International Journal of Research and Review
[Indexed, Peer-Reviewed Journal]
Volume 13; Issue: 9; September 2026
Website: www.ijrrjournal.com
Case Report *E-ISSN: 2349-9788; P-ISSN: 2454-2237*

Orofacial Manifestations and Comprehensive Dental Management in a Pediatric Patient with Williams-Beuren syndrome: A Case Report

Merin Mathew[1], Supriya S[2], Veronica Rose Puthenpurackal[3], Aparna Krishnan[4], Danu Dayakar P S[5]

[1,2,3,4,5]Department of Pediatric & Preventive Dentistry, Govt. Dental College, Alappuzha, Kerala

Corresponding Author: Merin Mathew

DOI: https://doi.org/10.52403/ijrr.20260924

ABSTRACT

Williams–Beuren syndrome (WBS) is a rare multisystem genetic disorder caused by microdeletion at chromosome 7q11.23. This case report describes the orofacial manifestations and management of a 11year old male child with WBS. The medical history revealed global developmental delay, mild intellectual disability, impaired gait due to limb weakness, limited verbal communication, hypothyroidism, and a history of seizure managed with antiepileptic medication. The child was also receiving iron and folic acid supplementation for hematinic deficiency. Premature greying of the scalp hair in addition to characteristic craniofacial features were observed. Intraoral examination revealed poor oral hygiene, generalized marginal gingivitis, black extrinsic staining of the teeth, proclination of maxillary incisors, multiple retained primary teeth with extensive dental caries and delayed eruption of the permanent dentition. Following medical evaluation, the retained primary teeth were extracted. Oral hygiene instructions, dietary counselling, and periodic follow-up were advised to the guardians. This case highlights the importance of comprehensive medical assessment, individualized treatment planning, preventive dental care, and multidisciplinary management to achieve favorable oral health outcomes in children with Williams Beuren syndrome.

Keywords: Williams-Beuren syndrome; William syndrome; Delayed tooth eruption; Dental caries in syndrome; premature greying of hair.

INTRODUCTION

Williams–Beuren syndrome (WBS) is a rare neurodevelopmental disorder resulting from a hemizygous microdeletion of chromosome 7q11.23, involving the elastin (ELN) gene and several adjacent genes. The syndrome has an estimated prevalence of approximately 1 in 7,500–10,000 live births. Individuals with WBS commonly exhibit characteristic facial features, developmental delay, intellectual disability, behavioral abnormalities, cardiovascular defects, connective tissue abnormalities, and endocrine disturbances.

The oral manifestations of WBS include delayed eruption of teeth, malocclusion, spacing, enamel defects, microdontia, hypodontia, increased caries susceptibility, gingivitis, and poor oral hygiene secondary to cognitive and motor impairment.

Management of these patients requires careful medical assessment, behavioral guidance, preventive strategies, and coordination with the child's physician, particularly in the presence of cardiovascular disease or seizure disorders.
This case report describes the orofacial manifestations and dental management of an 11year old child with Williams Beuren syndrome with retained carious primary teeth and delayed eruption of the permanent dentition.

CASE REPORT

An 11-year old child diagnosed with Williams Beuren syndrome was reported with the chief complaint of multiple decayed teeth and difficulty in chewing.
The child's medical history revealed global developmental delay, mild intellectual disability, impaired gait due to limb weakness with severe deficit in verbal and nonverbal communication (Figure 1). Extraoral features included microcephaly, right plagiocephaly, proptosis, large cornea, greyish hair (Figure 2).
The patient was under antiepileptic medication for recurrent seizures and thyroxine for hypothyroidism. The child was receiving ferrous sulphate and folic acid supplementation for iron and folate deficiencies. There was no history of congenital heart disease, hypertension, or other cardiovascular abnormalities. General examination demonstrated developmental delay and impaired gait. Impaired cognitive development limited the child cooperation for examination.

