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CASE REPORT Open Access

Clinical and radiological presentation of familial Mayer-Rokitansky Küster-Hauser syndrome in three sisters with literature review

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Abstract

Background: Mayer-Rokitansky Küster-Hauser syndrome MRKHS represents class I of congenital Müllerian anomaly, which resulted from interruption of embryonic development of the paramesonephric ducts in early pregnancy. It is characterized by uterine and proximal vaginal aplasia/hypoplasia associated with variable degree of cardiac, renal, and skeletal anomalies.

We aimed to review and analyze clinically and radiologically MRKHS relying on three young sisters' cases who presented with primary amenorrhea and were found to have features of MRKHS.

Case presentation: Three sisters aging 17, 20, and 25 years old presented with primary amenorrhea. Clinical workup was performed followed by ultrasound and MRI of the abdomen and pelvis, spine X-ray, audiogram, echocardiogram, hormonal study, and karyotyping. The three sisters had normal sex hormones and mature secondary sexual characteristic features. Additionally, cardiac valvular regurgitation and renal hypoplasia were recognized. Cytogenetic confirmed normal female 46 XX karyotype. MRI showed variable size and appearance of Müllerian remnant tissue of the uterus and proximal vagina.

Conclusion: MRKHS shows variable size and appearance of Müllerian remnant structures; however, it seems that the smaller the volume of remnant tissue the more severe associated anomalies. Associated valvular cardiac disease is documented, which was not reported before.

Keywords: Mayer-Rokitansky Küster-Hauser syndrome (MRKHS), Müllerian duct Primary amenorrhea, Congenital anomalies, Karyotype

Background

Mayer-Rokitansky-Kuster-Hauser syndrome MRKHS, also known as Mullerian agenesis, is a rare congenital disorder that occurs in females. It is caused by embryologic underdevelopment of the Mullerian duct, resulting in an absent or underdeveloped uterus and upper part of the vagina [1]. Females with MRKH have normal 46, XX female karyotype with normal development of secondary female sexual characteristics and external genitalia, as the functional ovaries are present, but absent menstruation [2].

Although a rarity, with an incidence of 1:5000 live female births, it is considered one of the most common causes of primary amenorrhea [1, 2].

The reproductive abnormalities of MRKH syndrome results from incomplete development of the Mullerian duct which normally develops into the uterus, cervix, fallopian tubes and upper two-thirds of the vagina at the age of 6–8 weeks of gestation [3].

There are two types of MRKHS. Type I is classified as only the reproductive organs are affected whereas type II is classified if other systems are involved. The kidneys

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might be abnormally formed, positioned, and/or a kidney may be absent. Hearing loss, heart, or skeletal abnormalities particularly of the vertebral column are other associated anomalies [3, 4].

The underlying pathogenesis of this condition is due to a combination of genetic and environmental factors such as exposure to chemicals during early pregnancy, medication, smoking, or viral illness. Although specific factors are often unknown, the increased number of familiar aggregates raises the hypothesis of genetic cause [5–8]. Most cases are sporadic; however, in this case series, we are reporting and analyzing a rare condition of familial occurrence of MRKHS in three sisters, and we believe there are few similar cases all over the world. We have also reviewed the literature on MRKHS in between sisters using PubMed search also the MRI radiological findings of 128 cases.

Case presentation

Three sisters aged 17, 20, and 25 years, presented to the gynecology clinic with primary amenorrhea. There was no family history of congenital anomalies or similar problems. They have a fourth sister with a normal menstrual cycle and is married with two kids. Their parents are cousins. Their mother was healthy and had her menarche at 12 years of age. She had unremarkable past obstetric history and was not exposed to drugs, radiation, or other environmental factors antenatally and all were delivered vaginally.

Extensive radiological, lab, and genetic investigations were requested to look for other associated abnormalities. The investigations included an ultrasound of the Abdomen and pelvis, MRI pelvis, X-ray spine, karyotyping, and hormonal study for (LH, FSH, prolactin, TSH, estrogen, testosterone, and progesterone) audiogram and cardiac echogram.

