Orocraniofacial Findings and Dental Management of a Pediatric Patient with Dubowitz Syndrome

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Dubowitz syndrome is a rare genetic condition characterized by microcephaly, dysmorphic facial features and delayed general growth. It is transmitted through autosomal recessive inheritance. The purpose of this report is to describe the oral, craniofacial and systemic characteristics of a 7-year 11-month-old boy with Dubowitz syndrome and the dental management provided. The pediatric dentist should possess the ability to recognize this rare alteration, to provide dental treatment and to refer for the necessary medical and multidisciplinary treatment.

Keywords: *Dubowitz syndrome, orocraniofacial findings, dental management.*

INTRODUCTION

Dubowitz syndrome is a rare genetic disorder of unknown etiology. It was first reported by Dubowitz in 1965, when informed about two English siblings who exhibited low birth weight, unusual facial appearance and skin eruptions.[1] Initially, the disease was confused with Bloom syndrome until it was recognized as a different condition.[2,3] Gross *et al*. defined the disorder after they studied three new cases and followed one of Dubowitz's original patients.[4]

The syndrome is an autosomal recessive condition with similar presentation in both sexes and all races; however, most reported cases have been Caucasians and Asians. The patients' parents are usually healthy; although the antecedent of consanguinity marriages should always be sought.[5] The responsible genetic mutation of Dubowitz syndrome has not yet been fully identified.[3]

Affected patients exhibit delayed prenatal and postnatal growth and microcephaly.[5-8] An important aspect is the presence of mental retardation, which it have been described as moderate or severe in 15% and 10%, respectively.[3,7,9,10] Behavior problems include hyperactivity, shyness, short attention span, occasional speech disabilities and high-pitched or hoarse voice.[6] Other characteristics are eczema, especially in the face, extremities and hands; foot deformities, syndactily of the toes; predisposition to cancer (leukemia, lymphoma and neuroblastoma); immune deficiencies; respiratory infections; and gastrointestinal symptoms such as feeding difficulties, frequent vomiting and chronic diarrhea.[2,3,11]

Facial anomalies include narrow face, micrognathia, high sloping forehead, flat supraorbital ridges and broad and depressed nose bridge continuous with the forehead; sparse eyebrows and frontal hair; large, prominent, low-set ears with or without dysplasia; slanting palpebral fissures, hypertelorism, blepharophimosis, ptosis, telecanthus, epicanthal folds and strabismus.[7,12] The main oral and dental alterations reported are high and narrow or submucosal cleft palate, bifid uvula, caries, macrodontia, oligodontia, delayed eruption in both dentitions, crowding and malalignment, midline diastema, conical incisors and fused or double teeth.[2,7,9]

The diagnosis of Dubowitz syndrome is based mainly on the craniofacial and cutaneous manifestations, and prenatal diagnosis is not reliable. Differential diagnosis includes Bloom syndrome, Smith-Lemli-Opitz syndrome, Seckel's syndrome, fetal alcohol syndrome, and others in which the eczema is a characteristic finding.[2,7] Prognosis of the disease is poor because of malnutrition and gastrointestinal disorders, recurrent infections of the skin and upper respiratory tract. In older children, the higher risk of cancer also compromises life expectancy. Nevertheless, the mean life span is considered normal, with concurrent growth and behavioral difficulties.[11]

The purpose of this report is to describe a case of a 7-year 11-month-old Mexican boy with Dubowitz syndrome—his systemic, orocraniofacial findings and the dental management provided.

