Maxillofacial Fibrous Dysplasia

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ABSTRACT
Fibrous dysplasia (FD) is a rare and benign bone disorder, where normal bone and bone marrow are replaced by fibrous connective tissue mixed with irregular bone trabeculae. FD located in the maxillofacial area might lead to bone deformity, facial asymmetry, hearing loss, nasal congestion, pain, paresthesia, dental malocclusion, and in severe case it can cause blindness. Describe the maxillofacial fibrous dysplasia disorder and its management. A female patient with progressive type of polyostotic fibrous maxillofacial dysplasia who underwent partial infrastructure maxillectomy and mass contouring. Fibrous dysplasia located in the maxillofacial area is a rare case and requires a complete examination and treatment with surgery.

Keywords: *Fibrous dysplasia, maxillofacial, maxillectomy, bone disorder.*

1. INTRODUCTION

Fibrous dysplasia (FD) is a non-neoplastic lesion and relatively uncommon benign bone disorders, where normal bone and bone marrow are replaced by fibrous connective tissue mixed with irregular bone trabeculae [1]. Generally, FD is a rare case and often occurs as asymptomatic lesion. FD which affects the maxillofacial area can cause bone deformity, facial asymmetry, hearing loss, nasal congestion, pain, paresthesia, dental malocclusion, and blindness–in severe case [2,3].

Fibrous dysplasia is a rare disease. Research conducted in China only found 266 cases jof fibrous dysplasia during 1994 to 2009.[4] The incidence of FD is the same for both men and women, [5] but, other study have shown that the incidence ratio of FD in men and women.

2. CASE REPORT

A 45-year-old woman came to ENT – HNS Clinic with nasal congestion. (The patient had been informed and gave consent to be featured in this article) The patient said that she continuously felt this symptom for the past three years and had been getting worse. She mentioned that for the past 1.5 years the symptom had gotten worse and followed by runny nose with thick mucus that sometimes has unpleasant odors, headache, and right ears buzzing. The patient also experienced pain and demonstrated enlargement on the right side of the face, then a hard lump appeared on the right side of the palate. The symptoms are not followed by nosebleed, mucus drains down to the throat, olfactory disorder, sneezing, itchy nose, loose teeth, blurred vision, and no lump on the neck. The patient denied any other significant medical history, including family health history with tumor.

On physical examination, the general condition of the patient was good, conscious, not anemic, did not appear short of breath, patient blood pressure was 130/80mmHg with heart rate 84x/minute and respiratory rate 20x/minute, afebrile temperature, general heart examination value within normal limits, mesocephalic head, and no palpable abdomen (spleen and liver). Examination of the patients' local status shows that, the face looks asymmetrical, there appears to be a prominent deformity of the anterior wall of the right maxillary sinus, which is hard on palpation, with indistinct borders, also there is no nasal deformities and crepitus. Anterior rhinoscopy examination showed that the lateral wall of the right nasal cavity was pushed medially. Hyperemic mucosa, concha edema, mass, or discharges were not found. Palpation on those lateral wall of the nasal cavity gives the impression of a hard mass in the maxillary sinus. Examination on the left

nose showed that the nasal septum was deviated to the left and there was no mass found in the left nasal cavity.

The right and left ears did not show any abnormalities, but the light reflex of the right tympanic membrane appeared to be reduced. Examination of the throat was all within normal limits, there was a lump on the right hard palate with 3x1x1cm in size, the lump surface was smooth, not fragile, and did not easily bleed, also palpation of the mass was felt hard. There is no palpable enlargement of lymph nodes in the neck.

