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CASE REPORT OF MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME

Radiodiagnosis

Manjeera Boddepalli* Resident, Department of Radiodiagnosis, Katuri Medical College and Hospital, Chinakondrupadu, AndhraPradesh, India. *Corresponding Author

Mahesh Krishna Anne Resident, Department of Radiodiagnosis, Katuri Medical College and Hospital, Chinakondrupadu, Andhra Pradesh, India.

Ramakrishna. N Professor, Department of Radiodiagnosis, Katuri Medical College and Hospital, Chinakondrupadu, Andhra Pradesh, India.

ABSTRACT

The Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH syndrome), also called Rokitansky syndrome or vaginal aplasia of the uterus, is a congenital condition characterized by the absence of the uterus and vagina, however, ovaries are present and the external genitalia are normal. There are two different forms of this syndrome: typical and atypical form. We report a case of atypical form of MRKH syndrome in a 15year old female who presented with primary amenorrhoea. On examination, blind vagina noted and uterus not palpable per rectally. On sonographic evaluation, Uterus was not visualized. Bilateral renal fossa are empty with ectopic right kidney. On MRI, Post contrast studies showed rudimentary uterine tissue and right ectopic kidney is noted in pelvis. Karyotyping revealed 46XX. MRKH atypical form should be in mind if there are features of its history, physical examination, US and MRI evaluations in a patient with primary amenorrhea.

KEYWORDS

Mayer–Rokitansky–Kuster–Hauser, vaginal aplasia of the uterus, primary amenorrhoea.

INTRODUCTION:

The Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH syndrome), also called Rokitansky syndrome or vaginal aplasia of the uterus, is a congenital condition characterized by the absence of the uterus and vagina, however, ovaries are present and the external genitalia are normal.

CASE REPORT:

A 15 year old female patient was brought to the hospital by her mother with complaint of primary amenorrhoea. Growth and developmental history from childhood was normal.

On Examination of the patient there were no signs of webbed neck, syndactyly, polydactyly. The patient had TANNER Stage 4 breast development. Axillary & pubic hair were normal. External genitalia examination revealed normal Labia majora and minora. Blind vagina was noted. No hymenal orifice. Urethral orifice was normal. On Per rectal examination, uterus was not palpable.

INVESTIGATIONS:

Routine lab investigations are within normal limits.
Karyotyping – 46XX ; Normal female karyotype .
Ultrasound Abdomen :

- Non visualisation of uterus.
- Bilateral empty renal fossa with right ectopic kidney.
- MRI:

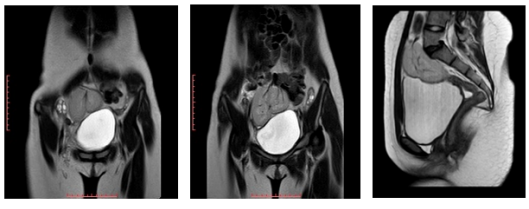

FIG 1: Coronal and saggital T2W images of pelvis showing non visualisation of uterus and normal ovaries on both sides.

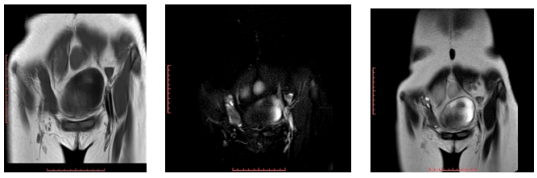

FIG 2: T1W, STIR & T2W images showing Well defined lesions, isointense on T1W and hyperintense on T2W & STIR noted in the bilateral inguinal region inferiomedial to the ovaries, measuring 1.7 X 1.1 cm on right side and 2.2 X 1.9 cm on left side.

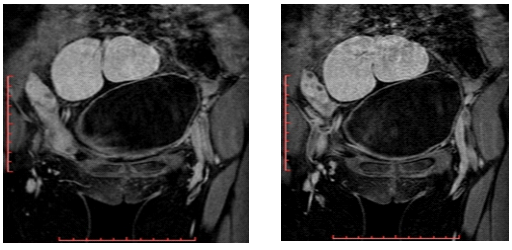

FIG 3: POST CONTRAST IMAGES: The lesions are showing enhancement on post contrast S/o Rudimentary Uterine tissue

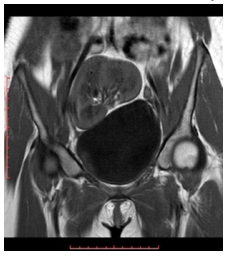

FIG 4: Both kidneys are not visualised in renal fossa. A 8.6x5.4cm ectopic kidney noted on right side of pelvis.

The patient was diagnosed as a case of MRKH syndrome (TYPE B)
The nature of the problem was discussed with the patient and her parents. They were counselled about the need for vaginal reconstruction and were referred to higher centre for possible vaginoplasty.

DISCUSSION:

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) is an anomaly that belongs to class I Mullerian duct anomalies.

There are two different forms of this syndrome[1] :
The typical form (type A) of this syndrome is characterised by the congenital absence of the uterus and upper 2/3 vagina with normal ovaries and fallopian tubes.
The atypical form (type B) of the syndrome includes associated abnormalities of the ovaries and fallopian tubes and renal anomalies.

Clinical presentation:

Primary amenorrhea with normal development of secondary sexual

characteristics.

Associations[2]:
The syndrome is often associated with alterations in the urinary or skeletal system which include:

- Vertebral anomalies: may be present in ~10% of cases.
- Renal anomalies: such as renal agenesis, ectopic kidney, fused kidney, renal hypoplasia, and horseshoe kidney may be seen in 30-40% of patients with the syndrome.

DIFFERENTIAL DIAGNOSIS:
Androgen insensitivity syndrome has male karyotype, presence of rudimentary ectopic testis and the absence of both uterus and ovaries.

Among the diagnostic tools, the utility of MRI for congenital Genito urinary diseases has been increasingly recognised. The final diagnosis was achieved by association of imaging findings with presence of karyotype 46XX[3].

This patient requires a multi-disciplinary approach with indicated anatomic treatment, the surgical or non-surgical creation of a neovagina, which may allow these patients to have a normal sex life[4,5].

CONCLUSION:
MRKH atypical form should be in mind if there are features of its history, physical examination, US and MRI evaluations in a patient with primary amenorrhea.

CONFLICT OF INTEREST:
There is no conflict of interest among the authors.

FUNDING STATEMENT:
There is no funding for the case report.

ETHICAL APPROVAL:
Necessary approval was taken from the Institution and the patients for carrying out this work

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