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Case Report

Dental management of oculodentodigital dysplasia: a case report

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Abstract: Oculodentodigital dysplasia is an extremely rare autosomal dominant pleiotropic disorder. The syndrome is characterized by abnormal facial features, central nervous system involvement, syndactyly and clinodactyly of fourth and fifth fingers, dry and lusterless hair, generalized enamel hypoplasia and odontodysplasia. Combination of odontodysplasia, poor oral hygiene, and parental neglect can lead to extensive destruction of tooth structure and the treatment options become limited. Early diagnosis with a proper treatment plan and meticulous oral hygiene program helps eliminate the necessity of multiple tooth extractions. This case report describes the comprehensive dental treatment aimed at rehabilitation of function and aesthetics of the dentition in an 8-year-old boy with oculodentodigital dysplasia. (J Oral Sci 52, 337-342, 2010)

Keywords: dental enamel hypoplasia; mouth rehabilitation; odontodysplasia.

Introduction

Oculodentodigital dysplasia (ODDD) is a highly penetrant autosomal dominant disorder, with variable expression (1). ODDD is caused by mutations in the *GJA1* gene located on human chromosomes 6q22-q23, encoding the gap junction protein Connexin 43 (Cx43) (2,3). Oral and dental manifestations of ODDD consist of oval palate (4), coronoid hypoplasia (5), mandibular retrognathism (6), enamel hypoplasia of the deciduous as well as permanent dentition and delayed tooth eruption (6). Histological examination shows enamel dysplasia, dentin hypocalcification, pulp denticles and hypercementosis (5).

Dental manifestations of the syndrome including generalized enamel hypoplasia, thin dentinal walls, extensive caries leading to early pulp involvement, and often open apices present challenges in providing dental treatment for these patients. A conservative approach should be taken to maintain the integrity and aesthetics of the patient's permanent dentition. The conservative treatment options require knowledge of the best choices for the presenting clinical situation, i.e., functional requirements and the inherent aesthetic properties of each material. The reported case describes the comprehensive dental treatment of an 8-year-old white boy diagnosed with oculodentodigital dysplasia (7).

Case Report

An 8-year-old boy was referred to the Department of Paediatric Dentistry, Tabriz University of Medical Sciences for treatment of a dental abscess related to the left lower first premolar.

Dental findings included yellowish color of the teeth with extensive carious lesions, hypoplastic appearance of the atypically-shaped teeth which were smaller than normal, and premature eruption of permanent canines and premolars. The parents reported that the lower right and left central incisors and lower left lateral incisor were missing and there was no history of extraction (Fig. 1). Panoramic radiograph revealed shortened teeth with 'ghost' appearance, unclear lamina dura, decreased thickness of

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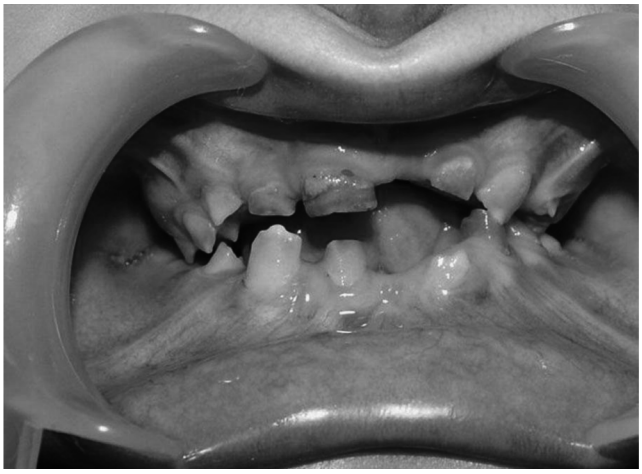

Fig. 1 Dysplastic teeth and multiple caries seen before treatment.

