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Research Paper

Skull Base Fibrous Dysplasia - Endonasal Endoscopic Approach : A Case Report

Medical Science

KEYWORDS : fibrous dysplasia,skull base,monostotic,polyostotic,endoscopic endonasal approach.

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ABSTRACT

Fibrous dysplasia is a non-neoplastic, expansile lesion of unknown origin. In about one-fourth of cases this disorder affects the head and neck area, where the mandible and maxilla are the most frequently involved sites. Its localization to the skull base is a rare event. Since the disease slowly progresses, its management is delayed until significant clinical symptoms or non-well-tolerated aesthetic deformities are present.We present a case of fibrous dysplasia of skull base in a 59 years old female. This tumor like growth was not restricted to the skull base, but also compressed the orbit and the globe. Endoscopic endonasal removal of the mass with a drill was performed under general anesthesia. No residual tumor was observed 4 months later.

CASE REPORT

A 59 year old female presented to ENT OPD with the chief complaint of nasal obstruction since 2 years and protrusion of right eye since 2 months.On anterior rhinoscopic examination,a mass was obstructing entire right nasal cavity which was smooth in appearance and bony hard in consistency.The left nasal cavity was normal.On general examination,right eyeball was displaced forwards and downwards.Region above the right eyeball was soft in consistency.On snellen's chart testing,patient's vision was 6/6 in both eyes.CT scan of PNS was done which showed a mass occupying entire right nasal cavity and ethmoids reaching upto right skull base with a typical ground glass appearance.Right frontal sinus was filled entirely with a soft tissue mass with erosion of the floor of sinus.

Based on clinical symptomology and typical 'ground glass'appearance on CT scan,a provisional diagnosis of fibrous dysplasia of right skull base with secondary mucocele of right frontal sinus causing displacement of right eyeball was made. Decision was taken to operate the patient with endoscopic approach with the aim to prevent further displacement of eyeball and loss of vision.Sinonasal mass was removed using drill and sent for histopathological examination.On relieving the obstruction at frontal sinus ostia ,spontaneous drainage of mucocele was established with the resolvement of proptosis on table.

A post-operative CT PNS was obtained 4 days later after nasal pack removal which showed clearance of disease in the sinonasal region.Histopathological report confirmed the diagnosis of fibrous dysplasia .Patient was followed up weekly for a period of 4 months which showed persistence of patency of frontal sinus ostia.

DISCUSSION

Fibrous dysplasia is a noninherited developmental anomaly of bone in which normal bone marrow is replaced by fibro-osseous tissue.[1, 2] This condition was first described in 1942 by Lichtenstein and Jaffe [3] ; hence, fibrous dysplasia is sometimes referred to as Lichtenstein-Jaffe disease. The disease process may be localized to a single bone (monostotic fibrous dysplasia) or multiple bones (polyostotic fibrous dysplasia).[4]

Fibrous dysplasia represents about 5% of benign bone lesions[3]; however, the true incidence is unknown, as many patients are asymptomatic. Monostotic fibrous dysplasia accounts for 75-80% of the cases.

Fibrous dysplasia is a slowly growing lesion that usually appears during periods of bone growth and is thus seen in those in early teen and adolescent years. Polyostotic fibrous dysplasia accounts for 20-25% of cases, and patients tend to present at a slightly earlier age (mean age, 8 y).[5]

Pregnancy can cause increased growth of the lesion as well as secondary changes of aneurysmal bone cyst formation. However, males and females are equally affected, although the polyostotic variant associated with McCune-Albright syndrome is seen more frequently in females.[3]

Any bone may be affected by fibrous dysplasia. The common sites of involvement, in decreasing order of frequency are the femur, tibia, skull and facial bones, pelvis,ribs, upper extremities, lumbar spine, clavicle, and cervical spine. The dysplasia may be unilateral or less commonly,

bilateral. In patients with polyostotic disease, the most commonly involved bones are the craniofacial bones, ribs, and metaphysis or diaphysis of the proximal femur or tibia, and the

lesions are often found on one side of the body.[6,7] The abnormal skin pigmentation also tends to be present on the same side. The craniofacial bones are affected in about 10% of cases of monostotic fibrous dysplasia and in 50%–100% of cases of polyostotic fibrous dysplasia.[8,9,10]

