

Limb Body Wall Complex

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Abstract: Limb body wall complex, also known as body stalk anomaly, is characterized by a combination of developmental abnormalities involving neural tube, body wall and limbs. Most of them are diagnosed by ultrasonography in the prenatal period and are not continued till birth. Usually it has a normal karyotype but body stalk anomaly may also be associated with trisomy 16[1]. The chance of recurrence is very low in subsequent pregnancies. There is no correlation with parents' age or fetal gender.

Key Words: Ultrasound, Gastroschisis, limb body wall complex, abdominal defects.

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I. Introduction:

Limb body wall complex is a rare congenital anomaly. Incidence is between 1 in 14,000 to 1 per 31,000 pregnancies[2] [3].

Limb body wall complex is due to the breakdown of ectodermal placode and subsequently involves the early embryonic folding process. The three essential characteristic features of body stalk anomaly includes :

- encephalocele or exencephaly or facial clefts.
- Thoraco or abdominoschisis.
- Limb deformity.

Other abnormalities usually associated includes spinal cord abnormalities, abnormalities of umbilical cord and membranes and persistent extra embryonic coelomic cavity. Etiology of body stalk anomaly is not clear. It is believed to be a severe form of amniotic band syndrome.

II. Case Report:

A 24 year old primi came with history of 5 months amenorrhea and complaints of foul smelling white discharge per vaginum. No history of consanguineous marriage or any drug intake. Dating scan was not done. Her gestational age according to LMP was 21 weeks and 4 days. According to USG her gestational age was 18 weeks and 5 days. Femur length was 2.96 cm corresponding to 19 weeks.

Ultrasound study of the fetus showed following abnormalities.

- Thorax appears narrow occupied completely by heart. Heart showed occasional bradycardia.
- Lungs not imaged clearly.
- Echogenic structure measuring ~ 1.8 x 1.9 cm noted anterior to the abdomen and superior to the umbilical cord attachment site – gastroschisis[13].(Fig. 1.1) (Fig. 1.2)
- Absent right lower limb.
- Rocker bottom foot noted in left lower limb.(Fig. 1.3) (Fig. 1.4)
- Deformed lower dorsal, lumbar and sacral spine[12] (Fig. 1.5)

