Dubowitz Syndrome: Report of a Case With Emphasis on the Oral Features

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ABSTRACT

Dubowitz syndrome is a rare condition in which the affected individual presents with dysmorphic facial features and manifests growth retardation. Although the condition is well reported in the medical literature, the dental manifestations have not been discussed in great detail. Some of the dental features reported include macrodontia, hypodontia, delayed eruption, and midline diastema. The purpose of this case report was to describe a young Chinese patient with Dubowitz syndrome with specific oral features. (J Dent Child 2005;72:100-103)

KEYWORDS: DUBOWITZ SYNDROME, DELAYED ERUPTION, MALALIGNMENT OF TEETH

Dubowitz syndrome, which has an autosomal recessive mode of inheritance, manifests as: (1) intrauterine and postnatal growth retardation; (2) microcephaly; and (3) a characteristic facial appearance. The condition is rarely reported in the literature, and very few have even mentioned the syndrome's oral characteristics.

In 1965, Dubowitz described 2 English siblings, said to have Bloom syndrome,[1] who exhibited "familial low birth weight dwarfism with unusual facies and a skin eruption." Later, Opitz [2] recognized that these cases had features inconsistent with Bloom syndrome and chose to call it "Dubowitz syndrome."[3]

This syndrome is now regarded as being a rare autosomal recessive disorder.[4,5] Most reported cases have been Caucasians, and the sex ratio is approximately 1:1.[4-7] The parents of the affected individuals are usually healthy, and the pedigrees are free of nonconsanguineous marriages.[6,7]

Intrauterine and postnatal growth retardation have been obligate symptoms in most reported cases,[5] while delayed postnatal weight gain and dwarfism are routinely present.[5] A delayed increase in head circumference[5] and, hence, microcephaly are consistent findings.[5,8] Most cases, however, appear to be mentally normal or, at worst, only mildly retarded.[4] They are often shy and hyperactive and exhibit short attention spans.[4,5] Speech disorders also have been reported in some cases.[5]

The dysmorphic facial features include a narrow face, micrognathia, a sloping forehead, and flat, supraorbital ridges with the bridge of the broad nose being continuous with the forehead.[4,5] Low-set ears with or without dysplasia[7,9] and posteriorly angulated ears have been reported.[10]

The eyes and periorbital area demonstrate telecanthus, hypertelorism, ptosis, and/or blepharophimosis. Slanting palpebral fissures have occurred in more than half of the reported cases,[5] while epicanthal folds and strabismus can also occur.[4,5]

The voice may be high-pitched and hoarse.[4,5] Other signs of incomplete morphogenesis[5] include foot deformity, such as syndactyly of the toes, and retarded bone age.[5]

Hematological and malignant disorders have been reported.[11,12] Also, susceptibility to recurrent infections of the respiratory and the GI tracts are frequently reported symptoms, which suggests the possibility of an immunodeficiency.[6,7] Although ventricular septal defects have been noted in several cases,[6] cardiac defects and other internal malformations are considered to be atypical of this syndrome.[7]

Many cases exhibit hypoplasia of the ectodermal tissues,[5] symptoms of which include sparse, thin hair, hypoplastic eyebrows,[5] and eczema. Wilhelm and Mehes[5] and Opitz,[2] however, believe that this is a coincidental feature of the disorder.

When a cleft palate has been present,[4] it has often been in the form of a submucosal cleft.[6] The only other reported oral manifestations have been delayed eruption,[5] dental crowding and malalignment, a midline diastema, and rotated mandibular incisors.[6] In addition, the teeth have been said to be conical shaped or macrodont, or there may be congenitally missing maxillary central incisors and double teeth. As the oral and dental manifestations have not always been mentioned, the occurrence rate of these dental anomalies may have been underestimated or occurred as coincidental findings.

