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# Recommendations for dental management of diastrophic dysplasia: a rare case report

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**Abstract. – BACKGROUND: When treated in a dental chair, patients with deformities such as scoliosis often require special consideration.**

**CASE REPORT: Here is a case of 9 years old Saudi child who was reported with dental problems. The objective of this study is to provide a guideline for dental management of diastrophic dysplasia.**

**CONCLUSIONS: Diastrophic dysplasia is a rare and nonlethal skeletal dysplasia, autosomal inheritance recessive, recognized by the infant's dysmorphic changes at birth. Diastrophic dysplasia is not a common hereditary disorder, however, the characteristic of this condition and guidelines for dental treatment should be known to a pediatric dentist, particularly at a major medical center.**

*Key Words:*
Diastrophic dysplasia, Hitchhiker appearance of thumb, Cauliflower ear, Scoliosis.

## Introduction

When a team of professionals work together to overcome health problems, a special child can benefit of great health care. Medical professionals, including dentists, must be familiar with specific diseases and syndromes that may be covered by the term "special". This knowledge is imperative in managing special patients effectively.

Diastrophic dysplasia (DD) is a sort of inherent osteochondral dysplasia with an autosomal latent legacy design, originally depicted by Lamy and Maroteux[1] in 1960. It is caused by homozygous or compound heterozygous changes within the diastrophic dysplasia sulphate transporter (DTDST) quality on chromosome 5q32[2-4]. It has been found in most white populations but is significantly more prevalent among Finns[5].

The term diastrophic dysplasia is inferred from the Greek word "diastrophs" which implies truly bent or bowed[2,6]. It is distinguished by micromelia dwarfism, progressive scoliosis, respective talipes equinovarus, numerous digital deformities (short fingers, synostosis of proximal interphalangeal joints, unusual thumb embedding which is called a "hitchhiker", situated as within act of catching a ride, due to deformation of the first metacarpus), short stature (the reason it also knows as diastrophic dwarfism), characteristic ear distortions (cauliflower ears) and sometimes a cleft palate[2,7,8].

## Case Report

A 9-year-old Saudi male diagnosed with diastrophic dysplasia was referred to the pediatric dentist for the management of dental problems. The patient had consanguineous parents, 6$^{th}$ rank among eight siblings and was born as a full-term by lower segmental caesarian section delivery. On physical examination, it was observed that the patient was active and mentally alert. The patient was with short limb, bilateral talipes equinovarus, 6 toes on both feet, 'hitchhiker' thumb and scoliosis. Full body radiographic evaluation showed bifid left first rib, lumbar lordosis and bilateral tibia hemimelia (Figure 1). An extraoral examination showed hypotrophic ears (Figure 1). Intraoral examination showed over-retained 61 and 62; palatially erupting 21; carious 54; 55; 65; 74; 75; 84 and 85, and remaining roots of 64 (Figure 2). The dental diagnosis for the case was multiple caries and unilateral single anterior crossbite in reference to (irt) 21 with high labial frenal attachment.

The treatment plan sequence was followed as shown hereby: behavior modification by Tell, Show and Do (TSD) technique, preventive phase



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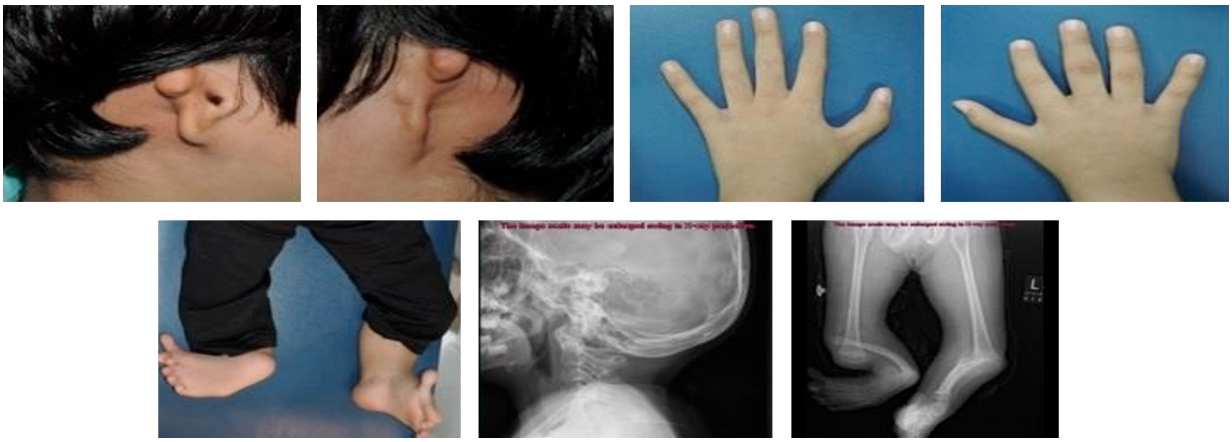