When only the cranial and facial bones are affected, the term craniofacial fibrous dysplasia is used. Van Tillburg analyzed skull lesions from 144 patients identified in the literature and noted that the frontal bones were most commonly involved followed by the sphenoid, ethmoid, parietal, temporal, and occipital bones.[11]

The etiology of fibrous dysplasia has been linked with a mutation in the GNAS1 gene that encodes the alpha subunit of the stimulatory G protein-coupled receptor, Gsá, and is

located at chromosome 20q13.2–13.3.[12,13] The activating mutations occur post-zygotically, replacing the arginine residue amino acid with either a cysteine or a histidine amino

acid. All cells that derive from the mutated cells manifest the dysplastic features. The clinical presentation varies,depending on the location of the mutation in the cell mass and the size of the cell mass during embryogenesis when the mutation occurs.[21] Severe disease may be associated with an earlier mutational event that leads to a larger number or a more widespread distribution of mutant cells. The sporadic occurrence of these diseases and the characteristic lateralized pattern of skin and bone involvement in the polyostotic forms of fibrous dysplasia suggest this mosaic distribution of abnormal cells. The Gsá mutation was first identified in patients with McCune-Albright syndrome, and the Gsá

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gene has also been linked to other endocrine tumors and human diseases.[14]

Radiographic features

Plain film

- ground-glass opacities
- may be completely lucent (cystic) or sclerotic
- well circumscribed lesions

CT

- ground-glass opacities : 56 % [15]
- homogeneously sclerotic : 23 %
- cystic : 21 %
- well-defined borders
- expansion of bone, with intact overlying bone
- endosteal scalloping may be seen [16]

MRI

MRI is not particularly useful in differentiating fibrous dysplasia from other entities as there is marked variability in the appearance of the bone lesions, and they can often resemble tumour or more aggressive lesions.

- **T1** - heterogeneous signal, usually intermediate
- **T2** - heterogeneous signal, usually low, but may have regions of higher signal
- **T1 C+ (Gd)** - heterogeneous contrast enhancement [15]

Nuclear Medicine

Demonstrates increased tracer uptake on Tc^{99} bone scans (lesions remain metabolically active into adulthood)

Differential Diagnosis

Lesions that may suggest fibrous dysplasia include simple bone cysts, nonossifying fibromas, osteofibrous dysplasia, adamantinoma,low-grade intramedullary osteosarcoma, and Paget disease.

Treatment

Many lesions are discovered incidentally on radiographs and are asymptomatic. If the radiographic findings are characteristic of fibrous dysplasia, a biopsy is not indicated. Such lesions ordinarily pose no risk for pathologic fracture or deformity, and only clinical observation is warranted. Follow-up radiographs should be made every six months to verify that there has been no progression. In newly identified cases, a bone scan is needed to exclude a diagnosis of polyostotic disease. When polyostotic disease is found, a referral to an endocrinologist for endocrine and metabolic testing is paramount so that associated endocrine abnormalities can be diagnosed and treated early.

Lustig(2001) observed twenty-one patients with histopathologically confirmed fibrous dysplasia involving the skull base over a 15-year-period (1983-1998).

The ethmoids were most commonly involved (71%), followed by the sphenoid (43%), frontal (33%), maxilla (29%), temporal (24%), parietal (14%), and occipital (5%) bones. The most common presenting features included atypical facial pain and headache, complaints referable to the sinuses, proptosis and diplopia, hearing loss, and facial numbness. Surgical treatment, guided by clinical presentation, ranged from simple biopsy with conservative follow-up to craniofacial resection.[17]

Cai M, Ma L(2012) did a retrospective analysis of collected data for 36 patients with histopathologically confirmed fibrous dysplasia involving the skull .

