CASE HISTORY REPORT

ABSTRACT

Opitz G/BBB syndrome is a genetic condition characterized by several abnormalities along the midline of the body, such as hypertelorism, craniofacial deformities, and dysphagia. This study reports the clinical features of Optiz syndrome and its importance in the knowledge of patients who are developmentally challenged as a whole, in order to establish adequate dental treatment for a certain clinical case. A 19-year-old patient visited the Paulista University for a dental treatment. The extraoral examination revealed ocular hypertelorism (wide-spaced eyes), oblique eyelids, epicanthus, low-set cart, and intellectual disability. During the intraoral examination, large caries lesions were observed surrounding the braces of the fixed orthodontic appliance and poor oral hygiene. Preventive and restorative treatments were carried out. It was concluded that the knowledge of patients with special needs as a whole is mandatory for an adequate dental treatment. This is a case report that highlights the importance of dentist and interdisciplinary care attendance for all patient systems, the examination and analyses should not be restricted to the oral cavity.

KEY WORDS: Opitz G/BBB syndrome, pathology, dental treatment

Dental treatment of a patient with Opitz G/BBB syndrome

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Introduction

The Opitz G/BBB syndrome is a rare genetic disorder characterized by several abnormalities along the midline of the body. Its exact way of genetic transmission is still unknown, although it can be related to the autosomal dominant inheritance or X-linked syndrome. There are two forms of Opitz syndrome, which are distinguished by their genetic causes and patterns of inheritance: X-linked Opitz syndrome, which is caused by mutation in a specific gene MiD1 (midline 1) on chromosome X, and autosomal dominant form, which is caused by mutation in the still unidentified gene on chromosome 22[1–6] caused by deletion of a small piece of chromosome 22, specifically 22q11.2, because of this researchers consider this condition as part of the 22q11.2 deletion syndrome.[7–9] Signs and symptoms of the syndrome include congenital heart disease (74% of subjects), palatal abnormalities (69%), velopharyngeal incompetence, submucous cleft palate, facial features (predominantly in Caucasians), and learning disabilities (70–90%).[5,6,9–13] Seventy-seven percent of individuals have immune deficiency, regardless of their physical presentation. In addition, they have hypocalcemia (50%), significant feeding problems (30%) caused by severe dysphagia, nasopharyngeal reflux prevalence of cricopharyngeal muscle, abnormal cricopharyngeal closure and/or diverticulum.[13–16] Renal anomalies (37%), hearing loss, laryngeal-tracheal-esophageal abnormalities, growth hormone deficiency, autoimmune diseases, seizures, and skeletal abnormalities are also reported.[13,16–18] Craniofacial findings include ear abnormalities, nasal, hooded eyelids, ocular hypertelorism, cleft lip and palate, facial asymmetry with aspects of "be crying," craniosynostosis and, eventually, long face and malar flatness.[17–20]

Other features include hypertelorism (wide-spaced eyes), hypospadias, particularly in male (urethral opening at the distal portion of the penis), cleft lip with or without cleft palate (50%) or just a cleft palate, also high-arched palate and thin upper lip.[21] Obstruction of the anal opening (imperforate anus) reported in 50% of the cases. Abnormalities such as flat nasal bridge, low-set ears, thin upper lip can also be observed, but these features vary among the affected individuals, even within the same family.[1,2] Signs and symptoms of autosomal dominant form of the condition are compatible to those observed in X-linked, which include cleft lip with or without cleft palate, while only the cleft palate is the most common in autosomal dominant form. In female, when the

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syndrome is X-linked, usually are slightly affected, often hypertelorism is the only sign of the disease.[1–5]

The diagnosis of Opitz syndrome G/BBB is determined based on clinical findings and is classified into major and minor results based on the frequency of occurrence. A family history consistent with X-linked inheritance supports further diagnosis.[4,20,22,23]

Structural abnormalities of the central nervous system (CNS) have occasionally been reported in patients with Opitz syndrome, which include inversion and left hippocampus dilatation of the temporal horn ipsilateral normal and corpus callosum,[24] hypoplasia of the corpus callosum, and cerebellar hypoplasia.[25] In individuals identified with mutation in MiD1, their brain MRI show midline malformations in 40% of the cases, and agenesis or hypoplasia of the corpus callosum and/or cerebellar vermis.[26]

This report proposes a description of general and oral clinical condition of a patient with Opitz G/BBB syndrome, and tries to understand and propose an appropriate treatment for the case.

