

CASE REPORT

Major omphalocele with liver protrusion. Liver dysmorphism. A case study

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Abstract

In the Renaissance (1634), Ambroise Paré described for the first time the omphalocele, highlighting the serious prognosis of this malformation. The incidence of omphalocele varies between 1/2000–1/6000 births, both sexes being equally affected. We present the case of a male infant, born at term after a pregnancy that was not followed by the family doctor or by the specialist obstetrician and who died after five days. On the anterior abdominal wall in the umbilical region, there was a formation of about 10 cm in diameter, covered by a membrane apparently avascular through whose transparency there could be observed a blue-reddish tumor, of discreet firm consistency that while breathing compressed the apparently avascular membrane that covers it. Intraoperative there was noticed liver herniation by parietal defect, operating the liver reintegration in umbilical cavity, subsequently the child dying from a cardio-respiratory arrest. The particularity of this case is on one hand the liver herniation without intestinal loops, the most common liver hernia being with intestinal loops and, on the other hand the presence of hepatic dysmorphism. We also mention the trilaminar structure of omphalocele membrane sac, which makes us think that the herniation was secondary, after the tenth week, through a defect of the anterior abdominal wall closure in the umbilicus region. The combination of these elements creates the uniqueness of this case.

Keywords: omphalocele, hepatocele, hepatic dysmorphism.

Introduction

In the Renaissance, in 1634, Ambroise Paré described for the first time the omphalocele, highlighting the serious prognosis of this malformation. In the 19th century, Hamilton succeeds in the first surgical healing, and in 1899, Ahlfeld introduces the conservative treatment of omphalocele, applying dressings with alcohol. In 1957, Grob resumes conservative treatment, but this time using mercurochrome dressings. Calder describes for the first time gastroschisis. The incidence of omphalocele is difficult to assess, often being ruptured or confused with gastroschisis. In the U.S.A., the incidence is estimated at 1/2000 births [1], in Colombia 1/4000 births [1] and in the UK the incidence of this malformation is estimated to be one in 6000 births [2]. During 1960–1970, omphalocele was three times more frequent than gastroschisis but the last twenty years gastroschisis is 2–3 times more frequent than omphalocele [1]. The highest ratio of gastroschisis/omphalocele is in Scandinavia, Northern Europe and the United States [3]. Mothers of children with gastroschisis are younger than those of children with omphalocele, and the preterm birth is also more common children in gastroschisis [3]. The rural/urban ratio is generally uniform in all parts of the world [3]. The two sexes are equally affected by this malformation [4, 5]. There have been reports of familial transmission of omphalocele, but these cases remain isolated and could not be genetically demonstrated [6]. Some studies have found the presence of previous miscarriages in mothers of children with omphalocele, but not being able to establish with certainty a direct causal link [7].

Materials, Methods and Results

We present the case of a male infant, born at term after a pregnancy that was not followed by the family doctor or by the specialist obstetrician and who died after five days. Mother had no infections during pregnancy, has not been subjected to medical treatments, did not work in a toxic environment, but has worked strenuously in the field and occasionally consumed alcohol during pregnancy. Fetus of 3000 g and 45 cm long, at birth had poor general state, dyspnea, cyanotic, cold extremities, mottled teguments with an umbilical formation of about 10 cm in diameter, covered by a membrane apparently avascular through whose transparency there could be observed a blue-reddish tumor, of discreet firm consistency that while breathing compressed the apparently avascular membrane that covers it. In the midst of this formation, there is the sectioned and tucked umbilical cord. Omphalocele base of approximately 10 cm. Lung: bilateral present vesicular murmur, no rales. Cardiac: AV=120 beats/min., rhythmic, without souffle. At birth, it presented circular cord, cesarean birth being performed. On the fourth day, the general condition worsened, the tegument becomes cyanotic and "coffee ground" vomiting occurs. During the same day, the general condition gradually worsened and tachycardia occurred (200 beats/min.), tachypnea, with blood pressure dropping. In the fifth day of life, the child has a serious malaise, agitation, "coffee ground" vomiting, cyanotic, cold extremities, tachycardia and agitation. Surgery was necessary. During the emergency surgery, the omphalocele is reintegrated into the peritoneal cavity and there are

observed the following: on the diaphragmatic area of the liver, at the boundary between the left and the right hepatic lobe, there was a spherical formation of 6/5/3 cm, with translucent capsule, smooth, brown, representing a part of the liver protruding in omphalocele. The intestine had normal construction and position (Figure 1).

At liver reintegration, which was under a membrane, apparently avascular, multilayered, and especially after closing the surgery stage, the cardiac and pulmonary

activity stopped and after a resuscitation of about one hour and 20 minutes, death has been declared. Necropsy did not bring other pathological elements. The subsequent morphopathological microscopic examination concluded the following: diffuse microvesicular steatosis, sinusoidal stasis marked and isolated the areas with massive porto-biliary fibrosis; the presence of subcapsular cystic formations with massive fibrin deposits and abundant acute inflammatory infiltrate (Figures 2-4).

