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CASE REPORT: DIAGNOSIS AND MANAGEMENT OF BODY STALK ANOMALY

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ABSTRACT

Background: Body stalk anomaly is a rare and severe malformation syndrome , occurring only 1 of 10.000 live births. The patholphysiology and trigger factor of body stalk anomaly are still unknown and characterized by : abdominal wall defects, thoracoo and abdominochisis, Severe kyphoscoliosis, a truncated or absent umbilical cord, and limb irregularities are characteristic features of body stalk anomaly. Potential factors contributing to this condition encompass early amnion rupture due to direct mechanical pressure or the presence of amniotic bands, vascular disruption in the early embryo, or an abnormality in the germinal disk.Early antenatal diagnosis is crucial for patient management despide the low life expectancy, and cooperative teamwork is needed to investigation the abdnormalities encountered at birth. the chromosomal testing is performed for congemital anomaly investigation. **Case Presentation:** We describe a case of body stalk anomaly that was identified through antenatal ultrasound evaluation during the initial visit. The findings were confirmed during the fetomaternal ultrasound examination, the finding were as follows : a single live fetus in utero in transverse lie position, absent amniotic fluid, scoliosis of the vertebrae, an abdominal wall defect, visceral organs (liver and intestines) protruding from the defect. The patient underwent elective caesarean section due to the tranverse lie positionof the fetus and severe oligohydramnios. The baby was born weigh in 2700 grams, measuring 30 cm in length, and with apgar score of 1/1. The observed congenital anomaly include : the placental adhering to the fetal abdomen, absence of an umbilical, visceral organs (liver, spleen, small intestine, and large intestine) protruding from a large abdominal wall defect. Both upper limbs are normal , the right lower limb has short femur with tibia and fibula, four toes and a band from the right foot to the pelvis. The left limb is not formed, with low set ears, placental thicketing, a singke umbilical artery, and undeveloped anus. **Conclusion:** Ultrasound examination is primary diagnostic tool to detect Body Stalk Anomaly (BSA) ,with ultrasound , doctors can visualize the internal structure of the fetus and conditions intra uterine. Diagnosis of chromosomal abnormalities in fetus is one of most important challrnge in perinatology .

Keywords : *Bodystalk Anomaly , ultrasound , cromosom, defect abdominal wall , limb abnormalities*

INTRODUCTION

Birth defects are common. Of all newborns, 2 to 3 percent have major congenital abnormality detectable at birth and 80 percent do not have an obvious etiology, and of those with an identified caused, nearly 95 percent of causes have chromosomal or genetic origins[1].

Body stalk anomaly is an exceptionally rare, extremely severe, and lethal abdominal wall abnormality. It constitutes a profound deformity affecting the limbs, thorax, and abdomen, marked by an absence of abdominal wall development. This results in the peritoneal cavity being exposed to the extraembryonic coelom, with the fetus being attached to the placenta. Other characterized are thoracoscizis, abdominosciazis, an intestinal featuring ,limb defect and exencephaly This condition is extremely rare with an incidence of 1 per 14,000 and 1 per 31,000 pregnancies in large epidemiologic studies[2-5].

Ultrasound

Accurate prenatal diagnosis is crucial for effective patient care. A key factor in ultrasound assessment involves determining the defect's position relative to the umbilical cord insertion. Ultrasound performed a pivotal role in evaluating essential features of abdominal wall defects and serves as a highly valuable tool for diagnosing these conditions. The screening sensitivity of ultrasound is approximately 80%. Ultrasonography (USG) is the preferred imaging modality for assessing the fetus before birth. The earliest point at which an omphalocele can be identified is at 12 weeks of gestational age[4,6].

Routine ultrasound scans have greatly enhanced the capacity to identify this congenital anomaly before birth, showcasing a high level of both precision and accuracy. Prenatal diagnosis using ultrasound hinges on the observation of freely-floating herniated intestines without any protective membrane, in conjunction with a normally positioned umbilical

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cord. Consistent and consecutive ultrasound assessments are imperative due to the possible hazards, such as bowel blockage, peritonitis, intestinal perforation, and limitations in fetal growth[4,6,7].

Chromosome screening and FISH

The chromosomal abnormality may arise due to deviations in chromosome structure or quantity. While conventional karyotyping analysis is the established method for prenatal screening of chromosomal abnormality, it is acknowledged to be a procedure that demands significant time and effort.

The FISH test utilized the AneuVysion technique, which includes three α-satellite DNA probes targeting chromosomes X, Y, and 18. These consist of three centromeric probes and two locus-specific probes, which are applied to the sample using two separate hybridization slides[9].