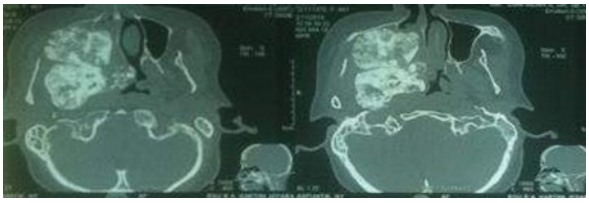

Figure 1 Example CT-scan of Paranasal Sinus Cavities with axial contrast cut

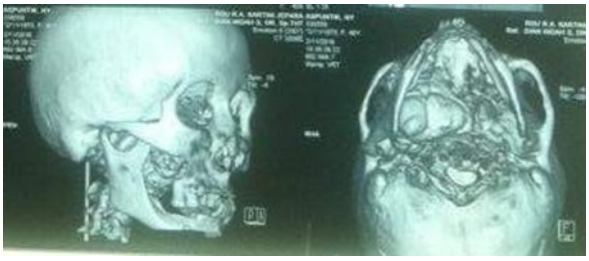

Figure 2 CT-scan of Paranasal Sinus Cavities 3D cranium sections

A CT-scan showed hyperdense lesions filling the right maxillary sinus pushing on the medial wall of the maxillary sinus and the maxillary sinus floor. The lesion extends to the right nasopharynx and right parapharyngeal space. Examination using contrast found that there were no enhancements (Figure 1 and Figure 2). Then the patient underwent a blood laboratory examination in preparation for surgery, the results were within normal limits. EKG examination shows a normal sinus rhythm.

Patient was then referred to the dental and oral department for dental examinations and prosthetic planning for dental implant placement. The dental and oral department then scheduled a dental prosthetic insertion procedure 1 to 2 months after the maxillectomy. The patient underwent infrastructure partial maxillectomy surgery. Maxillectomy was performed using a lateral rhinotomy approach with a Weber-Ferguson incision. This surgery was performed by tearing down the anterior wall and media wall of the maxillary sinus, then the hard palate and right-sided mole was removed. During surgery, a white, hard mass, filled the right maxillary sinus, was found and pushing on the medial wall of the maxillary sinus and right hard palate. The mass was cleaned using a chisel and a drill. The mass of the parapharynx was thinned using a drill. Some of the masses were sent to the Anatomical Pathology Department to be further examined. After the mass can be removed thoroughly, the maxillary sinus and right nasal cavity are then covered with a solid tampon roller. The incision on the skin is sutured layer by layer. The nasogastric tube is then threaded through the patient's left nose (Figure 3 and Figure 4).

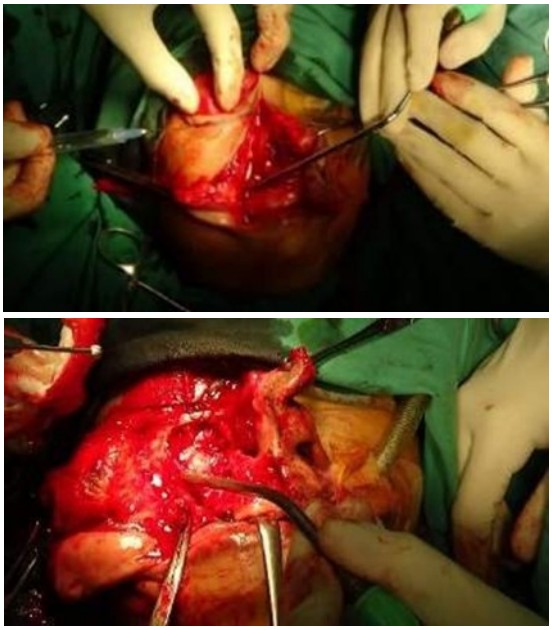

Figure 3 Infrastructure partial maxillectomy

The patient was then programmed for monitoring on vital signs, general condition and bleeding from the nose and throat. Nasal tampons were maintained until the third postoperative day, only the outer gauze was changed every day, or if it was full of blood oozing. The patient's diet is programmed via NGT. Patients were given RL infusion therapy 20tpm, IV Ceftriaxone 2g/24 hours, IV Methylprednisolone 125mg/12 hours, IV Tranexamic acid 500mg/12 hours, IV Ketorolac 30mg/8 hours, and IV Ranitidine 50mg/12 hours.

