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CASE REPORT

Early prenatal diagnosis of a lumbo-costo-vertebral syndrome

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Abstract Lumbo-costo-vertebral syndrome (LCVS) is a rare type of lumbar hernia with associated abnormalities of the vertebral bodies, ribs, and trunk muscles. Only a few cases have been reported in the literature, all of which were diagnosed after birth. We present a case of LCVS diagnosed early in the second trimester of pregnancy using two- and three-dimensional ultrasound. In our case, the associated anomalies were: multiple costovertebral anomalies, lumbar hernia, anal imperforation, left hand supernumerary digit, and clubfoot.

Keywords Lumbo-costo-vertebral syndrome · Congenital lumbar hernia · Ultrasound · Prenatal diagnosis

Introduction

Congenital lumbar hernia is a rare type of hernia. Association of congenital lumbar hernia with anomalies of the ribs, spine, and muscles constitutes lumbo-costo-vertebral syndrome. No more than 30 cases of LCVS have been published in the literature. All reported cases were diagnosed in childhood or even in the adult period. We present a case of LCVS diagnosed during early pregnancy, which is the first case reported prenatally to the best of our knowledge.

Case

During the first trimester ultrasound screening, a 28-year-old patient (gravida 0 para 0) being treated for primary infertility was found to have an increased fetal nuchal translucency (3.04 mm) and a subsequent elevated first trimester risk. The first trimester-combined screen indicated a low risk for 21, 18, and 13 trisomies. The 16-week scan revealed a mass protruding through the abdominal wall in the left lumbo-costal region containing small bowel. Bi-dimensional scan and three-dimensional examination showed absence of lower ribs on the left side, scoliosis, and spinal disruption (Figs. 1, 2). Other detected anomalies were agenesis of the left kidney and dysplasia of the right kidney (Fig. 3). The patient and her family were counseled regarding the prognosis of their child based on the cases in the literature. Surgical repair should begin early in childhood, and may involve simple closure in the case of small defects or even prosthetic materials for large defects. Despite the possibility of a good prognosis, understanding the potential complications, the patient chose to end the pregnancy. The pathological exam confirmed the ultrasound abnormalities and also revealed the presence of anal imperforation, left hand supernumerary digits, and left clubfoot (Fig. 4). One year later the patient got pregnant again and delivered a healthy 3100-g term newborn.

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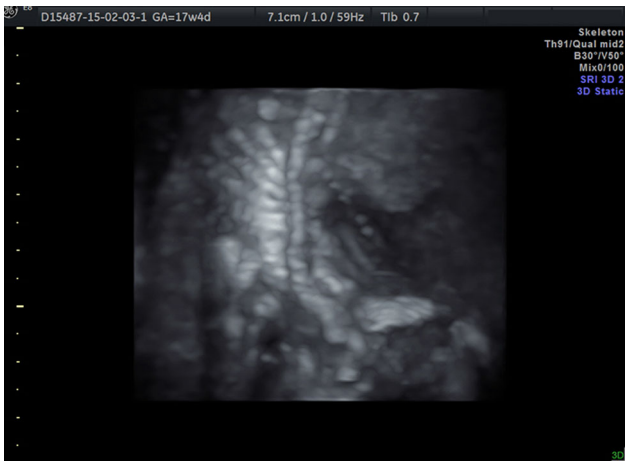

Fig. 1 Three-dimensional ultrasound: left side absent ribs and scoliosis (skeletal mode)

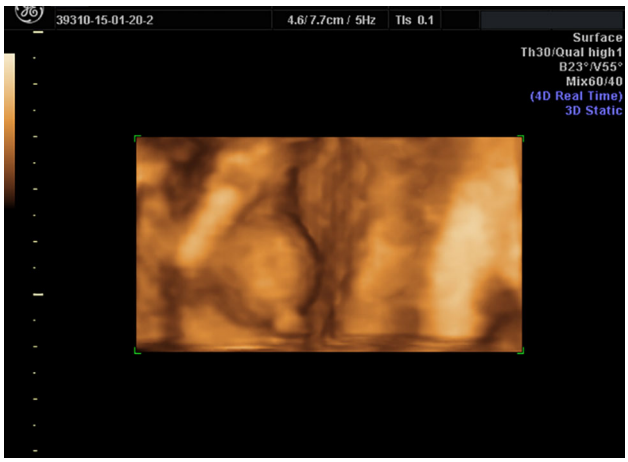

Fig. 2 Three-dimensional ultrasound: lower chest and upper abdominal bulge (surface mode)

