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Case report of Larsen syndrome presenting as acute onset quadriparesis

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Abstract : Larsen syndrome is one of the rare inherited disorders which affects the development of bones in the body. It is associated with multiple anomalies in different systems. Larsen syndrome involves skeletal system causing features like multiple joint dislocations, hypermobility of the joints, club foot, developmental dysplasia of hip usually teratologic type and supernumerary carpal and tarsal bones. It also involves spinal abnormalities like cervical kyphosis, thoracic scoliosis which can lead to compression on the cord and leading to myelopathy kind of picture which might need surgical treatment. Typical facial features include mid face hypoplasia, nasal bridge prominence, wide set eyes. There are usually of short stature and normal intelligence. In this case report we are presenting a case of Larsen syndrome who presented to us with quadriperesis secondary to subluxation at C3-4 and C4-5 with cervical kyphosis. The patient was a 15 year old boy presented with difficulty in walking for a period of 10 years, weakness of both the upper and lower limbs for a period of 3 months after a trivial fall. He had typical facial features like prominent forehead, wide set eyes. He underwent three stage cervical decompression and instrumented fusion. At 9 months follow up he has improved by 2 grades in his upper and lower limbs but he needs support for daily activities. X ray and CT scan shows fusion between C3-C6.

Keyword : Larsen syndrome, multiple joint dislocations, cervical kyphosis, quadriparesis.

Introduction: Larsen syndrome was first described by Larsen in 1950 as a rare genetic disorder which is associated with a wide variety of different symptoms. Classic features include dislocation of large joints, cervical kyphosis with subluxation, facial abnormalities, skeletal malformations, etc. Cervical spine abnormalities can be associated with progressive myelopathy.

Case: A 15 year old boy born to consanguineous couple, with delayed motor milestones till 3 years of age, presented with complaints of difficulty in walking for 10 years. He developed weakness of both the upper limbs and worsening weakness of lower limbs after a fall 3 months before presentation. 2 weeks before admission he had a second fall with further worsening of both upper and lower limb weakness. On examination he was of short stature, had wide intercanthal distance, prominent forehead, hyperlaxity of multiple joints, deformity in the spine, bilateral club feet, weakness in both the upper and lower limbs. Shoulder muscles were grade 2/5, elbow, wrist 3/5, fingers 1/5 , bilateral hips 3/5, knees 2/5, ankle dorsiflexion 0/5, EHL and FDL were 3/5 and plantar flexion 2/5. Biceps, triceps reflexes were exaggerated with clasp knife rigidity. He had a reverse supinator reflex. Knee and ankle jerks were exaggerated along with clonus, bilateral Hoffman's positive and bilateral plantar reflexes were extensor. There were no sensory deficit.

