

Case Report: Dental Considerations in a Pediatric Patient with Waardenburg Syndrome

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Introduction

Waardenburg syndrome is a rare hereditary condition caused by mutations that affect the development of neural crest cells, resulting in abnormalities in melanocyte distribution and craniofacial formation. Common clinical manifestations include sensorineural hearing loss, pigmentary disturbances of the hair, skin and eyes, as well as distinct craniofacial features such as dystopia canthorum (eyes set further apart), broad nasal root, and medial eyebrow flare. Although not a primary feature, dental anomalies such as hypodontia, peg-shaped lateral incisors, and delayed eruption have been reported, reflecting the underlying disruption in neural crest cell development.

Though estimated prevalence is approximately 1 in 40,000, clinical presentation remains highly variable, resulting in potential undiagnosis of milder forms. As a result, these patients may present in dental settings without a proper medical diagnosis. Because of their involvement in routine and emergency care, pediatric dentists are uniquely positioned to identify craniofacial and phenotypic abnormalities, especially in cases involving trauma where comprehensive evaluation is required. Failure to recognize syndromic features may lead to incomplete medical histories, inappropriate treatment planning, and missed opportunities for interdisciplinary referral. This case highlights the role of the pediatric dental provider in recognizing clinical signs suggestive of underlying genetic conditions and facilitating appropriate referral and care coordination.

Objective

- Identify the characteristic craniofacial dysmorphism and pigmentary abnormalities of Waardenburg syndrome during pediatric dental examinations.
- Evaluate the clinical significance of early detection and the necessity of interdisciplinary referral to mitigate diagnostic delays and secondary complications
- Integrate syndrome-specific dental anomalies; including eruption disturbances, hypodontia, and structural defects into a comprehensive, multidisciplinary treatment plan.

Case Presentation

A 10-year-old female presented for an emergency dental visit following traumatic injury. Clinical examination revealed increased mobility of teeth #7, #9, and #10, and fracture with avulsion of the cervical two-thirds of tooth #8. The patient reported pain, tenderness to palpation, and discomfort with jaw opening. Medical history was non-contributory with no medications or known drug allergies. Further evaluation identified pigmentary abnormalities of the eyes, dystopia canthorum, and a broad nasal root. Dental findings included peg-shaped maxillary lateral incisors (#7, #10) and congenitally missing mandibular incisors (#24, #25) on radiographic examination. Family history was notable for similar features in a maternal relative. Findings were consistent with **Waardenburg syndrome**.

Key Findings	Post Op Care
<ul style="list-style-type: none"> Trauma: Mobility (#7, #9, #10), cervical fracture/avulsion (#8) Symptoms: Pain, TTP, pain on jaw opening Craniofacial: Dystopia canthorum, broad nasal root Dental: Peg laterals (#7, #10), missing #24, #25 Family Hx: Positive 	<ul style="list-style-type: none"> Soft diet Acetaminophen Follow up in 1 week
Management	Clinical Significance
<ul style="list-style-type: none"> Irrigation with NaCl + debridement Isolation with retractor and cotton rolls Flexible splint (NiTi wire, UR6 - UL6) Bonded with flowable composite A2 Erupting teeth excluded 	<ul style="list-style-type: none"> Highlights role of pediatric dentist in identifying previously undiagnosed symptoms Emphasizes importance of family history + pattern recognition Supports need for interdisciplinary referral

Figures



Figure 1: Pre-operative photos taken 9/22/2025.

