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Case Report

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MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME WITH GONADAL DYSGENESIS: CASE REPORT

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ABSTRACT

This is a report of 18 year old teenage girl with primary amenorrhoea and underdeveloped secondary sexual characters. The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome with gonadal dysgenesis, also referred as müllerian agenesis or aplasia is caused by embryologic underdevelopment of the müllerian duct, with resultant agenesis or atresia of the vagina, uterus, or both. It is characterized by congenital aplasia of the uterus and the upper two thirds of the vagina in women showing under development of secondary sexual characteristics and a normal 46, XX karyotype.[1-2] The incidence of Mayer-Rokitansky-Kuster-Hauser syndrome was not clearly established, but studies indicate a variation of 1/4,000 and 1/5,000 live births of the female sex.[2]

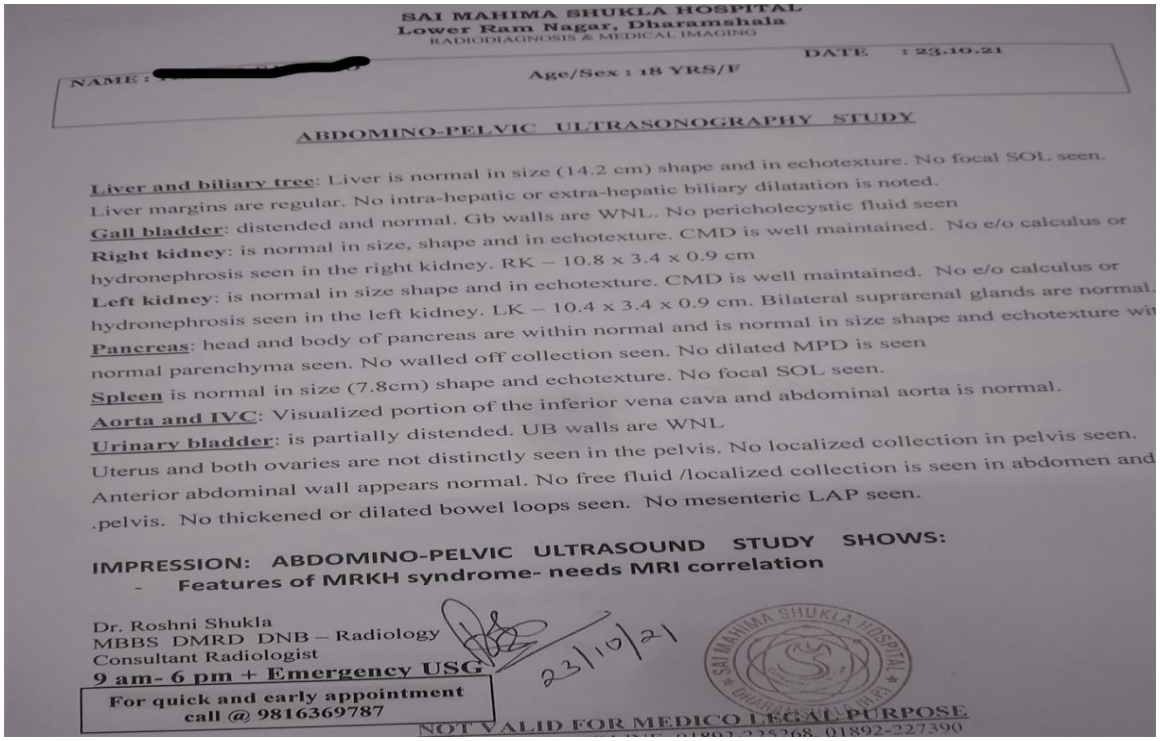
KEYWORDS: Mayer-Rokitansky-Küster-Hauser, Mullerian duct, Primary amenorrhoea.

INTRODUCTION

Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH syndrome) is named after its most famous 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 includes agenesis of the uterus and vagina, while Kuster then observed a correlation with urological defects. For this reason, this condition is also known as MRKH syndrome.[1] Type I (isolated) MRKH is less frequent than Mullerian duct aplasia, Renal dysplasia, and Cervical Somite anomalies (MURCS) association. Polycystic ovaries and ovarian tumors have been described in association with MRKH Type II.[1-2] Congenital malformations of female genitalia are often a challenge for doctors, requiring a great knowledge of embryonic development of the genital tract due to the wide variety of possible diagnosis. We report here a rare case of 46XX phenotype with gonadal dysgenesis and mullerian agenesis.

