Orofacial Findings and Dental Management of Williams-Beuren Syndrome

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Williams-Beuren syndrome is a rare congenital disorder involving the cardiovascular system, mental retardation, distinctive facial features, and tooth anomalies. The aim of the present report is to show a 10-year-old girl with Williams-Beuren syndrome, her general and orofacial clinical characteristics and the dental management.

Keywords: *Williams-Beuren syndrome*
J Clin Pediatr Dent 36(4): 401–404, 2012

INTRODUCTION

Developmental anomalies of the dentition are frequently observed in several craniofacial syndromes; during childhood, various aberrations in number, size, shape and position may lead to the appearance of dentofacial problems in oral function, esthetics and dental occlusion;[1] also, the dental management of these problems represents a challenge for the professional who attends children in the dental office.

Williams-Beuren syndrome is a rare congenital disorder that was first described in 1961 as an association among supravalvular aortic stenosis, mental deficiency and characteristic facies that were present in 4 children.[2] Later, Beuren *et al* reported the syndrome independently and expanded the phenotype to include peripheral pulmonary artery stenosis and dental malformations.[3]

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The disorder is characterized by growth and developmental deficiencies; cardiovascular defects; unusual dysmorphic facial features ("elfin" facies); dental anomalies and several specific conductive and neurological alterations, including mild-to-severe mental retardation; renal disorders; and microcephaly.[4,5] Patients with this syndrome exhibit, among the main craniofacial abnormalities, wide mouth, thick lower lips, tongue thrusting, full prominent cheeks, long philtrum, nose with depressed nasal bridge, heavy orbital ridges, medial eyebrow flare, low ear implantation, anterior inclination of the maxilla, and retrusive mandible. They also show some abnormalities both in primary and permanent teeth: high incidence of caries, malocclusions, enamel hypoplasia, supernumerary teeth, oligodontia, microdontia, taurodontism, pulp stones, excessive interdental spacing, short roots, and aberrant tooth shape including peg-shaped teeth.[1,6-8]

Although the true incidence of the syndrome is unknown, it is estimated to be 1 *per* 20,000–50,000 live births;[8,9] in Norway, however, an incidence of 1 in 7,500 live births was reported.[10] It affects both sexes equally, in all ethnic groups, around the world.[1] From the genetic point of view, Williams-Beuren syndrome usually occurs as a sporadic new dominant condition, although parent-child transmission has been reported.[11] Its etiology is not thoroughly clear; however, it is thought to be caused by submicroscopic heterozygous deletion of human chromosome No. 7. Several of the TFII-I family have been proposed as responsible for tooth anomalies during odontogenesis.[12]

Diagnosis of Williams-Beuren is based on recognition of the characteristic pattern of typical dysmorphic facial features, developmental delay, short stature, connective tissue abnormalities affecting the cardiovascular system, a particularly cognitive profile with learning difficulties, and sometimes transient infantile idiopathic hypercalcemia.[9,13] In order to confirm the diagnosis, specific molecular genetic tests and prenatal testing on fetal cells obtained from the

chorionic villus at about 10 to 12 weeks' gestation or amniocentesis at 15 to 18 weeks are available; such prenatal tests are rarely carried out since most cases are a single occurrence in a family, so there are no prenatal indicators for pregnancy risks. There is no specific treatment for Williams-Beuren syndrome. The systemic and local anomalies and developmental and cognitive disabilities should be approached in an individualized manner, according to the characteristics of each particular case. Treatment should be addressed from a multidisciplinary perspective, mainly through early stimulation, phonoaudiology, psychopedagogy and physical and occupational therapy.[14,15]

The aim of this report is to present the case of a 10-year-old girl with Williams-Beuren syndrome, her clinical characteristics, dental management, diagnosis and treatment procedures, and the short- and medium-term prevention plan.

CASE REPORT

A 10-year 9-month-old girl and her parents visited the Postgradute Pediatric Dentistry Clinic, requesting a routine dental appointment. She was the parents' first pregnancy, diagnosed with mild mental retardation, some language problems, complete vaccination scheme, and normal weight. Pediatric physical evaluation revealed a short stature, left-side kyphoscoliosis (Figure 1) and clinodactyly (Figure 2).

