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Clinical Presentation and Outcome of Fibrous Dysplasia in Patients attending Sahiwal Teaching Hospital, Punjab.

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Abstract

Objective: To study the clinical presentation, management and outcome of Fibrous Dysplasia in patients admitted in Neurosurgery Department of Sahiwal Teaching Hospital, Punjab.

Methods: This retrospective descriptive observational study was conducted in the months of January-June, 2021 from the past records of 20 patients with histologically confirmed Fibrous Dysplasia. The data was obtained from the hospital laboratory data base admitted between January 2017 to January 2021. The age of the patients, gender, socioeconomic status and length of hospital stay was also recorded. Clinical presentation and exact location of this tumor on x-rays, surgical intervention techniques and clinical outcomes were tabulated.

Results: This condition was common in the age range of 25-50 years, affecting both genders equally, was more prevalent in low socioeconomic group and prolonged the length of stay of patients to 3-4 weeks. It was observed from the radiographs of the patients that ethmoid bone was most commonly eroded (75%) followed by sphenoid (60%) and frontal bone (25%), 2 cases involved the maxillary bone (10%) and in one case it infiltrated the zygomatic bone (5%). The most common presenting features included visual deterioration and proptosis. Surgical intervention ranged from biopsy and conservative treatment to craniotomy and resection. There was no mortality recorded.

Conclusion: Patients with fibrous dysplasia most commonly present with visual deterioration. Although it is a benign disease, but surgical intervention is required for cosmetic purposes.

Keywords: Fibrous dysplasia, computed tomography (CT), magnetic resonance imaging (MRI).

Introduction

Fibrous dysplasia (FD) is a rare disease caused by a postzygotic mutation of DNA[1] that leads to abnormal fibroblast proliferation, defective osteoblast differentiation, and increased bone resorption.[2] It is a rare skeletal disorder in which fibrous tissues grow in place of a normal bone. This happens when the bone-forming cells do not mature and produce excessive fibrous tissues, which leads to weakening of the bones, bone deformities and fracture.[3] Fibrous dysplasia normally affects a single bone, usually the long bone in the arms and legs or the skull. It may also involve multiple bones.[4] If it involves a single bone, it is called monostotic and if it involves more than one bone, it is called polyostotic fibrous dysplasia.[4] The causative factor is unknown but monostotic is the most common, mildest form, occurring at the age of 20-30 years with involvement of ribs and craniofacial bones; forming 70% of the cases overall in the world.[5] Polyostotic is comprised of the remaining 30%, has a severe onset and is mostly found in children, resulting in skeletal and craniofacial deformities.[5] Monostotic never leads to polyostotic stage.[6] Both are entirely different entities. Fibrous dysplasia is diagnosed during the early years and is present throughout life if not surgically removed. Gender, ethnicity, environmental exposure, race, and geographical location have no influence on the disorder. It can occur in anyone.[7]

Globally, fibrous dysplasia represents 2.5 - 5% of benign bone lesions.[8] Very few cases of fibrous dysplasia have been reported in Pakistan and after an extensive literature search we have concluded that no study has so far been nationally conducted there has been no study conducted on the epidemiological incidence of this condition in our country. This condition remains symptomless for many years and shows clinical manifestations mostly during late 30s. Sahiwal Teaching Hospital is a public sector hospital located in the heart of Sahiwal city, where hundreds of patients visit the out-patient department every day. The neurosurgical unit has a heavy patient toll presenting with diverse clinical symptoms. As fibrous dysplasia is a rare clinical entity and yet there are patients occasionally admitted and treated with this condition, this study was designed to retrospectively gather relevant radiological clinical findings of these patients and correlate them with their clinical presentation and demographic details admitted from 2017 onwards.

Methods:

This retrospective descriptive observational study was conducted in the months of January-June, 2021 on the past history records of 20 patients with histologically confirmed FD. The data was obtained from the hospital laboratory data base of patients with FD admitted between January 2017 to January 2021. The age of the patients,

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gender, socioeconomic status and length of stay was also recorded. Moreover, any record of associated genetic disorder, including McCune-Albright syndrome, polyostotic FD with skin pigmentation or endocrinopathies, such as hyperthyroidism, hyperparathyroidism, acromegaly, diabetes mellitus, and Cushing syndrome, which could be associated with fibrous dysplasia, were thoroughly explored for from the previous records and ruled out.

