Osteogenesis imperfecta and pneumatization of bone—
A hidden reality

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Osteogenesis imperfecta is a bone disorder in which three factors that influence bone strength (quality, mass, and geometry of osseous tissue) are affected. Pneumatization refers to the development of air-filled cavities inside the bone. Knowledge about these air-filled cavities provides valuable information for understanding the diagnosis and spread of various pathologic entities like hemangiomas, giant cell tumors, eosinophilic granulomas, and even metastatic tumor deposits. This paper presents a case in which radiologic, clinical, biochemical, and karyotypic evaluations lead to a diagnosis of osteogenesis imperfecta. ORTHODONTICS (CHIC) 2013;14:e150–e155. doi: 10.11607/ortho.947

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Osteogenesis imperfecta (OI) is a heterogenous group of conditions affecting bone mass and fragility. It is a highly variable disease that occurs secondary to an abnormality in type I collagen synthesis or extracellular secretion. Associated craniofacial and dental manifestations may include dentinogenesis imperfecta (DI), hypoplastic maxilla, and hypodontia. The steps in the diagnosis, modification in treatment planning, and clinical procedures are worthwhile to discuss to alert orthodontists about the various special needs of these individuals.

Pneumatization refers to the development of air-filled cavities inside the bone. Tremble1 reported that accessory air cells could be found in 10 sites within the temporal bone, and one of them is in the zygomatic process of the temporal bone. The pneumatized articular eminence of the temporal bone (PAT) is usually identified as an asymptomatic radiolucent defect in the zygomatic process of the temporal bone with an appearance similar to mastoid air cells. Prevalence of these defects using panoramic radiographs was reported in the range of 1.62% to 2.5%.5,6

PAT closely resembles aneurysmal bone cyst, hemangioma, giant cell tumor, eosinophilic granuloma, fibrous dysplasia, and metastatic tumor deposits.5,4,7–10 However, unlike PAT, these entities are symptomatic and characterized by expansile, destructive radiolucent lesions in the zygomatic bone. PAT is usually detected accidentally and must be differentiated from the abovementioned radioluencies within the zygomatic arch. The panoramic radiograph is an essential diagnostic record in orthodontics for routine examination of the teeth.
CASE REPORT

A 16-year-old male patient (normal gait, height 156 cm) reported to the department with a chief complaint of anteriorly displaced maxillary anterior teeth and spacing between the teeth (Fig 1). Pre-orthodontic treatment records such as impressions, profile photographs, and panoramic radiographs were recorded. Panoramic radiographs revealed a multilocular radiolucent lesion on the right articular eminence (Fig 2). Since the lesion was asymptomatic clinically, it was kept under observation. There were multiple pulp chamber obliterations in relation to the mandibular premolars that were later confirmed by intraoral periapical radiographs (Figs 3 and 4). Intraoral examination showed their texture and their surrounding structures. But sometimes asymptomatic bony lesions such as PAT may be missed during routine examinations.

The aim of this article is to present a case where the sequence of diagnosis—from the observation of a radiolucent lesion in the articular eminence area (PAT) through the performance of different diagnostic tests—leads to a final diagnosis of OI.
and color to be normal (Fig 5). A thorough physical examination revealed the presence of blue sclera (Fig 6). These initial radiographic and clinical findings suggested a bony disorder.

Blue sclera is commonly associated with three conditions: fetal rickets, OI, and osteopetrosis. All of these bone disorders show hematologic changes; therefore, to delineate these disorders, blood serum calcium, phosphorus, and alkaline phosphatase levels were investigated. Interestingly in this case, alkaline phosphatase was increased, and serum calcium and phosphorus were within normal range. Osteopetrosis was excluded from differential diagnosis since the acid phosphate would be increased.

Further, consultation with an orthopedic surgeon revealed a history of bowing legs, hyperlaxity of joints, and frequent ligament tear during childhood, which prevented him from playing outdoor sports. The radiographs of the long bones showed increased periosteal reaction (resembling Codman triangle) in the right fibula (Fig 7). In addition, the articular facets were radiopaque, whereas trabeculae and cortices were normal.

Patients with fetal rickets have cupping and splying of the knee joint. In patients with subclinical fetal rickets, the ulnar bone will be involved in spite of normal blood investigation. Both of these findings were absent in the present case; therefore, the diagnosis of fetal rickets was excluded (Figs 8 and 9).
Patients with osteogenesis imperfecta may show presence of wormian bones, usually small and few in number, which never fuse with the surrounding major bones over the course of the patient's life. These can be visualized in a posteroanterior radiograph, which was performed to confirm their presence in this case (Fig 10). There exist many other craniofacial syndromes involving bones that would have similar features; therefore, genetic karyotyping was considered. The karyotyping result in this individual was normal (Fig 11), which excluded other craniofacial syndromes. The karyotyping only detects abnormality in chromosome number, but OI is a genetic disease affecting the COL1A1 and COL1A2 genes. Although gene mapping is required for definitive diagnosis in such cases, many researchers still do not consider gene mapping as a specific diagnostic test for OI. In this particular case gene mapping was not done after evaluating the cost-benefit ratio.

Because the radiolucent lesion in the right articular eminence was asymptomatic and nondestructive for 6 months, a diagnosis of PAT was made.

**DISCUSSION**

In OI all three factors that influence bone strength, ie, quality, mass, and geometry of osseous tissue, are affected. This leads to a decrease in bone strength and resistance to fracture. Further presence of PAT makes bone more porous. Knowledge of PAT is of particular relevance to orthodontists because it resembles pathologies radiographically. It causes minimal resistance in bone and facilitates the spread of various pathologic lesions such as tumors into the joint, inflammation, and fractures. Panoramic radiography is considered the preliminary method to diagnose these defects due to economical factors and the low radiation dose when compared to computed tomography (CT).

The presence of PAT in the temporomandibular joint (TMJ) has been associated with acute temporal bone fracture, implying that surgical procedures involving a TMJ affected by PAT can be complicated. Individuals with PAT can have perforation owing to firm dissection or forceful flap retraction during TMJ surgeries.
In certain individuals, TMJ dysfunction might require a fossa-eminence prosthesis to correct the effects of the pathology. When PAT is situated between the condyle and skull base, perforation may occur as a result of the screws. Hence, such defects should be evaluated thoroughly using a CT scan before complex surgeries are planned.

Certain procedures like orthognathic surgery, implant placement, and orthodontic tooth movement may be critical because of the nature of bone growth and healing in OI individuals. Implant failures were reported to be most likely in such individuals. Therefore, consideration should be given to fixed partial dentures in lieu of dental implants. The important consideration for the orthodontist is that all OI individuals will have some degree of compromised dentinal structure. The treatment plan should be modified to minimize heavy stress on the teeth, especially during debonding, which could lead to enamel fracture. Alternatively, this can be treated by banding of such teeth.

PAT is considered in the differential diagnosis of many pathologies of zygomatic bone, but the literature does not suggest any association between pneumatization and OI. PAT may be associated with OI, or it may be just an accidental finding in this case. Further investigation is required to delineate the role of PAT in such disorders.

CONCLUSION

In this case the sequence of diagnosis from the observation of a radiolucent lesion in the articular eminence area through the performance of different diagnostic tests led to the final diagnosis of OI. It highlights the fact that bony disorders like OI can affect orthodontic treatment plans and outcomes as well as the importance of radiologic findings like PAT that may appear in the diagnostic records of orthodontic patients. Orthodontists should be aware of such conditions to successfully modify the treatment plan and improve the individual’s outlook with a perfect smile.
REFERENCES