

## **Mayer-Rokitansky-Kuster-Hauser syndrome with congenital absence of right radial artery: a case report**

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**Abstract:** Mayer Rokitansky-Kuster Hauser syndrome is a malformation complex comprising of absent vagina and absent or rudimentary uterus. MRKH syndrome may be attributed to initial affection of intermediate mesoderm leading to (by the end of 4th week of fetal life) an alteration of cervicothoracic somites and pronephric duct. The latter induces the differentiation of mesonephric and then Wolfian and Mullerian ducts. We present a case of 21 year old married female with primary amenorrhea and normal secondary sexual characteristics with congenital absence of radial artery. The diagnostic investigations demonstrated occurrence of Mayer Rokitansky-Kuster Hauser syndrome with unilateral absence of radial artery.

**Keywords:** Intermediate mesoderm, pronephric ducts, Mayer Rokitansky-Kuster Hauser syndrome.

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### **INTRODUCTION**

Mayer-Rokitansky-Kuster-Hauser syndrome is an uncommon condition, with an incidence of one in 4000–5000 female births [1,2], and is the second most frequent cause of primary amenorrhea after gonadal dysgenesis[3]. Such a disorder is a form of Müllerian agenesis characterized by vaginal atresia, and uterine/tubal abnormalities which may include absence or hypoplasia of the uterus and Fallopian tubes. The patient's present karyotype 46, XX and normal secondary sexual characteristics, as the ovaries are present and functional, but menstruation is absent [4]. Such syndrome is classified into three types according to the involvement of structures other than the ones related to the reproductive system. The typical syndrome (type I) is represented by abnormalities restricted to the reproductive system. The second one (type II) is an atypical syndrome, with the presence of asymmetric uterine remnants and abnormal uterine tubes. Such syndrome type may be associated with ovarian disease, congenital renal, bone abnormalities and hearing defects. A third one, the so called MURCS type(MU-Mullerian duct aplasia,R-Renal agenesis/ectopia,CS-cervical somite dysplasia.), involves uterovaginal hypoplasia or aplasia, renal, bone and cardiac malformations [1–3]. Renal malformations include: unilateral agenesis, horseshoe kidney, renal

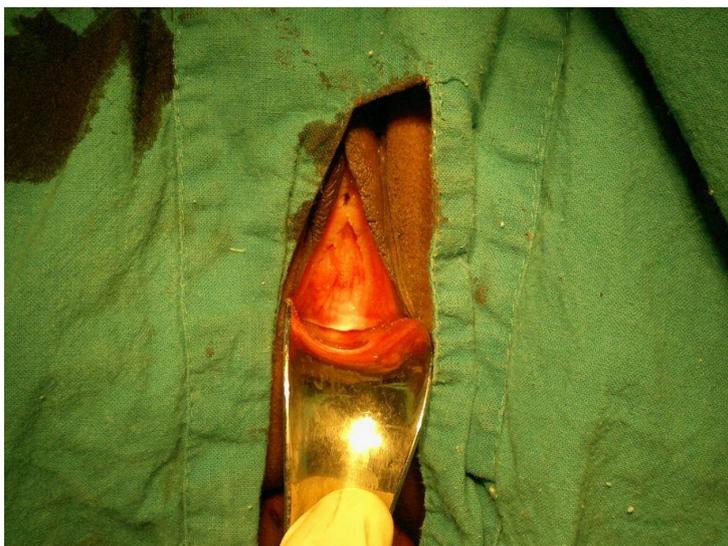
hypoplasia, ectopic kidneys and hydronephrosis. Bone malformations occur particularly in the vertebrae, most commonly with vertebral fusion (particularly cervical vertebrae), Klippel-Feil syndrome and scoliosis. Cardiac alterations and digital alterations such as syndactyly and polydactyly are rarer than those previously mentioned [1]. The syndrome aetiology remains unknown, but the increased number of cases in familial aggregates raises the hypothesis of a genetic cause [1]. The present report describes the case of a 21 year old female with primary amenorrhea and normal secondary sexual characteristics submitted to clinical, radiological investigations and diagnostic laparoscopy.