**Figure 1.** General features of diastrophic dysplasia.

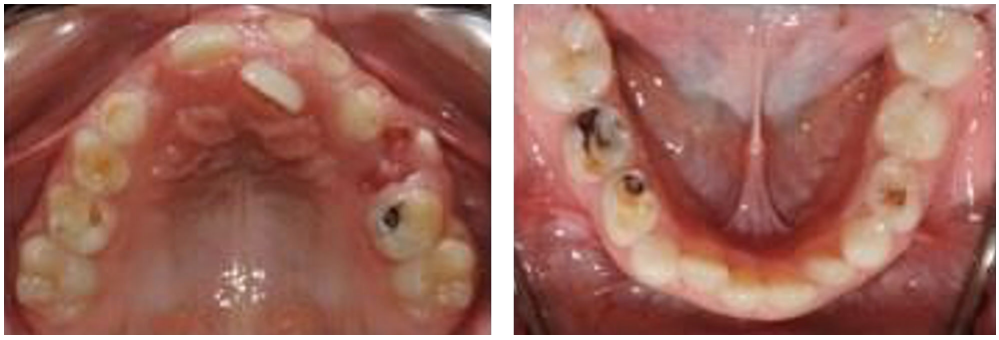

**Figure 2.** Intra-oral findings.

by educating both patient and parents about oral hygiene, restorative and surgical phase and orthodontic phase. The restorative and surgical phase was planned under local anesthesia. During the restorative phase, pulpectomy was done for 84 and 85; and 54; 55; 74; 75; 84 and 85 were restored with stainless steel crown (Figure 3). In the surgical phase extraction of 61, 62 and remaining roots

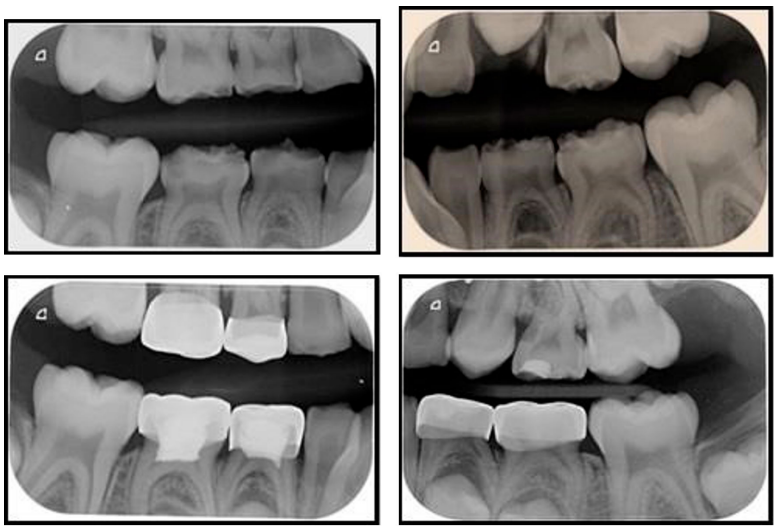

**Figure 3.** Pre-operative and post-operative radiographs.

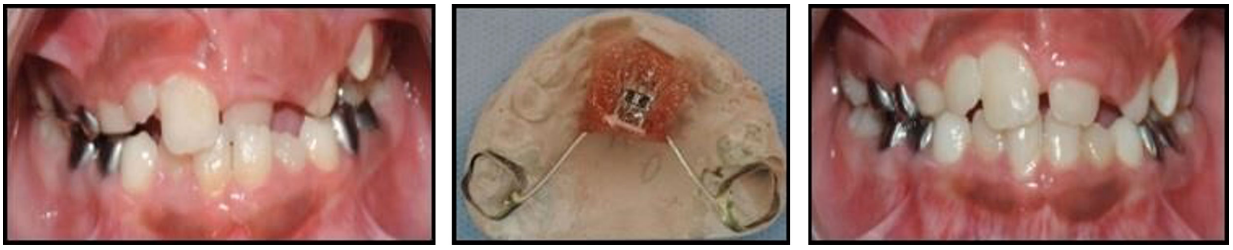

**Figure 4.** Cross-bite correction with expansion screw.

of 64 were performed. The orthodontic phase was planned for alignment of 21 with the help of the expansion screw (Figure 4). For high frenal attachment, Z-plasty frenectomy was performed (Figure 5). After 6 months follow-up found normally developing dentition and upper and lower permanent canines appears to be erupting normally (Figure 6 and 7).