Figure 1: Extraoral image with impaired gait

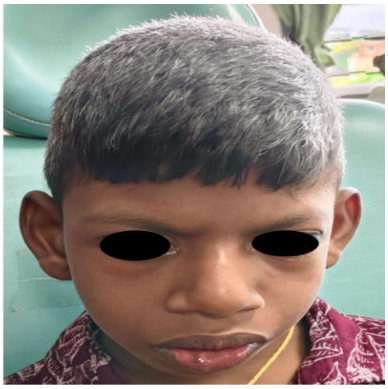

Figure 2: Hair with greyish appearance

Intraoral examination revealed generalized gingival inflammation with poor oral hygiene, multiple grossly decayed primary teeth and delayed eruption of the permanent dentition. Proclined maxillary anterior teeth were present. Generalized black extrinsic staining of the teeth was observed, which was considered to be associated with prolonged iron supplementation with no obvious soft tissue abnormalities (Figure 3)

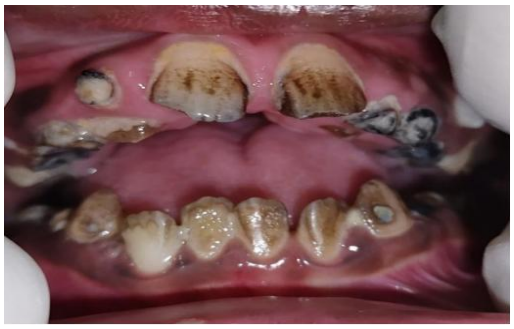

Figure 3: Intraoral image with black staining and multiple decayed teeth

Extraction of the affected retained primary teeth was planned considering their extensive destruction and the delayed eruption of their permanent successors. Due to the child's cognitive impairment, poor cooperation, and behavioral limitations, treatment was completed over four appointments with oral sedative Triclofos sodium syrup in conjunction with local anesthesia.
All planned extractions were performed uneventfully with no intraoperative or postoperative complications. Supragingival ultrasonic scaling was done to remove the extrinsic stains on permanent anterior teeth (Figure 4). Patient's guardians were given

counselling on oral hygiene maintenance, diet and the importance of periodic dental follow-ups especially to monitor the eruption of the permanent dentition.

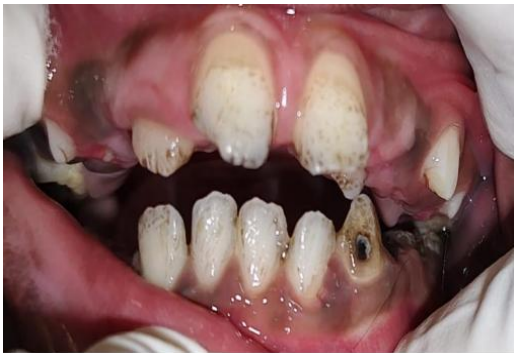

Figure 4: Postoperative image

DISCUSSION

Williams Beuren syndrome (WBS) is a multisystem genetic disorder with a wide spectrum of clinical manifestations, many of which have important implications for dental management. Although cardiovascular abnormalities, particularly supravalvular aortic stenosis, are considered the hallmark of the syndrome, their absence has been reported in a proportion of affected individuals, highlighting the variable phenotypic expression of WBS **[1,2].**

In the present case, despite the confirmed diagnosis of WBS, no cardiovascular abnormalities were identified, allowing routine dental treatment to be performed safely following medical consultation.

The patient exhibited several oral findings that are consistent with previous reports in the literature, including poor oral hygiene, generalized gingival inflammation, retained grossly decayed primary teeth, delayed eruption of the permanent dentition, and proclination of the maxillary anterior teeth **[3,4].** Poor oral hygiene is often secondary to cognitive impairment, reduced manual dexterity, and dependence on caregivers for daily oral care **[5].** Delayed eruption is a recognized dental manifestation of WBS and may contribute to prolonged retention of primary teeth, resulting in increased susceptibility to dental caries and the need for extraction **[3,4].** The presence of mild intellectual disability and limited verbal communication in this patient might have contributed to inadequate plaque control and increased caries risk, emphasizing the need for early preventive dental intervention and active caregiver participation in maintaining oral hygiene **[6].**

Premature greying of the scalp hair was another notable finding in the present patient. Although premature greying is not considered a characteristic clinical feature of Williams–Beuren syndrome, documenting such findings may help broaden the phenotypic description of this rare disorder **[2,3].** In the present case, the child had a history of hypothyroidism as well as vitamin B12 and folic acid deficiency, all of which have been reported as potential contributors to premature hair greying through their effects on melanocyte function and melanin synthesis. Therefore, the premature greying observed in this patient is more likely to be associated with these underlying endocrine and nutritional abnormalities rather than Williams–Beuren syndrome.

Generalized black extrinsic staining was observed on the teeth. This finding was most likely associated with prolonged iron supplementation prescribed for the patient. Iron-containing medications are known to produce extrinsic tooth discoloration through the deposition of iron compounds on the tooth surface, particularly in the presence of dental plaque **[8].** Supragingival ultrasonic scaling effectively removed these stains and improved the esthetic appearance of the dentition.

