CASE REPORT

MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME

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Mayer-Rokitansky-Küster-Hauser Syndrome is a congenital malformation in which there is failure of the Müllerian ducts to develop resulting in absent uterus and fallopian tubes and variable malformations of the vagina. Ultrasonography reveals absence of uterus with normally visualised ovaries.

Keywords: Mayer-Rokitansky-Küster-Hauser Syndrome, Müllerian agenesis, Congenital, 46XX

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INTRODUCTION

Müllerian agenesis, also called Mayer-Rokitansky-Küster-Hauser Syndrome (MRKH) is a congenital malformation characterised by a failure of the Müllerian ducts to develop, resulting in missing uterus and fallopian tubes and variable malformations of the upper portion of the vagina.[1] It is the second most common cause of primary amenorrhoea after gonadal failure (such as from Turner syndrome). Its incidence is 1 in 4,000 to 5,000 female births and it affects all races equally.[2] An individual with this condition is hormonally normal; that is, they will enter puberty with development of secondary sexual characteristics. However they discover the condition during puberty, as the menstrual cycle does not start (primary amenorrhea). Their chromosome constellation is 46XX. Ovaries are intact and ovulation usually occurs.

CASE REPORT

A 16-year-old girl came to Gynaecology Outpatient Department with complaint of primary amenorrhoea. She came for a medical check-up since her mother wanted to get her daughter married off.

On examination, patient had normal general physical examination. There was no acne. She had normal breast development (Tanner stage V) and axillary hairs were present. Genital examination showed adult pubic hair (Tanner stage V), and normal female external genitalia. She had normally placed urethra and a vaginal opening. Patient refused per speculum examination. Trans-abdominal ultrasound revealed an absent uterus. She had normal sized ovaries with small follicles in it and vagina was also present (Figure-1).

On the basis of ultrasound a diagnosis of absent uterus was made keeping in view the MRKH syndrome. Both kidneys were normal on ultrasound with no agenesis or ectopia. There was no musculoskeletal abnormality or hearing problem. Patient's hormonal profile like serum LH, FSH, progesterone and testosterone levels were within normal limits. MRI pelvis showed normal sized ovaries with follicles in it bilaterally with normal fallopian tubes and vagina but absent uterus and cervical tissue. The patient and her mother were counselled and made aware about her chances of having a normal married life but inability to bear children.

DISCUSSION

Müllarian agenesis, commonly referred to as MRKH Syndrome, is a rare disorder, which was first reported in the 1830s. The condition is named after August Franz Joseph Karl Mayer, Carl Freiherr von Rokitansky, Hermann Küster, and GA Hauser.[3] At 5 weeks gestation, the Müllerian ducts stop developing. The uterus, cervix, and upper two thirds of the vagina form from the fused caudal ends of the Müllerian ducts. Fallopian tubes develop from the un-fused upper ends; the renal system simultaneously develops from the Wolffian (i.e., mesonephric) ducts. Ovarian function is preserved because the ovaries originate within the primitive ectoderm, independent of the mesonephros.

Aetiology may be either mutation in a major developmental gene or a limited chromosomal imbalance. The recent study shows that MRKH is caused by heterozygous mutation of the WNT4 gene on chromosome 1p36.[4] This gene is responsible for the development of the paramesonephric duct, the embryonic precursor to female reproductive organs, such as the cervix, Fallopian tubes, ovaries, and much of the vagina. Without the WNT4 gene, the Müllerian duct is either deformed or absent, meaning that it never develops into the female reproductive organs.

There are two different types of this syndrome. Type I is characterised by an isolated absence of the proximal two thirds of the vagina, whereas type II is marked by other malformations including vertebral, cardiac, urologic and otologic anomalies. Of those suffering from the latter form, 40% will have kidney abnormalities, 10% will have hearing problems, and 10-12% will have skeletal abnormalities. Strubbe *et al*[5] also suggested these two separate subgroups. The term GRES (genital, renal, ear, skeletal) syndrome was suggested for the latter type.

Trans-abdominal ultrasonography is a simple and non-invasive method, and must be the first investigation. It reveals an absence of the uterus. However, a quadrangular retro-vescical structure may be wrongly identified as a hypoplastic or juvenile uterus. It corresponds to the vestigial lamina located underneath

the peritoneal fold, where utero-sacral ligaments attach. The vestigial lamina shows no cavity, no hyper-echogenic line, which normally corresponds to the uterine mucous membrane.[6] Renal malformations, must be systematically evaluated during this scan. However, ultrasonography is an operator-dependent technique that can fail at identifying anatomical structures of the pelvis.

MRI is a non-invasive technique that provides a more sensitive and more specific means of diagnosis than ultrasonography. It is increasingly becoming an essential diagnostic tool complementary to laparoscopy. The uterine aplasia is best characterized on sagittal images, while vaginal aplasia is best evidenced on transverse images. T2W imaging in the coronal plane is helpful for ovarian localisation. Moreover, MRI can be used at the same time to search for associated renal and skeletal malformations. Consequently, MRI facilitates the surgical approach, e.g. vaginal reconstruction. But also by the different reproductive potential and psychological impact.[7]

The management includes psychological counselling, as patient might suffer from anxiety, depression and face lot of psychological distress at diagnosis. Behavioural problems of the adolescent patients can be avoided by early appropriate guidance and reassurance. The choice of procedure and patient age at reconstruction depend upon individual anatomy, fertility potential and psychological and social factors. Treatment which consists in constructing a neovagina is generally offered to patients who have vaginal atresia and when they are ready to start sexual activity. Laparoscopy is not only useful for diagnosis of uterine malformations but can also be valuable for any treatment required for this type of malformation along with creation of an artificial vagina.[8]

Moreover, everyday improvement of medical technologies allows, in many countries, women to appeal for *in vitro* fertilization and surrogate pregnancy to bypass the absence of inner genital tract.

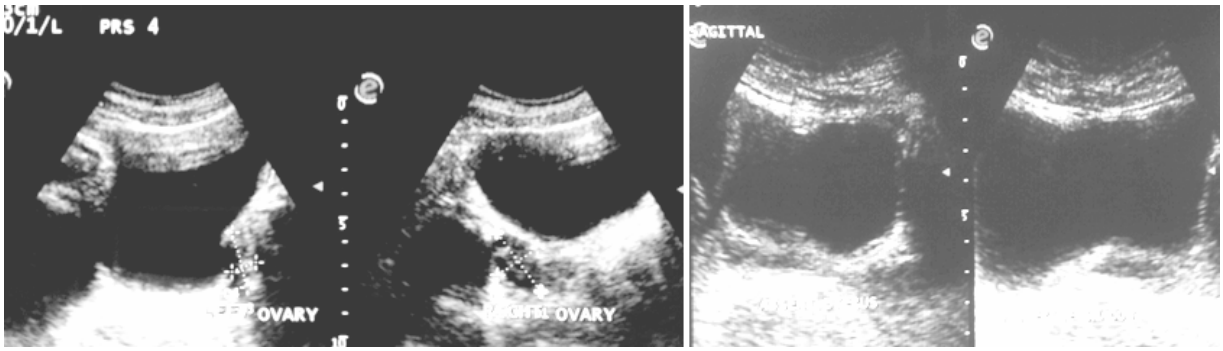

Figure-1: Ultrasound showed absent uterus behind the urinary bladder, normal sized ovaries. Vagina was present

CONCLUSION

MRKH is a congenital malformation with absence of uterus with normally visualized ovaries. Ultrasonography is a simple and non-invasive method, and must be the first investigation. It reveals absence of uterus with normally visualized ovaries. MRI is more sensitive and specific for further evaluation.

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