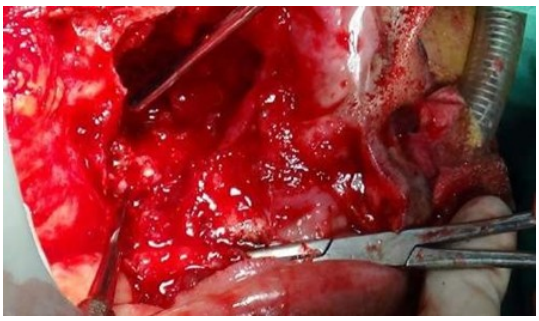

Figure 4 The FD mass has been removed from the right maxillary sinus

The first day after surgery the patient experienced swelling on the right side of the face, facial pain,

difficulty in opening the mouth and difficulty in swallowing. Physical examination showed asymmetrical face, edema on the right side of the face, good approximation suture, and no pus was found. The patient was programmed to remove the tampon on the third postoperative day, change the outer dressing every day, daily diet via NGT, and having mobilization gradually. Previous therapy was continued.

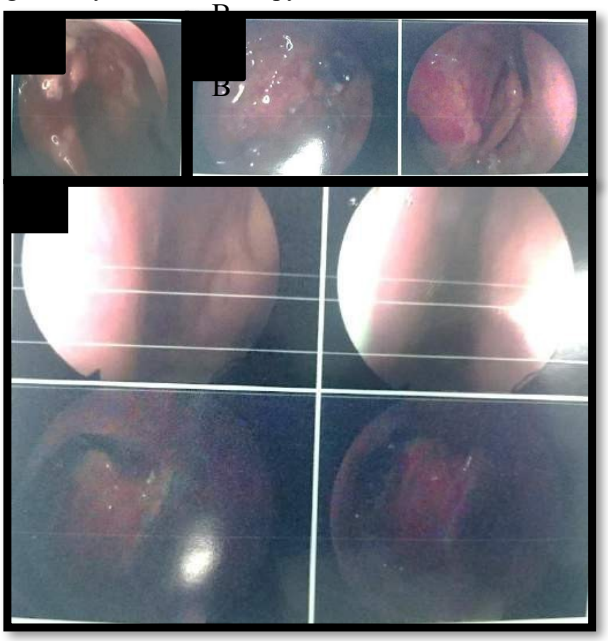

Figure 5 Postoperative evaluation: (A) H7 postoperatively, (B) 1 month postoperatively, (C) 5 months postoperatively

The nasal tampons were removed on the third postoperative day. The removal of the tampon was initially planned in stages, but because there was no bleeding, a complete removal of the tampon was performed. After the tampon is removed, it is evaluated using a nasoendoscopic examination. Evaluation using endoscopy found that the right nasal cavity and right maxillary sinus had no mass, crusting or bleeding. There was an oroantral fistula without visible ulcers or necrotic tissues. An evaluation performed five months after surgery found an oroantral fistula. Anterior rhinoscopy examination still found no mass growth or necrotic tissue (Figure 5).

3. RESULT AND DISCUSSION

Fibrous dysplasia (FD) is a non-neoplastic lesion and relatively uncommon benign bone disorders, where normal bone and bone marrow are replaced by fibrous connective tissue mixed with irregular bone trabeculae [1]. Generally, FD is a rare case and often occurs as asymptomatic lesion. FD which affects the maxillofacial area can cause bone deformity, facial asymmetry, hearing loss, nasal congestion, pain, paresthesia, dental malocclusion, and blindness–in severe case [2,3].

Fibrous dysplasia is broadly classified into three categories of disease: monostotic FD/MFD (affecting single bone), polyostotic FD/PFD (affecting several bones), and McCune-Albright syndrome (MAS). MAS is classically defined by the clinical triad of PFD, *café-au-lait* skin spots (light brown to dark like coffee *latté*), and endocrinopathy including precocious puberty (PP) [3].

