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CASE REPORT

Oral health condition in a 6.5-year-old girl with Prader-Willie syndrome – case report

Stan zdrowia jamy ustnej u 6,5-letniej dziewczynki z zespołem Pradera-Williego – opis przypadku

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Abstract

Introduction. *The Prader-Willi syndrome is associated with an abnormality occurring within chromosome pairs (15q11-13). During the neonatal period, PWS children are characterized by genital hypoplasia and muscle hypotension. Hyperphagia manifests itself in the pre-school period leading to manifest obesity. The characteristic features of PWS also include low growth, small hands, mental retardation, and rebellious behavior.* ***Aim of the study.*** *To present characteristics of a case of a 6.5-year-old patient with PWS.* ***Case report.*** *The patient presented for dental treatment accompanied by her mother. Medical history was taken and examinations were performed: extraoral and intraoral. Photo documentation was made. The patient was 112 cm tall and weighed 23.7 kg. The speech is unclear, but understandable. Up to 6 years of age, no hyperphagia or excessive anger had been observed in this patient, but in recent months these issues became more prominent. At age 3, she underwent dental treatment twice under general anesthesia. At presentation, all milk teeth were affected by caries, treated or removed for that reason. There were no carious or hypoplastic lesions on the surface of permanent teeth.* ***Conclusions.*** *Due to such features as dense saliva, hyperphagia, acromicria, and behavioral and emotional factors PWS patients are particularly prone to developing stomatognathic*

Streszczenie

Wstęp. *Zespół Pradera-Williego (PWS) związany jest z nieprawidłowością występującą w obrębie 15 pary chromosomu (15q11-13). W okresie noworodkowym dzieci z PWS charakteryzuje niedorozwój narządów płciowych oraz hipotonia mięśniowa, a także pewne charakterystyczne cechy w budowie twarzoczaszki. W okresie przedszkolnym objawia się hiperfagia – niezaspokojony głód prowadzący zwykle do znacznej otyłości. Charakterystyczną cechą PWS jest również niski wzrost, niewielkie dłonie, upośledzenie umysłowe oraz zachowania o charakterze buntowniczym.* ***Cel pracy.*** *Opis przypadku 6,5-letniej pacjentki z PWS.* ***Opis przypadku.*** *Pacjentka zgłosiła się z matką do gabinetu stomatologicznego. Zebrano wywiad lekarski oraz przeprowadzono badanie przedmiotowe: zewnątrzustne i wewnątrzustne. Wykonano dokumentację fotograficzną. Pacjentka ma 112 cm wzrostu i waży 23,7 kg. Mowa dziecka jest niewyraźna, ale zrozumiała. Do 6 roku życia nie zaobserwowano u pacjentki hiperfagii ani napadów gniewu, jednak w ostatnich miesiącach cechy te nasiliły się. W 3 roku życia była dwukrotnie poddana leczeniu zabiegom stomatologicznym w znieczuleniu ogólnym. Wszystkie zęby mleczne są dotknięte próchnicą, leczone lub usunięte z jej powody. Nie stwierdza się zmian próchnicowych ani hipoplastycznych na powierzchni zębów stałych.* ***Wnioski.*** *Ze względu na gęstą ślinę,*

KEYWORDS:
Prader-Willi Syndrome, oral health, dental caries

HASŁA INDEKSOWE:
zespół Pradera-Willego, zdrowie jamy ustnej, choroba próchnicowa

diseases. Patients with PWS and their carers should be encouraged to take preventive actions at home and in the dental office. Early loss of milk teeth and certain facial dysmorphic features necessitate orthodontic treatment in PWS patients.

hiperfagię, akromikrię oraz czynniki behawioralne pacjenci z PWS są szczególnie narażeni na choroby układu stomatognatycznego. Pacjenci z PWS i ich opiekunowie powinni być zachęcani do działań profilaktycznych przeprowadzanych w domu oraz w gabinecie stomatologicznym. Wczesna utrata zębów mlecznych oraz pewne cechy dysmorficzne twarzy sprawiają, że pacjenci z PWS często wymagają leczenia ortodontycznego.