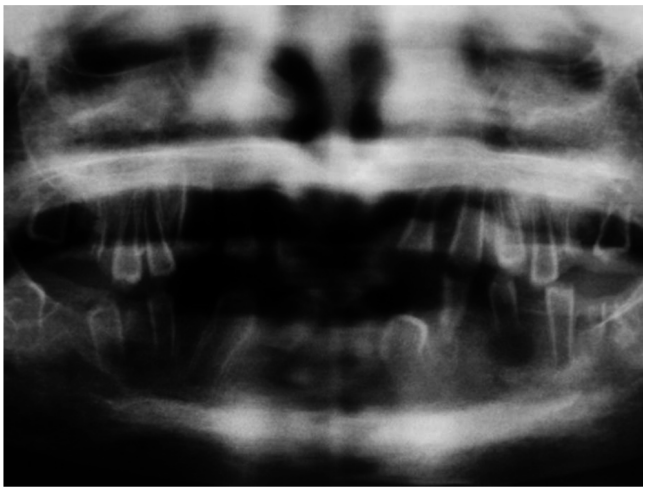

Fig. 2 Panoramic radiograph of the patient showing shortened teeth with 'ghost' appearance, generalized enlarged pulp chambers, and decreased thickness of dentin. Note the periapical radiolucency of lower left first premolar.

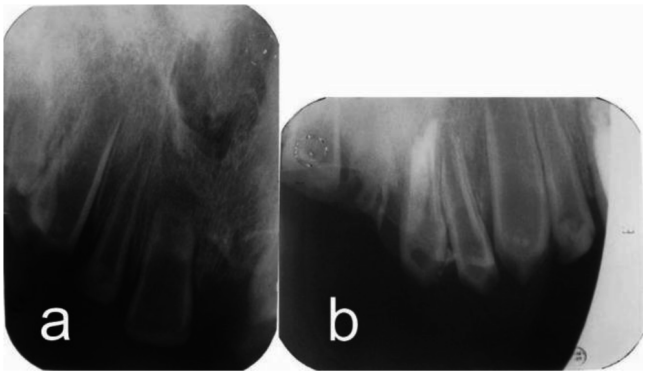

Fig. 3 (a) Short length and diffused calcification of right upper central incisor. Pulp calcification is seen in lateral incisor; (b) MTA plug is seen in the apex of right upper second premolar. Also note the calcification of coronal pulp in canine and first premolar.

dentin, and lack of contrast between enamel and dentine (Fig. 2). The existing permanent teeth were found to have taurodontism. Periapical radiographs of the teeth revealed irregular calcification and pulp denticles in most of the teeth evaluated (Figs. 3a and b). The permanent upper canines reached full eruption during the course of the treatment which lasted for two months, indicating an accelerated rate of eruption. During the same period, the second permanent molars also erupted.

The patient's five year old brother had frontal bossa, clinodactyly, hyperkeratosis of hands, mild enamel hypoplasia of primary teeth, and a bi-rooted right upper primary canine. The patient's parents who were second cousins also had clinodactyly. Considering all the signs observed in the family, oculodentodigital dysplasia was suspected, and therefore, the patient was referred to a pediatric hospital for further evaluation.

The patient's ophthalmologic examination revealed microphthalmia, microcornea, congenital cataract, chronic uveitis, glaucoma and reduced vision. Conventional radiography of both hands revealed agenesis of second phalanges of the fifth fingers and osteopenia of the wrists. No congenital heart disease was reported after examination by the specialist.

Autosomal dominant trait of the syndrome was suggested by genetic evaluation. To confirm the clinical diagnosis, a genome-wide search for the location of ODDD locus was performed using Short Tandem Repeat Polymorphisms (STRPs). Evidence of linkage between ODDD and markers from chromosome 6q22-q23 was detected. Missense mutation in *GJA1* (Connexin 43) exon 2 was found.

Before visiting the Department of Pediatric Dentistry, left upper and lower first permanent molars, left upper central incisor, and right lower first permanent molar had been extracted. On the first appointment, under local anesthesia (2% lidocaine with 1:100,000 epinephrine; Dentsply Pharmaceutical, York, PA, USA), pulp tissue of the abscessed left lower first premolar was extirpated using a Hedström-file and irrigated with 2.5% sodium hypochlorite. A calcium hydroxide dressing (Dentsply Herpo, Petrópolis, RJ, Brazil) was then given for 14 days and the tooth was restored with light-cured glass-ionomer (GC Fuji II LC, Tokyo, Japan) temporarily.

After resolution of the acute symptoms, in the next appointment, diagnostic dental casts of both arches were prepared for precise evaluation. The overall treatment objective, procedures involved, limitations, and alternative treatments were explained to the parents, and they signed an informed consent form.

