Dentinogenesis Imperfecta in patient with Osteogenesis Imperfecta: Case Report

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INTRODUCTION: ETIOLOGY AND EPIDEMIOLOGY

Osteogenesis Imperfecta (OI) is a genetic disorder of type I collagen, characterized by bone fragility, leading to frequent bone fractures. OI is classified into four basic types I, II, III, and IV.[7] OI most commonly has autosomal dominant inheritance, but it can also present due to autosomal recessive, or sporadic mutations. The prevalence of OI is 4-10 in 100,000 individuals[3].

Dentinogenesis Imperfecta (DI) is a genetic disorder affecting the development of dentin that can be present alone (DI Type II or III) but is also present in about 50% of patients with OI types III or IV (DI Type I). [3,1]
The incidence of DI is 1 in 8000.[1]

DIAGNOSIS AND MANAGEMENT

Diagnosis is made by DNA analysis of the COL1A1 gene and clinical features. Patients with OI type III often present with severe bone fragility and deformity, very slow growth, blue sclera, DI, and loss of hearing.[3] DI presents clinically with amber translucence or gray discoloration of crowns and enamel fractures due to weak underlying dentin.[3] Radiographically, crowns are bulbous, roots are short, and pulp canals are obliterated. Patients with DI will commonly present with midface hypoplasia resulting in class III malocclusion and unilateral or bilateral crossbites.

Intravenous bisphosphonates (IVBP) are antiresorptive medications designed to increase bone strength in many conditions, including OI.[6] While these medications can improve the quality of life for patients, they also pose a risk of medication-related osteonecrosis of the jaw (MRONJ).[6] MRONJ is "the progressive destruction and death of bone that affects the mandible and maxilla of patients exposed to treatment with medications known to increase the risk of disease, in the absence of a previous radiation treatment".[4]

The risk of developing MRONJ among osteoporotic patients treated with to IVBPs or RANK-L inhibitors is 0.02-0.3%.[8] However, in two systematic reviews with a combined total of over 500 children treated with BPs, no cases of MRONJ were reported. [8]

CASE REPORT

This case report describes the dental management of an 11-year-old female with a medical history significant for OI type III, DI, cervical kyphosis status post occiput to thoracic arthrodesis. She presented to the dental clinic at Healthy Smiles for Kids (HSK) of Orange County for a dental consultation. Her medical history includes prior history of right and left femur fractures. She also endured a humerus fracture after spinal surgery possibly caused by the blood pressure cuff. Her medical condition is currently managed with Valium as needed and IV hydrocodone/pamidronate every three months.

CLINICAL AND RADIOGRAPHIC FEATURES

Clinical examination was significant for overall small stature, active carious lesions, existing restorations, and over retained primary teeth. The patient's teeth had generalized amber translucence with localized missing enamel. Due to the patient's extensive spinal surgeries, extension and flexion of the neck was limited.

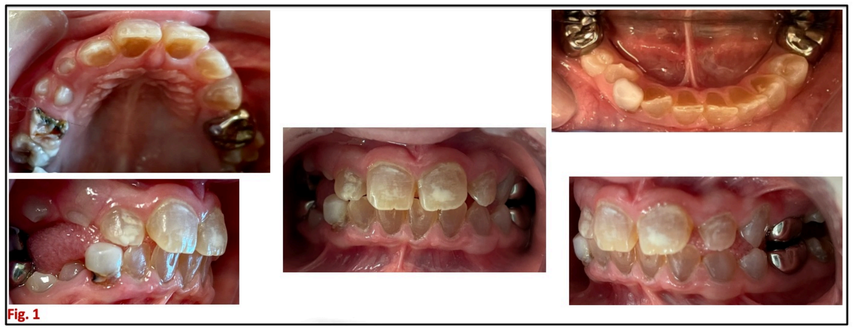
Fig. 1

Figure 1. Intraoral photos exhibiting amber translucent crowns and break down of permanent molars

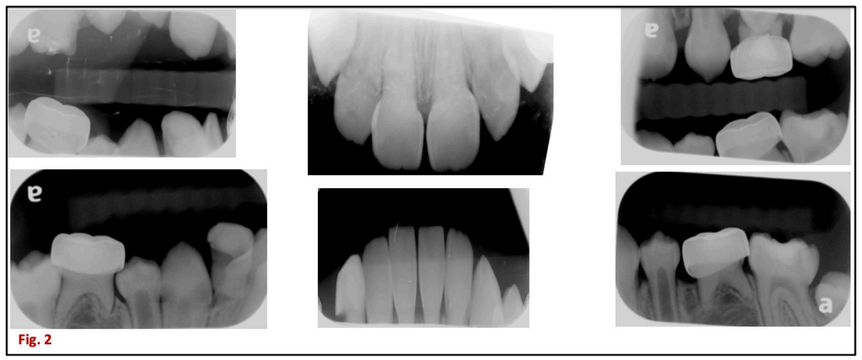
Fig. 2

Figure 2. Evidence of variable pulp canal obliteration and large pulp chambers, short roots of primary teeth with possible missing permanent mandibular premolars, and caries

TENTATIVE TREATMENT PLAN

The patient is planned for comprehensive dental treatment under general anesthesia. Consultations were sent to her pediatrician, endocrinologist, and orthopedist regarding necessary special considerations, precautions, and timing. The orthopedist recommended "extreme caution with patient handling to reduce fracture risk." The endocrinologist also advised using chlorhexidine after treatment for several days.

The HSK management of OI protocol has been adapted from protocols intended for adults[2] and in consultation with the OI clinic in Omaha, Nebraska. For patients receiving long-term bisphosphonate therapy (>2years):

- All invasive treatment (including extractions), must be scheduled 3 weeks prior to the next bisphosphonate infusion.
- Antibiotic prophylaxis should be considered if treatment includes surgical or traumatic procedures, especially sites with active infection.
- Consideration for antibiotic therapy for patients under long-term bisphosphonate therapy:
  - >14 years old with an infusion within the last 5 years
  - <14 years old with an infusion within the last 2 years
- If possible, delay elective procedures until cessation of bisphosphonate therapy.
- Procedures should be scheduled as long as possible from the last infusion with a 3-week healing period for any surgical wounds prior to the next infusion.
- 0.12% chlorhexidine gluconate mouth rinse should be used twice per day for 5 days before and after surgery or until the surgical site has healed.
- Any extraction sites should be sutured.

To reduce the risk of MRONJ, our patient's dental surgery was scheduled at least 1 month before the next infusion to allow healing post-extractions. Chlorhexidine was prescribed and she was instructed to rinse 2 times per day, five days prior to and after surgery.

Unfortunately, due to coordination of dental treatment with bisphosphonate therapy, the pandemic, and other illnesses, our patient's treatment has been postponed. A tentative date for her treatment will be scheduled prior to her next infusion. Based on current clinical and radiologic examination, mobile primary teeth will be extracted and the permanent molars are planned for SSCs.

CONCLUSION

This case report presents a patient with OI and DI and demonstrates the complex multidisciplinary approach that is necessary to provide appropriate care. A team of geneticists, endocrinologists, orthopedic surgeons, physical therapists and pediatric dentists should work together in coordinating patient care. Patients with OI and DI should establish a dental home to begin planning long-term management. Ideally, a thorough clinical and radiographic dental examination should be completed prior to the initiation of bisphosphonate therapy to plan invasive treatment appropriately. Preventive recommendations should be emphasized: maintaining good oral hygiene, nutritional counseling, and routine oral examinations.

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