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Mayer-Rokytansky-kuster-Hauser Syndrome: A Rare Congenital Anomaly-Report of Two Cases

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Abstract:

Mayer -Rokytansky-Kuster- Hauser Syndrome (MRKH) is a rare congenital anomaly, in which derivatives of mullerian duct is partially or completely absent whereas ovaries are normally present in a female patient. The patients usually present to physician with primary amenorrhea in adolescent period, when secondary sexual characteristics develops normally.Surgical correction permits normal sexual function and, possibly, reproduction with assisted techniques in MRKH patients. So,early diagnosis is very important to offer possible treatment options to the parents and patients and also to avoid psychological impact on patient. Here, we have described two cases of MRKH with different presenting complaints.

Keyword: *Mayer-Rokytansky- Kuster- Hauser Syndrome (MRKH), Mullerian duct, primary amenorrhea*

Introduction:

Mayer- Rokytansky-Kuster- Hauser syndrome, also known asmullerian agenesis syndrome, is a rare congenital anomaly in which derivative of mullerian duct is not developed or under developed in a genetically female patient. So, in this condition, uterus, vagina or both may be absent or rudimentary.[1] Patients with this condition is usually diagnosed at adolescent period when patient present with primary amenorrhea. But patient's secondary sexual characteristics develop normally as development of ovary is not affected.[2] Mullerian agenesis may be an isolated anomaly or may be associated with other anomalies like renal agenesis, vertebral anomaly and cardiac anomaly.[3]

Here we described 2 cases of MRKH syndrome, who were diagnosed at early age, when patient presented to us with another congenital anomaly in external genitalia.

Case description:

Case 1: A 6 year old girl presented with absence of clitoris since birth. The patient was born after an uneventful pregnancy. There was no family history of amenorrhea or infertilityor such type of abnormality.Genital examination revealed absence of clitoris, normal labia majora & minora and also separate urethral and vaginal orifices. Systemic examination revealed no cardiac or skeletal abnormality.The abdominal ultrasonography revealed non visualized uterus and also normal sized kidney on both side.On hormonal assay, FSH, LH and oestrogen was 0.50mIU/ml, 0.72 IU/L and 18.96 pg/ml respectively. All were in normal prepubertal level. Cytogenetic karyotyping revealed 46, XX karyo-typing (Fig 3). MRI of pelvic organs (Fig 1 & 2) revealed absence of uterus and upper part of vagina. Ovaries were present on both side. We diagnosed her as mullerian agenesis syndrome. We counseled the parents about development of secondary sexual characteristic and surgical options for reconstruction of vagina at adolescent period.

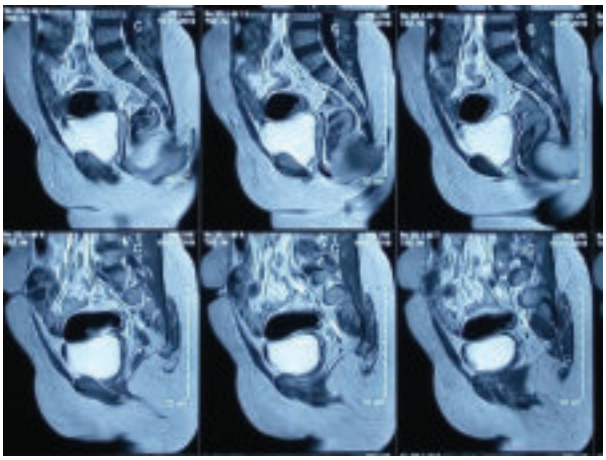

Figure 1 : *Sagital section of MRI of pelvic organ showing no uterus in between rectum and bladder (case 1)*

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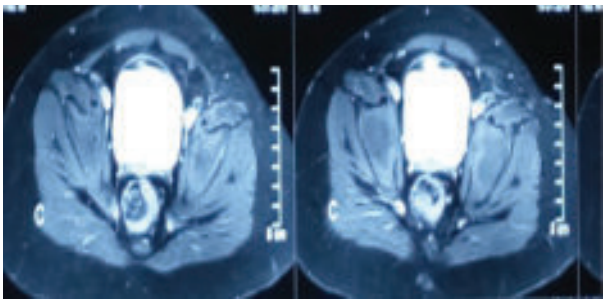

Fig.-2: *Axial section of MRI of pelvic organ showing no uterus in between rectum and bladder (case 1)*

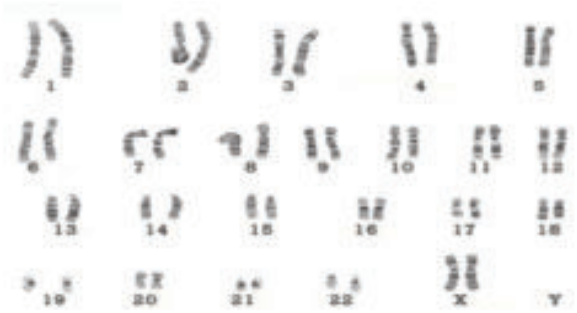

Fig.-3: *Cytogenetic karyotyping showing 46, XX (case1)*

Case 2: One anxious mother came to us with their 2ndfemale child at the age of 7year, who was operated at 2.5 years of age due to left sided irreducible painless inguinal hernia. Operation was done by a paediatric surgeon. At the time of operation, surgeon found solid mass at inguinal area. He excised the mass& sent for histopathological examination. Surgeon informed the parents that the masses might be gonad (ovary). When we examine the patient, her physical findings were normal. Genital examination showed normal female genitalia, 2 separate urethral and vaginal orifices. Her family history was also not significant.Ultrasonography of pelvic organs revealed nonvisualized uterus and vaginal canal. Ovary like structure were seen in right tuboovarian area on ultrasonography. Left ovary could not be seen. Kidneys on both side were normal.

