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Mayer-Rokintansky-Kuster-Hauser Syndrome: A Case Report.

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ABSTRACT

The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome affects 1 out of 4500 women. It is characterized by the congenital absence of the upper third of the vagina, uterus and tubes. It is usually associated with renal malformations and patients show normal phenotype and genotype. The MRKH syndrome usually remains undetected until the patient presents with primary amenorrhea despite normal development of secondary sexual characteristics, so imaging evaluation can demonstrate in one setting, non-invasively, the anomalies in development of genital tract. Age at diagnosis is usually between 15-18yrs. Such a case is reported and a critical review of clinical evaluation, diagnosis and the management alternatives of patients with MRKH syndrome available in medical literature is made. The paper is intended to help establish the best diagnostic criteria and treatment options for a comprehensive therapeutic approach to MRKH patients.

Keywords: MRKH, Mullerian agenesis, Gonadal dysgenesis, MURCS, Vaginoplasty.

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INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser syndrome is an uncommon condition, with an incidence of one in 4000–5000 female births and is the second most frequent cause of primary amenorrhea after gonadal dysgenesis. Such a disorder is a form of Mullerian agenesis characterized by vaginal atresia and uterine/tubal abnormalities which may include absence or hypoplasia of the uterus and Fallopian tubes. The patients present karyotype of 46, XX and normal secondary sexual characteristics, as the ovaries are present and functional, but menstruation is absent.

Such syndrome is classified into three types. Type I is represented by abnormalities restricted to the reproductive system. Type II is an atypical syndrome, with asymmetric uterine remnants and abnormal uterine tubes associated with ovarian disease, congenital renal and bone abnormalities and hearing defects. Type III, so called MURCS type, involves uterovaginal hypoplasia or aplasia, renal, cardiac and skeletal malformations. Renal malformations include: unilateral agenesis, horseshoe kidney, ectopic kidneys and hydronephrosis. Bone malformations occur particularly with vertebral fusion and scoliosis. Cardiac alterations and digital alterations such as syndactyly and polydactyly are rarer than those previously mentioned [1-5].

The syndrome aetiology remains unknown, but the increased number of cases in familial aggregates raises the hypothesis of a genetic cause.

MATERIAL AND METHODS

An 18 year old female with primary amenorrhoea has reported to gynaecology out patient care, Kamineni Hospital, Narketpally, Hyderabad, Andhra Pradesh, India. There was no suggestive history in her family. On examination her secondary sexual characteristics were normal. External genitalia showed 1cm long vagina and neither cervix nor uterus was felt on bimanual examination. A complete workup, in the form of ultrasonography, hormonal profile and karyotype had been done to establish the diagnosis.

OBSERVATIONS

On clinical examination, patient showed the development of secondary sexual characteristics compatible with her chronological age. At gynaecological examination a grooved urethra with elevated edges was observed. Speculum examination was performed (Figure 1). Transabdominal ultrasonography demonstrated streaked ovaries and vagina showed an absent proximal one third with a blind end (Figure 2). MRI confirmed the absence of uterus (Figure 3). The serum hormonal levels were within normal range (Leutinising hormone 11.84 mlU/ml, Follicle stimulating hormone 5.2 mlU/ml, Prolactin 576 mlU/ml, Estradiol 664 mlU/ml).

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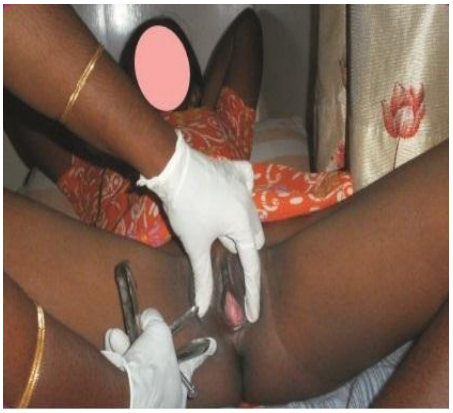

