

Craniofacial Fibrous Dysplasia- a Rare Case Report in a Tertiary Eye Hospital

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Abstract

Purpose: Fibrous dysplasia (FD) is a rare fibro-osseous lesion of the osseous structures of the body. Craniofacial FD confined to contiguous bones of the craniofacial skeleton. Purpose of the study is to report such a rare case which was diagnosed and managed in the outpatient department in our tertiary eye hospital. **Method:** A 12-year-old girl complaint of painless swelling over right side of eyebrow for 4 months. Facial examination revealed well-defined bony swelling rounded in shape with hard consistency, nontender; diameter 10x10 mm; overlying skin free; no regional lymphadenopathy. Visual acuity was 6/6. Anterior and posterior segment revealed normal. Ocular motility was full. No other swelling was present in body and café-au-lait spots were absent. Routine investigations such as hemogram, serum calcium and serum alkaline phosphatase (ALP) were performed. Among them ALP was raised to 300 U/L. CT scan showed a radio dense mass with ground-glass appearance involving right frontal bone whose expansion causing facial asymmetry. An incisional biopsy with histopathological analysis was done. It showed irregular bony trabeculae in Chinese script pattern scattered within fibrous stroma. The bone appeared woven rather than lamellar. MRI suggest diffuse thickening of right frontal bone. **Result:** Clinical history, radiographic assessment and histological features suggested that it was a case of craniofacial FD involving right side of frontal bone. **Conclusion:** Detection of early suspicious sign for diagnosis and early initiation for management is crucial to prevent the long-term complication with avert the process of optic nerve compression.

(J.Natl.Inst.Ophthalmol.2022;5(1):46-50)

Introduction

Fibrous dysplasia (FD) is an uncommon nonhereditary, skeletal developmental anomaly where the normal bone was replaced by excessive proliferation of cellular fibrous connective tissue mixed with bony trabeculae¹. FD is a benign disorder which does not spread. It may arise as a

single lesion referred to as monostotic or can occur with multiple lesions known as polyostotic. The disorder is diagnosed earlier in children and young children.² These conditions have a slight female predilection.³ FD of bone evolve from activating missense mutations in Gs alpha gene in pluripotent embryonic stem cells.⁴ Craniofacial FD (CFD) is one of the three types of FD which affects the bones of the craniofacial complex involving cranial base, vault with mandible and maxilla. The bones commonly involved are maxilla (12%) and mandible (12%), involvement of the ethmoid, sphenoid, frontal and temporal bones are infrequent. The affected bones show expansion, thickening and sclerosis. Depending on the involved bone patients may have visual abnormalities, hearing disturbances, facial asymmetry and tooth displacement.⁵ We report a case of CFD in a 12-year-old female patient. The exact incidence and prevalence of the disorder is unknown. Mild cases may go undiagnosed, making it difficult to determine the true frequency of FD in the general population. The monostotic form is more common than the polyostotic form; according to some reports by a ratio of 4:1; it seems to be a rare disease.

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Received: 22 Aug. 2022

Accepted: 06 Nov. 2022

The severity and specific symptoms of FD can vary greatly from one person to another. Most affected individuals only have one bone involved and often there are no associated symptoms (asymptomatic). Many times, FD is discovered incidentally when x-rays are performed for another reason. Conversely, some affected individuals can have multiple bones affected and develop severe and potentially disabling or disfiguring symptoms. In most affected individuals, onset of symptoms is usually in childhood; it is unusual for the onset of the disorder to occur after 10 years.

FD lesions may progressively grow and expand until an affected bone finishes growing. These lesions can eventually cause affected bones to become abnormally weakened, misshapen, and prone to fracture. Bone pain can also occur and may be severe in some patients.



Figure 1: Affected girl showing facial asymmetry

Case Report

A 12-year old girl visited our hospital with complaints of painless swelling over the right side of eyebrow for 4 months. Clinically, the patient presented with facial asymmetry. There was no history of trauma, trismus, and diminished vision, loosening of teeth or epistaxis. Facial examination revealed well-defined bony hard swelling over the right temporal side of eyebrow. On local examination the swelling was rounded in shape with hard consistency whose diameter was 10x10 mm, No tenderness on palpation, overlying skin was free, no regional lymphadenopathy. Ocular exam revealed visual acuity was 6/6 in each eye. Anterior and posterior segment was normal. Ocular motility was full.