Case report

A 19-year-old female patient (leucoderma Caucasian), born in São Paulo, Brazil, from a low socioeconomic income, visited, accompanied by her mother, the clinic of the Center of Study of Special Patient (CEAPE) at Paulista University, São Paulo. The reason for consultation was "toothache when chewing." Her medical-dental history, current and past medical history, was studied to determine the general health of the patient.

During the anamnesis, it was reported that the patient had the Opitz G/BBB syndrome (confirmed by pathology report). Her family history reported diabetes and hypertension. She had a premature birth in the 8th month of gestation by cesarean section. During birth, the child had jaundice, generalized hypotonia, and altered reflexes, being diagnosed pathologically.

Medical evaluation, as described by the doctor, reported cardiac anomaly, interventricular defect, dysphagia, reflux, involvement of the upper respiratory tract and lungs, bronchitis and recurrent pneumonia, myopia, mental deficiency, growth hormone deficiency, and skeletal anomalies.

The extraoral physical examination findings reported short stature (4 feet and 75 inches), weighing 78 lb, webbed neck, long fingers with endings in the tip and square nails (Figure 1). In addition, the face was elongated, contained craniofacial asymmetry with respect to "be crying," broad and flattened forehead (Figures 2A and B), ocular hypertelorism (Figure 3), strabismus, oblique and hooded eyelids, epicanthal folds, nasal bridge flat, hypotonic cheeks, micrognathia, and low-set ears.

During the intraoral examination, it was noted that thin and flabby upper lip, high palate and fibrosis in the palate center area (Figure 4), hypotonic tongue, and short lower lip frenum.

The patient's teeth had upper and lower braces (Figure 5), her mother reported that the patient has been through orthodontics treatment for 4 years. There were extensive demineralized white spot lesions with some cavitation around the brackets (Figure 5); the elements 13, 16, 22, 23, 24, 26, 34, 36 and 46 other dental elements presented with white spots on the face cervical vestibule. Clinical examination also showed the absence of dental elements 17, 27, 35, and 45 (Figure 6).

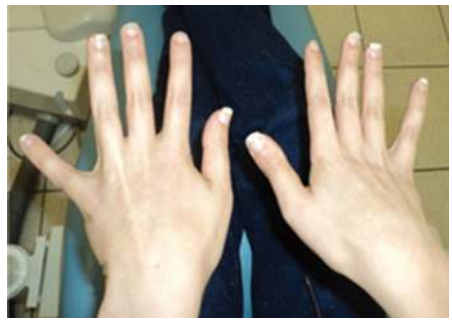

Figure 1. Appearance of hands showing elongated fingers and square nails.

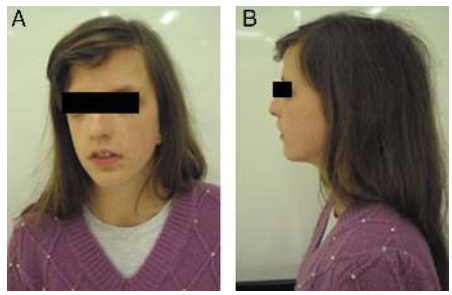

Figure 2. (A) Front view showing facial asymmetry. (B) Profile view demonstrating flattening of the malar.