Figure 1

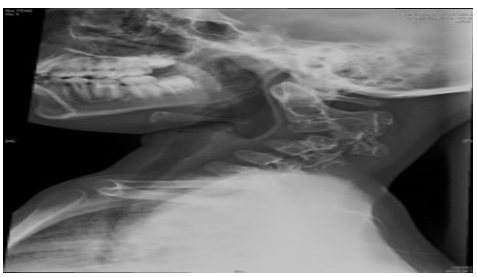

Figure 2

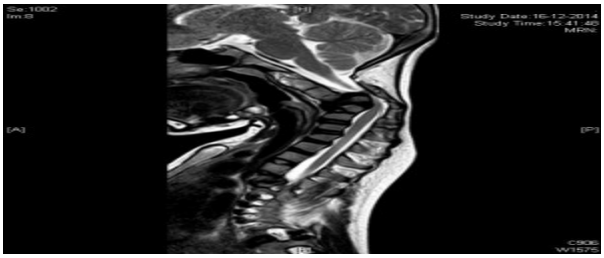

Figure 3

Imaging studies revealed - mid cervical kyphosis with dislocation of C3 C4 joint and subluxation of C4 C5 joint. The C4 vertebra was hypoplastic and was posteriorly migrated into the cervical canal. MRI showed dysplastic C4 and C5 bodies with posterior angulation causing severe spinal canal compromise and myelomalacic changes As a first stage, he underwent C2-C6 laminectomy, decompression and C2, C7-T1 pedicle screw fixation. Without adding the rods he was positined supine for an anterior procedure. At this time of surgery he developed cardiac arrest and was resuscitated. Further surgical procedure was deferred and was shifted to intensive care unit. He had worsening of neurology with grade 0 power of all the muscles groups, but with intact pin prick and light touch sensations. He was put on skull traction with gradual increase in weights. He showed improvement in his motor power in both the limbs. After 2 weeks he underwent anterior C4, 5 corpectomy, C3-6 fibula grafting and plating followed by posterior rod application on previously applied pedicle screws. After the surgery a Minerva cast was applied which was continued for 3 months.

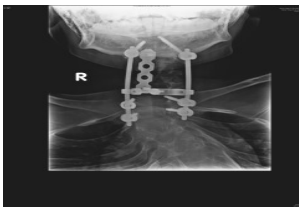

Figure 4

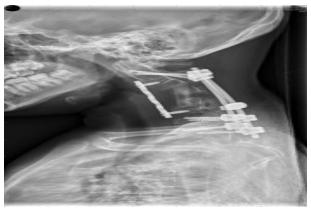

Figure 5

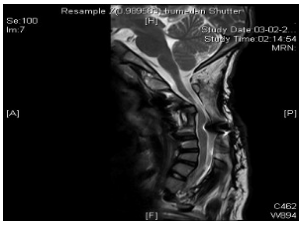

Figure 6

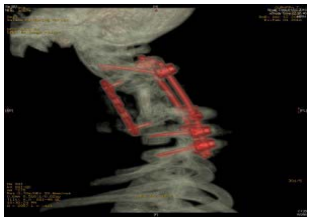

Figure 7

He gradually became better and at 9 months follow up, the motor power improved to 4/5 in shoulder muscles, elbow flexion 4/5, extension 3/5, wrist and finger muscles with a fair hand grip and lower limb muscle power was Hip muscles 3/5, knee 3/5, ankle dorsiflexion and plantar flexion 3/5, EHL was 3/5. His hand grip improved and he can hold spoon and eat food. His deep tendon reflexes are exaggerated. He was made to stand with tilt table and is now able to sit with support. He is undergoing rehabilitation and graduated physiotherapy. He is still dependent for his daily activities.

Discussion:

Larsen et al described this rare genetic disorder in 1950(1). Characteristic features of the syndrome are hypermobility of joints, multiple joint dislocations and facial abnormalities. Incidence was estimated to be one in 100,000(2). Both autosomal dominant or classic form and recessive forms were described. Classic form is caused by mutations in the FLNB (Filamin B) gene. This mutation may occur spontaneously or can be inherited as autosomal dominat. (3) Mutations in the gene CHST3 (Carbohydrate Sulfotransferase 3) were identified in the recessive trait of the Larsen syndrome.(3) Skeletal and joint abnormalities include dislocations or subluxations of major joints like hip, knees and elbows, supernumerary carpal bones which sometimes may be fused.(3) Hypermobility of joints leading to floppy fingers especially thumb. Spinal abnormalities like kyphosis or scoliosis along with dysplatic vertebrae. Facial features include mid facial hypertrophy leading to prominent forehead, wide intercanthal distance, depressed nasal bridge. Cervical spine abnormalities like kyphosis, dysplastic vertebra (flattened vertebrae(4), dysraphism, cleft in laminae, etc (3)) are common in patients with Larsen syndrome which may lead to compromise in the canal causing weakness and sometimes death. Approximately a dozen cases of Larsen's syndrome managed surgically for cervical spine abnormalities were mentioned in the literature. (5) Our patient had cervical kyphosis with dysplatic subluxed vertebrae with cord compression leading to myelopathy for which he underwent staged surgery. He is gradually improving but still is dependent for his daily activities.

Conclusion: Larsen syndrome patients with cervical myelopathy secondary mid cervical kyphosis and subluxation will benefit from surgical treatment and may need multi stage surgeries.

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