



## CASE REPORT

# Prader-Willi Syndrome, Oral and Dental Findings with a Multidisciplinary Care and Management Case Report

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### Abstract

Prader-Willi Syndrome (PWS) is a complex genetic, neurodevelopmental disorder that affects both males and females equally, causing a broad range of physical, cognitive, and behavioural challenges. Patients exhibited symptoms including low muscle tone, hyperphagia, and increased risk of obesity. PWS has three different molecular genetic classes. The most common cause is the deletion of the paternal copy of chromosome 15q11-q13. Healthcare providers are likely to observe mild cognitive impairment and behavioural problems, such as self-injury, compulsions, and sudden outbursts during diagnosis. Currently, there are no approved therapeutics for managing PWS; however, controlling hyperphagia and improving food-related complications are proposed as effective strategies. A ten-year-old Arab male diagnosed with PWS attended the orthodontic clinic, presenting with concerns about the appearance of his teeth. Clinical examination revealed a high palatal arch, dysmorphic features, a slightly arched foot, and extremely fistled fingers, indicative of developmental abnormalities. The patient underwent an EEG, which revealed epileptiform discharges in the frontal-temporal region, and an EMG for seizure evaluation. Additionally, dental examination revealed severe tooth wear on the maxillary permanent central incisors and the mandibular molars bilaterally.

**Keywords:** Prader-Willi syndrome, Periodontics, Orthodontics, Hypotonia, Behavioural management, Multidisciplinary approach

### Introduction

Prader-Willi syndrome (PWS) is a rare genetic disorder primarily caused by genomic imprinting, an error in the genomic mechanism. The prevalence of PWS is notable, affecting a segment of the population. Gutierrez and Mendez reported that PWS affects approximately half a million people globally and occurs in about 1 of every 20,000 live births. Without effective management of food-seeking behaviours and other associated challenges, individuals with PWS face a significant risk of reduced life expectancy.

Notably, PWS manifests in early life, most commonly through poor sucking ability and feeding difficulties during infancy. According to Sarkar *et al.*, affected children are unlikely to thrive and often demonstrate growth retardation accompanied by low muscle tone. In addition, PWS is associated with hypogonadism in both male and female children. The condition also contributes to motor and cognitive deficiencies, as well as multiple hormonal deficiencies. Although individuals with PWS tend to live longer than those with other congenital conditions, the syndrome carries

significant long-term health implications. Poorly managed PWS frequently results in morbid obesity, a major contributor to morbidity.

In addition to morbid obesity, PWS predisposes individuals to several chronic conditions, including diabetes, cardiovascular disease, orthopaedic complications. In severe cases, inadequate management can lead to death. Mortality in PWS has been attributed to various causes, most commonly respiratory failure, cardiac disease, and gastrointestinal complications. These comorbidities reflect the complex nature of PWS.

Individuals with PWS are at increased risk of developing oral diseases, such as enamel hypoplasia, caries, and tooth wear from attrition, erosion, periodontal disease, and delayed tooth eruption. These complications are exacerbated by decreased saliva flow, leading to increased dental caries. Hypotonia among PWS patients also affects oral musculature, increasing the risk of oral diseases. Preventive strategies, such as maintaining effective oral hygiene, attending regular dental check-ups, and receiving fluoride treatments, are essential to mitigate these risks.

PWS exposes patients to various health risks, underscoring the importance of effective management. Despite this need, medication and treatment non-adherence remain significant challenges, largely due to the behavioural aspects of the condition. The objective of this report is to present a detailed case analysis of a 10-year-old PWS patient. The report aims to contribute novel insights to existing research on dental health by examining the underexplored oral needs of individuals with PWS. Additionally, it seeks to raise awareness among general dental practitioners (GDPs) about the complexities of managing patients with PWS.

## Case Presentation

A 10-year-old male of Arabic ethnic background, diagnosed with PWS, attended the Dental and Maxillofacial Centre (DMFC) / Royal Medical Services (RMS) with his father, presenting with concerns about the appearance of his teeth. Consent was obtained from the father to use the medical records for case reporting purposes.

## Medical History

The patient was born with dysmorphic craniofacial features and was admitted to the neonatal intensive care unit at 25 days of age for 10 days due to convulsions, which were managed with phenytoin, and anaemia, for which he received a cell transfusion. During the early months of infancy, he was also diagnosed with glucose-6-phosphate dehydrogenase deficiency, feeding difficulties, and jaundice.

Further investigations conducted at an early age included an EEG, which revealed epileptiform discharges in the frontal and temporal regions, and an EMG for seizure detection. As he grew older, dysmorphic features became more pronounced, including a high palatal arch, tight tongue, extremely fistled fingers, mildly arched feet, and generalized mild hypotonia. The patient underwent tonsillectomy and adenoidectomy at the same hospital without complications.

## Dental History

The patient attended the orthodontic dental clinic at the DMFC/RMS at the age of 10, seeking evaluation of the appearance of his upper front teeth. Extraoral examination revealed short stature and obesity (Figure 1). Additionally, the patient exhibited speech impairment and generalised hypertonia.



