

Congenital Lumbar hernia with lumbo-costo vertebral syndrome with ipsilateral short femur, tibial and fibular hemimelia

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ABSTRACT

Congenital lumbar hernia is a rare anomaly. Congenital lumbar hernia is usually associated with lumbo-costo vertebral syndrome. Lumbo-costo vertebral syndrome is a condition that includes spinal anomalies like hemivertebrae, absent ribs, and abdominal wall muscle hypoplasia. It usually presents as a symptomatic large mass in lumbar triangle (either superior or inferior lumbar triangle). Congenital hernias occurred due to defects in the musculoskeletal system. These defects occur probably due to single somatic defect which occurs between 3-5 weeks of gestational age with resultant malformation of vertebral bodies, ribs and abdominal musculature. Incomplete form of lumbo-costo vertebral syndrome shows congenital hernia, spinal anomalies with no rib abnormalities. Congenital lumbar hernia occurs in superior lumbar triangle in lumbo-costo vertebral syndrome due to absence of 11th and 12th ribs with attenuation and weakened attachments of the involved adjacent musculature. We report a case of 2-year female child having swelling right lumbar region since birth due to congenital lumbar hernia with associated spinal anomalies.

Keywords: Lumbar hernia, lumbo-costo vertebral syndrome, congenital, abdominal wall defect

CASE REPORT

A 2-year old female child presented with swelling in right lumbar region since birth which increased on crying. Her right thigh, leg and foot were small in size (Figure 1). Ultrasound of abdomen showed a large defect in abdominal wall in right lumbar region through which herniation

of bowel loops was noted suggestive of lumbar hernia (Figure 2). Antero-posterior and lateral radiographs of dorso-lumbar spine were taken in view of abnormal curvature in dorso-lumbar spine. Radiographs revealed marked scoliosis in lumbar spine with convexity towards left. D12 vertebra showed butterfly vertebra with hypoplastic right half of vertebral body. L1 to L3 verte-

Figure 1 – Photograph of patient showing (A) - right lumbar hernia, (B)- small sized right leg and foot.



bral bodies showed butterfly vertebrae. There was partial agenesis of the Sacrum with absence of S4-S5 sacral segments. Right 11th and 12th ribs were absent. Discontinuity in pro-peritoneal fat planes was noted in right lumbar region through which bowel loop herniation was seen suggestive of lumbar hernia (Figure 3). Radiograph of right thigh and leg showed short right femur and hy-

poplastic (hemimelia) right tibia and fibula (Figure 4). MRI of Lumbo-Sacral spine showed partial sacral agenesis with absence of S4-S5 sacral segments, scoliosis of lumbar spine with convexity towards left. Butterfly D12 vertebra with hypoplastic right half, butterfly L1-L3 vertebrae, widened spinal canal in lumbar region. Syringomyelia was noted at D12-L1 level and defect

Figure 2 – Ultrasound in right lumbar region (A, B) – showing defect in parietal peritoneum (marked by white arrow) in right lumbar region with herniation of colon suggestive of lumbar hernia.