Anatomical location of fibrous dysplasia lesion in patients with the help of CT scans, patient's complaints recorded during history taking, methods of intervention through surgery and outcomes of treatment were all tabulated in these 20 patients with a diagnosis of fibrous dysplasia, retrospectively identified through reviews of clinical charts and computerized pathologic records.

Radiologic records, either computed tomography (CT) or magnetic resonance imaging (MRI) or plain x-rays, were attained wherever possible; radiographic information was otherwise obtained from reports or notations in medical records. Only those subjects were included in the study who had histopathologic confirmation of the disease and whose radiographic records were present in the hospital data base. The patients with missing radiographic or demographic records were not included in the study.

Results

Average age at presentation was between 25-50 years. There was a 1:1 male-female ratio (10 males and 10 females), and the patients belonged to poor socioeconomic class. Their average length of hospital stay was around 3-4 weeks, from the time of admission to the time of discharge after surgery. The most common anatomical location was ethmoid bone followed by sphenoid and frontal bone.

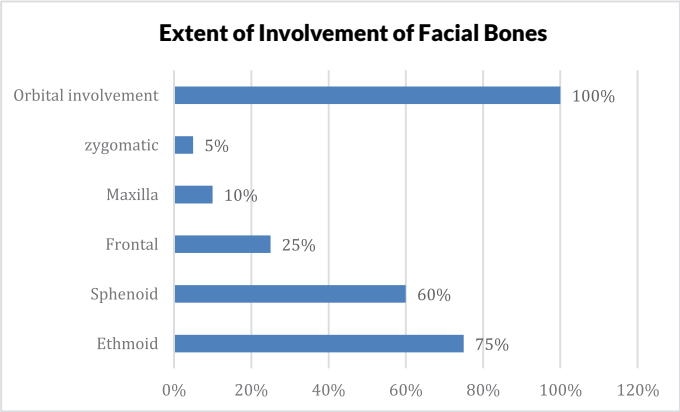

Figure 1: Percentage of facial bone involvement in patients

The most common clinical presentation of fibrous dysplasia was visual deterioration and proptosis (70%). Headache was found in 50% of patients whereas lockjaw was reported in 10% patients only.

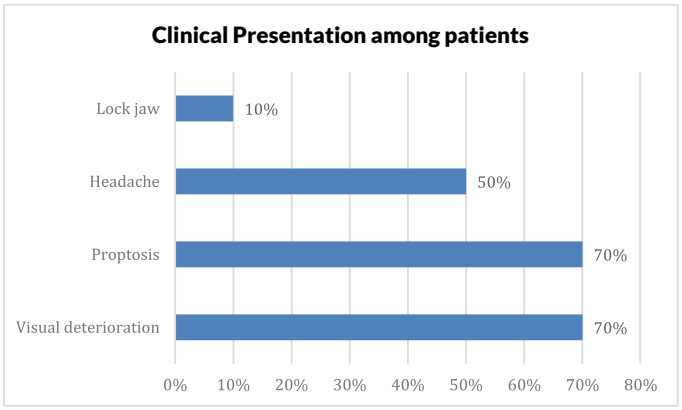

Figure 2: Clinical presentation among patients

Majority of patients underwent endonasal biopsy (50%), 40% patients underwent frontal craniotomy and excision due to compression of optic chiasma to relieve pressure, to improve the vision and 10% patients underwent frontal craniotomy with orbital decompression. One patient was blind and hence was sent home for interval craniotomy. Rest were followed up to monitor visual deterioration. There was no mortality recorded.

