International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Mehrotra R et al. Int J Reprod Contracept Obstet Gynecol. 2013 Dec;2(4):683-686
www.ijrcog.org
pISSN 2320-1770 | eISSN 2320-1789

DOI: 10.5455/2320-1770.ijrcog20131239

Case Report

Mullerian and gonadal dysgenesis: an unusual presentation

Ragini Mehrotra[1]*, Kamini Tyagi[2], Amita Yadav[2]

[1]Department of Obstetrics & Gynaecology, AIIMS Bhopal, M.P., India
[2]Department of Obstetrics & Gynaecology, MLN Medical College, Allahabad, U.P., India

Received: 2 September 2013
Accepted: 21 September 2013

***Correspondence:**
Dr. Ragini Mehrotra,
E-mail: raginimehrotra@gmail.com

ABSTRACT

The estimated prevalence of a unicornuate uterus is rather low, approximately 0.3% of the whole population, 0.6% of the infertile population, and 0.2% of the fertile population. Of the mullerian defects, however, unicornuate uterus is found in 3 to 13% of women. Ovarian maldescent occasionally occurs in patients with a normal uterus, the incidence is reported to be 20% when the uterus is absent (Rokitansky-Kustner - Hauser syndrome) and as high as 42% in cases of unicornuate uterus. The occurrence of Mullerian anomalies with concomitant gonadal developmental abnormalities is very rare. Case reports about the unicornuate uterus with ovarian abnormalities are very limited in the literature. We present a case of 17 year old girl with primary amenorrhoea and hematometra, for which she was operated and later on she was diagnosed as a case of vaginal and cervical atresia with unicornuate uterus together with ipsilateral ovarian agenesis and contralateral ectopic ovary.

Keywords: Unicornuate uterus, Mullerian anomalies, Ovarian maldescent, Hematometra, Rokitansky- Kustner-Hauser syndrome

INTRODUCTION

Uterine malformations are the result of major disturbances in the development, formation or fusion of the paramesonephric (mullerian) ducts during fetal life. The unicornuate uterus is caused by the non-development of one mullerian duct or the failure of the contralateral side to migrate to its proper location. The unicornuate uterus covers a wide range of anatomical entities. Possibilities are a unicornuate uterus with a communicating or non-communicating rudimentary horn, a rudimentary horn without uterine cavity, and the isolated unicornuate uterus. The estimated prevalence of a unicornuate uterus is rather low, approximately 0.3% of the whole population, 0.6% of the infertile population, and 0.2% of the fertile population[1]. Of the mullerian defects, however, unicornuate uterus is found in 3 to 13% of women.[1,2,3] The presence of a unicornuate uterus is associated with increased obstetric complications like early miscarriage, ectopic pregnancy, abnormal fetal presentation, intrauterine growth retardation, and premature labor. Undescended or ectopic ovaries are characterized by the attachment of their upper pole to an area above the level of the common iliac vessels. Although ovarian maldescent occasionally occurs in patients with a normal uterus, the incidence is reported to be 20% when the uterus is absent (Rokitansky-Kustner - Hauser syndrome) and as high as 42% in cases of unicornuate uterus. Bilaterality occurs more often in women with congenital absence of the uterus.[4] Despite the well-known association of undescended ovaries and unicornuate uterus, ovarian maldescent is reported only sporadically, suggesting the possibility that many cases go unrecognized.[5] The occurrence of Mullerian anomalies with concomitant gonadal developmental abnormalities is very rare. Case reports about the unicornuate uterus with ovarian abnormalities are very limited in the literature. We present a case of a unicornuate uterus together with ipsilateral ovarian agenesis and contralateral ectopic ovary.

We describe here the embryological background of mullerian duct anomalies and undescended ovaries.