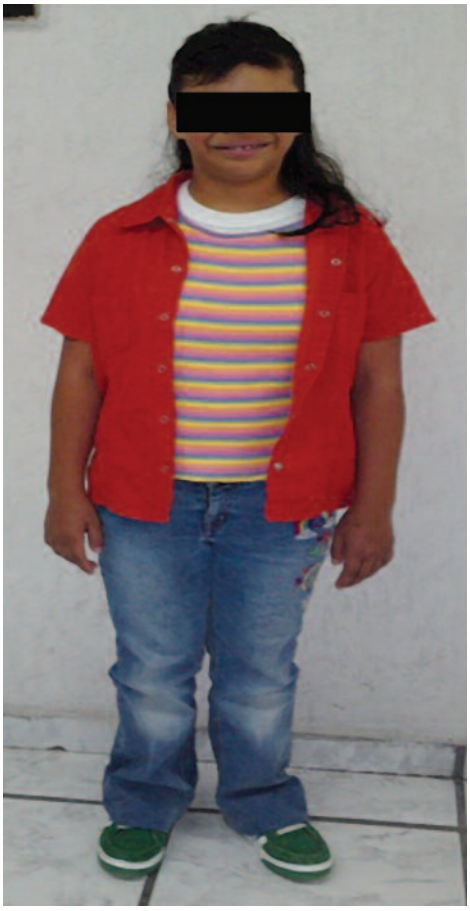

Figure 1. View of patient's whole body

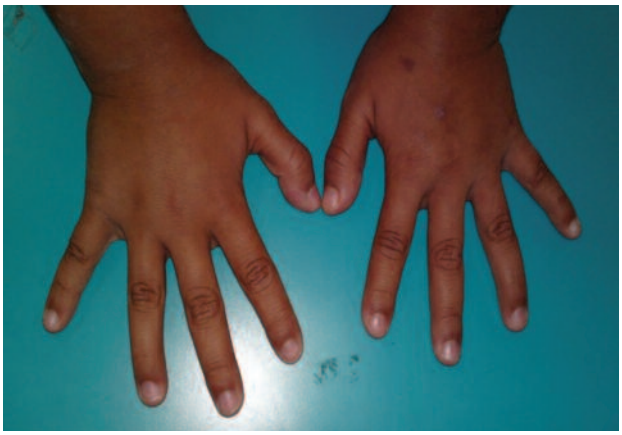

Figure 2. View of hands. Note clinodactyly

For the medical history, her parents reported an at-birth subdural hematoma caused by forceps; she did not start to walk until age 2. At that age she was diagnosed with Williams-Beuren syndrome and hypercalcemia. In addition, the girl had a unique asthma episode at 6 years, and she was diagnosed with attention deficit disorder at 7 and chickenpox at 8. Also at 8, she underwent open-heart surgery to correct aortic stenosis, and she submitted to 2 catheterizations during her life. According to the parents, the child had a regular level of oral hygiene, low sugar consumption, and never practiced any harmful oral habit. Bottle feeding was withdrawn at the age of 1. She had been orally checked only once, together with a complete dental cleaning, at a previous appointment, 4 four months before. Craniofacial examination (Figure 3) showed a symmetric dolicofacial face, thick lips, hypertelorism, wide and depressed nose, sparse, bushy eyebrows, normal TMJ, convex profile with apparent mandibular retrognatism and short chin. Intraoral examination revealed the mixed dentition stage, wide dental arches, wide maxillary anterior spacing, mandibular anterior crowding, mild enamel hypoplasia of the 4 permanent first molars, and evidence of caries in the primary molars (Figure 4). Dental treatment consisted of placing stainless steel crowns on the mandibular right first and second primary molars and extraction of the maxillary left first primary molar without placing a space maintainer. All of these procedures were carried out under local anesthesia without the necessity of

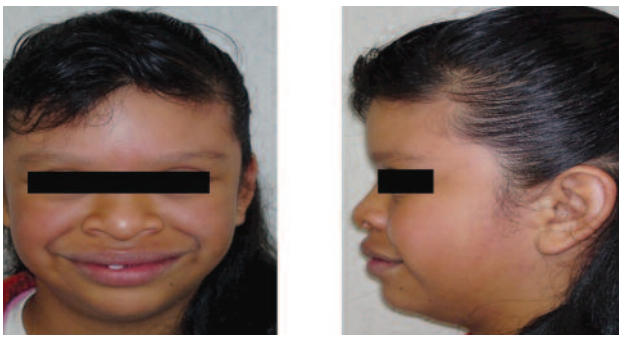

Figure 3. Craniofacial views.

