

Case Report	
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Title	Congenital Lumbar Hernia in an Infant with Phocomelia – An Unusual Presentation
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1) Keywords:

Lumbar hernia, Phocomelia. Meshplasty

- 2) **Abstract:** This case report presents a rare instance of congenital lumbar hernia in an infant diagnosed with phocomelia and bilateral congenital talipes equinovarus (CTEV). The patient, an 11-month-old female, exhibited progressive swelling in the left flank region from birth, necessitating surgical intervention. A comprehensive analysis of her condition, surgical approach, and literature on similar cases is documented, contributing to the understanding of congenital lumbar hernias.

Introduction: Congenital lumbar hernia is an uncommon anatomical defect occurring when the abdominal contents protrude through a defect in the lumbar region, potentially leading to significant clinical complications if left untreated [1]. Phocomelia, characterized by limb malformations with varying degrees of severity, adds complexity to the presentation and management of such hernias. This case illustrates the interplay between these two conditions and the resultant implications for surgical repair, postoperative care, and long-term recovery in affected infants.

Case Presentation: An 11-month-old female child, a known case of phocomelia and bilateral CTEV, presented with a noticeable mass in the left flank present since birth. The mass increased in size, particularly evident during crying episodes (Figure 1,2). Born at 37 weeks to a 21-year-old mother via normal vaginal delivery, the infant required no resuscitation at birth, being the first child from non-consanguineous parents. Physical examination revealed an 8×6 cm swelling in the left lumbar region, which was soft, reducible, non-tender with positive cough impulse. Systemic examination remained otherwise unremarkable. Radiological assessment including chest and abdominal X-rays & Ultrasonography of the abdomen indicated an abdominal wall defect in the right lumbar region, herniation of bowel loops, rib anomalies, and mild scoliosis (Figure 3). Patient was subjected to surgical intervention. The patient was positioned in the right lateral decubitus position. A transverse incision was made over the left lumbar region, revealing a hernial sac in the inferior lumbar triangle of Petit containing bowel loops, which were subsequently reduced (Figure 4). The edges of the defect measured approximately 4×2 cm was primarily closed. Overlying muscle layer were approximated and a 6×4 cm polypropylene mesh was placed

over it. Layered closure was performed. The post-operative recovery was uneventful, and the patient was discharged on post-operative day 4, meeting all recovery milestones.

Images:



FIGURE 1: Patient with phocomelia with CTEV with left lumbar hernia.



FIGURE 2: Patient having left lumbar hernia depicted with a lumbar bulge.



FIGURE 3: X ray abdomen erect suggestive of a left lumbar bulge (suspected hernia)