Embryology of mullerian duct anomalies and ectopic ovaries

During the indifferent stage of duct development, between the 5th and the 8th week of pregnancy, two pairs of ducts appear and coexist in all embryos, the mesonephric ducts or Wolffian ducts and the paramesonephric ducts, better known as the mullerian ducts. Male differentiation is instigated by the presence of a single factor encoded on the Y chromosome; female differentiation occurs in its absence. In the absence of mullerian-inhibiting substance which is produced by the sertoli cells in the developing testis, the paired mullerian ducts ultimately develop into the structures of the female reproductive tract. The structures include the fallopian tubes, uterus, cervix, and upper part of the vagina. The ovaries and lower part of the vagina have separate embryologic origins not derived from the mullerian system. Complete formation and differentiation of the mullerian ducts into the different parts of the female reproductive tract depend on completion of three phases of development, i.e. organogenesis, fusion, and septal resorption. Lateral fusion is the process during which the lower segments of the paired mullerian ducts fuse to form the uterus, cervix, and upper vagina. Bicornuate uterus and didelphys uterus are the result of a failure of fusion. The term vertical fusion occasionally is used to refer to fusion of the ascending sinovaginal bulb with the descending mullerian system. Complete vertical fusion forms a normal patent vagina, while incomplete vertical fusion results in an imperforate hymen. After the lower mullerian ducts fuse, a central septum is present, which subsequently must be resorbed to form a single uterine cavity and cervix. Failure of resorption is the cause of a septate uterus. Ovaries and the lower vagina are not derived from the mullerian system. If both the mullerian ducts do not develop fully, uterine agenesis or hypoplasia is found. A unicornuate uterus is the result of a non-development of one mullerian duct. Depending on the status of the rudimentary horn, four-basic classifications of a unicornuate uterus can be made - (1) no rudimentary horn (2) no cavity (3) communicating and (4) non-communicating. During the third month of fetal life, the ovaries descend from the posterior abdominal wall near the kidneys to the point just inferior to the pelvic brim. The descent is guided by the gubernaculum, a cord of mesenchyme connected to the lower pole of each gonad. Undescended ovaries are uncommon and mostly associated with an absent uterus (Rokitansky-Kustner-Hauser syndrome) or a unicornuate uterus.

CASE REPORT

17 year old girl who had not attained menarche presented with cyclical pain in lower abdomen for one year. She had enlarged uterus corresponding to 16 weeks size gravid uterus. Ultrasonography showed hematometra and absence of both the ovaries. She was operated for hematometra drainage. Low vaginal septum was excised, vaginal space was dissected but the cervical bulge was not having any opening. Another septum was excised and both the lips were visualised but there was no opening like external os. Cervical canal was not negotiated with dilators. Immediate decision for exploratory laparotomy was taken. Operative findings showed unicornuate enlarged uterus with ipsilateral (Right) ovarian agenesis (Figure 1). On left side uterus was attached to pelvic wall with fibrous connective tissue. Left tube, round ligament, broad ligament and left ovary were not found. Hysterectomy was done. We were unable to find out ectopic ovary, kidneys and both the ureters were normally present.

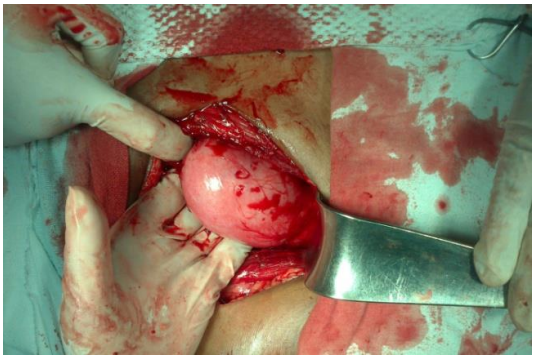

Figure 1: Intraoperative view of unicornuate uterus showing smooth right side without tube, ovary and any peritoneal fold while left side is attached with the pelvic wall also showing no tube and ovary.