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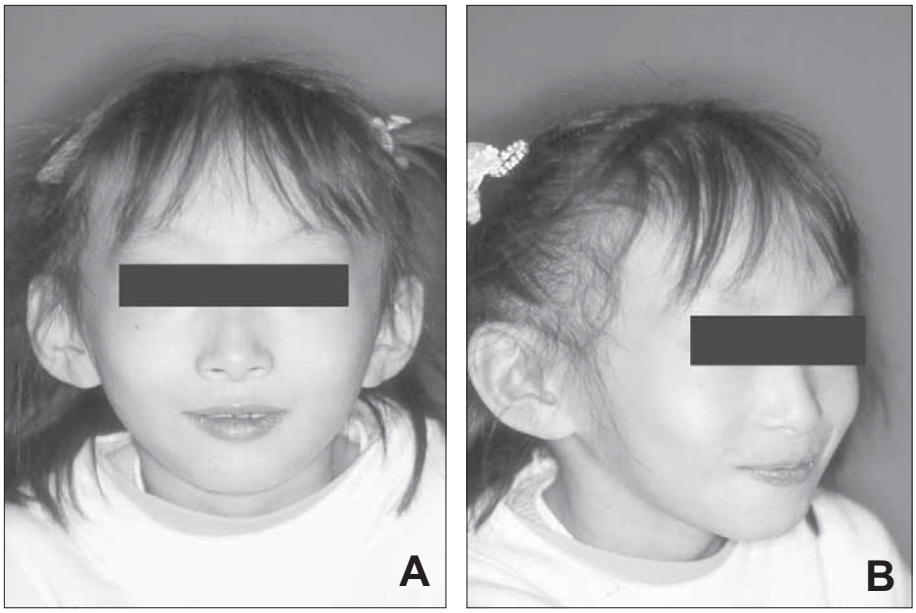

Figure 1. The face is small and triangular in shape, and there is micrognathism and retrognathism. Epicanthal folds, telecanthic hypertelorism, and blepharophimosis are also exhibited.

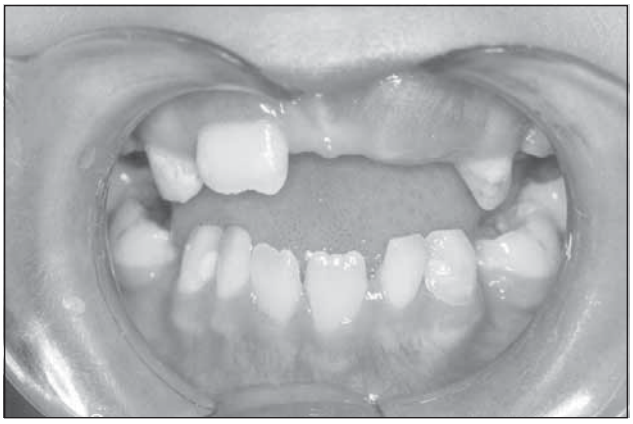

Figure 2. At the age of 8 years, 5 months, there was delayed eruption of tooth 9 and distal eruption of tooth 8, while teeth N and P were retained.

The purpose of this case report was to describe a girl with general features consistent with Dubowitz syndrome, including many of the rarely reported oral anomalies.

CASE REPORT

The patient was not diagnosed as having Dubowitz syndrome until she was 6 years old. Following genetic analysis, she was diagnosed by the attending. The girl was born at full-term via normal delivery, and her birth weight was 2.6 kg. Information about the duration of her delivery was unavailable. No major defects had been identified in her internal organs. She had allergies to soap and some spices, which required the use of a special soap prescribed by her dermatologist. In addition, she was mildly developmentally delayed.

At 5 years of age, she was found to be suffering from Early Childhood Caries, which necessitated the administration of general anesthesia for the extraction of numerous carious primary teeth. Subsequently, there was no dental review until she was 6 years old, when she received a dental checkup followed by preventive and restorative care from the School Dental Care Scheme. She was not managed, however, by a multidisciplinary team of dental specialists. At age 8, she was referred by an orthodontist to the authors for treatment of hypodontia in the permanent dentition.

Neither her 16-year-old elder sister nor her immediate family demonstrated any similar anomalies. She was attending the second grade, albeit in a special class within a school for normal children. She was a natural, outgoing girl who was extremely inquisitive. She made conversation with everyone she met and was friendly and affectionate. She was not hyperactive, although her mother reported that she did have a short attention span when doing her homework. This improved, however, with time and maturity. Her mother, who was extremely supportive and reassuring, demonstrated good interpersonal dynamics with her daughter.

The patient was found to be short at 107 cm (below the third percentile) and weighed only 14 kg (below the third percentile) at the age of 8 years, 3 months. Nevertheless, her body was normally proportioned. She presented with a small triangular face proportional to her small head (Figure 1a and b). Her mandible was micrognathic and slightly retrognathic. The nasal bridge was continuous with her forehead when viewed in profile, and she demonstrated telecanthic hypertelorism and blepharophimosis. Epicanthal folds were present that clearly covered the medial aspects of both eyes, which were relatively large compared to her small face and exhibited ptosis. The outer aspect of her eyebrows was scanty.

