CASE REPORT

MULLERIAN AGENESIS – A REPORT OF THREE CASES

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ABSTRACT: Agenesis is a rare abnormality of paramesonephric ducts, which result in complete absence of uterus and fallopian tubes. In this article, cases of absent development of Mullerian ducts are presented, followed by a brief discussion of etiology, clinical presentation, diagnosis and management of the patient in Mullerian agenesis. Differential diagnosis in a patient presenting with primary amenorrhoea and normal external genitalia is discussed.

INTRODUCTION: Mullerian agenesis is a Class I anomaly of development of Mullerian ducts in which Uterus, Fallopian tubes, Cervix and lower two thirds of the vagina are not developed ². It presents with primary amenorrhoea with or without vaginal atresia (Mayer-Rokitansky-Küster-Hauser syndrome). However, the ovarian development is not hindered because of the different embryological pathways for the development of ovaries and Mullerian ducts.

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Case 1: A 14yr girl was referred to the department of Radiology with primary amenorrhoea. She had no cyclical abdominal pain.

On examination, she had normal secondary sexual characters. She had female external genitalia. Pervaginal examination was not done.

Patient was advised abdomino pelvic sonography.

In pelvic sonography, uterus and cervix could not be visualized. Abdominal sonography showed normal kidneys.

Patient was taken up for MRI of the abdomen and pelvis. Axial, coronal and sagittal planes including T1, T2, T2 STIR & Fat Sat, 3D TRUFISP sequences were obtained.

Uterus and cervix were not visualized. However, distal two third of vagina was present. Left ovary was visualized and is normal and right ovary was not visualized (Patient has a history of pelvic surgery). Both kidneys showed normal appearance. No hydronephrosis noted.

Karyotyping was not done due to non-affordability of the patient.

Fig 1: T2 turbo spin echo image showing absence of uterus and cervix. UB- Urinary bladder, R- Rectum.

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Fig 2: T2 STIR coronal image showing the presence of left ovary. UB – Urinary bladder; OV – left ovary.

Case 2: A 22 Yr girl presented with primary amenorrhoea and was worked up in the similar manner. On examination, she had normal secondary sexual characters. She had female external genitalia. Pervaginal examination was not done. Abdominal and pelvic sonography showed normal kidneys, non-visualized uterus and cervix. Both ovaries are visualized and showed normal sized follicles in them.

MRI abdomen and pelvis showed absent uterus and cervix. T2 weighted sequences showed normal kidneys. No evidence of hydronephrosis noted. Karyotyping was not done.

Fig.3: T2 turbo spin echo sagittal image showing absence of uterus and cervix. UB – Urinary bladder; R – Rectum.

Fig 4: T2 STIR axial image showing both ovaries.
UB – Urinary bladder; ROV – Right ovary;
LOV – Left ovary

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Case 3: A 22 Yr old female patient was referred to the department of Radio diagnosis with a complaint of primary amenorrhoea and was worked up in the similar manner.

On examination, she had normal secondary sexual characters. She had female external genitalia. Pervaginal examination was not done.

Abdominal and pelvic sonography showed non-visualized uterus and cervix. However, only right kidney was visualized. Left kidney was not seen in the renal fossa. Both ovaries were visualized. Left ovary was anechoic. (? Follicular cyst)

MRI abdomen and pelvis showed absent uterus and cervix. T2 weighted sequences showed normal right kidney. Left kidney was not visualized in the abdomen and pelvis. No evidence of hydronephrosis noted in the right kidney.

Karyotyping was not done.

Fig. 5: T2 STIR coronal image showing absence of uterus and vagina.
UB – Urinary bladder; R – Rectum.

Fig. 6: T2 sagittal image showing both ovaries.
Left ovary is cystic.
ROV – Right ovary; LOV – Left ovary

Fig. 7: T2 STIR coronal image showing absent left kidney and compensatory hypertrophy of right kidney.
RK – Right kidney.

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DISCUSSION: The frequency of congenital absence of the vagina and uterus is not entirely clear, although reported incidences vary from 1 in 4,000 to 5,000 female births to 1 in 20,000 hospital admissions.

In women presenting with primary amenorrhea, the disorder is fairly common, second only to gonadal dysgenesis as a cause¹.

Mullerian anomalies are grouped as class 1 genital abnormalities according to the recommendations of the American Fertility Society².

This class further is subdivided into two types³:

Type A: symmetrical lack of development in the Mullerian ducts

Type B: the lack of development is asymmetrical. Type B is also associated with ovarian and renal anomalies.

Etiology: The etiology of Mullerian agenesis is unknown. The fetal gonad has the potential to differentiate into either a testis or an ovary. The critical factor that signals the bipotential gonad to develop into a testis is a gene called the H-Y antigen, located on the short arm of chromosome Y. The Sertoli cells of the fetal testis produce a substance known as anti-Mullerian hormone (also known as Mullerian-inhibiting factor or Mullerian-inhibiting substance) that causes the Mullerian structures to regress in utero, so that in the presence of testis, the female internal genital structures will not develop.

Therefore, after 6 weeks gestation, the absence of Mullerian-inhibiting factor in the female fetus promotes bidirectional growth of the paired Mullerian ducts along the lateral aspect of the gonads in conjunction with simultaneous regression of the mesonephric ducts. Interruption of Mullerian duct development during this time gives rise to aplasia or hypoplasia of the vagina, cervix, or uterus⁴.

Clinical presentation: The woman with Mullerian agenesis has normal external genitalia and normal functioning ovaries. The phenotype, therefore, is normal female with normal secondary sexual characteristics. The degree of malformation of the organs involved in the Mayer-Rokitansky-Kuster-Hauser syndrome varies. The fallopian tubes may be normal and the uterus vestigial or vice-versa. In 30-50% cases, there is an associated renal anomaly³.

Management: General physical examination commonly reveals a normal phenotypic woman with well-developed secondary sexual characteristics. However, vaginal examination may either reveal a short or absent vagina.

In evaluating patients with suspected Mullerian agenesis, the first investigation should be pelvic and renal tract ultrasonography because of its simplicity and low cost. If the ultrasonographic findings are indeterminate or incomplete, an MRI should be performed, as the information is much more precise⁵.

Urinary tract abnormalities must be excluded because they may be associated with Mullerian agenesis. This should be done by ultrasound scan, MRI, CT scan, or intravenous urogram,

Hysterosalpingography has no role in Class I Mullerian duct anomalies.

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Differential diagnosis: The main differential diagnosis of Mullerian agenesis is testicular feminization syndrome. The karyotype in this condition is, however, 46XY. In addition, the hormone profile in Mullerian agenesis will be typically that of a woman. Failure to obtain a karyotype, therefore, may miss the diagnosis of testicular feminization syndrome with the resultant retention of testes that could become malignant later in life⁶.

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