At a subsequent appointment, after administration of local anesthesia to the left upper and lower quadrants, caries were

removed using a No. 1 round bur on a slow-speed handpiece and an excavator. Pulp exposure was seen in the left upper second premolar, which revealed a necrotic pulp. Considering the fragility of the teeth, a rubber dam could not be placed and, thus, isolation was accomplished using cotton rolls. Due to thin dentinal walls of teeth, minimal filing was performed, and after irrigation with 2.5% sodium hypochlorite solution, root canals were dressed with calcium hydroxide paste for 14 days and restored with light-cured glass-ionomer temporarily.

In teeth without pulpal involvement following caries removal, the pulp was protected with a liner (Dycal Ivory; Dentsply De Trey, Konstanz, Germany), and the prepared cavities were filled with light-cured glass-ionomer until a composite full-veneer was placed later.

In the following appointment, the same treatment procedures were performed in the right upper and lower quadrants. Initial filing of the root canal revealed dispersed calcification and non-vital pulp of the upper right central incisor. After irrigation with sodium hypochlorite solution, the root canal was filled with calcium hydroxide paste and the tooth was restored with light-cured glass-ionomer temporarily. Final endodontic therapy was performed on the teeth after two weeks.

Continuing with the right side in the next appointment, the upper lateral incisor and second premolar were exposed during caries removal, revealing a hyperemic pulp which indicated irreversible pulpitis. Due to incomplete root formation, apexification with Mineral Trioxide Aggregate (MTA; Dentsply, Tulsa Dental, OK, USA) was considered for these teeth. Minimal filing to the working length (14 mm in the lateral incisor and 13 mm in the second premolar) was performed followed by irrigation. The apical 3-mm of root canals was filled with MTA and a periapical radiograph was taken (Fig. 3b). Then, a moist cotton pellet was placed in the canals and teeth were restored temporarily with light-cured glass-ionomer. In a subsequent appointment, teeth were reinforced with fibre post (D.T. Composipost RTD, France) and restored with light-cured composite. Later, the second premolar was covered with an SSC (Unitek Stainless Steel Crowns, 3M ESPE, St. Paul, MN, USA).

Upper right central incisor had a short working length (10 mm) and malformed root canal, which made the use of fibre-post impossible (Fig. 3a). Therefore, the total root canal length was filled with MTA, and the coronal part of the teeth was filled with light-cured composite. The tooth was subsequently restored with the direct full-veneer composite technique.

Lower left first premolar also had a short working length (8 mm). Total canal length was filled with MTA, and subsequently restored with light-cured composite until full-coverage with SSC.

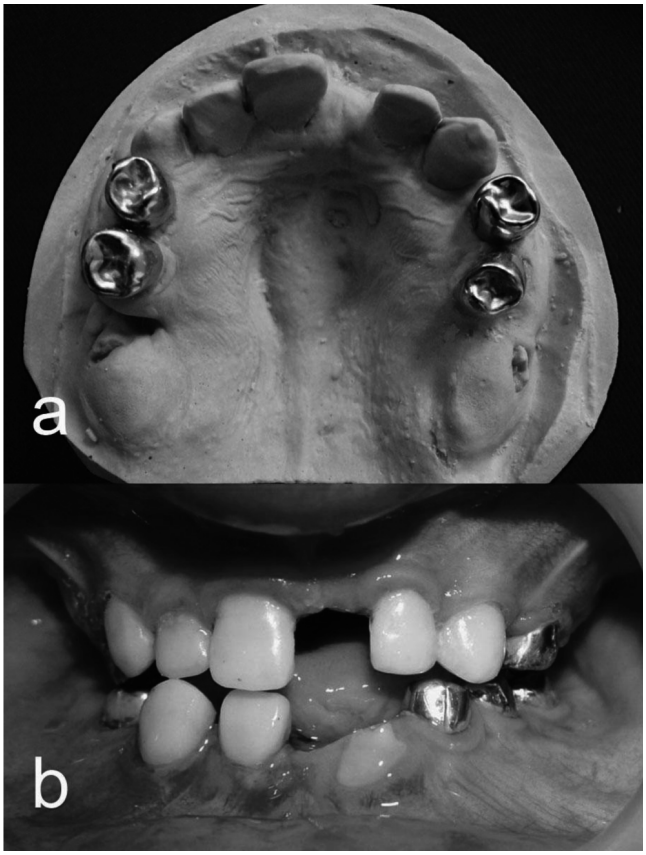

Fig. 4 (a) Adapted SSCs on the cast; (b) Composite full-veneers of anterior teeth. Some SSCs are also seen.