Introduction

Prader-Willi syndrome (PWS, Willi-Prader syndrome) was first described by Prader, Labhart, and Willi in 1956. PWS is a genetically determined syndrome and occurs in 1 in about 15,000-25,000 births, constant for both genders.[1,2] This syndrome is associated with an abnormality occurring within chromosome pairs (15q11-13). The most frequent cause of this syndrome is the deletion of the 15q11.2-q13 paternity chromosomal fragment (about 70%), less frequent include the disomy of chromosome 15, imprinting paternity implantation, or chromosomal rearrangement within 15q11.2-q13 (less than 1%).[3-5]

Diagnosis of the syndrome is often difficult especially in the youngest patients. The diagnosis is made using the Holm scale, which divides the symptoms into primary (1 point), secondary (0.5 points) and auxiliary (0 points). In children under 3 years of age the minimum number of points required to diagnose the syndrome is 5, while in children over 3 years the minimum is 8 points. In 2001, *Gunay-Aygun* et al. proposed other diagnostic criteria for the Prader-Willi syndrome, taking into account the variable clinical manifestation of the disorder depending on the patient's age. Preliminary diagnosis requires confirmation by genetic testing.[3-8] During the neonatal period, children with the Prader-Willi syndrome are characterized by genital malformation and muscle hypotension, which results in disturbed sucking reflex. Children have trouble eating and therefore do not gain weight or develop properly. There are also some characteristic features in the structure of the face. At the pre-school level, these features become less pronounced, but then hyperphagia is emerging at the forefront – unsettled hunger over which the patient has no control usually leading to considerable obesity. It should be noted here that Prader-Willi syndrome is the most common genetic cause of weight disorders. The patient does not feel satiated, which is probably due to impaired hypothalamic-pituitary function and endocrine pathway regulation by the axle. The characteristic feature of PWS is also low growth, so growth hormone therapy is often undertaken to favour a change in muscle-to-fat ratio and normalization of baby growth. Patients also manifest symptoms of mild to moderate mental retardation and have brighter complexion and hair colour than their relatives. Older patients exhibit obsessive-compulsive behaviours, anger and aggression, sleep and even psychotic disorders.[1,3,6] Persons with the Prader-Willi syndrome also require special attention from the dentist. Hypothyroidism disturbs the natural mechanisms of oral cleansing. Oral diseases also favour the frequent consumption of cariogenic foodstuffs as a substrate for caries-forming bacteria and a characteristic kind of saliva. Saliva secreted by patients with PWS is dense and viscous (contains a higher percentage of proteins and ions), and its secretion is 20% lower than in people without this disorder, so its natural oral cleansing capacity is limited.[9] Improper salivation promotes demineralisation of the enamel and formation of cavities, but it can also result in periodontal disease and mucosal lesions. Prader-Willi syndrome often presents hypoplastic lesions within the enamel, which also makes it more susceptible to caries. Small size of hands and limited motor abilities, as well as mental retardation make self-cleaning of teeth often ineffective, while rebellious behaviour can also impede hygienic practices performed by carers.[10]

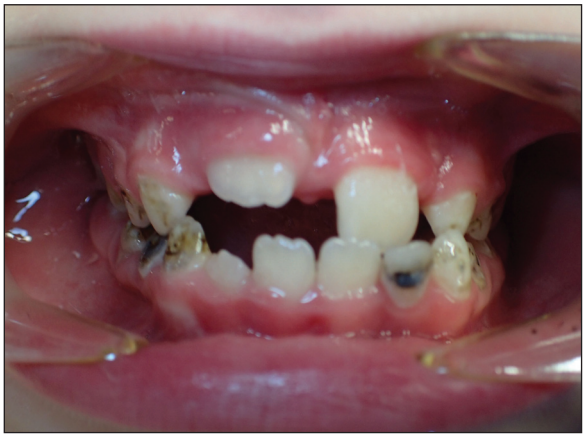

Fig. 1. *Patient – mixed dentition.*
Pacjentka – uzębienie mieszane.

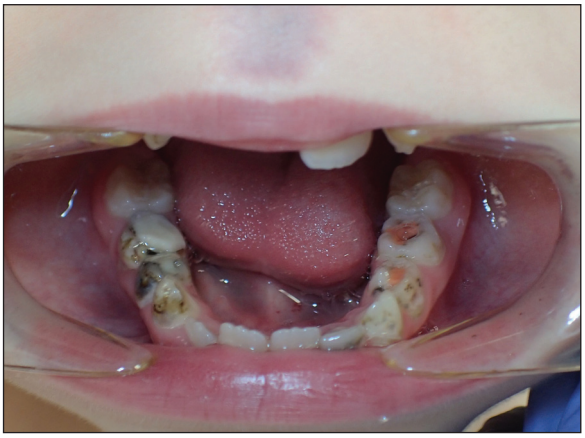

Fig. 2. *Mandibular dental arch.*
Łuk zębowy dolny.