CASE PRESENTATION

We report a case of 18-year-old girl who presented with no onset of menstrual cycle (primary amenorrhea). At clinical examination, the patient demonstrated development of secondary sexual characteristics not compatible with her chronological age. Pubic, axillary hair growth and breast development were scored, respectively, Tanner stages II and III. The patient had no evidence of facial dysmorphism, webbing of the neck or skeletal abnormalities. She has no history of chronic diseases and denies excessive exercise, medications, anorexia, clinical hypothyroidism or hyperandrogenism. No family history of amenorrhea. Her mother attained menarche at the age 14 years. On physical examination: weight 52 kg, height of 1.60m, female phenotype, symmetrical breasts, the genital examination hairiness gynecoid, rudimentary clitoris, immature labia majora and minora. An endocrine study including pituitary, ovarian, and thyroid evaluation was performed and revealed hypergonadotrophic hypogonadism (Luteinizing Hormone: 20.62 IU/L, Follicle Stimulating Hormone: 65.40 IU/L, Oestradiol: 24.24 IU/L) with normal level of prolactin and Thyroid Stimulating Hormone. At gynaecological examination a grooved urethra with elevated edges was observed. Vaginal and speculum examination was not performed. Pelvic ultrasonography: uterus and both ovaries are not distinctly seen in the pelvis (Figure 1). On magnetic resonance imaging (MRI) T2- weighted image of pelvis shows the absence of uterus and ovaries in pelvis.



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Lower Ram Nagar, Dharamshala
RADIODIAGNOSIS & MEDICAL IMAGING

NAME : [redacted] DATE : 23.10.21
Age/Sex : 18 YRS/F

ABDOMINO-PELVIC ULTRASONOGRAPHY STUDY

Liver and biliary tree: Liver is normal in size (14.2 cm) shape and in echotexture. No focal SOL seen. Liver margins are regular. No intra-hepatic or extra-hepatic biliary dilatation is noted.
Gall bladder: distended and normal. Gb walls are WNL. No pericholecystic fluid seen
Right kidney: is normal in size, shape and in echotexture. CMD is well maintained. No e/o calculus or hydronephrosis seen in the right kidney. RK – 10.8 x 3.4 x 0.9 cm
Left kidney: is normal in size shape and in echotexture. CMD is well maintained. No e/o calculus or hydronephrosis seen in the left kidney. LK – 10.4 x 3.4 x 0.9 cm. Bilateral suprarenal glands are normal.
Pancreas: head and body of pancreas are within normal and is normal in size shape and echotexture wit normal parenchyma seen. No walled off collection seen. No dilated MPD is seen
Spleen is normal in size (7.8cm) shape and echotexture. No focal SOL seen.
Aorta and IVC: Visualized portion of the inferior vena cava and abdominal aorta is normal.
Urinary bladder: is partially distended. UB walls are WNL
Uterus and both ovaries are not distinctly seen in the pelvis. No localized collection in pelvis seen. Anterior abdominal wall appears normal. No free fluid /localized collection is seen in abdomen and .pelvis. No thickened or dilated bowel loops seen. No mesenteric LAP seen.

IMPRESSION: ABDOMINO-PELVIC ULTRASOUND STUDY SHOWS:
- Features of MRKH syndrome- needs MRI correlation

Dr. Roshni Shukla
MBBS DMRD DNB – Radiology
Consultant Radiologist
9 am- 6 pm + Emergency USG
For quick and early appointment call @ 9816369787

23/10/21

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NOT VALID FOR MEDICO LEGAL PURPOSE

Figure 1: Ultrasonographic report showing MRKH Syndrome.

