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Case presentation of isolated Mayer -Rokitansky -Kuster -Hauser (MRKH) Syndrome- A Short Review of Two Cases

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ABSTRACT:

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH) is a congenital condition with an unclear cause that is characterized by mullerian agenesis and may be linked to cardiac, skeletal, or renal malformations. Most incidents are infrequent. Two typical cases of MRKH syndrome are presented here. When a patient presents with primary amenorrhea, a comprehensive diagnostic workup should encompass hormonal screening, physical examination, chromosomal analysis,.and history collection. A healthy ovary is indicated by normal levels of hormones , follicle stimulating hormone (FSH) and luteinizing hormone (LH). The precise anatomical deficiencies, however, are highlighted by imaging modalities like MRI and USG. In conclusion, the absence of a regular menstrual cycle in a young, healthy-looking adolescent girl should raise the possibility of Mayer-Rokitansky-Kuster-Hauser syndrome. Regarding her potential for conception in the future and the various treatment options (both surgical and non-surgical), the patient should get appropriate counseling.

KEYWORDS: Amenorrhea, Agenesis, Infertility, MRKH.

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INTRODUCTION:

Congenital hypoplasia of the uterus and upper vagina is a characteristic of the Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome The incidence of MRKH syndrome has been estimated as 1 in 4500 newborn girls .Its characteristics include the normal 46, XX, female chromosomal pattern, normal fallopian tubes, ovaries, and external genitalia, as well as the partial or whole absence (agenesis) of the uterus with an absent or hypoplastic vagina. Pubic hair growth and breast development are also typical. Abnormalities of the skeleton and/or

kidneys are frequently seen. Frequently, primary amenorrhea the absence of menstruation by age 16—is the first detectable symptom of MRKH condition. [1,2,3]

Mayer (1829) and Rokitansky (1838) were the first to describe this syndrome. Rokitansky found vaginal and uterine agenesis, while Mayer reported some vaginal duplication Kuster identified the urologic connection in 1910, and Hauser distinguished between MRKH and androgen insensitivity syndrome (AIS) in 1961.[4]

Three groups correspond to MRKH syndrome[5]

1. Isolated uterovaginal agenesis is the defining characteristic of Type I/Typical MRKH.
2. Type II/Atypical MRKH: It is linked to abnormalities of the ovaries and kidneys
3. MURCS linked MRKH: This third kind includes hearing impairment, renal, skeletal, and cardiac abnormalities, as well as Mullerian duct aplasia, renal aplasia, and cervico-thoracic somite dysplasia relationship.[6]

CASE HISTORY:

CASE 1

A sixteen year old girl revealed that she had previously experienced primary amenorrhea. She did not report experiencing any urinary or gastrointestinal complaints. Following was the diagnostic workup for her primary amenorrhea.

Background: Her family did not have a history of primary amenorrhea, delayed menses, or any other endocrine disorders. She was 13 years old when her breast development (thelarche) began. She never reported experiencing periodic stomach ache. Smell perception abnormalities were not present in the past. There was no prior history of radiation, chemotherapy, or surgery to the abdomen or inguinal area.

Physical Examination: Her height was normal and her growth parameter was normal. Her BMI of 18.75 kg/m2 was noted. Vital signs were normal, with a blood pressure of 110/68 mmHg. She displayed typical secondary sexual traits during physical assessment. Tanner staging IV was reached for pubic hair growth, axillary hair development, and breast development. Her external genitalia appeared normal, and her vaginal opening was visible. No gonad, inguinal hernia, or palpable abdominal tumor was present. An aperture was visible. No inguinal hernia, gonads, or palpable abdominal mass was present in the inguinal area.

Imaging: The right and left ovaries measured 2.8 X 2.3 cm and 2.5 X 1.4 cm, respectively, on the pelvis during ultrasonography. There was no visible uterus, which may indicate an aplastic uterus. The kidney, ureter, and bladder were all visible on the MRI scan without any abnormalities. Although the uterus could not be detected, a bud on the postero-inferior part of the bladder was observed, which may indicate an aplastic or hypoplastic uterus. The ovaries had several little follicles and were normal in terms of size, shape, and intensity.

