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Letter to the editor

Dental Management of Patients with Prader Willi Syndrome

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Prader–Willi syndrome (PWS) is a genetic disorder which occurs with a frequency of about one in 10,000–30,000 live newborns. Both males and females, and all races are equally affected. PWS is a complex disorder with multiple disabilities, and the main defect is found in the hypothalamus. Child with PWS at the age between 2 and 3 years becomes constantly hungry and if the diet is not controlled, often leads to obesity and type 2 diabetes. Because of the presence of xerostomy, people with PWS have tendency to develop cavities and periodontal disease, and early application of preventive and prophylactic measures is very important. Before invasive dental treatment, consultation with a physician who treats the underlying disease is necessary, and the application of general anesthesia is often required due to mental retardation. If the pediatric dentists become more familiar with PWS, the diagnosis and treatment of oral pathology will start earlier, which is essential to improvement of the general health and the quality of life and care for these individuals. Multidisciplinary approaches are necessary for dental management of the orofacial problems in patients with PWS.

Keywords: Prader Willi Syndrome, dental treatment, periodontal disease

Prader–Willi syndrome (PWS) is a genetic disorder (typically new mutation) which was identified in 1956, and occurs with frequency of about one in 10,000–30,000 live newborns. The condition is named after Andrea Prader, Heinrich Willi, and Alexis Labhart, who described it in detail in 1956. Both males and females, and all races are equally affected. Despite the fact that PWS is a rare disease, it is one of the ten most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified. Approximately 75% of all patients have a del (15q) (1). Hartin et al. published the first feasibility study of a newly developed droplet digital polymerase chain reaction (ddPCR) examining DNA copy number differences in the PWS imprinting center (IC) region of patients with IC defects. Their cohort study included 17 individuals and concluded that ddPCR which is a cost-effective method can detect the presence of microdeletions in PWS, and have an important role in genetic counseling and recurrence risks for families (2).

PWS is a complex disorder with multiple disabilities, and the main defect is found in the hypothalamus. PWS belongs to the group of eating disorders and is associated with anorexia nervosa, bulimia nervosa, binge eating disorder, and pica. Many of these conditions carry a social stigma. In some developed countries special care dentistry services exist for young people, and without the input of dental profession all these conditions could have a detrimental effect on the primary and permanent dentition (1).

Symptoms of the disease are noticed very

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early. At birth, the child has a problem with breast-feeding because of the weak muscles and slow development. Usually, around 2 to 4 years of age, children receive a pronounced appetite, which is probably due to changes in the hypothalamus. Child becomes constantly hungry and if the diet is not controlled, often leads to obesity and type 2 diabetes. Also, death can occur due to cardio-pulmonary insufficiency (3).

Children with PWS show intellectual impairment, but are well-intentioned, and able to cuddle. They are emotionally unstable and often show surprising behavioral problems. Children are of low IQ, which requires constant care throughout their lives, and they rarely can live independently. Cryptorchidism is also very often present in males with PWS (4).

PWS individuals may also present other comorbidities, such as sleep disorders, scoliosis, constipation, dental issues and coagulation disorders. The protocol of the Children's Institute at Universidade de São Paulo was based on four main issues: diet, exercise, recombinant human growth hormone (rhGH) therapy (5), and behavioral and cognitive issues. According to the Prader-Willi eating pyramid, the diet must include a caloric restriction of 900 kcal/day and daily aerobic exercises and postural therapy (6). Also, other physical exercises were revealed to be safe and effective for improving the physical fitness of patients with PWS (7).

Analysis of 20 PWS patients of both sexes in order to determine whether the facial appearance was reflected in changes in the bony architecture with cephalometric roentgenograms revealed that a characteristic bony model might be created for PWS which could be of use in diagnosis and in the treatment of PWS patients by their orthodontist (8). Their face is elongated with sloping eyes and prominently nose saddle. Due to the presence of xerostomy, the mucous membrane of the mouth as well as the mouth itself are dry, and due to the expressed weak oral muscles, self-cleaning of the oral cavity is absent. Because of this, there is halitosis, a high prevalence of caries and periodontitis, and the appearance of teeth erosion (3). Different drugs are used to reduce appetite, but with little success. Once the patient gets weight, it becomes impossible to reduce it.

Some disorders with systemic physiological effects have a big impact on the dental hard tissues development. Among them are cystic fibrosis, HIV/AIDS, leukaemia, Alstrom syndrome, hypophosphatasia, PWS, tricho-dento-osseous syndrome, tuberous sclerosis, familial steroid dehydrogenase deficiency, and epidermolysis bullosa. An increased knowledge by dental practitioners is required about relevant treatment strategies and prognosis, disease processes and etiology of these diseases for better dental management and increasing their quality of life (9). It was also point out that Fragile X, Rett, and Prader-Willi syndromes are accompanied by varying degrees of mental retardation and challenging behaviors, and that dentists must be familiar with anticonvulsant, antihypertensive, antidepressant, and antipsychotic medications and as well as central nervous system stimulants (10).

Many case reports of PWS exist in the literature. One of them that was reported by Salako and Ghafouri, described a 5-year-old child with PWS who showed a severe form of dental caries, highlighting the importance of the early dental consultation and preventive dental procedures (11). Hsiao et al. in their case report described a 10-year-5-month-old girl from Japan with all typical symptoms associated with PWS. The girl had enamel hypoplasia, crowding over the anterior teeth, and narrow dental arch. Also, the mesio-distal widths of the present teeth were small in comparison with the national average. A retardation of the growth of the maxilla and mandible was found with X-ray cephalometric analysis. Dental treatment was performed by general anesthesia (12). A 24-year-old man with PWS with a class III malocclusion, featuring maxillary hypoplasia and severe enamel

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deficiency was described by Banks et al. Orthodontic alignment treatment, and surgical advancement of the maxilla were performed for the patient. The improvement of the final occlusion and increase of the short clinical dental crown heights were conducted by orthodontic treatment and restorative treatment to augment vertical facial height (13). A 3-year-old boy who presented grade II obesity, difficulty of locomotion, hypotonia, and history of cardiopathy was also described (14). A dental treatment under general anesthesia was planned for the extraction of the primary molar element 74 and atraumatic restorative treatment technique in the other teeth (14). In another study, a white 15-year-old male patient with dental plaque, gingivitis, viscous salivation, inserted gum and tongue ulcerations was described. The patient had also herpes encephalitis which was revealed by oral biopsy (15). Yanagita et al. described a 20-year-old Japanese man with PWS. His clinical and radiographic findings confirmed periodontal disease, anterior open bite, crowding and attrition of the lower first molars. Authors pointed out the importance of long-term follow-up of oral health care by dental practitioners, and their role to prevent dental caries and periodontal disease in patients with PWS (16). 15 patients with PWS were also followed at the Centre for Human Genetics of the University Hospital of Leuven. Nine of the patients were cavity-free, while missing and filled teeth scores (DMFT/dmft) ranged from 0 to 28. Body mass index (BMI) ranged from 16 to 42.6, and was not associated with caries experience or erosive tooth wear (17).

In conclusion, before invasive dental treatment, consultation with a physician who treats the underlying disease is necessary, and mental retardation often requires the application of general anesthesia. Because of the tendency of people with PWS to develop cavities and periodontitis, early application of preventive and prophylactic measures is necessary.

If the pediatric dentists become more familiar with PWS, the diagnosis and treatment of oral pathology will start earlier, which is essential to improvement of the general health and the quality of life and care for these individuals. Multidisciplinary approaches are necessary for dental management of the orofacial problems in patients with PWS.

Conflict of interest

The authors declared no conflict of interest.

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