**Introduction**

Larsen syndrome is an extremely rare genetic condition that occurs in about one in every 100,000 births. It is characterized by congenital dislocations of multiple body joints along with other unusual features of the face, hands, and bones.

**Case Summary**

The subject of this report is a Hispanic female infant delivered by spontaneous vaginal delivery at 40 weeks gestation to a 20-year-old G1P0 woman. Pregnancy was uneventful and the results of routine maternal prenatal screening tests were negative. The mother denied family history of congenital birth defects. Labor and delivery progressed normally without an uncomplicated induction. Apgar scores were 7 and 8 at 1 and 5 minutes respectively.

At birth, the infant appears in no apparent distress, is alert, and has a weight of 3,333 g (20%), a length of 47 cm, and a head circumference of 34 cm (20%).

Examination of the head and neck is significant for short neck, prominent forehead, flat face, depressed nasal bridge, hypertelorism, micrognathia, small mouth and cliff palate. Musculoskeletal examination showed dislocated elbows, hips and knees, shortened proximal and upper lower extremities, bilateral club feet and rocker-bottom deformity of right feet, clinched hands and overlapping fingers of both hands. There were no abnormal pulmonary, cardiovascular or abdominal findings.

**Discussion**

Larsen syndrome is an osteochondrodysplasia characterized by large joint dislocations and characteristic craniofacial abnormalities. The cardinal features of the condition are dislocations of the hip, knee and elbow joints, with equinovarus and equinovarus foot deformities. Spina bifida, marked in most infants, are also present. Craniofacial anomalies include hypertelorism, prominence of the forehead, a depressed nasal bridge, and a flattened midface. Other birth defects such as structural heart defects, cleft palate, cataracts, extra or small bones of the ear, and abnormalities of the vertebrae may also be present.

Respiratory problems are frequently seen at birth because of laxity of the trachea. Feeding and swallowing difficulties are common. Cervical spine instability is a very serious complication of Larsen syndrome as it can cause compression of the spinal cord and lead to paralysis or death. Hearing loss is also a well-recognized complication.

Both recessive and dominant patterns of inheritance have been described. Some cases are sporadic. Bicknell et al. (2007) identified several different heterozygous mutations in the FLNB gene in 20 unrelated patients with Larsen syndrome.

Larsen syndrome should be suspected in any baby having multiple joint dislocations at birth. Currently, there is no routinely available genetic test to confirm the diagnosis and, thus, diagnosis must be based on clinical and x-ray findings.

Treatment requires a multidisciplinary approach and varies according to the symptoms of a particular child. Joint problems require long-term orthopedic care. Regular medical examinations are crucial to assess the condition of the bones, joints, spine, heart, and eyes. MRI of the neck is recommended in children to screen for cervical vertebral problems. Hearing should be evaluated on a periodic basis, especially in children, because of the potential for conductive hearing loss. Bone age determinations are also recommended periodically to screen for cataracts.

**Conclusion**

Larsen syndrome should be suspected in any baby having multiple joint dislocations at birth. The effects of the syndrome vary markedly from person to person. Treatment is generally focused on the specific problems of the individual. Death can occur due to cardiopulmonary arrest, and long-term problems can result from compression of the cervical spine. Respiratory complications are also frequent, with recurrent infections, as in our patient.

**References**

3. Online link - http://www.omim.org/entry/150250