

Diastrophic Dysplasia – Variant

A 5-year-old boy was referred for evaluation of skeletal deformities and difficulty in walking for last six months. He had history of bilateral radial head subluxation in early childhood. His height was 103 cm (10th centile), upper: lower segment ratio 1.02 and arm span 100 cm. He had lumbar lordosis and bony widening of elbows and knees; both ears were swollen (cauliflower ears) (**Fig. 1**). Hands and feet appeared normal. The skeletal radiographs revealed metaphyseal flaring of humerus, femur and tibia, short broad neck of femur and adducted metatarsals with medial twisting (**Fig. 2**). A diagnosis of diastrophic dysplasia-variant was made.

Diastrophic dysplasia is a cause of short-limb dwarfism which presents with progressive joint



FIG. 1 Widening at elbow and knee (A1) and characteristic cauliflower ear (A2).



FIG. 2 Radiological images showing widening at metaphyseal ends of femur and tibia (B1) and incurvation of metatarsals of right foot (B2).

deformities and soft tissue contractures. Diastrophic variants are taller and less severely affected. Patients may show characteristic cauliflower ears. Other physical characteristics include hitchhiker's thumbs (thumb that is abducted over a short first metacarpal), cleft palate, clubfoot and dislocation of hip, patella and radial head. The tubular bones show metaphyseal widening. Lumbar lordosis may result due to hip contractures while scoliosis or kyphosis may develop in 80% patients during disease course. Differential diagnoses include: achondroplasia (typical skull and vertebral anomalies without joint contractures, thumb or ear deformities), and spondylo-epiphyseal dysplasia (short-trunk dwarfism with involvement of vertebrae, hip and proximal epiphyseal centers of long bones).

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