Dental Treatment for Patients with Rett Syndrome: A Case Report

Anjelica Tyrell, DDS[1], Brittany Waters, DMD[1,2]

[1]Division of Dentistry, Children's Healthcare of Atlanta, Atlanta, GA, [2]Department of Pediatrics, Emory University School of Medicine, Atlanta, GA

Abstract

Introduction: Rett Syndrome (RS) is a rare, progressive neurodevelopmental disorder, primarily affecting females, caused by gene mutations of MECP2 on the X chromosome impacting neurologic development and cognitive function. RS is characterized by a period of normal development followed by regression and loss of previously acquired behavioral, motor and cognitive skills, muscular hypotonia, epilepsy, scoliosis, and autistic traits.

Case Report: This case report details the dental rehabilitation completed at Children's Healthcare of Atlanta for an eight-year-old female diagnosed with Rett Syndrome. Prompt treatment was recommended to mitigate the risk of infection and address suspected pain from a primary tooth fractured due to caries. The full mouth dental rehabilitation completed in an operating room under general anesthesia will be reviewed, along with its rationale, and underlying medical implications associated with this condition.

Background

Rett Syndrome (RS) has an incidence of 1 in 10,000 to 15,000 female births. RS cases in males are extremely rare and result in severe phenotypes, often leading to early mortality. A Rett Syndrome diagnosis is based on genetic testing and clinical features.[3]

There are four stages of Rett Syndrome:

1. **Early Onset stage:** This stage lasts from six to eighteen months of life, during which the child develops normally and shows few symptoms.[2]
2. **Regression/Rapid Destructive stage:** At the onset of the rapid destructive stage, the child begins to lose previously acquired skills, including purposeful hand movements, speech, and social interaction. Motor difficulties, including tremors, rigidity, and ataxia are also common.[2]
3. **Plateau/Pseudo-stationary stage:** The rate of decline slows in this stage, and some recovery and improvements are observed. However, there is a high likelihood patients will continue to have cognitive and motor impairments, including epilepsy.[2]
4. **Late Motor Deterioration stage:** This stage typically starts after ten years of age. Children develop more severe physical disabilities, such as scoliosis, seizures, and respiratory problems.[2]

Systemic Manifestations

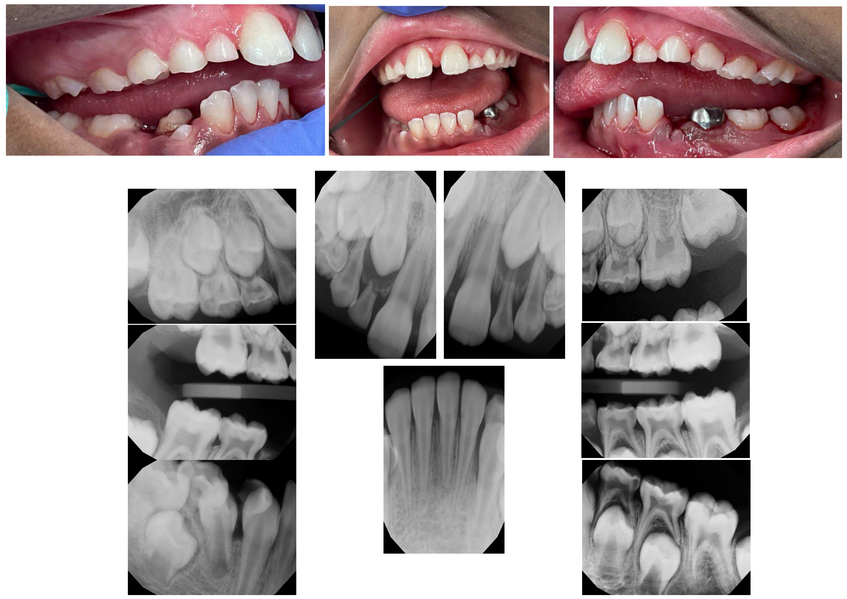
Rett Syndrome affects multiple systems, including the gastrointestinal, cardiovascular, musculoskeletal and central nervous systems.

Neurological and Musculoskeletal: Neurological manifestations of RS are seen in all four stages. In the early stages, children have delays in reaching developmental milestones, hypotonia, and distinctive repetitive hand movements like hand wringing and clapping. As the syndrome progresses, 90% of children will have seizures, varying from non-convulsive status epilepticus to generalized seizures, including tonic and focal onset. Additionally, children may have apnea during wakefulness and sleep, as well as excessive somnolence. Over time, children will experience cognitive decline, loss of social skills, and inability to perform tasks independently. In the later stages, musculoskeletal problems like gait abnormalities, dystonia, and scoliosis become more prevalent. Children have tremors, lack of coordination, an unsteady or wide-based gait, and an eventual inability to walk independently.[5]

Cardiovascular: Cardiovascular involvement is common in children with RS due to dysregulation of the autonomic nervous system. Some manifestations include bradycardia, tachycardia, QT prolongation, and arrhythmias, including life-threatening arrhythmias, such as Torsades de Pointes and ventricular fibrillation. Due to the risk for QT prolongation, routine ECG monitoring and avoiding medications that prolong the QT interval is recommended.[2,4]

Gastrointestinal: Gastrointestinal issues affect 75% of children with RS.[8] Complications include dysphagia, gastroesophageal reflux disease, constipation, and abdominal distension. Severe gastrointestinal issues, such as chronic erosive esophagitis or severe reflux, lead to feeding and nutrition challenges. There is an increased risk of chronic pulmonary complications due to aspiration.[4,7]

Case Report



Chief Concern: "My daughter's bottom right tooth broke off while eating this weekend. Unsure if she is in pain due to her being non-verbal."

