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Mayer-Rokitansky-Küster-Hauser syndrome as a cause of primary amenorrhea: a case report

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Abstract*: Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare cause of primary amenorrhea. It is defined by congenital aplasia of the uterus and upper two-thirds of the vagina in women with normal development of secondary sexual characteristics. Diagnosis is based mainly on magnetic resonance imaging (MRI). We report the case of a 21-year-old girl who consulted for primary amenorrhea, with present and well-developed secondary sexual characteristics. Biological workup revealed normal ovarian function and gonadotropic axis. Pelvic ultrasound and magnetic resonance imaging revealed complete agenesis of the uterus, the upper two-thirds of the vagina and the left kidney, confirming the diagnosis of Mayer-Rokitansky-Küster-Hauser (MRKH) type II syndrome. The purpose of this case report is to evoke the diagnosis of MRKH, in the presence of primary amenorrhea in a young woman with well-developed sexual characteristics, and also to look for specific signs on imaging, particularly MRI.*

Keywords : Primary amenorrhea, utero-vaginal aplasia, MRI, case report

Introduction

Primary amenorrhea is the absence of menstruation at the age of 15 in patients with normal growth and secondary sexual characteristics. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome remains a rare cause of primary amenorrhea [1], and is defined by congenital aplasia of the uterus and upper two-thirds of the vagina in women with normal development of secondary sexual characteristics and a normal karyotype (46, XX) [2]. We report the case of a 21-year-old girl who consulted for primary amenorrhea, with no clinical or biological abnormalities, in whom pelvic ultrasound and magnetic resonance imaging revealed Mayer-Rokitansky-Küster-Hauser syndrome.

Case report

The patient was a 21-year-old girl with no previous history of primary amenorrhea. Clinical examination revealed female external genitalia, well-developed breasts and other secondary sexual characteristics. The patient was a virgin, so the vaginal examination was not performed. The hormonal work-up confirmed normal ovarian function, with 17-esradiol at 100 pg/ml, as well as the gonadotropic axis, with FSH at 7 mIU/ml and LH at 4 mIU/ml, and normal testosterone levels. The genetic study revealed a constitutional karyotype of 46 XX. Ultrasound and pelvic MRI showed complete agenesis of the uterus and the upper two-thirds of the vagina (Figure 1, Figure 2 ,3), leading to the diagnosis of Mayer-Rokitansky-Küster-Hauser syndrome.

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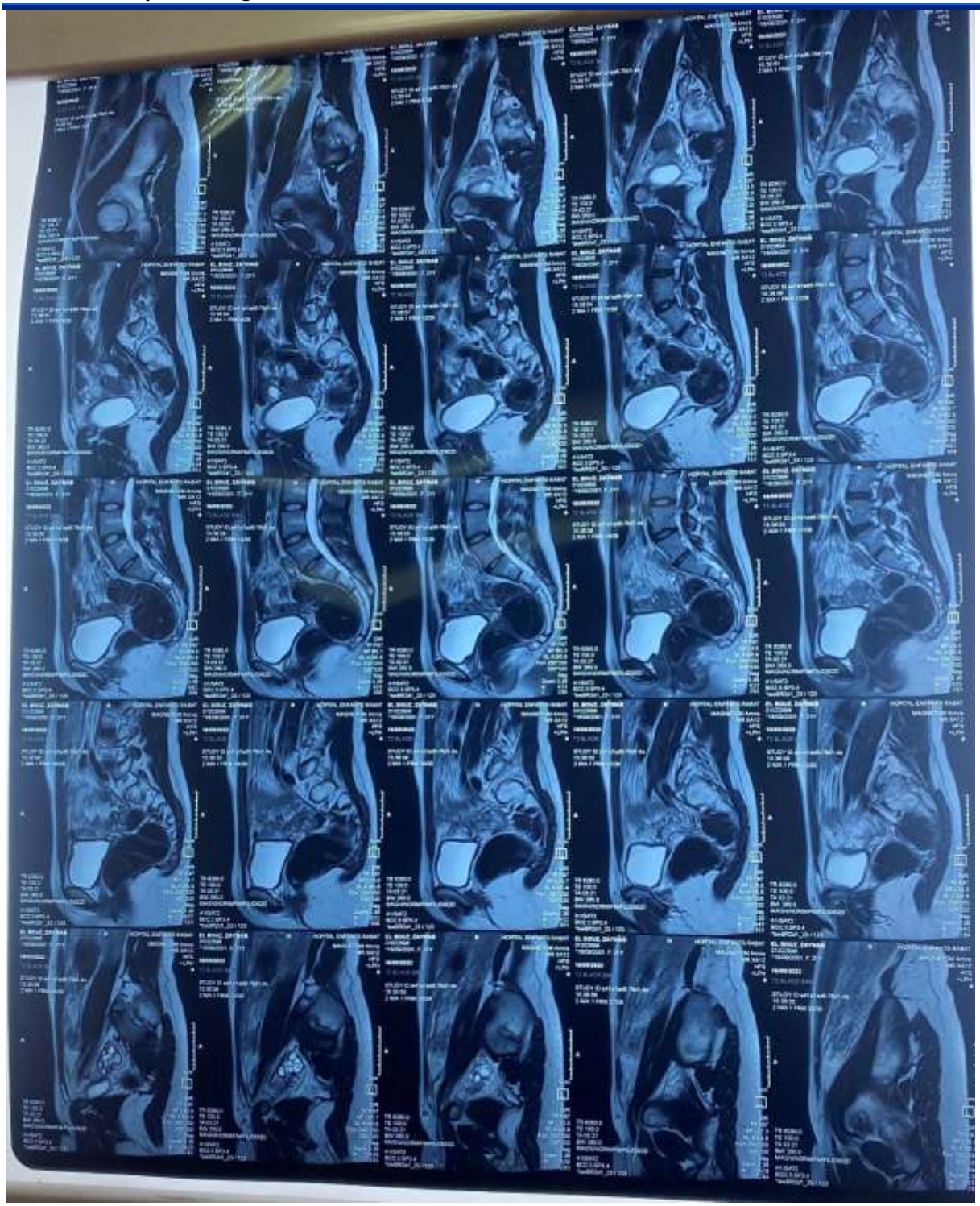