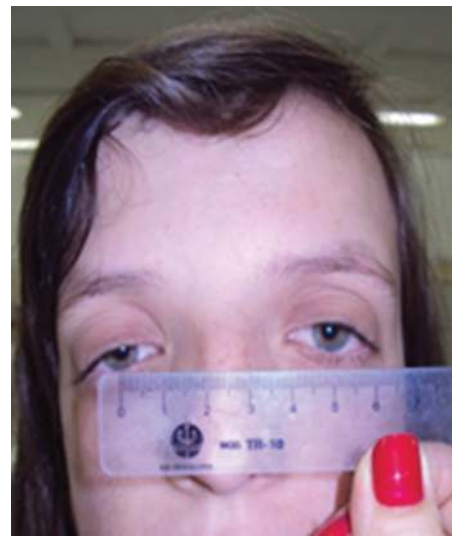

Figure 3. Interpupillary distance (hypertelorism).

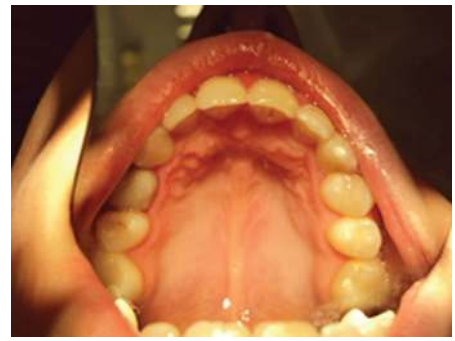

Figure 4. Feature ogival palate with fibrosis of palate center, where it is observed restorative treatment completed.

Figure 5. Upper and lower arcade with fixed orthodontic appliances.

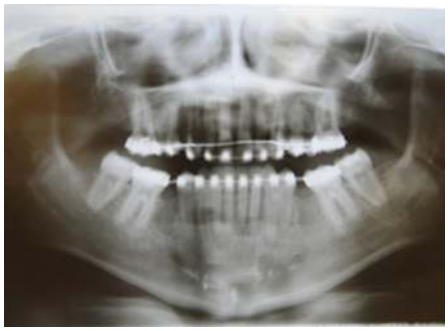

Figure 6. Panoramic radiography, where it is observed the absence of dental germs 18, 28, 38, and 48, besides the absence of the elements 17, 27, 35, and 45.

The elements 13, 14, 15, 16, 23, 24, and 25 had 100% biofilm accumulation and bleeding gingival, and the element 26 had signs of gingival inflammation with bleeding on probing, the diagnosis was generalized gingivitis, salivary flow was moderate, but low salivary pH around 4.5 demonstrating that the patient had high activity of caries.

As complementary tests were performed, panoramic radiograph showed the absence of third molars germs, as shown in Figure 6, and expansion of pulp cameras in most teeth also confirmed by periapical radiographs (Figures 7A-C), which also confirmed the carious lesion diagnosis.

A request was made to the orthodontist for the removal of the braces to restore oral health of the patient. After removal of the braces, carious lesions were found on the buccal surfaces (Figures 8A and B) and proximal of the elements 13, 16, 22, 23, 26, 36, and 46 plus 11 and 21, the element 24 was diagnosed with extensive damage to mesial-distal occlusal caries and pulp involvement, which was diagnosed by periapical radiography and thermal tests. An immediate treatment protocol was conducted to provide oral health education to the patient and her mother. The measures for adequate oral environment included periodontal procedures with prior medication of prophylactic antibiotics as recommended treatment protocol by the American Heart Association (AHA) for patients presented with heart disease (2.0 g of amoxicillin 1 hour before the procedure) after performing, scaling, and