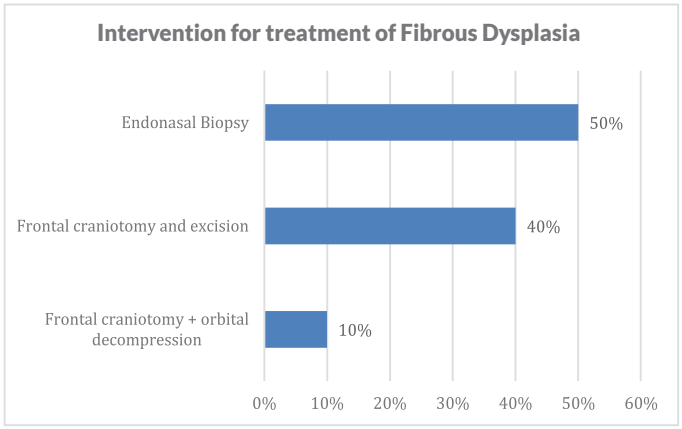

Figure 3: Selected intervention for treatment of Fibrous Dysplasia

The results of chi-Square test revealed that the patients who underwent Frontal craniotomy + orbital decompression had involvement of ethmoid and sphenoid in developing fibrous dysplasia were 2(10%). The patients who underwent Frontal craniotomy and excision had involvement of ethmoid sphenoid (5 (25%), ethmoid and frontal bone (2(10%)) and sphenoid and zygomatic bone (1(5%) in developing fibrous dysplasia. The patients who underwent endonasal biopsy had involvement of ethmoid sphenoid bone (4(20%)), ethmoid and Frontal bone (3(15%)), Maxillary bone (2(10%)), and Ethmoid and maxillary bone (1(5%)) in developing fibrous dysplasia.

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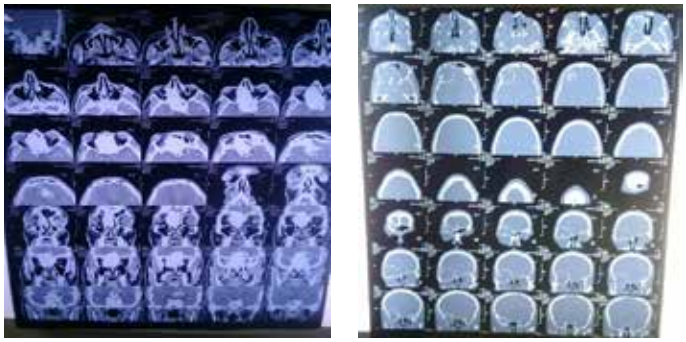
Table 1: Comparison of intervention selection related to bone involvement

	Ethmoid Sphenoid	Ethmoid Frontal	Maxilla	Ethmoid Maxilla	Sphenoid Zygomatic	X2	Sig.
Frontal craniotomy + orbital decompression	2(10.0%)	0(0.0%)	0(0.0%)	0(0.0%)	0(0.0%)	6.327	0.611
Frontal craniotomy and excision	5(25.0%)	2(10.0%)	0(0.0%)	0(0.0%)	1(5.0%)		
Endonasal Biopsy	4(20.0%)	3(15.0%)	2(10.0%)	1(5.0%)	0(0.0%)		

Figure 4 (A and B). Axial and coronal view of lesion in patient involving ethmoid and sphenoid bone causing compression of optic chiasma and showing orbital involvement.



Discussion

Fibrous dysplasia is a rare bone disorder, commonly associated with pain, deformity and fractures, which may significantly impact quality of life[1, 2] The records obtained from the hospital indicated that the patients who had undergone surgery were all suffering from craniofacial fibrous dysplasia and there were no mortalities recorded. Surgical intervention was opted for by all patients for many reasons, including pain relief, cosmetic reasons and for fear of fractures and loss of functions.[10] Patient education regarding the risk of fracture is important. In cases of craniofacial disease, patients should be coached to monitor for evolving cranial nerve deficits including vision and hearing loss. There were no cases reported involving cranial nerve deficit in this study, but the main concern of patients was severe pain in the face radiating to head region. It was observed that the function of nearby cranial nerves was not compromised in any of the patient but loss of vision was the most common complain which probably was due to compression of optic nerve.

Since there is no medical treatment for fibrous dysplasia, the only alternative we are left with is surgical intervention, which should be carefully planned based on the patient's clinical presentation. The basic purpose of surgery is to preserve the function and not only excise the lesion. Lesions that do not cause any symptoms and are found by chance should be followed up by serial CT or MRI scans and not considered for surgical resection if they are benign.[10] More often, however, the surgeon is faced with a lesion and without a diagnosis. If a lesion cannot be readily classified by radiologic studies, open biopsy and surgical excision are warranted. However, it must be remembered that fibrous dysplasia is a benign process, and therapy should be guided by the patient's clinical presentation.[11]

Rare case reports have been reported across Pakistan by surgeons and physicians with unusual presentations involving pain and swelling in face region[12,13], including both polyostotic and monostotic FDs. This condition is usually associated with mutations in Guanine Nucleotide binding protein (GNAS) gene and are a clinical presentation of syndromes like Mccune Albright, Jaffe' and Lichtenstein disease.[14,15] In these patients, there was no record found of any of these disorders. It was unusual to find as many as 20 cases of fibrous dysplasia among patients visiting this hospital over a span of 4 years, when these cases are rarely reported elsewhere. This population should be further explored in order to determine the etiology of this rare condition. As this is a genetic disease, further studies should be performed to get exact demographics of our population.