For the upper left second premolar (working length, 13 mm), the same procedures that were performed for the upper right second premolar (MTA plug and fibre-post placement) were followed.

Due to the disproportionate small size and malformation of premolars and unfeasibility of chair-side SSC adaptation, SSCs were first adapted on the dental cast before final adaptation in the mouth. Preparation of premolars was limited to reduction of cusp tips and the axial walls were left intact. After tooth preparation, impressions of both arches were taken. In order to adjust the SSCs, they were cut bucco-lingually and/or mesio-distally and soldered, and then, polished using mullets (Fig. 4a). All SSCs were fitted and cemented in two appointments.

In addition, direct composite full-veneers were placed on anterior teeth after minimal reduction of the enamel (Fig. 4b). In order to replace the missing upper left central incisor, a fibre-reinforced composite (FRC) bridge was considered. A composite pontic (Heliomolar, Ivoclar Vivadent, Liechtenstein) was directly made on the FRC

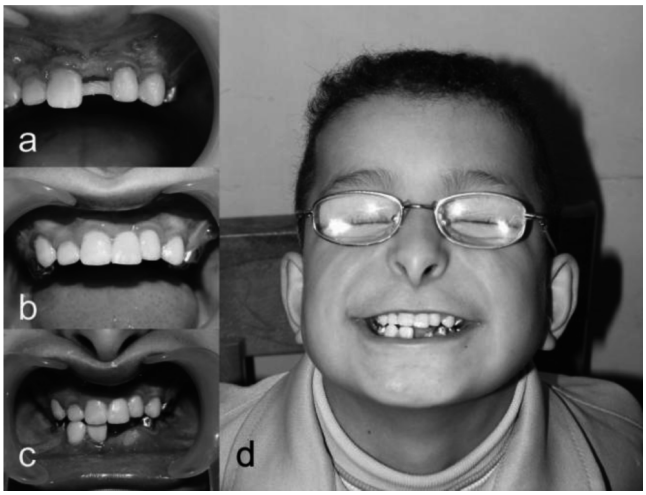

Fig. 5 (a) Fibre glass in place; (b) Replaced missing left upper central incisor using FRC bridge; (c) Post-treatment intra-oral view; (d) Patient's frontal view after treatment.

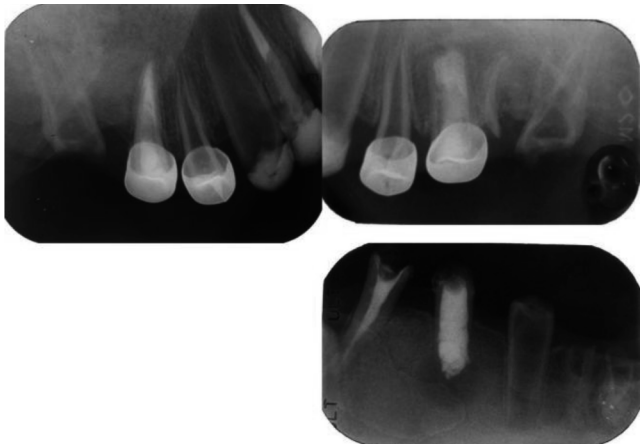

Fig. 6 Post-treatment radiographs of treated teeth.

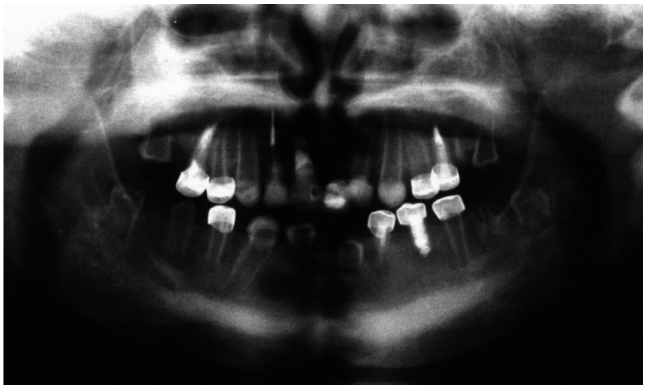

Fig. 7 Panoramic radiograph at 7-month follow-up. The periapical radiolucency of left lower first premolar has markedly decreased.

base (Figs. 5a, b).

