Clinical Case Reports International

Case Report
Published: 15 Jul, 2023

Short Umbilical Cord Syndrome: Antenatal Diagnostic

Fathi M, Eya K, Montacer H, Ameni M and Dalenda C*

Department of Obstetrics and Gynecology, Tunis Maternity and Neonatology Center, Tunis El Manar University, Tunisia

Abstract

Short Umbilical Cord Syndrome (SUCS) is a rare condition characterized by an abnormally short umbilical cord associated with a complex set of disruptive abnormalities having in common the failure closure of the ventral body wall.

This abstract presents an antenatal diagnostic case study of a patient diagnosed with Short Umbilical Cord Syndrome at 13 weeks of gestation.

The case study involved a pregnant woman who underwent routine ultrasound examinations during the first trimester. The ultrasound revealed several fetal malformations and a significantly shortened umbilical cord in addition to altered fetal movements and reduced amniotic fluid volume.

Five days after the ultrasound, the fetus died in utero, and the patient underwent an induction and fetal autopsy confirming the diagnosis of SUCS.

This case study underscores the significance of antenatal diagnosis in identifying Short Umbilical Cord Syndrome and implementing appropriate management strategies to optimize pregnancy outcomes.

Although the prognosis for SUCS is poor if all components are present, there are minor cases where malformations could be managed postnatally.

Early detection of SUCS through routine ultrasound examinations allows healthcare providers to closely monitor affected pregnancies and develop tailored delivery plans, thereby minimizing potential risks to both the mother and the fetus. Timely interventions and comprehensive follow-up care are crucial in ensuring the best possible outcomes for pregnancies affected by Short Umbilical Cord Syndrome.

Introduction

Short umbilical cord syndrome, also known as the limb-body wall malformation complex and the body stalk anomaly, is a poorly defined sporadic group of congenital anomalies characterized by a complex set of disruptive abnormalities having in common the failure closure of the ventral body wall. This disorder is characterized by a short or absent umbilical cord and disruption of the lateral body wall, spine, limbs, face, and cranium, isolated or in combination.

Early detection of SUCS through antenatal diagnosis is crucial for effective management and optimizing pregnancy outcomes.

In recent years, advancements in prenatal screening and diagnostic techniques have improved the ability to identify structural abnormalities and anomalies during routine ultrasound examinations. The detection of SUCS during these screenings provides healthcare providers with valuable information for developing appropriate management strategies and implementing targeted interventions to mitigate potential risks.

This case report presents a unique instance of antenatal early diagnosis of Short Umbilical Cord Syndrome in a pregnant woman. The case highlights the importance of early first trimester ultrasound examinations in identifying this rare condition and underscores the subsequent management considerations necessary especially for ensuring the well-being of both the mother and the fetus if the case is mild and to minimize the mother's psychological trauma if the case is severe or lethal.

Understanding the antenatal diagnostic process and management considerations for Short Umbilical Cord Syndrome is essential for obstetricians, radiologists, and other healthcare professionals involved in prenatal care.

OPEN ACCESS

Correspondence:
Kristou Eya, Department of Obstetrics and Gynecology, Tunis Maternity and Neonatology Center, Tunis El Manar University, Tunisia

Received Date: *26 Jun 2023*
Accepted Date: *11 Jul 2023*
Published Date: *15 Jul 2023*

Citation:
Fathi M, Eya K, Montacer H, Ameni M, Dalenda C. Short Umbilical Cord Syndrome: Antenatal Diagnostic. Clin Case Rep Int. 2023; 7: 1572.

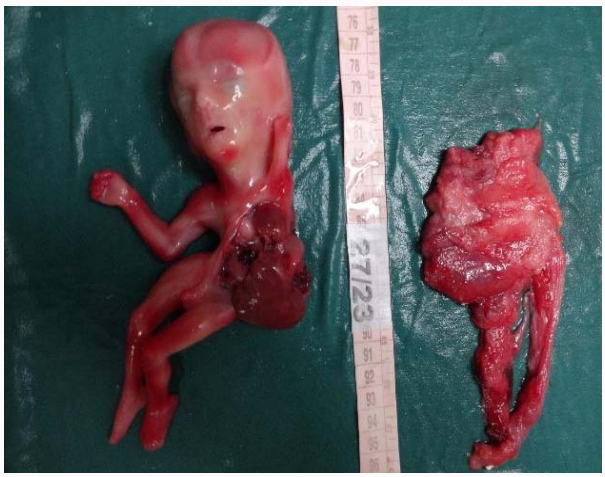

Figure 1: Left: Anterior view of the fetal abdominal wall defect and marked scoliosis. Right: Fragmented trophoblast with an attached remnant umbilical cord.

