

Case Report

A Rare Sporadic Polymalformative Syndrome with Congenital Abdominal Wall Defects. Caregivers and Parents Working Together Make Shared Ethical Decisions

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Abstract: Improvements in ultrasound technology and the appropriate timing of antenatal ultrasound has led to refined prenatal diagnosis and enhanced accuracy of diagnosis of fetal anomalies and makes it possible to treat or not at birth, because they have diverse etiology and prognosis. They has severe congenital common finding as thoracoabdominal defects, spinal cord abnormalities, positional limb deformities, abnormalities of umbilical cord and ectopia cordis. The technological revolution and radical new treatment modalities in patient care cause ethical dilemmas when therapeutic possibilities are very varied. Healthcare workers at all levels of care, after detection of fetal defects need to get involved to provide better family management to reduce pain and suffering. Avoid a futile treatment is ethical obligation since the time of Hippocrates.

Keywords: Body Stalk Anomaly, Thoracoabdominal Syndrome, Pentalogy of Cantrell, Amniotic Band Syndrome, Bioethics, Palliative Care

1. Introduction

Ultrasound technology can be used to detect most birth defects before birth, conditions that require immediate and adequate support. Healthcare workers at all levels of care, after detection of fetal defects need to get involved to provide better family management to reduce pain and suffering.

Clinical finding's brought together sporadic polymalformative syndromes diagnosed by prenatal ultrasonographic studies with diverse etiology and prognosis. They had severe congenital common finding as thoracoabdominal defects, spinal cord abnormalities, positional limb deformities, abnormalities of umbilical cord and ectopia cordis. Body stalk anomaly (BSA), thoracoabdominal syndrome (THAS), pentalogy of Cantrell and amniotic band syndrome (ABS) are rare similar fetal polymalformative syndromes, though there are certain

differences between them. [1, 2, 3]

BSA is a combination of developmental abnormalities involving spine and neural tube, body wall, and the limbs with persistent extraembryonic coelomic cavity. ABS is characterized by the presence of thin membrane-like strands attached to fetal body parts and causing constrictions and amputations membranes and persistent extra embryonic coelomic cavity. The spectrum of anomalies depends on which part gets entrapped and at what point of gestation. Limb body wall complex (LBWC) is a fatal form of ABS who mimes BSA, THAS and Cantrell's Syndrome. Various hypotheses was proposed to explain the pathogenesis of ABS and LBWC including early amnion disruptions, embryonic dysplasia, and vascular disruption in early pregnancy.

BSA has abdominal wall defects in which the abdominal organs lie outside of the abdominal cavity in a sac of amnioperitoneum with a very small umbilical cord. BSA is a

fatal anomaly and, hence, its early diagnosis aids in proper management of the patient as THAS, a rare disorder that is present at birth characterized by a combination of defects of sternum, congenital diaphragmatic hernia, absence of the pericardium, omphalocele and congenital heart diseases.

The hypotheses proposed to explain the pathogenesis of BSA is a germinal disk abnormality. A complete failure of body folding along cephalic, caudal, and lateral axes during the sixth postmenstrual week. [4] Aberrant cephalic folding leads to a defect in the thoracic wall and epigastrium, which allows development of ectopia cordis. Aberrant lateral folding results in herniation of the midabdominal contents into a large amnioperitoneal sac, which inserts peripherally onto the placental chorionic plate in lieu of a very short umbilical cord. Due to the extrusion of the intraabdominal contents, the spine and thoracic cavity do not develop symmetrically, which results in severe scoliosis and abnormalities of malrotation of the spine. It has been associated with cocaine abuse and short age mothers. It is mostly considered to occur randomly from unknown cause.

