

An Interesting case of Mayer-Rokitansky-Kuster-Hauser Syndrome Type II with renal agenesis malformation

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Background & Introduction

- Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKH) is a rare condition affecting 1 in 4500 women [1] with Congenital absence of upper the vagina and uterus as the primary feature.
- The commonest abnormalities associated with MRKH syndrome type II are renal dysplasia and various skeletal malformations, typically vertebral. Less frequently it is associated with cardiac malformations & hearing impairment [1].
- Isolated features of the main malformations e.g. renal agenesis are often not investigated in all patients affected by MRKH. Therefore, It is important to raise awareness of this rare but important diagnosis & associated renal malformations, to ensure timely investigation & referral.

Case Report

- We present a 27 year old nullip referred for evaluation of daily non-specific abdominal pain. She denied urinary, bowel symptoms or dyspareunia. She was noted to have a single kidney on USS which prompted further evaluation.
- **PMH:** She was fit and well with no previous surgery. **DH:** No regular medication or allergies.
- **SH:** married, living with her husband., non smoker
- **OE:** HR 82 RR 19 BP 120/85 normotensive, BMI16, Negative urine dip with no haematuria. Normal development of secondary sexual characteristics.
- **Bloods:** Normal haemoglobin, renal and thyroid function, HCG <1. I
- **Investigation** with Ultrasound abdomen, CT renal tract and Pelvic MRI, revealed uterine aplasia with absence of fallopian tubes and cervix with associated right renal agenesis in keeping with MRKH Type 2. The Vagina appeared normal, the left ovary was visualised anterior to the inguinal canal and the smaller right ovary appeared in the right inguinal region. The patient was referred onto fertility services for further counselling.

Discussion

- In this case, the patient had unilateral renal agenesis and an L4 haemangioma in the vertebral body in keeping with MRKH Type 2.
- These incidental findings maybe detected on evaluation of other symptoms and MRKH should be considered as a differential diagnosis in such cases.

Conclusion

- The Incidence of MRKH syndrome has been under-evaluated mainly because, until recently, it was viewed as a female-specific sporadic disorder [1]. Isolated features of the triad of main malformations, including kidney agenesis and/or skeletal defects, were consequently may not get investigated in all patients.
- In patients presenting with incidental renal agenesis, clinicians should consider this rare diagnosis and ensure patients are investigated and referred appropriately, preventing diagnostic delays and misdiagnosis.

References:

[1] Pai A, Shakir M. Mayer-Rokitansky-Kuster-Hauser syndrome type II: A rare case. *Indian J Hum Genet.* 2013;19(1):113-115. [2] Griffin et al. Congenital absence of the vagina. The Mayer-Rokitansky-Kuster-Hauser syndrome. *Ann Intern Med.* 1976;85:224-36.