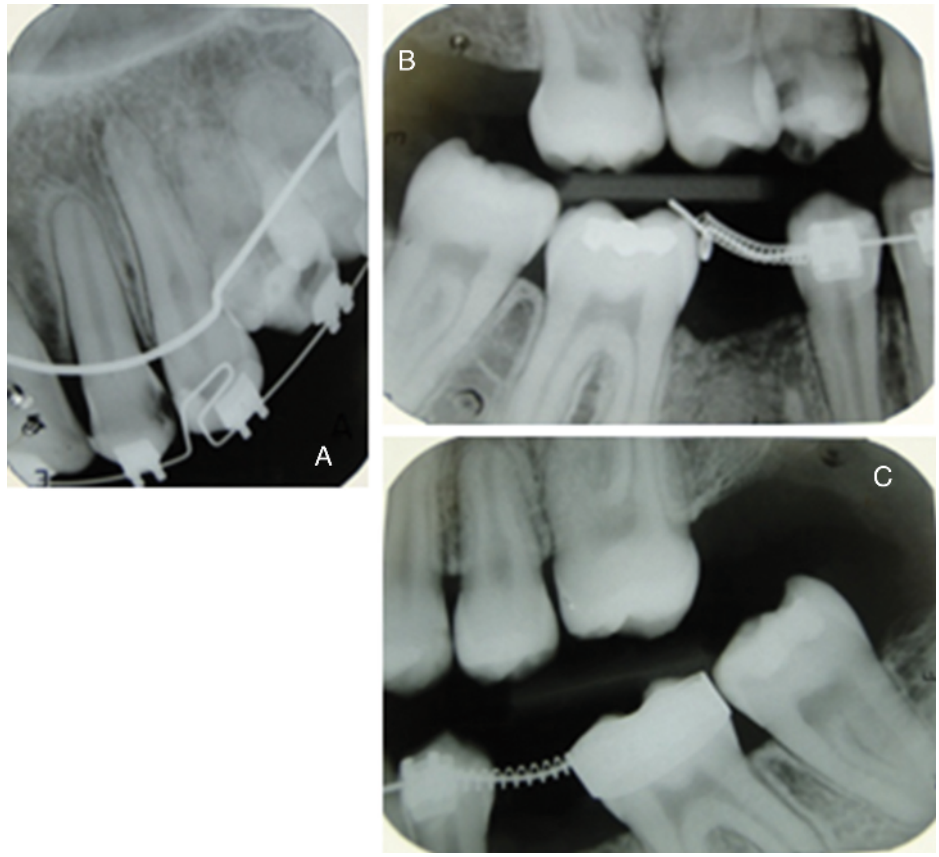

Figure 7. (A) Periapical radiograph shows deep caries that will need root canals treatment. (B), (C) Bite-wing radiography shows deep caries that will need root canals treatment.

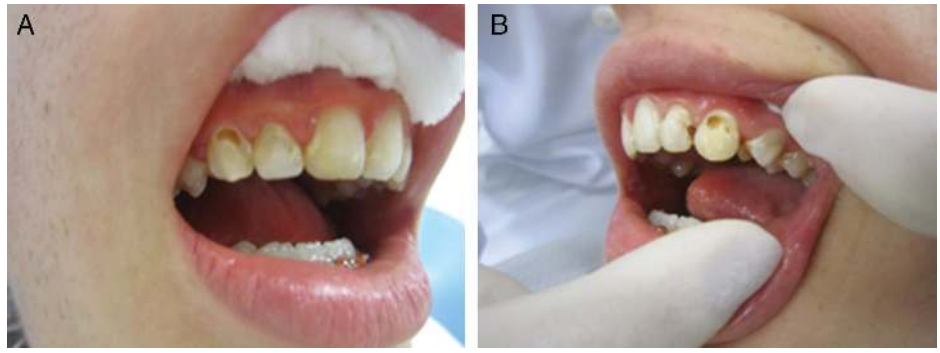

Figure 8. (A), (B) Appearance immediately after removal of a device is shown where caries lesions in the left and right.

root planning the upper and lower jaw. To treat white spot lesions without cavitation, fluoride therapy with fluoride varnish was used once a week for 7 weeks. In addition, the remineralization of structures performed and dental sealants were placed on the occlusal surfaces of the elements 14, 15, 34, and 44. In line with the recommended treatment protocol, endodontic treatment of the tooth 24 with previous antibiotic therapy medication was planned as mentioned above, this protocol was used only for the canal root l instrumentation procedure and post-instrumentation phase, the patient was not medicated. At the end, it was restored with microhybrid composite. The other dental elements 13, 16, 22, 23, 26, 35, 36, and 46 were restored with composite resin in their buccal surfaces (Figure 4).