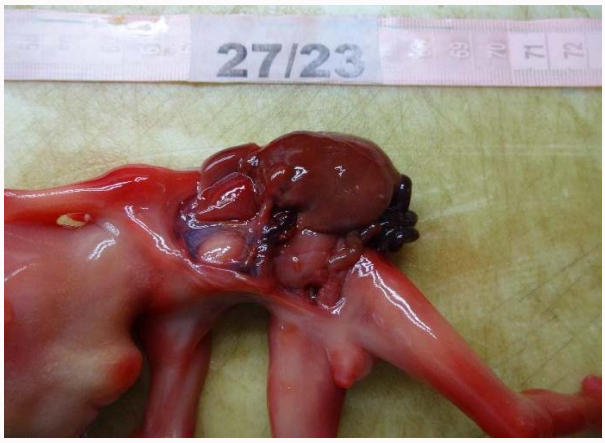

Figure 2: Laparoschisis extended to the thoracic and pelvic area with evisceration including the heart, left lung, the liver, intestines and the bladder.

Observation

Madam S, a 30-year-old pregnant woman Gravida 2, Para 2, the first child is 2 years old, in apparent good health.

The patient has no pathological history, and the current pregnancy is at a term of 13 weeks of gestation when she was referred to us for a routine ultrasound examination. As part of her comprehensive prenatal care, a dating ultrasound was performed at 7 weeks' amenorrhea, confirming the pregnancy and term.

The ultrasound performed at our department was the second one she undergoes during her pregnancy, and it revealed bilateral cystic hygromas, severe scoliosis of the spine, malposition of the fetal heart, and an abdominal wall defect (Figure 1), altered fetal movements and reduced amniotic fluid volume are also noted, adding to the complexity of the case. Five days after the ultrasound, the fetus died in utero, and the patient underwent an induction and fetal autopsy combined with a psychological care for the patient.

At pathology, the fetus had marked scoliosis of the spine (Figure 2) and a 3.5-cm nuchal hygroma (Figure 3).

Bilateral talipes deformity and rocker-bottom feet were present. The heart and left lung were externalized, whereas the right lung remained within the thorax. The abdominal organs were normally formed. The right kidney was absent, and the left kidney was dysplastic. A midline defect that extended down to and involved the bladder and external genitalia was present. An imperforate anus was also present. The umbilical cord extended from the defect and measured 4 cm in length. The trophoblast was fragmented (10 g in weight) with an attached umbilical cord remnant.

Discussion

Short Umbilical Cord Syndrome (SUCS) is also referred to as a body stalk anomaly or limb-body wall complex. The diagnosis of SUCS is made when 2 out of 3 of the following anomalies are present: Exencephaly or an encephalocele, a thoracic and/or abdominal wall defect, a thoraco- and/or abdominoschisis, and limb anomalies [1].

Other internal anomalies such as cardiac anomalies, hypoplasia of the lungs, absence or hypoplasia of the diaphragm, gut malrotation or atresia, absence of the gallbladder, renal agenesis or dysplasia, and genital abnormalities have also been reported [2].

This complex lesion is thought to occur due to a vascular insult resulting in a short or absent umbilical cord and disruption of the lateral body wall, face, cranium, spine, and limbs. These abnormalities may occur as isolated defects or in combination. Several researchers have proposed possible causes for this anomaly [3]. One hypothesis is that alterations in blood flow led to disruption and incomplete development of embryonic tissue due to hemorrhagic necrosis and anoxia during the fourth through sixth week of development. This disruption in blood flow results in a large spectrum of defects to the developing embryo, such as adhesion to the amnion and persistence of the extra-embryonic coelom [4]. Others speculated that this syndrome is a result of early amnion disruption or an early error in embryonic development. SUCS is also thought to result from rupture of the amnion between the third and fifth week of embryogenesis. Others have postulated that these defects are produced by either vascular disruption or mechanical compression [5].