Case 1

A 17-year-old female patient, a secondary school student, presented to the gynecology clinic with primary amenorrhea and cyclic colicky abdominal pain. The patient was diagnosed at 15 years of age to have cardiomyopathy. She had a height of 159 cm, and a weight of 104 kg, and her BMI was 41. She was pale and tired. Her breasts were well developed at Tanner stage V in addition to normal pubic and axillary hair development. Gynecological examination revealed normal external genitalia while speculum examination was not performed, as the patient is single. Her hormonal profile (FSH, LH, prolactin, TSH, estradiol, progesterone, and testosterone) was within normal limits. Serum urea, creatinine, blood sugar, and liver enzymes were also normal. An abdominal ultrasound examination showed an absent uterus with normal ovaries. She then underwent abdomen and pelvic MRI, which showed normal ovaries with an absent uterus and proximal vagina (rudimentary remnants) and normal kidneys. Echocardiography revealed moderate left ventricular hypertrophy LVH, grade I diastolic dysfunction, ejection fraction EF 45%, dilated four chambers, grade II mitral regurgitation MR, grade I tricuspid and aortic regurgitation TR/AR. A cytogenetic evaluation revealed 46, XX karyotype, thus confirming the diagnosis of MRKH syndrome. Audiogram was normal. The patient and her parents were counselled about the findings and implications on infertility.

Case 2

A 20-year-old female patient, a college student, presented with primary amenorrhea. The patient has cyclic colicky abdominal pain. She was admitted to the hospital at 17 years of age with meningoencephalitis. At that time, an incidental pelvic ultrasound finding showed an absent uterus. Her height was 163 cm, weight 124 kg and BMI was 46.7. Detailed history and clinical examination were performed. The patient demonstrated normal secondary sexual characteristics with normal breast, axillary and pubic hair development. Gynecological examination revealed normal female external genitalia on inspection, while speculum examination was not performed, as the patient was single. Her hormonal profile (FSH, LH, prolactin, TSH, estradiol, progesterone, and testosterone) was within normal limits. Serum urea, creatinine, blood sugar, and liver enzymes were also within normal. Trans-abdominal examination showed an absent uterus, while both ovaries were present and polycystic. Abdomen and pelvic MRI examination was also conducted and showed normal ovaries with good volume, but the uterus and the vaginal canal could not be visualized (rudimentary remnants). The kidneys were normal. An echocardiogram showed good systolic and diastolic function. EF 60% with mild grade I TR and MR. The audiogram was normal.

Cytogenetic testing revealed a normal female chromosome pattern (46, XX), thus confirming the diagnosis of MRKH syndrome. The patient and her parents were counselled regarding the findings and implications on fertility and childbearing.

Case 3

A 25-year-old female patient, unemployed, and only finished intermediate school, presented with primary amenorrhea that was not properly investigated previously, in addition to cyclic colicky abdominal pain and recurrent urinary tract infection. Her height was 160 cm, weight was 108 kg, and BMI was 42. Detailed history and clinical

Table 1 MRKHS MRI findings of five large sample sized studies in comparison to our study

Authors	Bhayana A et al. 2019 [4]	Boruah, Deb Kumar et al. 2017 [5]	Hall-Craggs et al. 2013 [6]	Yoo R et al. 2013 [2]	Kara, Taylan et al. 2013 [15]
Sample size	25	11	61	15	16
Lateral bud	Rt: present in 23/25 Lt: present in 18/25 It is caudal and anterior to ovaries Volume is 0.5	Bilateral in 9/11 Rt bud was 2.26 ± 3.3 left bud was 1.27 ± 1.1	54 were bilateral, and 7 were unilateral	All has bilateral bud Volume was 2.64 ± 0.65	–
Paramedian/midline bud:	18, 20/25	3/11		In 87% (13/15) of the cases	Uterine hypoplasia or remnant uterus was detected in 11 patients (68.8%)
Fibrous bands	25/25	7/11	–	All	–
Vagina	Proximal is rudimentary and the distal is present in 25/25	Upper 2/3rd is absent in nine patients	Absent vagina in 22 patients (33%). The mean vaginal length was 2.0 cm (range, 1.0–6.5 cm)	The upper two-thirds was absent in all cases, whereas the lower one-third was present in 93% (14/15) of the cases	
Ovaries	25/25	11/11 present, one patient has ectopic in the Lt inguinal canal and Rt iliac fossa The mean volume of Rt ovary was 4.74 ± 2.58 and Lt ovary was 4.65 ± 2.2	Bilateral ovaries were present in 54 patients Ectopic ovaries were found in 27 patients. The ectopic ovaries were mainly in the pelvis. The range of ovarian volume was 1.0–33.1 mL, with a mean volume of 10.1 mL	All patients had bilateral ovaries located in the pelvis	Ovaries were seen in 10 patients (62.5%), 2/10 were ectopic
Renal	Agenesis of kidney: 3/25 Ectopic pelvic kidney 1/25			Unilateral renal agenesis	Renal agenesis 2 Horseshoe kidney 1 Ectopic kidney 2 Urachal remnant 1
Spine	Lumbosacral transition: 2/25 sacral agenesis 1/25 Scoliosis 1/25			Four cases: (sacralization of lumbar vertebra in 3 cases, mild scoliosis in one case)	-L5 spina bifida -Bilateral hemisacralization, -Roto-scoliosis in only one case -Coccygeal vertebral fusion1
Cardiac	–	–	–	–	–
Type	Type I 19/25, type II 6/25	Type I 8/11, type II 3/11		Type I 9/15 Type II 4/15	Type I 10/16 Type II 6/16

