

Prader Willi Syndrome: Orofacial Findings and its Management

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Abstract

Prader willi syndrome (PWS) is classical but sporadic genetic microdeletion syndrome affecting both male and female and first known human genomic imprinting disorder. The cardinal features of PWS are characterised by complex behavioural problem and phenotypic presentation which changes with age. The diagnosis is mainly by specific clinical features, extra oral and intraoral findings which is confirmed by genetic testing. This case report aims to highlight the orofacial features, role of pediatric dentist, importance of awareness of paediatrician and medical team towards the orofacial condition and its effect on overall systemic health.

Keywords: Prader Will Syndrome; Childhood Central Obesity; Orofacial; Hyperphagia

Abbreviation

PWS: Prader Willi Syndrome

Introduction

PWS (OMIM Number 176270) is rare, neurogenic and complex disorder which has clinical features such as short stature, hypogonadism, characteristics behaviour problem such as obsessive-compulsive disorder, intense preoccupation with food, and depression, low muscle tone as well as involuntary urge to eat constantly which lead to severe obesity [1]. The diagnosis is mainly by evaluating presenting well defined clinical features and confirmed by genetic test which detects nearly 100% of patient with high specificity [2]. PWS has orofacial features like almond shaped eyes with upslanting palpebral fissures, triangular mouth, narrow face, micrognathia, thin upper lip, prominent forehead with intraoral findings such as poor oral hygiene, xerostomia, increase viscosity of saliva [3]. PWS requires primary care physicians and multidisciplinary approach for treatment that include ophthalmologists, paediatrician, endocrinologists and pediatric dentist. Therefore, detailed description of oro-facial features in 11 year old female child with PWS is described. The aim is to emphasize the need of early involvement of pediatric dentist in multidisciplinary team for the management of oro-facial condition.

Case Report

A 11 year old female of 42 kg reported with the chief complaint of yellowish discoloration and decayed teeth. During the complete anamnesis, natal history reported that the child was delivered at term by lower segment cesarean section with low birth weight of 1.8 kg, and did not cry immediately. She has history of feeding problem in infancy, hyperphagia with increasing age and delayed developmental motor milestone. General physical examination revealed some characteristics features such as severe central obesity, prominent forehead,

strabismus, upslanting palpebral fissures, upslanting eyes, hypertelorism, small hand, hypogonadism, exhibit mood swing, hyperactive, aggressive behaviour when contradicted and involuntary eating associated with hyperphagia (Figure 1a and 1b). Based on clinical (major and minor criteria) and genetics test confirmed the diagnosis of PWS. Intraoral and extraoral examination revealed generalised enamel hypoplasia, multiple dental caries wrt 16 26 36, poor oral hygiene, viscous saliva, gingivitis, positive gingival bleeding and mouth breathing habit (Figure 2). Panoramic radiograph revealed grossly carious teeth wrt 16 26 36 (Figure 3). The patient underwent oral prophylaxis for her gingivitis, oral rinse with chlorhexidine were instructed. The patient received prophylactic fluoride varnish application. The roots of a destroyed tooth wrt 36, 46 and root canal treatment wrt 16 followed by coronal restoration were planned. The patient was kept under regular follow up every three months showed marked improvement in oral hygiene status, gingival condition and oral mucosa. She is also under regular follow-up with pediatric endocrinologists for her obesity and other concerned systemic condition.



Figure 1a: Short stature with small hand, feet and central obesity.



Figure 1b: Upslanting palpebral fissures, almond shaped eyes, hypertelorism and mild strabismus, fish mouth appearance.



Figure 2: Intraoral photograph revealed Poor saliva production, viscous saliva and multiple carious tooth.



Figure 3: Panoramic radiograph (OPG) revealed grossly carious teeth wrt 16 26 36.

Discussion

PWS is rare multi systemic complex genetic disorder caused by lack of expression of gene on paternally inherited chromosome 15q 11.2-q13 region which may be due to paternal deletion (65 - 75%), maternal uniparental disomy (20 - 30%), and imprinting defect (1 - 3%) [4]. It occurs with an incidence of 1/10,000-1/30,000 described by Prader, Labhart and Willi in year 1956 [5]. DNA methylation technique will confirm the diagnosis and able to differentiate it from Angelman syndrome [4].

The phenotype of patient is mainly due to hypothalamic dysfunction which is responsible for hyperphagia, hypogonadism, temperature instability, high pain threshold, hypersomnia and multiple endocrine abnormalities [6].

The oral finding observed in our case is in accordance with previous reported literature which includes multiple dental caries leading to destruction of crown structure, generalized enamel hypoplasia, poor oral hygiene, gingivitis (bleeding on probing) and mouth breathing. Thick viscous saliva, decrease salivary flow rate, increase amount of salivary ions and proteins may be the underlying cause of oral findings [7]. The PWS patient having characteristics neurobehavioral alterations provides challenge to the pediatric dentist and its team because it requires behaviour management strategies for achieving patient's cooperation and provision of care for preventing and treating oral condition that will help in promoting the general and oral health of the patient. PWS can affect oral health through a person's ability to cope and cooperate with dental care, and through the daily inability to manage daily oral hygiene. The approach for the management should include screening, preventive and treatment programme for dental caries, gingival health, oral hygiene and other oral conditions [8].

The precocious diagnosis and anticipatory care is necessary for minimising the risk of progression of oral disease in advanced stage. Once the diagnosis of PWS is confirmed, the greatest benefit for patient can be achieved by taking holistic view of oral and systemic health, and working with medical and social team to achieve the most appropriate care and treatment through an integrated care pathway [5].

Therefore, the present case was treated first with behaviour management protocol, preventive schedule such as regular fluoride varnish application, oral hygiene instructions followed by oral prophylaxis, extraction of roots. Regular recall and follow up of child was done for assessing the oral hygiene status and practices.

Conclusion

PWS is rare genetic disorder displaying typical clinical presentation and intraoral findings. Pediatric dentist should be aware of the characteristics phenotypic presentation of the PWS with orofacial and intra oral condition. Similarly, medical team should be aware of intraoral condition and its consequences. Early diagnosis and proper referral can lead to a better quality of life in children suffering from PWS. The low salivary flow rate, thick and viscous saliva, higher risk of gingival inflammation, preventive schedule with regular professional cleaning is particularly important for individuals with PWS.

Source of Support

Nil.

Conflict of Interest

Nil.

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