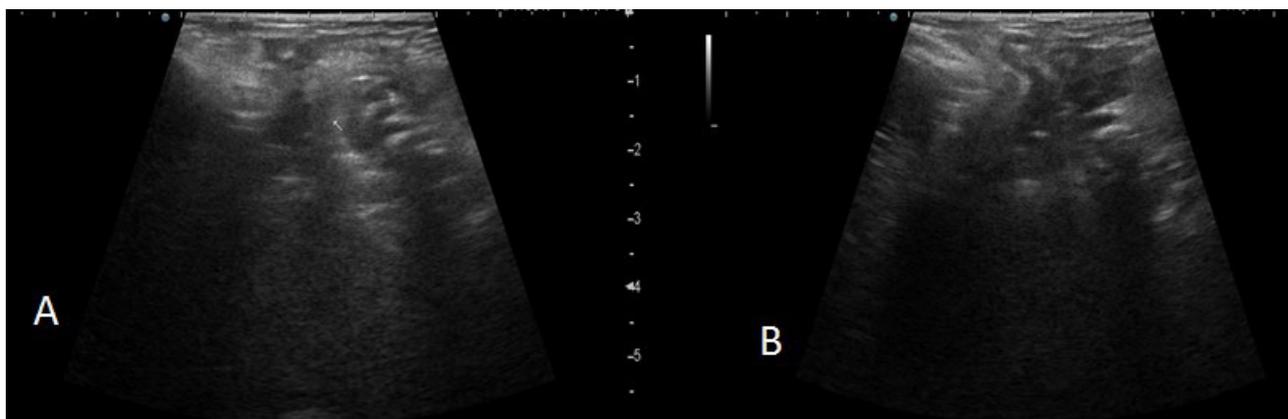


Figure 3 – Radiograph of lumbosacral spine AP (A), lateral (B) showing multiple vertebral anomalies in lumbar spine, absent right 11th and 12th ribs with right lumbar hernia.

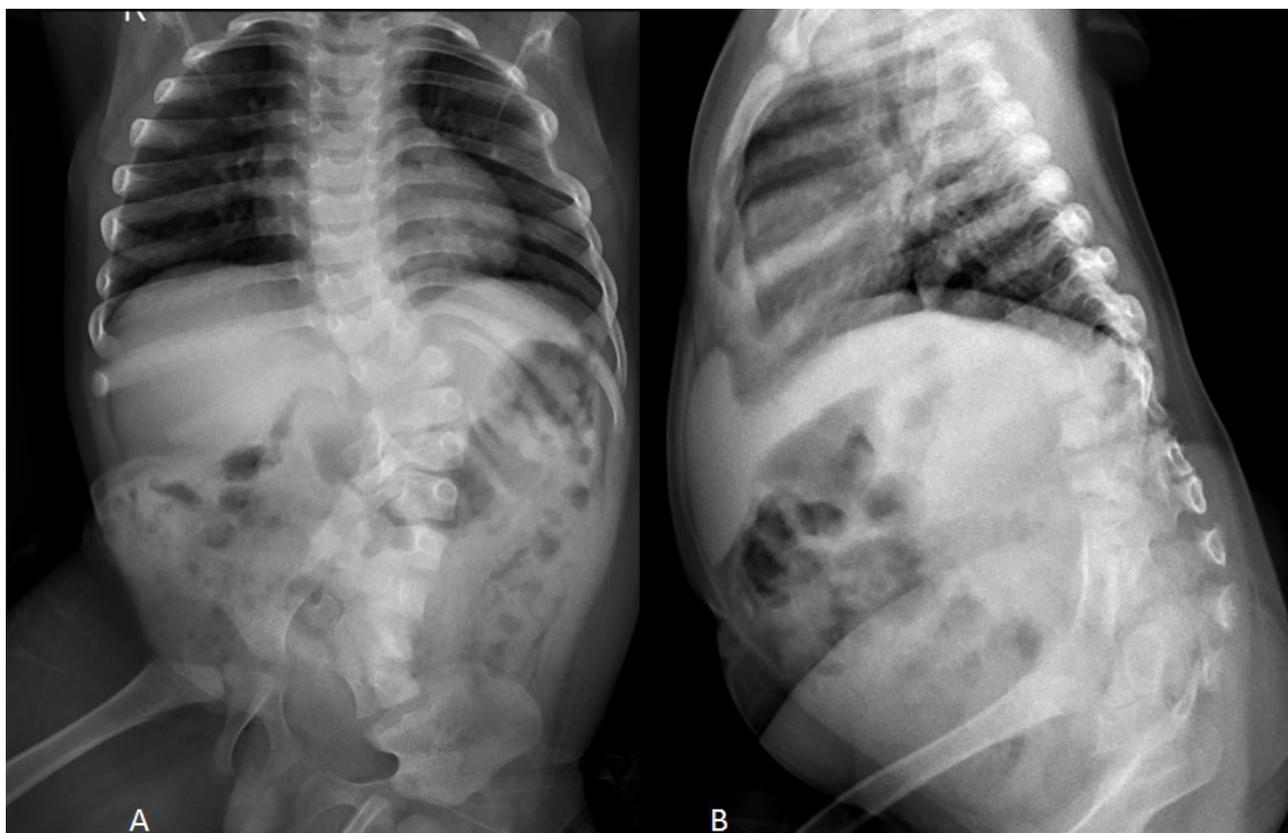


Figure 4 – Radiograph of both thighs AP(A), right leg AP (B)- showing short right femur and right tibial and fibular hemimelia.