THAS (OMIM 313850) occurs with varying degrees of severity, potentially causing serious life-threatening complications. When all five defects are present, this is referred to as complete pentalogy of Cantrell. Professor Rivka Carmi, an Israeli pediatrician and geneticist confirm THAS as a X-linked dominant disorder with markers in the region Xq22-q27. [5] Additional results indicated that the TAS gene is located between the DXS425 and HPRT loci (Xq25-q26.1). [6] The specific symptoms and severity can vary from one fetus to another; some babies may have mild defects with incomplete expression of the disorder and others may have serious, life-threatening complications. The affected individuals will not necessarily have all of the symptoms. [7]

There are similarities and differences between fetal abdominal wall defects which are very important for the correct patient management. Most abdominal wall defects are detected antenatally, but prenatal diagnosis may influence timing, and location of delivery and this facilitates postnatal multidisciplinary approach. The goal of surgical treatment is to place the herniated viscera back into the abdomen and close the wall defect. Options include primary closure, staged approaches and delayed closure; or palliative care.

The size of the defect, the presence of associated anomalies and respiratory failure at birth are significant predictors of mortality. [8] In these cases, the question is whether survival is possible because infants are likely to have serious lung diseases due both to prematurity and to the effects of the defects and life-threatening complications. In case of doubt, aggressive delivery room interventions cannot be characterized as physiologically futile and they may be temporarily initiated, but if attending physician think this temporary success does not alter the probability of survival he need to change the level of care. Caregivers have no ethical obligation to provide futile treatment.

For those newborns where death is unavoidable there is an urgent need to initiate informed consent processes with parents in order to formulate a consensus of palliative care. An

important part of this process is to evaluate the parent's values in order to include their in the consensus. It has been suggested that parents' experience generate less guilty when the neonatologist starts discussion about palliative care and end of life decision. Exploration of personal parents values is needed. [9, 10]

2. Case Report

A 41-year-old woman, G5 P3 Cs1 Ab0, had regular prenatal care before 14 weeks of gestation without problems. She had a no consanguineous married life of six years without past or present illness but cocaine use by both parents. At 19 weeks of gestation fetal scan was performed and an anterior thoracoabdominal defect with a partial extrathoracic heart and big supraumbilical omphalocele was recognized in this (until now) uneventful pregnancy during the second ultrasonographic evaluation. There was no family history of congenital anomalies, genetic abnormalities, or history related to ectopia cordis. The woman was admitted to a tertiary level perinatal hospital at gestational age of 22 weeks. Diagnosis of anterior midline supraumbilical abdominal wall defect, deficiency of the anterior diaphragm, defect of the lower sternum, defect in diaphragmatic pericardium, partial thoracic ectopia cordis and severe congenital kyphoscoliosis was made in a 46xx fetus without structural or numeric chromosomal anomalies. An unfavorable prognosis to the fetus was predicted and conservative management of prenatal care had been chosen by family.

The woman was referred to our zonal hospital at gestational age of 24 weeks. The diagnosis of congenital fetal anomaly was Cantrell's pentalogy. Despite unfavorable prognosis to the fetus, mother had chosen conservative care and offer palliative and psychological care by a multidisciplinary team consisting of a perinatologist, a obstetrician, a neonatologist, a *psychologist* and nurses should counsel parents prenatally with frankness and with accurate medical information about the delivery mode and possible infant care.

Parents choose autonomously a cesarean section delivery informed about the possibility of fetus demise during labor due to prolonged cardiac compression, damage of herniated viscera, or rupture of omphalocele sac in partum.

Cesarean section was performed at 34 weeks of gestation. The newborn was a female of 1880g weight who scored 6 (1min) and 6 (5min) on Apgar scale at birth, with hypotonia, weak cry, and generalized cyanosis. The physical examination revealed split sternum with partial thoracic ectopia cordis by anterior thoracoabdominal wall defect; serious kyphoscoliotic curvature of the spine; supraumbilical left omphalocele with stomach, small and large bowel loops, liver, gallbladder and spleen inside the sac with umbilical cord vessels in the wall. Umbilical cord was short and had three vessels. Low implant ears was an additional finding. He had no neural tube defects and permeable anus. Figure 1

Ectopic heart with partial absence of the pericardium was beating in the limite of an hipoplastic chest, at a rate of 100/min with remittent bradycardia. After birth, the onphalocele was

covered with warm saline-soaked sterile dressing and assisted with positive pressure ventilation with the lower target range of oxygenation. An x-ray of the spine was taken. Figure 2



Figure 1. Female with split sternum, partial thoracic ectopia cordis by anterior thoracoabdominal wall defect.