**Figure 1:** Extraoral features of Prader-Willi syndrome

*Note: Highlighted feature - short stature, obesity, speech impairment, and generalised hypertonia. These symptoms were indicative of PWS*

## Intraoral Examination

### Soft tissues

Bleeding from the gingiva and tongue tie.

### Oral hygiene

The patient's oral hygiene was assessed as fair, with a plaque score of 10% using disclosing tablets (MIRA-2-Ton Tablets). A thorough periodontal examination revealed bleeding on probing in all quadrants (Code 1), indicating poor oral hygiene.

### Dentition: Caries and Tooth wear (TW)

Severe TW was observed on the maxillary permanent central incisors and the mandibular molars bilaterally (Figures 2, 3). Signs of bruxism were also present, likely related to the patient's cognitive and behavioural issues, as reported by the father, who noted episodes of stress and anxiety-related tantrums.



**Figure 2:** Extraoral facial photograph (frontal view)

*Note: Highlighted features - gingival bleeding, poor oral hygiene, dental caries, and tooth wear*

The extraoral features were indicative of PWS.



**Figure 3:** Intraoral photographs showing severe tooth wear (TW) in both maxillary and mandibular arches

The severe TW observed during the intraoral examination provided further evidence of manifestations associated with PWS.

## Occlusion

The patient presented with a Class III incisor relationship on a mild skeletal Class III base, with an average vertical dimension and skeletal asymmetry, characterized by a chin deviation to the right. This was complicated by a complete overbite extending to the teeth. A reversed overjet of 3 mm was noted, along with mild crowding in the lower left segment and moderate crowding in the upper left segment. The lower centreline was shifted 2 mm to the right. There was a tendency toward crossbite involving the upper left first premolar, displacement of the upper left second premolar, and bilateral Class III canine and molar relationships, as illustrated in the images below (Figure 4).



**Figure 4:** Intraoral photographs showing reverse overjet of 3 mm, mild crowding, and crossbite

*Note: The mild skeletal Class III malocclusion is attributed to the protruded mandible relative to the maxilla. This malformation resulted in a larger*

*overbite, characterized by excessive overlap of the upper incisors over the lower incisors.*

The patient was assessed using the Index of Orthodontic Treatment Need (IOTN), a standardized tool developed by Brook and Shaw over three decades ago that is widely used in orthodontics to assess the treatment needs of children.<sup>5</sup> The IOTN consists of two components: the dental health component (DHC) and the aesthetic component (AC), which together determine whether treatment is indicated for dental health or aesthetic reasons.

The patient's IOTN was 3b/7, a level for which the National Health Service recommends prioritization as a complicated case requiring orthodontic treatment.

A comprehensive orthodontic examination was conducted, and a customised treatment plan was developed. The DMFC oral hygienist provided the patient with thorough oral hygiene instructions, given the increased risk of caries and gingivitis in children with PWS. The syndrome is also associated with decreased salivary flow and increased salivary acidity, further emphasizing the importance of strict oral hygiene practices. Considering the patient had difficulty using his hands, he was advised to use a special handle toothbrush to improve grip and brushing efficiency.

### **Radiographic Investigations**

No indications were found for either extraoral or intraoral radiographic examination.

After a comprehensive orthodontic examination, the patient's occlusion was categorized as IOTN 3b/7 and considered a high-priority case for orthodontic treatment.

The orthodontic treatment plan included an annual review of skeletal Class III progression, followed by interceptive treatment with a removable upper appliance. The risks and benefits associated with the treatment plan were explained to the patient's parents.

### **Treatment**

The patient was first seen at the DMFC prior to the COVID-19 outbreak, which delayed dental intervention. After the outbreak plateaued, he

was reviewed in January 2021 at the orthodontic clinic with the same complaints regarding dental appearance. The final treatment plan was categorized into three phases: periodontic, restorative, and orthodontic treatment.

The patient attended the periodontology clinic at the age of 13 to improve his oral hygiene, which was challenging to maintain due to behavioural and cognitive problems as well as anxiety. Simple behavioural techniques were employed to facilitate acceptance of treatment. Detailed oral hygiene instructions were provided to the patient and his parent, and full-mouth root surface debridement was subsequently performed over multiple appointments under topical anaesthesia. Ultrasonic scalers were used to thoroughly remove plaque deposits from the tooth surfaces. The periodontist recommended conservative restoration for the eroded teeth. Five months later, the patient reattended the periodontology clinic, but showed no improvement in oral health, as indicated by plaque and gingival scores.

As a result, the patient was referred to a specialist for conservative management of the tooth wear observed in the upper anterior teeth. He was then referred to the restorative clinic, where the upper central and lateral incisors were restored with a resin composite (Universal Restorative A1 Body Shade, 3M Filtek) to improve the eroded crowns.

The patient later visited the orthodontist to initiate an interceptive treatment plan. It involved the use of an upper removable appliance with a posterior biteplate and an anterior screw to procline the maxillary incisors. The risks and benefits were explained to the mother, who consented to the treatment. The patient was recalled several times over a four-year period, with orthodontic reviews scheduled every 4-6 weeks.