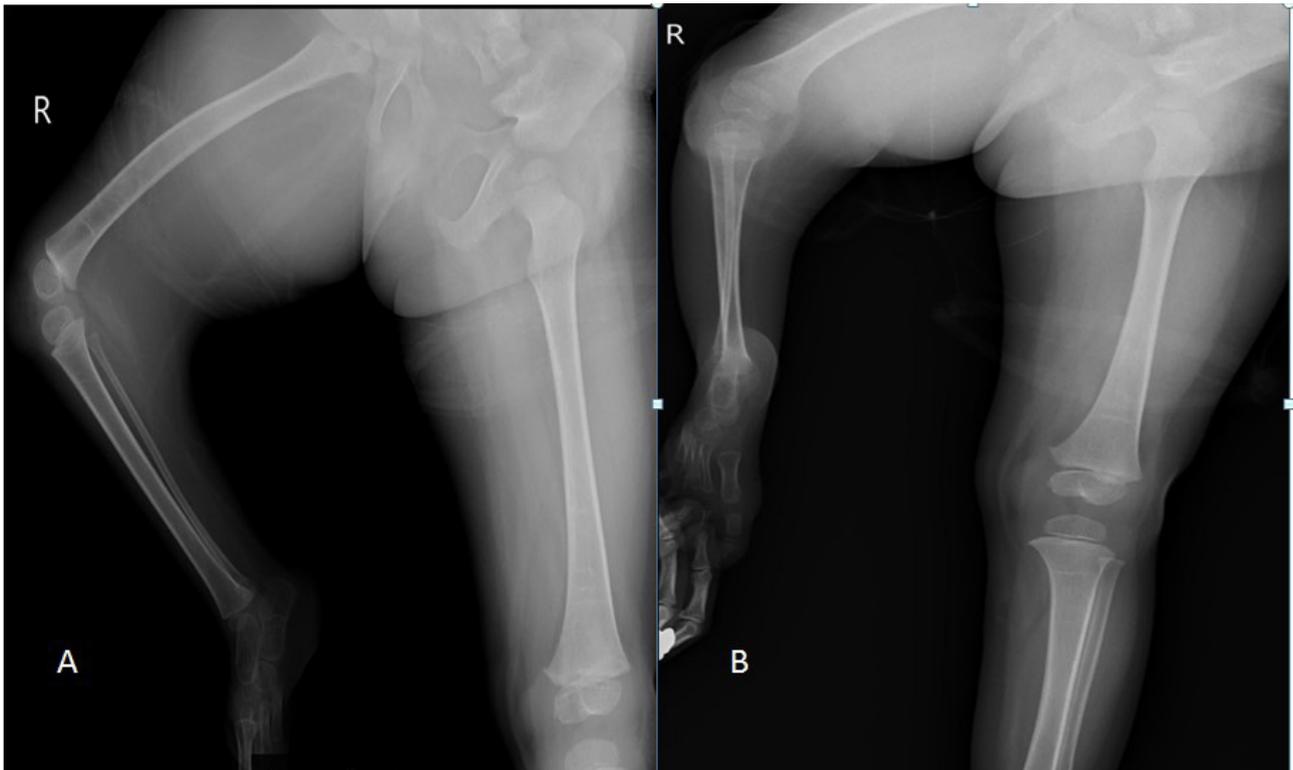


Figure 5 – MRI Lumbo sacral spine Coronal STIR (A,B)- showing right lumbar hernia containing colon(A) and multiple vertebral anomalies in lumbar spine(B).