Systemic examination revealed normal. There was no swelling present in body and café-au-lait spots were absent. Routine investigations such as hemogram, serum calcium and serum alkaline phosphatase (ALP) were done. All parameters were within normal limits except ALP. ALP was raised to 300 U/L. The computed tomography (CT) scan was done to evaluate the bony status. CT scan showed a radio dense mass with ground-glass appearance involving right frontal bone and its expansion causing facial asymmetry. CT scan showed thickening of the frontal region with loss of anterior wall of frontal sinus. An incisional biopsy was obtained from the lesion and histopathological analysis was done. Macroscopy showed two pieces of bony hard-tissue greyish white in color measuring 1 cm × 0.5 cm and 0.5 cm × 0.5 cm approximately. Microscopic examination showed irregular bony trabeculae in Chinese script pattern scattered within fibrous stroma. Bony trabeculae showed no osteoblastic rimming. Poorly formed metaplastic bone was separated by cellular fibrous connective tissue stroma. The bone appeared woven rather than lamellar. Based on the clinical history, radiographic assessment and histological features of the lesion, a diagnosis of craniofacial fibrous dysplasia FD was suggested which involving right side of frontal bone. MRI suggest diffuse thickening of right frontal bone. No treatment for FD affected frontal bone was done because there were no ocular symptoms and patient was unwilling too. Patient was kept on observation and periodic follow up.





Figure 2: Radiological finding of affected girl



Figure 3: Histological finding of fibrous dysplasia

Diagnosis

A diagnosis of fibrous dysplasia is based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests. Individuals with mild forms of monostotic FD may be diagnosed incidentally when receiving an x-ray for another reason. Specialized imaging techniques may be used to evaluate bone. Such imaging techniques include computerized tomography (CT) scanning and magnetic resonance imaging (MRI). The abnormal tissue in FD resembles ground glass when seen on x-ray. These tests may be used to determine how extensively bones are affected.

A bone scan, also known as bone scintigraphy, is used to determine the extent of bone disease and may be used when a solitary FD lesion is found to confirm a diagnosis of monostotic FD or to discover whether additional affected areas are present (polyostotic FD). During this test, a

harmless radioactive dye is injected into an arm vein. A special camera that can track the dye as it travels through bone is used to create a picture of the skeleton and determine all affected areas.

Bone biopsy is the surgical removal and microscopic examination of a small sample of affected tissue. A bone biopsy can reveal characteristic changes to bone that occur in individuals with FD and may be necessary to distinguish a FD lesion from other types of growths or tumors if it is unclear after an x-ray.

A highly sensitive, specific form of polymerase chain reaction (PCR) has been used to detect somatic mutations of the *GNAS1* gene that characterize FD. PCR is a laboratory test that has been described as a form of “photocopying.” It enables researchers to enlarge and repeatedly copy sequences of DNA. As a result, they are able to closely analyze DNA and more easily identify genes and genetic changes (mutations). In FD, a specific form of PCR testing can detect activating mutations of *GNAS1* in peripheral blood cell.

Treatment

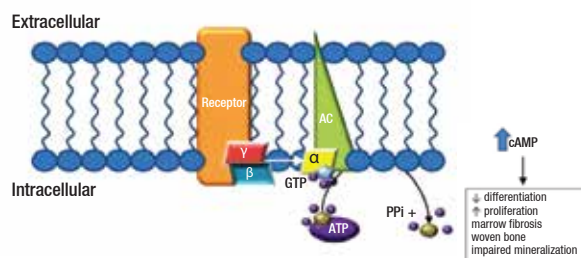
The treatment of FD is directed toward the specific symptoms that are apparent in each individual. Treatment may require the coordinated efforts of a team of specialists. Pediatricians, general internists, orthopedic surgeons, endocrinologists, and other healthcare professionals may need to systematically and comprehensively plan to treat the patient. Psychosocial support for the entire family is essential as well.

Specific therapeutic procedures and interventions may vary, depending upon numerous factors, such as disease progression; size of the lesion(s); the presence or absence of certain symptoms; an individual’s age and general health; and/or other elements. Decisions concerning the use of particular drug regimens, surgical treatments and/or other treatments should be made by physicians and other members of the health care team in careful consultation with the patient based upon the specifics of his or her case; a thorough discussion of the potential benefits and risks, including possible side effects and

long-term effects; patient preference; and other appropriate factors. Being seen by a physician(s) with familiarity in treating individuals with FD is recommended. Individuals with FD have been treated with drugs known as bisphosphonates such as pamidronate or alendronate. These drugs reduce bone turnover by inhibiting bone resorption. Calcium and vitamin D may be given along with the drug. Surgery is often used to treat individuals with FD, although most physicians recommend a conservative strategy.

Discussion

Fibrous dysplasia (FD) is a rare bone disorder. Bone affected by this disorder is replaced by abnormal scar-like (fibrous) connective tissue. This abnormal fibrous tissue weakens the bone, making it abnormally fragile and prone to fracture. Pain may occur in the affected areas. As children grow, affected bone may become misshapen (dysplastic). The severity of the disorder can vary greatly from one person to another. Any part of the skeleton can be affected, but the long bones of the legs, the bones of the face and skull (craniofacial area), and the ribs are most often affected. FD is usually diagnosed in children or young adults, but mild cases may go undiagnosed until adulthood. FD of the craniofacial region can cause a variety of symptoms depending on the type and specific location of the lesions(s). Such symptoms can include pain, nasal congestion, misaligned or displaced teeth, uneven jaws, and facial asymmetry, in which one side of the face does not match the other side. FD in the craniofacial region can alter the facial features resulting in an abnormally prominent forehead (frontal bossing), bulging eyes (proptosis) and difference in the vertical positions of the eyes so that the eyes are uneven (vertical dystopia). The degree of facial abnormality can vary greatly from one person to another. The shape of the skull may be altered in certain cases.