Table 2 Anomalies associated with type II MRKHS[a]

Site of anomaly	Description	Associated risk
Renal anomalies	Renal agenesis, dysplasia, hypoplasia, and ectopia	Infection Stone. Hydronephrosis
Skeletal malformations	-Vertebral dysplasia - Malformed or missing ribs - Scoliosis -Elevation of scapula -Sacralization and lumbarization -Spina bifida	Limited cervical motion Short neck Sprengel deformity
Abnormalities of the head and face	Micrognathia (small jaw) Cleft lip, cleft palate, facial asymmetry	
Hearing	Hearing loss (conductive or sensory)	
Abnormalities of the extremities	(ectrodactyly): absence of a portion of one or more fingers or toes (syndactyly): webbing of the fingers or toes Duplicated thumb Absent radius	
Heart malformations	Atrial septal defect ASD Tetralogy of Fallot Pulmonary stenosis	

[a] Summarized results from all cases reviewed in our paper

examination were performed. The patient demonstrated normal secondary sexual characteristics with normal breast, axillary, and pubic hair development. Gynecological examination revealed normal female external genitalia on the inspection while speculum examination was not performed, as the patient was single. Trans-abdominal examination showed a rudimentary uterus and proximal vagina while both ovaries were present and appeared polycystic, the left kidney was hypoplastic, and the bladder wall was thickened. An abdominal and pelvic MRI examination was also conducted and confirmed the same findings. Echogram revealed mild LVH with good systolic function and grade I MR and AR. The audiogram was normal.

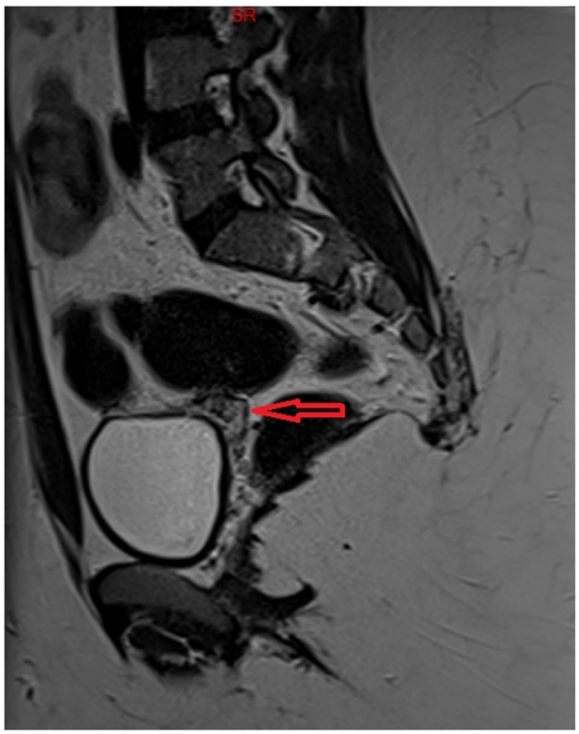

Fig. 1 Sagittal T2WIs pelvic MRI showed midline uterine bud (red arrow) behind the dome of the urinary bladder with cystic cavitation

Cytogenetic testing revealed 46, XX female karyotype, thus confirming the diagnosis of MRKH syndrome similar to the first case. The patient and her parents were counselled regarding the findings and implications on fertility and childbearing.

