OLGU SUNUMU CASE REPORT

Prenatally Diagnosed Limb Agenesis in A Case of "Limb Body Wall Complex" At 18th Week of Gestation

Onsekizinci Gebelik Haftasında Prenatal Tanı Konulmuş Bir "Ekstremite Vücut Duvarı Kompleksi" Olgusunda Ekstremite Agenezi

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ABSTRACT A case of gastroschisis and kyphoscoliosis at 18th week of gestation was further investigated and was found that he has lower limb agenesis and described as "limb body wall complex". Prenatal ultrasonography is an important tool to diagnose important syndromes such as "limb body wall defect" which is incompatible with life.

Key Words: Prenatal diagnosis, limb deformities, congenital, gastroschisis

ÖZET Gastroşizis ve kifoskolyoz olan bir olgu onsekizinci gebelik haftasında detaylı incelenerek alt ekstremite agenezine sahip olduğu tespit edilmiş ve "ekstremite vücut duvarı kompleksi" tanımlanmıştır. Prenatal ultrasonografi bunun gibi yaşamla bağdaşmayan anomalilerin tanımlanmasında önemli bir yere sahiptir.

Anahtar Kelimeler Prenatal tanı, konjenital ekstremite deformiteleri, gastroşizis

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The Limb Body Wall Complex (LBWC) is a rare polymalformative syndrome. It is generally defined as consisting of two of the three following abnormalities: a) myelomeningocele and/or caudal regression, b) thoraco- and/or abdominoschisis and c) limb defects.[1] The cause of the limb body wall complex is controversial. There are different theories about this abnormality. The prognosis for infants with limb body wall complex is universally fatal. Herein, we present a case of LBWC detected in early pregnancy via prenatal ultrasonography.

CASE REPORT

A 26-year-old primigravida woman was referred for further investigation at 18 weeks' gestation because of fetal gastroschisis and kyphoscoliosis. A review of the history revealed that the parents were both healthy. The mother did not report any past history of medical problems or recent infectious episodes. In addition, she had no exposure to teratogenic agents such as cocaine, tobacco or alcohol. The family history was also noncontributory.

A level II sonographic examination revealed anterior body wall defect through which whole abdominal organs protrude and floating in the amniotic fluid and also having kyphoscoliosis (Figure 1). Additionally, left lower limb agenesis (Figure 2) and vesical exstrophy were diagnosed. Fetal kidneys and diaphragm could not be visualized. Umbilical cord was consisting of three vessels. Fetal growth was consistent with gestational age and no other fetal abnormality was detected. Amniocentesis revealed a 46, XY karyotype. Under the diagnosis of LBWC parents are informed about this anomaly. Because of the poor prognosis, the couple chose to give up the baby. Termination was undertaken subsequently.

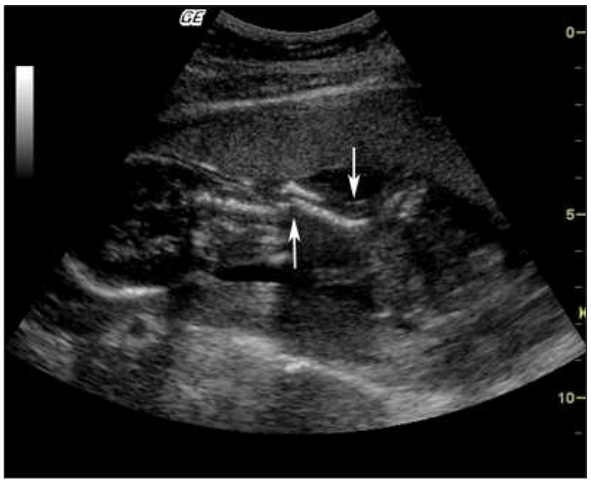

FIGURE 1: Sonographic appearance of limb body wall complex anomaly. Spinal column of the fetus can be observed. There is marked kyphoscoliosis (White arrows).