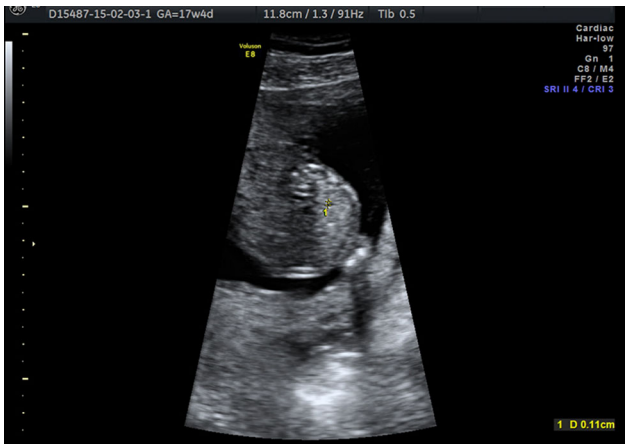

Fig. 3 Two-dimensional ultrasound image: hyperechogenic unilateral kidney (axial section)

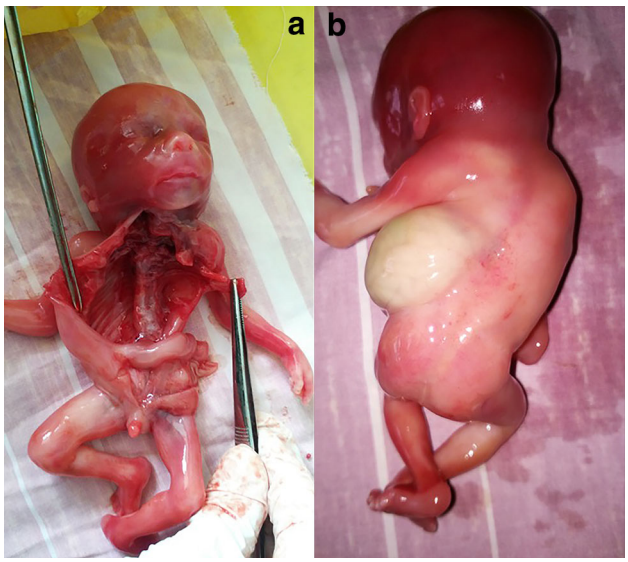

Fig. 4 Clinical picture showing the left lumbar hernia. **a** Costo-vertebral anomalies, scoliosis, and absence of the left ribs. **b** Hernia apparently in left lumbar region

Discussion

Congenital lumbar hernias are rare entities that may occur anywhere in the lumbar region between the 12th rib and the iliac crest. The parietal defect usually appears in the superior lumbar triangle of Grynfelt (located between ribs and quadratus lumborum and internal oblique muscles), and less often in the inferior triangle of JL Petit (which is more superficial) [1]. The location of the hernia in this area explains the frequent association of the costal agenesia [2].

The association of lumbar hernia and anomalies of the ribs, spine, and muscles constitute lumbo-costo-vertebral syndrome, which is one of the most rare entities. The first case of LCVS syndrome was published by Touloukian in 1972 [3]. Since then there have been approximately 30 reported cases with LCVS in the literature. All the cases reported to date were diagnosed after birth. Our case is the first one diagnosed during pregnancy using 2D and 3D ultrasound.

Currently, the etiology of this condition is unknown. No genetic or molecular causes have been identified. Although the exact etiology is not known, Touloukian [3] postulated that the LCVS anomalies could be due to a single somatic defect occurring during 3–5 weeks of gestational age, resulting in malformations of abdominal musculature, ribs, and vertebral bodies. Between 3 and 5 weeks of gestational age, the mesoderm differentiates into somites, which further differentiate into the sclerotome (that forms the vertebrae and costal process), the myotome (that forms the skeletal muscles of the trunk), and the dermatome (which contributes to the deep layers of the skin and the

subcutaneous tissue). Any disturbance, such as anoxia, during this stage could result in vertebral, costal, and parietal wall defects [1]. The vertebral defects are represented by hemivertebrae and posterior spinal dysraphism. The rib anomalies range from hypoplasia to agenesis of local ribs and bilateral rib defects.