Hormonal Evaluation: Her levels of prolactin, TSH, LH, and FSH were all within normal ranges. Karyotyping: She has a karyotype of 46, XX, according to chromosomal research. Therefore, Mayer-Rokitansky-Kuster-Hauser syndrome was diagnosed based on the workup mentioned above.

CASE 2:

A 24 year old married woman arrived with a history of lower abdominal pain and no signs of menstruation yet. She also reported itchy

vaginal discharge white in color and copious in amount.

There was no prior history of gastrointestinal issues or allergies. After a thorough history and examination, she was evaluated for primary amenorrhea.

History: Her mother and sister have never experienced a delayed menstrual cycle. She was 13 years old when her breasts started to develop. No prior history of abdominal operations was present. Physical examination results showed that she was growing normally, her vital signs were normal, her blood pressure was 120/80 mmHg, and her secondary sexual characteristics were normal. Axillary hair was present, pubic hair growth was in tanner staging IV, and breast development was in tanner stage 4. Her external genitalia were normal, and her vagina was visible but short and blinded. Per abdomen shows no discernible bulk, no organomegaly or pain. The right and left ovaries measure 25 x 21 mm and 23 x 10 mm, respectively, and the uterus is not visible. The bilateral kidneys are not visible independently, but a single kidney measuring 99 x 51 mm is visible in the pelvis(Figure-1).


Spleen: Shows normal size, shape and echotexture.

Pancreas: Shows normal size, shape and echotexture.

Kidneys: B/L kidneys are not separately visualized however there is a single kidney seen in pelvis measuring 99X51mm. Cortico medullary differentiation is maintained. Pelvicalyceal system is normal. No calculus or mass lesion is seen.

Bladder: Shows normal lumen. Wall thickness is within normal limits. No calculi or focal mass lesion is seen.

Uterus is not visualized

Ovaries: B/L ovaries are normal in size, shape and echotexture. No mass is seen
Rt. Ovary: 25X21mm Lt. Ovary: 23X10mm

No retroperitoneal lymphadenopathy or intraperitoneal free fluid is seen.
Bowel loops are showing normal peristaltic movements.

IMPRESSION: Findings are suggestive of uterus is not visualized however B/L ovaries are normal with a single pelvic kidney.

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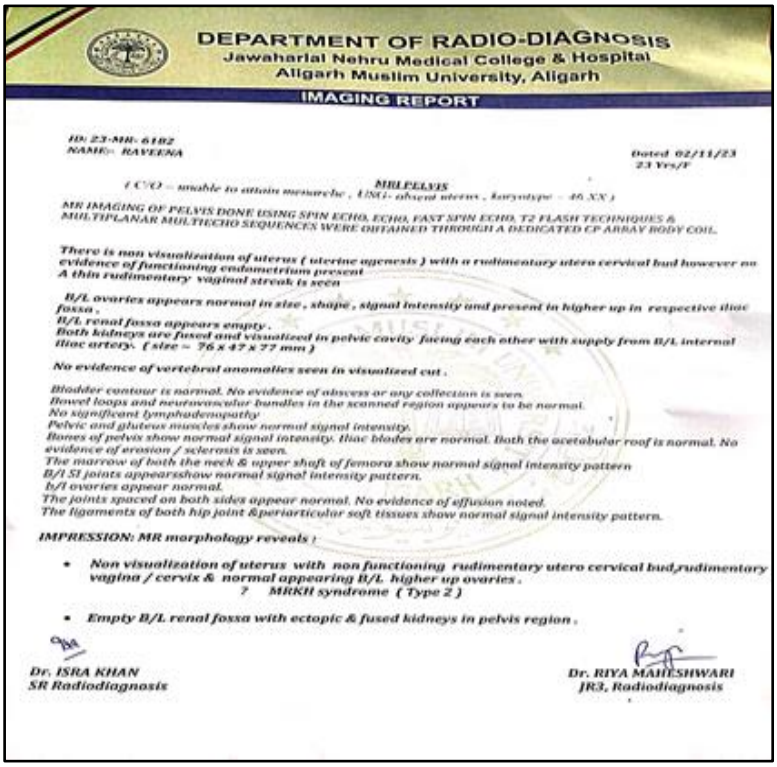
Figure-1: USG before treatment