Analytical and literature review

A PubMed non-filtered search results for MRKHS among sisters were reviewed using the following keywords: Mayar Rockitansk/sisters/siblings; a result of seven applicable papers were recognized, and none of them reported this syndrome in three sisters.

B Geyoushi et al. (2007) "Primary amenorrhea in non-identical twins: an improbable cause" [7] presented a case of 18-year-old dichorionic twins. She showed a well-developed secondary sexual characteristic features with primary amenorrhea and abdominal pain that was approved to be MRKHS.

Huepenbecker SP et al. (2017) "Two sisters with Mayar Rockitanski-Kuster-Hauser syndrome and serous adenocarcinoma of the ovary" [9] presented a case of two sisters with ovarian adenocarcinoma in 2010; the two sisters were known to have MARKHS since 1970.

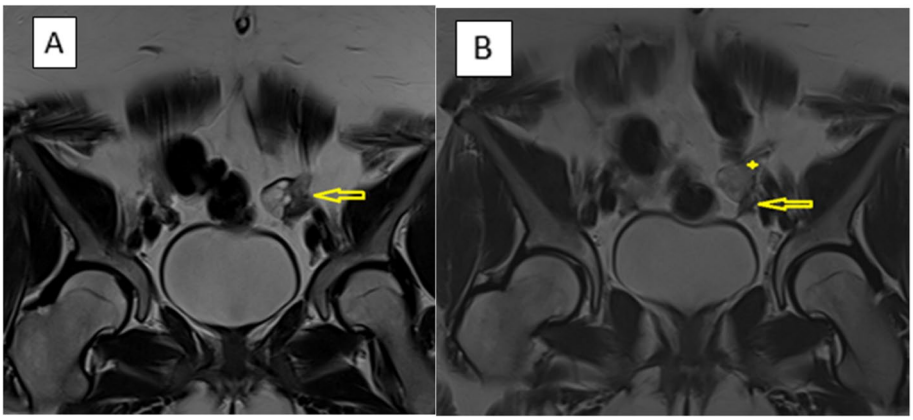

Fig. 2 **A**, **B** Coronal T2WIs pelvic MRI showed the course of lateral uterine bud (yellow arrow) extends from the lateral side of ovary (yellow star) (**A**), downward just superior to the level of bladder dome (**B**) Note the ectopic high location of ovary (yellow star)

S. Kula et al. (2004) "Mayar Rockitanski-Kuster Hauser syndrome" [10] reported two sisters with MRKHS with associated pulmonary stenosis.

Xue Ma et al. (2016) "Familial occurrence of Mayar Rockitanski-Kuster Hauser syndrome" [11] reported a case of two sisters: 16- and 6-year-olds with MRKHs and more investigation among the family revealed other non-first relative familial cases.

Ugonna A Duru et al. (2009) "Discordance in Mayar–von Rockitanski-Kuster-Hauser syndrome noted in monozygotic Twins" [12] reported a case of a 17-year-old monochorionic–monoamniotic twin with MRKHS but her twin sister was with the normal reproductive system.

Katharina Rall et al. 2015 (2015) "Typical and atypical associated findings in a group of 346 patients with Mayar Rockitanski-Kuster Hauser syndrome" [13] studied

