

A Young Asian Girl with MRKH Type B Syndrome: A Case Report

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Abstract

Mayer-Rokitansky-Kuster-Hausler (MRKH) syndrome is a rare congenital Müllerian duct malformation. Its incidence is 1 in 4500 female live births. It is one of the common causes of primary amenorrhea. The clinical features include primary amenorrhea, normal development of secondary sexual characters and blind vagina. Diagnosis is confirmed by ultrasound and MRI pelvis. The radiological features are aplasia of uterus and upper two third of vagina. Normal 46 XX genotype is found. It is of two types, type A which is isolated type and type B which is associated with other anomalies such as renal, cardiac, skeletal and adnexal pathologies. Treatment includes psychological counselling and surgical creation of neovagina. Pregnancy can be achieved by assisted reproductive techniques in woman with normal ovarian function. The authors hereby discuss the case via a case report of 16 year old Asian girl who presented with primary amenorrhea and found to have MRKH type B syndrome.

Keywords

Primary amenorrhea, Müllerian duct, MRKH syndrome

I. Introduction

MRKH syndrome is a rare congenital disorder characterised by uterine and vaginal malformation. It occurs due to failure of development of Müllerian duct leading to aplasia or hypoplasia of uterus and vagina. Its incidence is 1 per 4500 female births [1-3]. It is of two types:

- Type A: isolated utero-vaginal aplasia or rokityansky sequence.
- Type B: associated with other malformations such as Renal, cardiac, skeletal, fallopian tube or ovarian pathologies.

Woman with this syndrome presents with primary amenorrhea. It is characterised by normal secondary sexual characteristics, normal genotype, normal ovarian function in most of the cases and absent or underdeveloped uterus and upper part (2/3) of vagina.

Thus, authors hereby are going to review the case of MRKH syndrome through a case of 16 year old girl who presented with primary amenorrhea and was diagnosed with MRKH type B syndrome.

II. Case Report

A young sixteen year old Asian girl presented to the outpatient department of our hospital with complaint of primary amenorrhea. There was no history of cyclical abdominal pain. There was no significant past medical history or any surgery done in past. No similar history was present in first or second degree relatives. On examination, she had normal secondary sexual characteristics with normal breast development, presence of axillary and pubic hair. On local vaginal examination, blind

vagina was found. On per rectal examination, uterus was not palpable.

Blood investigations were done including haemogram and hormonal profile which were found to be in normal range. Value of FSH was 3.47 mIU/ml, LH was 4.32 mIU/ml, PRL was 12.3 ng/ml and TSH was 2.32 mIU/ml.

Ultrasonography was advised which revealed absent uterus with non visualisation of bilateral ovaries. For further confirmation of diagnosis, MRI abdomen with pelvis was done, on which following findings were found:

- Agenesis of uterus (figure I).
- Agenesis of upper one third of vagina (figure I).
- Absent bilateral ovaries (figure II).
- Absent left kidney with malrotated ectopic pelvic right kidney (figure III).

On karyotyping, normal 46 XX genotype was found. Hence diagnosis of MRKH type B syndrome was made. Patient and her family was counselled and patient was further advised for vaginal neoplasty.

III. Discussion

Primary amenorrhea is defined as failure to achieve menarche till age of 14 years in absence of normal secondary sexual characters or till 16 years irrespective of secondary sexual characters [4]. The most common cause of primary amenorrhea is gonadal pathology followed by MRKH syndrome [5].

The female reproductive tract develops from a pair of Müllerian ducts [6]. Following structures are derived from Müllerian duct: fallopian tube, uterus, cervix and the upper two-thirds of the vagina. The ovaries and lower third of the vagina have different

embryological origins. Ovaries are derived from germ cells that migrate from the primitive yolk sac while lower one third of vagina is derived from sinovaginal bulb.

Normal development of the Müllerian ducts depends on the completion of three phases: organogenesis, fusion and septal resorption. During first phase i.e. Organogenesis, bilateral Müllerian duct is formed. Failure of this phase results in agenesis or hypoplasia of uterus or unicornuate uterus. During second phase, fusion of Müllerian ducts leads to formation of uterus with a central septum. Failure of this step results in a bicornuate or didelphys uterus. Septal resorption is the third phase during which resorption of the central septum occurs. Failure of this stage results in a septate or arcuate uterus.