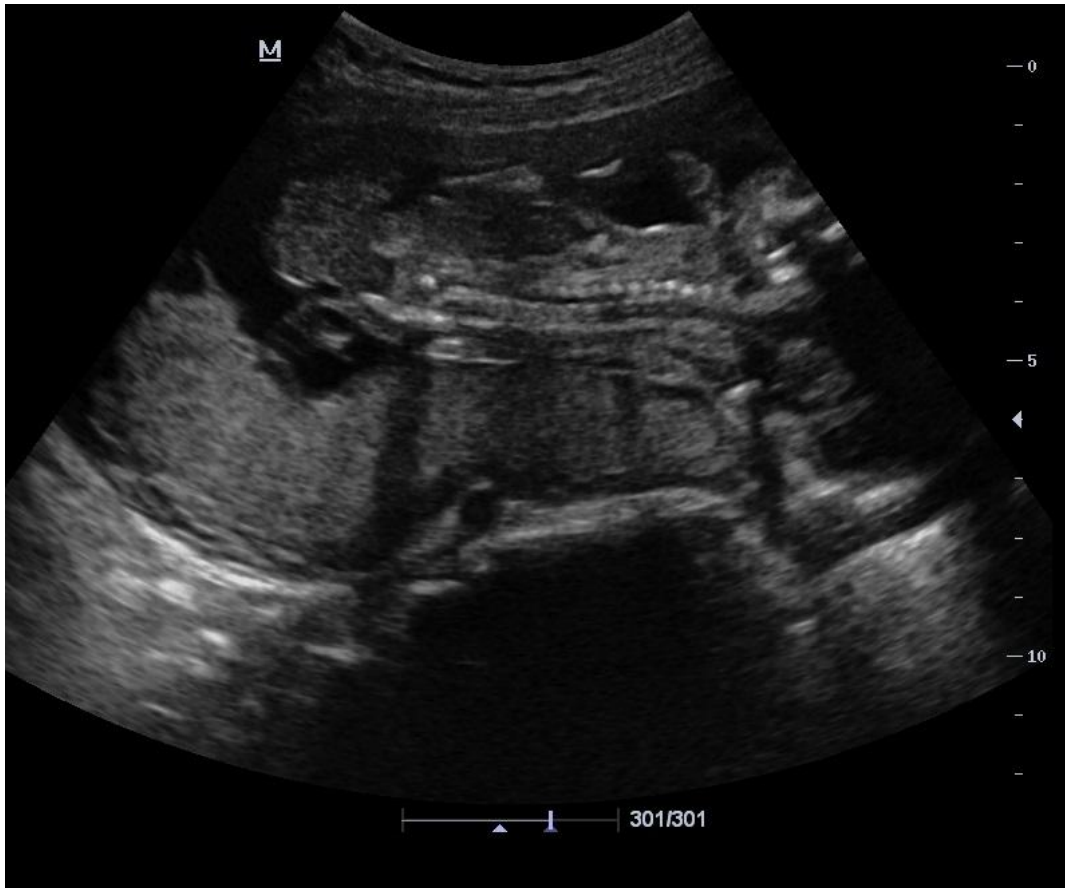


Fig. 1.1 Gastroschisis



Fig. 1.2 Gastroschisis

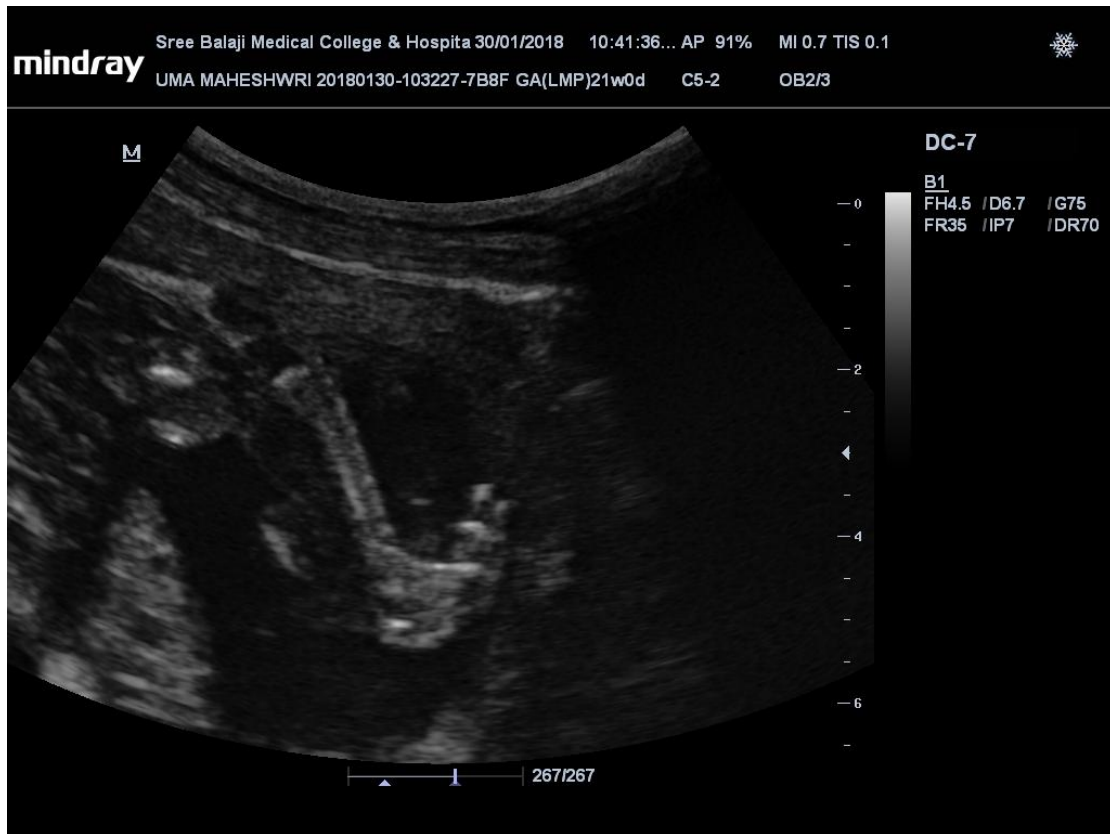


Fig. 1.3 Rocker bottom foot



Fig. 1.4 Rocker bottom foot



Fig. 1.5 Deformed lower dorsal, lumbar and sacral spine

The mother was counseled to terminate the pregnancy after Ultrasonography.

Following features were noted in the still born:

- Umbilical cord was short. (Fig. 2.1)
- Exencephaly.(Fig. 2.1)
- Large midline defect of anterior abdominal wall with bowel loops and liver herniating through the abdominal wall defect – Gastroschisis. (Fig. 2.2)
