International Journal of Health Sciences
September 2016, Vol. 4, No. 3, pp. 15-18
ISSN: 2372-5060 (Print), 2372-5079 (Online)

Published by American Research Institute for Policy Development
DOI: 10.15640/ijhs.v4n3a3
URL: https://doi.org/10.15640/ijhs.v4n3a3

# Congenital Malformations of a Fetus in a Single Horn of a Bicornuate Uterus: A Case Study

**Brittany Stanek, PA-S[1], Justin David, PA-S[2] & Daniel Coovert, PhD[3]***

**Abstract**

A bicornuate uterus, or "heart-shaped" uterus, occurs secondary to incomplete fusion of the Müllerian ducts and lack of resorption of the uterine septum around 5 weeks of gestation. It is estimated that about 1-3% of women have some degree of uterine malformation, with bicornuate uteri consisting of about 25% of those anomalies. Pregnancy in a bicornuate uterus has been shown in correlation with recurrent miscarriage, preterm labor, and malpresentation of the baby. We follow a case of a 29 year old female with a known bicornuate uterus through her second pregnancy. She presented with live intrauterine pregnancy without complication until 16 weeks gestation when a congenital malformation was found via ultrasound. Serial ultrasounds revealed three large defects, which lead to the patient electing for dilation and evacuation at 20 weeks gestation due to a low chance of survival of the fetus. There is some research that shows a higher rate of birth defects in mothers with a bicornuate uterus. It is unclear whether or not the congenital malformations of the fetus were correlated to the bicornuate uterus of the mother.

**Keywords:** Bicornuate uterus, gastroschisis, fetal malformations

## 1. Introduction

Congenital uterine anomalies are defined as any malformation of the uterus that occurs during fetal development. Many sources report about 1-3% of women to have some degree of uterine malformation (Bermejo et al., 2009; Raga, 1997). The most common uterine anomaly is a bicornuate uterus, accounting for about 25% of all anomalies (Bermejo et al. 2009). A bicornuate uterus, or "heart shaped" uterus, occurs due to incomplete fusion of the Müllerian ducts, and further lack of resorption of the retained septum. When there is failure in fusion, it results in a bicornuate uterus that is heart shaped with a retained septum dividing the uterus into two separate horns (Bermejo et al., 2009; Carlson, 2004).

Most cases go unnoticed until recurrent miscarriages occur. About 36% of all pregnancies within a bicornuate uterus will end in a miscarriage, and about 25% of women with a bicornuate uterus suffer from recurrent pregnancy loss (Matsaseng et al., 2012).

## 1. Case Report

A 29 year old female, G2P0 with known history of having a bicornuate uterus. Past medical history is significant for asthma and GERD, taking albuterol and omeprazole. In 2012, the patient presented to her OBGYN complaining of irregular menses.

[1] Department of Applied Medicine & Rehabilitation, Indiana State University, Terre Haute, Indiana, USA.
[2] Department of Applied Medicine & Rehabilitation, Indiana State University, Terre Haute, Indiana, USA..
[3] Department of Applied Medicine & Rehabilitation, Indiana State University, Terre Haute, Indiana, USA.

Previously, while on oral contraceptives, she had regular menses with cycle about 28-30 days lasting about 5-6 days. At this time, she was having spotting in between her cycles. Her urine pregnancy test was positive in the office and her first beta-human chorionic gonoadotropin (beta-HCG) was 18 mIU/mL. Measured again 48 hours later, the level remained at 18 mIU/mL. When measured again 48 hours later, however, her level decreased to less than 3 mIU/mL. Her physician then concluded she had miscarried. In order to confirm the diagnosis, a transvaginal ultrasound was performed and she was found to have a bicornuate uterus.

Two years later, the patient presented again to her OBGYN desiring to become pregnant, and an MRI of her uterus was ordered. The MRI showed a bicornuate uterus measuring 7.6cm longitudinal by 3.8cm AP by 5.6cm transverse. The uterus is tilted to the right of the midline. Both horns are equal in size with a 2cm septation between horns. The endometrium was measured at 4mm in thickness and was within normal range (Figure 1).