CASE REPORT

A 25-year-old female patient referred by the obstetrics specialist of Kerinci Regional General Hospital, with a diagnosis of G1P0A0 at full term pregnancy of 40-41 weeks and major congenital abnormalities. The patient was referred to Dr. M Djamil Padang Hospital for comprehensive management of her baby. On the Fetomaternal ultrasound examination at Dr. M Djamil Padang Hospital, it was observed that: There is a single live fetus in the intrauterine position, lying transverse with the head in the lower back. There is vertebral scoliosis, amniotic fluid is depleted, there is no abdominal wall (defect), and visceral organs (liver, spleen, and intestines) protrude from the defect and are fused with the placenta. Other major congenital abnormalities are not clearly visible. Biometric measurements: BPD: 94 mm, FL: 65 mm, and HL: 58 mm. The impression from the ultrasound examination is: Severe oligohydramnios with suspicion of a body stalk anomaly.

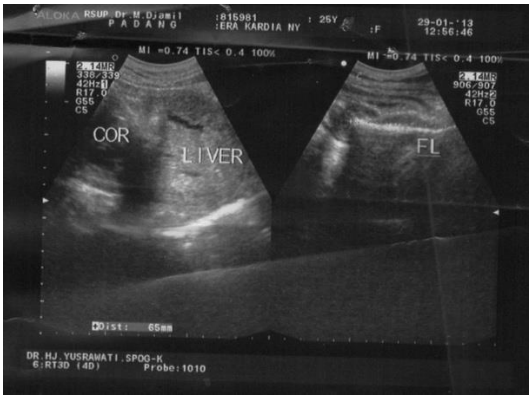

Figure 1. ultrasound report

Termination of pregnancy in this patient is an elective cesarean section due to indications of fetal transverse position and severe oligohydramnios, with collaboration between the perinatology and pediatric surgery teams. The baby was born with a weight of 2700 grams and a length of 30 cm, with an Apgar score of 1/1. Congenital abnormalities found in the neonate are: The placenta is attached to the fetal abdomen, there is no umbilicus, visceral organs (liver, spleen, small intestine, and large intestine) protrude from a large abdominal wall defect. Both upper extremities are normal, the right lower extremity has a short femur with tibia and fibula, there are 4 fingers, and there is a band from the right foot to the pelvis. The left extremity is not formed, low-set ears, placental thickening, single umbilical artery, and the anus and genitalia are not formed.

For further examination, a placental sample was sent for chromosome analysis. Chromosome analysis using Fluorescence in situ Hybridization (FISH) revealed that the patient's sex chromosomes are mosaic, specifically XO (38%) and XX (62%), with chromosomes 13, 18, and 21 appearing in duplicates, indicating they are normal. The presence of mosaic chromosomes, including normal (XX) and monosomy X (XO), generally occurs due to non-disjunction during post-fertilization mitosis. The FISH technique cannot rule out the presence of chromosome number abnormalities other than sex chromosomes (13, 18, 21) and structural chromosome abnormalities.

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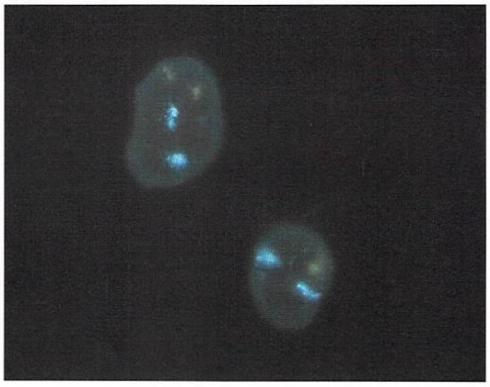