- Baby was kyphotic.
- Absence of right lower limb. (Fig. 2.2)
- Rocker bottom foot in left lower limb. (Fig. 2.3)

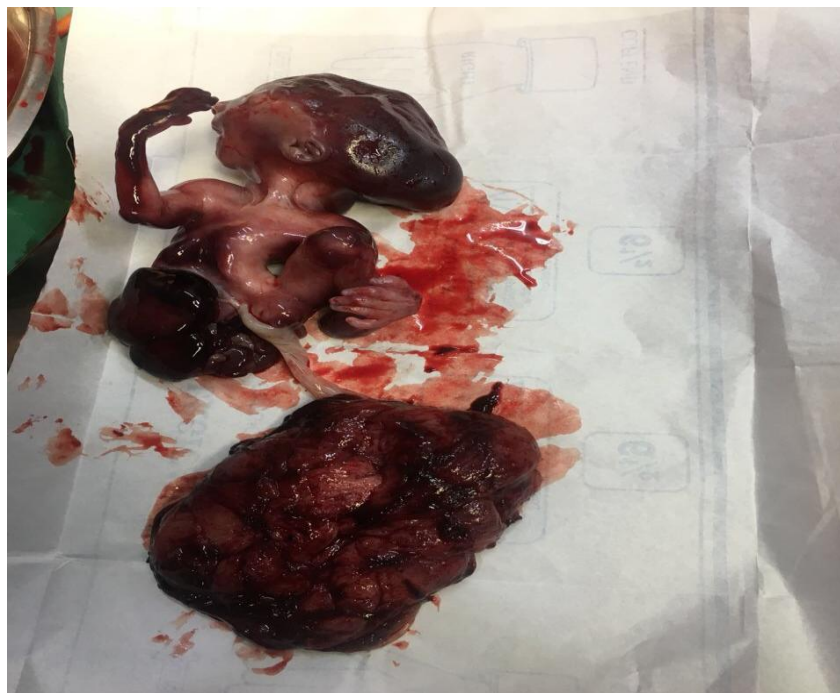


Fig. 2.1 short umbilical cord and exencephaly



Fig. 2.2 Gastroschisis and absent right lower limb



Fig. 2.3 Rocker bottom left foot

X – RAY of the fetus showed: (Fig : 3.1 – 3.3)

- Kyphosis of spine
- Left rocker bottom foot.
- Absent right lower lim.
- Lung hypoplasia.
- Extravasation of liver and intestine.
- Soft tissue swelling noted in the occipital region of skull.