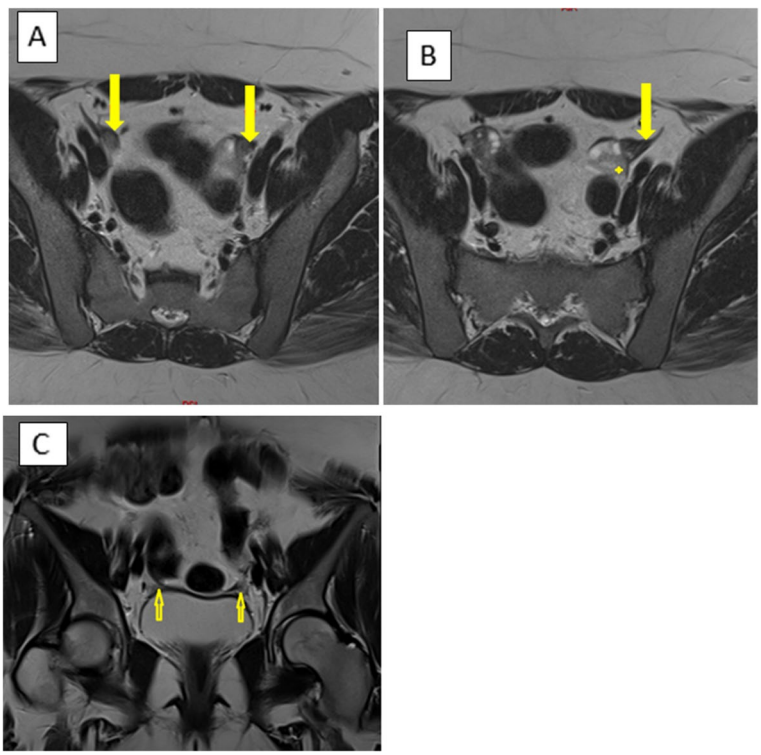

Fig. 3 **A**–**C** Axial T2WIs pelvic MRI: **A**, **B** showed the lateral uterine bud (closed yellow arrow) located lateral high in the pelvis and abutting the ovaries (yellow star). **C** Fibrous bands seen bilaterally above dome of the bladder (yellow open arrow)

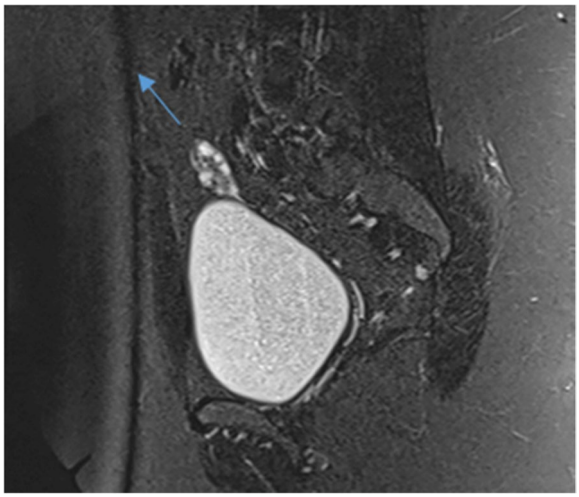

Fig. 4 Sagittal T2 STIR showed polycystic appearance of ovary (base of blue arrow)

around 346 MRKHS patients, they found around 39 siblings, and the incidence of malformation was studied as well; 57.6% renal, 44.4% skeletal, and 30.8% other malformations. 53.2% of the sample had type I MRKHS and 41.3% had type II.

Higher incidence of linked malformation in siblings of Mayar-Rockitanski-Kuster-Hauser syndrome patient was studied by M. Wottgen et al. in 2008 [14] by using a questionnaire for 73 MRKHS patients, they did not find siblings involvement, but there was 13 out of 103 siblings who had about a three-time increase in musculoskeletal malformation in comparison with a normal population.

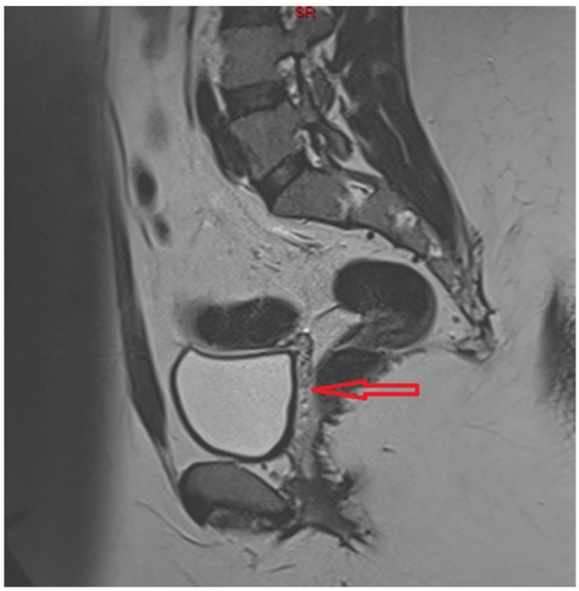

Fig. 5 Sagittal T2WIs pelvic MRI showed hypoplastic upper two-third of the vaginal with cystic remnants (red arrow)

Table 3 Cardiac echogram and audiogram results of our patients

	First patient (17 years old)	Second patient (20 years old)	Third patient (25 years old)
Echogram	Moderate LVH Grade I diastolic dysfunction EF 45% Dilated four chambers Grade II MR Grade I TR Grade I AR	Good systolic and diastolic function EF 60% Trace TR, MR	Mild LVH with good systolic function Grade I MR Mild AR Trace AR EF 60%
Audiogram	Normal	Normal	Normal

Literature of MRI findings in MRKHS of five large sample-sized studies was also reviewed and summarized in Table 1.