Figure 1 – Appearance of the liver at necropsy: aberrant “lobe” liver.

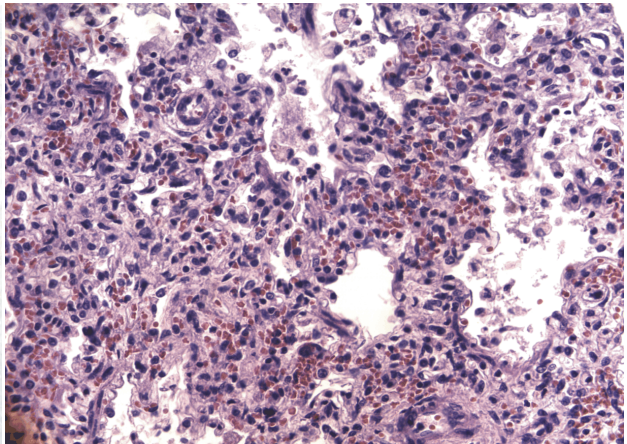


Figure 2 – Section through the right lobe of the liver. HE stain, 40×.

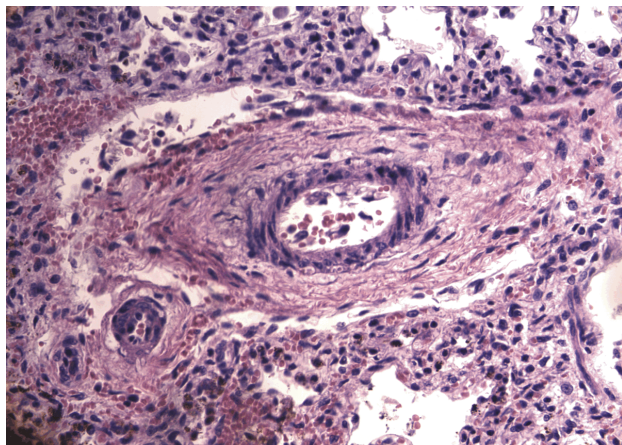


Figure 3 – Section through aberrant “lobe” liver. HE stain, 40× (image 1). Appearance of the liver with the same structure as the rest of the parenchyma.

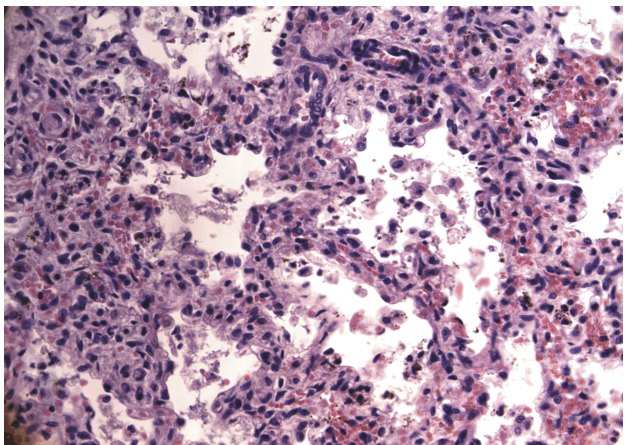


Figure 4 – Section through aberrant “lobe” liver. HE stain, 40× (image 2). Appearance of the liver with the same structure as the rest of the parenchyma.

Discussion

Omphalocele is a herniation of abdominal viscera through a much-enlarged umbilical ring, and should be distinguished from laparoschisis (gastroschisis) which is a paraumbilical congenital evisceration. In case of the omphalocele, the tegument is missing, the tumor formation is covered by a translucent pearly avascular membrane, formed by peritoneum, Wharton jelly and amniotic foil; umbilical cord is inserted in the anterior pole of the omphalocele, its elements being dispersed at the base. Gastroschisis is the presence of a right paraumbilical small parietal defect by which the transverse colon or bowel loops eviscerate; the umbilical cord is normally inserted, there is no bag and in all cases, the malrotation appears due to the lack of coalescence process [8].

At the beginning of the sixth week of gestation, continuous stretching of the small intestine, derived from

the middle intestine, combined with increased abdominal pressure due to increased abdominal viscera, especially liver, causes herniation of intestinal loops through the umbilicus, and produces the rotation of 90° in the counterclockwise direction, rotation that occurs around the vascular axis represented by the superior mesenteric artery. In the tenth week, the middle intestine retracts into the peritoneal cavity-taking place at the same time as the umbilical orifice restriction. At fourth week, the abdominal wall is separated only by somatopleure, then taking place through differentiation and proliferation the abdominal wall formation, which is being finalized until week 10, and currently the intestine fully reaches the abdominal cavity [9]. The presence of a parietal defect of the ventral abdominal wall in the umbilical region is assumed that is due to insufficient dorsoventral curvature of the embryonic body between weeks 4 and 8, and due to insufficient differentiation of somitic mesoderm, which

normally forms the connective tissue of the skin and anterior abdominal wall muscles [10]. Thus, omphalocele may be due to either insufficient return of bowel loops in the abdominal cavity, in these cases, the hernia sac is formed solely from amnion or a secondary herniation occurring after the tenth week, due to a defect of the anterior abdominal wall closure in the umbilicus region, when the hernial sac is formed of peritoneum, subserous fascia and amnion. However, parietal abdominal defect can be located above the umbilicus, in which case it may evaginate the liver or heart, or lower when it may evaginate only the small intestine.