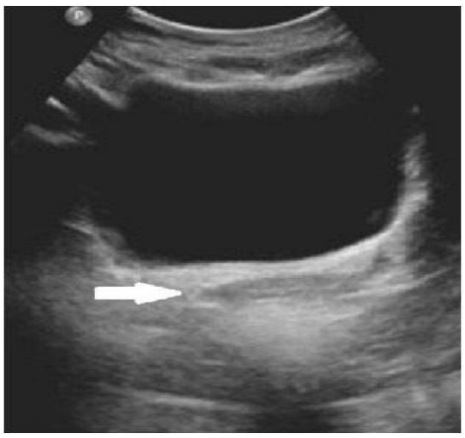

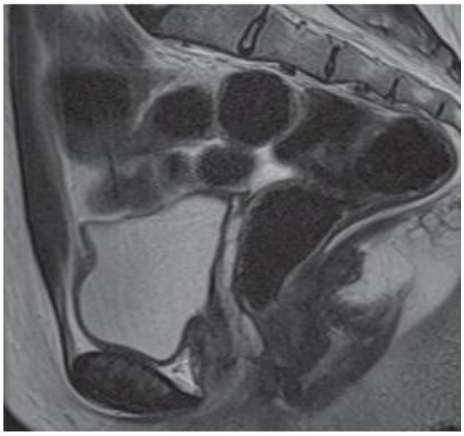

Genetic evaluation revealed a karyotype of 46, XX and micro deletion at 17q12, thus determining the diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome. The patient was submitted to surgical correction of the vagina (vaginoplasty).

DISCUSSION

The uterus, cervix and upper two thirds of the vagina form from the fused caudal ends of the mullerian ducts. The fallopian tubes develop from unfused upper ends. The renal system simultaneously develops from the wolffian (ie, mesonephric) ducts. The skeleton, which is derived from the embryonic mesoderm, is vulnerable to developmental disturbances at this time. In MRKH syndrome at approximately 5^{th} week of gestation, mullerian ducts stop developing. Ovarian function is preserved because the ovaries originate within the primitive ectoderm, independent of the mesonephros.

Although Mayer-Rokitansky-Kuster-Hauser syndrome was previously thought to be a sporadic anomaly, familial cases support the hypothesis of a genetic aetiology and are receiving increased attention. Frequent association with other malformations of renal, skeletal and auditory systems suggests the involvement of HOX genes. Mammalian HOX genes are well known for their role during organogenesis of axial skeletal, urogenital differentiation and

hindbrain development. Although the precise gene has not yet been identified, this syndrome appears to be transmitted in an autosomal dominant fashion, with incomplete penetrance and variable expressivity.

Imaging studies such as ultrasonography and magnetic resonance imaging are necessary to allow the determination of anatomic characteristics of the syndrome. Laparoscopy is indicated only in cases where the evaluation by the two previous imaging methods is inconclusive. Once the diagnosis is established**,** a clinical investigation should be undertaken to identify possible associated malformations.

The indicated anatomic treatment is the surgical or non-surgical creation of a neovagina, which may allow these patients to have a normal sex life. Patients who want to have children should be encouraged to adopt or the possibility of having biological children by means of assisted reproduction techniques should be suggested, considering that the presence of functional ovaries in these women allow the production of normal ovules [6-9].

CONCLUSION

Imaging studies such as ultrasonography and magnetic resonance imaging are necessary to allow the determination of anatomic characteristics of the syndrome. Because of the typical anatomic alterations, Mayer-Rokitansky-Kuster-Hauser syndrome generates anxiety and psychological distress with consequences on the patient's quality of life, which definitely require a multidisciplinary approach. Thus the impossibility of pregnancy may be alleviated by the recent developments in the management of this syndrome surgically or non-surgically, by the passage of time, by counselling, by family's support and by support groups.

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