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Case Report

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Clinical Manifestations and Pediatric Dental Management of Beckwith-Wiedemann Syndrome in a 13-Year-Old Indian Patient: A Case Report

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Abstract

Background: *Beckwith-Wiedemann Syndrome (BWS) is a rare congenital genomic imprinting disorder characterized by overgrowth, hemihypertrophy, and variable craniomaxillofacial manifestations. Pediatric dentists are often the first specialists to encounter the oro-dental manifestations of this syndrome and play a pivotal role in early recognition, preventive management, and timely referral.*

Case Description: *A 13-year-old male who reported to the Department of Pediatric and Preventive Dentistry. Clinical examination revealed left-side hemi-hyperplasia, facial asymmetry, short fingers with clinodactyly of the fifth digit, hyperpigmentation of the knees and neck, and a Trendelenburg gait, which were confirmed as Beckwith-Wiedemann Syndrome following pediatric medical evaluation. Intraoral examination revealed generalized moderate dental fluorosis (Dean's Grade 3), Class I occlusal caries on upper permanent first molars, retained primary right upper canine, and impacted right maxillary canine. Treatment provided included full-mouth scaling, preventive resin restorations (PRR) on teeth 16 and 26, fluoride counseling, and dietary modifications and comprehensive interdisciplinary referral.*

Conclusion: *This case underscores the importance of pediatric dentists in identifying syndromic features, performing targeted radiographic evaluation, and coordinating multidisciplinary care for patients with BWS. Early diagnosis and a structured preventive protocol are essential to optimize oral health outcomes in such rare genetic disorders.*

Keywords: *Beckwith-Wiedemann Syndrome; Dental Fluorosis Hemihypertrophy; Impacted Canine; Pediatric Dentistry; Preventive Resin Restoration.*

INTRODUCTION

In the science of pediatric dentistry, a specialty dealing children from birth to adolescence encompasses array of clinical conditions, systemic problems, developmental malformations and syndromes [1-3]. Beckwith-Wiedemann Syndrome (BWS) is a rare congenital overgrowth disorder occurring in approximately 1 in 10,300 live births and it affect different parts of the body [4]. It arises from genetic and epigenetic alterations at the 11p15.5 imprinting center, which regulates fetal and postnatal growth [4]. The incidence is equal in males and females with the notable exception of monozygotic twins that show a drastic excess in females. In addition to clinical heterogeneity, BWS also exhibit etiologic molecular heterogeneity. A variety of genetic and or epigenetic alterations in growth regulatory genes on chromosome 11p15.5 are associated with specific phenotype-epigenotype/genotype correlations including different recurrence risks. 85% of BWS cases usually occurs sporadically but familial transmission occurs in 15% of cases. This panethnic genetic disorder generally affects children growth and babies born with BWS appear larger than normal a condition referred as macrosomia. Some children look taller than other children with similar age. However, the growth will slow down by the age of 8 in

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BWS. BWS is also known as Beckwith Wiedemann spectrum. This is due to variation in symptoms and their severity. BWS is characterized by a wide spectrum of hallmark clinical features, including macrosomia, gigantism, macroglossia, anterior abdominal wall defects, organomegaly, hemihyperplasia, ear creases or pits, and neonatal hypoglycemia and an increased risk of embryonal tumors. The classical features such as exomphalos, macroglossia and gigantism were first described by Beckwith and Wiedemann and hence the name for the syndrome. The orodental manifestations of BWS are diverse and include macroglossia, skeletal malocclusions most frequently Class III open bite, delayed eruption, dental crowding, diastema, enamel defects, and tooth shape anomalies. Hemihyperplasia, when present, can lead to pronounced facial asymmetry and arch-length discrepancies, further complicating occlusal development [5-7]. Pediatric dentists are uniquely positioned to recognize these early signs, initiate appropriate preventive therapy, and coordinate comprehensive care with orthodontists, oral and maxillofacial-surgeons, geneticists, and pediatricians [8]. This case report presents the dental findings and pediatric dental management of an adolescent male with BWS, emphasizing the role of the pedodontist in syndrome identification, growth assessment, and interdisciplinary treatment planning. The aim of the present research paper is to show case occurrence of Beckwith-Wiedemann Syndrome in a 13-Year-Old Indian patient emphasizing about clinical manifestations and preventive management of dental problems.