Discussion

The typical clinical presentation of this syndrome, as in our patients, is primary amenorrhea with cyclic colicky pain [1–4].

Clinical examination usually reveals well-developed secondary female sexual characteristics with a short or absent vaginal canal; however, the virginity of the patients usually limits the vaginal evaluation.

Hormonal profile (FSH, LH, estradiol, progesterone, prolactin, and testosterone) was within normal limits. The karyotype is required to rule out androgen insensitivity syndrome [1–6].

The top differential diagnosis of MRKHS are androgen insensitivity and transverse vaginal septum with an imperforate hymen, which were excluded confidently in our patients and the diagnosis of MRKHS type II was confirmed.

Imaging studies such as ultrasound and MRI are necessary to assess the anatomic features of this syndrome. Ultrasonography is the initial method of choice to confirm the absence of the uterus from its normal location, which is posterior to the urinary bladder, while the ovaries are typically present; however, their ectopic location sometimes makes them hard to be identified on ultrasound.

MRI pelvis is the most sensitive and specific imaging method to assess this syndrome as well as the associated anomalies.

For our three sisters patients, we performed an MRI of the pelvis and abdomen (Siemens 1.5 Tesla), the following sequences were adapted: coronal T2WIs, axial: T1WIs, T2WIs, STIR sagittal T1WIs, T2WIs.

MRI imaging aimed to assess the following [4–8]:

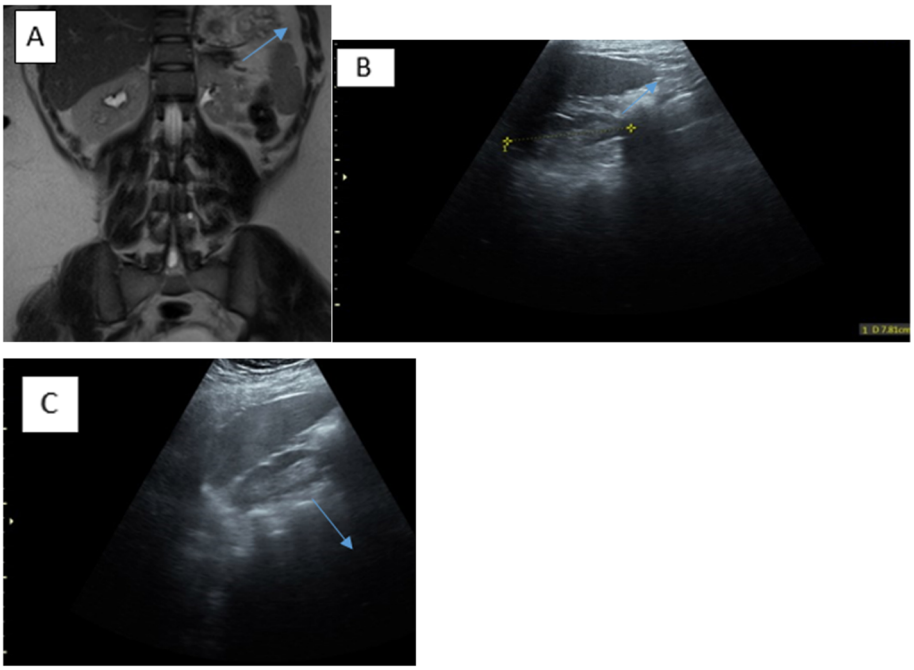

Fig. 6 **A** Coronal T2WIs MRI showed Lt kidney hypoplasia (base of blue arrow). **B**, **C** Renal ultrasound showed Lt kidney hypoplasia with preserved fetal lobulations, long axis of kidney measures about 7.8 cm (base of blue arrow)

1- The ovaries: presence, location (pelvic or extra pelvic, and if they are pelvic in location, are they normally located or ectopic high in the pelvis), size, and the presence of follicular cysts or masses.
2- The uterus: presence and the remnant uterine bud location. Usually, the rudimentary tissue may be recognized in three main locations [4–6]:

   a. Triangular midline/paramedian located posterosuperior to the dome of the urinary bladder (usually triangular)
   b. Lateral remnant tissue in both sides close to ovaries
   c. Two fibrous bands that connect the lateral tissue to the midline one.