In our case, the presence of multilayer membrane, leads to a more insufficient differentiation of the somitic mesoderm, translated by secondary herniation of abdominal viscera and the second element of this particular case is the liver protrusion through hernial ring, the liver being formed at the end of the tenth week. This hepatic protrusion is put on the account of liver size, knowing that at 30 days, liver weight is equal to the embryo weight and at birth, it occupies more than half of the abdominal cavity, the ratio between the liver weight and of body weight being 1/20; in adults this ratio becomes of 1/33. Omphalocele, as mentioned, especially in the major form, is often associated with other malformations. There have been described cases of association with colon atresia and Hirschsprung's disease [11], with complete absence of the large intestine [12] or with ileal prolapse and omphalomesenteric duct [13]. Medical literature mentions a case where the omphalocele was associated with intestinal malrotation, urinary bladder exstrophy and medulloblastoma, omphalocele association with malformations of the nervous system rarely occurring [14]. There is met a frequent association between omphalocele and cardiovascular malformations, the medical literature describing malformations and various associations such as: abnormalities of the inferior vena cava, being quoted its angulations at the junction with the right atrium or even the absence of the inferior vena cava [15–17]; association with interatrial septal defect and coronary sinus [18]; association with the left heart hypoplastic syndrome and left bronchus hypoplasia [19] or aorta coarctation [20]. Other malformations associated with omphalocele would be pulmonary hypoplasia [21], pseudocyst of giant multilobular umbilical cord [22], intrapericardial diaphragmatic hernia [23], trisomy 1q [24], trisomy 18 [20], radius aplasia [20], intellectual disabilities or hypospadias, the latest being associated with chromosomes deletion 2q22, 1q22.3 [25]. However, there have also been mentioned some late pathological associations of omphalocele such as cardiac valvular defects, being known the omphalocele frequent association with cardiac malformations, but a clear causal link could not be established [26]. The most frequent malformations associated with omphalocele are common mesentery, malrotation, intestinal atresia or volvulus [27–32]. In the minor omphalocele there has been reported urinary bladder herniation [33]. There have been reported cases when the omphalocele sac herniated a liver cystic teratoma together with liver falciform ligament [34]. However, omphalocele cases when liver herniates are very rare. Liver involvement is exceptional and in the case of gastroschisis [35] rather

can be found engaged with the gall bladder, uterus, fallopian tubes, urinary bladder or undescended testicle. Kluth D and Lambrecht W consider that the gastroschisis is more probably a small broken omphalocele rather than an entity with its own development [36]. Omphalocele presence is also encountered in Cantrell–Heller–Ravitch syndrome, which assumes the existence of five defects: ectopia cordis, abdominal wall defect (omphalocele most frequently, but gastroschisis may exist), defect in the sternum, anterior diaphragm and diaphragmatic pericardium. The disease is extremely rare. It occurs at about five births in a million. So far there have been reported 90 cases of Cantrell's syndrome, of which very few formed a complete picture of the syndrome. In one case of Cantrell's syndrome, there has been reported a form of liver dysmorphism, in which the liver presented three separate lobes [37]. Recent studies have shown that folate and vitamin B₁₂ used around the period of conception can prevent omphalocele [38]. Regarding the omphalocele treatment, opinions differ and vary depending on its size. Dunn JCY and Fonkalsrud EW report that despite surgical improvements, mortality exceeds 10% [39]. Generally, if the omphalocele is an isolated lesion, the prognosis for survival after postnatal surgical correction exceeds 90%; in such cases, prenatal diagnosis may contribute to improved survival prognosis [40]. The present case is a pathological rarity, on one hand due to liver protrusion alone and on the other hand due to liver dysmorphism. Omphalocele association with other malformations significantly reduces the chances of survival, the most serious being the cardiac malformations which actually dictate the prognosis. In this case, due to the unfavorable evolution, the closure of the omphalocele was tried, and although the abdominal wall allowed parietal defect closure, cardiorespiratory arrest occurred.

✚ Conclusions

Due to the presence of multilayer membrane, we consider that the omphalocele development is due to insufficient differentiation of somitic mesoderm, translated by secondary herniation of abdominal viscera, also influenced by the liver protrusion through hernial ring, the liver being formed at the end of the tenth week. This hepatic protrusion is considered to be due to liver size, knowing that after 30 days, the liver weight is equal to embryo weight and at birth, it occupies more than half of the abdominal cavity, the ratio between liver weight and body weight being 1/20; in adults, this ratio is 1/33.

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