At the time of reporting, the patient was nearly 15 years old and was still undergoing orthodontic treatment. The care plan incorporated a combination of treatment approaches.

### **Discussion**

PWS is a complex condition affecting a significant proportion of children worldwide. The prevalence

of PWS is moderate, with most cases resulting from a deletion of the paternally inherited 15q11.2-q13. PWS occurs due to genetic changes affecting a specific region of the chromosome, typically during conception or early foetal development. The National Institutes of Health has further noted that these alterations may also arise in the early stages of foetal development. Additional changes, including maternal uniparental disomy and imprinting centre defects, have also been identified as causes of PWS, although such scenarios are rare.

Although PWS results from genetic alterations, it is not hereditary, as evidenced in this case, where the patient's siblings did not exhibit the condition. Therefore, it is imperative to clarify that PWS is not directly related to the consanguinity noted in the patient's history. Albanghali reported that the prevalence of consanguineous marriage is high in Arab countries, with at least 40% of the population married to close relatives, primarily cousins. While consanguineous marriages are linked to several health risks, they do not contribute to the prevalence of PWS. Chromosomal deletions, the primary cause of PWS, are not inherited through typical Mendelian patterns or linked to parental consanguinity. Thus, while consanguinity provides a cultural and genetic background for the patient's family, it is not considered a contributing factor in the development of PWS in this case.

Additionally, PWS is associated with behavioural and cognitive challenges. These challenges may limit patients' adherence to treatment plans and recommendations. Research has linked cognitive impairment with reduced compliance and adherence levels to treatment, even for patients with higher educational levels. PWS-related behaviours, such as rigidity, temper tantrums and anxiety, also interfere with patients' adherence to treatment. These challenges may explain the non-adherence, which necessitated multiple hospital recalls in this case. Consequently, tailored treatment modules are crucial to mitigate these challenges.

The recommended treatment plan is tailored to ensure that the patient's needs are met. This approach targets the specific cognitive and behavioural challenges for each patient. Amaro *et*

*al.* recommended pharmacological interventions to manage the complex cognitive and behavioural difficulties. For instance, psychotropics can mitigate behavioural and neuropsychiatric problems. In addition, multidisciplinary treatment was prioritized, as it provides a more holistic approach to addressing health issues and ensures that a wide range of specialised skills are utilized to manage specific health conditions. This approach comprises a multidisciplinary team of professionals, including doctors, therapists, and caregivers, who can effectively manage the complex cognitive and behavioural challenges in PWS patients.

Dental management for patients with PWS also requires a multidisciplinary approach, considering the cognitive and behavioural challenges associated with the condition. Early interventions are recommended, along with regular dental checkups, to promote oral health and prevent complications such as those reported in the present case. Driscoll *et al.* recommended dental evaluation at least every six months after tooth eruption or every 3-4 months for persistent dental issues.<sup>4</sup> These regular checkups help mitigate the risks of caries and enamel hypoplasia. Early interventions may include fluoride applications, pit and fissure sealants, dietary counselling, and reinforcement of at-home oral care routines. Such measures not only support the maintenance of oral health but also reduce the likelihood of requiring more invasive procedures.

### Significance

The significance of this case lies in its detailed exploration of the oral and dental challenges faced by PWS patients and the multidisciplinary approach used to manage these issues. The case emphasizes the increased risk of oral diseases in PWS patients, such as enamel hypoplasia, caries, tooth wear, gingival bleeding, and delayed tooth eruption. These issues are linked to factors such as decreased saliva flow, hypotonia affecting the oral musculature, and behavioural challenges, including bruxism. The analysis also highlights the importance of a collaborative treatment plan that incorporates periodontic, restorative, and orthodontic interventions. This approach addresses the complex needs of PWS patients, including their

cognitive and behavioural challenges, to improve oral health and overall quality of life. The report underscores the need for individualized care plans that consider the patient's behavioural and cognitive limitations, such as using special toothbrushes for improving grip and employing simple behavioural techniques to promote treatment adherence. Additionally, the case aimed to educate GDPs about the complexities of managing PWS patients, emphasizing the importance of early interventions, regular dental checkups, and preventive strategies to mitigate oral health risks. By presenting a detailed case analysis, it adds novel insights to the existing research on dental health in PWS patients, addressing an underexplored area and encouraging further studies. Overall, this case highlights the need for a holistic, multidisciplinary approach to manage the oral health challenges of PWS patients, while also raising awareness and contributing to the broader understanding of the condition.

### Conclusion

PWS is a complex condition with severe medical and oral health influences and treatment challenges that GDPs need to be aware of. In the report, a multidisciplinary treatment plan was formulated through collaboration with various dental professionals. The treatment plan for such a condition should be based on a thorough clinical and oral examination, the patient's behaviour and level of cooperation, and parents' input to ensure meeting the patient's special needs in a way that improves the quality of life and overall health.

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