3- The vagina: if it is present (proximal and distal parts) or replaced by rudimentary remnants.
4- Any associated anomalies like renal, musculoskeletal (MSK), and cardiac. The associated anomalies which were documented in all reviewed papers/references during our article preparation (around 15 papers) were summarized in Table 2.

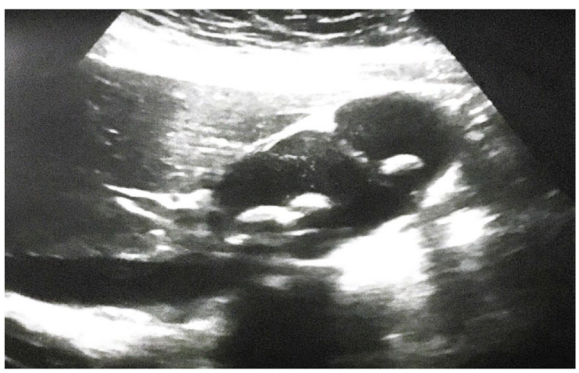

Fig. 7 Transabdomenal ultrasound of gall bladder showed multiple gall bladder stones

Measurements were performed using (MPTronic medical software EZ.DICOM CD VIEWER version3 2.8.0), the volume was calculated using the formula length*height*width *0.523.

The midline Müllerian remnant tissue was seen in one patient (Fig. 1), and the volume is around 4.812 cc, while the lateral uterine bud was seen bilaterally in two sisters, and unilateral in the youngest sister, the former has only Rt bud that measures around 1.156. The volume of the lateral bud for the second and third sisters were Rt 2.711/Lt 2.9 cc7, Rt 1.749/Lt 3.295 cc respectively. The lateral buds abut the ovaries which are located high in the pelvis and extend from superolateral to anteromedially in all patients (Figs. 2 and 3).

Table 4 Radiological results of our cases in pelvic MRI and spine X-ray

		First patient (17 years old)	Second patient (20 years old)	Third patient (25 years old)
Uterus	Paramedian/midline uterine bud tissue	Absent	Present Paramedian bud: seen posterior to the dome of urinary bladder in the midline 1.6 × 2.5 × 2.3 cm Hypointense on T2WIs) with cavitation	Absent
	Lateral uterine bud tissue Rt and Lt bud	Present: Extends from lateral side of ovaries downward medially Rt bud 1.7 × 1 × 1.3 (1.156 cc) Lt bud absent Hypo intense on T2WIs No cavitation	Present: Extends from lateral side of ovaries downward medially Rt bud 1.6 × 1.8 × 1.8 (2.711 cc) Lt bud 1.7 × 2.4 × 1.4 (2.987 cc) Hypo intense on T2 WIs No cavitation	Present: Extends from lateral side of ovary downward to posteriormedially Rt bud: 1.9 × 1.6 × 1.1 cm (1.749 cc) Lt bud: 3 × 1 × 2.1 cm (2.987 cc) Hypo intense on T2 WIs No cavitation
	Fibrous band	Present on Rt side	Present on both side	Absent
Proximal 2/3 of vagina		Hypoplastic	Hypoplastic	Hypoplastic
Distal 1/3 of vagina		Normal	Normal	Normal
Ovaries		-Present in the pelvis in normal location with follicular cysts Rt ovary 2.3 × 1.2 × 2 cm (2.887 cc) Lt ovary 1 × 0.6 × 1 cm (0.319 cc)	High pelvis location with follicular cysts Rt ovary (1.7 × 2.6 × 1.9 cm) (4.392 cc) Lt ovary 2.4 × 2.3 × 2 cm (5.773 cc)	High pelvis location with few follicular cysts (polycystic appearance) Rt ovary 3.4 × 2 × 1.9 cm (6.757 cc) Lt ovary (1.7 × 2.6 × 2 cm) (4.623 cc)
Kidneys		Normal	Normal	Hypoplastic Lt kidney around 7.5 cm
Spine		Normal	Normal	Normal
Others		Gall bladder stones	–	-Diffuse urinary bladder wall thickening -Polycystic appearance of ovaries

STAIR was the best MRI sequence that detects ovaries. The ovaries were present in the three sisters bilaterally, but ectopic high in the pelvis and showed follicular cysts (Figs. 2, 3, and 4). However, the largest ovary size showed a polycystic appearance. The Rt ovaries measures for the three sisters 2.887 cc, 4.392 cc, and 6.757 cc, respectively, and the Lt one measures 0.319 cc, 5.773 cc, 4.623 cc, respectively.

