CT and MR Imaging in a Large Series of Patients with Craniofacial Fibrous Dysplasia

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Summary

Background: In this retrospective review of patients with craniofacial fibrous dysplasia (FD), the clinical and radiological findings of CT and MR scan were analyzed.

Material/Methods: The study material included 32 patients, at 9 to 68 years of age that were directed for differential diagnostics of several disorders in the head. We recorded CT and MRI data related to the lesion number, location, sidedness, appearance, and sex of the cases with craniofacial FD.

Results: Of 32 patients involved in this study, 17 had monostotic and 15 had polyostotic involvement pattern. Bones most commonly involved by monostotic involvement in females were, in descending order, mandibular, maxillary, and sphenoid bones, while the sphenoid bone was involved the most in males. Leontiasis ossea was observed in 2 patients. Sclerotic and mixed lesion types were more common in both females and males. In T1- and T2-weighted MRI sequences, hypointensity was more common compared to hyperintensity or heterogeneous intensity. The type of enhancement of lesions was found similar after contrast medium administration.

Conclusions: In the presence of craniofacial FD during CT or MRI imaging of the head, a detailed description of FD lesions may provide an important clinical benefit by increasing radiological experience during the diagnostics of this rare disorder.

Keywords: Craniofacial Abnormalities • Fibrous Dysplasia of Bone • Magnetic Resonance Imaging • Tomography Scanners, X-Ray Computed

Background dysplasia (FD) as an entity was described partly by von Recklinghausen in 1891, and in detail by McCune and Bruch in 1937, which was immediately succeeded by coining of the term “fibrous dysplasia” by Lichenstein in the following year [1–3]. Knowledge about the clinical and radiological course and behavior of FD has increased considerably for the last 70 years. FD is a congenital but non-heritable, non-neoplastic, slowly progressive disorder of the bone-forming mesenchyme, presenting as a solitary focal area, or generalized multifocal areas in the bone structure with inadequate maturation from woven to lamellar bone. FD is one of the rare disorders [4] and the prediction of incidence and prevalence is difficult to establish with certainty because of asymptomatic undetected cases. However, the FD lesions may be not rare, and they make up between 5% and 7% of benign bone tumors [5].

FD lesions may involve one bone (monostotic FD), multiple bones (polyostotic FD), or be a component of McCune-Albright syndrome (MAS), which was known with a classical triad of polyostotic FD, café-au-lait skin macules, and endocrinopathies [3,6–9]. Although FD lesions can be located in a portion of any bone in the body, it is usually found in the proximal femur, tibia, humerus, ribs, and craniofacial bones, in decreasing order of incidence. Although multiple bones can be affected at once with a preference to
the one side of the body, polyostotic cases can affect multiple adjacent bones or multiple extremities. However, FD lesions do not spread to other bones since the pattern of involved bones was determined congenitally [3,6].

Cranial or facial bones are affected approximately in 30% of the patients [10,11]. The most common presenting symptom in craniofacial FD is a gradual, painless enlargement of the involved bone or bones in the craniofacial region, clinically seen as facial asymmetry, sometimes as severe deformity with devastating functional and aesthetic consequences for the affected individuals [12–20].

Increase in awareness and familiarity with different imaging varieties of FD lesions helps early diagnosis and management of patients and eases to deal with the complications and their management. For this purpose, we attempted to contribute to the knowledge related to the imaging features of FD.

The aim of the study was to report the findings of CT and MR imaging in patients with craniofacial FD and to discuss the imaging assessment of craniofacial FD.

Material and Methods

Patients

In this study, thirty-two patients were reviewed retrospectively to present craniofacial FD detected in patients subjected to differential diagnostics of craniofacial lesions. CT imaging was the preferred option in twenty-five patients. A total of 2 patients underwent MRI only. Five patients underwent both CT scan and MRI. A total of 32 patients...
(13 male and 19 female) with craniofacial FD and mean age of 28.3±15.3 years (range, 9–68 years) were included in this study from 2002 to 2012. The mean ages of females (n=19) and males (n=13) were 27.1±15.3 (9 to 67 years) and 30.1±15.7 (9 to 68 years), respectively. The approval of Human Ethics Committee of our university was obtained and patient charts were retrospectively reviewed.

**Clinical findings**

A painless mass was the most common symptom, and in many patients the mass was found incidentally. A total of 32 patients experienced different degrees of headache, from slight to very severe pain. Four patients had a progressive hearing loss. Five patients had vertigo. Diplopia, nasal obstruction, and numbness were rare symptoms. Vision loss was not common (2 patients) but was the most serious symptom. None of the patients had abnormal skin or cutaneous pigmentation. A routine laboratory examination, including serum calcium, phosphorus, and alkaline phosphatase levels was within normal limits. Thoracic, abdominal, and cardiovascular examinations of all patients were normal.