INTRAORAL FEATURES

At the time of presentation, the patient was in the early mixed dentition. Unfortunately, her mother was unsure whether she had any congenitally missing primary teeth. The teeth present, in her mouth, were 3, A, C, 8, H, J, 14, 19, K, M, N, 24, 25, Q, R, T, and 30. Her permanent teeth appeared to be large in size compared to her small face. The amalgam restorations on the occlusal surfaces of teeth K and T were defective, and there was recurrent caries. In addition, space loss had occurred following the extraction of the maxillary first primary molars. All of her permanent teeth were sound. There was no obvious enamel hypoplasia, and the color and quality of the enamel was within normal limits. Both arches, which were parabolic in shape, were flared posteriorly. Her palate was relatively flat, while the labial and buccal sulci were shallow. Her saliva flow was not confirmed to be normal, since no salivary function tests were performed. Her mouth opening was severely re-

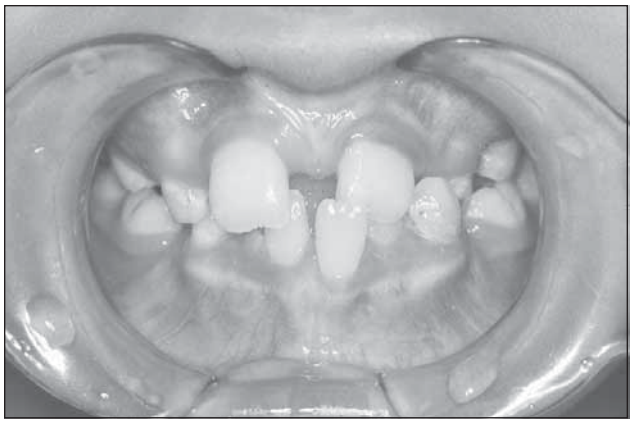

Figure 3. By age 9, tooth 9 had erupted in crossbite with 24, which displaced 24 buccally. Hence, tooth 9 was trapped between the labially displaced 24 and lingually erupting 23. Tooth 9 erupted distally into the space of G, leaving a 5–mm space between teeth 9 and 8. Teeth 23 and 26 erupted lingually to teeth 24 and 25.

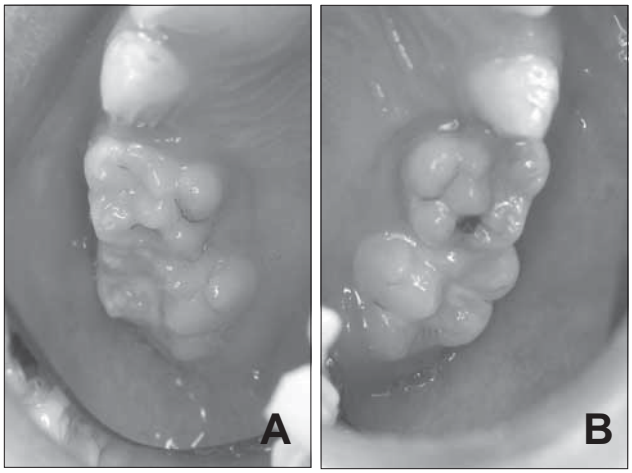

Figure 4. Accessory cusps on the occlusal surfaces of teeth A, J, 3, and 14.

stricted, measuring 30 to 35 mm (40 to 45 mm is normal for an 8-year-old girl).

By the age of 8 years, 5 months, there was delayed eruption of tooth 9, and tooth 8 had erupted distally into the extraction site of D, while teeth N and Q were retained (Figure 2).

When re-examined 7 months later, tooth 9 had erupted. There was a large midline diastema between teeth 8 and 9. Tooth 9 was in crossbite with tooth 24, which was displaced labially due to the traumatic occlusion. Teeth 23 and 26 erupted lingually (Figure 3). All maxillary first permanent and second primary molars had accessory cusps present in the center of the occlusal surfaces (Figures 4a and b). The mesial marginal ridges were also extremely prominent. Table 1 summarizes the dental findings in the primary and permanent dentitions.

Table 1. Summary of Oral and Dental Features Present in the Reported Case

Primary dentition	Permanent dentition
Delayed exfoliation	Delayed eruption
	Hypodontia
	Transpositions
	Anterior crossbite
Accessory cusps on molars	Crowding
Taurodontism	Midline diastema
	Accessory cusps on molars
	Taurodontism of molars
	Shallow buccal and labial sulci
Shallow buccal and labial sulci	Micronagthism and retrognathism

RADIOGRAPHIC FINDINGS

Radiographically, the patient exhibited several interesting dental features. Apart from the delayed eruption of her maxillary and mandibular anterior teeth, teeth 7 and 10 were congentially missing (Figure 5). The presence of crowding was confirmed in both arches. Space loss had occurred in the maxillary premolar regions, and teeth 8 and 9 had erupted distally into the spaces that 7 and 10 should have occupied. Taurodontism was observed in all the primary and permanent molars (Figure 5).