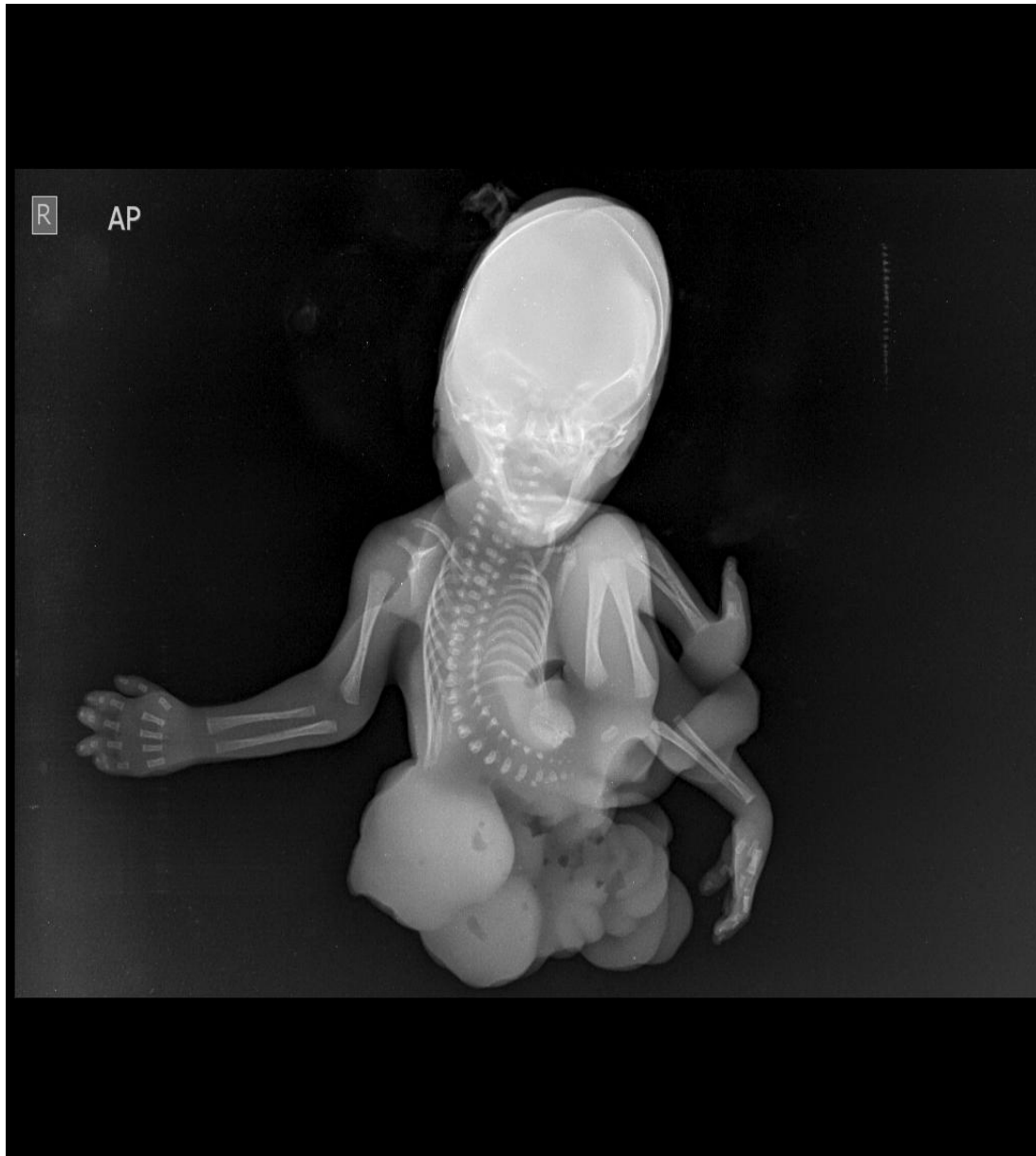


Fig. 3.1



Fig. 3.2



Fig. 3.3

MRI of the Fetus. (Fig : 4.1 – 4.4)

- Lower half of the body is not normally developed
- Liver , spleen and almost the entire bowel loops are seen outside the peritoneal cavity.
- Dorso-lumbar spine is seen curved with convexity to right side.
- Umbilical cord is seen attached to the lower end of the poorly developed lower half of the body.
- Only left lower limb is seen.
- Left ileum is seen high placed just below the costal margin.
- Left hip is in fixed flexion. Left femur, tibia and fibula are normal.
- Rocker bottom appearance of left foot.
- Long and short bones of both upper limbs are normal.
- Spalding sign is seen in skull, diffuse edema involving the fetal cranium and body seen.
- Skull shows overriding of bones.