CASE REPORT

A 13-year-11-month-old male patient presented to the Department of Pediatric and Preventive Dentistry, College of Dental Sciences, Davangere, with the chief complaint of forwardly placed upper front teeth. The patient was a ninth-grade student who was born full-term by normal vaginal delivery following an uneventful pregnancy. His postnatal history revealed breastfeeding for 18 months, followed by bottle-feeding for one month, and his developmental milestones were attained within the normal age range. There was no significant family history of similar craniofacial anomalies or malocclusion, and no history of consanguinity. General physical examination revealed several characteristic abnormalities, including left-sided hemihypertrophy (Figure 1) resulting in bodily asymmetry, short fingers with clinodactyly involving the fifth finger and toe (Figure 2 & 3), hyperpigmentation over the neck and knees and a Trendelenburg gait. These findings raised suspicion of an underlying syndromic condition, prompting referral to a pediatrician, where the diagnosis of Beckwith–Wiedemann syndrome (BWS) was confirmed. Intraoral examination demonstrated good oral hygiene, complete permanent dentition with retained primary tooth (Figure 4). Considering the established association of BWS with craniofacial abnormalities, delayed tooth eruption, and altered dental development, a comprehensive radiographic assessment was performed to evaluate the stage of dental maturation, eruption pattern, and associated dentoalveolar findings (Figure 5), as well as the underlying skeletal pattern and pharyngeal airway dimensions using a lateral cephalogram (Figure 6 & 7). The detailed clinical and radiographic findings of the patient are summarized in Table 1.

Table 1: Clinical and Radiographic findings

Diagnostic Evaluation		**Findings observed**
Intraoral examination (Figure 4)	Enamel Alterations	Generalized moderate dental fluorosis (Grade 3, Modified Dean's Index)
	Caries Status	Class I occlusal caries on permanent upper molars (16, 26)
	Dentition status	Bilateral Class I molar relation, increased overjet, crowding in mandibular dentition, posterior unilateral cross-bite on right side.
	Eruption Anomalies	Retained deciduous maxillary right canine (13)
Radiographic Examination (Figure 5 & 6)		Panoramic (OPG) radiograph demonstrated impacted maxillary right canine (13) in addition to retained primary right canine (53) (Figure 5) Lateral cephalogram showed reduced upper and lower airway space (Figure 6 & 7)
Developmental Dental Age		12–13 years

Following comprehensive clinical and radiographic evaluation, a multidisciplinary treatment plan was formulated with emphasis on preventive, restorative, and interceptive care. Initial management included full-mouth ultrasonic oral prophylaxis, followed by individualized oral hygiene instructions to reinforce plaque control and long-term preventive practices. Preventive resin restorations (PRRs) were subsequently placed on the permanent maxillary first molars (16 and 26) using a minimally invasive composite resin technique to conservatively manage incipient occlusal carious lesions while preserving sound tooth structure. Considering the presence of generalized dental fluorosis, individualized caries risk assessment was performed, and dietary counseling was provided with recommendations tailored to the patient's local fluoride exposure and oral health needs. Given the complexity of the malocclusion and the impacted maxillary left permanent canine, an interdisciplinary approach was adopted, and the patient was referred to the Departments of Orthodontics and Oral and Maxillofacial Surgery for comprehensive evaluation and definitive management of the impacted canine, arch-length discrepancy, and underlying skeletal pattern. As an interceptive measure, extraction of the retained primary maxillary left canine (53) was planned to promote the spontaneous eruption of the impacted permanent canine (13) or, if required, facilitate subsequent surgical exposure and orthodontic traction.