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IMAGING REPORT

ID: 23-MR- 6182
NAME:- RAVEENA

Dated 02/11/23
23 Yrs/F

(C/O – unable to attain menarche , UMI- absent uterus , karyotype – 46 XX)

MRI PELVIS

MR IMAGING OF PELVIS DONE USING SPIN ECHO, ECHO, FAST SPIN ECHO, T2 FLASH TECHNIQUES & MULTIPLANAR MULTIECHO SEQUENCES WERE OBTAINED THROUGH A DEDICATED CP ARRAY BODY COIL.

There is non visualization of uterus (uterine agenesis) with a rudimentary utero cervical bud however no evidence of functioning endometrium present
A thin rudimentary vaginal streak is seen

B/L ovaries appears normal in size , shape , signal intensity and present in higher up in respective iliac fossa .
B/L renal fossa appears empty .
Both kidneys are fused and visualized in pelvic cavity facing each other with supply from B/L internal iliac artery. (size – 76 x 47 x 77 mm)

No evidence of vertebral anomalies seen in visualized cut .

Bladder contour is normal. No evidence of abscess or any collection is seen.
Bowel loops and neurovascular bundles in the scanned region appears to be normal.
No significant lymphadenopathy
Pelvic and gluteus muscles show normal signal intensity.
Bones of pelvis show normal signal intensity. Iliac blades are normal. Both the acetabular roof is normal. No evidence of erosion / sclerosis is seen.
The marrow of both the neck & upper shaft of femora show normal signal intensity pattern
B/l SI joints appearsshow normal signal intensity pattern.
b/l ovaries appear normal.
The joints spaced on both sides appear normal. No evidence of effusion noted.
The ligaments of both hip joint &periarticular soft tissues show normal signal intensity pattern.

IMPRESSION: MR morphology reveals :

- Non visualization of uterus with non functioning rudimentary utero cervical bud,rudimentary vagina / cervix & normal appearing B/L higher up ovaries .
  ? MRKH syndrome (Type 2)
- Empty B/L renal fossa with ectopic & fused kidneys in pelvis region .

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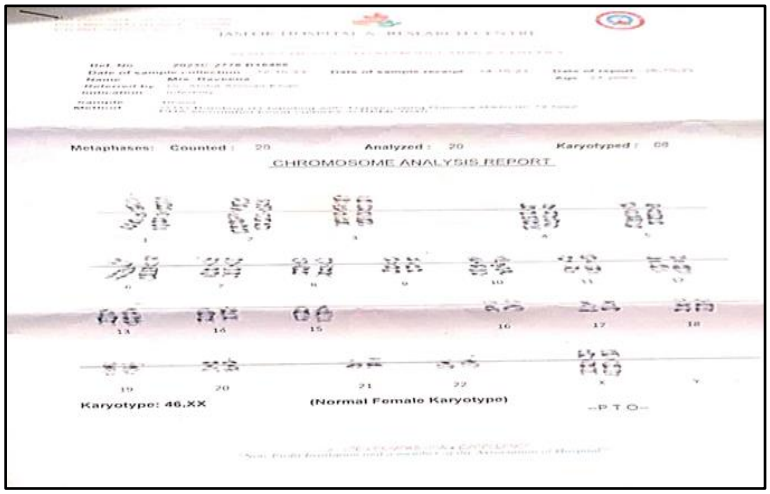
Figure-2 MRI report of pelvis



Metaphases: Counted : 20 Analyzed : 20 Karyotyped : 05

CHROMOSOME ANALYSIS REPORT

Karyotype: 46,XX (Normal Female Karyotype)