Medical History: 8-year-old female with Rett syndrome, developmental delay, fine motor delay, periodic limb movements of sleep, gait abnormality, sleep disorder, spasticity, hypotonia, autism spectrum disorder, seizure disorder

Dental History: Patient routinely clenches and grinds teeth, congenitally missing maxillary permanent lateral incisors

Medications: Trofinetide, Clonazepam, Baclofen, Diazepam, Levetiracetam, Ondansetron, Calcium carbonate

Social History: Lives at home with mother and father

Oral Hygiene: Parents brush patient's teeth 1-2x/day with fluoride toothpaste, do not floss

Diet: Patient has a varied diet by mouth, drinks water, juice, and sports drinks, routinely pouches food in her cheeks

Caries Risk Assessment: High

Extraoral Exam: Facial symmetry, no swelling or tenderness, no lesions observed on lips or face

Intraoral Exam: Mixed dentition, generalized attrition, staining on first permanent molars, dental caries

Treatment Plan: The Children's Healthcare of Atlanta pediatric dentistry department recommended comprehensive dental rehabilitation in the operating room under general anesthesia. The treatment plan aimed to remove the fractured and carious tooth #S to reduce the risk of infection. After obtaining a full mouth series of intraoral radiographs and completing a dental exam in the operating room, we aimed to restore optimal oral health by treating existing dental caries, providing preventive treatment in the form of sealants on the first permanent molars, and completing interceptive extractions of the maxillary primary lateral and canines.

Completed Treatment: extractions: #C, #D, #G, #H, #M, #S, stainless steel crown: #L, preventive resin restoration: #3-O, sealants: #A, #B, #I, #J, #K, #T, #14, #19, #30, dental prophylaxis, and fluoride varnish application

Craniofacial And Dental Manifestations

Craniofacial: Microcephaly is a common feature in children with RS, characterized by a head circumference smaller than normal due to reduced brain growth. This condition becomes apparent during early childhood as brain development slows over time. Conversely, some children initially present with normal head size but later develop macrocephaly during their first year of life. This enlargement of the head may remain as the child grows.[6]

Children with RS may have a distinctive facial appearance with a small chin, thin upper lip, downturned mouth, and a high, narrow forehead. Often there is reduced expressiveness in facial movements due to deficient neuromuscular function. Abnormalities in posture and coordination (ataxia) are frequent and may affect craniofacial alignment and overall appearance. [6]

Dental: The most common dental complications reported in patients with RS is bruxism and non-physiological tooth wear. The high prevalence of bruxism is associated with involuntary muscle movements resulting from neurodevelopmental and neuromuscular disturbances. Development of malocclusions and unfavorable growth patterns is influenced by the behavioral and physical characteristics associated with RS. Incorrect mandibular positioning and oral habits like thumb sucking, mouth breathing, and tongue thrusting can contribute to high arched palates and malalignment of the dentition, maxilla, and mandible.[1]

Children with RS may have oral manifestations of nutritional deficiencies. Poor coordination of oral musculature may result in sialorrhea. Conversely, pharmacological management of RS may induce side effects such as xerostomia, glossitis, and gingivitis. The high incidence of caries in this patient population is multifactorial. [1]

Conclusion

Rett Syndrome is a rare genetic condition that has systemic and oral implications. This case report reinforces the need for dental professionals to be vigilant in the early dental management in individuals with RS. By recognizing the high risk of bruxism, malocclusion, and low salivary flow in patients with RS, pediatric dentists can implement early preventive strategies such as fluoride treatments, customized oral hygiene plans, and scheduled routine examinations to evaluate for malocclusion or caries. Proactive dental care and continued dental education can help minimize the need for invasive treatments and improve the overall oral health of patients with RS.

References

1. Al-Hathlol W, Bokhari R, Alzahrani N, Alkuwaiti E. Challenges in the Dental Management of Rett Syndrome under General Anesthesia: A Rare Disease. Case Rep Dent. 2022;2022:4038221. Published 2022 Feb 1. doi:10.1155/2022/4038221
2. Bricker K and Vaughn BV (2024) Rett syndrome: a review of clinical manifestations and therapeutic approaches. Front. Sleep 3:1373489. doi: 10.3389/frsle.2024.1373489
3. Hagberg B, Aicardi J, Dias K, Ramos O. A progressive syndrome of autism, dementia, ataxia, and loss of purposeful hand use in girls: Rett's syndrome: report of 35 cases. Ann Neurol. 1983;14(4):471-479. doi:10.1002/ana.410140412
4. Kyle SM, Vashi N, Justice MJ. Rett syndrome: a neurological disorder with metabolic components. Open Biol. 2018;8(2):170216. doi:10.1098/rsob.170216
5. Lopes AG, Loganathan SK, Caliaperumal J. Rett Syndrome and the Role of MECP2: Signaling to Clinical Trials. Brain Sci. 2024;14(2):120. Published 2024 Jan 24. doi:10.3390/brainsci14020120
6. Mahdi SS, Jafri HA, Allana R, Amenta F, Khawaja M, Qasim SSB. Oral Manifestations of Rett Syndrome-A Systematic Review. Int J Environ Res Public Health. 2021;18(3):1162. Published 2021 Jan 28. doi:10.3390/ijerph18031162
7. Somerville H, Tzannes G, Wood J, et al. Gastrointestinal and nutritional problems in severe developmental disability. Dev Med Child Neurol. 2008;50(9):712-716. doi:10.1111/j.1469-8749.2008.03057.x
8. Tan, X., Zhu, B., Li, Y. et al. An ambulatory dental treatment of a child with Rett syndrome and limited mouth opening under muscle relaxant-free general anesthesia: a case report. BMC Anesthesiol 24, 3 (2024). https://doi.org/10.1186/s12871-023-02379-4