Body stalk anomaly: Three months of survival. Case report and literature review

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ABSTRACT
Body stalk anomaly is composed of a set of genetic component abnormalities that are still rather unknown. This anomaly consists of a large defect in the abdominal wall closure, anatomical defects of the pelvis and lower limbs, severe scoliosis, and pulmonary hypoplasia. In addition to these deformities are heart disease and neural tube closure defects. Because of the association of these severe deformities, the cases described in the literature have proven to be almost entirely incompatible with life, resulting in abortion and stillborn fetuses. Therefore, the present article describes a case of body stalk anomaly that survived for nearly three months, the first of its kind in Latin America.

The disease known as body stalk anomaly, limb body wall complex, or body stalk complex is a very rare anomaly (1:14,000 to 42,000 pregnancies; 1: 7500 fetuses from 10 to 14 weeks of gestation) [1–3]. This medical condition is characterized by a complex anomaly of the anterior abdominal wall, severe kyphoscoliosis, rudimentary umbilical cord, and anatomical defects of the pelvis and lower limbs. Cardiac disorders, craniofacial defects, and several other malformations may also be associated [4–6]. The pathogenesis of this variable spectrum of anomalies is not well understood, but it is usually incompatible with life, progressing to miscarriages or stillborn fetuses. Prior literature shows only three reports of survival for more than one month, since the death most commonly occurs in fetal or perinatal period. The present study illustrates a case of this serious congenital malformation with an 84-day survival, which is most likely the first case of its kind reported in Latin America.

Our aim is to describe the rare occurrence of the body stalk complex and the importance of multidisciplinary care to provide the proper perinatal management for this newborn.

1. Case report

A pregnant woman, at 36 weeks of pregnancy, in her second pregnancy, was admitted to the Clinical Hospital at the Federal University of Minas Gerais (UFMG/Ebserh) and was submitted to a C-section due to a fetal breech and malformations observed by morphological fetal ultrasound (suspicion of gastroschisis, congenital clubfoot, and anidramnio).

The newborn (NB) was a male baby, who was hypotonic, cyanotic, and bradycardic. Oropharyngeal aspiration and oxygen mask ventilation were immediately performed to prevent hypothermia, presenting swift improvement in cyanosis. The baby presented an Apgar score of 5/8 and a weight 2295 g ($p < 10$).

Multiple defects were identified: bulky ruptured omphalocele (liver and gall bladder, intestines and pancreas, and externalized testis), pelvic asymmetry, and atrophy of the lower limbs with clubfoot (Figs. 1 and 2).

Shortly after birth, the NB was referred to a Neonatal Care Unit for clinical stabilization, where he underwent endotracheal intubation and catheterization of the umbilical vein in order to administer electrolyte solutions and antibiotics.

After receiving an evaluation from the pediatric surgery team, we decided to keep the NB in the operating room, where we performed right femoral vein catheterization by dissecting the great saphenous vein, and then conducted a surgical correction of the
abdominal defect due to a large visceros-abdominal disproportion. As the giant omphalocele was ruptured, and the double-sided screen was not available at the moment, the protection of exposed intestines was made with the suture using the baby's own membrane (amnion, Wharton's jelly, and peritoneum), associated with the use of a bovine pericardium screen to cover the exteriorized portion of the liver.

During the postoperative period, the patient received intensive care in the Neonatal Care Unit, including prolonged parenteral nutrition. Conservative local treatment of omphalocele was implemented through the topical application of silver sulfadiazine to facilitate the epithelization of the remaining phase in order to preserve the amniotic membrane (Fig. 3).

As for other associated anomalies, it is worth noting that the NB presented myelomeningocele and defects of the thoracic and lumbosacral spine. In addition, no significant cardiac abnormalities were observed in echocardiographic examinations, except for the patent ductus arteriosus of 4.4 mm. The transfontanellar ultrasound (USTF) presented results that were within the normal range. The karyotype was 46, XY; blood screening for phenylketonuria, hemoglobinopathies, cystic fibrosis, and hypothyroidism proved to be negative; and abdominal ultrasound revealed right hydronephrosis.

Due to the persistence of food intolerance, a CT scan of the abdomen was performed, which revealed a specific gastric compression caused by the liver and indicated the need for a nassoenteric tube to facilitate food intake. The patient presented anemia and hypoalbuminemia, and received blood transfusions.

The NB developed anasarca, respiratory failure, pulmonary hypertension (confirmed by echocardiography), and carbon dioxide retention, requiring high levels of mechanical ventilation. These conditions are possibly secondary to pulmonary hypoplasia associated with giant omphalocele. Later, due to prolonged mechanical ventilation, it was necessary for the NB to undergo tracheostomy.

Despite intensive care, the patient developed severe cardiorespiratory failure, abnormal liver function, electrolyte disturbances, and an acid base. On the 84th day after birth the NB died.

2. Discussion and literature review

The body stalk anomaly is a set of very rare congenital malformations. In Denmark, between 1970 and 89, a prevalence of 3.4% of live or stillbirths were born with a defect in the abdominal wall [7,8].

The affected fetuses or NBs present multiple malformations, the most common of which include severe scoliosis, pulmonary hypoplasia, pelvis defects, giant omphalocele, and, in some cases, a short or an absence of the umbilical cord [9–13]. This anomaly is also called limb-abdominal wall complex [1] and can be classified into two types according to the malformation phenotype: type I is complicated with craniofacial defects and type II is complicated by ventral wall defects [9].

Almost all of the affected fetuses and NBs quickly evolve to death [14–16], mainly due to pulmonary hypoplasia, which causes respiratory failure and severe pulmonary hypertension. There is a consensus in fetal medicine that body stalk anomaly is fatal, as few
The first case of body stalk anomaly in Latin America with a long time survival was reported in 2007 by the Japanese authors, a country of greater economic and social development, which has advanced technologies for the perinatal management of patients with severe and rare anomalies, such as cases of body stalk anomaly.

Despite the fatal evolution, it can be conclude that it is important to report the experience of managing complex cases in pediatric surgeries such as this. Therefore, this case contributed to our knowledge of how to apply proper surgical tactics in an attempt to prolong survival and improve the quality of life of some children with giant omphalocele associated with varying forms of body stalk anomaly.

### 3. Final comments

It is important to mention the importance of the ethical aspects [5,35] that are absolutely necessary when a medical staff is faced with the challenge of treating children with rare and complex diseases, especially those that include very limited or even fatal outcome survival, as is the case of body stalk anomaly. Generally, hospitalization may be prolonged, generating both an emotional and a financial burden on the family and the healthcare system in general. Intensive care treatment can still cause distress and pain to the patient and family, due to high mortality. In this context, multidisciplinary care is fundamental in order to make a reflection on the best decision to be made regarding NB care limits, highlighting the family approach, taking their fears and anxieties into consideration. We, as healthcare professionals, should not only seek to prolong life, but also to minimize the suffering of patients and provide full support to the NB’s parents.

### 4. Conclusions

The present study reports on an experience with the treatment of most likely the first case of body stalk anomaly in Latin America, with a survival of 84 days, in order to highlight the importance of prenatal diagnoses and multidisciplinary perinatal care. Currently, advances in neonatal intensive care have made the survival of patients with this complex and rare congenital malformations possible, but is important to reflect on the ethical challenges in the approach taken for these patients and their families, taking into account the appropriate therapeutic limits for each case.

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