



Bony Cyst in a Ground Glass Matrix: A Rare Case Report of Craniofacial Fibrous Dysplasia with Secondary Aneurysmal Bone Cyst

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Abstract

Keywords

- ▶ craniofacial fibrous dysplasia
- ▶ secondary aneurysmal bone cyst
- ▶ tumor

Fibrous dysplasia has various ways of presentation including syndromic associations and secondary changes within the lesion. We present a case of a 21-year-old female with craniofacial fibrous dysplasia, presenting with proptosis and intermittent blurring of vision due to focal fibrous dysplasia involving the frontal bone with secondary aneurysmal bone formation that was provisionally diagnosed on imaging and confirmed on histopathology. This case demonstrates the typical imaging findings of fibrous dysplasia with seldom encountered secondary aneurysmal bone cyst formation and also discusses about the pathology and management of the craniofacial fibrous dysplasia.

Introduction

Fibrous dysplasia (FD) is a nonhereditary disorder in which abnormal differentiation and maturation of osteoblasts occur. It is characterized by replacement of normal bone with fibrous stroma intermixed with immature and haphazardly arranged trabeculae of woven bone.¹

Craniofacial FD involves skull and facial bones only and presents as an enlarging mass with symptoms mainly due to mass effect.² On radiological examination, it has variable presentations including homogeneously dense or mixed cystic pattern. A sudden increase in size of the mass may point toward a secondary aneurysmal bone cyst or malignant transformation with such cases being rare.³

Case Report

A 21-year-old woman with 1-year history of right eye proptosis, intermittent blurring of vision presented with progressive worsening of her symptoms. On ophthalmological examination, pupils were reactive with no restriction of eyeball movement in any direction of gaze. Her nasal exami-

nation was unremarkable. She underwent magnetic resonance imaging (MRI) with correlative computed tomography (CT) for the evaluation of proptosis.

On CT scanogram image (▶**Fig. 1**), expansile lytic lesion with ground glass matrix was seen involving skull and facial bones. On MRI (▶**Figs. 2 and 3**), an ill-defined altered signal intensity lesion with internal fluid-fluid level was seen along the superomedial aspect of right orbit involving frontal and adjacent ethmoid sinuses with mass effect over the right superior rectus muscle. There was diffuse expansion of adjacent frontal bone with heterogeneous signal on both T1- and T2-weighted images. No significant internal enhancement was seen within the lesion. Correlative CT (▶**Fig. 4**) was done that demonstrated diffuse ground glass attenuation in the expanded right frontal bone involving the superomedial aspect of right orbit and right ethmoid bone. A central lytic lesion was seen that corresponded to the lesion showing fluid-fluid levels on MRI.

The provisional radiological diagnosis was of craniofacial FD with cystic degeneration and secondary aneurysmal bone cyst formation. The patient underwent right pterional craniotomy with gross total excision of the tumor. Intraoperatively,

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Fig. 1 Computed tomography scanogram image showing expansile lytic lesion with ground glass matrix involving skull and facial bones.

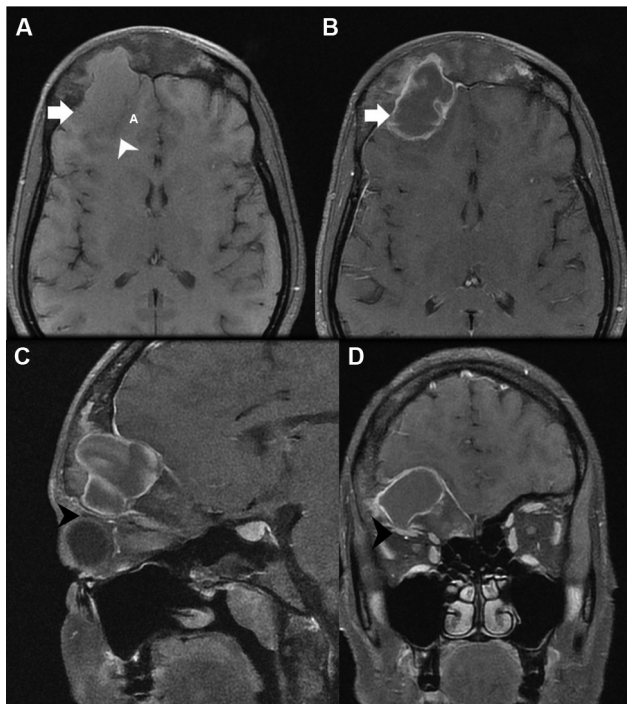


Fig. 2 (A) Magnetic resonance imaging (MRI) axial T1 image showing diffuse expansion of frontal bone with predominantly hypointense signal with areas of hyperintensity. The cystic lesion (white arrow) has T1 isohyperintense signal compressing and displacing the underlying right frontal lobe (arrowhead). (B) MRI axial T1 post-contrast image showing enhancement of cyst wall with no internal enhancement (white arrow). Mild enhancement is seen within the dysplastic frontal bone. (C) MRI sagittal T1 post-contrast image shows rim-enhancing lesion compressing the right orbit without any intraconal extension. The orbital capsule is preserved with mass effect over underlying right superior rectus muscle (arrowhead). (D) MRI coronal T1 post-contrast coronal images showing the lesion causing mass effect over the right superior rectus muscle, distorting it (arrowhead). Normal signal is seen within the optic nerve. The orbit is displaced inferiorly.