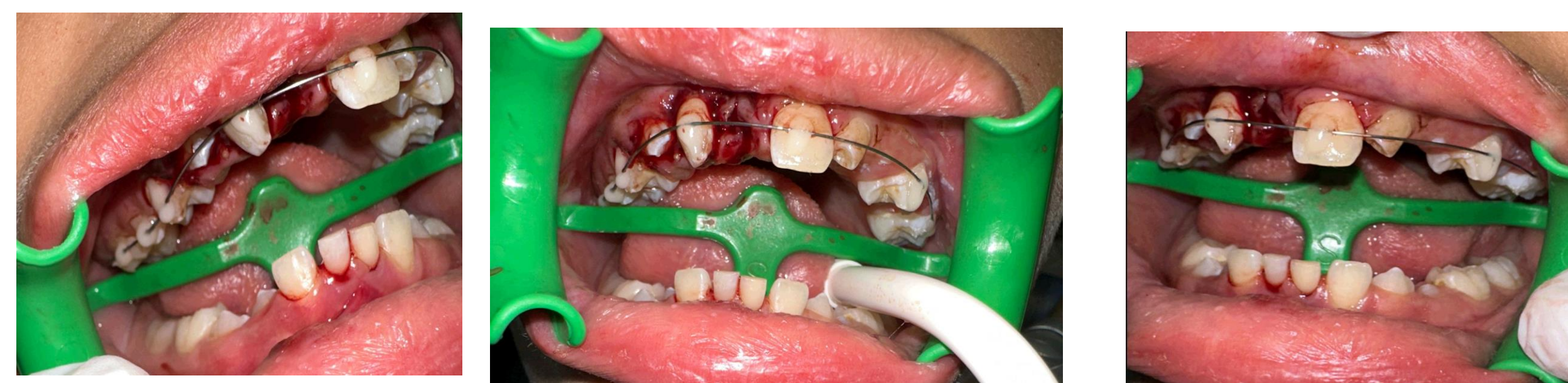


Figure 2: Post operative photos taken 9/22/2025.



Figure 3: Phenotypic appearance of child diagnosed with Waardenburg Syndrome.



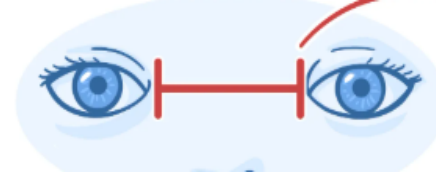


Figure 4: Panoramic and occlusal radiographs highlight the dental anomalies found in patient dentition. Congenitally missing #24 + #24, peg laterals present.

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Discussion

Waardenburg syndrome demonstrates significant phenotypic variability, which can complicate timely diagnosis, particularly in pediatric patients without a previously established medical history. In this case, the patient presented for management of acute dental trauma following avulsion of a permanent tooth. While immediate priorities included stabilization and splinting, the comprehensive extraoral and intraoral examination, an essential component of pediatric dental care, revealed findings suggestive of an underlying syndromic condition, including pigmentary abnormalities, characteristic craniofacial features, and notable dental anomalies.

Category	Clinical Findings	Clinical Significance	Dental Care Implications
Extraoral Features 	Heterochromia iridis, dystopia canthorum, broad nasal root	Classic phenotypic markers of Waardenburg syndrome, may be subtle/easily overlooked	Prompts consideration of syndromic diagnosis and need for interdisciplinary referral
Intraoral Features 	Peg-shaped maxillary lateral incisors, congenitally missing teeth	Reflect underlying disturbances in neural crest cell development	Affects arch development, spacing, eruption patterns, long-term restorative planning
Audiologic Risk 	Potential sensorineural hearing loss	High prevalence in Waardenburg syndrome patients	Requires modified communication strategies and behavior guidance
Developmental Considerations	Mixed dentition phase	Critical window where dental and craniofacial anomalies become evident	Pediatric dentists play key role in early detection and monitoring

Although standard protocols for management of dental avulsion remain unchanged, failure to recognize an underlying condition may lead to challenges in care delivery, including ineffective communication, increased anxiety, or misinterpretation of patient behavior. Early identification of both systemic and dental manifestations allows the pediatric dentist to initiate timely referral to audiology, genetics, and orthodontics, facilitating comprehensive, coordinated care and reducing the risk of delayed diagnosis and long-term complications. This case highlights the critical role of the pediatric dentist as both a clinician and an early diagnostician. Integrating careful assessment of dental anomalies with extraoral findings enhances the ability to recognize syndromic patterns, supports prompt interdisciplinary referral, and ultimately improves long-term functional and esthetic outcomes for the patient.

Conclusions

Diagnostic Vigilance: This case underscores the significance in role of the Pediatric dentist with the early detection of syndromic conditions. Even in emergency settings, a systematic clinical and radiographic evaluation is vital for identifying undiagnosed systemic conditions presenting with concurrent dental and craniofacial anomalies and allows for early intervention.

Interdisciplinary Coordination: Prompt recognition facilitates essential referrals to audiology and genetics. Early intervention prevents complications from delayed diagnosis and ensures a holistic approach to the patient's functional and developmental needs.

Comprehensive Dental Planning: Management must extend beyond acute concerns to include long term strategies for:

- Congenitally missing teeth (**hypodontia**).
- Esthetic rehabilitation of structural anomalies, such as **peg-shaped laterals**.
- Psychosocial Impact:** Addressing dental esthetics during childhood and adolescence is critical, as oral health outcomes directly influence a patient's **quality of life** and psychosocial development.
- Proactive Guidance:** Early identification allows the clinician to transition from reactive treatment to a proactive, multidisciplinary care model that optimizes both oral health and long-term esthetic outcomes.