## Discussion

Diatrophic dysplasia is a rare autosomal recessive inheritance disorder a type of congenital osteochondro dysplasia present at birth. The characteristic signs are caused by physeal, epiphyseal, and articular cartilage impairment all over the body[5]. Diastrophic dysplasia is also known as DD, diastrophic dwarfism, diastrophic nanism syndrome and DTD. The range and severity of symptoms and physical findings associated with diastrophic dysplasia may vary greatly from case to case. It appears to be uncommon in Saudi Arabia compared with the incidence in other countries, especially Finland. Here a case of 9-year-old Saudi child who was reported with multiple dental problems and was managed accordingly. A similar case of cleidocranial dysplasia having impacted teeth were managed by Inchingolo et al[9] by surgical orthodontics. Dental management of such cases depends on lots of factors like dental age and chronological age of the patient as well as it needs a multidisciplinary approach which focuses on preventive and symptomatic management[9].

At the movement, there is no single treatment for diastrophic dysplasia but trials looking at the effect of growth hormone and genetics are ongoing. The best possible treatment consists of a sensible diet, regular gentle exercises like swimming and routine check-up by the pediatricians.

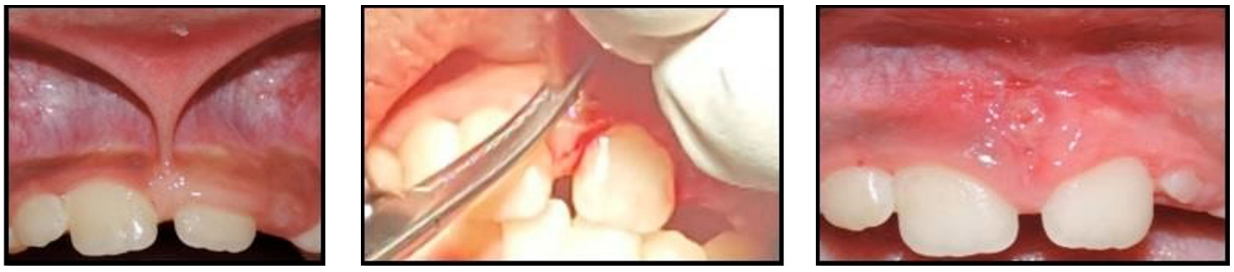

**Figure 5.** Z-plasty frenectomy.

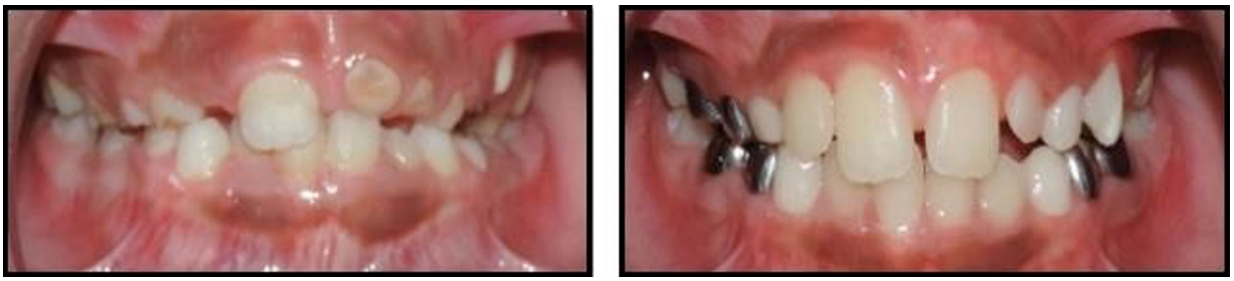

**Figure 6.** Pre-operative and post-operative (after 6 months) intra-oral.

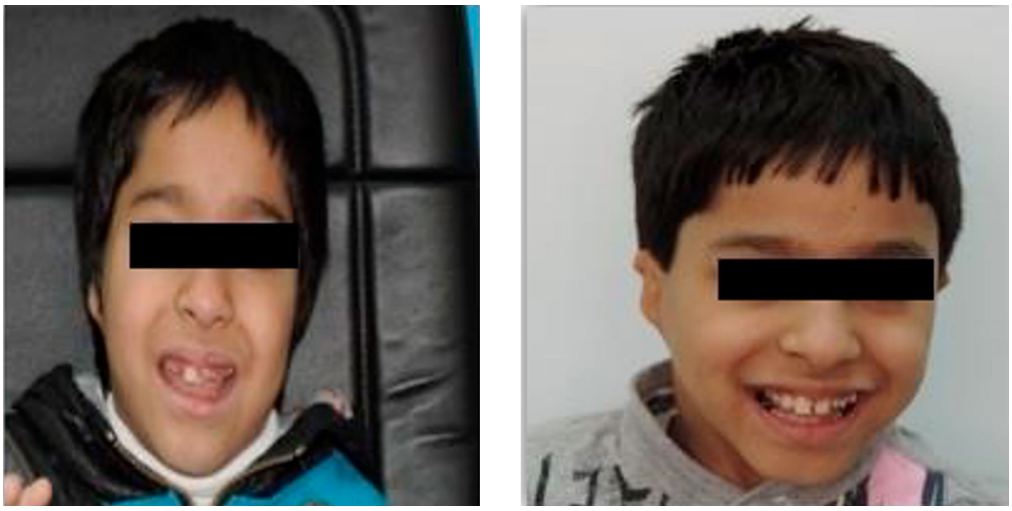

**Figure 7.** Pre-operative and post-operative (after 6 months) extra-oral.

### *Recommendations*

Treating a patient in a dental chair with deformations such as scoliosis requires extraordinary consideration. The following guidelines are used for patients with such deformities (Ruiz[6]):

*A. Before dental procedures:*

1. Utilize behavior administration strategies directly related to IQ (like with any compromised patient).
2. Assess the emotional and mental well-being of the patient.
3. Utilize family and medical histories, which may be supportive in setting up needs for the patient's treatment.
3. Record a detailed dental history.
4. Assess cleft palate, if present, plan either surgical or prosthodontic care.
5. Assess the seriousness of the patient's condition.
6. Consider referral to other specialties, making rehabilitation easier.
