

Larsen Syndrome

An eleven year old male child born to a nonconsanguineous couple presented with multiple joint dislocation since birth. He had mild motor delay. Examination showed presence of short stature. There was no microcephaly. He had flat facies, prominent forehead, depressed nasal bridge, and hypertelorism (**Fig. 1**). He had bilateral rhizomelic shortening of upper limbs, spatulate and dislocated thumbs (**Fig. 2**), bilateral elbow, ankle, and hip dislocation (**Fig.3**). Examination of parents did not reveal any features of Larsen syndrome. X-rays of long bones showed presence of bilateral tibio-femoral and patellar dislocation at knees and dislocation at hip, ankles and thumbs. He also had hypoplastic fibula on right side. X-ray spine showed presence of short and thick pedicles, kyphosis and hypoplastic superior articular facets. There was no atlanto axial dislocation. Child was referred to orthopedic surgeon for aggressive orthopedic management.

Larsen syndrome (OMIM 150250) is a complex syndrome with genetic heterogeneity, and with both autosomal dominant and autosomal recessive patterns of inheritance. Mutations in gene encoding filamin B (FLNB) result in Larsen syndrome. This gene has an important role in vertebral segmentation, joint formation and endochondral ossification and is also mutated in atelosteogenesis types I and III, and in spondylocarpotarsal syndromes. Autosomal dominant form is characterized by flat facies, joint hypermobility, congenital multiple joint dislocations, especially of the knees and talipes equinovarus. The mid-face is hypoplastic with a depressed nasal bridge. Cleft palate may be present. Osteoarthritis involving large joints and progressive kyphoscoliosis are potential complications. Airway obstruction caused by tracheomalacia and bronchomalacia may be life threatening. All affected individuals should be evaluated for cervical spine instability and caution should be taken during anesthesia because of the mobile arytenoids cartilage as well as the potentially dangerous spinal anomalies. Important differential



FIG. 1 *Facial dysmorphism. Note flat nasal bridge and prominent forehead.*



FIG. 2 *Note dislocations at thumb.*



FIG. 3 *Multiple dislocations at large joints.*

IMAGES

diagnosis is Otopalatodigital syndrome type 1 characterized by pugilistic facies, hearing loss, paddle shaped metatarsal bones, no juxta calcaneal bones and no supernumerary carpal bones.

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Calcinosis in Juvenile Dermatomyositis

A 5 year old boy was diagnosed to have JDM (Juvenile dermatomyositis) at the age of 2 years and had received treatment with corticosteroids, methotrexate and hydroxychloroquine for three years. The muscle power improved while on therapy. He presented with swelling around the knees and ankles for last two months. X-ray showed massive calcinosis around ankle joint (**Fig 1, 2**).

Calcinosis is a sequel of juvenile dermatomyositis. Its pathogenesis is not well

understood. It is said to occur in up to 30% of patients with JDM. The sites most frequently affected are the elbows, knees, digits and extremities, although it may occur virtually anywhere over the body. Longstanding active disease, especially when associated with delay in initiation of therapy is supposed to be an important risk factor for development of calcinosis. There is no specific treatment.

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FIG. 1 *Calcinosis around ankle joint.*

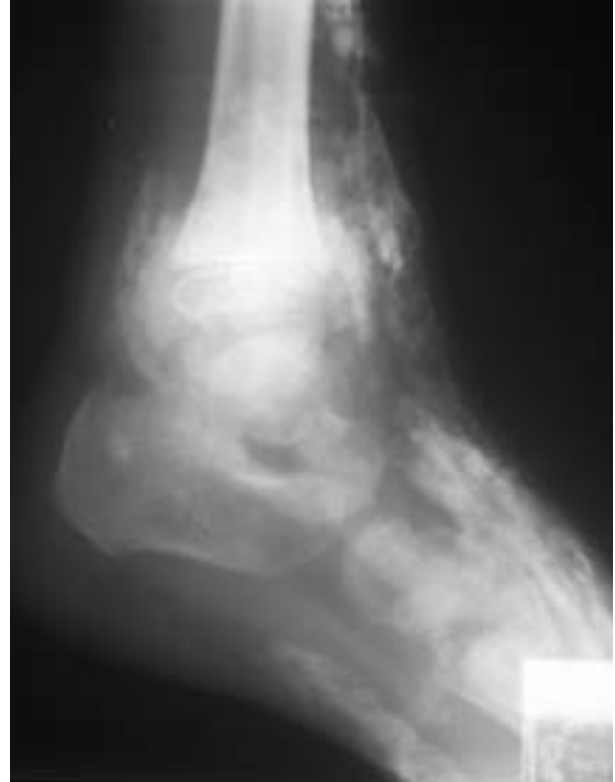


FIG. 2 *Calcinosis around ankle joint (lateral X-ray).*