Treatment of lower right second premolar and lower left canine was postponed due to incomplete eruption. Because there was no evidence of root resorption in the left lower primary canine, following pulp therapy, an SSC was placed to protect the tooth, similar to the premolars. The remaining roots of upper right first permanent molar were removed surgically.

The patient's post-treatment view is shown in Figs. 5c, d. Post-treatment radiographs are shown in Fig. 6. A 3-month follow-up was recommended to check restorations and give preventive oral hygiene instructions as well as for application of fluoride varnish and monitoring the eruption of unerupted teeth.

On the follow-up appointment after 7 months, the condition of restorations and oral hygiene were satisfactory. In the panoramic radiograph, periapical radiolucency of the lower left first premolar had markedly disappeared (Fig. 7).

Discussion

To the best of our knowledge, there is no documented report of comprehensive dental management of ODDD to date, other than extractions (8). For optimal treatment planning for these patients, dentists should consider factors such as type and severity of involvement, the presence of infection, the patient's age and expectations of the child and parents.

In the present case, the primary aims of dental treatment were to preserve arch integrity as well as to improve the patient's nutritional, aesthetic and psychological status. Considering the young age of the patient, alveolar bone preservation is considered of great importance. This is emphasized when the early eruption of permanent teeth is taken into account; except for the left lower primary canine, all the present teeth of the patient were permanent at this age. The teeth were structurally defective, and thus, particularly susceptible to caries and potential fractures resulting from trauma. Tooth extractions that would pose as an immediate treatment modality under normal circumstances in such a case, will result in compromised development of the alveolar process, and considering the socio-psychological complications of such an aggressive approach, it seems reasonable to consider a conservative treatment plan in management of the present case.

The present patient suffered from recurrent dental abscesses, which could be a challenge to maintain good oral health and favorable masticatory function. Clinically, multiple caries were present and enamel and dentin were extensively involved. Radiographically, this condition resembled odontodysplasia, as described in previous reports

(9,10). Radiographic similarities between the teeth in this case and odontodysplasia could justify the comparison of treatments; however, there is no consensus on the treatment of odontodysplasia. The management of regional odontodysplasia is somewhat controversial and revolves around the question of whether to remove the affected teeth. Although many clinicians prefer to extract the anomalous teeth as soon a diagnosis of regional odontodysplasia is made, some prefer to retain them until skeletal growth is complete as long as they are free of infection (10). However, due to involvement of all teeth in our patient, multiple extractions would be considered an aggressive approach. Therefore, attempts were made to retain the teeth as much as possible. The agreed-upon treatment plan after consultation with other dental specialists included endodontic treatment of pulpally involved teeth, stainless steel crowns for posterior teeth and direct full composite veneers for the anterior teeth, and a fibre-reinforced composite bridge to replace the upper central incisor.

Many studies have reported enamel hypoplasia as a consistent feature of dental involvement in these patients (4,6,8). A previous report describing the dental management of ODDD has documented gross dental caries secondary to severe enamel hypoplasia in a 30-month-old child with hypoplastic enamel and multiple caries (8). In this study, the treatment consisted of SSCs of posterior teeth and canines, and extraction of pulpally involved primary anterior teeth (8). In our case, enamel hypoplasia, enlarged pulp cavities, thin dentinal walls and multiple pulp exposures with open apices were encountered in all permanent teeth, which complicated the treatment to a greater extent.

In the present case, SSCs were used in the case of posterior teeth as a protective measure against their susceptibility to caries and fracture. In the anterior teeth, to protect the defective enamel and dentine and to provide aesthetics, complete crown coverage using direct composite restorations was selected. In teeth with pulpal involvement, the treatment of choice to achieve our objectives was pulp therapy with MTA as an apical seal and subsequent final obturation. Final treatment of the posterior teeth comprises the more challenging part of the treatment. Due to thin dentinal walls, preparation of teeth for cast restoration was not feasible. Full coverage with composite restoration is also prone to wear and, hence, would not be a suitable final restoration.

Despite the fact that the performed treatment met the expectations of the patient and the parents as it improved the child's masticatory function and aesthetics, our patient's vision and walking, unfortunately, deteriorated during the observed period, consistent with evidence in the literature that indicated worsening of neurological signs and symptoms of the syndrome towards the second decade of life (11,12).

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