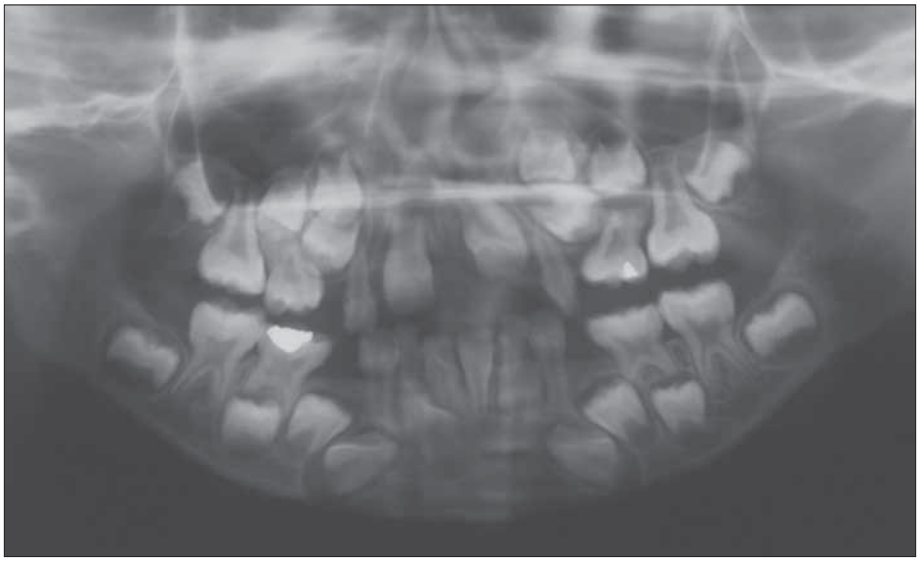

Figure 5. Panoramic radiograph at 8 years, 5 months old showing congenitally missing teeth 7 and 10, and possible transposition of teeth 11 and 12. Also, taurodontism of all the primary and permanent molars is evident.

DISCUSSION

The lack of reports in the dental literature on oral features in Dubowitz syndrome patients can be explained

by the fact that most of the published reports were made by medical practitioners. Those oral features that were reported in the literature are not consistent. The authors believe that the dental features noted in the presented case might not be present in all Dubowitz patients. The accurate description and reporting of oral features identified in each individual with Dubowitz is essential to advance our knowledge of the syndrome.

Furthermore, dental findings can be useful as a diagnostic tool to identify possible individuals with Dubowitz syndrome. Although this condition is relatively rare, it is conceivable that pediatric dentists will encounter children with this syndrome. Hence, it is important that the dental profession be made aware of this syndrome's characteristics and have the knowledge to plan and carry out short- and long-term treatment regimens for these children. Dentists should also be prepared to refer these patients to relevant centers where multidisciplinary services are available for their management.

The prolonged retention of the primary teeth requires a much higher standard of preventive care. To some extent, however, this is dependent on the patient's compliance.

The provision of good dental esthetics and self-esteem are of a higher priority in these children who already look different from their peers. It may be necessary, even as interim measures, to provide early closure of the diastema, redistribute the spaces, and create composite build-ups of the primary teeth (to mimic the missing permanent teeth) or insert resin-bonded bridges to replace missing teeth.

The presence of the accessory cusps on the molars could be a potential problem, as occlusal interferences may occur that require occlusal equilibration. The presence of a restricted opening of the mouth is consistent with TMJ malformations that may cause discomfort, so early intervention might be required when there are early signs of discomfort.

As was evident in this case, postnatal growth can be severely retarded in children with Dubowitz syndrome. This can affect the timing of the various corrective procedures such as orthodontic therapy and orthognathic surgery. The retarded bone age is also a factor that should be considered if orthognathic surgery is contemplated.

The treatment plan for this reported patient commenced with education and reinforcement of good oral hygiene practices. This was followed by thorough prophylaxis and the application of topical fluoride varnish. The recurrent caries and defective amalgam restorations were removed, and the teeth were restored. A simple fixed orthodontic appliance, in conjunction with a bite block, was used to correct the localized crossbite between teeth 9 and 24. Further definitive orthodontic treatment will be required in the future. This will be best managed by an orthodontist in the next few years once more permanent teeth have erupted.

A number of difficulties were encountered when trying to provide the necessary oral care. The patient's extremely small mouth and limited opening, coupled with the flared dental arches, made it difficult to take impressions and to fit orthodontic bands. The construction of custom impression trays, combined with the application of petroleum jelly to the lips and the commisures of the mouth, made such procedures slightly easier.

This case study identifies some of the rarely reported oral features of Dubowitz syndrome to identify those findings which may be consistent with the syndrome rather than those that are coincidental.

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