The presence of proclined maxillary incisors also corresponds with the malocclusion patterns frequently described in WBS. Craniofacial growth disturbances, altered muscle function, and abnormal connective tissue development have been proposed as contributing factors **[7,9].**

Behaviour management remains an important consideration in children with WBS because cognitive impairment, developmental delay, anxiety, and communication difficulties may limit cooperation during dental treatment.

Comprehensive medical evaluation before treatment is essential, particularly because patients with WBS may present with cardiovascular disease, hypertension, endocrine abnormalities, seizure disorders, or other systemic conditions that can influence dental care **[1,2].** The history of epilepsy and hypothyroidism in this patient required careful review of the medical status and medications before treatment planning. Dental treatment in patients with WBS should be individualized according to the patient's systemic condition, behavioural status, and medical risk assessment. Behavioural guidance techniques, shorter appointments, effective communication with caregivers, and stress reduction are particularly important because many affected children exhibit anxiety, hypersensitivity to sound, and cognitive impairment.

The extraction of retained primary teeth was undertaken to eliminate sources of infection and facilitate the eruption of permanent successors. Periodic follow-up is essential to monitor the eruption pattern and identify the need for future orthodontic intervention, as malocclusion and delayed dental development are common in WBS **[3,4].** Preventive strategies, including reinforcement of oral hygiene measures, dietary counselling, topical fluoride application, and regular recall visits, remain fundamental components of long-term dental care in these patients **[6].**

This case highlights the importance of individualized treatment planning and interdisciplinary collaboration in the management of children with Williams–Beuren syndrome. While previous reports have described the general oral manifestations and dental management of individuals with WBS, the present case describes a pediatric patient presenting with extensive caries involving retained primary teeth, delayed eruption of the permanent dentition, significant communication limitations, and multiple medical co morbidities, highlighting the practical challenges of individualized dental care. Early recognition of the oral manifestations, appropriate medical assessment, and implementation of preventive dental care can significantly improve oral health outcomes and quality of life in affected individuals.

CONCLUSION

Williams–Beuren syndrome presents unique challenges for dental practitioners because of its multisystem involvement and associated developmental and behavioral characteristics. Comprehensive medical assessment, early preventive intervention, and individualized dental treatment are essential to improve oral health and quality of life. This case highlights the importance of recognizing the oral manifestations of WBS and adopting an interdisciplinary approach to dental management.

Declaration by Authors

Acknowledgement: None
Source of Funding: None
Conflict of Interest: No conflicts of interest declared.

REFERENCES

1. Pober BR. Williams-Beuren syndrome. N Engl J Med. 2010 Jan 21;362(3):239-52.
2. Morris CA. Williams Syndrome. Adam MP, Bick S, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2026.
3. Axelsson S. Variability of the cranial and dental phenotype in Williams syndrome. Swed Dent J Suppl. 2005;(170):3-67.
4. Hertzberg J, Nakisbendi L, Needleman HL, Pober B. Williams syndrome--oral presentation of 45 cases. Pediatr Dent. 1994 Jul-Aug;16(4):262-7.
5. Joseph C, Landru MM, Bdeoui F, Gogly B, Dridi SM. Periodontal conditions in Williams Beuren syndrome: a series of 8 cases. Eur Arch Paediatr Dent. 2008 Sep;9(3):142-7

6. Klingberg G, Broberg AG. Dental fear/anxiety and dental behaviour management problems in children and adolescents: a review of prevalence and concomitant psychological factors. Int J Paediatr Dent. 2007 Nov;17(6):391-406.
7. Wong D, Ramachandra SS, Singh AK. Dental management of patient with Williams Syndrome - A case report. Contemp Clin Dent. 2015 Jul-Sep;6(3):418-20.
8. Watts A, Addy M. Tooth discolouration and staining: a review of the literature. Br Dent J. 2001 Mar 24;190(6):309-16.
9. Vavetsi K, Panagopoulou O, Koromantzos P, Fryssira H, Bobetsis SA, Emmanouil D, Bobetsis YA. Oral manifestations of nine individuals with Williams syndrome. A case series. Spec Care Dentist. 2024 Mar-Apr;44(2):438-449.

How to cite this article: Merin Mathew, Supriya S, Veronica Rose Puthenpurackal, Aparna Krishnan, Danu Dayakar P S. Orofacial manifestations and comprehensive dental management in a pediatric patient with Williams-Beuren syndrome: a case report. *International Journal of Research and Review*. 2026; 13(9): 239-243. DOI: *https://doi.org/10.52403/ijrr.20260924*

******