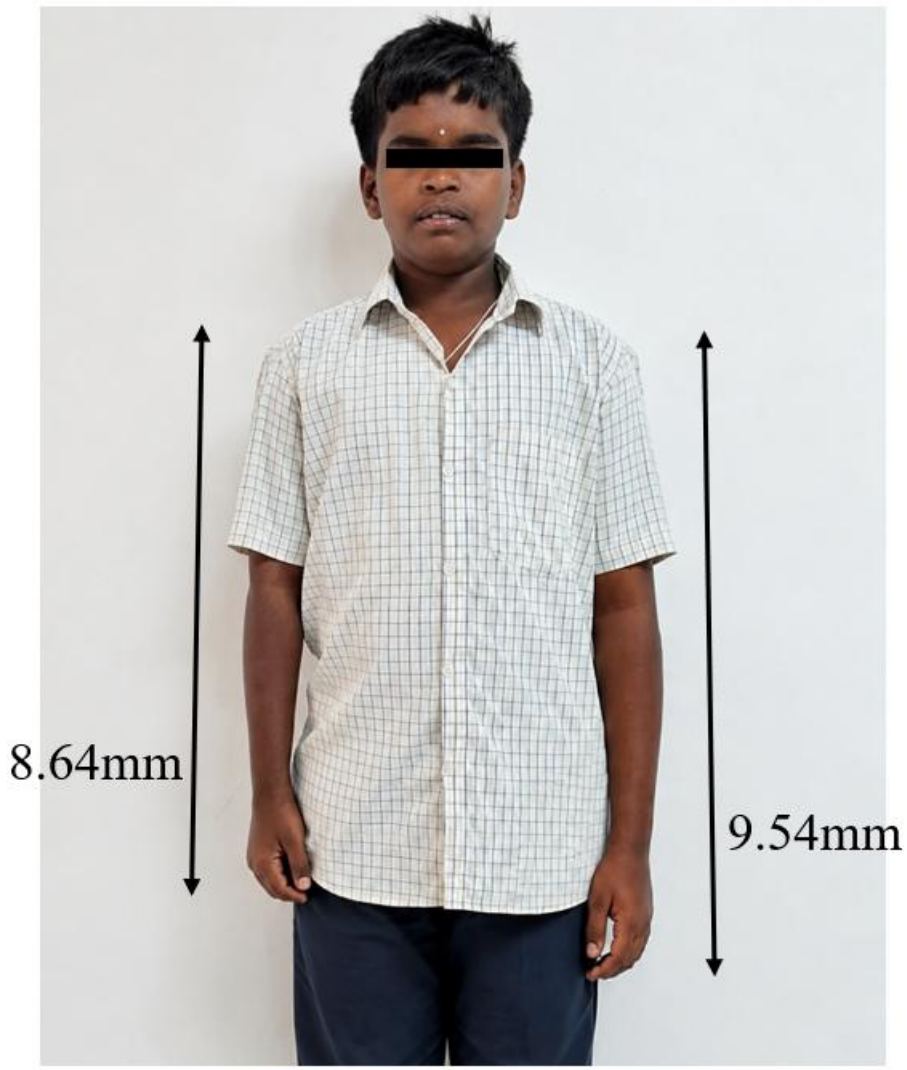


Figure 1: Left side Hemihypertrophy

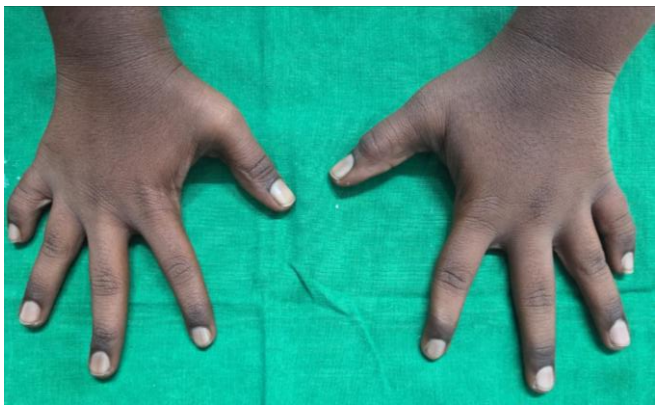


Figure 2: Short fingers and clinodactyly of 5th Finger



Figure 3: Short fingers and clinodactyly of 5th Toe



Figure 4: Intraoral photographs of the patient



Figure 5: Orthopantograph of the patient showing impacted maxillary right canine

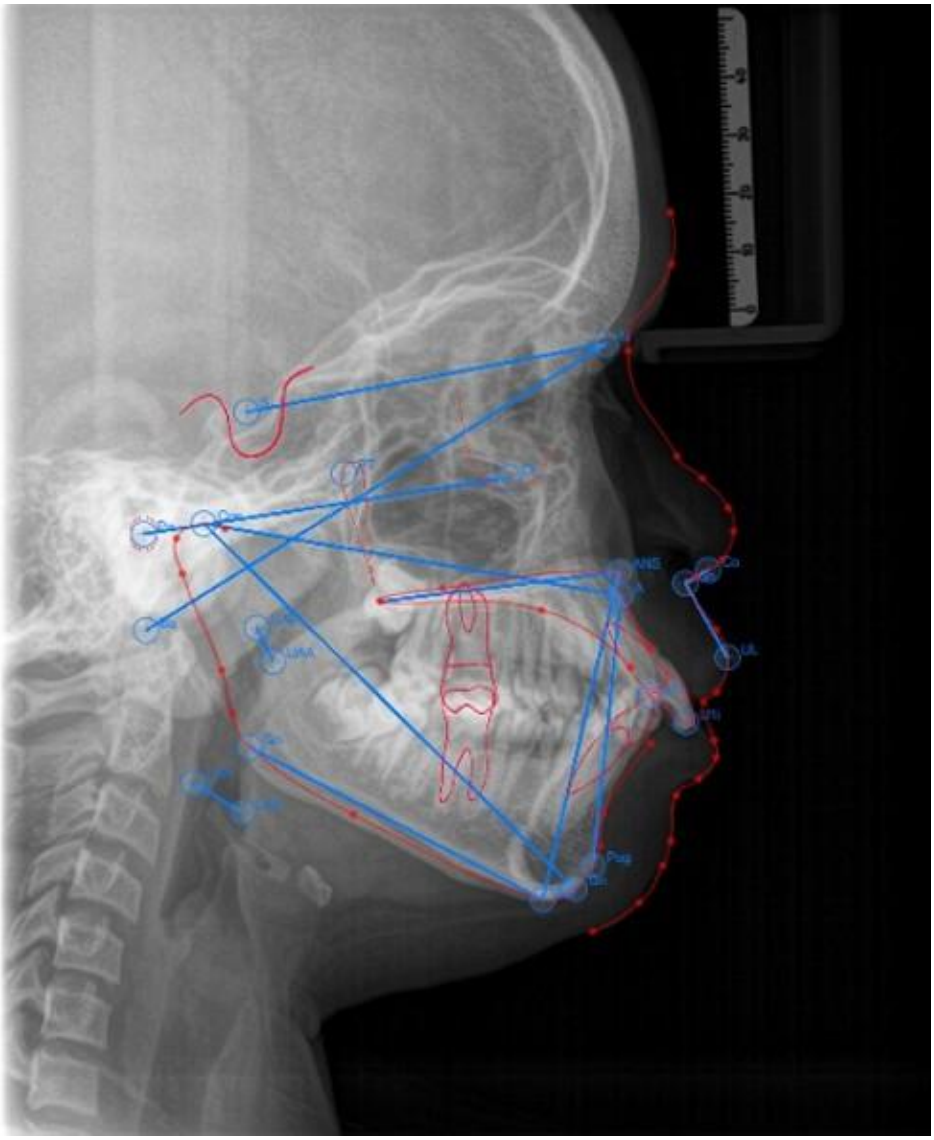


Figure 6: Lateral Cephalogram of the patient showing reduced upper and lower airway space

Dentition

Measurement	Value / Status
Upper incisor to Point A (4.00mm - 6.00mm)	11.17mm Forward position of upper incisors
Lower incisor to A-Pog (1.00mm - 3.00mm)	6.77mm Forward position of lower incisors

Airway

Measurement	Value / Status
Upper Pharynx (15.00mm - 20.00mm)	6.34mm Reduced upper airway space
Lower Pharynx (11.00mm - 14.00mm)	9.95mm Reduced lower airway space

Figure 7: Important findings of Lateral Cephalogram



Figure 8: BWS Facies, characteristic feature of BWS syndrome

DISCUSSION

Beckwith–Wiedemann syndrome (BWS) is a rare congenital overgrowth disorder, and despite its well-recognized systemic manifestations, reports describing its oral and craniofacial features from a pediatric dental perspective remain scarce. BWS patients show increased growth rate during the last months of pregnancy and in the first few years of an infant life. Growth

parameters classically exhibit weight and height around the 97th percentile with head size closer to the 50th percentile. However, adult show normal range in heights compared to normal people. Sometimes, abnormal growth is marked by macroglossia or hemihyperplasia. Due to macroglossia individuals can experience deglutition problems, speech and sleep related disorders. Affected BWS cases show recognizable facial gestalt which is more often called as 'BWS facies' characterized by prominent eyes with infraorbital creases, facial nevus flammeus, macroglossia, full lower face with a prominent mandible, posterior helical pits, anterior earlobe creases and midfacial hypoplasia (Figure 8). Gradually BWS facies normalizes across childhood and hence while diagnosing BWS in adults or adolescents requires careful assessment of childhood photographs. Although development appears normal in these individuals, because of chromosome 11p15.5 duplication, serious perinatal complications like uncontrolled hypoglycemia and prematurity is observed. hypoglycemia is reported in 30-50% of babies with BWS which is likely caused by islet cell hyperplasia and hyperinsulinemia.