Figure 2. Result FISH Probe kromosom 18

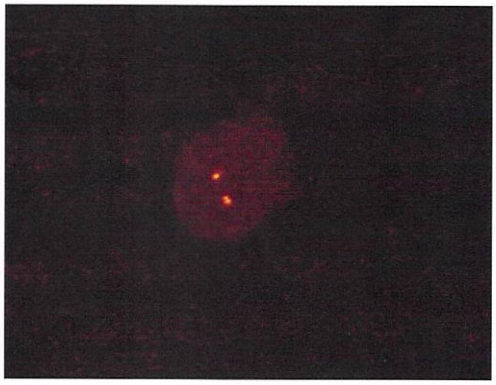

Figure 3. Result FISH Probe kromosom 18

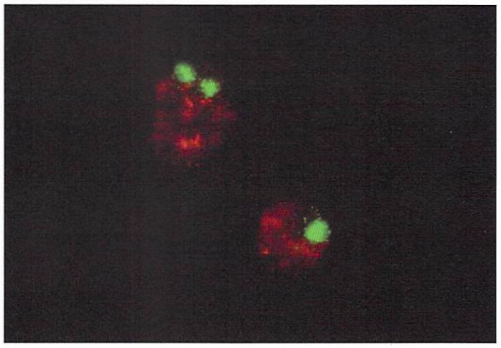

Figure 4. Result FISH Probe kromosom x dan Y

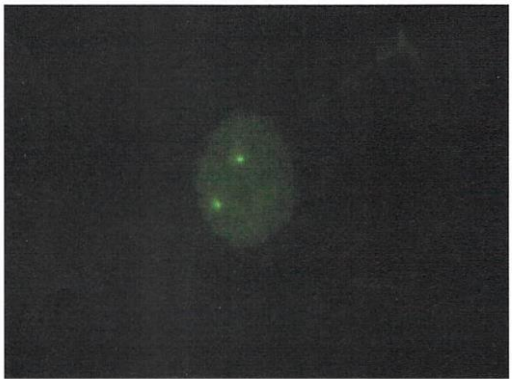

Figure 5. Result FISH Probe kromosom 13

The karyotyping analysis result is as follows: the number of chromosomes in each studied cell is mos 45,X(20)/46,XX(20), which means there are 2 populations of cells (mosaic), namely: a. 45,X, which means there are 45 chromosomes with one X chromosome (monosomy X), found in 20 studied cells (50%). b. 46,XX, which means there are 46 chromosomes with XX as the sex chromosomes, found in 20 studied cells (50%).

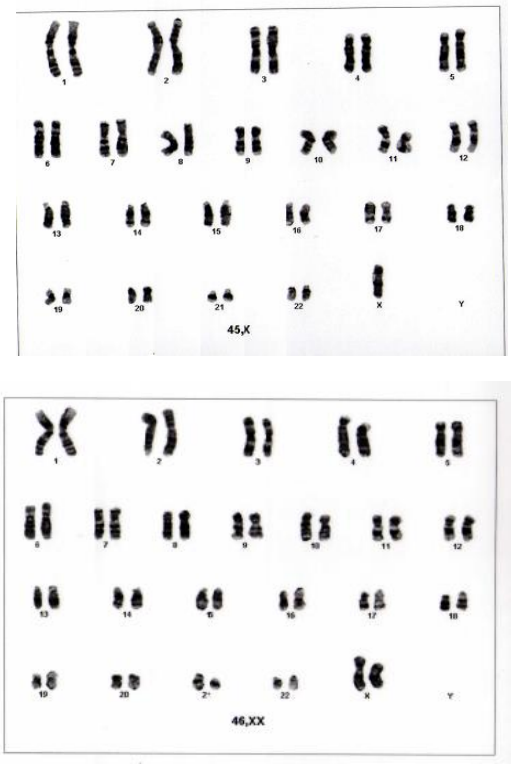

Figure 5. karyotyping

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DISCUSSION

The 25-year-old patient diagnosis of G1P0A0, at full term pregnancy of 40-41 weeks, along with a major congenital anomaly. In the full-term pregnancy, the patient had check-ups at Kerinci Regional General Hospital and was subsequently referred to Dr. M Djamil Padang Hospital for further examination at the fetomaternal department and comprehensive management of her delivery. Based on the ultrasound examination at the fetomaternal department, it corresponds to the presentation of body stalk anomaly. This includes an abdominal wall defect, visceral organs (liver and intestines) protruding from the defect and fused with the placenta, vertebral scoliosis, limb abnormalities, and severe oligohydramnios.

Ultrasound is a critical tool in diagnosing body stalk anomaly, as it allows for detailed visualization of fetal anatomy and abnormalities. However, it's important to note that this condition is rare and complex, and diagnosis can be challenging even with advanced imaging techniques.

The termination in this patient was performed through elective cesarean section, which was in line with the indications of transverse fetal position and severe oligohydramnios. The management of delivery with congenital anomalies was done in collaboration with the pediatric surgery and perinatology teams. The congenital abnormalities observed in the neonate include: placenta attached to the fetal abdomen, absence of an umbilicus, visceral organs (liver, spleen, small intestine, and large intestine) protruding from a large abdominal wall defect. Both upper extremities are normal, the right lower extremity has a short femur with tibia and fibula, there are 4 fingers, and there is a band from the right foot to the pelvis. The left extremity is not formed, low-set ears, single umbilical artery, and the anus and genitalia are not formed. This presentation corresponds to the diagnosis of Body Stalk Anomaly.

The limb body wall complex, also referred to as body-stalk syndrome, is an infrequent condition marked by severe deformities. The majority of fetuses do not reach full term, either experiencing spontaneous abortion or requiring medical intervention. Among the few that do, most are stillborn. Long-term survival after birth is exceedingly uncommon, with only one documented case, in which the child faced profound physical challenges. The cause of this condition remains unknown[6].

