Body stalk anomaly in monozygotic twinning: a case report

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We describe a case of concordant body stalk anomaly in a monozygotic twin. Autopsy of the fetus showed abnormalities compatible with the maldevelopment of embryonic folding. Abdominal viscera were in a sac covered by the amnion and were attached directly to the placenta. The anus was not visible and no discernible external genitalia were noted. Other findings included a neural tube defect and a rectal duplication as an enteric cyst. Umbilical cord had only one vein and an artery. No abnormalities were found on pathologic examination of the placenta.

Although we encountered cases previously with gastroschsis and omphalocele, this was the first case of body stalk anomaly that we recognized as an enteric cyst, which is extremely rare in twins.

Key words: body stalk anomaly, monozygote twinning, embryonic body fold defect.

In early embryogenesis, maldevelopment of the body folding may cause a variety of abdominal defects such as pentalogy of Cantrell, gastroschisis, omphalocele, body stalk anomaly and cloacal extrophy.

The body stalk anomaly associated with agenesis of the umbilical cord or with a very short umbilical cord with multiple abnormalities is characterized by a large and irregular abdominal wall defect[1-3]. Two hypotheses have been postulated to explain this pathology. The first is maldevelopment of body folds when the trilaminal embryo is transformed into a cylindrical embryo, and the second is the syndrome complex resulting from mechanical teratogenesis following rupture of the chorion or yolk sac[3]. Body stalk anomaly in monozygotic twins is quite rare. There have been only three cases reported in the literature[3].

Because of its rarity and the lack of diagnostic criteria, prenatal diagnosis of body stalk anomaly is very difficult; however, there have been antenatally diagnosed cases with ultrasonography (USG)[4].

The body stalk anomaly, which is quite rare and lethal, may be associated with neural tube defects, intestinal atresia, genitourinary and skeletal defects, and chest wall abnormalities[1].

In our autopsy report, we tried to identify the features of the case and took a brief look at the literature.

Case Report

Our case was the second pregnancy of a 28-year-old female. A twin male fetus at 36th gestational week, clinically diagnosed as omphalocele, died within a few minutes after birth. No information was obtained about the prenatal period or family history. The analysis of this karyotype could not be done, nor was there ultrasonographic data concerning the follow-up of the prenatal period.

Autopsy Findings: The fetus had a large abdominal defect through which all the abdominal organs were exposed. The herniated organs were covered with a thin transparent membrane which was attached directly to the placenta without having a distinct border with the placental amniotic sac. The liver, small and large bowel, spleen and stomach were inside the herniated sac (Fig. 1). the anus was obliterated. External genitalia were not identified.

Placenta was diamniotic monochorionic, and had two centrally located umbilical cords, which were close to one another. The umbilical cord of the fetus in our case on to which the amnion was attached was 15 cm long and consisted of an artery and a vein. The other umbilical cord had 3 vessels.

There was marked kyphoscoliosis of the vertebra, and there was a cystic lesion, 15 cm in diameter, in the sacral region (including gluteal area). The lesion was covered by skin, and it was filled with a serous fluid. The interior of the cyst was smooth and the cyst was associated with the vertebral column (spina bifida) via a bone defect 3 cm in diameter (Fig. 2). The lungs were atelectatic. No other pathology was detected in the rest of the organs.

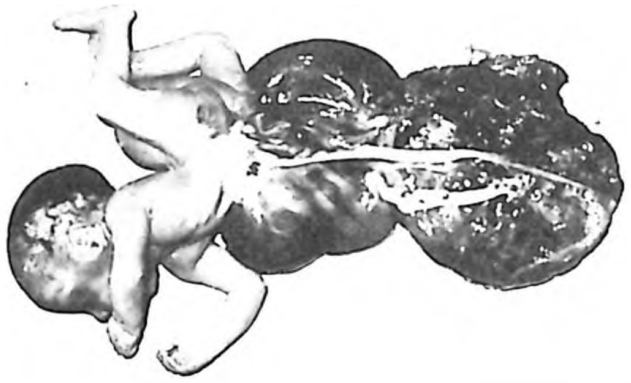

Fig. 1. The herniated organs were covered with a thin transparent membrane which was attached directly to placenta without having a distinct border with placental amniotic sac.

Fig. 2. There was a cystic lesion, 15 cm in diameter, in sacral region (including gluteal area). The lesion was covered by skin, and was filled with a serous fluid.

Discussion

The body stalk is closely involved in the process of lateral folding during the fourth to eighth week of embryonic life. It is derived from the umbilicus and remnant yolk sac and is covered by the amnion. Therefore, any deviation from the normal developmental process may cause severe abdominal wall defects, abnormal development of the hindgut and absence or very severe reduction of the umbilical cord[4].

Although the incidence of abdominal wall defects is said to be higher[1], the incidence of the body stalk anomaly was only six in 252,000. Lakshminarayana et al[2] determined a rate of one in 50,000, and in one other study it was found to be one in 14,273 births. Ours was the only body stalk anomaly previously reported[5].