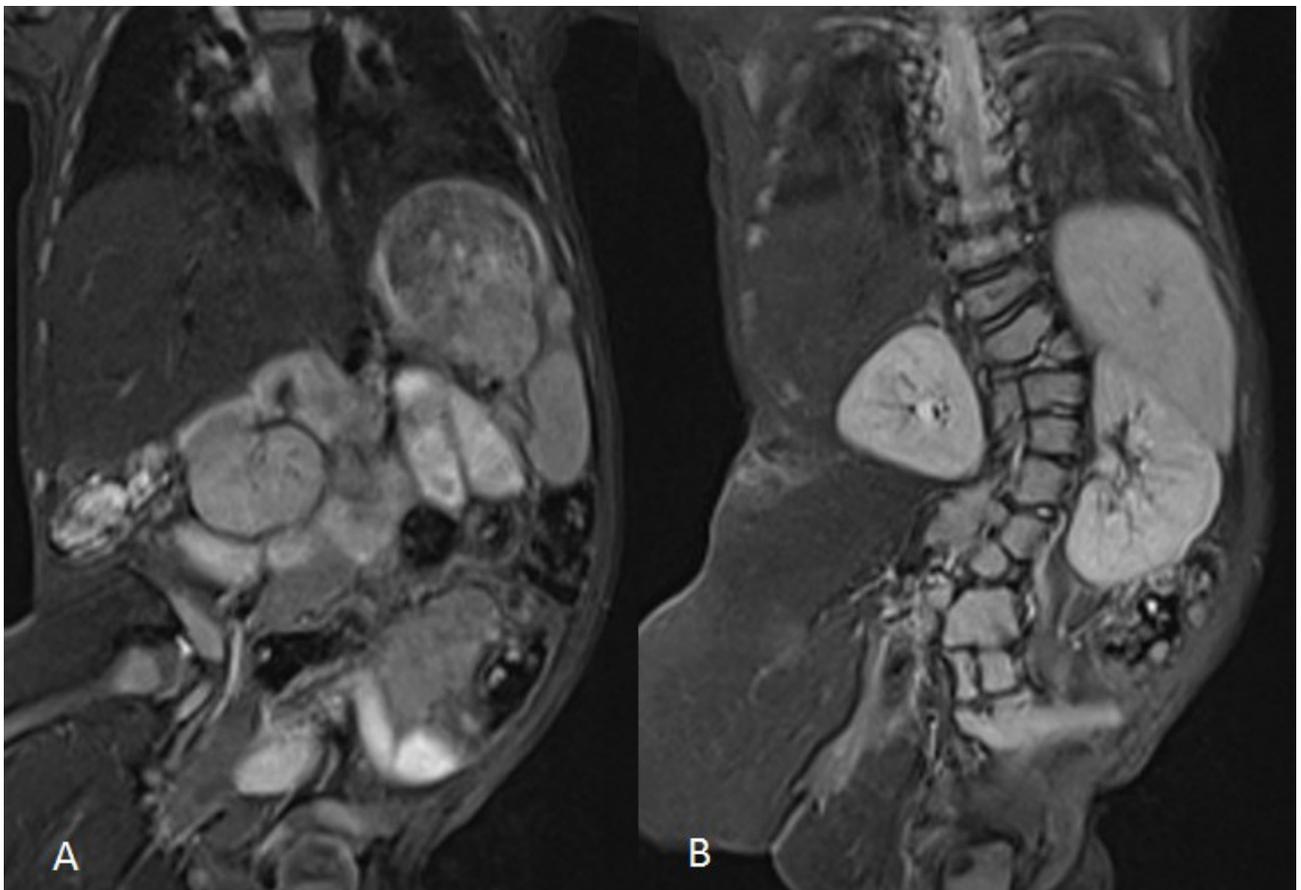
