

## Case Report

# Mayer-Rokitansky-Kuster-Hauser syndrome with gonadohypoplasia: a rare case report

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**Received:** 27 May 2016

**Accepted:** 02 June 2016

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### ABSTRACT

MRKH (Mayer Rokitansky Kuster Hauser) syndrome is a congenital abnormality seen in one out of 5,000 women characterized by the agenesis of the vagina, cervix, and uterus. It is also associated with kidney, bone and hearing difficulties. The ovaries are present with a normal function similar to that of a healthy reproductive woman's by producing eggs and female hormones. Chromosomes are the normal 46xx female karyotype. We report this rare syndrome in a 26-year-old female where she had presented with complaints of absence of uterus with the absence of left kidney. She didn't attain menarchy, secondary sexual characters are well developed. Small right ovarian follicular cyst with a rim of ovarian tissue was observed. She had undergone vaginoplasty.

**Keywords:** Congenital abnormality, Karyotype, Primary amenorrhea

### INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser syndrome is a rare congenital abnormality that affects women. It is characterized by the absence of the vagina, cervix, and uterus, which affects one in every 5,000 women.<sup>1</sup> They show primary amenorrhea with the development of normal secondary sexual characteristics during puberty. The disorder is generally of type I and type II. Type I which occurs as an isolated finding and type II occurs with abnormalities of additional organ systems mainly the kidneys and the skeleton.<sup>2</sup>

The exact cause of MRKH syndrome is idiopathic, but there is some evidence of genetic origin. Mainly in familial cases, MRKH is of familial origin with an inherited autosomal dominant trait showing variable expression and incomplete penetrance. Polygenic multifactorial inheritance may be another cause of MRKH syndrome.<sup>3</sup> Diagnosed is based on detailed patient history, characteristic symptoms, a variety of specialized tests such as specialized imaging techniques.

Transabdominal ultrasonography followed by MRI is usually done. Ultrasound depicts the uterus and vagina and also evaluates the kidneys which are a simple, non-invasive procedure that lacks radiation. MRI is also non-invasive and is generally more sensitive than an ultrasound which evaluates the uterus and vagina simultaneously along with evaluates the kidney and skeleton. Karyotyping is also performed to rule out other conditions.<sup>4</sup>

Females with MRKH syndrome have a normal 46, XX karyotype. In women diagnosed with MRKH syndrome, levels of LH (luteinizing hormone), FSH (plasmatic follicle stimulating hormone) and 17 $\beta$ -oestradiol are normal, proving the integrity of ovarian function. The treatment of MRKH syndrome depends on the age of affected individual at the stage of diagnosis. Counseling is the first choice to be opted primarily to have Psychological support. The treatment of vaginal aplasia consists of creating a neo-vagina for sexual intercourse. Treatment may be either surgical or nonsurgical. Usually, nonsurgical techniques are considered the first-line

approach. Plastic surgery can be done for artificial vagina (vaginoplasty). Females with MRKH syndrome cannot bear children (infertile). But by using in vitro fertilization of their own eggs and surrogate pregnancy they can have a child. The absence of kidney may result in increased susceptibility to urinary tract infections and renal calculi for which appropriate therapy is given.<sup>5</sup>

## CASE REPORT

A 26 year old female patient had presented with complaints of absence of the uterus with the absence of left kidney. She did not attain menarchy and secondary sexual characters were well developed. Her appetite, bowel, bladder habits were found to be normal. On systemic examination normal heart sounds were heard, bilateral air entry was normal, normal vestibular breath sounds were heard, the abdomen was soft and no organomegaly, higher mental functions, cranial nerves are normal.

Ultrasonography had shown the absence of left kidney, uterus, and ovaries. Rest of the abdomen was normal. Her biological markers were Estradiol-310.57, Progesterone-0.64mIU/ml, Prolactin-5.97ng/ml, Follicle Stimulating Hormone-5.64 mIU/ml, Luteinizing Hormone-8.44 mIU/ml, Sodium-136mmol/lit, Potassium-4.1mmol/lit, Hemoglobin-12g/dl, WBC-11000 cells/cum. Urine analysis: Color-Straw yellow color, P<sup>H</sup> -6, Specific gravity-1.015, Pus cells - 2-3, RBC-Nil, Epithelial cells-2-3, Casts-Nil, Crystals-Nil, Urobilinogen-0.2, Ketones-Nil.



**Figure 1(a) and (b): Chromosomal analysis showing 46, XX female karyotype.**

Her chromosomal analyses were found to be karyotype 46XX shown in Figure 1. Computerized tomography had shown the absence of the left kidney and uterus, small right ovarian follicular cyst with the rim of ovarian tissue and small streak left ovary was present. Based on these investigations she was diagnosed with Mayer-

Rokitansky-Kuster-Hauser syndrome for which she had undergone McIndoe's Vaginoplasty.

**Table 1: Laboratory analysis.**

Laboratory analysis	
Number of cells counted	25
Number of cells analysed	25
Number of cells karyotyped	05
Banding	G band
Average band resolution	450-550

## DISCUSSION

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a congenital disorder with uterine aplasia and upper 2/3 vaginal aplasia in phenotypically normal females. Women show normal 46, XX karyotypes normal functional ovaries and external genitalia<sup>6</sup>. There are two subtypes of MRKH: Type I (typical) or isolated and type II (atypical).

Among which the frequency of type II is much greater. The typical form is characterized by normal Fallopian tubes with symmetric muscular buds which are referred as the Rokitansky sequence, which affects the caudal part of the Mullerian duct (upper vagina and uterus). Type II shows asymmetric hypoplasia of buds with or without dysplasia of the Fallopian tubes.

This is often associated with renal defects (ectopia of one or both kidneys or unilateral agenesis or horseshoe kidney, in about 40-60% of patients).<sup>7</sup> The etiology of MRKH syndrome is idiopathic, but there is clear evidence of genetic origin. Polygenic multifactorial inheritance may be another cause of MRKH syndrome. Mainly in familial cases, MRKH is inherited as an autosomal dominant trait with variable expressivity and incomplete penetrance.<sup>8</sup>

In women diagnosed with MRKH syndrome, levels of luteinizing hormone, plasmatic follicle stimulating hormone and 17 $\beta$ -oestradiol are normal proving the integrity of ovarian function.<sup>9</sup> She had presented with karyotype 46, XX chromosome with normal development of secondary sexual characteristics and normal LH, FSH levels. Treatment depends on the age of individual at the stage of diagnosis. Counseling is the first choice to be opted to have Psychological support. Treatment may be either surgical or nonsurgical. Plastic surgery can be done for artificial vagina (vaginoplasty).

Females with MRKH syndrome cannot bear children (infertile). But by using in vitro fertilization of their own eggs and surrogate pregnancy they can have a child.<sup>10</sup> Skeletal abnormalities also require physical therapy, reconstructive surgery, and other medical management depending upon the severity of the bone deformities. The absence of kidney may result in increased susceptibility

to urinary tract infections and renal calculi for which appropriate therapy is given.<sup>11</sup>

*Funding: No funding sources*

*Conflict of interest: None declared*

*Ethical approval: Not required*

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**Cite this article as:** Chandini M, Ramineni HB, Rakesh Y, Babu YN, Vidyadhara S. Mayer-Rokitansky- Kuster-Hauser syndrome with gonadohypoplasia: a rare case report. *Int J Res Med Sci* 2016;4:3045-7.