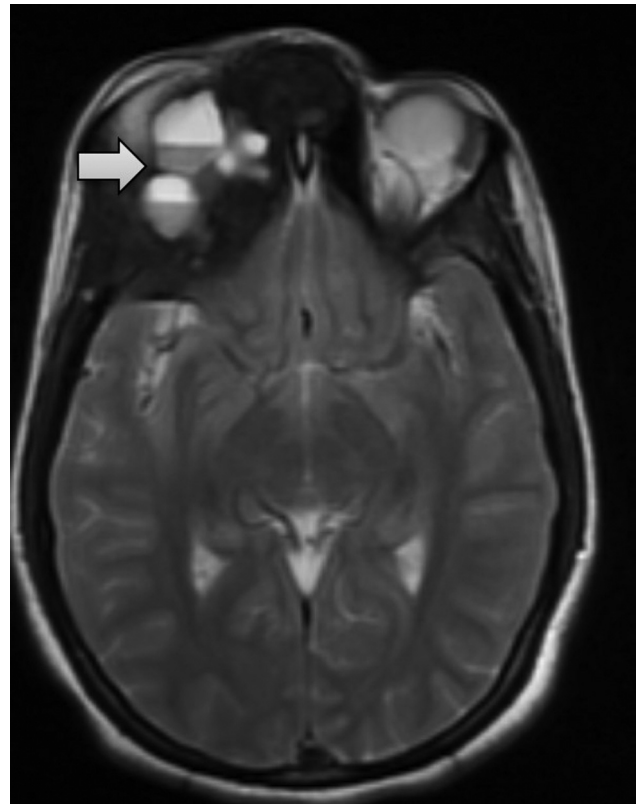


Fig. 3 Magnetic resonance imaging axial FRFSE T2 image showing fluid-fluid levels within the level along the superomedial aspect of right orbit (white arrow). It is compressing and displacing the underlying right frontal lobe. FRFSE, fast recovery fast spin echo.

a cystic lesion was seen in the right frontal sinus that was extending up to the medial orbital wall and ethmoid sinus. The posterior wall of frontal sinus was deficient with hypertrophy of medial orbital wall. The expanded bone as well as the cyst wall was sent for histopathological diagnosis that confirmed the radiological findings.

Discussion

FD occurs due to a sporadic mutation in the α -subunit of the Gs stimulatory protein. FD can either present as an isolated condition, wherein it has been eponymously named Jaffe-Lichtenstein disease, or it may present as a part of syndromes like McCuneAlbright and Mazabraud syndromes.⁴ With regard to the natural course of monostotic FD, it never progresses to polyostotic variety but also does not spontaneously resolve. FD is not static but undergoes histopathological, radiological, and clinical evolution over time with maximum growth occurring during childhood with the lesions becoming quiescent after puberty.

FD is a developmental anomaly with involvement of skull and facial bones in 10 to 25% of patients with its monostotic form.² It usually affects young adult patients similar to our case. Monostotic form is the most common craniofacial variety, with polyostotic type less common.⁵ It usually presents with cranial asymmetry or facial deformity with mass effect on cranial structures presenting as proptosis, globe dystopia,

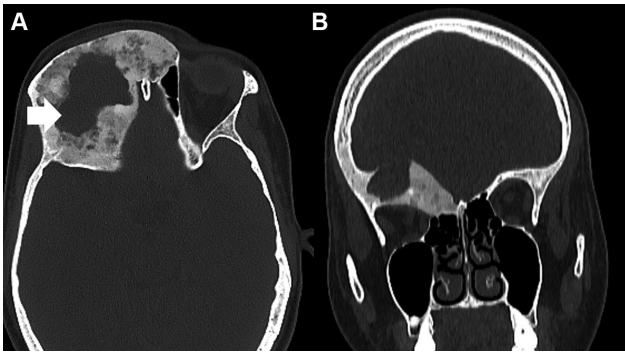


Fig. 4 (A) Computed tomographic (CT) axial bone window demonstrates characteristic diffuse ground glass attenuation in the expanded right frontal bone involving the superomedial aspect of right orbit, right ethmoid bone, bony nasal septum, and nasal bone. Central lytic lesion is seen (white arrow). (B) CT coronal bone window showing central lytic lesion with the dysplastic bone involving the right cribriform plate and reaching up to the crista galli.

