Oral Manifestations in a Patient with Rett Syndrome: Case Report

CASE WESTERN RESERVE UNIVERSITY
School of Dental Medicine

Nicole Piening DDS; Gerald Ferretti DDS, MS, MPH; Margaret Ferretti DMD, MPH; Ying An DDS, PhD; Maria Botros, DDS
Department of Pediatric Dentistry, Rainbow Babies and Children's Hospital
Case Western Reserve University School of Dental Medicine

University Hospitals Rainbow Babies & Children's
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Introduction

Rett Syndrome is a genetic neurological and developmental disorder that primarily affects females and disrupts normal brain development. Rett syndrome are caused by a mutation in the MECP2 gene. It leads to a loss of motor skills and language. Typically, babies with Rett syndrome develop normally during the first 6-9 months, after which they begin to lose previously acquired abilities, such as crawling, walking, and communicating. Oral manifestations of Rett Syndrome can include a high palate, anterior open bite, bruxism, gingivitis, periodontitis, gingival hyperplasia, xerostomia, glossitis, erythema multiforme, and delayed eruption of teeth.

In this case report, we present a case of a 15-year old female patient with Rett Syndrome who exhibits delayed eruption of permanent teeth.

Case Report

A 15-year-old patient was consulted at University Hospitals Rainbow Babies & Children's Hospital in August 2024 with a chief complaint of primary teeth not exfoliating. Mom reported patient has not seen a dentist in 4 years and the previous dentist did not want to extract at that time due to her medical history. Mom reported the patient has lost a couple of baby teeth since last dental visit. Mom reported the patient has a special diet that consist of no crunchy food or food needs to be cut into small pieces. Past medical history is significant for Rett Syndrome, severe malnutrition, non-verbal, developmental delay and possible seizures. The patient is currently going through evaluations for seizure diagnosis. The patient is currently taking calcium carbonate, cholecalciferol and acetaminophen.

A limited exam was completed at time of consult and it was determined due to the extent of treatment and patient cooperation a comprehensive oral examination was needed under general anesthesia (GA). Extra-oral exam reveals microcephaly with normal facial features. Clinical examination revealed the patient has generalized unerupted permanent teeth, with most primary teeth still present (A, B, C, E, F, G, H, I, J, K, L, M, R, S, T) (see Figure 2A,B), attrition, gingivitis, and calculus. No soft tissue pathology. Clinical caries were present on teeth 7, 8,9, 10. Radiographic exam reveals the permanent teeth exhibited fully formed roots with a delayed eruption pattern (Figure 2C-I), over-retained primary teeth and confirmed finding of caries on 7, 8, 9, 10.

The following treatments were completed on the patient: Extractions were completed on (A, B, C, E, F, G, H, I, J, K, L, M, R, S, T) due to being over-retained and caries. Two 3.0 chromic sutures were placed on LL and LR to help with healing and gum tissue approximation. Due to caries, composite restorations were placed on 7-DIFL, 8-IF, 9-ILF, 10-IF. Tooth 9 received an IDPC with Theracal. The patient also received a prophylaxis cleaning and fluoride treatment.

The patient will be followed by pediatric dentistry on a 2 year OR recall. Emphasized to mom the restorations may fracture due to bruxism. Future treatment includes full resin crowns on 7, 8, 9, 10. The patients delayed eruption will continue to be monitored.