Monostotic FD which affecting craniofacial bone only accounts for 10% of cases, whereas polyostotic FD accounts for more than 50% of cases. Fibrous dysplasia, which only affects the craniofacial skeleton is known as craniofacial FD. The prevalence of craniofacial FD was also higher in polyostotic FD (71% of cases) than monostotic FD (10% of cases). The maxillary bones are more susceptible to FD than the mandibular bones. Also, it is more common for women to have craniofacial FD than man [6]. Another study conducted in Hong Kong from 1982 to 2004 showed that there is no significant difference in the incidence of FD in both sex [7]. In addition, it was also found that 8 out of 9 FD cases affected the entire hemimaxilla and only one case affected the posterior maxilla alone [7]. The patient in this case report was a 45-year-old woman with FD affecting the maxilla, nasopharynx and right parapharynx. The FD lesion found in this patient was classified as a craniofacial MFD because it affects more than one craniofacial bone.

Fibrous dysplasia is a non-inherited genetic disorder induced by mutation of the GNAS1 gene on chromosome 20, which encodes the alpha sub-unit of the G-protein stimulatory receptor (Gsα). This mutation occurs after the zygotic phase. This mutated Gsα receptor induced several complex problems; in bones, it will be induced autonomous bone growth via parathyroid hormone receptor pathway, on the skin via melanocyte stimulating hormone receptors, on the ovaries via follicle stimulating hormone receptors, and in the thyroid and pituitary gland via thyroid hormone and growth hormone receptors [8].

Generally, FD signs and symptoms involved: pain, bone deformity and fracture, often asymptomatic. Alkaline phosphatase serum occasionally appears to be elevated, but the levels of calcium, parathyroid hormone, 25-hydroxyvitamin D, and 1.25-dihydroxyvitamin D within normal limits in most cases. Craniofacial fibrous dysplasia can take place in any bone on the head and face, so the most common signs and symptoms usually include facial pain, headache, cranial asymmetry, facial deformity, tooth displacement, malocclusion, also visual and hearing impairment. [5] The most common symptoms found in patients with craniofacial FD are facial deformities, vertical dystopia (eye position that is not aligned vertically), proptosis,

and facial asymmetry [9]. FD lesions on the face can be broadly classified into three categories; quiescent (stable, no growth found), non-aggressive (slow growth), aggressive (fast growth, often accompanied by pain, paresthesia, pathological fractures, and transformation into malignancy) [9].

Commonly, the diagnosis of FD is often made radiologically. Pagetoid image or also known as ground glass pattern appears as a picture of fibrosis areas that emerge as a mixture of radiolucent and dense areas, which found in 56% of cases. A sclerotic pattern is present in 23% of cases, characterized by a uniform dense pattern. Another pattern is a cystic pattern which is present in 21% of cases depicted like a shell on the bone, which emerged as a curved or oval shape with high dense surround [10]. The MSCT scan performed on the patient in this case report showed an image that matched the ground glass pattern where the mixture of lucent and dense areas is found. In line with the literature, most of the patients with FDs come up with ground glass image (Figure 6).

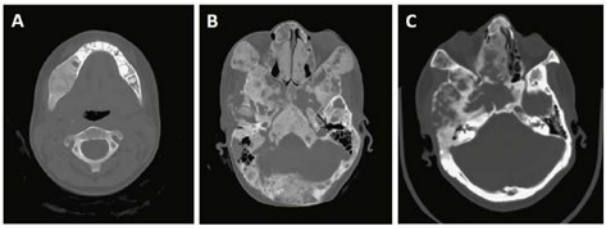

Figure 6 Examples of radiological features of fibrous dysplasia. (A) Ground glass. (B) Mixed.(C) Cystic

The diagnosis is confirmed by biopsy whenever possible. Biopsy is often risky because in FD cases there is often a lot of vascular tissue found, so there is still a risk of bleeding [9].