The cut specimen showed small amount (10 ml) of dark chocolate colored collected blood into the uterine cavity. Endometrium was apparently normal. Uterine walls were thick (i.e. myohyperplasia). Tubal ostium was present on left side, from inside the cavity which was communicating only upto half of thickness of myometrium (Figure 2). On right side no tubal ostium was present, internal os was present but cervical canal was not patent (Figure 3). On 5th postoperative day she was sent for MRI in the search of ectopic ovary and MRI was suggestive of ovary in left inguinal canal and near the lumbar vertebra (Figure 4).

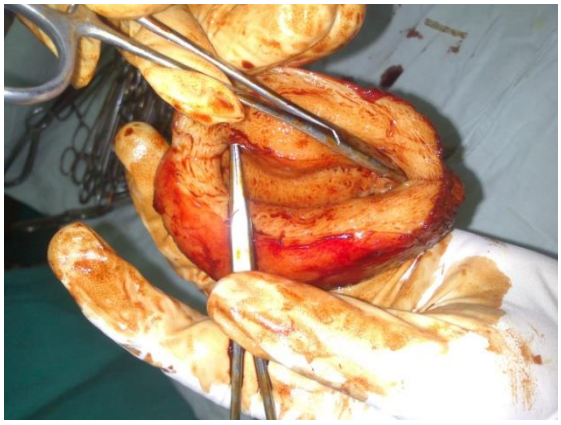

Figure 2: Cut specimen of uterus showing blind left tubal ostium and atretic cervical canal.

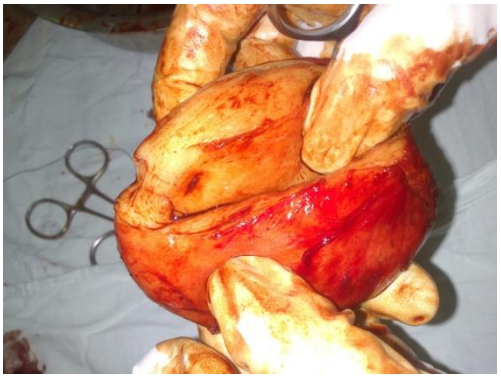

Figure 3: Cut specimen of uterus showing blind left tubal ostium.

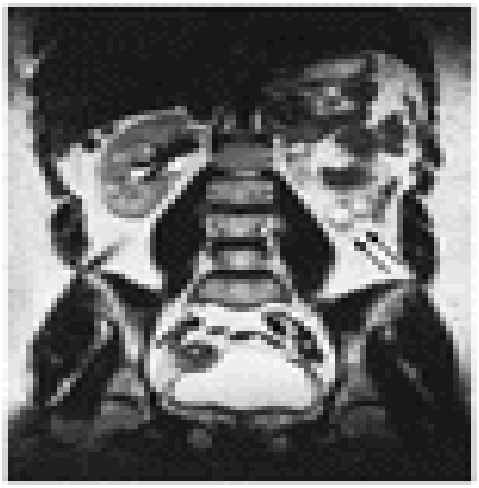

Figure 4: Coronal T_2- weighted MRI scan showing a left sided ectopic ovary in inguinal canal.