### **CASE REPORT**

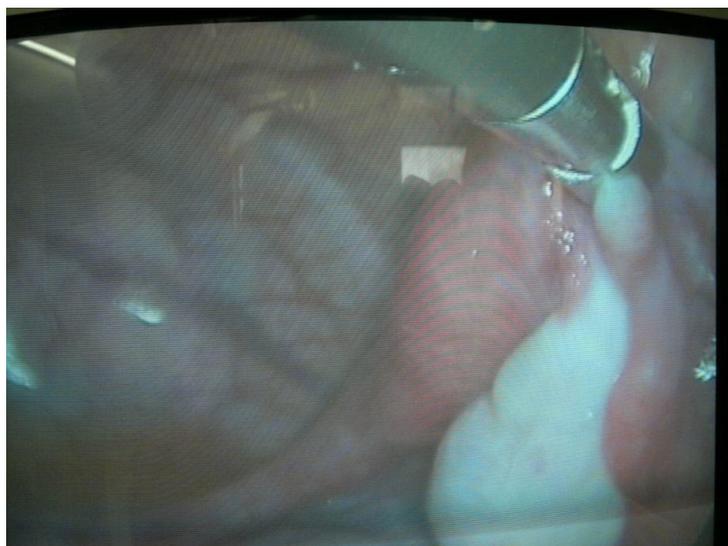
A 21 year old married Hindu female with no history of consanguinity in the family presented to gynae OPD with complaints of primary amenorrhea and primary infertility. She had no history suggestive of cyclical menstruation. She had active married life of 3 years & had no difficulty in intercourse. On general physical examination she was averagely built and averagely nourished & all the vital parameters were normal except for her right radial pulse which was absent. On clinical examination, the patient demonstrated a development of secondary sexual characteristics compatible with her chronological age.

On gynaecological examination vulva was apparently normal .Pelvic examination showed her vulva, labia minora and majora, and clitoris as normal and well estrogenised. The vagina was blind ending with a length of 6 cm .Cervix was not there. On P.V examination uterus and adnexa couldn't be palpated. On P.R examination uterus couldn't be felt but small masses of around 3 cm×2cm could be felt. All other investigations were normal except for USG which showed small infantile uterus and adnexa. Kidneys were normally placed in USG KUB region. X-ray chest PA view, lumbosacral spine and cervical spine were normal.ECG was normal. Her thyroid profile and FSH, LH, and estrogen levels were normal.

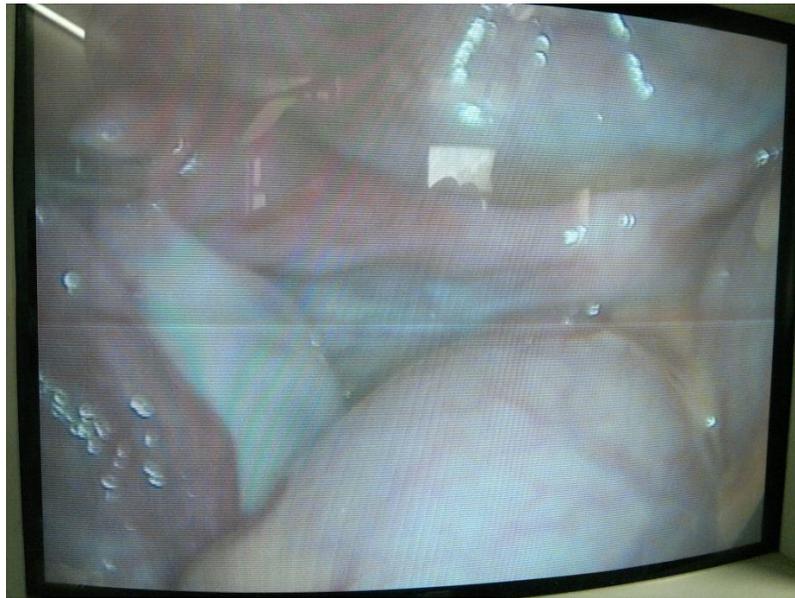
Buccal smear was chromatin positive indicating genetic sex to be female. Cardiologist was consulted for the absent right radial pulse which was diagnosed to be congenital absence of right radial artery.MRI and karyotyping could not be done due to financial constraints. She was posted for diagnostic laparoscopy which showed two small rudimentary uterine bulbs with well developed fallopian tubes with well developed ovaries. So she was diagnosed to be a case of Mayer Rokitansky-Kuster –Hauser syndrome with absence of right radial artery. Her post operative period was uneventful and was discharged with the advice of IVF with embryo transfer to a gestational carrier or adoption.