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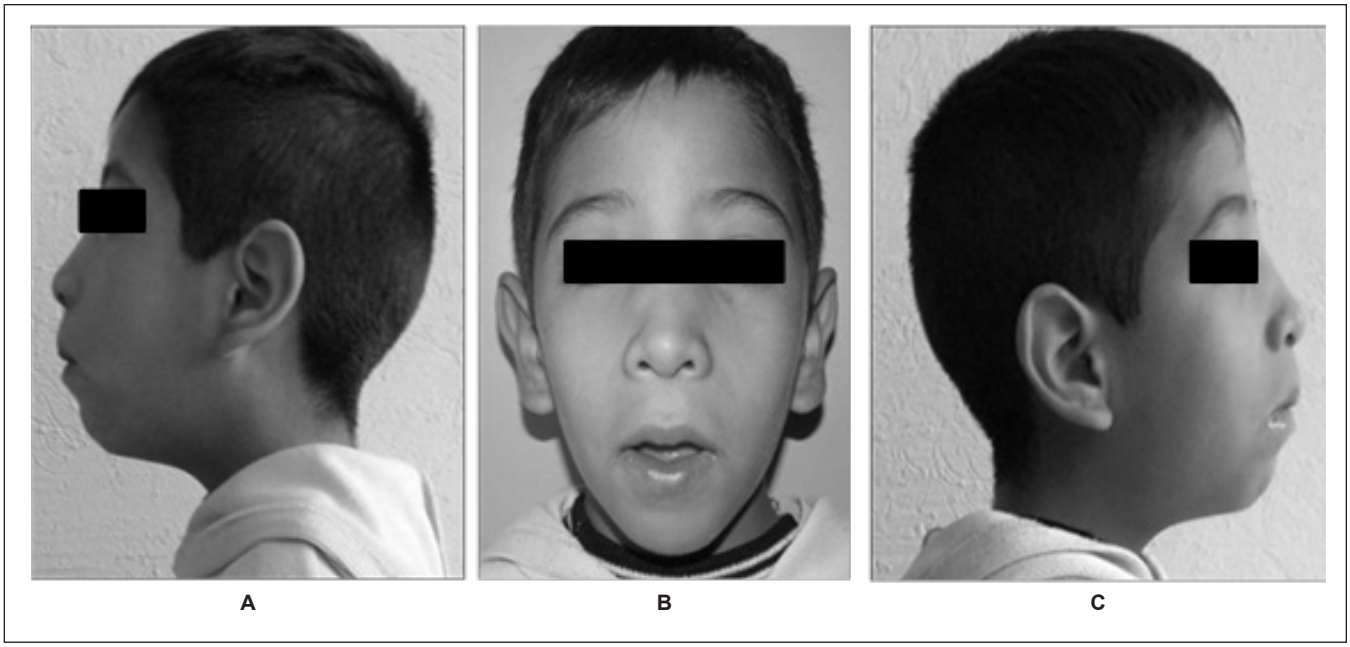

Figure 1. Facial views: A. Left side; B. Front, C. Right side.

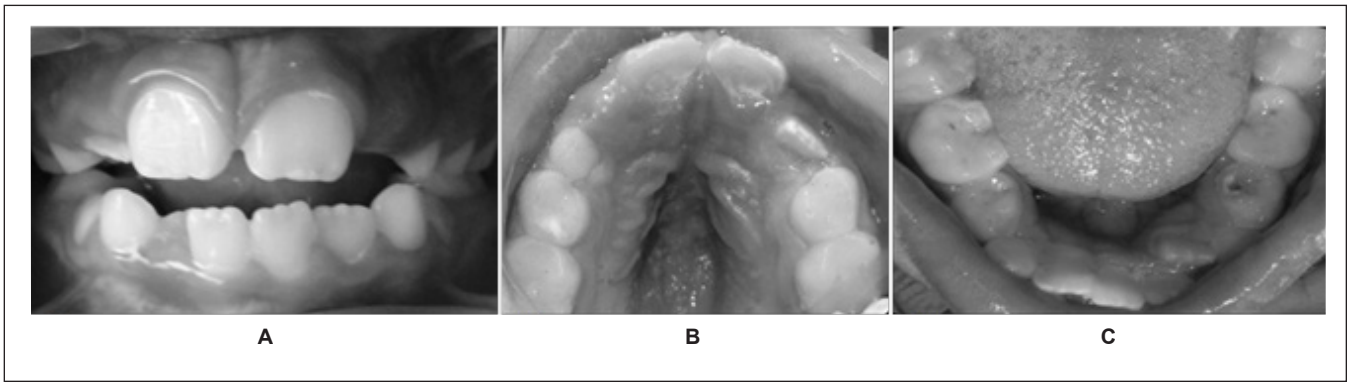

Figure 2. Intraoral views: A. Front; B. Upper arch C. Lower arch.