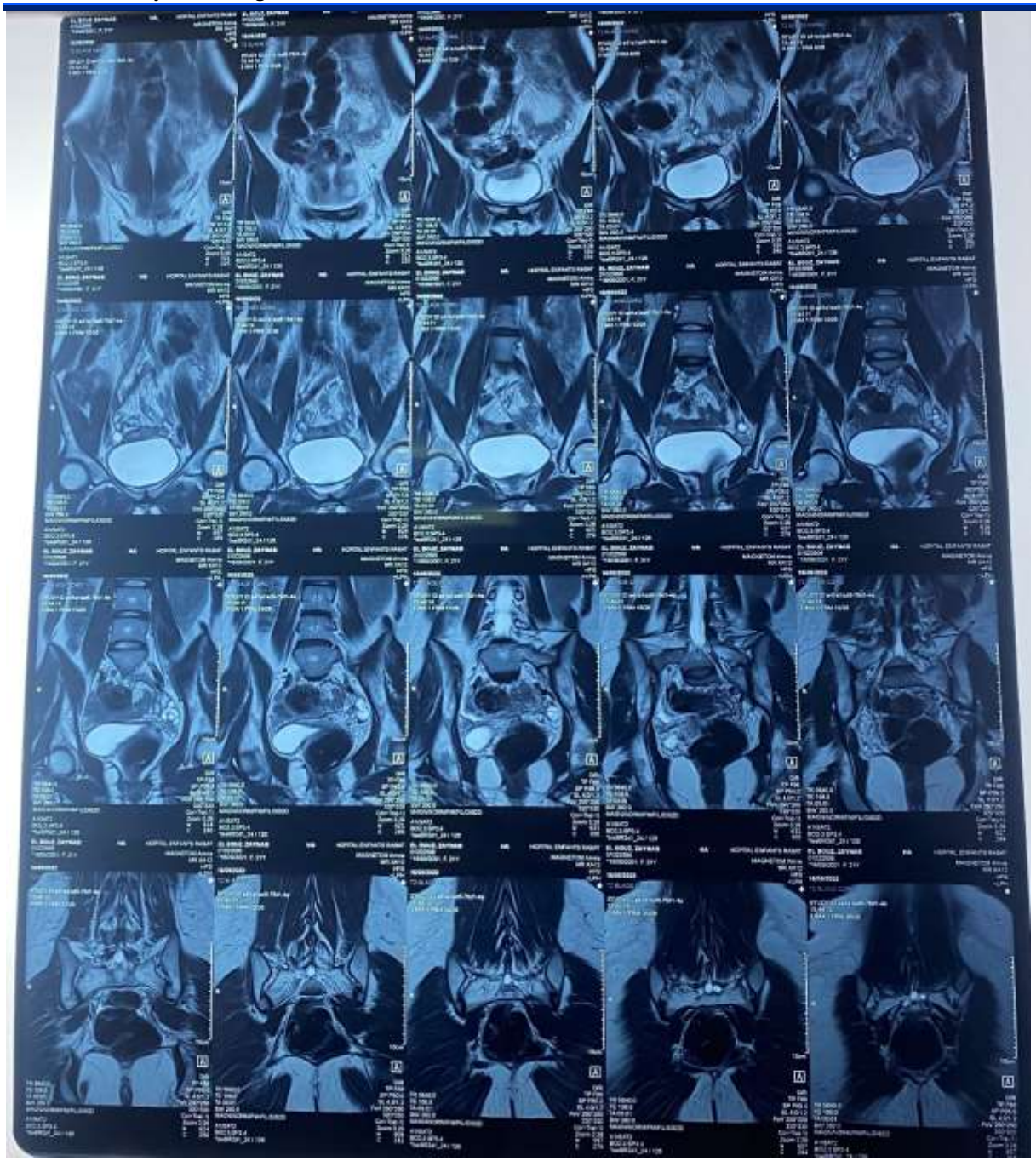

FIGURE 1,2,3 : Pelvic MRI: sequence; uterine agenesis and upper 2/3 of vagina

Discussion

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is defined by congenital aplasia of the uterus and upper two-thirds of the vagina in women with normal development of secondary sexual characteristics and a normal karyotype (46, XX) [2]. Two clinical forms have been described: MRKH type I, corresponding to isolated uterovaginal agenesis, and MRKH type II, characterized by incomplete agenesis and/or associated with other congenital malformations [3]. Our patient had MRKH type II. The incidence is estimated at one in 4,500 women [1]. The main clinical sign is primary amenorrhea; secondary sexual characteristics are present and well developed [4]. Genetic workup shows a normal blood karyotype (46, XX) with no visible chromosomal abnormalities [5

]. The endocrine workup (plasma FSH, LH and 17-estradiol) is normal, testifying to the integrity of ovarian function and the gonadotropic axis (FHS, LH) [5]. In our patients, hormonal and genetic tests were normal. Ultrasound examination via the suprapubic approach is a first-line method, which allows the diagnosis to be suspected by showing the absence of a uterine structure between the bladder and the rectum. Nevertheless, a quadrangular retrovesical structure may be mistakenly identified as a hypoplastic uterus, corresponding to the vestigial lamina located beneath the median part of the transverse peritoneal fold. An associated renal malformation should also be systematically sought during this ultrasound examination [6].