Body stalk anomaly can be confused with short umbilical cord syndrome. Some believe that the latter is a variant of the stalk anomaly, while other authors say that it is a severe form of amniotic band syndrome, since amniotic bands are present in 40 percent of cases of body stalk anomaly[4].

Sacral vertebral anomalies, such as bifid vertebra in which the defect is large, and hemivertebra in which the defect is smaller, may be associated with hindgut enteric fistulas[6-8].

The cystic lesions of the sacral and coccygeal regions may derive from sequestered intestinal duplication or target remnants, or may occur as a part of the neuroenteric fistula complex. The sacral cysts are usually localized in presacral, intraspinal and postsacral regions; they may be large and multiloculated or multiple and small. The hindgut cyssts are surrounded by varying degrees of muscle layer, whereas tailgut cysts consist of isolated bundles of muscle in which no smooth muscle property is seen[8]. In our case, the cyst was lined by simple cuboidal epithelium and a small portion of the wall was made up of primitive nerve cells and nerve fibers (Fig. 3). For this reason it was accepted as neuroenteric cyst. The results of immunohistochemical stains in epithelial and nerve cells were S-100: positive (Fig. 4) and focal mucin positivity in epithelial cells (Fig. 5).

In our case, the umbilical cord was short and had one artery and a vein, which is not a common feature of body stalk anomaly in monozygotic twins. However, the presence of one artery accompanying some anomalies, such as ventral abdominal defect, has been reported.

Since body stalk anomaly is lethal, it necessitates eliminating omphalocele and gastroschisis in differential diagnosis[9].

In conclusion, although body stalk anomaly is rare, in order to exclude it, it is necessary to

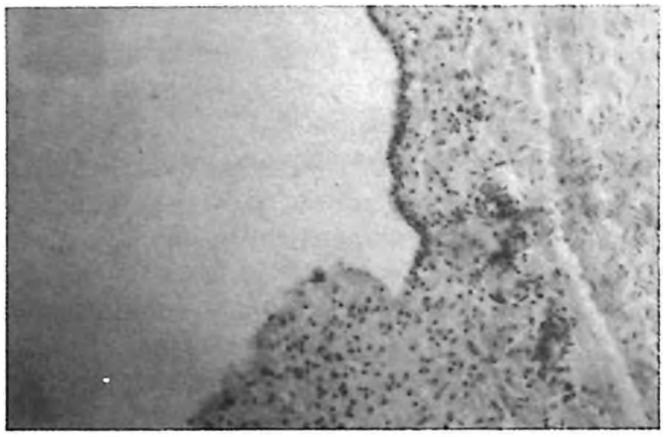

Fig. 3. The cyst was lined by simple cuboidal epithelium and a small portion of the wall was made up of primitive nerve cells and nerve fibers, (H+E X 200).

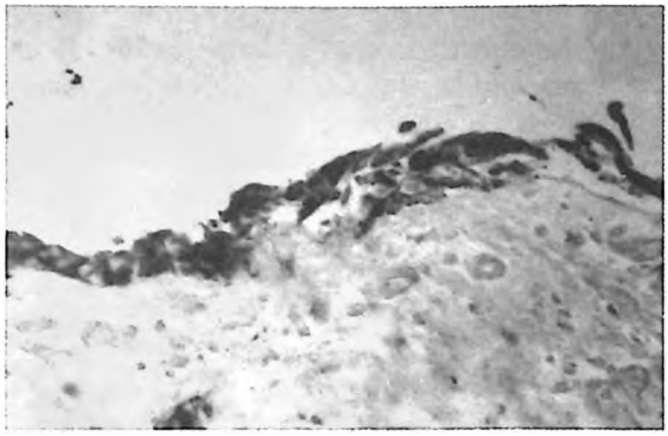

Fig. 4. S-100 positivity in epithelial cells and nerve cells, (S-100 X 400).

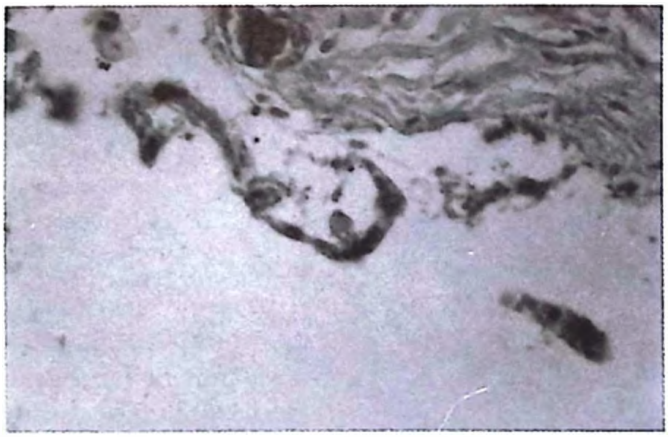

Fig. 5. Focal mucin positivity in epithelial cells, (Alcian Blue X 400).

identify the features of abnormalities in a fetus with an abdominal wall defect, especially omphalocele. The rarity of body stalk anomaly in monozygotic twins and association of neural tube defect and neuroenteric cyst are the particular features of our case.

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