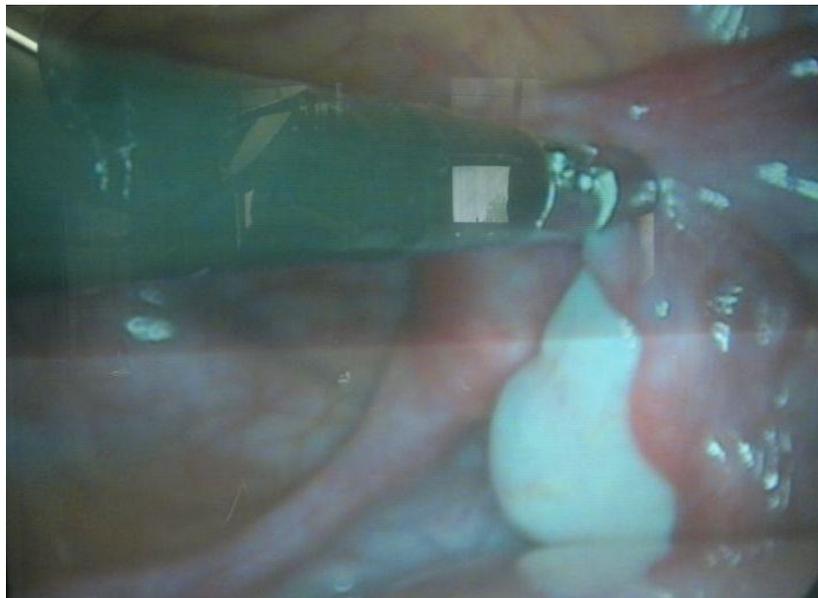
**Fig-1:Per speculum examination showing short vagina (nearly 6 cm) with cervical agenesis**



**Fig-2:Laposcopic view showing right sided uterine bulb with well developed fallopian tube and ovary**



**Fig -3: Laparoscopic view showing left uterine bulb with well developed fallopian tube and ovary**



**Fig-4:Laparoscopic view showing right sided uterine bulb with well developed fallopian tube and ovary**

#### **DISCUSSION:**

The typical clinical presentation of this syndrome is primary amenorrhea, in association or not with cyclic colic pain, in an adolescent with secondary sexual characteristics compatible with age, with no sign of virilization. Gynaecologic examination may detect either absence of the vaginal canal or vaginal shortening [1,2,5-8].

Imaging studies such as ultrasonography and magnetic resonance imaging, in association or not with laparoscopy, are necessary to allow the determination of the anatomic characteristics of the syndrome. Ultrasonography is the initial method of choice. This method can demonstrate the absence of the uterus

between the bladder and the rectum [1, 5, 9]. The vestigial lamina may be confused with the uterus, as it is found in its habitual site. Also, renal anomalies may be observed in cases of type II syndrome [1]. Magnetic resonance imaging is the most sensitive and specific imaging method in the evaluation of this syndrome, not only for allowing the acquisition of multiplanar images, but also for allowing the acquisition of sequences with fat saturation. It allows a good definition of anatomical alterations such as uterine agenesis, as well as evaluating ovaries, vagina and associated anomalies [1, 4, 5, 9]. Laparoscopy is indicated only in cases where the evaluation by the two previous imaging methods is inconclusive and provided this method allows the definition of a therapeutic strategy.

Once the diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome is established, a clinical investigation should be undertaken to identify possible associated malformations [1, 4]. Gardner *et al* reported a 32 year old woman with fusion of 2 cervical vertebra and unicornuate uterus who had radial ray anomalies with bilateral thenar hypoplasia and absent radial pulses. This case might represent an incomplete form of MURCS association or an example of an overlap between the MURCS and VATER (vertebral, anal, tracheoesophageal, radial) associations[10]. Amin MU *et al* reported a case of Mayer-Rokitansky-Kuster-Hauser syndrome associated with urogenital sinus anomaly, solitary left sided pelvic kidney and vesicovaginal communication manifesting as leakage of urine from vagina since birth due to absence of anterior wall of vagina and posterior wall of urethra[11]. The final diagnosis is achieved by the association of the imaging findings with the presence of the karyotype 46, XX. The differential diagnosis should be made with other situations where the patient presents primary amenorrhea and normal secondary sexual characteristics, such as congenital absence of uterus and vagina, isolate vaginal atresia with androgen insensitivity syndrome and transverse vaginal septum with imperforate hymen[1].

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, thus requiring a multidisciplinary approach [5, 6]. 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 [1–6]. As the surgical approach is chosen, uterine remnants can be removed to avoid future endometriosis [1]. 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 ova[12]. Even with the recent developments in the management of this syndrome, its diagnosis causes significant psychological distress, affecting the patients' quality of life because of the absence of menstruation and impossibility of pregnancy. The distress caused by the diagnostic may be alleviated by surgical or non-surgical treatments, by the passage of time, by counselling, by family's support and by support groups [12].

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