In addition to costal, vertebral, and parietal defects, a multitude of other organ anomalies can appear randomly according to cases published in the literature. The associated malformations described are limb defects (arthrogryposis, clubfoot), genitourinary anomalies (undescended testis, renal agenesis, ureteropelvic junction obstruction), cardiac atrial septal defect (ASD), caudal regression anomalies, cloacal exstrophy, anorectal malformation, abdominoschisis, diaphragmatic hernia, focal nodular hypoplasia of liver, and neural tube defects [1, 4–7]. There are reports of cases associated with neurofibromatosis type I and neuroblastoma, suggesting that neural involvement (neuropraxia) may be the underlying lesion in muscle weakness and lumbar hernia defect [8, 9]. In our case, we have along with parietal defect, scoliosis, absent lower ribs, and hemivertebrae, several other anomalies: anal imperforation, right renal agenesia, clubfoot, and supernumerary digits.

Because of these possible associations, cases diagnosed with LCVS require an extensive workup to detect and diagnose various other congenital anomalies. For prenatal diagnosis, modern imaging techniques such as 3D ultrasound and HDlive can be useful, especially for making the differential diagnosis with other fetal anomalies [10–12]. The differential diagnosis includes gastroschisis, omphalocele, limb body wall complex, and Prune belly syndrome. In our case, both 2D and 3D ultrasound proved to be useful for the diagnosis.

If the diagnosis is established after birth (such as in all cases published in the literature), contrast-enhanced CT scan of the abdomen along with plain radiology helps in the detection of all the associated anomalies of the other organ systems.

The prognosis of LCVS depends on the associated malformations. Early elective surgery is the treatment of choice and is recommended before 12 months of age because the hernial defect may enlarge with growth, making primary closure with surrounding tissue difficult. Surgical repair with local tissues is preferable and generally possible [2, 4]. When the size of the defect is large and there is extensive muscular hypoplasia, there is a need for prosthetic material [2, 4].

Conclusion

LCVS is a rare syndrome usually diagnosed after birth and during childhood. We discovered the anomaly early in the second trimester of pregnancy. Our finding emphasizes the importance of the ultrasound scan because in this way we can avoid pathologies with long-term morbidity.

Compliance with ethical standards

Conflict of interest No author has any potential conflicts of interest.

Ethical statements All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1964 and later versions. Informed consent was obtained from the patient for being included in the study.

References

1. Fakhry SM, Azizkhan RG. Observations and current operative management of congenital lumbar hernias during infancy. Surg Gynecol Obstet. 1991;172:475–9.
2. Stamatiou D, Skandalakis JE, Skandalakis LJ, et al. Lumbar hernia: surgical anatomy, embryology, and technique of repair. Am Surg. 2009;75:202–7.
3. Touloukian RJ. The lumbocostovertebral syndrome: a single somatic defect. Surgery. 1972;17:181.
4. Akcora B, Temiz A, Babayigit C. A different type of congenital lumbar hernia associated with lumbocostovertebral syndrome. J Pediatr Surg. 2008;43:e21–3.
5. Krishnamurthy S, Kapoor S. An incomplete form of lumbocostovertebral syndrome in association with atrial septal defect, arthrogryposis and clubfeet. Indian J Pediatr. 2009;76:411–3.
6. Harris K, Dorn C, Bloom B. Lumbocostovertebral syndrome with associated VACTERL anomalad: a neonatal case report. J Perinatol. 2009;29:826–7.
7. Al-Salem AH, Abu-Srair H, Qaissaruddin S. Focal nodular hypoplasia of the liver with the lumbo-costovertebral syndrome. J Pediatr Surg. 1996;31:1282–4.
8. Rimmelin A, Dias P, Salatino S, et al. Colonic lumbar hernia secondary to congenital bone anomalies in a case of neurofibromatosis type I. J Radiol. 1996;77:279–81.
9. Lafer DJ. Neuroblastoma and lumbar hernia: a causal relationship? J Pediatr Surg. 1994;29:926–9.
10. Grigore M, Iliev G. Diagnosis of sacrococcygeal teratoma using two and three-dimensional ultrasonography: two cases reported and a literature review. Med Ultrason. 2014;16:274–7.
11. Grigore M, Iliev G, Gafitanu D, et al. The fetal abdominal wall defects using 2D and 3D ultrasound. Pictorial essay. Med Ultrason. 2012;14:341–7.
12. Grigore M, Mares A. The role of HDlive technology in improving the quality of obstetrical images. Med Ultrason. 2013;15:209–14.