

Limb Body Wall Complex

Sonography Case Study

Introduction

Limb body wall complex is a congenital disorder defined by many internal abnormalities.¹ Some of these abnormalities include abdominoschisis, scoliosis, facial cleft, thoracic wall defect, ectopia cordis, or limb abnormalities such as clubfoot, absent limbs, bradydactyly, and polydactyly. The congenital disorder can be seen in fetal ultrasounds, and is most commonly recognized if the fetus has limb defects or thoracoabdominoschisis with gastroschisis.²

Classifications

Van Allen et al. was one of the first to notice commonalities in the fetal defects in 1987 and proposed the idea that to diagnose an infant with limb body wall complex, it must have two of the three main abnormalities: exencephaly or encephalocele with facial clefts, thoraco and or abdominoschisis, and limb defects. The diagnosis of limb body wall complex today still involves these three main abnormalities.³⁻⁵

Russo et al. later developed two apparent phenotypes in 1993. Phenotype I included the facial clefts, amniotic adhesions, craniofacial defects and amniotic band syndrome, whereas Phenotype II had imperforated anus, lumbosacral meningocele, kyphoscoliosis, placental anomalies, urogenital anomalies, but no craniofacial defects.^{1,5,6} In 2007, these phenotypes were revised and Sahinoglu et al. developed and proposed new classifications. In Type I, the fetus would have an intact thoracoabdominal wall, normal placenta and umbilical cord but would have craniofacial defects, similar to Russo's Type I. Included in Sahinoglu's Type II, the fetus would have supraumbilical, thoracoabdominal wall defect with the herniation of the organs, covered by the amniotic sheet that connects to the skin margin, and deformed umbilical cord with normal cloacal parts. Type III is characterized by infraumbilical abdominal wall defects with the placenta being attached to the skin at the site of the defect. Similar to Type II, there is herniation of the organs, but into the extraembryonic coelom cavity. Type III also has cloacal structure malformation or no cloacal structures present in the fetus at all.^{1,5}



Figure 1.

Image shows fetus one with the liver on the outside of the abdominal cavity.⁷



Figure 2.

Image demonstrates fetus one in a "Buddha" position indicating abnormal lower limbs with caudal regression.⁷



Figure 3.

Image shows fetus two with bowel and liver on the outside of the abdominal cavity.⁷



Figure 4.

Image shows fetus two's pelvis with the absent of lower limbs.⁷

Case Study

A female in her late 20's presented to a clinic for a follow up exam in regards to her pregnancy, in which the first clinic was questioning the possibility of conjoined twins. The pregnancy for the woman was unplanned and she was taking lithium at the time of her conception.

The ultrasound exam revealed two fetuses present that were not conjoined. The gestational age of the fetuses measured 12 weeks and both presented with multiple anomalies. The scan showed both fetuses with normal heart rates and normal head anatomy for their gestational age. However, fetus one had the heart and liver located on the outside of the body, (See Figure 1) thick nuchal translucency, and an abnormal kyphotic spine. Fetus one also appeared to be in a potential Buddha pose indicating abnormal legs with caudal regression (See Figure 2). Likewise fetus two had the heart and liver on the outside of the body and an abnormal spine, but presented with no lower extremities (See Figures 3 & 4).

Conclusion

Limb body wall complex is a congenital disorder characterized by many physical abnormalities. The main criteria used to diagnose this disorder includes the fetus having two of the three abnormalities: exencephaly or encephalocele with facial clefts, thoraco and/ or abdominoschisis, and limb defects. The thoracic and abdominal wall defects need to be looked at with careful consideration as they may have no association with limb body wall complex and have a higher prognosis. Limb body wall complex is classified by the extent of the abnormalities present and is subdivided into phenotypes based on the anatomy affected. Ultrasound can be used in the early detection of the physical abnormalities and helps create a more definitive diagnosis, and can be confirmed with a postpartum physical examination using the assistance radiographic images.

References

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