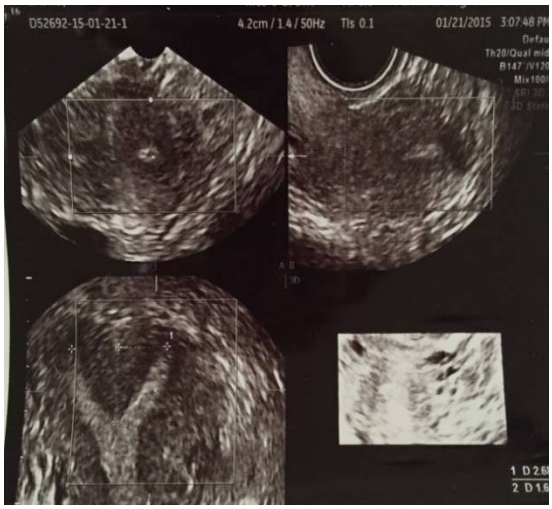


**Figure 1:** Transvaginal ultrasound showing bicornuate uterus

The patient obtained pregnancy with last menstrual period 5/16/15, making her estimated due date 2/23/16. She was referred to a maternal fetal medicine specialist to be followed more closely. The patient had her first ultrasound at 9 weeks gestation confirming intrauterine pregnancy in the left horn of the bicornuate uterus (Figure 2). The antenatal period was uneventful and the patient's cervical length was checked bimonthly for cervical incompetence until 16 weeks gestation. At this point, the patient received her second ultrasound, and there was found to be an abdominal wall defect with suspected gastroschisis. Follow up ultrasound was performed at 18 weeks gestation confirming gastroschisis. There was also what appeared to be a diaphragmatic hernia. Repeat ultrasound again was performed at 19 weeks (Figure3), which confirmed the hernia, and found that the fetus had a congenital heart defect known as truncus arteriosus. With the combination of these three congenital defects, the maternal fetal medicine specialists gave the baby a very low percent chance of making it full term and a very poor prognosis of living after birth. The patient elected to have a dilation and evacuation in early October, 2015.

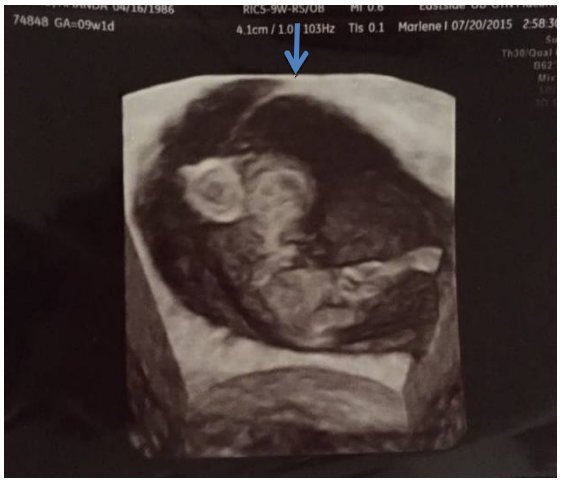


**Figure 2:** ultrasound at 9 weeks gestation showing intrauterine pregnancy in left horn of the uterus. Arrow indicates the intrauterine septum.

The postoperative period was uneventful and then patient was sent home the same day. The fetus was sent for cytogenetic and mutational analysis. Cytogenetic results showed an absence of chromosomal defects. Genetic mutational analysis did not reveal any mutations in the genes most commonly associated with the observed congenital defects. Both the patient and her husband then underwent genetic screening, which turned out to be negative. The patient was counseled to sustain two normal menstrual cycles before again trying to conceive. She was instructed to continue taking her prenatal vitamins and she to double her amount of folic acid to 4mg.

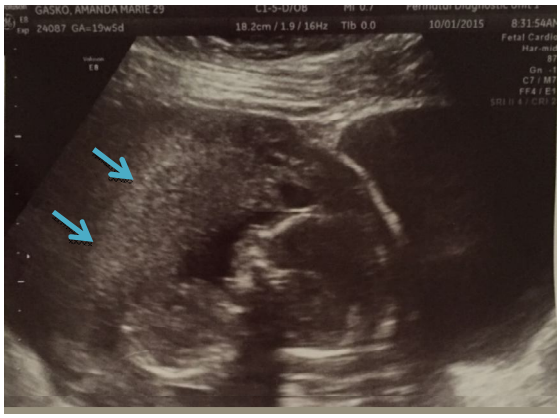


**Figure 3:** ultrasound at 19 weeks gestation showing gastroschisis. Arrows corresponds to abdominal contents outside the abdominal wall. Asterisk indicates fetal cranial vault

## 2. Discussion

Having the condition of a bicornuate uterus has been associated with recurrent miscarriage, preterm labor and malpresentation of the baby (Matsaseng and Kruger 2012). There is very limited research that links bicornuate uteri to fetal deformity. The patient followed had one previous miscarriage that may have been caused by the bicornuate uterus. Many times when a bicornuate uterus is present, there is a rudimentary horn and a fully developed horn. If the fetus implants into the rudimentary horn, there is a much higher chance of miscarriage. In the case of the patient reviewed, both of her uterine horns were equal in size as mentioned above. It is unclear whether or not the miscarriage was secondary to the bicornuate uterus (Matsaseng and Kruger 2012).