The diagnosis of this patient is confirmed by anamnesis, physical examination, and conventional radiology. The signs and symptoms experienced risky by our patients are in line with the literature, where the patient experienced nasal congestion, lumps on the cheeks and palate, facial pain, asymmetry, and hearing loss. However, there were no malocclusion or visual impairment experienced by our patients. An examination conducted using CT scan found that there was a ground glass image on the os-maxilla, ethmoid, and right sphenoid. Biopsy was not performed in this patient to confirm the diagnosis. However, the decision of not using the biopsy was taken considering the location of the lesion was in the sinuses and the consistency of the mass tends to be solid. Consequently, there was a high risk of bleeding and it render difficult if the biopsy procedure was performed. According to the literature, a biopsy may not be performed as the diagnosis can be made using the results of the patient's anamnesis, physical, and radiological examination. Based on the above findings, the clinical diagnosis of the patient's disease was fibrous dysplasia polyostotic with aggressive type.

Management of FD in both children and adults is provided based on the clinical findings and biological behavior of the lesions. So far, histology has not been able to determine the disease prognosis precisely because there are no biomarkers to determine the behavior of these fibro-osseous lesions [9]. Pediatric patients will be more susceptible to this case considering the potential for bone growth, transformation into malignancy, and its association with other tumors.

In the case of quiescent or stable Fibrous dysplasia, in which the patient does not experienced any facial deformities, it is recommended to do an observation once a year to find out whether any alteration occur. Examinations on the patient experience, physical inspection including sensory nerve evaluation around the lesion, and CT examination are performed at each visit in the first 2 visits, then interval checkup can be given based on clinical findings. Surgical action such as contouring is required if the patient concerned about the deformity of her/his face. Resection of the entire mass can be performed in monostotic FD, but it is difficult to perform in PFD and MAS. Regular examination post-operation is still needed to ensure recurrences and other deformities are absence [11].

Patients with non-aggressive FD should ideally be awaited until they reach a quiescent state and have reached the bone maturity age before surgery. Surgery for certain cases of non-aggressive FD can be considered, such as in adolescence patient who are experiencing psychosocial development and often disturbed by the presence of facial deformities. The patient and family should understand and be aware of the potential for recurrences that often occur and when the lesion cannot be completely removed. Polyostotic fibrous dysplasia and MAS most of the time cannot be resected because of the extensive nature of the lesions. Repetitive surgeries for contouring and extensive reduction in tumor mass are required to achieve a proportional facial shape. [9]

Patients with extensive growth fibrous dysplasia (aggressive type) typically come with pain or paresthesia/anesthesia. Depending on the location of the lesion, the patient may also experience visual impairment, epiphora, hearing loss, nasal congestion, pain, and malocclusion. Patients in this group are recommended to undergo immediate evaluation including CT examination. A biopsy of the lesion area needs to be done before surgery. Therapy can be given

in the form of surgery ranging from contouring to complete removal of the tumor mass. [9]

The progression of FD into malignancy is reported to occur in less than 1% of cases. Menon et al., reported that the risk of MFD and PFD developed into malignancy was 0.4%, whereas in MAS was 4%.[12] The most common malignancy is osteosarcoma, however fibrosarcoma, chondrosarcoma and malignant fibrohistiocytoma have also been reported. Immunohistochemical examination is necessary in these cases to differentiate FD from malignancy. Immunohistochemical tests that can be done are MDM2 and CDK4 which will generate positive results in cases of malignancy [9].

Treatment given to this patient was surgery. The surgery performed in this patient was a partial infrastructure maxillectomy. This method was used because the FD mass fills the right maxillary sinus and extends downward to the hard palate, while the orbital floor was still intact. The rest of the tumor behind the maxillary sinus was not completely removed, but contouring is performed to ensure that the nasal airway remains patent. This action is in accordance with the literature since this is a case of progressive type of polyostotic FD, so that surgical removal of the mass and contouring was performed.

4. CONCLUSION

This article reported a 45-year-old woman with a progressive polyostotic fibrous dysplasia. Partial infrastructure maxillectomy and mass contouring were performed on the patient. There were no mass recurrences in the evaluation performed at one- and five- months post-treatment.

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