Hormonal assay revealed FSH, LH and Oestrogen were respectively 0.47mIU/ml, 0.69 IU/L and 15 pg/ml. All the hormone level was in prepubertal level. Cytogenetic karyotyping revealed a 46, XX. MRI of pelvic organ (fig 4) revealed absence of uterus and also upper part of vagina. Ovary was absent on left side and present on right side. Our diagnosis was Mayer- Rockytansky- Kuster- Hauser syndrome. We counseled the parents about development of secondary sexual characteristic and surgical options for reconstruction of vagina at adolescent period.

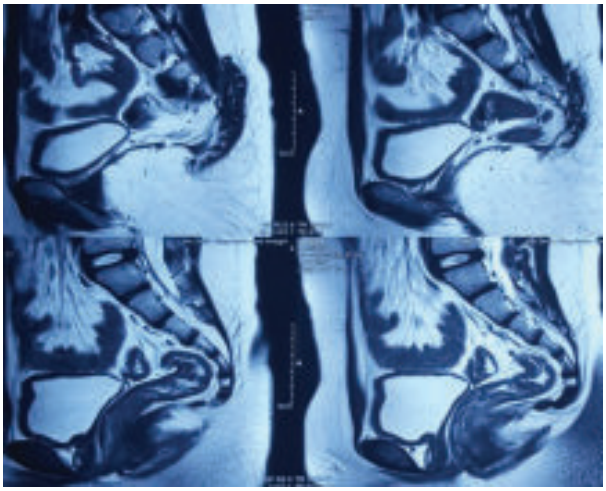

Figure 4: *MRI of pelvic organ (sagital section) showing absence of uterus and vagina in pelvis (case2)*

Discussion:

The mullerian and wolffian ducts are essential for the development of the female and male reproductive system, respectively. In genetically female, the mullerian ducts mature to become fallopian tubes, uterus, cervix and upper two thirds of the vagina (lower third is derived from the urogenital sinus), while the Wolffian duct degenerates. Developmental abnormalities, such as uterine and vaginal agenesis or the duplication of the uterus and vagina are preceded by any disruption of this physiological process.[4] Mullerian agenesis syndrome or MRKH syndrome is characterized by complete or partial failure of development of derivatives of mullerian duct in a cytogenetically 46,xx female.[1] Sometimes, vagina may appear as a small dimple below the urethra. Ovarian function is preserved because the ovaries originate within the primitive ectoderm, independent of the mesonephros. The exact pathogenesis of Müllerian agenesis with or without associated malformations is now well described and its etiology remains unknown. So, its familial occurrence is of considerable interest. MRKH was previously thought to be a sporadic anomaly, but familial cases support the hypothesis of a genetic etiology. Although the precise gene has not yet been identified, MRKH syndrome appears to be transmitted in an autosomal dominant fashion, with incomplete penetrance and variable expressivity.[3,5] MRKH syndrome is subdivided into two types: type I (isolated) or Rokitansky sequence and Type II or MRKH with associated anomaly.[6] MRKH syndrome type 2 is more frequently associated with renal, vertebral, and to a lesser extent,

auditory and cardiac defects.[7] In our both cases there was no positive family history. So, they may be sporadic cases. In the second case, patient had ectopic ovary which was operated. Histopathology report was not available. In a previous study on MRKH patients, about 38% patient showed ectopic ovary.[8] Wang Y. et al. also stated that MRKH patients may have complete uterine agenesis or rudimentary uterus without endometrium remnant.[8] It usually remains undetected until the patient presents with primary amenorrhea despite normal female sexual development at adolescent period. Our 2 cases were diagnosed at an early age, which is unusual. We could diagnose the patient at this age because patients presented with another anomaly and we considered them for detailed evaluation of that anomaly. As our 1st case has both ovary, she has the potential for development of secondary sexual characteristics. 2nd case has unilateral ovary and she will be needed to follow up for development of secondary sexual characteristics. Surgical correction permits normal sexual function and, possibly, reproduction with assisted techniques in MRKH patients. In recent years, infertility treatment options through in vitro fertilization have become available as part of the long-term care of these patients.[9] In our both cases, possibly normal sexual function and reproduction with assisted technique will be possible.The timing of the creation of a neovagina is elective, but treatment should be postponed until late adolescence to allow informed consent and compliance. In some studies, long-term follow-up has shown that vaginas created during childhood have high failure rates and require additional procedures for the creation of a functional vagina.[10,11]

Conclusion:

Mayer- Rockytansky-Kuster-Hauser syndrome (MRKH), a rare congenital abnormality, is frequently associated with various vertebral, renal and cardiac malformations. Ectopic ovary is also a frequent association. However, there is no reported case of MRKH associated with absence of clitoris. To diagnose the case of MRKH in early life, any patient with external or internal genital abnormality should be thoroughly evaluated.

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