Case Report

Fibrous Dysplasia of Skull: An Uncommon Presentation

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Abstract

Fibrous dysplasia is a developmental disorder of bone that can occur in monostotic or polyostotic form. Monostotic form is more frequent, usually seen in older children and young adults, commonly occurring in ribs, femur, tibia and craniofacial bones. Polyostotic fibrous dysplasia is less common, affecting as few as two bones to as much as 75% of the skeleton, predominantly involving the femur, pelvis and foot. We present a case of fibrous dysplasia involving the skull in a 29-year-old woman. On magnetic resonance imaging (MRI), a well-defined lytic lesion was identified in the outer cortex of the left parietal lobe for which two differential diagnoses were made: Langerhans cell histiocytosis (LCH) and lymphoma. The patient underwent craniotomy followed by mesh cranioplasty. The atypical location of the lesion initially obscured the diagnosis, which was ultimately confirmed through histopathological analysis.

Keywords: Fibrous dysplasia, Monostotic, Polyostotic, MRI, Parietal lobe, Histopathological analysis

Introduction

Fibrous dysplasia is a benign fibro-osseous lesion which may present in two forms: monostotic form and polyostotic form.[1] Out of these two forms, the monostotic form is more common, accounting for nearly 75% of cases of fibrous dysplasia.[1] Although fibrous dysplasia can occur at any age, the most common age group for the monostotic form and polyostotic form is between 20-30 years of age and below 10 years, respectively.[2] However, there is equal distribution between both the sexes.[3] Fibrous dysplasia is a genetic disease that has been linked to an activating mutation in the gene that encodes the α subunit of stimulatory G protein (Gsα) located at 20q13.2-13.3, resulting in upregulation of cAMP.[4] It is characterized by swelling, bone pain, bone deformities and pathological fracture. However, some patients may have associated endocrine dysfunction, precocious puberty and cutaneous hyperpigmentation.[5] Complications like nerve compression and malignant transformation are extremely rare.[5] In patients who report increasing pain that persists during rest, wakes them at night or is associated with an enlarging mass, malignancy should be suspected.[3] Fibrous dysplasia is a key characteristic feature of McCune-Albright syndrome[6] and it is the most common tumor of the ribs. Monostotic fibrous dysplasia commonly involves ribs, proximal femur and craniofacial bones.[3] However, polyostotic fibrous dysplasia commonly involves the femur, pelvis and foot.[3] In cases involving the craniofacial bones, fibrous dysplasia is more commonly observed in the zygomatic-maxillary complex and anterior cranial base than in the calvaria, and thus neurological symptoms like seizures are rarely seen in cases of fibrous dysplasia.[7] This report describes the case of a 29-year-old female with headache and seizures associated with fibrous dysplasia involving the left parietal lobe.

Case Report

A 29-year-old female presented to the Neurosurgery OPD with complaints of headache, vomiting and seizures for the past 5 years. Headache was sudden in onset and got aggravated with sound, lack of sleep and exposure to bright light. She was found to be a patient of migraine and was on medication for the same. Seizures were on and off with a frequency of 1-2 per year, sudden in onset followed by loss of consciousness. The patient was subjected to MRI that revealed a well-defined lesion in the outer cortex of the left parietal lobe appearing as a lytic lesion with a punched-out margin (Fig. 1).

On the basis of MRI findings and clinical suspicion, two differential diagnoses were made by the surgeons: Lymphoma and Langerhans cell histiocytosis (LCH). Surgery was performed, i.e., craniotomy followed by mesh cranioplasty, and the entire specimen from the left parietal lobe was sent to the department of Pathology for histopathological examination. Gross examination revealed a bony tissue measuring $3.4\times3.3\times1$ cm. One complete cross-section was taken, divided into two halves, and one half was given for decalcification and later on processed.

Microscopic examination showed branching and anastomosing irregular bony trabeculae of woven bone lacking osteoblastic rimming, interspersed in abundant fibrous stroma composed of cytologically bland spindle cells without prominent cytological atypia along with normal-looking mature bony trabeculae having marrow spaces with hematopoietic cells (Fig. 2).