**CT examinations**

The examinations were performed with the use of multidetector CT (128-slice CT unit, Aquilion, Toshiba, Japan) with standard parameters. CT examinations were performed with and without contrast medium enhancement, in axial sections with 2-mm collimation using 130 kV, 125 mA values.

Multiplanar reconstructions (MPR) of CT images in the bone window were used to evaluate the location and extent of the lesion in the craniofacial region. Additionally, 3-dimensional volume-rendered images were obtained from axial images on a separate workstation to display vascular and osseous structures (Figures 1–3).

**MRI examinations**

MRI examinations were performed using a 1.5 Tesla MRI machine (Excelart, Toshiba, Tokyo, Japan) with standard head coils. The MR images were obtained in three planes (axial, sagittal, and coronal) with T1-weighted (T1W) spin echo (SE) (repetition time [TR]=440 msec, echo time [TE]=15 msec, flip angle [FA]=90°), and T2-weighted (T2W) fast SE (FSE) (TR=5000 msec, TE=90 msec, FA=90°) sequences. In MRI examinations, no contrast medium was used. The MRI appearances were noted on T1W, T2W and post-contrast T1W images.

**Statistical analysis**

We recorded CT and MRI data related to the lesion number, location, sidedness, appearance, and sex of the cases with craniofacial FD. Data were expressed as numbers and percentages. MRI data were analyzed with chi-square test. Significance was determined at the p<0.05 level.

**Results**

Overall, craniofacial FD was more common in females than in males. Of 32 patients involved in this study, 17 had monostotic and 15 had polyostotic involvement pattern on CT and MRI examinations. Polyostotic involvement was more common in females. Bones most commonly involved by monostotic involvement in females were, in descending order, mandibular, maxillary, and sphenoid.

![Figure 2. A 27-year-old woman with monostotic FD. (A) 3D-CT image showing an expansile osseous lesion in the maxillary bone (short white arrow), and (B) unenhanced axial CT on bone window showing a sclerotic pattern (long white arrow).](image-url)
bones, while the sphenoid bone was involved the most in males. Leontiasis ossea was observed in 2 female patients. Parietal involvement was present in only one male patient. Maxillary involvement was absent in polyostotic involvement in females and in monostotic involvement in males. The frontal bone was by far the most commonly involved bone in polyostotic involvement in females, while frontal, sphenoid, and temporal bones were the most commonly involved bones in males (Table 1).

Lesions were assessed as sclerotic, cystic or mixed in terms of findings according to the density characteristics on CT images.

MRI demonstrated the lesions to be homogeneously hypointense to normal bones and skeletal muscles on FSE T1W, and hypointense to normal bones and skeletal muscles on SE T2W images in all patients. All lesions showed some degree of enhancement on post-contrast T1W images (Figure 4).

Figure 5 presents the number of females and males according to radiological patterns of CT findings in the study population. In females and males, the ratios of monostotic FD (47.4% vs. 61.5%, respectively) and polyostotic FD (52.6% vs. 38.5%, respectively) were found to be comparable.
The sclerotic and mixed types of lesions were more common in both females and males. Figure 6 shows the type and number of osseous involvements of craniofacial FD in the study population. Single and 2–3 bone involvement was meaningfully more frequent, and leontiasis ossea was present in 2 cases. Figure 7 displays the findings of MRI signal intensities and contrast medium enhancement features in the study population subjected to MRI (n=7), with all of the patients being administered contrast medium. On T1- and T2-weighted MRI sequences, hypointensity was more common compared to hyperintensity or heterogeneous intensity. The type of enhancement of lesions was found similar after contrast medium administration.

Only nine cases were treated surgically. The treatments included local curettage, local resection of related bone, excision of related bone and reconstructive plate internal fixation, excision of related bone and rib graft, and excision of related bone and ceramic prostheses reconstruction. The remaining patients did not accept any treatment. All patients were advised to undergo regular clinical and radiological follow-ups.