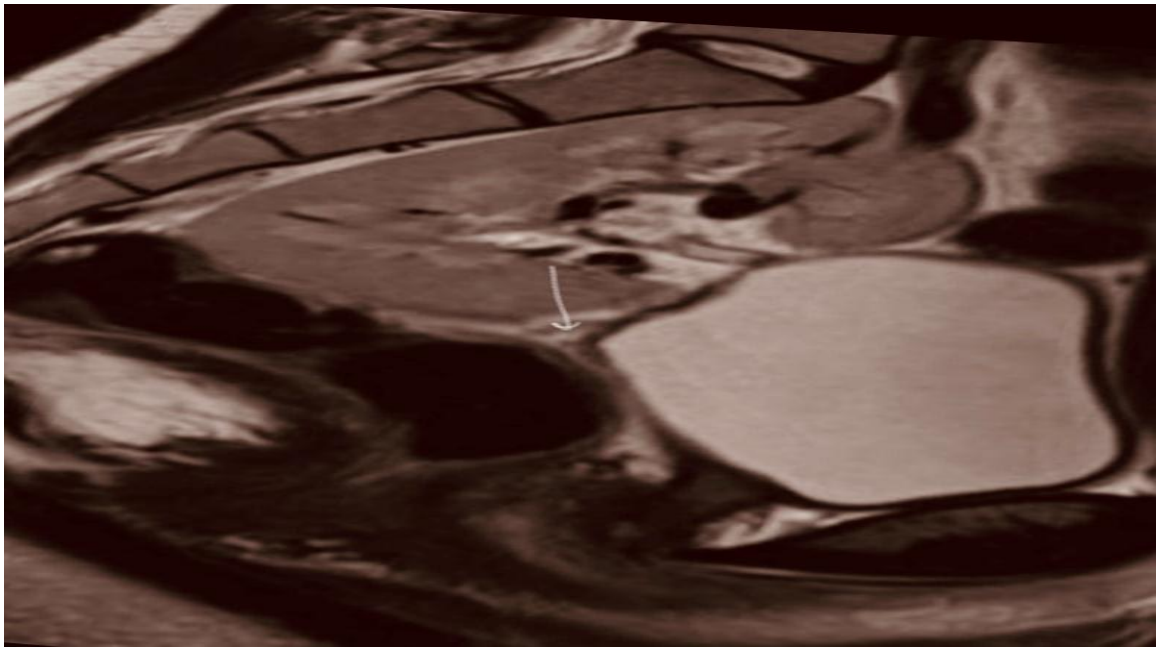

Figure 2: T2- weighted image of pelvis shows the absence of uterus and ovaries in the magnetic resonance imaging of pelvis.

DISCUSSION

Patients with MRKH syndrome typically present during adolescence with primary amenorrhea defined as absent menstrual periods at the age of 16 following normal puberty and development of secondary sex characteristics. Other complaints at referral include dyspareunia/apareunia and (cyclic) abdominal pain. Patients may be referred after an incidental finding of vaginal or uterus agenesis, but if examined by imaging at young age such findings may be false interpretations of the prepubertal uterus. Median age at referral has been reported to be 17.5 years (interquartile range: 16–19).[3]

The normal development of the female reproductive tract depends on the interaction between genetic, hormonal and environmental factors for the differentiation of the Müller and Wolff ducts, and the urogenital sinus.[4] Changing these factors can result in a wide spectrum of abnormalities of the reproductive tract, including imperforate hymen, vaginal agenesis or atresia,

incomplete fusion of the Müllerian ducts and Müllerian aplasia.[5] There may be various forms of Müller duct abnormalities, ranging from small anatomical variations to total aplasia. The most common cause of abnormalities is the Mayer-Rokitansky-KusterHauser syndrome (MRKH). It affects up to 90% of women with vaginal agenesis.[4-5] Gonadal dysgenesis with female phenotype is defined as the absence or incomplete development of ovaries. It is the most common cause of primary amenorrhea. 46,XX gonadal dysgenesis is relatively rare form of gonadal dysgenesis and wide variations are seen in its clinical presentation.[6] The association of gonadal dysgenesis and MRKH syndrome is extremely rare. In the present case, both gonadal agenesis and mullerian agenesis existed in the same patient.

Ultrasound examination 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.[7] However, due to technical difficulties, the results can sometimes be inconclusive.[7] MRI is the imaging modality of choice for the confirmation of diagnosis and is also useful in the identification of any associated malformations.[8] Preibsch et al. have shown an excellent correlation between MRI and laparoscopy findings in patients with MRKH syndrome.[9]

The diagnosis of MRKH syndrome has a significant psychological burden on patients and family because of the associated infertility.[7] The distress can be decreased by psychological counseling and support groups. Treatments include progressive vaginal dilators or surgical creation of a neovagina. Assisted reproductive techniques and surrogacy may be options with regard to fertility.[10]

CONCLUSION

The association of gonadal dysgenesis and Mayer Rokitansky kuster hauser syndrome is very rare and appears to be coincidental, independent of chromosomal anomalies. The patient and family was given all information regarding MRKH syndrome as well as treatment options regarding infertility.

Conflict of interest: All authors declare they have no conflict of interest.

Financial disclosure: Nil.

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