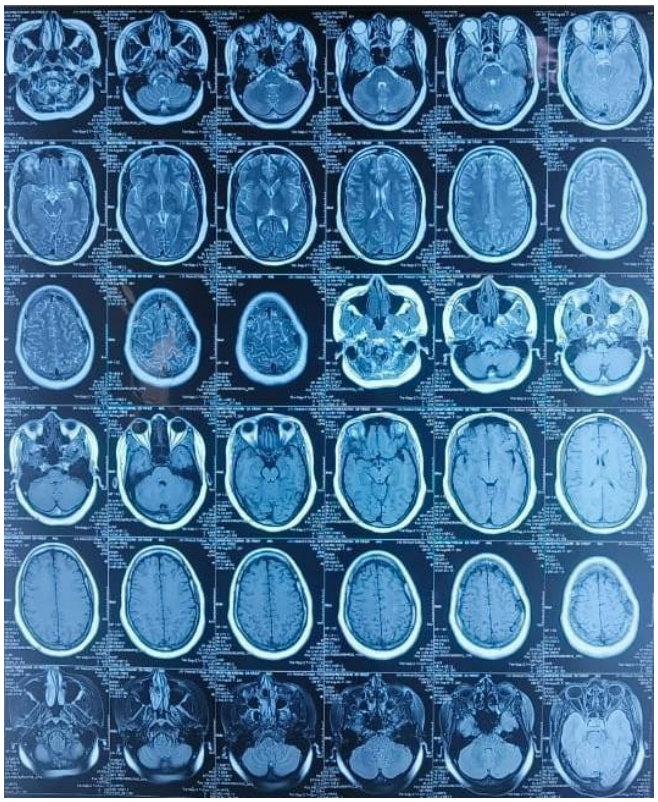

Figure 1: MRI: A well-defined lesion in the outer cortex of the left parietal lobe appearing as a lytic lesion with punched-out margins.

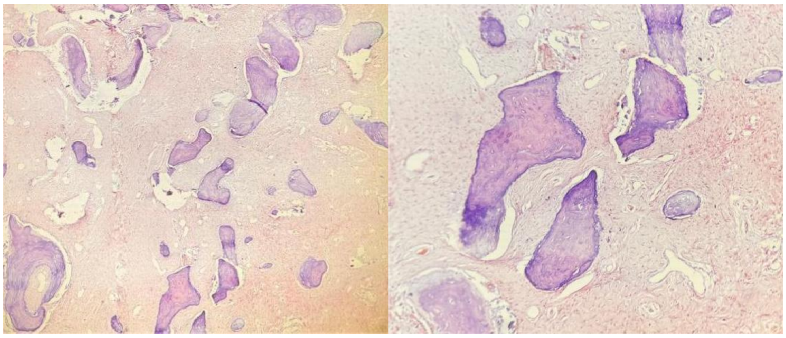

Figure 2: a: H&E X 100 showing branching and anastomosing irregular bony trabeculae of woven bone (C-shaped) with no conspicuous osteoblastic rimming, interspersed in abundant fibrous stroma. b: H&E X 400 showing branching and anastomosing irregular bony trabeculae of woven bone with no conspicuous osteoblastic rimming, interspersed in abundant fibrous stroma.

Discussion

Fibrous dysplasia is not a recent discovery. It was named as "osteitis fibrosa disseminata" by Freund in 1934. It was renamed as "fibrous dystrophy" by Jacobson in 1937.[8] However, the terms fibrous dysplasia and polyostotic fibrous dysplasia were first proposed by Lichtenstein in 1938.[9] The term craniofacial dysplasia is used when only cranial and facial bones are involved. Craniofacial bones are involved in 10% cases of monostotic fibrous dysplasia and 50-100% cases of polyostotic fibrous dysplasia.[10] Lusting et al. conducted a study on fibrous dysplasia of the skull and concluded that the most common bones involved were ethmoid bones followed by sphenoid, frontal, maxilla, temporal, parietal and occipital bones.[11] The initiation and progression of fibrous dysplasia have been caused by an activating mutation in the GNAS1 gene.[11] Fibrous dysplasia is characterized by a mixture of cellular fibrous stroma and trabeculae of immature woven bone in varying amounts. Common signs and symptoms of fibrous dysplasia include swelling, bone pain, bone deformities and pathological fractures.[7] The MRI findings of fibrous dysplasia of the skull in our case revealed a well-defined lesion in the outer cortex of the left parietal lobe appearing as a lytic lesion with punched-out margins, which had a strong resemblance to Langerhans cell histiocytosis and lymphoma, thereby leading to an unavoidable dilemma in reaching a conclusive diagnosis. In our patient, there was involvement of the outer cortex of the parietal lobe which might have resulted in compression of underlying structures and irritation of meninges related to the motor cortex, which may have contributed to seizures.

In the case of LCH, microscopy shows infiltration of sinuses by Langerhans cells with abundant, pale eosinophilic cytoplasm, irregular and elongated nuclei with prominent nuclear grooves and folds, fine chromatin and indistinct nucleoli.[12] However, microscopic examination of lymphoma shows atypical lymphoid cells which may be immunoblasts or centroblasts having vesicular nuclei, prominent nucleoli, scant cytoplasm and occasional mitotic figures.[13] As there were no such findings in our case, thus LCH and Lymphoma were ruled out.

Conclusion

Fibrous dysplasia of skull bones is a rare entity which may mimic other lesions on radiology like lymphomas or Langerhans cell histiocytosis. The final diagnosis can be made on histopathology, but neurosurgeons and physicians shall keep fibrous dysplasia in the differential diagnoses of the lytic lesions of the skull bones.

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