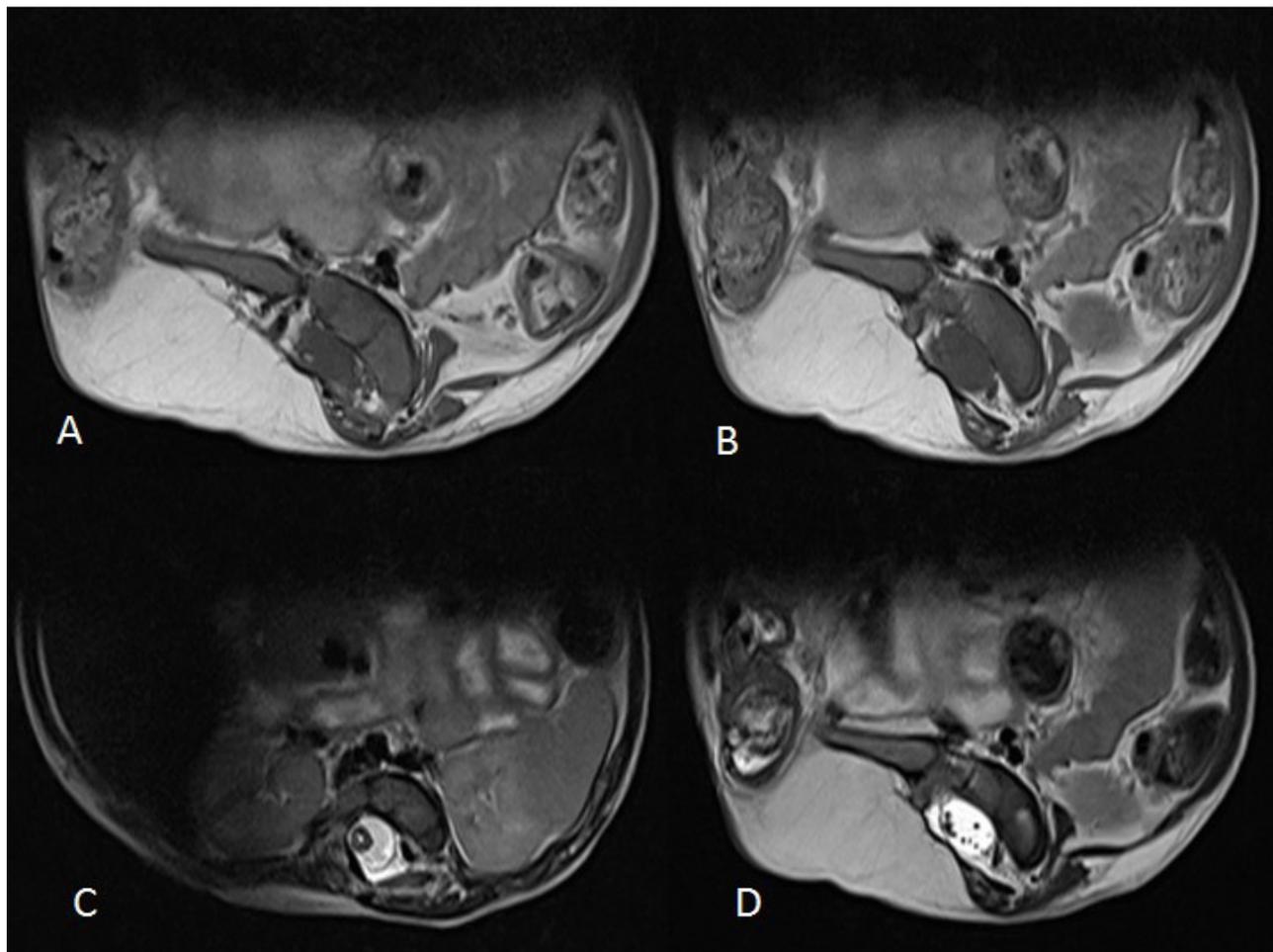


