

Case report

Lumbo-Costo-Vertebral Syndrome; with Associated Unique Constellation of Congenital Anomalies

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Lumbocostovertebral syndrome with associated congenital lumbar hernia is a rare entity. We present a case of lumbocostovertebral syndrome with a unique constellation of associated anomalies i.e; Arnold-Chiari type 2 malformation, ectopic kidney and inguinal hernia. Embryological background of lumbocostovertebral syndrome with associated other congenital anomalies is also discussed. All cases of congenital lumbar hernia should also be investigated for associated other such congenital anomalies.

Key Words: Lumbocostovertebral Syndrome; Congenital Lumbar Hernia; Congenital Inguinal Hernia; Arnold-Chiari Malformation

Introduction

Lumbo-costo-vertebral syndrome (LCVS) is a condition that includes hemivertebrae, absent ribs, and abdominal wall muscle hypoplasia presenting as congenital lumbar hernia. Congenital lumbar hernia (CLH) is a rare anomaly, with around 70 reported cases. CLH associated with lumbocostovertebral syndrome is an infrequent condition, with around 20 cases been reported in literature so far. Some cases of lumbo-costo-vertebral syndrome have also been reported with associated other congenital defects.

We are reporting a patient with lumbo-costo-vertebral syndrome with associated unilateral inguinal hernia, ectopic kidney and Arnold-Chiari type II malformation, which is a unique constellation of anomalies. To our knowledge it is the first reported case of its nature.

Case Report

A 10 days old male neonate presented with swellings over back and right side of abdomen since birth with appearance of another swelling over inguinal area by the 9th day of life. Being breech presentation

and twin pregnancy, he was born through caesarean section at 35 weeks of gestation. His birth weight was 2.4 kg with APGAR of 7 and 9 at 1 and 5 minutes respectively. There was no history of maternal diabetes or preeclampsia. There was no family history found for birth defects, genetic disorder or consanguinity.

**Figure 1:**

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On examination, he had OFC of 31 cm and length of 37 cm. He was found to have a swelling at the lower back region, bluish in color, soft, 4 x 4cm in size, well-defined and irreducible (Meningocele). There was another swelling in the right lumbar region, measuring 5 x 5 cm, ill-defined, compressible but irreducible. A reducible swelling over right inguinal area was also noticed with inability to get above that swelling. Thoraco-lumbar scoliosis with convexity towards the right side was present. The neurological examination of the lower limbs and rest of the systemic examination were normal.



Figure 2:



Figure 3:

Radiological investigations revealed T9-S1 hemi-vertebrae with hypoplastic ribs (right sided; T7- T9), absent ribs(right sided;T10,T11 and T12) and thoraco-lumbar scoliosis.Cranial USG showed hydrocephalus (Arnold-Chiari type 2 malformation). Abdominal USG showed right ectopic,mal-aligned kidney with right lumbar swelling and right inguinal canal showing segment of herniated gut in it (lumbar hernia and inguinal hernia).

Multidisciplinary approach involving the Pediatrician, Pediatric Neurosurgeon, Pediatric general surgeon, Developmental Pediatrician and Physiotherapist was adopted. Meningocele was repaired by the Pediatric Neurosurgeon with uneventful post-operative recovery. Oral Acetazolamide was given for hydrocephalus. Inguinal Hernia repair is in plan and the child is on regular follow up of Pediatrician, Pediatric Neurosurgeon, Pediatric Surgeon, Developmental Pediatrician and Physiotherapist.

Discussion

Lumbocostovertebral syndrome (LCVS) is a condition that includes hemivertebrae, absent ribs, and abdominal wall muscle hypoplasia. Congenital lumbar hernias are an uncommon type of hernia and usually presents as an asymptomatic large mass in the superior lumbar triangle (Grynfeltt-Lesshaft triangle) or in the inferior lumbar triangle (Petit triangle) [1]. Various other malformations have been associated with LCVS. These include the VACTERL association, syndactyly, focal nodular hyperplasia, posterior spinal dysraphism, Arnold-Chiari malformation, caudal deficiency, tethered spinal cord, renal agenesis, ureteropelvic junction obstruction, cloacal exstrophy, cryptorchidism, lumbar, diaphragmatic, inguinal and sciatic hernia, absent tibia, lung hypoplasia and supranumerary digit [2-9]. Our case describes a rare combination of congenital lumbar hernia, right inguinal hernia, right ectopic kidney and Arnold-Chiari type II malformation associated with LCVS.

In 1972, Touloukian [5] described the lumbocostovertebral syndrome as a single somatic defect that leads to various anatomical defects including the trunk musculature, vertebrae and ribs. He described a case of a newborn with ribs hypoplasia, hemivertebrae, and bilateral lumbar hernias. He ascribed these anomalies to agenesis of a somite during 3rd to 5th week of embryogenesis. At the gestational age of 3 to 5 weeks, the mesoderm differentiates into somites that are further differentiated into the sclerotome (vertebrae and costal processes), the myotome (skeletal muscles of the trunk) and the dermatome (deep layers of the skin and the subcutaneous tissue) [10]. Any interruption during this stage of development may result in defects in the structure of vertebrae, ribs and abdominal wall [11]. It has been speculated that this anomaly occurs from a vascular accident early in embryogenesis which leads to a developmental field defect [5].

There is no specific genetic defect that has been identified yet for this syndrome, therefore leaving this syndrome as a clinical diagnosis only. Twin pregnancy may be a risk factor for the development of this syndrome. Identification of anomalies in Lumbo-costo-vertebral syndrome should therefore direct the clinician towards thorough evaluation for other associated anomalies. It is therefore important to investigate patients with vertebral and abdominal defects to rule out these related anomalies along with timely management with multidisciplinary approach.

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