FD can potentially cause a variety of neurological symptoms as areas of abnormal tissue development can compress nearby nerves. Specific symptoms are related to the specific nerves involved. For example, vision loss and hearing impairment can occur because of compression of optic and auditory nerves in the skull. However, vision loss and hearing impairment only occur in rare instances. It can be monostotic or polyostotic. The craniofacial bones are affected in 10%–25% of cases in monostotic forms and in 50% of cases in polyostotic forms. FD essentially affects children and young adult⁶ such as in the present case, a 12-year-old female. CFD is a benign, slowly progressive bone disorder in which normal craniofacial bones are replaced by fibrous tissue in which secondary metaplastic bone formation occurs.⁷ FD is relatively rare in the craniofacial region, (only 20% of all locations).⁸ Involvement of frontal, sphenoid, nasoethmoid and maxillary bones may result in nasal obstruction, sinus obliteration mainly frontal and maxillary sinus and subsequent sinusitis. Other features associated with CFD are dystopia, dysesthesias in the distribution of the trigeminal nerve, epiphora and also headaches.⁹ Latest researchers suggest that the activated G-protein Wnt/B-catenin signaling pathway is involved in modulation of bone formation. Patients with activating guanine nucleotide-binding protein, alpha stimulating mutations specifically showed activated Wnt/B-catenin signaling. The typical radiographic feature of FD is a radiolucent, hazy or ground-glass, pattern. The patterns are due to defective mineralization of immature abnormal bone and it is usually different from the radiographic appearance of normal bone.¹ There are three types of computed tomography (CT) images described as ground glass (56%), homogeneous dense (sclerotic) (23%) and radiolucent (cystic) (21%). These findings are characteristic of FD. Magnetic resonance imaging (MRI) also may help in assessing cranial nerve involvement and soft-tissue structures adjacent to the lesion. Bone scintigraphy is usually recommended to rule out the polyostotic variant of FD. On MRI, FD shows homogenous, moderately low signal intensity on T1-weighted images. Both CT and MRI are excellent imaging modalities in defining the compressive effect of

CFD on the orbit, optic canals and adjacent paranasal sinuses.⁵ Serum ALP is significantly high in FD. Elevated Serum ALP is usually a reliable marker for predicting the prognosis of patients with FD. There is no specific treatment exists for FD. Radical resection is the only technique to obtain complete resolution of FD. Wait-and-see is indicated in cases of stable lesions and ceases to grow once the patient reaches puberty. Reconstructive techniques allow obtaining adequate aesthetical and functional results. Aggressive lesions are treated by radical resection, except in pediatric patients with residual large defects in which it can be acceptable to try to resolve symptoms through bone shaving, reserving more aggressive treatments in cases of relapse or after skeletal maturity. Bisphosphonates are often used as medical treatment as they may reduce the increased bone resorption. They may alleviate bone pain in FD; to conclude, most of the cases of FD can be treated by conservative recontouring.

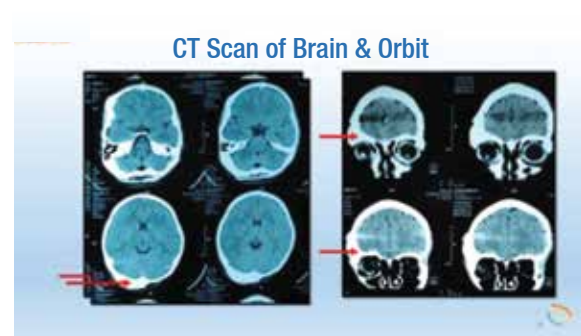
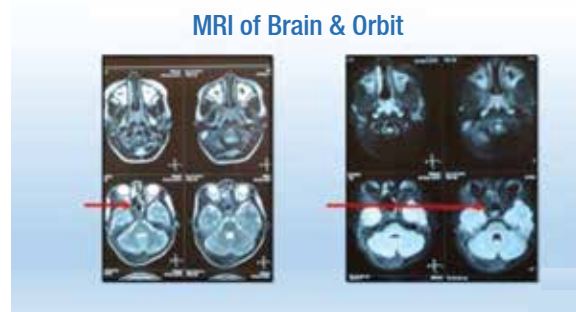


Figure 4: MRI and CT scan finding of Fibrous dysplasia

Conclusion

Fibrous dysplasia is a rare diagnosed disease. Detection of early suspicious sign for diagnosis and early initiation for management is crucial to prevent the long-term complication with avert the process of optic nerve compression.

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