI brain shows significant thickening of the frontal bones including the orbital roof, the floor of the anterior cranial fossa and nasal bones.

Figure 2-B, 2-C, 2-D: Axical MRI brain show thickening of the frontal bones associated with multiple cysts with intracystic fluid level seen at the right side of this enlarged bone.

Figure 2E: Coronal MRI brain shows enlargement of the frontal bone at the right side associated with multiple cysts with intracystic fluid.

2 years later, the patient came to the clinic complaining of an increase in the size of the swelling, and his colleagues were making fun of him. CT brain repeated and showed New development of four intradiploic bony lytic lesions; three are small, the maximum dimension of 8 mm while the fourth is the largest measuring 2 x 2.5 x 6 cm and seen at the right posterior frontal bone limited by the coronal suture. This large lytic lesion causes erosion of the outer table and rarefaction of the inner table. Erosion of the outer table is also seen at one of the small lytic lesions at the midline of the lower frontal bone measuring 7 x 14 mm. As per the patient's request for cosmetic reasons, he underwent bifrontal craniectomy and removal of two aneurysmal cysts, followed by 3D custom-made cranioplasty [**figure 3**].

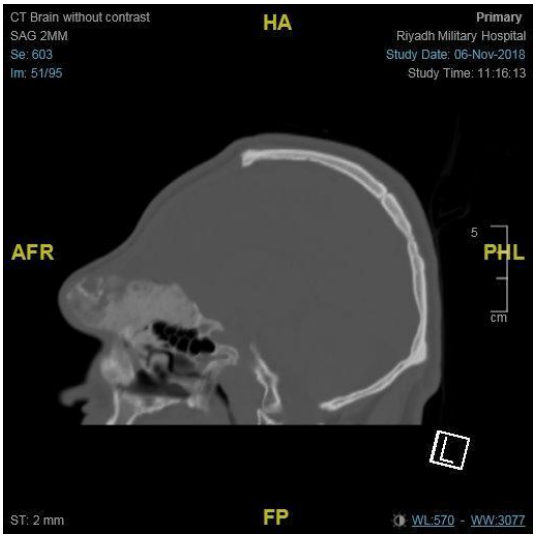

Fig. 3-A

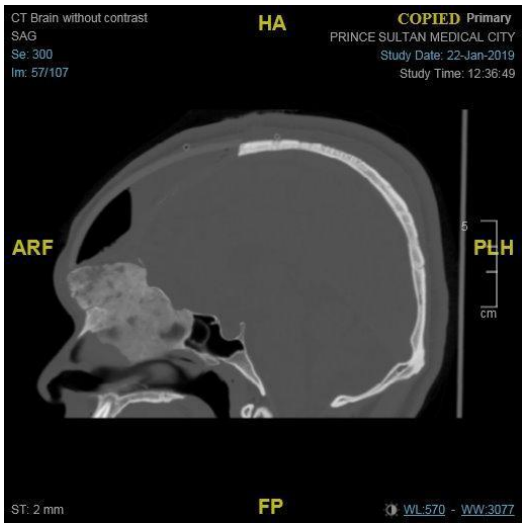

Fig. 3-B

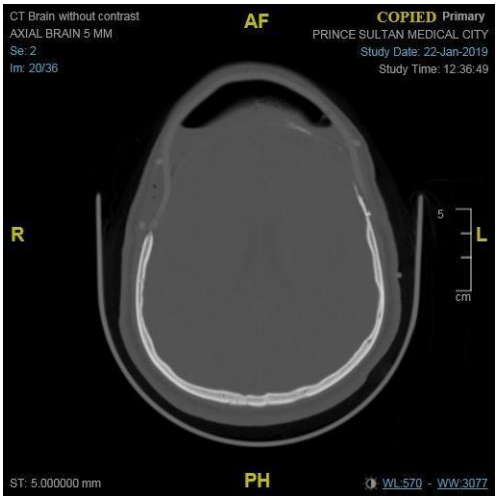

Fig. 3-C

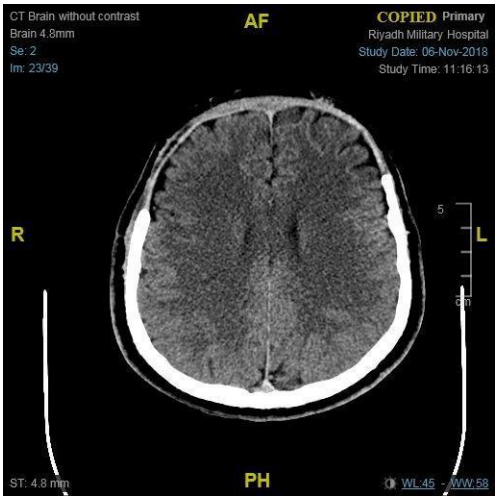

Fig. 3-D

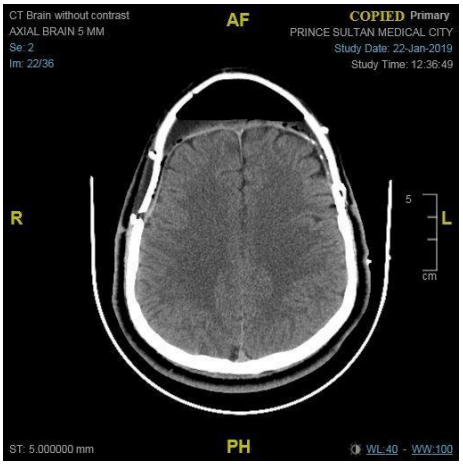

Fig. 3-E

Figure 3-A: Sagittal CT brain demonstrate a bifrontal craniectomy and removal of the two aneurysmal cysts.

Figure 3-B: Sagittal CT brain demonstrate a cranioplasty 3D custom made, after the bifrontal craniectomy and removal of the two aneurysmal cysts.

Figure 3-C, 3-D, 3-E: Axial CT brain of a post cranioplasty showing well reconstruction.