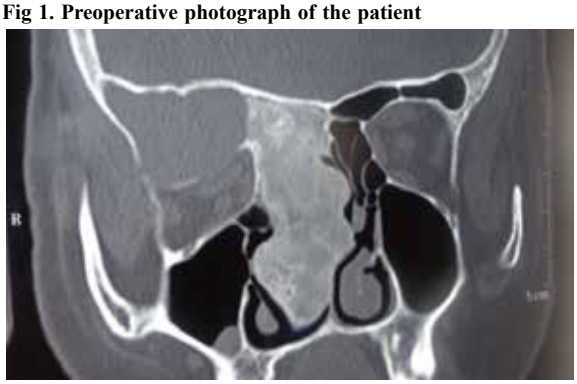
All 36 patients in this review were diagnosed with fibrous dysplasia involving at least part of the skull. In this study, the most commonly involved area of the skull was the frontal bone (52.78% of patients). The next most common area of skull was the temporal bone (30.56% of patients), followed by the sphenoid bone (25% of patients), the parietal bone (19.44% of patients), and orbital bone (13.89% of patients). The principal clinical presentation included headache, local lump, exophthalmos, visual disorder, cranial nerve paralysis, and facial malformation. These patients were treated by surgical treatment, and several of them underwent various degrees of reconstruction to optimize function.[18]

Rijuneeta,Ashok k gupta(2010) analysed 11 patients of fibrous dysplasia of paranasal sinuses with involvement of skull base. Four were managed by endonasal endoscopic approach,1 patient underwent right lateral craniotomy,1 patient had deridement using exrternal ethmoidectomy approach.Rest 5 cases were managed conservatively. [19]

CONCLUSION :

Fibrous dysplasias involving the skull base are rare . The most common treatment for these lesions has been based on surgical resection using an external approach. Only recently has the endonasal endoscopic approach been utilized for the partial or complete removal of these lesions. In this report, we attempt to outline the effectiveness of the endonasal endoscopic approach for the treatment of skull base fibrous dysplasia.

Fig 1. Preoperative photograph of the patient



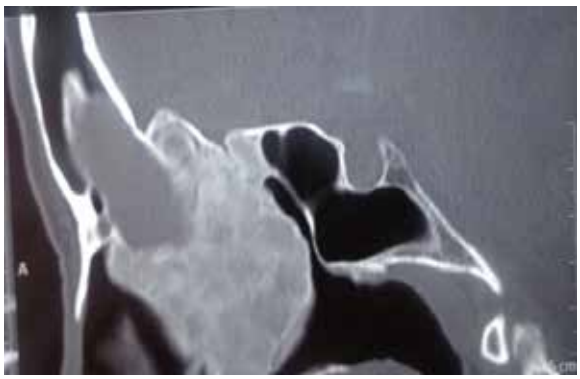
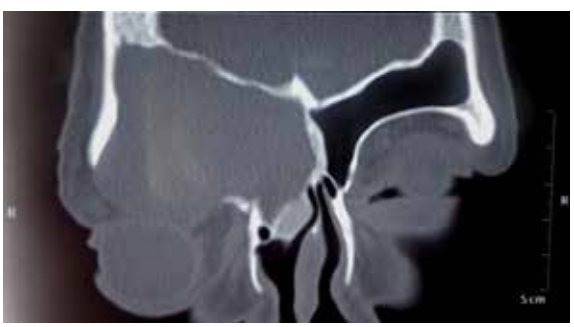


Fig 2. Computerized tomogrphic scans of of the paranasal sinuses showing mass involving right ethmoids with a typical "ground glass" appearance(A : coronal section,B : saggital section.Arrow)



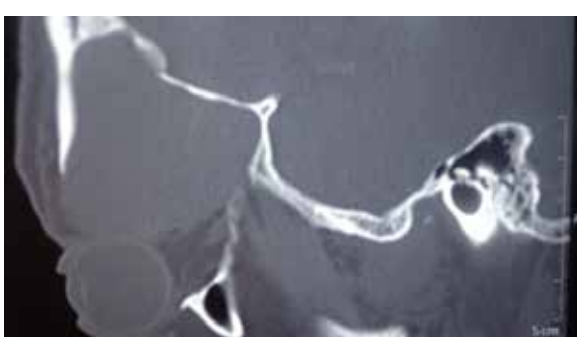


Fig 3 : Computerized tomographic scans of paranasal sinuses showing secondary mucocele of the right frontal sinus with erosion of right frontal sinus floor pushing the right eyeball downwards.(A : coronal section ,B : saggital section. Arrow)

Fig 4 : Post operative photograph of the patient

Fig 5 : Computerized tomographic scans of the paranasal sinuses done 2 months postoperatively showing clearance of disease from right ethmoids and right frontal sinus.(A : coronal section,B : sagittal section.Arrow)

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