The fetus in this case first presented with gastroschisis. This is an abdominal wall defect that allows for abdominal contents to protrude out of the abdominal cavity. This defect usually occurs around 5-8 weeks of gestation when the abdominal wall muscles are forming. It is usually not noticed until much later when the contents begin to form outside of the body wall. The defect requires immediate surgery after birth to repair the defect, and place the abdominal contents back into the cavity (Jones et al., 2016). The second defect noted to the fetus was a diaphragmatic hernia. Failure of a portion of the diaphragm leads to abdominal contents entering into the chest cavity. This can put pressure on the heart and lungs causing fetal distress (Aggarwal et al., 2016). Lastly, the third defect, and most severe found in the fetus was truncus arteriosus. This deformity is very rare and occurs in only 1% of male fetuses (Nourzad and Baghershiroodi, 2013). Persistent truncus arteriosus is defined as the lack of separation of the aorta and pulmonary artery past 4 weeks of gestation. Complications of this defect include cyanosis at birth, heart failure, and fetal demise (Nourzad and Baghershiroodi, 2013).

It is unclear whether or not the bicornuate uterus had an effect on the pregnancy in review. Throughout her pregnancy, her cervical length was checked and was never found to be incompetent, nor did she have any signs of decreased space in the uterus. One large study by Martinez-Frias et al., studied the risk for congenital anomalies in infants of mothers with a bicornuate uterus (Martinez-Frias et al., 1998). This study sampled 26,934 infants with congenital malformations using the Spanish Collaborative Study of Congenital Malformations surveillance system. Each mother was then categorized into having a bicornuate uterus or not having a bicornuate uterus. Results showed that mothers with bicornuate uteri had 4 times higher rate of fetal malformation than mothers with normal uteri. Specifically, five fetal defects were found significantly higher in bicornuate uterus mothers including limb deficiencies, nasal hypoplasia, teratoma, omphalocele, and acardia-anencephaly. It is difficult to determine the pathogenesis of the link between maternal uterine malformations and fetal defects (Martinez-Frias et al., 1998).

Although the fetus presented in this case did not have any of the defects found in the case control study, it is unclear whether or not she had a higher risk of fetal birth defects due to her bicornuate uterus. It is not unreasonable to conclude that having a bicornuate uterus predisposes women to having fetuses with malformations after reviewing the study. More research should be performed though to support this case control study, and to determine the percentage of risk for mothers with uterine anomalies.

## 3. References

Aggarwal A, Lohani R, Suresh V. (2016). Case series on anesthesia for video-assisted throacoscopic surgery for congenital diaphragmatic hernia in children. *Anesthesia: Essays and Researches*, 10(1): 128-131.

Bermejo C, Cantarero R, Diaz D, et al. (2009). Three-dimensional Ultrasonography in the Diagnosis of Müllerian Duct Anomalies. *Donald School Journal of Ultrasound in Obstetrics and Gynecology*, 3(1): 21-30.

Carlson BM. (2004). *Human Embryology and Developmental Biology*. St. Louis, MO: Mosby.

Jones AM, Isenburg J, Salemi JJ, et al. (2016). Inreasing Prevalence of Gastroschisis – 14 States, 1995-2012. *Morbidity and Mortality Weekly Report*, 65(2): 23-26.

Martinez-Frias ML, Bermejo E, Rodriguez-Pinilla E, et al. (1998). Congenital Anomalies in the Offspring of Mothers With a Bicornuate Uterus. *Pediatrics*, 101(4): 1-3.

Matsaseng T, Kruger TF. (2012). Laparoscopic Strassman's metroplasty for bicornuate uterus- is it relevant? *SAJOG*, 18(3): 85-87.

Nourzad G, Baghershiroodi M. (2013). A case report of truncus arteriosus communis and genetic counseling. *ARYA Atheroscler*, 9(4): 254-259.

Raga F, Bauset C, Remohi J, et al. (1997). Reproductive impact of congenital Müllerian anomalies. *Human Reproduction*, 12(10): 2277-2281.