CASE REPORT

Maxillofacial fibrous dysplasia: a diagnostic challenge

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SUMMARY

A 30-year-old woman presented for orthodontic treatment, with a chief symptom of a 'shifting bite' and concurrent facial asymmetry with aesthetic concerns. The patient had previously received treatment from several general dentists and several specialists, without accurate diagnosis. Radiological investigation coupled with biopsy confirmed a diagnosis of fibrous dysplasia. Proper diagnosis led to changes in the treatment plan and gave the patient realistic expectations about the options she had for the outcome of treatment. Prompt diagnosis by dental practitioners is critical to patient satisfaction and successful outcome; therefore, it is important to familiarise ourselves with the signs, symptoms and proper course of management of fibrous dysplasia.

common clinical and radiological findings to best treat their patients.

CASE PRESENTATION

A 30-year-old woman presented for orthodontic treatment, with a chief symptom of a 'shifting bite,' concurrent facial asymmetry and aesthetic concerns ([figure 1](#)). The patient felt that her bite had been getting worse over the past 5 years. She mentioned previous treatment from different dentists and specialists, the most recent being 3 years prior, with unsatisfactory results. Previous treatment included significant occlusal reduction on teeth #17 and #37 as well as orthodontic treatment; the treatments failed to correct the occlusal instability. The patient denied any other significant medical history, including facial trauma and medications. Neither her family nor social history was significant.

BACKGROUND

Fibrous dysplasia (FD) is a fibro-osseous disease characterised by the replacement of normal bone structures with fibrous tissue and woven bone; specifically, a defect is present in differentiation and maturation of osteoblast cells. FD is a relatively uncommon disorder.¹ FD is broadly classified into three categories of the disease: monostotic, polyostotic and McCune Albright syndrome. The term craniofacial FD is used for cases of FD that are confined to the bones of the craniofacial skeleton. These cases cannot be categorised as monostotic due to multiple bone involvement, commonly the zygomatic-maxillary complex. They are considered of greater importance to dental practitioners, because of the often corresponding changes to the facial structure. Signs and symptoms of the disease may include, but are not limited to: facial deformities, paraesthesia and malocclusion. Asymmetry and swelling are some of the most common symptoms. When FD is found in the facial skeleton, due to the slow progression of the disease, it may present entirely as an asymptomatic entity. In young patients, progression of the disease tends to decrease as the patient approaches puberty;¹ however, although rare, continued active disease and symptoms past puberty have been documented, often due to a change in hormonal balance from oral contraceptive use.^{2,3} There are very few established guidelines for the management of this disease, due to the diversified clinical presentations and varying degrees of severity.⁴ The presentation of this case in particular was interesting due to the unusually continuing symptoms at the patient's age. Although postpubescent FD is rare, dental practitioners should familiarise themselves with the

Clinical findings

The patient demonstrated enlargement of the right side involving the maxillary region. On close observation, the expansion spanned from the inferior orbital rim to the angle of the mandible ([figure 2](#)). On smiling, 5 mm of excessive gingival display was noted, with a visually observable maxillary cant ([figure 3](#)). The patient had no café-au-lait macules and noted no cranial nerve impairment. Additionally, there was no suggestive evidence of endocrinopathy.

Intraoral examination

Significant centric occlusion (CO) and centric relation (CR) shift ([figure 4](#)) when smiling were observed. In CR, the patient only occluded on teeth #2 and #31, with a 5 mm maxillary midline deviation to the left side. The mandibular midline was concordant with the maxillary midline. Overjet was asymmetrically progressing from 3 to 6 mm, from the patient's right to left. The patient did not have a crossbite relationship and there was no observed tooth mobility.

INVESTIGATIONS

Radiological investigation included a panoramic radiograph, showing a diffuse radiopaque region in the right maxillary bone spanning from the second premolar to the maxillary tuberosity in the anteroposterior direction. A gradual blending of normal trabecular bone into an abnormal pattern with ground glass appearance was appreciated ([figure 5](#)). There was no evidence of root resorption noted; however, significant displacement of the teeth had occurred. Owing to the fact there

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Figure 1 Frontal profile demonstrating right asymmetry of the face.

Figure 3 Frontal profile on smiling displaying excessive gingiva with maxillary cant.

Figure 2 Profile from below showing asymmetry of the right maxillary region.

Figure 4 Intraoral image showing asymmetry of the right maxilla, open bite on left side.

Figure 5 Panoramic radiograph showing radiopaque lesion across the right maxillary region.

was no evidence of café-au-lait spots and no suggestive evidence of endocrinopathies, further investigations to exclude polyostotic involvement were not performed.

To assess the extent of the lesion and to serve as a baseline advanced imaging, a cone beam CT was obtained. Volumetric analysis demonstrated a hyperdense area in the right maxillary region, with an ill-defined and blending trabecular pattern. In the axial slice, a thinned out but intact buccal cortical plate with expansion was noted (figure 6). Volume rendering showed a dense, thick right maxillary region. No other region was affected. A provisional diagnosis of FD was made. A referral for biopsy was requested and the lesion was found to be a benign fibro-osseous growth consistent with the diagnosis of FD.

Biopsy report: examination revealed multiple sections of a specimen composed of dense fibrous tissue and numerous trabeculae of dense viable bone. Plane polarisation confirmed a mix

Figure 6 Multiplanar reconstruction view exhibiting expansion of the right maxilla; thin buccal cortical plate and volume rendering displaying posterior open bite.

of haphazard woven and lamellar patterns of bone deposition. A diagnosis of benign fibro-osseous lesion consistent with FD was made.

