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MAYER–ROKITANSKY–KUSTER–HAUSER SYNDROME: SYNDROME OF MULLERIAN AGENESIS – A REPORT OF TWO CASES

Gynaecology

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ABSTRACT

The Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH syndrome), also called Rokitansky syndrome or vaginal aplasia of the uterus, is a congenital developmental disorder that is characterized by the absence of the uterus and vagina with normal ovaries and the external genitalia . It is seen in at least 1 out of 4500 women. MRKH may be isolated (Type I), or may be associated with renal, vertebral, and to a lesser extent, auditory and cardiac defects (MRKH Type II).Primary amenorrhoea is the commonest presenting feature seen in this clinical condition with normal secondary sexual characteristics. Here, i report two cases of MRKH syndrome, one in a 18-year-old unmarried female who presented with primary amenorrhea and the other in a 28-year-old female with primary amenorrhea and primary infertility.

KEYWORDS

Mayer–Rokitansky–Kuster–Hauser, amenorrhoea, aplasia, Mullerian, infertility .

INTRODUCTION

Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH syndrome) is named after its discoverer Baron Karl von Rokitansky (Czechoslovakia, 1804–1878), a physician and professor at the University of Vienna. In 1829 and in 1838, Mayer–Rokitansky described a syndrome that is associated with agenesis of the uterus and vagina, while Kuster then observed a correlation with urological defects. Therefore, this condition is also known as MRKH syndrome. Primary amenorrhea is the commonest presentation associated with this syndrome. Primary amenorrhea is defined as failure of attaining menarche by 13 years of age in the absence of secondary sexual characteristics or by 15 years of age in the presence of secondary sexual characteristics (1). There are various causes of primary amenorrhea, including primary ovarian insufficiency, outflow disorders of the female genital tract, Mullerian developmental anomalies, hypothalamic and pituitary disorders, and receptor abnormalities or enzyme deficiencies. MRKH syndrome is the second most common cause of primary amenorrhea after gonadal pathology(2). Herein, I report two cases of MRKH syndrome, one in a 18-year-old unmarried female who presented with primary amenorrhea and the other in a 24-year-old female with primary amenorrhea and primary infertility.

Case Reports

Case 1

18 years old unmarried female presented to the department of Gynecology at a tertiary care centre with the chief complaint of absence of menstruation. On examination , external genitalia was normal on inspection. Secondary sexual characteristics were within normal limits. For further workup, pelvic sonography was advised which revealed non visualised uterus with normal appearing bilateral ovaries. Therefore, MRI pelvis was done which also revealed non visualised uterus, cervix and upper vagina with normal bilateral ovaries (Fig 1 a & 1b). There was no other associated anomaly seen in this case. Therefore, diagnosis of MRKH (Type I) was made.

Case 2

24 years old married female presented to the department of Gynecology at a tertiary care centre with the chief complaint of inability to conceive. She had not attained menarche yet. There was no history of cyclical abdominal pain. On examination the external genitalia and the secondary sexual characteristics were within normal limits. The pelvic sonography was advised which revealed non visualised uterus and the ovaries. For further workup MRI pelvis was done which showed the non visualised uterus, cervix and upper vagina with normal bilateral ovaries (fig 2a & 2b). No associated anomalies were seen in this case. Therefore, diagnosis of MRKH (Type I) was made.

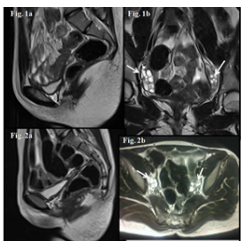

Fig 1a & b show the T2WI sagittal & coronal sections which reveal the absence of uterus, cervix & upper vagina with normally visualised bilateral ovaries (white arrows) in a 18 yr old female. Fig 2 a & b are the T2WI sagittal and axial sections of the 24 yr old female patient showing non visualised uterus, cervix and upper vagina with normal bilateral ovaries (white arrows).

DISCUSSION

Müllerian agenesis, eponymously referred to as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a spectrum of congenital anomalies of unknown etiology characterized by a variable degree of utero-vaginal agenesis in female with normal secondary sexual characteristics and a 46, XX karyotype. It is caused by interrupted normal development of the Müllerian duct system, which normally forms the uterus, cervix and the upper two-thirds of the vagina, during the fifth and sixth weeks of gestation (3). MRKH syndrome is generally divided into two types, with type I being seen in isolated cases of utero-vaginal agenesis, while the more common type II is seen in cases of utero-vaginal agenesis associated with extra-genital anomalies, including urologic (e.g. renal agenesis, pelvic kidney, and horseshoe kidneys), skeletal, auditory and cardiac anomalies. The most severe form of type II MRKH syndrome is Müllerian hypoplasia, renal agenesis, cervicothoracic somite dysplasia (MURCS) (4). Primary amenorrhoea is the commonest presentation. Pelvic sonography is often the first diagnostic test in the evaluation of patients with MRKH syndrome and can confirm the presence of ovaries and the absence of a uterus (5). However, due to technical difficulties, the results can sometimes be inconclusive(5).MRI is the imaging modality of choice for the confirmation of diagnosis and is also useful in the identification of any associated malformations with this clinical condition(6,7). Preibsch et al. have shown an excellent correlation between MRI and laparoscopy findings in patients with MRKH syndrome (8). The diagnosis of MRKH syndrome causes a significant psychological burden on patients because of the associated infertility (5). Treatment options include progressive vaginal dilators or surgical creation of a neovagina. Assisted reproductive techniques and surrogacy may be options for fertility in these cases (9).

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