CASE REPORT

A 7 year 11-month-old Mexican boy was presented by his parents to the Clinic of Pediatric Dentistry at San Luis Potosi University, Mexico, referred for comprehensive dental treatment. The parents reported that he was born at full-term via normal delivery and that his birth weight was 2.7 kg. They also reported consanguinity by the father's grandparents and the existence of paternal close relatives with mental disabilities, dwarfism, gigantism and epilepsy. Although delayed intrauterine growth had been detected, the child was not diagnosed with Dubowitz syndrome by genetic analysis until 5 months of age because he previously had manifested delayed psychomotor growth; lack of head support, body bearing and neck control; swallowing difficulties; attention deficit; malnutrition; deep palate and lack of ear growth. Since age 1 1/2 years, the patient had received physical, psychological and speech therapy. At 5 years, the child exhibited problems of enuresis and involuntary defecation; during that time, he showed mild delayed psychomotor growth and little socialization. At the time of the initial exam, the boy was taking risperidone 2 mg every night because of hyperactivity, irritability and restlessness. While attending first grade, he was found to have low weight and height according to somatometric studies (below the 10 percentile); he also exhibited mild left scoliosis when standing. The parents had not reported the presence of anomalies related to the digestive, respiratory or cardiovascular systems and recent laboratory studies showed normal hematologic counts.

Extraoral examination (Figure 1 A–C) showed microcephaly, dolicocephaly (adenoid facies), triangular face, strongly convex profile, sparse hair, wide forehead with prominent glabella, hypertelorism, epicanthus, broad and depressed nasal bridge, severe mandibular micrognatia and retrognatia, lip incompetence and limited mouth opening.

Intraoral examination (Figure 2 A–C) revealed poor oral hygiene with generalized plaque, mouth breathing (noted by his otolaryngologist) with a triangular, narrow and deep palate; an anterior open bite of approximately 4 mm, produced by a pronounced tongue thrust, and severe mandibular anterior crowding. He was in the mixed dentition stage with absence of the primary maxillary left canine and subsequent space loss, partial eruption of both perma-

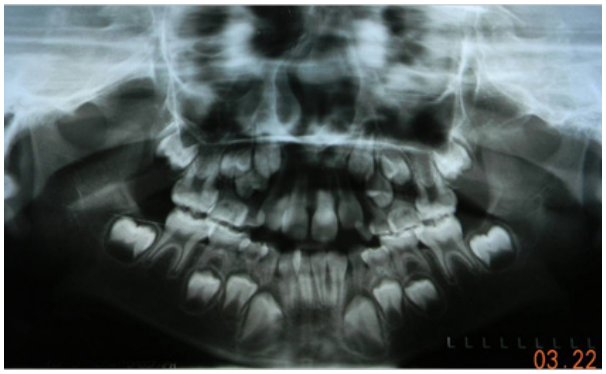

Figure 3. Panoramic radiograph.

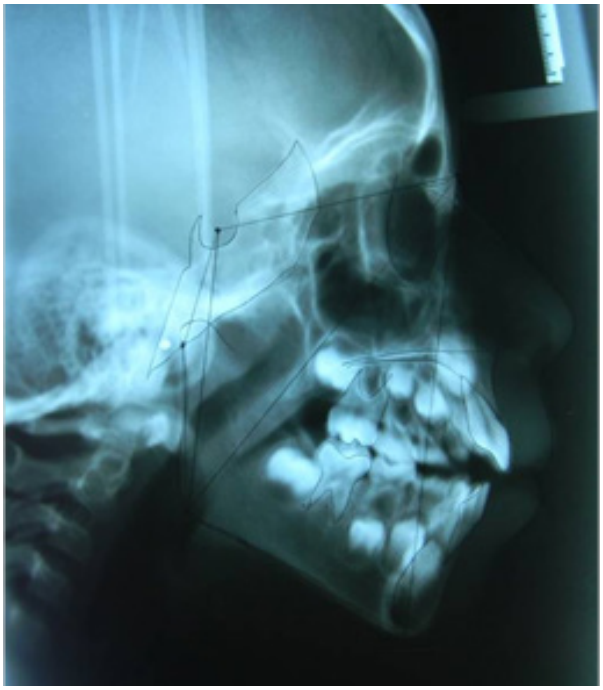

Figure 4. Lateral radiographic view.

nent maxillary lateral incisors, a bilateral class I molar relation and carious lesions in the primary mandibular second molars and first molar (in infraeruption) and maxillary left first molar. A panoramic radiographic view did not reveal any apparent dental or skeletal anomalies (Figure 3).