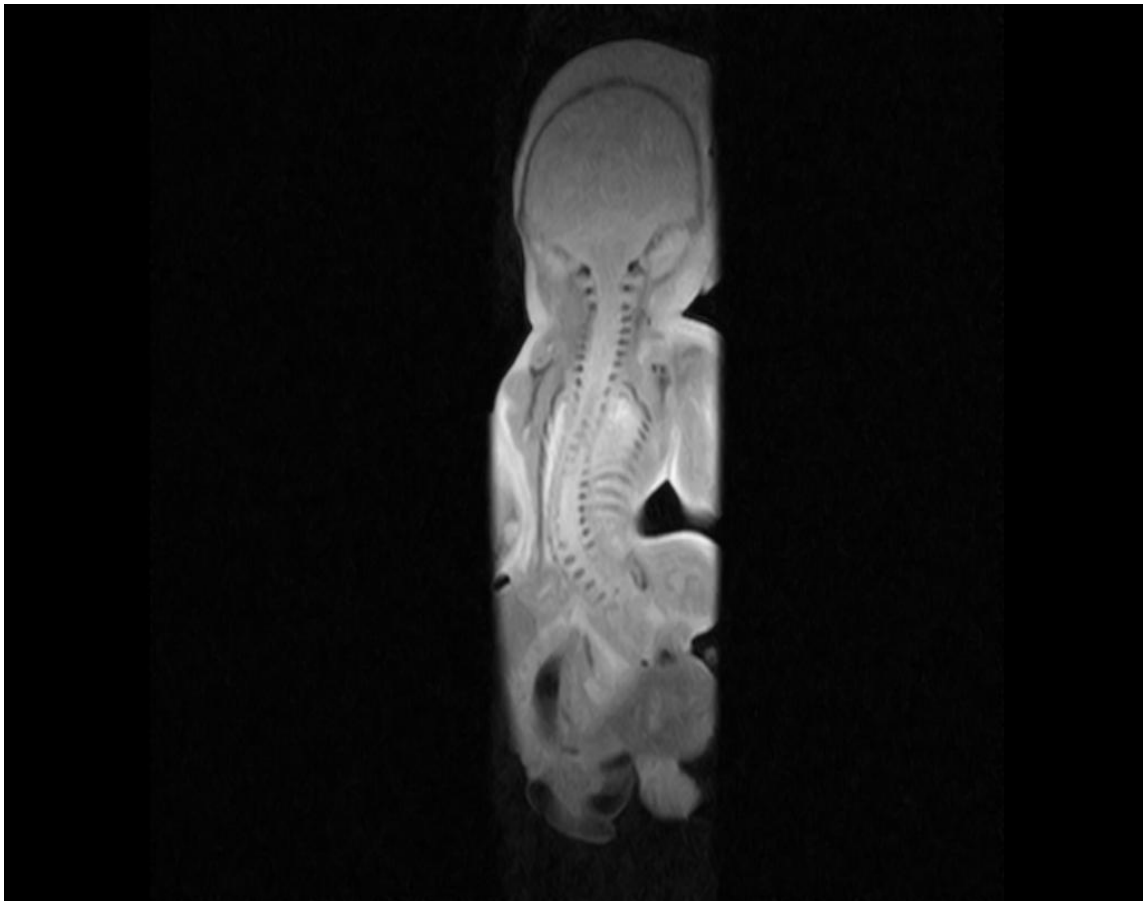


Fig. 4.1

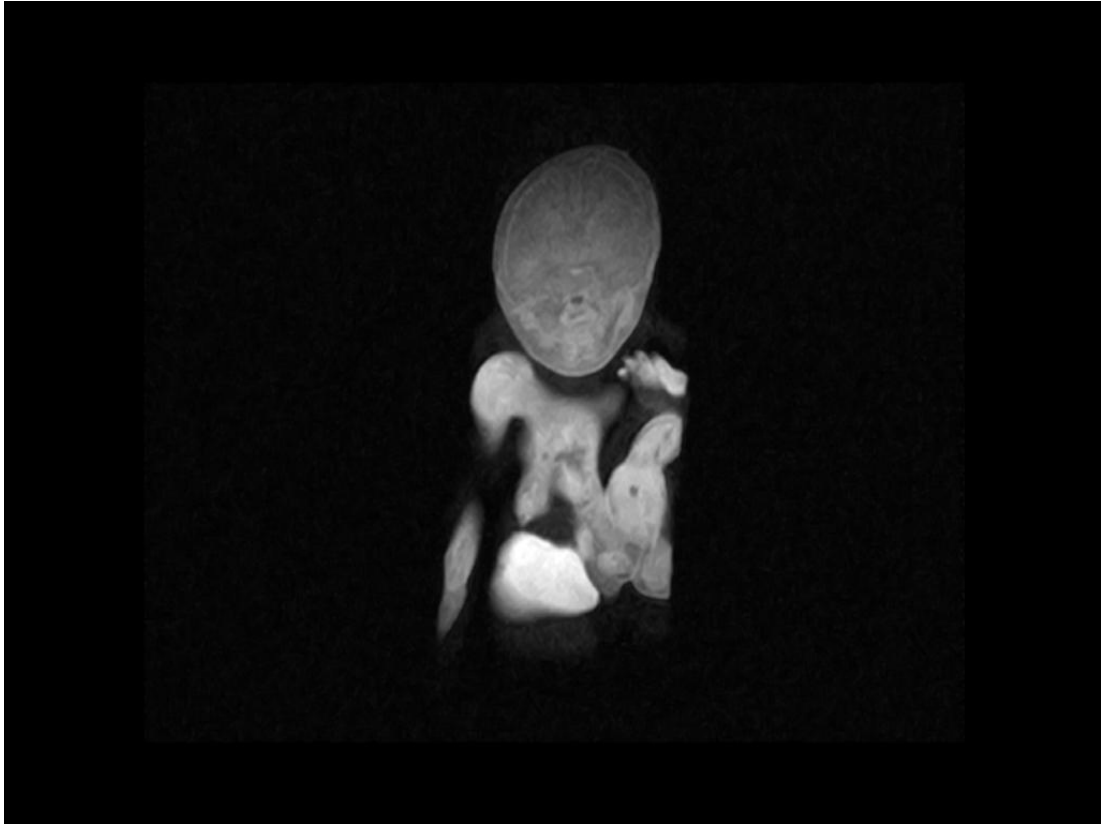


Fig. 4.2



Fig. 4.3



Fig. 4.4

III. Discussion:

The theories to prove Limb body wall complex include:

- 1) Early rupture of amnion[4],[5].
- 2) Vascular disruption theory[6].
- 3) Embryonic mal-development[7]

Early rupture of amnion: The foremost and the most accepted theory is an early rupture of the amnion before there is an obliteration of the coelomic cavity. It states that the primary rupture of the amnion induces the creation of fibrous bands from the chorionic surface, which entraps the fetal body parts.

Vascular disruption theory: Vascular disruption theory includes events that has negative influence on normal embryonic blood supply thereby disrupting the normal morphogenesis. Vascular disruption theory is supported by the evidence that cocaine use causes body stalk anomaly.

Embryonic mal-development: Limb body wall complex occurs from complete failure of body folding along all three axes (cephalic, caudal, and lateral). Mal-development of each of the four folds results in a distinct group of fetal structural abnormalities[10],[11]. Abnormal cephalic folding produces a form of pentalogy of Cantrell, lateral fold defects causes in omphalocele, and aberrant caudal folding may create abnormalities of cloacal exstrophy. Limb body wall complex is supposed to be due to faulty folding in all three axes with persistence of the extra-embryonic coelomic cavity. The various malformations associated with body stalk anomaly depend on the degree of aberrant development of each of the four folds.

Two main phenotypes of body stalk anomaly have been described:

- Fetuses with craniofacial defect: they show two specific characters - a) Encephalocele or exencephaly with facial clefts. b) amniotic bands or amniotic adhesions between cranial defects and placenta.
- Fetuses without craniofacial defect: they often present with urogenital abnormalities, anal atresia and lumbosacral abnormalities[7],[8].