Sagittal T2WIs recognized the rudimentary proximal vagina with normal distal vagina for all sisters (Fig. 5).

The three sisters had cardiac manifestations, mainly valvular disease. The condition was more severe with the youngest one (Table 3). Furthermore, the oldest sister had a hypoplastic kidney with preserved fetal lobulation (around 7.8 cm on the long axis) (Fig. 6).

We found that valvular cardiac defect is strongly associated with MRKHS in our cases such association was not reported before, in addition to that, we noted that the smaller volume of remnant tissue the more severe the associated congenital anomalies.

A gallbladder stone was seen in the youngest patient (Fig. 7) but it could be related to her morbid obesity.

The detailed MRI findings were summarized in Table 4.

The treatment of MRKS requires a multidisciplinary approach as they have not only the anatomical changes but also psychological stress, anxiety, and sometimes depression that result from concerns about not being able to have normal sexual life or getting pregnant.

Treatment is usually postponed until the patient is ready to start sexual activity. Surgical reconstruction

of neovagina or non-surgical (using vaginal dilators) may allow the patient to have a normal sexual life. Most patients with MRKHS have a rudimentary non-functioning uterus but the ovaries are functioning and can produce normal ovum that can be later fertilized by the husband's sperm using assisted reproductive techniques, and then transferred the embryos to a surrogate mother. Although this option might be not accepted in some communities due to religious beliefs or governmental laws as in our case, legal children adaptation may be a good alternative. Transplantation of the uterus has been reported and might be an option for MRKHS patients who intend to get pregnant.

Regarding cyclical abdominal pain and secondary endometriosis, it can be relieved by surgical resection of remnants of bud.

The diagnosis of MRKS syndrome induces severe psychological stress which causes a significant impact on a patient's quality of life; however, treatment whether surgical or non-surgical, in addition to psychological support and counseling for those patients may improve the quality of life and reduce anxiety and stress [1–3].

Conclusion and learning points

-MRKHS should be considered in case of primary amenorrhea, and multisystem investigation is mandatory to exclude associated anomalies.

-The MRI findings recognize (1) variable appearance of Müllerian remnant tissue of uterus and vagina, including paramedian and lateral uterine rudimentary bud, bilateral fibrous band, and rudimentary proximal vagina. (2) The presence of both ovaries, which could be ectopic.

-The smaller the volume of Mullerian remnant tissue, the more severe the associated anomalies.

-The valvular cardiac disease is associated with MRKHS, which was not reported before.

Abbreviations

MRKHS: Mayer-Rokitansky-Küster-Hauser syndrome; MRI: Magnetic resonance imaging technique; MSK: Musculoskeletal; NS: Not stated; AR: Autosomal recessive; AD: Autosomal dominant; MR: Mitral regurgitation; AR: Aortic regurgitation; TR: Tricuspid regurgitation; EF: Ejection fraction; LVH: Left ventricular hypertrophy; Rt: Right; Lt: Left; WIs: Weighted images.

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Authors' contributions

All authors have made substantial contributions to the conception, design of the work, acquisition, and interpretation of data. They have drafted and revised the manuscript, and they have approved the submitted version and the modified version. The authors have agreed both to be personally accountable for the authors' own contributions and to ensure that questions related to the accuracy or integrity of any part of the work are appropriately investigated, resolved, and the resolution documented in the literature.

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Availability of data and materials

The data that support the findings of this study are available from the computerized patient medical archived file in the hospital, but restrictions apply to the availability of these data, which were used under license for the current study, and so are not publicly available. Data are however available from the authors upon reasonable request and with permission of MOH hospitals.

Declarations

Ethics approval and consent to participate

Our research was approved by institutional ethics committee of faculty of medicine in Mutah University.

Consent for publication

Our research does not contain any personal data, photos, or clinical trial for medication or radiation or any hazard; however, written consent form was signed by the patients and their parents. Moreover, agreement of institutional ethics committee of faculty of medicine in Mutah University was also achieved.

Competing interests

The authors declare that they have no competing interests.

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