

Larsen Syndrome: A Case Report

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Abstract

Larsen syndrome was first described in 1950 by Larsen, Schottstaedt and Bost. This rare inherited disorder is characterized by congenital dislocation of multiple joints along with other anomalies of heart, face, hands and bones. Awareness of this condition and associated complications helps in better follow up and management of these patients.

Key words: Genu recurvatum, Short stature, Hypodontia, Bifid uvula, Submucosal cleft palate, Cardiac anomaly

Introduction

Larsen syndrome is a rare genetic disorder characterized by the association of multiple joint dislocations, skeletal abnormalities and atypical face including hypertelorism, depressed nasal bridge and prominent forehead. We report a nine years old female child with short stature, bilateral congenital dislocation of knees, hypodontia, bifid uvula and major cardiac anomaly.

The Case

A nine years old female child presented to us with the complaints of congenital bilateral dislocation of knees and difficulty in walking due to restricted flexion at the knee joint. She was born at term, third born to non-consanguineous parents. Pregnancy was uncomplicated and there was no exposure to any known teratogenic agent. Her birth weight was 1.4 Kilograms and at birth, her lower limbs were hyper-extended, touching her forehead. On the third day of life, she was evaluated for cardiac anomaly and 2-D-Echocardiography revealed a large subaortic VSD, aortic override, severe right ventricular outlet obstruction. Total intra-cardiac repair of Tetralogy of Fallot was done at eight years of age.

Her milestones were within normal limits, her intelligence was normal and academic performance was good. Her vision and hearing was normal. On examination, she had short stature and her height was 115 cm (87% of expected). She weighed 15 kg (58% of expected).

She had facial dysmorphism with hypertelorism, flat nasal bridge, prominent forehead and micrognathia

The central and lateral incisors in the lower jaw were missing in primary and secondary dentition. There was a submucosal cleft in the palate and bifid uvula.

She had hypermobility at wrist, elbow, and ankle joints and bilateral dislocation of knee joints with limited flexion at the knee joint. She also had spina bifida at S1 level. X-ray wrist joint did not reveal extra carpal bones, but in the Ankle, there were accessory tarsal bones with atypical calcaneus. No other family members were affected. Chromosomal analysis detected our case as 46XX (normal constitutional karyotype) (GTG band technique)

Discussion

Larsen syndrome is a rare genetic disorder with an incidence of about one in 100,000. It is characterized by joint hypermobility and multiple joint dislocations, especially of knees and feet¹. The characteristic facial features include midfacial hypoplasia with a depressed nasal bridge. Sometimes present are other birth defects such as structural heart defects, cleft palate, cataracts, extra bones of the wrist, and abnormalities of the vertebrae.

Percin et al. have reported a case of a fifteen year old Turkish girl with Larsen syndrome with maxillary prognathia, malocclusion, supernumary



Fig 1a: Showing facial dysmorphism with hypertelorism, depressed nasal bridge, micrognathia and broad forehead.



Fig 1b: Showing Hypodontia (Missing incisors in lower jaw).



Fig 1c: Showing Genu Recurvatum.



Fig 1d : Showing Extra Tarsal Bones

teeth, macroglossia and microdontia². Sanjani et al. have reported an eight year old Chinese boy with Larsen syndrome who had advanced periodontitis³. Hypodontia with missing central and lateral mandibular incisors has not been reported. Mukund D Rahalkar et al have reported a case of extra carpal bones⁴. Our case had extra tarsal bones.

Orhan et al. have reported a case of prenatally diagnosed lethal type Larsen-like syndrome associated with bifid tongue⁵. Klenn et al have reported a case of complete agenesis of anus with the presence of bifid uterus⁶. Bifid uvula in Larsen syndrome has not been previously reported.

Etiology of Larsen's syndrome is a generalized mesenchymal disorder that arises from a defective process of embryonal induction involving mesenchymal tissue. Sequence analysis of FLNB has revealed mutation in Filamin B.

Autosomal dominant and autosomal recessive forms of the condition have been proposed. A lethal form

of Larsen syndrome (Larsen like syndrome) has been described as a combination of Larsen phenotype and pulmonary hypoplasia. Anomalies such as syndactyly, cleft palate, severe short stature and major cardiac anomalies were more frequently seen in patients with the recessive form⁷. The only form of prenatal diagnosis available to date is ultrasound. Fetal ultrasound performed by a specialist at 18-20 weeks of pregnancy can sometimes reveal signs of Larsen syndrome. Knee dislocations and hyperextension, club feet, fixed flexion of elbows, wrists, and fingers, and some of the characteristic facial features can sometimes be noted by ultrasound in affected fetuses.

Treatment will vary according to the symptoms of a particular child. Joint problems require long-term orthopedic care. Hearing should be evaluated on a periodic basis, especially in children, because of the potential for conductive hearing loss. Ophthalmologic examinations are recommended periodically to screen for cataracts. The condition does not affect intelligence and children can expect to have normal school experiences, with the exception of physical education,

which will need to be adapted to each child's Needs. Reconstructive surgery is a viable option for restoration of knee stability and function. The spinal status of these children must be monitored through out their life.

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