

Case Report: Five-Year Follow-Up of a Patient with Familial Expansile Osteolysis

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Background

- Familial Expansile Osteolysis (**FEO**) is a rare autosomal dominant disease with key features that include abnormal tooth root resorption, bone dysplasia, & middle ear deafness.
- It has been documented in 3 unrelated family lineages (German, Irish, and American), & 2 individual American cases in 2003.
- Bone pain usually develops prior to the appearance of skeletal abnormalities and persists throughout the progression of the disease.
- FEO is caused by a gene mutation in **TNFRSF11A**, which encodes the receptor activator of NF-kappaB (**RANK**), resulting in abnormal osteoclast activation. This can result in problems with bone remodeling in several bones in the body, leading to pathologic fracture.
- External cervical root resorption (**ECR**) is a dynamic and aggressive loss of mineralized tooth structure seen in a permanent tooth's cemento-enamel junction. It is asymptomatic, can be an incidental finding, and radiographically is seen as asymmetrical with ill-defined borders.
- The findings associated with Familial Expansile Osteolysis and treatment build on the case report published by Macaraeg et al. (2020).**

Initial Findings (2019)

- 10-year-old female (S.M) arrives at Penn Dental Pediatrics
- Chief Complaint:** "We were referred here by the orthodontist because she has early resorption of primary teeth and root resorption of her permanent teeth."
- Medical and dental history:** Missing ossicles (profoundly deaf), finished phase 1 ortho
- Panoramic image was provided by her orthodontist from December 2018



Panoramic Radiograph December 2018



Periapical Radiograph
March 2019

- External root resorption involving the apical third of the root of the permanent mandibular central incisors is seen
- Referral to Genetics & Endocrinology Teams at Children's Hospital of Philadelphia**
- Confirmed Diagnosis: Familial Expansile Osteolysis (FEO)**
- Identified TNFRSF11A gene mutation**

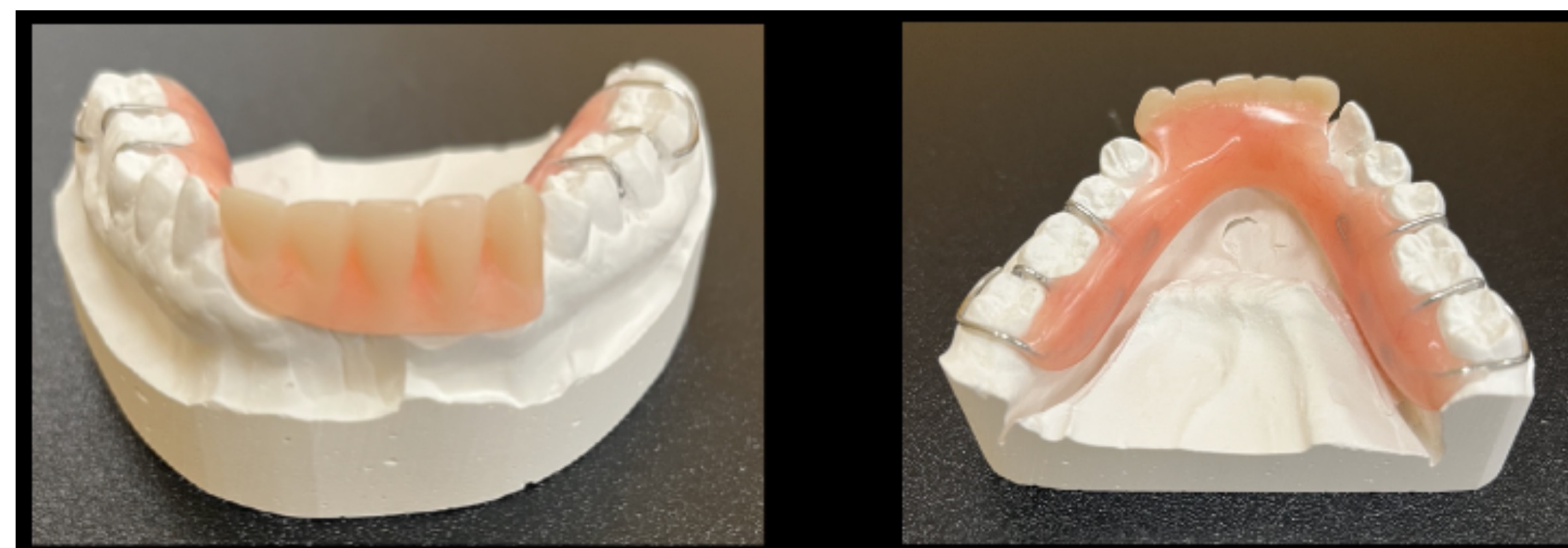


Intraoral Photograph
March 2019

Progression & Treatment

Treatment Rendered:

- Decorations**
 - June 14, 2021: #7, 10
 - December 4, 2023: #11, 22
- Extractions**
 - August 30, 2019: #24, 25
 - April 7, 2022: #8, 9
- Vital Pulp Therapy**
 - June 7, 2024: #19
- Surgical History:**
 - Arthroscopy Knee Meniscal surgery (2021)
 - Achilles Tendon Rupture (2023)
 - Ruptured triceps tendon (2023)
- S.M. was given an upper and lower removable partial denture in August 2019. Following subsequential loss of teeth, additional teeth have been added to the partial.
- S.M. began IV bisphosphonates following her diagnosis, which helped systematically.
 - Switched to oral bisphosphonates to hopefully aid with implant placement in the future.
- S.M. reports sensitivity and pain to cold, but all teeth test clinically vital.