The thoraco-abdominoschisis of SUCS is characterized by an anterolateral body wall defect with evisceration of the thoracic and or abdominal organs. The eviscerated organs remain in a short amniotic sac, limited anteriorly by the placental chorion and posteriorly by the fetal retroperitoneum. This results in the amnion being continuous with the fetal skin. Thoraco-abdominoschisis occurs more commonly on the left side (64%). The umbilical cord is often short and abnormal. Scoliosis occurs in 77% of cases [6]. The severe scoliosis usually appears L or U shaped and is due to the tight fixation of the fetus to the placenta due to the abnormally short umbilical cord. Midfacial clefts associated with encephalocele, exencephaly, and holocranium occur in 40% of cases as a consequence of early vascular disruption. Alpha-fetoprotein levels are elevated; however, the karyotype is usually normal.

Sonographically, a complex mass located lateral and anterior to the fetal chest or abdomen is usually appreciated. Continuity of the amnion to the margins of the body wall should imply a diagnosis of SUCS. Our case is unique in that the fetus also had a cystic hygroma. also unique to our case is the very early diagnosis of the SUCS [7].

The association of these malformations usually results in a disorganized mass composed of membranes and organs that, in conjunction with the oligohydramnios, may be very hard to evaluate

sonographically. Amnioinfusion may be helpful in some cases. Differential diagnosis includes pentalogy of Cantrell, omphalocele, or gastroschisis. However, severe scoliosis and limb abnormalities in the setting of an abdominal wall defect is usually consistent with short umbilical cord syndrome [6]. The prognosis for SUCS is poor if all components are present.

While this case report contributes valuable insights into the antenatal diagnosis and management of SCS, it is essential to acknowledge certain limitations. Firstly, as SUCS is a rare condition, the reported case represents a single instance, limiting generalizability. Additionally, the case report does not provide a comparison with other management approaches or a control group, which could have offered further insights into the efficacy of the interventions employed [5].

In conclusion, the presented case report sheds light on the significance of antenatal diagnosis and personalized management strategies in pregnancies affected by Short Umbilical Cord Syndrome. Early detection through routine ultrasound examinations enables healthcare providers to develop tailored management plans, closely monitor maternal and fetal well-being, and make informed decisions based on the severity of the fetal prognosis [7].

Conclusion

This case report highlights the significance of antenatal diagnosis and individualized management in optimizing pregnancy outcomes for patients with Short Umbilical Cord Syndrome (SUCS). Early detection through early first trimester routine ultrasound examinations allows for timely interventions and close monitoring, minimizing potential risks associated with ongoing severe SUCS and allowing potential actions to improve fetal prognosis in mild cases.

References

1. Callen PW. Ultrasound evaluation of fetal abdominal wall defects. In: Callen PW, editor. Ultrasonography in obstetrics and gynecology. 3rd Ed. Philadelphia, PA, WB Saunders Company. 1994. P. 385-7.
2. Saunders RC. Structural fetal abnormalities: The Total Picture. St Louis, MO, Mosby-Yearbook. 1996.
3. Patten RM, Van Allen MI, Mack LA, Wilson D, Nyberg D, Hirsch J, et al. Limb-body wall complex: in utero sonographic diagnosis of a complicated fetal malformation. Am J Roentgenol. 1986;146:1019-24.
4. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. Am J Med Genet. 1987;28:529-48.
5. Miller ME. Structural defects as a consequence of early intrauterine constraint: Limb deficiency, polydactyly, and body wall defects. Semin Perinatol. 1983;7:274-7.
6. Ballantyne J. Antenatal pathology and hygiene. Edinburgh, UK, Wm. Green & Sons, 1904.
7. Moerman P, Fryns J, Vandenberghe K, Lauweryns J. Constrictive amniotic bands, amniotic adhesions, and limb-body wall complex: Discrete disruption sequences with pathogenic overlap. Am J Med Genet. 1992;42:470-79.