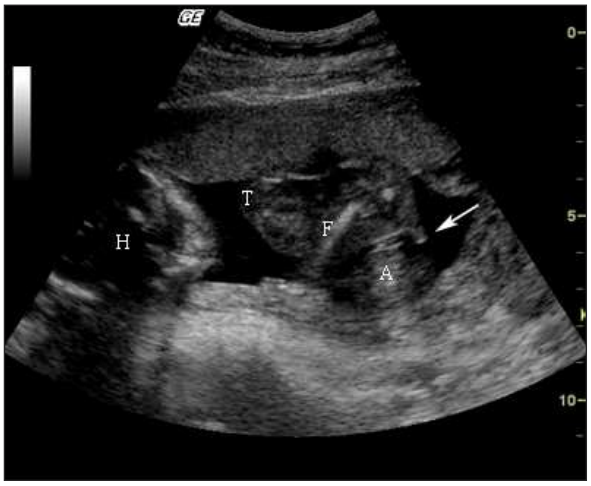

FIGURE 2: Detailed anatomic parts of the fetus can be seen (H: Head, T: Thorax, F: Right femur, A: Abdominal organs; liver and intestines). It is clear that left limb is absent below the pelvic level.

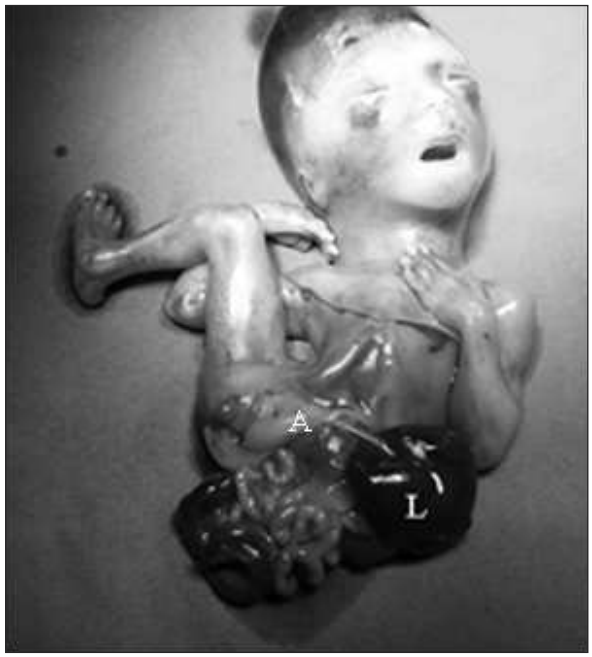

FIGURE 3: Picture of the fetus after abort. Anterior wall defect of fetus through which liver and intestines protrude is seen (A: Absent limb, L: Liver). Also left lower limb is totally absent.

After abortion, diagnosis confirmed as limb body wall complex, including; large anterior abdominal wall defect, kyphoscoliosis, vesical exstrophy and total left lower limb agenesis (Figure 3). Also, umbilical cord was short (12 cm) and had three vessels. An informed consent was taken from the couple about the use of photographs.

DISCUSSION

The incidence of limb body wall complex is 1 in 14.273 live births.[2] The pathogenesis of limb body wall complex is unclear. Three general pathogenic mechanisms have been anticipated for this malformation: amnion rupture, vascular disruption, and embryonic malformation.[3-6] An early rupture of the amnion with mechanical compression between the 3-5th embryonic weeks is most accepted theory.[3,5] The other one is the vascular disruption theory.[3,6] Malformation of the body stalk resulting from a defect in the germ disc leads to an abnormal body folding and amniotic cavity formation and a failure to obliterate the extraembryonic coelom.

This explains the absent or short umbilical cord and broad insertion of the amnio-peritoneal membrane onto the placental chorionic plate.[5]

Limb defects in LBWC are found in 96% of the cases and include club foot (32%), oligodactyly (12%), arthrogryposis/web (12%), absent limb (9%), single forearm bone (8%), single lower leg bone (6%), pseudosyndactyly (5%), split hand/foot (5%), radial/ulnar hypoplasia (4%), rotational defect (4%), and preaxial poly-dactyly (3%).[6] Also internal malformations in LBWC are found in 95% of the cases and contain cardiac anomalies (43%), absent diaphragm (74%), abnormal pulmonary lobulations (50%), gastrointestinal (100%), trilobulated liver (4%), polysplenia (4%), absent gall bladder (29%), renal (65%) and urogenital abnormalities (56%).[7]

Totally absent lower limb seen in our LBWC case, to the best of our knowledge have not been described earlier. Left limb is absent from the level of pelvis and folding of the vertebral column is seen which results the diagnosis of the kyphoscoliosis.

Because the LBW complex is incompatible with life, it is important to diagnose this abnormality prenatally, and to differentiate them from other anterior abdominal wall defects. In our case, the abdominal defect and kyphoscoliosis were initially suspected by another obstetrician, but the nature of the accurate diagnosis had not been made. Therefore, a careful ultrasound survey of fetal limbs, head and face is mandatory when cytogenetic analysis is normal in fetuses with abdominal wall defects.

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