The histopathological report illustrated sections of bone, soft tissues, intracystic nodule (irregular shape, soft, yellow-tan lesion), the lining of the cyst wall (white tan soft tissue) and bony trabecular formation referred to as "Chinese letter" configuration. That gives the conclusion of fibro-osseous lesions consistent with FD with cystic changes [**figure 4**].

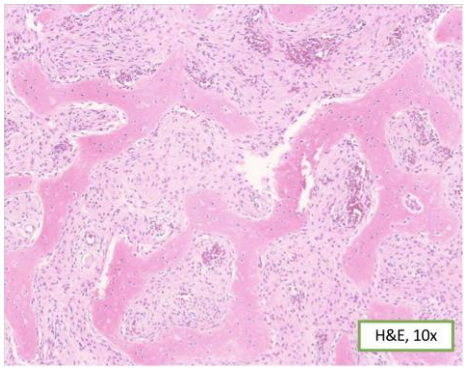

Fig. 4-A

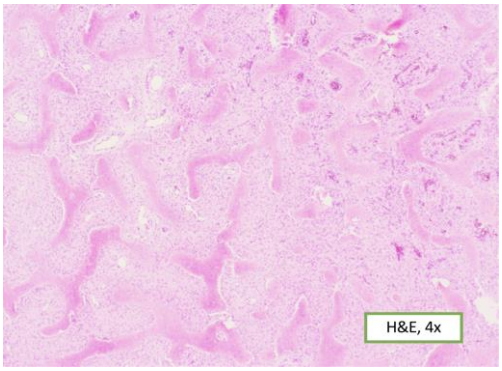

Fig. 4-B

Figure 4-A: At higher magnification power (100x) the bone trabeculae are seen to lack an osteoblastic rim. The stroma in between these particles consists of bland spindly fibroblasts that are devoid of cytological atypia and mitotic figures.

Figure 4-B: At a low magnification, the lesion is composed of bony trabecular forming irregular S and C like shapes, a configuration commonly referred to as "Chinese letter" configuration. This is the manifestation of failure of the woven bone to mature into lammelar bone.

Discussion

FD is a benign bony developmental anomaly characterized by a build-up of immature and disorganized fibrous tissue instead of the normal bone elements within the medullary and cortical bone. Malignant transformation has been reported to occur in 1-4%.**[8]** The condition is related to a postzygotic mutation in the GNAS-1 gene that encodes the subunit of α stimulatory G protein (Gsα) located on chromosome

20.[8] The mutation results in replacing the amino acids cysteine or histidine for arginine of the genomic DNA in the osteoblastic cells.[9] This allows for the activation of adenylate cyclase, overproduction of cyclic adenosine monophosphate (cAMP) and increased cell proliferation and inappropriate cell differentiation. As a consequence, this leads to the development of FD lesions with abnormal bone matrix, trabeculae, and collagen. [10]