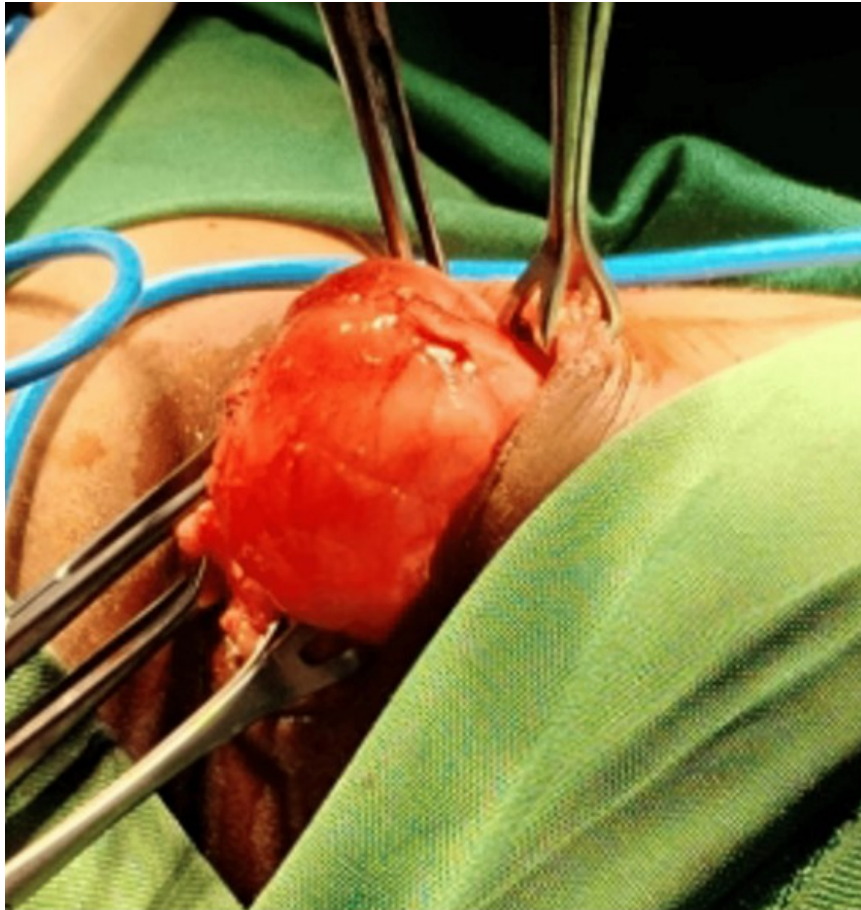


FIGURE 4: Intra-op image showing a large sac of left inferior lumbar hernia

Discussion: The management of congenital lumbar hernias, particularly in the presence of associated anomalies such as phocomelia, requires a meticulous surgical technique to ensure optimal outcomes. Patient had inferior lumbar hernia which is a rare presentation occurring due to somatic mutation during embryogenesis. When coupled with phocomelia puts forth both diagnostic and management challenges due to altered anatomy in the infant. The transverse incision approach utilized in this case is consistent with the recommendations put forth in the literature, where it has been shown to provide adequate exposure while minimizing tissue trauma. Additionally, transverse incisions facilitate better visualization of the hernia sac and surrounding structures, which is crucial in cases of inferior lumbar hernia coupled with anatomical variations like phocomelia [1]. In cases of inferior lumbar hernias, particularly in infants, the preservation of surrounding vascular and neural tissues during repair is imperative to minimize postoperative complications [2]. The incidence of congenital lumbar hernias is rare, with an estimated cases reported till date are 18 [3]. To the best of our

knowledge, a combination of phocomelia with bilateral CTEV with unilateral inferior lumbar hernia has not been reported yet. Additionally, advanced radiological investigation may provide useful anterior abdominal wall musculature information, but accessibility with affordability poses a challenge in a developing nation. However, pre-operative ultrasonographical assessment, which is readily available, in the hands of an experienced radiologist gives good details, helps planning, as in our case. The literature highlights the importance of early surgical intervention to reduce the risk of incarceration and strangulation of the herniated contents, which aligns with our case where the hernia was present since birth and increased in size during crying episodes. Records have shown that surgical repair performed within the first year of life has a high success rate, particularly when performed within the first six months [4]. Conversely, delayed surgical intervention is associated with significantly higher morbidity [4]. Our decision to proceed with surgical repair at 11 months follows the recommendation, emphasizing timely intervention 4 of 5 in the setting of evident symptoms and growth of the hernia [4]. Also, considering the large defect of inferior lumbar hernia in our case, prevention of complications was a mainstay thus meshplasty was advocated to close the defect. Postoperative care in infants with congenital lumbar hernias must be tailored to their specific needs, especially in patients with phocomelia, where mobility and overall recovery can be impacted by associated limb anomalies. The literature advises a multidisciplinary approach in postoperative care, focusing not only on wound healing and infection prevention but also on rehabilitation to promote mobility [5]. In the analysis of outcomes following surgical repair, a successful long-term recovery is expected with appropriate postoperative management, including physical therapy and follow-up assessments [5]. Such data supports our commitment to rigorous follow-up for this infant to monitor her recovery trajectory. The interaction between congenital lumbar hernias and phocomelia is relatively underexplored in existing literature. However, the surgical outcomes are greatly improved with tailored rehabilitation strategies to address the multifaceted challenges posed by limb malformations [5]. This emphasizes the importance of a collaborative care model involving paediatric surgeons, physical therapists, and occupational therapists.

Conclusions: Congenital lumbar hernia, although rare, is a known entity. Solitary management of the same is doable. However, when associated with anomalies like phocomelia with bilateral CTEV, it poses a challenge due to muscular weakness noted due to somatic mutations, thus posing a challenge as they may require hernioplasty and not herniorrhaphy. This case reinforces the need for ongoing research and review of surgical strategies for congenital lumbar hernias, particularly in the context of associated anomalies like phocomelia. Continued documentation of such cases will enrich the surgical literature, enhancing our understanding and bolstering evidence-based approaches in the management of these complex conditions.

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