--P T O--

Figure-3: Chromosome analsis

On MRI, the uterus is not seen, bilateral ovaries appear normal but are higher up with Non-functional rudimentary uterocervical bud and vagina/cervix. Reminiscent of MRKH syndrome (type 2) Pleural effusion in both kidneys, together with ectopic kidneys fused together in pelvis (figure2). Hormonal evaluation: Her blood TSH, serum LH, FSH, and prolactin were all within normal limits. 5. Karyotyping: based on chromosome analysis, her karyotype was 46,xx. Figure-3

DISCUSSION:

Patients with MRKH syndrome typically present with primary amenorrhea during adolescence

women with this condition have normal ovarian function and become mother through IVF-ET and surrogacy in high income countries.[7] Mullerian agenesis, or

Mayer-Rokitansky-Kuster-Hauser syndrome, is a spectrum of congenital malformations for which the exact origin is unknown. However, Mullerian aplasia and hyperandrogenic symptoms are caused by mutations in the WNT4 gene. In women with normal ovaries and secondary sexual features, utero-vaginal agenesis occurs. The uterus can be as simple as bilateral, non-cannulated muscle buds with normal ovaries, fallopian tubes, and endocrine and cytogenetic assessments.

It is inherited via the autosomal dominant mode. It is brought on by the aberrant development of mullerian ducts, and as a result, organs such as the kidney are also affected. When it comes to aberrant sexual development, such as cervicothoracic somite malformations and unilateral renal agenesis, MRKH is related to the Mullerian duct aplasia-renal agenesis-cervicothoracic somite dysplasia (MURCS) relationship. Our patient case 1 had MRKH TYPE 1.

Our patient case 2 had MRKH type 2 (MURCS) association, as evidenced by Mullerian agenesis and left kidney agenesis. Heart problems are uncommon in MRKH type 2, which can also affect the skeletal system, bones, and upper urinary tract. In 40% of instances, there are abnormalities of the urinary system, primarily unilateral renal agenesis, hypoplastic kidneys, kidneys shaped like horseshoes, and hydronephrosis. Imaging studies are mostly used to diagnose MRKH. [8]

Although transabdominal ultrasonography is the first line of inquiry, abdomino-pelvic magnetic resonance imaging provides more accurate and lucid information than the former Thus, even though our patient had previously undergone ultrasonography, we recommended that she get an MRI.[9]Congenital vaginal agenesis, low transverse vaginal septum, androgen insensitivity, as well as imperforate hymen, are among the differential diagnoses.

Knowing that they do not have a uterus or vagina causes worry and mental stress in young women with MRKH diagnoses. Thus, patient counseling comes before any kind of treatment, as it did in our instance.[10] Both non-surgical and surgical methods of neo-vaginal formation are used in treatment. In non-surgical techniques, the Franck's dilator method is used to lengthen and dilate the vagina.[11]Vaginal dilators, also known as Hegar candles, are applied to the perineal dimple for at least 20 minutes each day. With a success rate ranging from 78% to 92%, this technique is considered a first-line therapy due to its non-invasive nature and high effectiveness. Surgical procedures include the Vecchietti surgery, the Abbe-McIndoe operation, and sigmoidal colpoplasty. which are employed in the treatment of MRKH syndrome.

CONCLUSION:

Lack of thorough evaluation, delayed diagnosis, and unclear explanation all contribute to the underestimation of MRKH syndrome prevalence. Medical professionals ought to be well-prepared to advise a patient with MRKH syndrome on her options for future conception, diagnosis, and treatment options.

Taking into account the maturity of the patient is crucial, especially when undergoing neovaginal surgery. It is important to cope with any emotions of inadequacy or loneliness that the patient or their family may be experiencing.

Despite not being a fatal condition, MRKH syndrome diagnosis in adolescence has a profound impact on a patient's life. For some patients, there are options for conception and sexual interactions with appropriate care.

Consent of patients :
Whole study was carried out with the consent of patients

Limitation of study :
This is case details of two patients and further studies should be carried out in more number of cases for its scientific validation.

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Conflict of interest: Author declares that there is no conflict of interest.

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