Oral diseases in a patient affected with Prader-Willi syndrome

G.A. SCARDINA, G. FUCÀ, P. MESSINA

ABSTRACT. Background A case of Prader-Willi syndrome (PWS) in a 13-year-old girl is reported. The patient presents with systemic and oral manifestations of the disease. Case report The patient shows: obesity, hypotonia, mental retardation, small hands, prominent forehead, strabismus, hypoplastic teeth, poor oral hygiene, caries, oral candidiasis and thick, sticky saliva. This case is reported to underline the importance of the oral and dental problems of these patients. Caries and oral candidiasis are correlated with the reduced secretion of saliva and with poor oral hygiene. The role of paediatric dentistry is considered to be necessary for the prevention of oral complications of this syndrome.

Keywords: Prader-Willi, Oral, Caries, Candidiasis, Oral hygiene.

Introduction

Prader-Willi syndrome (PWS) is a complex genetic disorder that includes short stature, mental retardation or learning disabilities, incomplete sexual development, characteristic behaviour problems, low muscle tone, and an involuntary urge to eat constantly, which, coupled with a reduced need for calories, leads to obesity. PWS is identified in approximately one in 25,000 births. Because many affected persons are not diagnosed at an early age, this statistic is likely an underestimate. More realistic estimates of PWS prevalence range from one in 10,000 to one in 15,000. PWS affects both sexes equally and occurs in subjects of any race. Newborns with PWS are unable to suck well enough to intake sufficient nutrient, due to the low muscle tone. Often they must be fed through a tube for several months until muscle control improves. In the following years, children with PWS develop an increased interest in food and quickly gain excessive weight. The obesity that results is the major cause of illness and death in this disorder. Obesity in PWS can cause high blood pressure, breathing difficulties, diabetes and other problems. Subjects with PWS have a characteristic appearance and speech quality, significant learning disabilities or mental retardation, and various other problems. A number of these features must be present for a clinical diagnosis of PWS, and specific genetic tests are available to confirm diagnosis [Gunay-Aygun et al., 2001]. Early diagnosis of PWS gives parents an opportunity to manage their child’s diet and avoid obesity and its related problems from the start [Holm et al., 1993]. Although consensus clinical criteria for the diagnosis of PWS were published, genetic testing has become the standard because it detects nearly 100% of patients with PWS, is highly specific, and can diagnose PWS earlier than it could be possible basing on clinical criteria [Cassidy and McCandless, 2005; Crino et al., 2003; Varela et al., 2005]. Treatment of a child affected with PWS involves the primary care physician and a multispecialty team that includes an ophthalmologist to evaluate for myopia and strabismus, a paediatric endocrinologist for consideration of growth hormone treatment, a developmental paediatrician, and a paediatric dentist to manage caries, oral candidiasis and xerostomia [Bazopoulou-Kyrkanidou and Papagiannoulis, 1992; Friedlander et al., 2003].

Case report

Oral diseases in a 13-year-old female are described. Cytogenetic analysis in the family revealed a paternal microdeletion including SIM1 at 6q16.3.

Observation revealed a girl of short stature, with
marked obesity, prominent forehead, bitemporal narrowing, strabismus, small hands (Fig. 1). At birth she weighted 3 kg. Poor sucking caused difficulty in feeding during the first year of life. She developed polyphagia at the age of 5 years.

On the basis of the clinical and cytogenetic findings the diagnosis of PWS was made.

The teeth observed were:

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Poor oral hygiene (Fig. 2) as well as poor saliva production and viscous saliva were observed (Fig. 3). There were extensive deposits of plaque around all the teeth, particularly in the lower front region and lateral back region. Caries were observed (1.7, 2.7, 3.7, 4.7).

Periodontal examination showed gingivitis, periodontitis and positive bleeding gingival index. Radiographic examination showed a compromised temporomandibular articulation (Fig. 4). Enamel hypoplasia was observed (1.1, 1.4, 2.1, 2.5, 3.2, 3.3, 4.2, 4.6). Lingual epithelium alterations were also observed. Oral test confirmed the diagnosis of oral candidiasis (Fig. 5). Oral microsomia (<30 mm) and small hands made the maintenance of oral hygiene particularly difficult (Fig. 6).

The patient underwent deep scaling for her periodontal disease which resulted in a marked improvement of her periodontal disease. Anti-fungal therapy was administered (Micotef oral gel).