Conclusion

Patients with fibrous dysplasia most commonly presented with visual deterioration. Although it is a benign disease, but surgical intervention is required for cosmetic purposes.

Limitations of the study

As this is a rare condition, adequate sample size could not be obtained. Moreover, a prospective study could have gathered more information about the clinical presentation and outcomes of this rare disease. We could not obtain the serum alkaline phosphatase levels from the past records, which are significantly raised in this condition

References

1. Hopkins C, de Castro LZ, Corsi A, Boyce AM, Collins MT, Mara Riminucci M, Heegaard A,
Fibrous dysplasia animal models: A systematic review. Bone, 2021; Vol. 155, 2022,116270, https://doi.org/10.1016/j.bone.2021
2. Hartley I, Zhadina M, Collins MT, Boyce AM. Fibrous dysplasia of bone and McCune-Albright syndrome: a bench to bedside review. Calcif. Tissue Int., 104 (5) (2019), pp. 517-529
3. Kelly MH, Brillante B, Collins MT. Pain in fibrous dysplasia of bone: age-related changes and the anatomical distribution of skeletal lesions. Osteoporosis Int. 2008 Jan;19(1):57-63. doi: 10.1007/s00198-007-0425-x. Epub 2007 Jul 11. PMID: 17622477.
4. J S. Lee E J. FitzGibbon Y R. Chen H J. Kim L R. Lustig. Clinical Guidelines for the Management of Craniofacial Fibrous Dysplasia. Orphanet Journal of Rare Diseases 2012, 7(Suppl 1):S2 http://www.ojrd.com/content/7/S1/S2.
5. Traibi A, El Oueriachi F, El Hammoumi M, Al Bouzidi A, Kabiri el H. Monostotic fibrous dysplasia of the ribs. Interact Cardiovasc Thorac Surg. 2012;14(1):41-43. doi:10.1093/icvts/ivr048
6. Kushchayeva YS, Kushchayev SV, Glushko TY, et al. Fibrous dysplasia for radiologists: beyond ground glass bone matrix. Insights Imaging. 2018;9(6):1035-1056. doi:10.1007/s13244-018-

0666-6
7. Mahadevappa A, Patel S, Ravishankar S and Manjunath GV. Monostotic Fibrous Dysplasia of the Rib: A Case Report. Case Reports in Orthopedics, vol 2012, ArticleID 690914, 5 pages, 2012. https://doi.org/10.1155/2012/690914

8. Pruksapong C, Lohwongwatana B. Orbitocranial Fibrous Dysplasia: Immediate Reconstruction with Titanium 3-dimensional Printing. Plast Reconstr Surg Glob Open. 2020;8(9): e3114. Published 2020 Sep 21. doi:10.1097/GOX.0000000000003114
9. Kaynak BA. Conservative treatment of Fibrous Dysplasia. Pak J Med Sci. 2019;35(3):873-876.
doi: https://doi.org/10.12669/pjms.35.3.14.
10. Fang X, Liu H, Lang Y, Xiong Y, Duan H. Fibrous dysplasia of bone: Surgical management options and outcomes of 22 cases. Mol Clin Oncol. 2018;9(1):98-103. doi:10.3892/mco.2018.1636

11. Menon S, Venkatswamy S, Ramu V, Banu K, Ehtaih S, Kashyap VM. Craniofacial fibrous dysplasia: Surgery and literature review. Ann Maxillofac Surg. 2013;3(1):66-71. doi:10.4103/2231-0746.110088.
12. Kaynak, B. A. (2019). Conservative treatment of Fibrous Dysplasia: Conservative treatment of Fibrous Dysplasia. Pakistan Journal of Medical Sciences, 35(3). https://doi.org/10.12669/pjms.35.3.14

13. Siddique M, Bashir H , Niazi IK , Shamim A. Monostotic Osteofibrous Dysplasia of the Tibia. J Cancer Allied Spec 2018;2(1):9
14. Dumitrescu CE, Collins MT. McCune-Albright syndrome. Orphanet J Rare Dis. 2008 ;3:12. Published 2008 May 19. doi:10.1186/1750-1172-3-12.
15. Akkemik O, Erkus S, Kalenderer O, Dündar N. Jaffe Lichtenstein Type of Polyostotic Skeletal Fibrous Dysplasia With the Involvement of Cranial Bones. J Craniofac Surg. 2021 Nov-Dec 01;32(8):e705-e708. doi: 10.1097/SCS.0000000000007700. PMID: 33935141.