Because of the tendency for rebellious behavior, mental retardation, and frequently considerable damage to the teeth, ambulatory treatment is not always possible and may need to be performed under general anesthesia. In this case, PWS patients need careful assessment due to several risk factors. First of all, hypotension – characteristic feature of this syndrome –increases the possibility of pulmonary complications and delayed recovery of independent respiration, so muscle relaxants should be selected and used with caution. In addition, obesity, micrognathia, high palate vesicles, and scoliosis obstruct intubation and can cause complications such as acute respiratory failure and aspiration to the lungs. In addition, due to increased appetite, preoperative starvation may not be feasible, especially in patients with intellectual disability and anger. It is, therefore, necessary to monitor patients carefully. There are also difficulties in maintaining blood glucose levels due to changes in carbohydrate and lipid metabolism. Furthermore, many children affected by neurodevelopmental disorders also have a disorder of thermoregulation. Because patients may also have pharmacologically treated co-morbidities, great caution is required when selecting anesthetic agents.[11-13]

Case report

The patient reported to the dental office with her mother. Medical history was taken and extraoral and intraoral examinations were performed. Photographic documentation was made (Fig. 1, 2, 3). The patient, who was diagnosed with Prader-Willi syndrome, was born at 31 weeks of gestation through caesarean section due to bacterial infection of fetal waters. By this time, the pregnancy was not compromised and no abnormalities were detected during the control. The newborn weighed 1400 g and was intubated. In the early months of life, Prader-Willi syndrome symptoms such as muscle hypotension and feeding problems were suspected, but assumed to be consequences of prematurity. In the second year of life, the Prader-Willi syndrome was diagnosed by genetic testing and was found to be due to maternal chromosomal disomy 15. The patient is currently undergoing

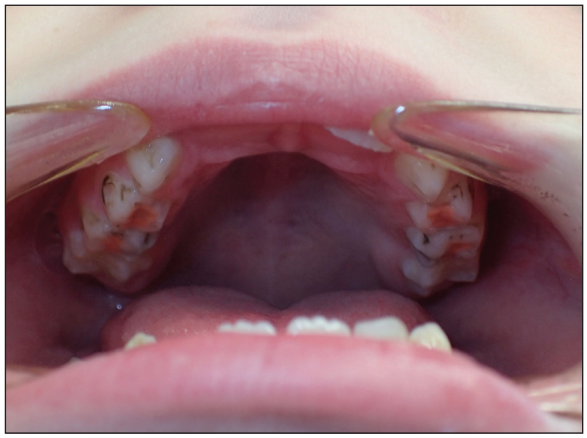

Fig. 3. *Maxillary dental arch.*
Łuk zębowy górny.

growth hormone therapy. She is 112 cm tall and weighs 23.7 kg. Her BMI is 18.9 which for the patient's age means overweight. Her growth is at 10 centile and weight at 75 centile. Although both of these results fall within the narrow range, the disproportion between them is greater than 50 centile points, which indicates an anomaly. The patient's figure is proportional, the hands are fine, with no significant deviations from the norm. The patient attends kindergarten together with healthy peers. She is cheerful and sociable, and her relationships with other children are good. The child's speech is blurred but understandable. In an interview, the patient's mother reports the presence of gallstones, but this is not associated with the occurrence of the syndrome. The patient also wears glasses because of a strabismus. The patient's mother reports that she has not observed excessive appetite and anger in her 6-year-old, but in recent months these elements are slightly on the increase. There were no episodes of skin nibbling or self-injury. From the dental point of view, the patient regularly attends check-ups, including preventive and curative visits. On the extraoral examination, it is stated that she has an amygdala shaped eyes. Other features of facial dysmorphism characteristic of Prader-Willi syndrome are not explicit. Skin discolouration can be observed, which is most likely the result of intubation. On intraoral examination, no pathological changes in the mucous membrane of the oral cavity are observed. The patient's mother and medical records do not mention the occurrence of such changes in the past. During an orthodontic evaluation, anterior occlusion was noticed – probably a consequence of the premature loss of upper incisors. Tooth 73 is in cross bite. At a later stage, orthodontic treatment may be necessary. Muscle hypothesis cannot be stated. The child's oral hygiene is moderately good. The mother of the patient says she brushes the child's teeth twice a day morning and evening with a manual toothbrush with fluoride toothpaste. The patient's mother is aware of proper dietary principles, and tries to implement them. The patient first came to the dental office at the age of 2 years, at which point the teeth of the patient had already been affected by advanced caries. An attempt was made to perform treatment, however, because of lack of cooperation, it was decided to induce general anesthesia. The patient at 3 years of age underwent dental surgery twice under general anesthesia. Treatment was successful. During these procedures extractions of teeth 52, 51 and 62 were performed, as well as conservative treatment of teeth 55, 54, 65, 64, 74, 75, 84. The other treatments were performed under the conditions of the dental office. Sedation or local anesthesia was not necessary. The patient was subjected to regular dentine impregnation treatment with 10% silver nitrate precipitated Lugol for 3 years. Replacement of leaky fillings was also performed. Presently, there are lower first molar teeth, lower central incisors and partly erupted upper molar teeth. There are no carious lesions on the surface of permanent teeth or hypoplastic lesions. The DMF index is 0. Teeth 52, 51 and 62 are missing – these teeth were removed a few years earlier due to changes in periapical tissues. The remaining milk teeth are affected by caries, df = 15. Changes in smooth surfaces were treated with impregnation, whereas lesions on the chewing surfaces of molar teeth were treated conservatively by the preparation of cavities and fillings with glass ionomer and compomer. Currently, the patient is regularly subjected to dental control, conservative treatment and fluoridation with Duraphat fluoride varnish.