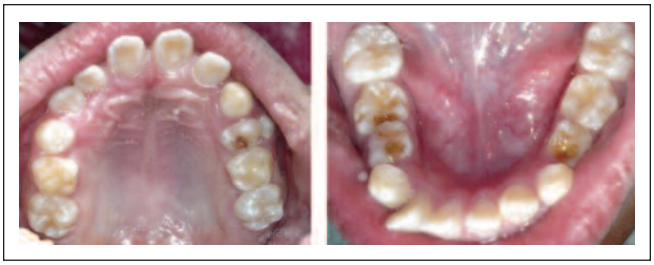

Figure 4. Intraoral views.

antibiotic prophylaxis. Although the child was overprotected by her parents, her behavior was characterized as willing and fairly cooperative, with some distraction episodes. Immediate preventive management consisted of placement of resin sealants on her 4 permanent first molars, teaching and reinforcing brushing technique and flossing, a complete prophylaxis, and fluoridated varnish applications on several enamel white spots. Orthodontic treatment was planned for the near future, and preventive control appointments were programmed every 3 months to motivate her brushing and administer new dental prophylaxis and fluoridated varnish applications.

DISCUSSION

Williams-Beuren syndrome is a rare genetic disorder, with the frequent occurrence of orofacial and dental abnormalities. Because of these problems, the pediatric dentist plays a significant role in the management of the affected patient.[16] The present case of this syndrome was considered to be of only mild seriousness, based on the unremarkable facial features (except the lip wideness), the slight mental and psychomotor deficiencies, and the almost normal physical development exhibited by the patient; however, other typical clinical manifestations such as aortic stenosis and hypercalcemia were present. It is worth noting that the cardiovascular abnormalities inherent in these patients must be taken in account by the pediatric dentist because heart damage can be so serious that even routine dental treatment (eg, local or general anesthesia) could put the patient's life at risk. Therefore, consultation with a pediatric cardiologist is strongly recommended for possible antibiotic prophylaxis and other managing considerations to minimize the risks. On the other hand, hypercalcemia is associated with high caries incidence, because of the enamel hypoplasia and hypomineralization, and with delayed tooth eruption.[17] Hypercalcemia has also been associated with placental abnormalities and elevated levels of—or hypersensivity to—vitamin D.[18] Other alterations reported as risk factors enhancing caries susceptibility in children and adolescents with the syndrome are anorexia and vomiting.[19]

A few cases of Williams-Beuren syndrome in children have been published in the dental literature. Boraz reported a case of an 8-year-old boy with multiple carious teeth who had been diagnosed with the syndrome 5 years previously. Because of his extremely uncooperative behavior, restorative treatment took place under general anesthesia with prior prophylactic antibiotic coverage. Amalgam restorations, pulpotomies, stainless steel crowns and extractions—all on primary teeth—and the placement of a lingual arch were performed.[16] Cobo *et al* described a girl of the same age with moderate mental deficiency, typical elfin facies, delays in somatic growth and tooth eruption, oligodontia and multiple microdontia; however, the patient was checked only for orthodontic treatment of a severe anterior open bite.[19] Onçag *et al* presented a boy of 8 who exhibited an important psychomotor developmental delay, serious mental deficiency, and advanced caries. Dental treatment consisted of tooth extractions and placement of space maintainers under local anesthesia since general anesthesia was contraindicated because of his severe cardiovascular problems.[17]

Management modalities in these cases and ours show that there are no contraindications for routine dental treatment such as that given to the girl described herein, except perhaps for patients with severe mental deficiency or a serious cardiac condition. However, as previously mentioned, it is always advisable to consult with a pediatric cardiologist before doing any dental procedure. Besides, early dental examinations and parent counseling are important in the management of compromised medical patients. Likewise, rigorous preventive and dietary programs must be individually designed—especially in children with severe enamel hypoplasia, high caries rates and vulnerable cardiac conditions—to minimize the risk of infection in the oral cavity.[16]

CONCLUSION

Because of systemic and local problems, particularly of the cardiovascular system, orofacial anomalies and possible mental deficiency, frequent dental examinations and proper preventive and dietary practices may have significant value in improving the quality of life of children with Williams-Beuren syndrome.

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