MRI is more sensitive and specific than suprapubic ultrasound. It enables a precise diagnosis thanks to the T2 sequence in sagittal and axial section, confirming uterine aplasia and the upper two-thirds of the vagina, and the normal appearance of both ovaries [7]. MRI can also be used to search for other associated malformations (kidney and bone). When a patient presents with primary amenorrhea and well-differentiated secondary sexual characteristics, the differential diagnosis is first of all vaginal atresia or a transverse vaginal septum, confirmed by careful clinical examination and the presence of a uterus on imaging [8 , 9]. The phenotype is very similar to that of MRKH, with primary amenorrhea, uterovaginal aplasia and possibly renal malformation. However, these abnormalities are associated with signs of hyperandrogenism (acne and hirsutism), correlated with plasma assays showing elevated testosteronemia [10]. Finally, androgen insensitivity syndrome is a male pseudohermaphroditism. The phenotype is female, with testes in the abdominal or inguinal position and a high testosterone level equivalent to that of the male subject. Treatment consists in reconstituting a neovagina, enabling the patient to enjoy a normal sexual life. Psychological support is essential for patients with MRKH syndrome [10].

Conclusion

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome should be considered in the presence of primary amenorrhea in a young woman with well-developed sexual characteristics. Diagnosis is based primarily on imaging, notably ultrasound and pelvic MRI. The latter is the examination of choice, using the T2 sequence to confirm utero-vaginal aplasia, ovarian integrity and the search for associated malformations, notably renal. Nevertheless, the possibility of confusion with other syndromes including uterovaginal anomalies requires knowledge of the various differential diagnoses.

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