The available dental literature is largely limited to isolated case reports and small case series [6,10], highlighting the need for further documentation of the clinical presentation and multidisciplinary management of affected individuals [13]. Pediatric dentists are often the first specialists to encounter the oro-dental manifestations of this syndrome and play a pivotal role in early recognition, preventive management, and timely referral. Similar observations have been reported in several rare syndromic case reports published in the pediatric dental literature, where oral manifestations served as an important clue for diagnosis and multidisciplinary management [12-14]. BWS is clinically diagnosed using a scoring system based on cardinal and suggestive features. Cardinal features include macroglossia, macrosomia, lateralized overgrowth (hemihyperplasia), multifocal or diffuse organomegaly, and anterior abdominal wall defects [4]. Our patient exhibited hemihyperplasia, limb anomalies (clinodactyly), and cutaneous hyperpigmentation, supporting the clinical suspicion of BWS. Hemihyperplasia in BWS can manifest as asymmetric growth of the face, trunk, or limbs, leading to significant orthodontic and orthopedic challenges [5,6].

In the present case, facial asymmetry was evident in the vertical and transverse dimensions, and the maxillary and mandibular arches displayed marked asymmetry. The arm-length discrepancy and digital anomalies further colloborated the systemic nature of the overgrowth [15]. Pediatric dentists must be vigilant in documenting these findings, as they directly influence arch-form prediction and space-management strategies [6,10,12]. The pediatric dental role in BWS extends beyond restorative and preventive care [6]. Pediatric dentists must coordinate with pediatricians to ensure adherence to tumor surveillance protocols, including abdominal ultrasonography and serum alpha-fetoprotein monitoring [16]. Airway evaluation constitutes an important component of the comprehensive assessment of patients with BWS because of their increased susceptibility to upper airway compromise and obstructive sleep apnea. In the present case, lateral cephalometric analysis demonstrated reduced pharyngeal airway dimensions, a finding that underscores the need for long-term airway surveillance and interdisciplinary follow-up, even in the absence of clinically evident macroglossia. Preventive care remains the cornerstone of pediatric dental management in BWS [16]. Oral prophylaxis, topical fluoride application, pit-and-fissure sealants, and dietary counseling are essential to mitigate the risk of dental caries and periodontal disease, particularly in patients with malalignment and cleaning difficulties [17-19]]. In this case, the existing preventive resin restoration and recent prophylaxis provided a foundation upon which further interdisciplinary care was built.

BWS affected individuals show increased frequency of malformations and medical complications like abdominal wall defects in the form of umbilical hernia, diastasis recti, omphalocele, visceromegaly involving any single or combination of organs such as liver, spleen, pancreas, kidneys and adrenals. It has been reported that fetal adrenocortical cytomegaly is a pathognomonic finding observed in BWS [20,21]. In addition to this, unilateral or bilateral renal anomalies like primary malformations, renal medullary dysplasia, nephrocalcinosis and nephrolithiasis have also been reported. Moreover, cardiac malformations like cardiomegaly were observed in 20% of children with BWS, which resolved spontaneously [20].

In differential diagnosis, other conditions like Costello syndrome, Perlman syndrome, Mosaicism for trisomy 8, Maroteaux-Lamy syndrome, Sotos syndrome, Simpson-Golabi-Behmel syndrome should be ruled out. Because most of genetic syndromes have features in common with BWS. For cases involving asymmetry as an isolated finding, it is important to determine whether the asymmetry represents hemihyperplasia (overgrowth) or hemihypoplasia (decreased growth). However, molecular testing is the confirmatory procedure to arrive at correct diagnosis. Therefore, it is always necessary to perform comprehensive clinical examination including appropriate investigations, clinical genetics consultation, biochemical testing, molecular testing and brain imaging [15].

The meticulous diagnosis of BWS requires no absolute criteria for its diagnosis. Because BWS is one among several other conditions associated with genetic-epigenetic alterations on chromosome 11p15. This spectrum includes children with features of BWS who do not fulfill clinical diagnostic criteria, as well as children with isolated hemihyperplasia or isolated Wilms tumor. It is suggested that presence of at least three major findings or two major findings and one minor fining helps in clinical diagnosis. Moreover, it is essential to consider the heterogeneous expression of this disorder and the role of positive molecular tests that may help in diagnosis even in cases with few cardinal clinical manifestations. In the case of a clinical diagnosis of BWS, molecular determination of altered methylation, a microdeletion at imprinting center 1(IC1) and/or IC2, or a mutation in CDKN1C would confirm the diagnosis [16-20].

CONCLUSION

Pediatric dentists play a pivotal role in the early recognition of syndromic conditions through careful evaluation of oral and craniofacial findings [22-24]. Timely preventive care, appropriate restorative treatment, interceptive management, and interdisciplinary collaboration are essential for preserving oral health, optimizing function, and improving the overall quality of life of children with special healthcare needs.

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