7. Assess the parental state of mind towards the patient's condition and educate them about issues with oral health problems and how the pediatric dental specialist can offer assistance.
8. Using proper equipment and instrumentation to prevent discomfort and damage to the patient.

*B. During dental procedures:*

1. Make the patient in the most comfortable position in the dental chair and demonstrate its safety and avoid prolonged positioning in the dental chair to prevent breathing discomfort, a potential kyphoscoliosis complication.
2. In patients with cleft palates, use compressed air and water sprays with particular care.
3. While making an impression for the dental cast, avoid using excessive impression material.
4. Encourage patient active participation.
5. Try to carry out surgical procedures in the same manner as with a patient who is physically normal.
6. While taking intraoral radiographs in a patient with cleft palate take special care.

*C. For general anesthesia:*

1. Patients with DD pose many challenges to the anesthesiologists[7,10,11].
2. The cervical stagnation should be considered, and safety measures must be taken during manipulation of the airway.
3. Extreme scoliosis can lead to lung disease and even pulmonary hypertension as well.
4. Intubation difficulties are usually secondary to a high incidence of cleft palate, posteriorly positioned both upper and lower jaws and laryngo-tracheal stenosis.
5. Neuraxial anesthesia is not contraindicated but is potentially difficult.
6. There is no known optimum dosage of the spinal and/or epidural so small doses with titration should be used.

## Conclusions

Diastrophic dysplasia is a very rare clinical condition worldwide. It shows specific skeletal and cartilaginous patterns which can be easily identified at an early stage. Such conditions need

a multidisciplinary approach, therefore, the characteristics and guidelines for the management of such cases must be known to pediatricians, anesthesiologists, and pediatric dentists, particularly at a major medical center.

### Conflict of Interest
The Author declares that there is no conflict of interests.

### Funding
The current study received no financial support.

### Informed Consent
Before starting the case, the parent was informed about the study and after getting written consent the case was started.

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