hypertelorism, nasal stuffiness, or with neurological symptoms due to compression of optic/vestibulocochlear nerves. Rarely it may present with scoliosis or features of brain compression.⁶ Craniofacial FD preferentially involves anterior craniofacial bones more than the lateral or posterior bones, with frontal bones being most commonly involved⁷ and has three patterns of involvement—ground glass pattern with combination of dense and radiolucent area of fibrosis, sclerotic—homogenous pattern, and a cystic variant with spherical or ovoid lucent area surrounded by dense bone.⁸ In due course of time, it tends to become heterogeneous with the appearance of cystic areas that are concerning for development of secondary ABC or sarcomatous conversion, as was seen in our case.⁴ However, formation of cysts within lesions of FD is not always indicative of secondary changes but can also be seen in the natural course of the disease with advancing age.⁴ Clinically, it may present as a painless swelling, local numbness, or as in our case with proptosis or nasal stuffiness when located in the anterior part of the skull.

CT is the modality of choice for the assessment of the extent and characterization of craniofacial FD, while MRI is the preferred modality for the assessment of complications related to it.⁴ On MRI, the lesions have varied signal intensity but all cases will show post-contrast enhancement, which may be patchy, central, rim, homogeneous, or a combination thereof with milder enhancement in inactive lesions.⁹ Active and multiple lesions can be detected in young patients with technetium 99m-methyldiphosphonate bone scan as an initial diagnostic scan and the lesions can be followed up on radiographs. FD lesions can mimic malignancy/metastasis on fluorodeoxyglucose positron emission tomography scans with increased uptake upon sarcomatous conversion.¹⁰

Imaging differentials for craniofacial FD include ossifying fibroma, cherubism, and central giant cell granuloma.¹¹ Ossifying fibromas typically present with bone expansion, intracortical bone lysis, and rim of sclerotic bone. Cherubism is inherited in an autosomal dominant pattern and involves cystic expansion of mandible and maxilla with sclerosis in later stages. Expansile lytic multiloculated lesions are seen in

central giant cell granuloma that is almost exclusively seen in the mandible with rare cases reported involving the skull base. Another possible differential for cystic change in a preexisting craniofacial FD is a mucocele that, due to blockage of sinus recesses by expanded bone, will have a variable appearance on cross-sectional imaging depending upon the protein content. It may be hypo-hyperdense on CT, hypo-hyperintense on T1- and T2-weighted images, and will show mural enhancement on post-contrast images.¹²

Increase in size of a preexisting FD may be due to secondary ABC formation or malignant transformation.¹³ Secondary ABC occurs due to disruption of blood supply to the lesion. It is seen as a lytic, expansile bony lesion, made up of multiple thin-walled communicating cystic cavities with different densities of blood.¹⁴ It is important to differentiate secondary ABC formation from cystic variant of FD as aneurysmal cysts usually expand much more rapidly than FD.⁴ Malignant transformation, which includes secondary osteosarcoma, fibrous sarcoma, chondrosarcoma, and malignant fibrous histiocytoma, occurs in <1% of patients with monostotic variety.⁸ On imaging, it will have enhancing soft tissue components with extensive surrounding destruction and will demonstrate marked diffusion restriction.

Management of craniofacial FD is guided by the extent of patient's facial dysmorphism, psychological impact on the patient, and the risk of secondary changes in the lesion. It can be dealt with a multidisciplinary approach including medical management in the form of bisphosphonates, ensuring optimal vitamin D levels and surgical correction by burring of the lesion or subtotal to total excision and reconstruction.¹¹

Conclusion

Craniofacial FD can cause facial disfigurement and psychological dysfunction and in rare cases, it may progressively enlarge with secondary cystic transformation to cause orbital, nasal, or auditory symptoms. Imaging helps in diagnosing the primary pathology, characterizing secondary transformation and in surgical planning. A multimodality approach for treatment is necessary with early recognition of this progressive disease including optimal follow-up, aiming to prevent complications in these patients.

Ethics Approval

Appropriate consent from the patient was obtained. Anonymity of patient has been maintained throughout the case report.

Consent to Participate

Appropriate consent from the patient was obtained.

Consent for Publication

All authors give consent for publication of the manuscript in your esteemed journal.

Authors' Contributions

All authors have contributed to the conceptualization, design, acquisition of data, drafting, and review of this article and approved the version to be published.

Conflicts of Interest

None declared.

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