Treatment and prognosis:

Limb body wall complex has extremely poor prognosis and is invariably fatal [14] [15]. Management is often supportive. Future pregnancies are however not thought to carry an increased risk of redeveloping the condition.

IV. Conclusion:

Body stalk deformity is a congenital condition causing multiple fetal abnormalities. The typical features can be detected ultrasonographically by the end of first trimester. Prenatal diagnosis of this anomaly would permit the early termination of pregnancy or avoidance of surgical intervention.

References:

- [1]. Y. Chan, N. Silverman, L. Jackson, R. Wapner, and R. Wallerstein, "Maternal uniparental disomy of chromosome 16 and body stalk anomaly," *American Journal of Medical Genetics*, vol. 94, pp. 284–286, 2000.
- [2]. Mann L, Ferguson-Smith MA, Desai K, Gibson AA, Raine PA. Prenatal Assessment of anterior abdominal wall defects and their prognosis. *Prenat Diagn*. 1984;4:427–35. [PubMed]
- [3]. Forrester MB, Merz RD. Epidemiology of abdominal wall defects, Hawaii, 1986-1997. *Teratology*. 1999;60:117–23. [PubMed]
- [4]. R. Torpin, "Amniochorionic mesoblastic fibrous strings and amnionic bands. Associated constricting fetal malformations or fetal death," *American Journal of Obstetrics and Gynecology*, vol. 91, no. 1, pp. 65–75, 1965. View at Publisher · View at Google Scholar · View at Scopus
- [5]. M. C. Higginbottom, K. L. Jones, B. D. Hall, and D. W. Smith, "The amniotic band disruption complex: timing of amniotic rupture and variable spectra of consequent defects," *The Journal of Pediatrics*, vol. 95, no. 4, pp. 544–549, 1979. View at Publisher · View at Google Scholar · View at Scopus
- [6]. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. *Am J Med Genet* 1987; 28:529.
- [7]. Gajzer DC, Hirzel AC, Saigal G, et al. Possible Genetic Origin of Limb-Body Wall Complex. *Fetal Pediatr Pathol* 2015; 34:257.
- [8]. Lockwood CJ, Scioscia AL, Hobbins JC. Congenital absence of the umbilical cord resulting from maldevelopment of embryonic body folding. *Am J Obstet Gynecol*. 1986;155:1049–51. [PubMed]
- [9]. Hartwig NG, Vermeij-Keers C, De Vries HE, Kagie M, Kragt H. Limb body wall malformation complex: An embryologic etiology? *Hum Pathol*. 1989;20:1071–7. [PubMed]
- [10]. Russo R, D'Armiento M, Angrisani P, Vecchione R. Limb body wall complex: A critical review and a nosological proposal. *Am J Med Genet*. 1993;47:893–900. [PubMed]
- [11]. Deruelle P, Hay R, Subtil D, Chauvet MP, Duroy A, Decocq J, et al. Antenatal diagnosis of limb body wall complex. *J Gynecol Obstet Biol Reprod (Paris)* 2000;29:385–91. [PubMed]
- [12]. Keppler-Noreuil K, Gorton S, Foo F, Yankowitz J, Keegan C. Prenatal ascertainment of OEIS complex/cloacal exstrophy - 15 new cases and literature review. *Am J Med Genet A*. 2007;143A:2122–8. [PubMed]
- [13]. Mitanchez D, Walter-Nicolet E, Humblot A, Rousseau V, Revillon Y, Hubert P. Neonatal care in patients with giant omphalocele: Arduous management but favorable outcomes. *J Pediatr Surg*. 2010;45:1727–33.[PubMed]
- [14]. Negishi H, Yaegashi M, Kato EH et-al. Prenatal diagnosis of limb-body wall complex. *J Reprod Med*. 1998;43 (8): 659-64
- [15]. Emanuel PG, Garcia GI, Angtuaco TL. Prenatal detection of anterior abdominal wall defects with US. *Radiographics*. 1995;15 (3): 517-30

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