Mayer-Rokitansky-Küster-Hauser syndrome

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DESCRIPTION

A 19-year-old female patient presented with a history of primary amenorrhoea. Her developmental milestones were normal. There was no history of systemic illness or chronic medication use. She noticed thelarche at the age of 11 years and developed pubic hair at 13 years. She was born as the first child to second degree consanguinity and her younger sibling (sister) had attained menarche at 12 years of age. There was no growth stunting. On examination, she was 159 cm tall (with a mid-parental height of 162 cm) and had normal appearing female external genitalia and breasts (Tanner stage-5) and also had normal axillary and pubic hair development. There were no dysmorphic features. Her karyotyping was normal (46,XX). The patient blood investigations were as follows: haemoglobin 13.8 g/dL (normal range12–15), serum creatinine 0.8 mg/dL (normal range0.6–1.2), follicle stimulating hormone 4.2 mIU/mL (normal range 0–4.5), luteinising hormone 8.9 mIU/mL (normal range 1.9–12.5), testosterone 38.2 ng/dL (normal range 50–120), anti-Müllerian hormone 6 ng/mL (normal range 2–6.8), oestradiol 160 pg/mL (normal range 30–400) and 17-hydroxyprogesterone 80 ng/dL (normal range <100).

An axial section of the MRI of the abdomen and pelvis showed complete absence of uterus, cervix and vagina, with normal ovaries. (figure 1). A coronal section displayed a single midline pelvic pancake kidney (figure 2). There were no vertebral body anomalies. A diagnosis of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome was made. MRKH syndrome is typically known by the congenital absence of the uterus and the upper part of the vagina in women who have normally developed secondary sexual characters along with a normal female 46,XX karyotype. The reported incidence is about 1 in 4000–5000 live births. They are usually sporadic, although familial cases have also been described, denoting a probable genetic cause.[1] Affected women have normal growth and pubertal development (normal breast and pubic as well as axillary hair development in the presence of primary amenorrhoea). This condition is also usually associated with renal defects (unilateral agenesis or ectopia of 1 or both kidneys, horseshoe kidney in 40% of patients), cervicothoracic anomalies (asymmetric, fused vertebrae, scoliosis and Klippel-Feil anomaly in about one-fifth). Biochemical evaluation is essentially normal as these patients have normal functioning ovaries. Management of this condition includes screening for associated renal and skeletal anomalies. Psychological counselling and surgical procedures to create a neovagina when the patient is ready to start sexual activity are the main modalities of treatment for MRKH syndrome.[2]

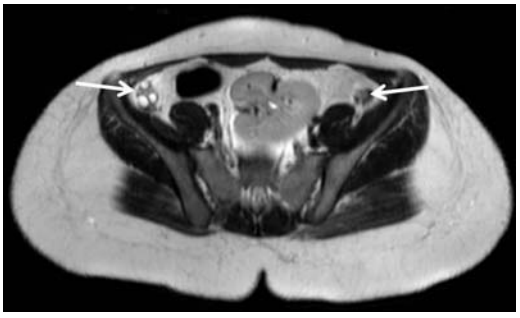

Figure 1 MRI of the abdomen and pelvis (axial section) showing complete absence of uterus, cervix and vagina, with normal ovaries.

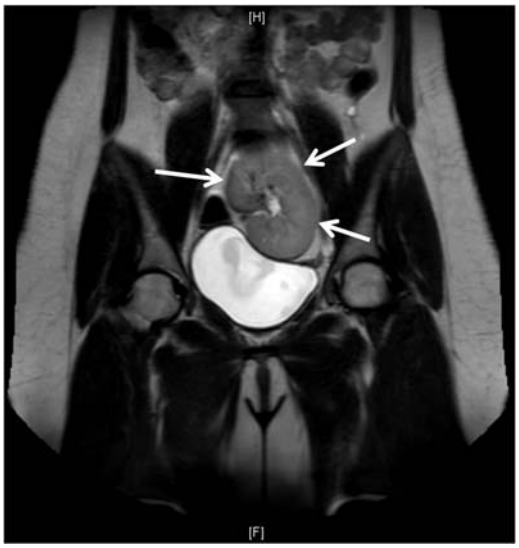

Figure 2 MRI of the pelvis (coronal section) displaying a single midline pelvic pancake kidney.

Learning points

- Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is diagnosed when a woman presents with primary amenorrhoea along with development of normal secondary sexual characters and normal female XX karyotype.
- MRKH syndrome is characterised by the congenital aplasia of the uterus and upper part of the vagina.
- Associated conditions such as ectopic or pelvic kidneys and skeletal deformities are seen in at least one-fourth of the patients.

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REFERENCES

1 Guerrier D, Mouchel T, Pasquier L, *et al*. The Mayer-Rokitansky-Küster-Hauser syndrome (congenital absence of uterus and vagina)-phenotypic manifestations and genetic approaches. *J Negat Results Biomed* 2006;5:1.
2 Morcel K, Camborieux L, Guerrier D; Programme de Recherches sur les Aplasies Müllériennes. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. *Orphanet J Rare Dis* 2007;2:13.