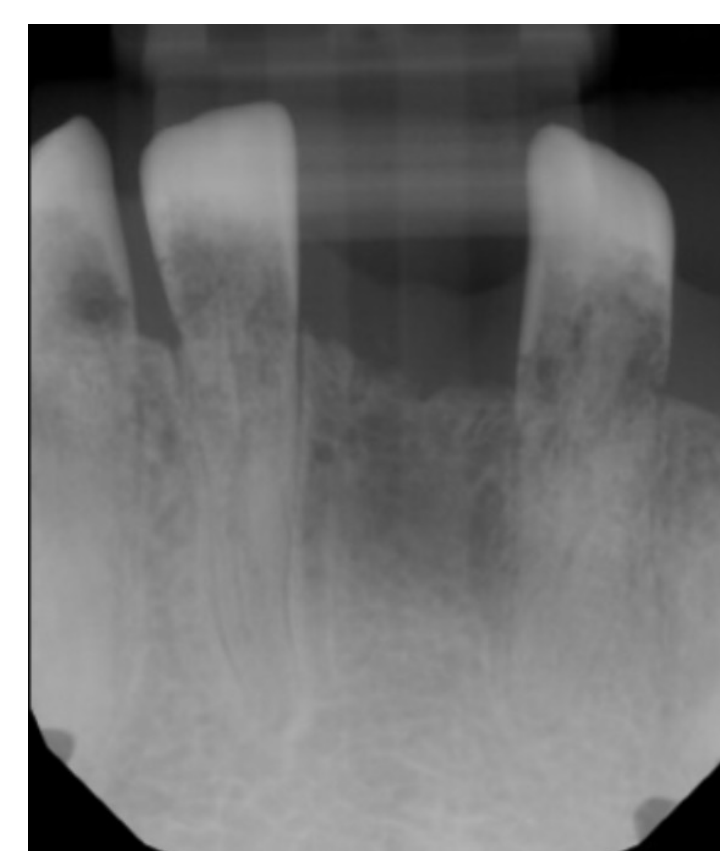


Updated Findings (2024/2025)

- Chief Complaint:** "Two month ago, I had pain in my lower front and left teeth. Currently, I do not have pain in my teeth after seeing the endodontic resident."
- Updated medical and dental history:**
 - Daily medications: Alendronate tablets 10mg, Calcium Carbonate tablets 500mg, & Vitamin D3 capsules
 - The resorption progress pattern matches the dental eruption sequence.**
- Treatment goals:**
 - Manage her symptoms based on pain and progression
 - Continue use of upper and lower removable partial denture



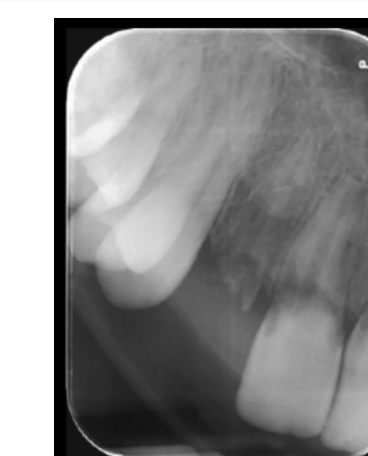
Intraoral Photographs
January 2025



Periapical Radiograph of
Remaining Permanent
Mandibular Incisors
January 2025

Radiographic Imaging

Example of Decoronation of tooth #7



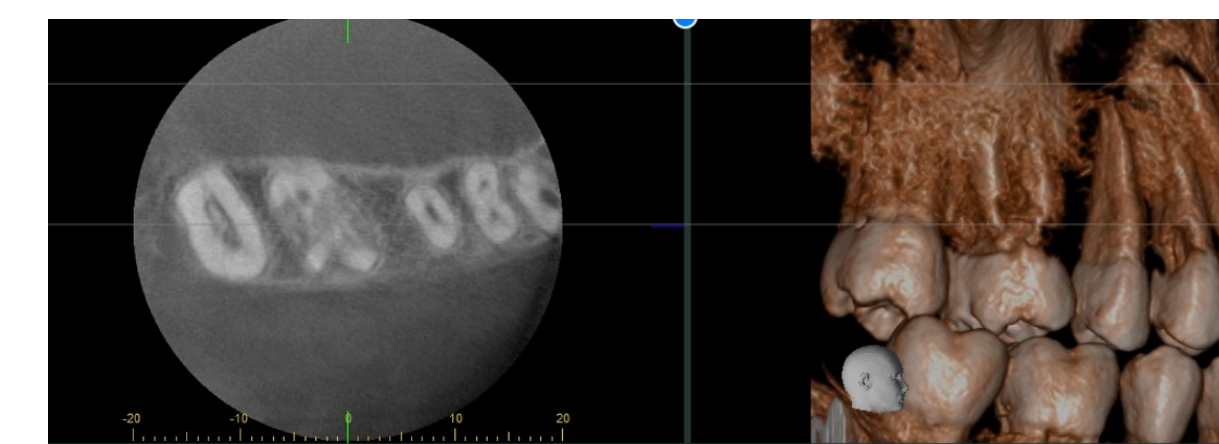
June 2021

Example of Vital Pulp Therapy of tooth #19



June 2024

Example of the rate of progression for external root resorption of tooth #3 (asymptomatic to pain)



August 2023

August 2024

January 2025

January 2025

Future Direction

- All treatment options performed have been unsuccessful in stopping the progression.**
- With the number of teeth affected, the surgical treatment of multiple ECR lesions has been challenging.
- At 16 years old, S.M. has a very high pain tolerance and tries to avoid cold foods & water
- Previously when the pain was unbearable, decoronation or extraction of the tooth helped.
- We anticipate that she will lose her teeth. Patient's pain and comfort are prioritized as the disease progresses.**
- Extracted teeth from previous years were sent to the National Institute of Dental and Craniofacial Research.
- Moving forward, future research is needed to determine the potential use of intracanal bisphosphonates and success of future implants.

References

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