Discussion

The radiological signs of craniofacial FD are very distinctive, visualized as a thin cortex with well defined borders and ground-glass appearance. Radiographically, the appearance varies regarding the degree of development and amount of bony matrix within the lesion. The radiographic appearance is more radiolucent and well defined in the early stages, and becomes mottled and more radiopaque as the disease progresses [18]. Since 1970s, CT has been increasingly used to define the location and extent of lesions of FD. A standard craniofacial CT scan, with slice thickness no greater than 3.75 mm, is the preferred imaging modality for the presence of FD in the skull base and facial bones [6]. The magnetic resonance imaging (MRI) appearances of FD are often not conclusive for the differential diagnosis of FD. Although CT findings can also be variable, CT more frequently leads to a specific diagnosis thanks to the characteristic ground-glass appearance of woven bone, seen on CT scans in most, if not all, cases of craniofacial FD [21,22].

Management of craniofacial FD remains difficult to perform due to the small number of cases presenting to each hospital and the great variation between its clinical presentations. FD is found predominantly in children and young adults although may often be unnoticed until middle age. In 70% of the lesions, FD is monostotic and asymptomatic, and may be identified incidentally. Craniofacial FD most commonly involves the bones of the face or the skull base, usually involving the sphenoid or temporal bones. FD commonly crosses bony sutures and monostotic FD at a single site may involve multiple bones. The polyostotic FD often causes deformation of bones, resulting in complications

| Case No. | MFD (n=9) | PFD (n=10) | Case No. | MFD (n=8) | PFD (n=5) |
|----------|-----------|------------|----------|-----------|-----------|
| 1 and 12 | Maxillary |            | 20       | Occipital | Frontal, zygomatic, ethmoid, sphenoid |
| 2        | Bilateral frontal | 21 | Ethmoid |
| 3        | Frontal, occipital, sphenoid | 22 | Mandibular |
| 4 and 9  | Sphenoid | 23 | Bilateral temporal |
| 5, 10, and 15 | Mandibular | 24 | Temporal |
| 6        | Leontiasis ossea | 25 and 28 | Sphenoid |
| 7        | Zygomatic, sphenoid | 26 | Mandibular |
| 8        | Frontal, zygomatic | 27 | Frontal |
| 11       | Clivus, occipital condyle | 29 | Temporal, ethmoid, sphenoid |
| 13 and 17 | Frontal, ethmoid | 30 | Frontal, ethmoid, sphenoid |
| 14       | Temporal | 31 | Fronto, sphenoid, temporal, occipital condyle, parietal |
| 16       | Leontiasis ossea | 32 | Clivus |
| 18       | Frontal, occipital, sphenoid, mandibular | 33 | |
| 19       | Clivus | |

MFD – monostotic fibrous dysplasia; PFD – polyostotic fibrous dysplasia.

(p>0.05). The sclerotic and mixed types of lesions were more common in both females and males.

Figure 6 shows the type and number of osseous involvements of craniofacial FD in the study population. Single and 2–3 bone involvement was meaningfully more frequent, and leontiasis ossea was present in 2 cases. Figure 7 displays the findings of MRI signal intensities and contrast medium enhancement features in the study population subjected to MRI (n=7), with all of the patients being administered contrast medium. On T1- and T2-weighted MRI sequences, hypointensity was more common compared to hyperintensity or heterogeneous intensity. The type of enhancement of lesions was found similar after contrast medium administration.

Table 1. The distribution of cases according to anatomical locations.
such as repeated fractures, limb length discrepancies, and bone pains. The wide spectrum of clinical presentations of craniofacial FD is reflected not only in the location and extent of the lesions but also in the types of CT or MRI findings. On CT scans, FD lesions may have homogeneous or heterogeneous density, or may be cystic in nature [3,21,23,24].

CT is the accepted imaging modality of choice in the diagnostics of craniofacial FD in any patient with typical symptoms of FD, such as asymmetric facial swelling. CT may also complement and enhance the interpretation of MRI appearances of lesions in the skull base and facial bones, especially if there is an MRI-detected bone-based lesion suspicious for a malignant neoplasm; in such cases, the typical areas of ground-glass density in CT may confirm the diagnosis of craniofacial FD. CT is a better radiological tool to assess the deformation of adjacent structures such as the optical canal. FD reveals three characteristic lesions on CT scans: ground-glass pattern (56%), homogeneously dense pattern (23%), and cystic variety (21%) [25]. Cystic variety is usually characterized by radiolucency surrounded by a dense rim of bone seen in FD occurring in the mandible, and rarely seen in the maxilla or other facial bones. The diagnosis of FD is usually easy in polyostotic FD and MAS because of the grossly hemimelic location of typical lesions.
compared to the monostotic forms. During the diagnostic workup, the most important roles of CT are to provide information on the size of the bone lesion, to detect some cortical erosions that may not be visible on plain radiographs, and to show fissures. CT is also useful to assess optic canal narrowing with a potential nerve compression. Density assessments can differentiate FD from other conditions such as osteomyelitis, langerhans granulomatosis, and some malignancies decreasing bone density significantly. In contrast, CT cannot rule out other cystic conditions which may be better visualized with MRI [4,8,21,25–30].

