Dental and Systemic Findings in Williams–Beuren Syndrome

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Introduction

Williams–Beuren syndrome (WBS) is a rare multisystem genetic disorder resulting from a **microdeletion of chromosome 7q11.23**. WBS is characterized by:

- **Cardiovascular anomalies** (e.g., bicuspid aortic valve).
- **Neurodevelopmental impairment** and endocrine dysfunction (e.g., hypothyroidism).
- **Distinctive orofacial features** and oral manifestations including enamel hypoplasia, tooth agenesis, and increased caries risk.

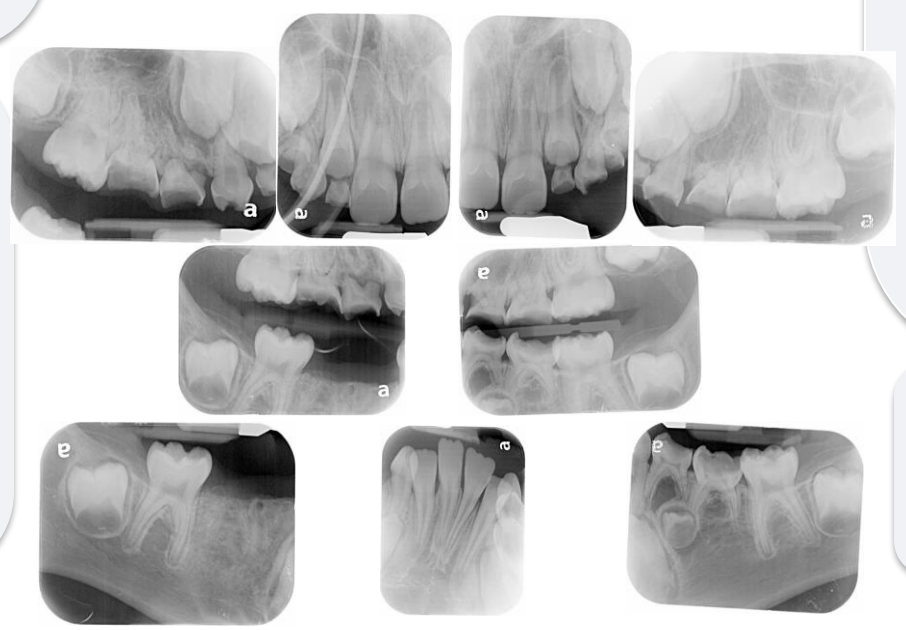
Case Report

- **Patient:** 7-year-old male with genetically confirmed Williams–Beuren syndrome
- **Medical History:** Developmental delay, autism spectrum disorder, hypothyroidism, feeding difficulties, and bicuspid aortic valve (cardiology clearance obtained prior to treatment)
- **Clinical Findings:** Poor oral hygiene, generalized enamel hypoplasia, high caries risk, and multiple grossly carious and fractured primary teeth
- **Behavioral Assessment:** Significant anxiety and limited cooperation precluded conventional in-office treatment
- **Chief Concern:** Caregiver-reported pain and difficulty eating

Radiographic Finding

full-mouth radiographic evaluation revealed multiple congenitally missing permanent teeth (#4, #5, #12, #13, #20, #28, #29; Universal numbering system).

Additional findings included extensive carious involvement, pulpal pathology, and compromised tooth structure among primary dentition, supporting the need for comprehensive treatment under general anesthesia.



Treatment

Comprehensive oral rehabilitation under general anesthesia due to extent of disease, behavioral limitations, and medical complexity

Procedures Performed:

- **Extraction of non-restorable primary teeth**
- **Placement of occlusal composite restorations on permanent teeth (#3, #14, #19, #30)**
- **Full-mouth debridement and fluoride application**

Postoperative Management: Caregiver received anticipatory guidance, dietary counseling, and oral hygiene instruction; patient placed on high-risk caries recall schedule

Patients with Williams–Beuren syndrome present with multiple risk factors for poor oral health, including enamel hypoplasia, tooth agenesis, and increased caries susceptibility. Developmental delay and behavioral challenges further complicate routine dental care and often necessitate treatment under general anesthesia.

Cardiovascular anomalies require thorough preoperative medical evaluation to minimize anesthesia-related risks. Early dental intervention is critical in this population. Preventive strategies—including fluoride therapy, dietary modification, and caregiver education—play a key role in reducing disease burden.

Clinical Significance

Pediatric dental providers are often among the first to identify oral manifestations of systemic conditions such as WBS. Early recognition allows for timely referral, coordinated care, and improved overall health outcomes.

References

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