Case Report

Complex Mullerian Malformation: A Rare Case of Hypoplastic Noncavitated Uterus in the Middle with Two Rudimentary Horns on Either Side

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Mullerian anomalies which cause infertility in women were described by different classification systems. We report a rare case of uterine anomaly in a 16-year-old patient presented with primary amenorrhea. Her diagnostic laparoscopy findings revealed two uterine rudimentary horns on either side of the upper pelvis with a hypoplastic noncavitated central uterus. The pathogenesis of this anomaly may not be clearly defined but it was stated that these occur due to the developmental defects in embryo. This case report is one of the rarest cases presented and may signify the Mullerian duct anomaly.

Keywords: Hypoplastic uterus, Mayer–Rokitansky–Kuster–Hauser, Mullerian anomaly, rudimentary horn

INTRODUCTION

Mullerian anomalies are rare and usually found in 1 of 4500 female cases of primary amenorrhea and 2%–8% cases of infertile women. They are mostly undiagnosed till the time of menarche. The anxious mother brings the child to the clinic to know the cause of her primary amenorrhea. Sometimes, married couples come with a history of infertility when this diagnosis is made.

Mullerian ducts differentiate into the fallopian tubes, uterus, and the upper part of the vagina during the intrauterine phase. When interruptions are found in the development of these Mullerian ducts, we encounter various malformations. These range from complete agenesis, hypoplasia, and fusion defects such as unicornuate uterus with or without a rudimentary horn, uterine didelphys, complete or partial bicornuate uterus, complete or partial septate uterus, and arcuate uterus. These anomalies are associated with normal functioning ovaries and normal appearing external genitalia. Segmental or complete agenesis or hypoplasia may involve the vagina, cervix, fundus, tubes, or any combination of these structures.

Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome is the most common example in this category. We now report a rare variant of MRKH seen with midline hypoplastic noncavitated uterus.

CASE REPORT

A young unmarried 16-year-old girl was brought to our center by her anxious mother on November 19, 2016, with a history of primary amenorrhea. Parents were married by third-degree consanguinity. Her younger sister attained menarche at 13 years of age. On examination, her breast and external genitalia looked normal with scanty pubic hair (Tanner’s Stage 3). Following the conservative approach, we did not do vaginal examination. She weighed 57.8 kg with a height of 154 cm.

Her transabdominal scan done on November 19, 2016, at our hospital showed a hypoplastic uterus measuring 3.4 cm × 1.4 cm, right ovary – 3.2 cm × 1.8 cm, and left...
ovary – 2.9 cm × 2.3 cm. We could not visualize the two rudimentary horns. Her karyotyping at our center was normal (46XX). Her hormonal analysis revealed normal levels of luteinizing hormone and follicle-stimulating hormone. She was put on cyclical hormones for 3–4 months, but there was no withdrawal bleeding following which she did not come for follow-up.

She came back to us after 3 years on June 2, 2019, with her two earlier ultrasonography reports done outside. The first on November 13, 2015 showed a hypoplastic uterus with normal looking ovaries and normal vagina, and the second on November 14, 2016 revealed uterine aplasia with bilateral rudimentary noncavitary horns with both ovaries normal, and on transrectal ultrasound, the vagina and rudimentary cervix were visualized. We were surprised to see the USG report of 2016 [Figure 1]. Hence, we decided to perform a diagnostic laparoscopy on June 7, 2019, which revealed a small hypoplastic uterus in the midline with two rudimentary horns on either side like MRKH with normal looking ovaries and tubes [Figures 2-5]. Her anti-Mullerian hormone, estradiol (E2), prolactin, and thyroid hormone levels were within normal limits.

The uterus in the middle had uterosacral ligaments on both sides with spread out ligament on the right side. On either side of the noncavitated uterus, there were two fleshy thick tube-like structures, at the end of which there were rudimentary horns on both sides with normal looking ovaries and tubes. This portion of anatomy looked like MRKH. Pelvic ultrasonography did not reveal any functioning endometrium in any of these structures. There was no family history of such condition, nor did her mother take any medicines or made any attempt to terminate this pregnancy. The etiology and pathophysiology of this anomaly is not known but looks like a variant of MRKH and Mullerian dysgenesis due to error in intrauterine development of the Mullerian system.

After adequate counseling and informed consent from the couple, we report the third case of a 16-year-old with hypoplastic noncavitated uterus in the middle with two rudimentary horns on either side (a variant of MRKH with noncavitated uterus in the middle).

**RESULTS**

This rare case of MRKH with noncavitated nonfunctional uterus in the middle is the second of its
kind as mentioned in literature. It is generally said that at the gestational age of 45–49 days, the Mullerian duct system does not grow further for reasons unknown. This occurrence is sporadic, etiology being unknown.

**Discussion**

Organogenesis of the female reproductive system is initiated from urogenital ridges which are developed at the 5th week of gestation. The incidence of Mullerian duct anomalies is between 0.001% and 10%.[5] Mullerian duct anomalies such as early developmental failure during 5 weeks of gestation may cause agenesis or hypoplastic uterus. Mullerian duct develops at 7 weeks of gestation. Fusion of Mullerian duct and formation of the uterus occurs at 10 weeks of gestation. This fusion is seen in the midline and progresses from the caudal portion to the cranial portion. The unfused parts of the Mullerian duct (cranial part) form fallopian tubes. Complete or partial developmental failure of the Mullerian duct may result in noncavitary rudimentary horns, which is one of the possible subtypes of unicornuate anomaly. The classification system of uterine anomalies is based on a single theory.[6] As ovaries develop from the primitive ectoderm and not from the Mullerian duct, there may not be abnormality in the female sexual development. Moreover, in the hypoplastic uterus, the organ is seen fully differentiated.[7-9]

In the literature, the first case was reported in the *Journal of Human Reproduction* by Nezhat and Smith, 1999. A nulligravid woman aged 18 years presented with severe dysmenorrhea was examined, and her diagnostic laparoscopy findings revealed unicorneuate uterus with two cavitated, noncommunicating rudimentary uterine horns stacked one on top of the other.[10] The second case was reported in the same journal by Sadik *et al.*, 2002. A 26-year-old female presented with primary amenorrhea, and infertility was evaluated, and her laparoscopic findings revealed hypoplastic noncavitated uterus and two rudimentary horns.[11] Controversy exists between genetic and familial etiology. It is a food for thought to consider a multifactorial pathogenesis, yet to be discovered.[12] There are two other recently reported cases of Mullerian duct anomaly, one with a longitudinal vaginal septum, two cervices, a uterine septum, and a single normal fundus was diagnosed during cesarean delivery following natural conception[13] and the other with a 23-year-old female with a partial uterine septum and a double cervix.[14]

**Conclusion**

We have come across many cases of MRKH syndrome, several of them who got married have come and had vaginoplasty for sexual function. Many such women came back to us for infertility, and some of them conceived through *in vitro* fertilization and embryo transfer through a surrogate with their own gametes. These children are all normal. Assisted reproduction is a boon to those who are unfortunately born with Mullerian anomaly, ovarian dysgenesis, premature ovarian failure, Turner’s syndrome, unicornuate uterus, and other such conditions. If the awareness is created among all doctors, such patients can be helped to complete their family.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published, and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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