Various studies have suggested the use of MRI as a diagnostic tool for FD [30–35]. MRI is a useful imaging modality for the evaluation of complex cases of FD such as patients with compression of neurological structures. The characteristics of FD may be quite non-specific in the interpretation of an MRI scan. In particular, the presence of a strongly enhancing bone-based expansile lesion on MRI may mimic more invasive neoplasms [30]. FD lesions may be characterized by a decreased signal and sharply demarcated borders on both T1- and T2-weighted images. MRI scan can lead to a misdiagnosis of FD [36]. The MRI characteristics of FD do not share the distinctive features seen on radiography or CT scan, and often resemble those of tumors. This is particularly a challenging problem when the lesion shows intermediate signal intensities on T1-weighted images and high signal intensities on T2-weighted images, and enhances brilliantly after the injection of contrast material. FD may be correctly diagnosed only when the signal intensities on both T1- and T2-weighted images are low in spite of contrast media administration [31]. The fibrous tissue is responsible for the low-intensity signals observed on MRI T1-weighted and spin-echo sequences. Variable-intensity signals, especially high-intensity signals on T2-weighted sequences, are a consequence of the heterogeneous nature of FD lesions. Non-specific liquid-intensity signals are encountered in cases of cystic FD lesions [4,31]. In the present study, MRI demonstrated the lesions to be homogeneously hypo- and isointense to normal bones and skeletal muscles on FSE T1W and hypointense to normal bones and skeletal muscles on SE T2W images in all patients. As the authors, we hypothesized that those findings were not specific for FD. Several studies have shown that MRI is helpful in determining soft tissue involvement, in defining the relation of the lesions to the orbit and the optic nerves preoperatively [25–28,31–34].

Chen et al. [21] presented the CT findings of their 46 patients’ follow-ups in their Craniofacial Center. In their series, painless swelling was the chief clinical problem in 78% of patients, followed by dental malocclusion in 22%. Clinical manifestations were reported to have occurred before 6 years of age in 34%, between 6 and 10 years in 27%, and at more than 10 years in 39% of patients. They noted that the average number of bones involved was 3.2 bones per patient and that involvement of more than one craniofacial bone occurred in 70% of patients. They found that the maxilla, orbital and frontal bones were most
commonly involved and that reassessment of CT findings revealed FD lesions as sclerotic or homogenous in 34%, mixed white and dark or heterogeneous in 55%, and cystic in 11% of cases. We think that the type of lesions changes with age. In our study, sclerotic and mixed types of lesions were more commonly present.

Sztuk et al. [29] reported accidental diagnosis of craniofacial FD in 22 patients during CT. They found monostotic FD in 18 cases (78.8%), polyostotic dysplasia in 4 cases (13.0%), and skull-face dysplasia in 9 cases (14%). According to their CT findings, FD lesions were presented as unclear and gradually turning into pathological tissue with different densities, and having trabeculated appearance, as well as sclerotic, lytic or mixed structure. They concluded that early lesions usually have lower density than the normal surrounding bone, giving the appearance of translucency.

Based on the CT and MRI appearances, Paget’s disease, cherubism, hyperparathyroidism, neurofibromatosis, chronic sclerosing osteomyelitis, ossifying fibroma, osteoma, eosinophilic granuloma, osteochondroma, chondromyxoid fibroma, meningoïd, and sarcomatous neoplasms can be considered as differential diagnosis for craniofacial FD. Generally, CT and MRI presentations of craniofacial FD can change considerably. This may result from gender and age-related changes in the nature of the lesions. Management of FD is usually conservative (i.e., follow-ups with CT or MRI) or surgical, depending on the location, extent, and clinical findings [25–30,32–34,37,38].

To our knowledge, this study is one of the most extensive studies on FD in the literature. However, there are some limitations of this study. First, a small number of cases prevents from drawing rational conclusions for clinical practice. Second, the data were collected retrospectively; CT, MRI and histopathological findings were not available for all patients.

Conclusions

In our opinion, CT plays a more important and useful role in assessing the actual extent of bone involvement, while MRI is more useful in cases with nervous system and/or soft tissue involvement. In particularly CT in combination with MRI can improve the diagnostic and treatment accuracy in FD.

Conflicts of interest

The authors declare that there is no conflict of interest.

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