Figure 6 – MRI Lumbosacral spine axial T1WI (A, B) and axial T2WI (C,D) showing right lumbar hernia containing right colon.



DISCUSSION

Congenital lumbar hernia is a rare anomaly. Earliest report was made in 1803.^{1,2} Congenital lumbar hernia is usually associated with lumbo-costo vertebral syndrome. Lumbo-costo vertebral syndrome is a rare condition with only 16 cases reported in English literature.¹ Lumbo-costo vertebral syndrome is a condition that includes spinal anomalies like hemivertebrae, absent ribs, and abdominal wall muscle hypoplasia. It usually presents as a symptomatic large mass in lumbar triangle (either superior or inferior lumbar triangle).¹ Lumbo-costo vertebral syndrome may be associated with other malformations like VACTERL association (Comprising vertebral anomalies, anal atresia, cardiac defects, trachea-oesophageal fistula, esophageal atresia, renal anomalies and limb abnormalities), diaphragmatic hernia, PUJ obstruction, renal agen-

esis, cloacal extrophy, cryptorchidism, hydrocephalus, focal nodular hyperplasia of the liver, spinal dysraphism, caudal deficiency, syndactyly.^{3,4} Lumbo-costo vertebral syndrome may be seen in infant of diabetic mother.¹ Lumbar hernia can be congenital (20% of total) or acquired (80%). Congenital hernias occurred due to defects in the musculoskeletal system. They occur individually or in association with other abdominal hernias, lumbo-costo vertebral syndrome and neurofibromatosis (Type1).⁵ Acquired lumbar hernia can be primary or secondary. Primary lumbar hernias (55%) are spontaneous without any causal factors like surgery, trauma, and infection. Secondary lumbar hernia (25%) occur due to blunt, penetrating or crushing trauma, surgical lesions, post-operative, infections in pelvic bone, ribs or lumbodorsal fascia, infected retroperitoneal hematomas, hepatic abscess. Tovloukian in 1972 first described lumbo-costo vertebral

syndrome.⁶ Lumbo-costo vertebral syndrome includes abdominal wall muscular hypoplasia, congenital absence of ribs, hemi-vertebrae and anterior myelomeningocele. These defects occur probably due to single somatic defect which occurs between 3-5 weeks of gestational age with resultant malformation of vertebral bodies, ribs and abdominal musculature. The crown rump length of embryo is 1.5 mm between 3-5 weeks of gestational age when mesoderm between the endoderm and ectoderm on either sides of notochord differentiation into somites which differentiate into the sclerotome (which form the vertebrae and costal process and the myotome (that forms skeletal muscle of tongue and the dermatome (which forms skin and subcutaneous tissue). Vertebral, costal and abdominal wall defects can occur if there is any disturbance during this stage like anoxia demonstrated in animal study. Neurulation occurs during the 3-5 gestational age during which formation neural groove, closure of anterior (25 days) and posterior (27-28 days) neuropores, appearance of ventral horn cells, formation of anterior and posterior roots and disc junction of neuroectoderm from the surface ectoderm occurs.⁷ Hence lumbo-costo vertebral syndrome can be associated with spinal cord malformations. Hence spinal dysraphism may be also associated.⁷ Other anomalies associated with LCVS are congenital sciatic hernia / absent tibia, post myelomeningocele. Rarely lumbo-costo vertebral syndrome may be associated with congenital subcostal hernia.⁸ Etiologies of both subcostal hernia and lumbar hernia are same in lumbo-costo vertebral syndrome. Anomalies associated with lumbo-costo vertebral syndrome are lung hypoplasia, supernumerary digits, undescended testis, hydrocephalus due to Arnold Chiari malformation, hypoplastic sacrum.⁹ Incomplete form of lumbo-costo vertebral syndrome shows congenital hernia, spinal anomalies with no rib abnormalities.¹⁰ Less than 50 cases of congenital lumbar hernia associated with other congenital anomalies have been reported in English literature.¹¹ Predisposition of lumbar hernia in superior triangle is due to multitude of factors like length and angulation of rib, size of the quadratus lumborum, serratus

posterior muscles, the insertion of latissimus dorsi between 11th and 12th ribs, variable insertion of external oblique muscle above the 12th rib.¹¹ Due to absence of 11th and 12th rib, the involved adjacent musculature is attenuated with weakened attachments. Hence congenital lumbar hernia occurs in superior lumbar triangle in lumbo-costo vertebral syndrome.¹¹ Other abnormalities associated are congenital club foot, arthrogryposis.¹¹

CONCLUSION

Congenital lumbar hernia is rare and occurs in superior lumbar triangle. It is often associated with spinal and rib anomalies in lumbo-costo-vertebral syndrome. Incomplete form of lumbo-costo vertebral syndrome shows congenital lumbar hernia, spinal anomalies with no rib abnormalities. Other associated congenital anomalies may be seen.

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