DIFFERENTIAL DIAGNOSIS

Other metabolic bone diseases, such as hyperparathyroidism, may produce a similar bone pattern. However, these diseases are bilateral and do not cause bone expansion. Paget's disease may produce a similar pattern and bone expansion, however, Paget's disease generally occurs in an older age group and involves the entire mandible (in contrast to the unilateral tendency of FD). It is of importance to differentiate chronic osteomyelitis from FD especially in children. Although osteomyelitis may result in enlargement of the jaws, new bone is laid down on the surface of the outer cortex, and examination may reveal evidence of the original cortex within the expanded portion of the jaw. FD, on the other hand, expands the bone from an internal aspect, displacing and thinning the outer cortex so that the remaining cortex maintains its position at the outer surface of the bone. Furthermore, the presence of sequestra can aid in the identification of osteomyelitis.⁵

TREATMENT

A non-surgical approach was chosen as the plan of action. The patient will be closely monitored for any signs of continued progression of the disease, and is beginning orthodontic treatment to correct the maxillary cant; the goal is to establish proper balanced occlusion. A surgical approach option was deferred because of the possibility of activating the lesion and thus leading to potential overgrowth.⁵

OUTCOME AND FOLLOW-UP

The patient is currently undergoing orthodontic treatment, with no existing complications, and will be followed up appropriately. More importantly, she is very appreciative of the thorough diagnostic work up, and relieved that she has been diagnosed correctly and that the therapeutic plan has been communicated

clearly. The proper diagnosis of FD will aid the orthodontist and consequently provide proper treatment for the patient.

DISCUSSION

The first medical documentation of FD was in 1891, by von Recklinghausen, but proper diagnostic protocol as well as management for this complicated disease still remains controversial. Benign fibro-osseous lesions of the jaws often present similarities histologically. FD and ossifying fibroma (OF), for example, often exhibit similar clinical, radiological and histological features. Hence, it is essential to follow strict parameters during diagnostic procedures. As no single investigation is pathognomonic to FD, a proper diagnosis can only be made after a full examination is conducted, including but not limited to: medical history, clinical examination and histological and radiological report. The ill-defined blended 'ground-glass' margins of FD may help differentiate it from other fibro-osseous lesions.⁵ In many cases, there is an accompanying expansion of maxillary bone. Biopsy may not be mandatory if radiographic and clinical findings are sufficient to diagnose FD.⁵ However, in our case, it was elected that a biopsy would be taken to provide an accurate diagnosis. The biopsy results, coupled with the cone beam CT and radiographic evidence, allowed for the diagnosis of maxillofacial FD. Nevertheless, gnathic and extragnathic FD can show distinct radiological and histological profiles that should be accounted for in routine diagnostic practice. Hence, the utility of GNAS as a molecular marker for FD has been recently reinforced by Patel *et al*,⁶ who found that no Arg201 codon mutations were found either in osseous dysplasia (OD) or OF cases.

Treatment

Though there are no uniformly accepted treatment protocols, surgical therapy is often used to correct FD, and to achieve normal function and aesthetics. Valentini *et al*⁷ examined the results of conservative and surgical treatment for maxillofacial FD in 68 patients over a span of over 20 years and concluded that in most cases of monostotic FD, surgical intervention is an

aggressive but acceptable treatment. Recurrence can range from 15% to 20%, however, and lesions often show surprising growth potential if surgically altered during active growth.⁸ A single blanket therapy to be used for complex craniofacial diseases can be difficult to establish due the huge variation in presentations. An individualised approach is suggested as the model treatment, with careful consideration of all factors involved. The decision to surgically intervene should be based on the patient's function, aesthetics and desires. After a lengthy discussion with our patient, a conservative treatment approach was chosen: careful monitoring of the disease and compensatory orthodontic treatment in order to improve function and aesthetics. The patient was asymptomatic for pain and did not wish to undergo a complex surgical procedure. In complex cases, communication with the patient is imperative. When the patient presented for treatment, she was in an anxious emotional state, and very apprehensive to continue with treatment, due to previous unsatisfactory experiences. Not only were her previous failed treatments a financial burden, she noted the facial asymmetries caused her to be more self-conscious in public, as well as placed distrust towards her oral healthcare providers. By listening to the patient and completing a full health history, radiographic work up, and clinical examination, the patient was able to be correctly diagnosed and informed. The patient stated she was much happier to proceed with treatment once trust had been established. It must be highlighted that patients with craniofacial FD should have yearly vision and hearing evaluations.

Conclusion

In cases of FD, the duration of orthodontic treatment may be longer than in conventional cases (no disease present), the results may be less than satisfactory and there is possibility of disease recurrence, however, as long as the patient is informed, practitioners should not shy away from treating FD in their office setting.

Learning points

- ▶ Fibrous dysplasia, although uncommon, can be very difficult to accurately diagnose and treat, due to the diverse clinical presentations and varying degrees of severity.
- ▶ Dental practitioners should familiarise themselves with the signs, symptoms and proper course of treatment.
- ▶ Treatment and care should be personalised to the patient's needs, desires and involvement sites.
- ▶ Communication is key for, patient treatment and expected outcomes.

Competing interests None declared.

Patient consent Obtained.

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