Clinical and Radiographic Presentation

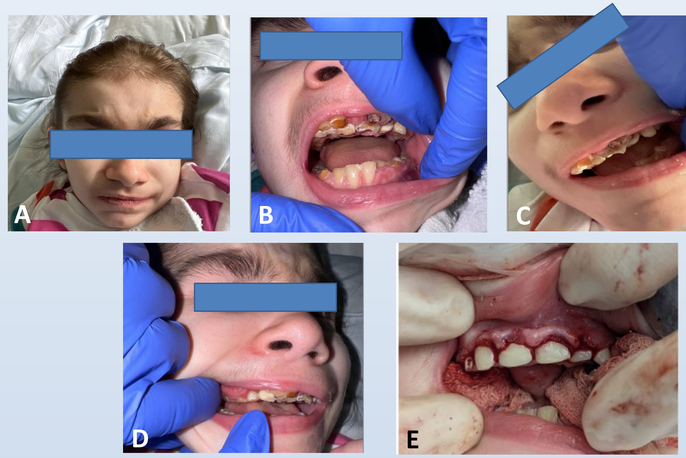

Figure 1 Extraoral and intraoral photographs of the patient

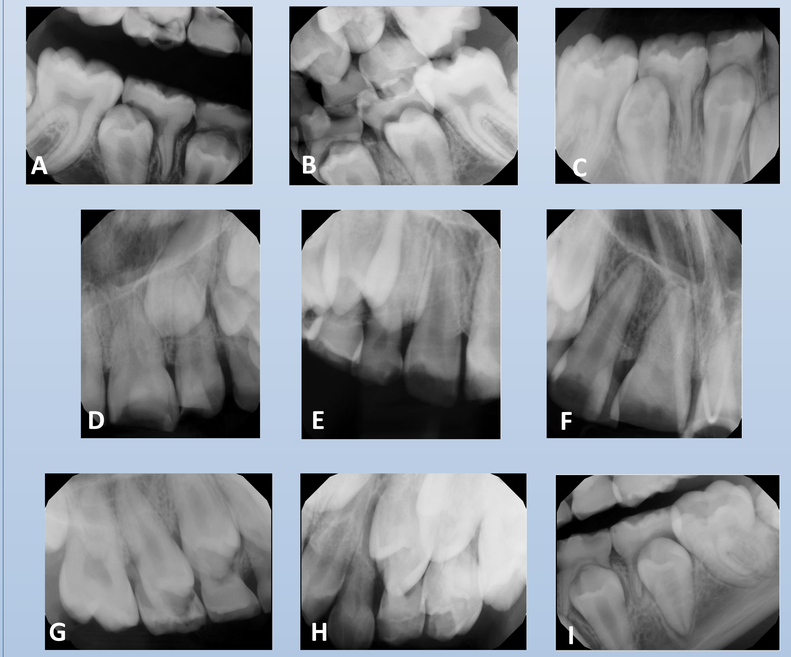

Figure 2 A-B Intraoral bitewings taken on 12/12/2024. C-I intraoral periapical radiographs on posterior and anterior regions.

Dental and Craniofacial Manifestations

Following dental and craniofacial anomalies were noted in our patient which is typical in a patient with Rett syndrome:

- Microcephaly
- Bruxism
- Delayed eruption of permanent dentition
- Gingivitis
- High arched palate
- Drooling

Discussion

Rett syndrome is a rare neurodevelopmental disorder associated with common oral health issues, including bruxism, poor oral hygiene due to motor dysfunction, increased risk of periodontal disease and caries, as well as habits like tongue thrusting and mouth breathing, contributing to malocclusion. Hypersalivation and swallowing difficulties further complicate oral health.

Although delayed permanent teeth eruption is observed in patients with Rett syndrome, generalized delayed unerupted permanent teeth can also be an indicator for endocrine imbalance such as: hypothyroidism, hypoparathyroidism, or growth hormone deficiency, which are not uncommon in such cases. Based on pediatric dental team's recommendation, patient's endocrinologist ran the lab work to rule out GH deficiency hypothyroidism and hypoparathyroidism. Her delayed eruption was ultimately determined to be caused by her severe malnutrition. This case highlights the importance of comprehensive health assessments in patients with delayed tooth eruption, as identifying underlying causes—whether nutritional, endocrine, or genetic—can guide appropriate interventions. Increased awareness among healthcare professionals and caregivers can lead to better management strategies and improved quality of life for those living with Rett syndrome.

The desired outcome for this patient would be for the teeth to erupt on their own. In this case, we hope the conditions will be improved after patient's severe malnutrition problem is resolved. However, if this does not happen, the prognosis of the teeth eruption is poor. In severe cases, surgical exposure may be needed. If teeth fail to erupt completely, prosthetic rehabilitation with implants, bridges, or dentures ensures function and aesthetics. Due to her medical history prosthetics may not be the best option. A multidisciplinary approach involving dentists, nutritionists, and medical specialists is essential for comprehensive care.

References

1. Mahdi, Syed Sarosh, et al. "Oral Manifestations of Rett Syndrome—A Systematic Review." *International Journal of Environmental Research and Public Health*, vol. 18, no. 3, 2021, p. 1162. National Center for Biotechnology Information
2. National Institute of Neurological Disorders and Stroke. (n.d.). Rett syndrome. *National Institutes of Health.*
3. Mayo Clinic. (n.d.). Rett syndrome. *Mayo Clinic.* https://www.mayoclinic.org/diseases-conditions/rett-syndrome/symptoms-causes/syc-20377227
4. Modafferi C, Tabolacci E, Grippaudo C, Chiurazzi P. Syndromic and Non-Syndromic Primary Failure of Tooth Eruption: A Genetic Overview. Genes (Basel). 2025;16(2):147. doi:10.3390/genes16020147.