The most common anomalies are cardiac, genitourinary, gastrointestinal, musculoskeletal, neural tube and head neck defects.

Among abdominal wall defects, gastroschisis and omphalocele stand out as the most prevalent types. Both involve the intestines protruding outside the body through an abdominal opening, but omphalocele originates from within the umbilicus and entails the presence of a surrounding membrane enclosing the exposed intestines. In contrast, gastroschisis lacks this membrane enclosure, and the abdominal wall opening typically does not involve the umbilicus. Chromosomal anomalies are identified in 40-60% of fetuses with exomphalos and encompass conditions like trisomies 18, 13, and 21, as well as Turner, Klinefelter, and triploidy syndromes[10,6].

Body stalk anomaly accompanied by craniofacial abnormalities arises from early vascular disruption. The craniofacial defects from a primary ectodermal defect in the embryonic disc, while some of the malformations emerge as secondary complications of this initial disruption in embryogenesis. The majority of limb body wall complex cases are typically identified during the second trimester[11].

The baby was born with a weight of 2700 grams and a length of 30 cm, with an Apgar score of 1/1. Long-term survival after birth is exceedingly uncommon, with only one documented instance reported, and in this case, the child experiences severe physical impairments[12].

In postnatal management, chromosome and karyotype examinations were performed to determine if there is a relationship between Body Stalk Anomaly and chromosomal abnormalities, and to prepare for future pregnancies. The FISH and karyotyping chromosome examinations showed a mosaic sex chromosome impression, 45,X(20)/46,XX(20), with chromosomes 13, 18, and 21 in duplicate. The results of the examination correspond to Body Stalk Anomaly, indicating that this anomaly is not always associated with chromosomal trisomy abnormalities.

Birth defects can arise from genetic issues stemming from mutations in one or more genes, chromosomal abnormalities, or environmental influences during pregnancy. Cytogenetics focuses on examining the arrangement and quantity of chromosomes through techniques like karyotyping. It

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delves into irregularities in chromosomes, which can arise from either congenital or acquired genetic conditions. These abnormalities can result from mistakes in the number or structure of chromosomes. Cytogenetic disorders affect approximately 2% of pregnancies in women over 35 years old, 1% of live births, and 6% of stillbirths. It's notable that over half of spontaneous abortions in the first trimester are attributed to chromosomal irregularities. In India, nearly half a million babies are born each year with malformations, with approximately 21,000 cases of Down syndrome (trisomy 21) occurring, translating to about 1 in every 1,150 births. This condition is more prevalent than any other genetic disorder. Additionally, in pregnancies where malformations are detected through ultrasound, the incidence is notably higher, ranging from 17% to 27%[13-15].

CONCLUSION

The presented case involves a 25-year-old patient diagnosed with G1P0A0 at full term pregnancy, complicated by a major congenital anomaly. The anomaly, identified as Body Stalk Anomaly, encompasses severe malformations including an abdominal wall defect, protrusion of visceral organs, vertebral scoliosis, limb abnormalities, and severe oligohydramnios. The rarity and severity of this condition often lead to limited postnatal survival, with only isolated cases reported. In this instance, the neonate faced significant physical challenges. Ultrasound is a critical tool in diagnosing body stalk anomaly, as it allows for detailed visualization of fetal anatomy and abnormalities. However, it's important to note that this condition is rare and complex, and diagnosis can be challenging even with advanced imaging techniques.

Chromosome and karyotype examinations were performed to determine if there is a relationship between Body Stalk Anomaly and chromosomal abnormalities, and to prepare for future pregnancies.

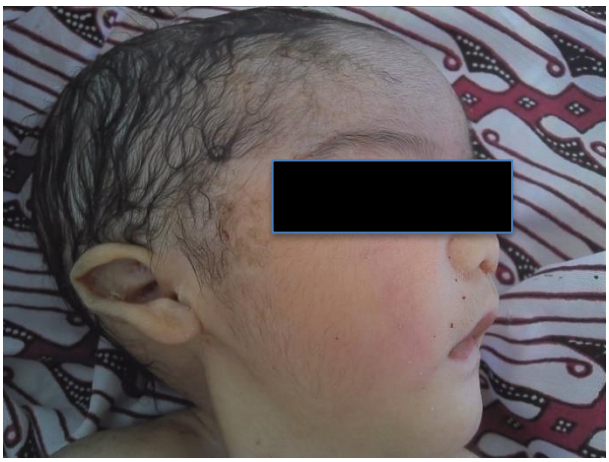

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Figure 6. fetus with body stalk anomaly

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