The three subtypes of FD are monostotic (Single bone), polyostotic (Multiple body's bones) and craniofacial (Confined to the craniofacial bones).[5] About 3% of polyostotic FD occurs as McCune-Albright syndrome (MAS) with its tried of Polyostotic, endocrine hyperfunction, and café-au-lait macules. [11,8] CFD can have a psychosocial impact on the patient due to its capability of causing considerable facial disfigurement and functional disability.[12] The bones commonly involved are mandible (12%) and maxilla (12%), involvement of the ethmoid, sphenoid, frontal, and temporal bones are infrequent. [13] The most common symptoms are facial asymmetry, swelling, and pain. [14] Depending on the affected bone, patients can also present with difficulty breathing, loss/decrease of vision, dystopia, proptosis, hearing loss, facial paralysis, numbness, and headaches. [15] Cystic mass lesions, including cystic degeneration and other cystic lesions such as aneurysmal bone cysts, are uncommon in FD and can undergo rapid enlargement. It is considered to be one of the causes of optic nerve compression and patients present with sudden vision deteriorations. [1] Visual impairment is an alarming symptom in those patients. [16] In which it can occur quickly, and can become permanent in a short time, systemic steroid use should be considered only as a temporary solution until the patient can undergo optic nerve decompression.23. [15] It has been found that FD has 3 histological categories. The classical form, irregularly shaped trabeculae of immature woven bone in a cellular, loosely arranged fibrous stroma arranged in a pattern referred to as a "Chinese writing pattern". The sclerotic/pagetoid, characterized by dense, sclerotic trabecular bone with complex systems of cement lines, associated with cranial bones. The sclerotic/hypercellular type, discontinuous bony trabeculae distributed in an ordered "often parallel pattern", associated with gnathic and skull base bones.[17,18] Both CT and MRI are excellent imaging modalities in diagnosing CFD and defining the constrictive effect on the orbit, optic canals and adjacent paranasal sinuses.[19] Computed tomography (CT) offering advantages in the evaluation of bone changes and extension with 3 different patterns; the ground-glass pattern is most common (56%) (Consists of admixture of dense (osseous elements) and radiolucent (fibrous components) in the medullary space), sclerotic type (23%) (Characterized as homogeneously dense masses), and the cystic pattern (21%) (Appears as a spherical lucency surrounded by sclerotic margin).[8] On magnetic resonance imaging (MRI), CFD lesions appear low- to iso- intense in T1w and show contrast enhancement. On T2w it appears as a high-intense lesion that is not as bright as the signal of malignant tissue, fat, or fluid.[20] The recommended treatment options of CFD are observation, medical therapy, surgical remodeling, or radical excision and reconstruction.[5] As clinical manifestations are differing from one patient to another, care of these patients must be tailored to their desires, lesion sites and

extension, comprehensive evaluation and multidisciplinary involvement. [8] Observation with periodic follow-ups is used in the small and asymptomatic lesions. Medical therapy includes; steroids for expansile lesions near the optic nerve causing visual symptoms, and bisphosphonates to reduce the pain and rate of growth of the lesion.[13,15,21] Even if medical therapy has a role in the management of symptoms, the mainstay of treatment remains surgery.[22] Among the innovations of radiological imaging and surgical advances, surgery is favored for cosmetics and relieves compressions on vital structures.[15] Surgical protocols of CFD range from conservative shaving/contouring to partial or complete surgical resection with immediate reconstruction.[23] The choice depends on several factors: age, site of involvement, rate of growth, aesthetic disturbance, functional disruption, patient preference, general health of the patient, possible morbidities, surgeon's experience and the availability of a multi-disciplinary team (neurosurgeon, ophthalmologist, maxillofacial surgeon and orthodontist). [1,7,13] The surgical procedure is usually postponed till puberty hoping that the lesion is stabilized as the bones reach maturity.[5] However, surgery is desired when there is a threat to the optic nerve that might lead to visual damage. [16] Chen and Noordho in 1990 divided the head and face into four zones as a treatment algorithm for the management of CFD. They are based on the considerations of anatomical structures, esthetic and functional concerns of the disease for surgery at these sites: The esthetically critical zone (Zone 1), involves fronto-orbito-malar regions of the face, radical excision and reconstruction with simple bone grafting is recommended. Not esthetically critical zone (Zone 2), involves the hair-bearing scalp, the intervention depends on the patient's need. The difficult surgical access zone (Zone 3), involves the central skull base with the sphenoid, pterygoid, petrous temporal bone, and mastoid, observation of lesions is advised. The difficult reconstructing zone (Zone 4), involves the tooth-bearing areas of the skull, maxilla and mandible, conservative management is recommended. [22,24] Once a management is performed, it is recommended to monitor management response, progression and regrowth with serial follow-up, CTs, Serum alkaline phosphatase (ALP), a marker for bone turnover, and urinary hydroxyproline.[7,12]

Conclusion

CFD is a benign boney developmental anomaly that can cause considerable facial deformities and functional disturbances. Good outcomes can be achieved with proper understanding, diagnosing, treatment planning and follow-ups. Both CT and MRI are excellent imaging modalities in diagnosing and visualizing the CFD effect on adjacent structures. As clinical manifestations are differing from one patient to another, the care of these patients must be tailored to their desires. The mainstay of treatment is surgery for cosmetics and relieves compressions on vital structures. Serial follow-ups with CT, serum alkaline phosphatase and urinary hydroxyproline are recommended to monitor treatment response, progression, or reactivation.

Acknowledgments

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