Medications to increase salivary flow and saliva substitutes were administered (Oralbalance gel).

Oral rinses with clorexidine were prescribed to prevent mycotic infection. The patient also received a prophylactic fluoride treatment.

The roots of a destroyed tooth were extracted (3.6).

The follow-up (clinical controls every three months) showed: a remarkable improvement of the periodontal disease (negative gingival bleeding index); reduction

![Fig. 1 - Small hands in PW patient.](image1)

![Fig. 2 - Poor oral hygiene in PW patient.](image2)

![Fig. 3 - Poor saliva production and viscous saliva observed in PW patient.](image3)

![Fig. 4 - Radiographic examination showed the compromised temporomandibular articulation.](image4)
of the incidence of new caries and the absence of mycotic infections. The oral mucosa appeared normal. The patient had been recently hospitalized for her obesity and routine systemic medical controls.

**Discussion**

Prader-Willi Syndrome was first described by the endocrinologists Prader, Labhart and Willi in a report describing an unusual pattern of abnormalities including decreased fetal activity, very poor muscle tone, feeding problems in infancy, underdeveloped sex organs, short stature and retarded bone age, delay of developmental milestones [Krautmann et al., 1981].

Major clinical findings described are: neonatal and infantile central hypotonia that improves with age (floppy baby); feeding problems in infancy and possible failure to thrive.

Facial features are: dolichocephaly in babies, narrow face, almond shaped eyes, thin upper lip and down turned corners of the mouth, bruxism [Krautmann et al., 1981; Wigren and Hansen, 2003].

Minor clinical findings described are: short stature, small hands and feet, narrow hands, eye abnormalities, hypo-pigmentation, thick, viscous saliva with crusting at the corners of the mouth, speech articulation defects [Krautmann et al., 1981; Wigren and Hansen, 2003].

The oral abnormalities of the patients suffering from PWS have included: low basal salivary secretion which persists after looking at food for 10 min, advanced dental caries with hypoplastic enamel, delayed eruption and cranio-facial abnormalities which include almond-shaped palpebral fissures, narrow bifrontal diameter and fish mouth appearance (Tables 1, 2) [Bazopoulou-Kyrkanidou and Papagiannoulis, 1992; Friedlander et al., 2003; Greenwood and Small, 1990; Salako and Ghafouri, 1995]. The uncontrolled polyphagia leads to obesity, which becomes more obvious during adolescence. Severe obesity, besides being the main handicap for those patients, limits life expectancy. Deaths have been reported to occur between the ages of 3 to 30 with the most common cause of death, complications of obesity [Vogels et al., 2004]. The case report is

**FIG. 5 - Oral candidiasis in PW patient.**

**FIG. 6 - Oral microsomia (<30 mm) in PW patient.**

**TABLE 1 - Dental concerns in PWS.**

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<td>• Poor oral hygiene may lead to rampant caries</td>
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<td>• Frequent exposure to carious food if food intake is not closely monitored due to hyperphagia</td>
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<tr>
<td>• Poor saliva production and viscous saliva compound caries risk</td>
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<td>• Caution with anaesthetics and sedative medications including pain medication due to unpredictable responses</td>
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**TABLE 2 - Dental recommendations in PWS.**

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<td>• Give Oral hygiene instructions to caregivers and review eating habits</td>
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<tr>
<td>• Medication to increase salivary flow or saliva substitutes may be beneficial for PWS patients who exhibit xerostomia</td>
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<td>• Use caution when sedating or prescribing medications due to unexpected responses</td>
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described to emphasise the importance of the oral problems of these patients. Enamel hypoplasia and early dental caries have been reported at an overall frequency of 40% with no significant difference in the incidence between the deletion and no deletion chromosome subgroup [Goldstone, 2004].

Caries and oral manifestation of candida infection are caused by excessive consumption of sugar, in connection with the hypoplastic enamel, the reduced secretion of sticky and thick saliva, and poor oral hygiene [Friedlander et al., 2003].

Conclusion
The case report of PWS demonstrates the classic abnormalities described in this syndrome, including poor oral hygiene and caries. It has been shown that oral candidiasis and periodontal disease can also occur. The oral manifestations of the disease can be managed by early diagnosis with preventive procedures. The role of the paediatric dentist is fundamental in the medical team for the management of the children with PWS.

References