Based on the clinical and radiographic information, we decided to place pit-and-fissure sealants on the four permanent first molars and resin restorations on all decayed primary molars. All restorative procedures were fully agreed to by the child's parents through a signed informed consent form and carried out under local anesthetics (2% lidocaine with epinephrine 1:100,000, 4 mg/kg). Patient's behavior was rated as definitely negative from the first appointment, according to Frankl's classification. Modeling techniques and psychological methods of behavior management during most dental procedures were used. A Jarabak cephalometric analysis was carried out (Figure 4), which revealed a narrowing of the upper airway because of adenoid hypertrophy, vertical facial growth, posterior rotation of the mandible and maxillary protrusion. Comprehensive orthodontic-orthopedic treatment was instituted, beginning with intercepting the tongue habit by means of a fixed upper appliance and subsequent placement of functional orthopedic appliances to manage the skeletal class II, maxillary lack of space and mandibular anterior crowding.

Control appointments were planned every 3 months in order to observe the occlusal evolution and eruption of the remaining permanent teeth, as well as to reinforce the preventive recommendations of brushing, flossing and diet.

DISCUSSION

Dubowitz syndrome is a pathologic condition that is manifested in the craniofacial complex and the oral cavity. The incidence has not been clearly established.[7] Approximately 150 cases have been reported, mostly in the USA, eastern Europe, Russia and Japan,[2,3,10] although few of these cases have been reported in the dental literature.[3,5] Though these reports describe oral features, they are inconsistent, which might be explained by its rare incidence. In our knowledge, this case is the third report about the oro craniofacial findings in a pediatric patient.

Pediatric dentists should be aware of the existence of this disease and that it can be diagnosed and identified by several features that may be found in the oral cavity, face and head of the affected children. Some of these features were exhibited by the patient reported in the present case such as microcephaly; broad, sloping and prominent forehead; broad nose bridge; hypertelorism; epicanthus; sparse hair; short neck; mandibular micrognathia and retrognathia; high and narrow palate and evident occlusal anomalies.

It is also important, when designing the dental treatment plan in children with Dubowitz syndrome, to take into account that, because of the delay in eruption of the permanent teeth, the primary teeth tend to remain for more prolonged periods. Appropriate preventive and restorative measures should be implemented to preserve these teeth, also keeping in mind that the aesthetic features can help improve the patient's self-esteem. Likewise, management of the malocclusion should include not only orthodontic therapy, interception of inappropriate oral habits and orthopedic appliances to stimulate mandibular and maxillary growth, but also the possibility of future orthognathic surgery, especially in cases of severe skeletal class II malocclusion, as in the present case. Clinicians should consider that, in children and adolescents with Dubowitz syndrome, bone growth is generally delayed relative to the chronologic age.[5]

Other aspects that should be carefully considered during treatment planning is the possibility of employing behavioral management techniques, both psychological and pharmacological, in order to obtain reasonable cooperation from the affected patient during dental treatment, especially in cases of mental retardation, hyperactivity and attention deficit. In the present case, we successfully used some of these methods, together with extensive verbal communication throughout the restorative period; however, the limited mouth opening exhibited by the child was an important factor in extending the duration of each appointment.

CONCLUSION

Pediatric dentists should possess the ability to provide high quality dental treatment, as well as refer and guide the parents toward other medical, dental and psychological specialists, not only for precise clinical and genetic diagnoses, but also to insure that the patient receives multidisciplinary management of the disease, thus improving the expectations and quality life of their children affected by this rare syndrome.

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