Introduction

Diastrophic Dysplasia is a rare skeletal dysplasia first described by Maroteaux and Lamy in 1960. Approximately 200 cases have been described in the literature. It’s a disorder of the cartilage and bone development. The disease is inherited in an autosomal recessive manner.

Patients with diastrophic dysplasia have epiphyseal involvement and are at increased risk for degenerative joint disease. Development and growth of cartilaginous structures are disturbed.

All skeletal dysplasia warrant multidisciplinary attention. Regular assessment by an orthopedist, geneticist, pediatrician, dentist, neurologist, and physical therapist will provide the most comprehensive treatment.

Abstract

Diastrophic Dysplasia is a disorder of the cartilage and bone development. The disease is inherited in an autosomal recessive manner. Siblings of affected individuals are at 25% risk for inheriting an abnormal allele from both carrier parents. Approximately 5% are new mutations.

Diastrophic dysplasia is one of several skeletal disorders caused by mutations in the SLC26A2 gene. This gene provides instructions for making a protein that is essential for the normal development of cartilage and for its conversion to bone. Mutations in the SLC26A2 gene alter the structure of developing cartilage, preventing bones from forming properly and resulting in the skeletal problems characteristic of diastrophic dysplasia.

Besides diastrophic dysplasia, mutations in the SLC26A2 gene can result in three other skeletal dysplasia conditions: Lethal achondrogenesis type II, achondrogenesis type 2 and mild recessive multiple epiphyseal dysplasia.

The diagnosis rests upon a combination of clinical, radiologic and histopathology features. The diagnosis is confirmed by molecular genetic testing for SLC26A2 (DJDST).

Case Presentation

(CC) A 2 month-old female presents with the chief complaint of difficulty breathing for 1 day.

PHYSICAL EXAMINATION:

Weight: 3 lbs (9% height: 44 cm (< 5%)) and a head circumference of 39 cm (5-10%); abnormal features, epicanthal folds (Figure 3), cauliflower ears (Figure 4), short limbs, small chest, pterygium (Figure 5), hitchhiker thumbs (Figure 2) and bilateral club feet.

Results

RADIOGRAPHIC FINDINGS:

- Short limbs
- Thracic platypodsphynx (Figure 5)
- Hypoplasia of the first metacarpal (Figure 6)
- Cervical kyphosis (Figure 7)
- Metatarsal veins

ECHOCARDIOGRAM:

- Patent Foramen-Ovale

Conclusions

Diastrophic dysplasia requires a multidisciplinary approach. Management principle is to maintain joint position and mobility as much as possible by physical means. Croup surgery is restricted to individuals with clinical or neurophysiologic evidence of spinal cord impingement.

Surgical correction of contractures is performed postpuberty. Annual monitoring of spinal curvature and joint contracture is appropriate.

As with many other skeletal conditions, the patient experienced respiratory insufficiency because of small rib cage and tracheal instability and collapsibility. A complication such as aspiration pneumonia is common. On occasion, the disease can be lethal at birth, but most affected individuals survive the neonatal period and develop physical limitations with normal intelligence.

Throughout life, the disorder appears to involve the skeleton, tendon, ligament and joint capsules causing restricted joint mobility. This worsens with age. Mental development and intelligence are usually normal.

Finally, all skeletal dysplasia warrant multidisciplinary attention. Regular assessment by an orthopedist, geneticist, pediatrician, dentist, neurologist, and physical therapist will provide the most comprehensive treatment.

References


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