DISCUSSION

A classification based on the degree of failure of normal development of the female genital tract was proposed in 1979 by Buttram and Gibbons[6] and modified in 1988 by the American Society for Reproductive Medicine. According to this classification, the unicornuate uterus represents class II. This malformation is the result of defective development of one of the two Mullerian ducts**.** Four subtypes of this anomaly have been described: communicating horn, noncommunicating horn, horn without cavity, and a unicornuate uterus without a horn. Our case was the last one. In the original study, Buttram and Gibbons reported that 31% of their patients with uterine abnormality have urinary anomalies in which congenital absence of a kidney was the most common. Urinary tract abnormalities occur more frequently in class I and class II uterine anomalies than with those in classes III, IV, or V.[7] But we have not found any congenital renal anomaly in our case. Pedro Acien[2], in his report in Human Reproduction in 1992, suggested that a unicornuate uterus may also be caused by complete agenesis of all or the some organs derived from one urogenital ridge, resulting in a unicornuate uterus and, on the contralateral side, no uterine horn or ovaries and renal agenesis or hypoplasia.[8] Therefore, the absence of one ovary in our case may be explained by the abnormal development of organs derived from unilateral urogenital ridge. Other ovary was found ectopic with the help of MRI. Different terms have been used to describe an undescended or ectopic ovary. Lachman and Berman[9] were the first to introduced usage of the term ectopic ovary which can be divided in three categories: (1) postsurgical implant, (2) postinflammatory implant, and (3) true embryologic ectopic ovary. From an embryological point of view, the presence of ectopic ovaries can be explained by a lack of caudal descent of the gonads into the true pelvis or by a retarded differential growth of that portion of the urogenital ridge which gives rise to both the gonads and the fallopian tube. During the third month of fetal life, the developing ovaries descend from a position near the kidneys to their final position in the true pelvis. Undescended ovaries may be unilateral or bilateral and can be associated with abnormalities of the mullerian ducts such as unicornuate uterus. In this case an MRI was performed and ectopic ovary was found in the inguinal canal. Although the prevalence of unicornuate uterus with undescended ovary is low, a correct diagnosis is mandatory. It is important to realize that reproductive and menstrual performance is jeopardized when uterine malformations and ectopic ovary are involved. The knowledge of the existence of an ectopic ovary was also of importance and relevance in this case because hematometra was diagnosed ie endometrium of unicornuate uterus was under normal cyclical hormonal changes. MRI has proven to be the best imaging method to explore an ovary in an anomalous position and to document associated malformations.[10,11,12] Our results indicate that MRI in cases of unicornuate uterus will detect undescended ovaries more frequently than is appreciated so far. This elegant method seems to be superior to ultrasound.

CONCLUSION

We presented a very rare clinical condition that demonstrates a unicornuate uterus without a horn, and ipsilateral ovarian agenesis with contralateral ectopic ovary concomitantly.

REFERENCES

1. Raga F, Bause C, Remohi J et al. Reproductive impact of congenital Mullerian anomalies. Hum Reprod 1997;12:2277-81.
2. Acien P. Incidence of Müllerian defects in fertile and infertile women. Hum Reprod 1997;12:1372-6.
3. Grimbizis GF, Camus M, Tarlatzis BC, et al. Clinical implications of uterine malformations and hysteroscopic treatment results. Hum Reprod Update 2001;7:161-74.
4. Dabirashrafi H, Mohammad K, Moghadami-Tabrizi N. Ovarian malposition in women with uterine anomalies. Obstet Gynecol 1994;83:293-4.

5. Verkauf BS, Bernhisel MA. Ovarian maldescent. Fertil Steril 1996;65:189-92.
6. Buttram VC, Gibbons WE. Mullerian anomalies: a proposed classification (an analysis of 144 cases). Fertil Steril 1979;32:40–6.
7. Buttram VC, Reiter RC. Surgical treatment of infertile female. Baltimore: Williams & Wilkins, 1985:149.
8. Acien P. Embryological observations on the female genital tract. Hum Reprod 1992;7:437.
9. Lachman MF, Berman MM. The ectopic ovary. A Case Report and Review of the Literature. Arch Pathol Lab Med 1991;115:233-5.
10. Marten K, Vosshenrich R, Funke M, et al. MRI in the evaluation of mullerian duct anomalies. Clin Imaging 2003;27:346-50.
11. Trinidad C, Tardaguila F, Fernandez GC et al. Ovarian maldescent. Eur Radiol 204;14:805-8.
12. Troiano RN, McCarthy SM. Mullerian duct anomalies: imaging and clinical issues. Radiology 2004;233:19-34.

DOI: 10.5455/2320-1770.ijrcog20131239
Cite this article as: Mehrotra R, Tyagi K, Yadav A. Mullerian and gonadal dysgenesis: an unusual presentation. Int J Reprod Contracept Obstet Gynecol 2013;2:683-6.