Figure 2. Severe scoliosis and malrotation of the spine.

Newborn's father present into the delivery room understood the conduct established by medical team, endorsing that the approach of discard surgical intervention was the most appropriate election. The newborn girl was transferred to the neonatal intensive care unit but when she was admitted, palliative care was indicated.

It was impossible to make a differential diagnosis between fetal abdominal wall defects such BSA (variable sized defects, abnormal cord insertion, membrane covered liver/bowel, ascites) and THAS including Cantrell's pentalogy (sternal, pericardial, diaphragmatic defects; ectopia cordis; large omphalocele). She died within 2 hours. The parents declined postmortem newborn's autopsy.

3. Discussion

Antenatal ultrasound made prenatal fetal anomalies diagnosis but in some cases as in this one can not establish viability and possibility of treat. These rare group of fetal

sporadic polymalformative syndrome diagnosed by antenatal ultrasonographic studies has diverse treatment and prognosis; they has similar congenital thoracoabdominal defects.

The differential diagnosis includes BSA, THAS, ABS and other fetal abdominal wall defects syndromes, for example OEIS complex (large omphalocele, exstrophy, imperforate anus, spinal anomalies complex). Body stalk anomaly is usually characterized by the presence of a major anterior body wall defect, limb deformities, kyphoscoliosis, an absent or short umbilical cord and/or craniofacial defects. Our patient had a normal karyotype, abnormal cord insertion, a membrane sac covered heart, liver and spleen with sternal, pericardial, and diaphragmatic defects; severe congenital kyphoscoliosis and thoracic hypoplasia.

In this case as many studies shows too, the level of experience of the ultrasound examiners often remains unclear. An faster indication for intrauterine MRI can give us more information compared to highly specialized, experienced ultrasound examiners in prenatal tertiary centers. Intrauterine MRI is now playing an additional role in fetal differential diagnosis but is stressful for families.

Ethical dilemmas are not new to perinatal medicine. With new technic and advances surgical treatment some congenital malformation can survive with high quality of life. With uncertain antenatal data and the parental decision to continue gestation, some ethical decision moved to the delivery room. Standards in evidence-based ethics have complimented evidence-based medicine guidelines. We use Tom Beauchamp and James Childress principle ethics approach: Autonomy, nonmaleficence, beneficence, and justice; and Carol Gilligan's Ethics of Care, moral approach on relationships instead of emphasizing autonomy and rules; from compassion instead of being justice-based and in connections with each human being in the scenario. [11, 12]

Importantly, the understanding between the family and the medical team was established, accepting that the approach of discard a surgical intervention and mechanic ventilation was the most appropriate. We argue that before and after birth and when the outcome is uncertain, all available options and detailed information should be fully explained to the parents.

Ethically speaking, beginning care with palliative intent or changing from intensive action to palliative care are not morally different although the processes and procedures are different. Exist the perception that initial treatment with palliative care may be easier and cause less suffering and pain to the infant than switching from intensive to palliative care in the reorientation of care. In case of doubt, aggressive delivery room interventions cannot be characterized as physiologically futile and they may be temporarily initiated. However, palliation of suffering and pain is necessary in both schemes.

4. Conclusions

MRI can be recommended with appropriate safety measures (a quick examen after 18 weeks of gestation) as in our case with unclear sonographic finding and possible therapeutic consequences for the care of the fetus or newborn

baby. MRI is an excellent supplementary method to ultrasound. Concretely, this means ultrasound first.

Ethical decision making cannot be conceived without the active and autonomous participation of the newborn's family.

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