At the end of this stage, reinforcing preventive oral health program, it was found, after 1, 3, and 6 months, that rehabilitation procedures were in good condition, in addition to observing a great improvement in oral hygiene.

Discussion

In this study, we observed several clinical findings, along with craniofacial and oral manifestations of the Opitz G/BBB syndrome. Although the cause is still unknown, the molecular genetic MiD1 testing can confirm the diagnosis.[2,23,27,28] The treatment of a patient with this syndrome requires a multidisciplinary service where physician, physiotherapist, ophthalmologist, and dentist, among others, each in their area of expertise, must act to ensure the patient a good quality of life.[5,6,9–13]

In handling patients with special needs, it is essential to carry out interconsultation with the patient's physician to obtain additional data in order to prevent problems associated with their underlying disease (mental disabilities, heart disease, etc.) that may arise during some risk procedures.

In this study, there were relationship between the main systemic manifestations and clinical features of the syndrome.[1,14,15,18–20] The literature also refers cardiac abnormalities in these patients,[5,13–15] therefore, as a protocol before performing dental treatments, it recommends cardiologic evaluation of patients with Opitz to verify the need for prescription antibiotic prophylaxis. In this report, the patient developed heart disease, which required prescription prophylactic antibiotics (amoxicillin 2.0 g, 1 hour before invasive dental procedures), as the protocol recommended by the AHA.

The presence of oral clefts is common in this syndrome[1,2,5,6] and has consequences in various aspects of functioning; however, in this case the patient had ogival palate and fibrosis in the palate center area (Figure 4), a finding that had not been reported in the literature, as well as the absence of tooth buds 18, 28, 38, and 48 (Figure 6) shown in the panoramic radiograph.

There was a challenge to obtain the information about clinical case from the patient's mother and her past dentist, primarily on the absence of the elements 17, 27, 35, and 45, as shown in Figure 6, which prevented the correct diagnosis of some demonstrations.

Regarding craniofacial amendments by individual who studied, they were met in accordance with the literature review.[9–12] The person responsible for the patient reports that in order to be able to correct the facial deformity of the patient an orthodontist's help was sought for braces installation, who nominated upper and lower braces. However, in absence of educational measures to oral care for both the patient and the individual responsible for her, especially for patients with special needs, may result serious harm to the patient's oral health. The presence of various caries, high bacterial biofilm index (100%) and presence of calculating (100%), and pH 4.5, demonstrated that the patient had alarmingly high risk of caries activity.

It must be remembered that this population of patients with special needs due to their physical and mental condition, have difficulty running their oral care in a satisfactory manner, requiring another person to perform it. Neglecting this fact by a dentist may lead to situations of clinical severity as happened in this case, where the orthodontic appliance is a contributing factor for the severity of the oral health of the patient.

Based on what was said above, it is fundamentally a change in attitude on the part of dental professionals with regard to the focus of attention. From the time that a patient is known as a whole, we must obtain better clinical results.

Conclusion

The great heterogeneity of craniofacial demonstrations and other pathologies associated with their underlying disease in patients with Opitz syndrome suggest a multidisciplinary approach to the monitoring and treatment of affected patients.

Fibrosis in the center of palate, enlargement of the pulp camera, and absence of third molars germs were not previously reported, as were observed in the patient referred to in this study.

Knowing the patient with special needs as a whole is extremely important to perform dental treatment; one should not neglect the contraindications of some treatments individually because ignorance can lead to clinical failure.

Competing interests

The authors have declared that they have no conflicts of interests.

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None.

Patient consent

The case report was approved by the Ethics Committee of Paulista University (643/09), and the patient signed the consent form.

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