Discussion

Although the Prader-Willi syndrome is a relatively rare disorder, several cases of oral health and case-control studies in larger patient groups have been reported in the literature. All authors confirm that PWS children are at high risk of oral disease. In our patient, all primary teeth were affected by caries, she is also at high risk of permanent dental caries but no carious lesions are present at the moment. *Bailleul-Forestier* et al. evaluated oral health in 15 PWS patients. Among patients, the DMFT/dmft ratio was between 0-28, and 9 patients were caries-free. There was no association between high BMI and carious lesions.[14] *Scardinia* et al. described a case of a 13-year-old girl with Prader-Willi syndrome who had hypoplasia of enamel and carious lesions. There was limited opening

of the mouth (30 mm) and small hands, which made it difficult to clean the mouth properly. There was also reduced salivation and related candidia infections in the oral mucosa.[15] *Saeves* et al. studied children and adults with PWS in terms of cavities of non-carious origin. They found that erosive and abrasive lesions are a serious problem in this group of patients often to the extent that prosthetic treatment is necessary. There was also a lower rate of salivation in patients with PSW who did not have a higher incidence of caries.[16-18] Many children with the Prader-Willi syndrome have a problem with proper oral cleansing. The common mental impairment negatively affects the ability to learn the proper brushing techniques. Characteristically, small hands impair the patient's manual skills, and the tendency for rebellious behaviour makes those affected reluctant to accept parental support. The case of a 20-year-old patient who often falls asleep without brushing his teeth due to uncontrolled drowsiness is common in PWS. Bad oral hygiene and insufficient saliva secretion are associated with an increased risk of periodontal disease and damage to hard tissue. *Yanagita* et al. described a case of a 20-year-old patient with Prader-Willi syndrome who was diagnosed with localized periodontitis due to poor oral hygiene and crowding of teeth that makes brushing difficult.[19] *Gadens* et al. described the case of a 10-year-old PWS patient. Intra-oral examination showed a significant accumulation of biofilm, presence of gingivitis, dry mouth and stomatitis.[20] *Saeves* et al. compared a group of PWS patients with a control group, finding that the Gingival Index was higher in the study group than in the control group.[17] Children with the Prader-Willi are usually mildly to moderately impaired. Many of them are prone to aggressive and rebellious behaviour, therefore, dental treatment can often be difficult to undertake and the procedure must be performed under general anesthesia. Our patient was treated twice under general anesthesia at the age of 3 years. During these procedures, extraction of three milk teeth was carried out and another seven received conservative treatment. There were no complications despite publications reporting the risk of general anesthesia in people with PSW due to childhood hypotension, obesity and morphological disorders of the stomatognathic system. *Soo Jeong Lee* and *Kwangwoo Baek* also described the case of a 3.9-year-old girl with PWS with multiple carious lesions treated under general anesthesia. Extractions of teeth 62, 74 and 84 were performed, while the remaining teeth were treated with steel crowns and composite materials.[13] *Ki Un Song* et al. described the case of an 18-year-old boy treated under general anesthesia. The reason for coming to the clinic was the injury to both upper central incisors caused by a fall. Initially, treatment with 30 mg of Midazolam was administered intramuscularly; however, due to the lack of patient's cooperation, only pulpectomy of tooth 11 and a temporary filling in tooth 21 were performed. The endodontic treatment of both incisors and permanent reconstruction were performed under general anesthesia.[12,13,20] Children with PWS have characteristic morphological facial features such as infantile dolichocephaly, narrow face or reduced biconcave size, amygdala eyes, small mouth, narrow jaw, gothic palate and others. Early loss of milk teeth due to advanced caries may result in orthodontic defects. *Gardens* et al. in the orthodontic study have identified a Class II defect, a gothic palate and anterior teeth crowding. Radiological examination revealed a delayed eruption of permanent teeth.[21-23]

Conclusions

1. Due to such features as dense saliva, hyperphagia, acromicria, and behavioral and emotional factors PWS patients are particularly prone to developing stomatognathic diseases.
2. Patients with PWS and their carers should be encouraged to take preventive actions at home and in the dental office.
3. Early loss of milk teeth and certain facial dysmorphic features often necessitate orthodontic treatment in PWS patients.

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