Based on the type of malformation, The American fertility association has classified Müllerian duct anomalies into 7 types as shown in figure IV. Of this, MRKH syndrome is class I Müllerian duct anomaly. It is further classified into:

- Type A: isolated syndrome characterised by absent uterus and upper two third of vagina with normal bilateral ovaries and no associated anomalies.
- Type B: associated with ovarian agenesis, renal anomalies, skeletal abnormalities, cardiac defects and hearing/ocular anomalies. Renal anomalies can be renal agenesis, ectopic kidney, hyperplasia, malrotation or horse shoe kidney.

In this case, patient was found to have MRKH type B syndrome with absent uterus and upper two third of vagina with bilateral ovarian agenesis and renal anomalies (left renal agenesis, right malrotated ectopic kidney in pelvis).

The incidence of MRKH syndrome has been estimated as 1 in 4500 female births [1-3]. The majority of cases appears to be sporadic [7], however familial cases have also been described [1,8-10].

For diagnosis of MRKH syndrome, imaging studies plays a significant role. The main options among imaging studies are ultrasound and magnetic resonance imaging (MRI). Ultrasound is the first modality which is easily accessible and readily available, on which uterus and bilateral adnexa can be visualised. However, it is not always effective in identifying underdeveloped Müllerian structures and ovaries, which are usually, located high in the pelvis, often at the level of the pelvic brim in these cases. Hence for confirmation and further management, MRI is the most useful diagnostic modality. It allows a good definition of anatomical alterations such as uterine agenesis, along with evaluation of ovaries, vagina and associated anomalies.

The main differential diagnosis of MRKH syndrome is androgen insensitivity syndrome (AIS), which is characterised by insensitivity to androgen receptors. However in AIS, genotype will be 46 XY, with normal testosterone levels and presence of intraabdominal testicles instead of uterus and ovaries.

Young women diagnosed with MRKH syndrome suffer from extreme anxiety and very high psychological distress when they are told they have no uterus and vagina. Thus, it is recommended that the patient and family should be counselled before and throughout treatment. Management of MRKH syndrome includes surgical creation of neovagina or vaginoplasty, which may allow the patients to have a normal sex life. In case of uterine hypoplasia, uterine remnants can be removed

to avoid future endometriosis. Patients who want to have children, can have biological children by means of assisted reproduction techniques in presence of functional ovaries, however in woman with ovarian agenesis, adoption should be encouraged [11].

IV. Conclusion

MRKH syndrome is a rare congenital syndrome but associated with acute distress to patient and family. Hence a woman with primary amenorrhea should be evaluated thoroughly and completely for making a proper diagnosis and further management accordingly. Family and patient should be counselled regarding prognosis of disease and future fertility.

V. References

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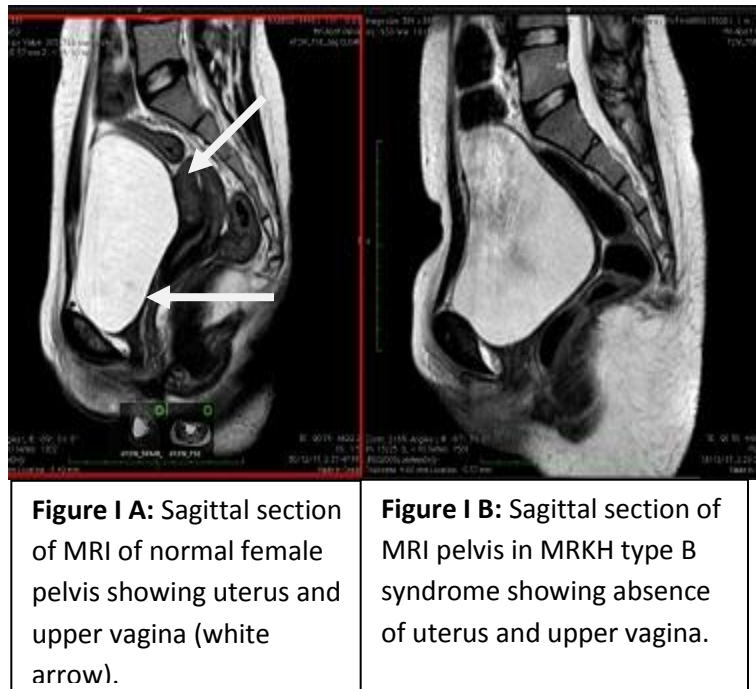


FIGURE I: Sagittal section of MRI pelvis in a normal female and a woman with MRKH type B syndrome.

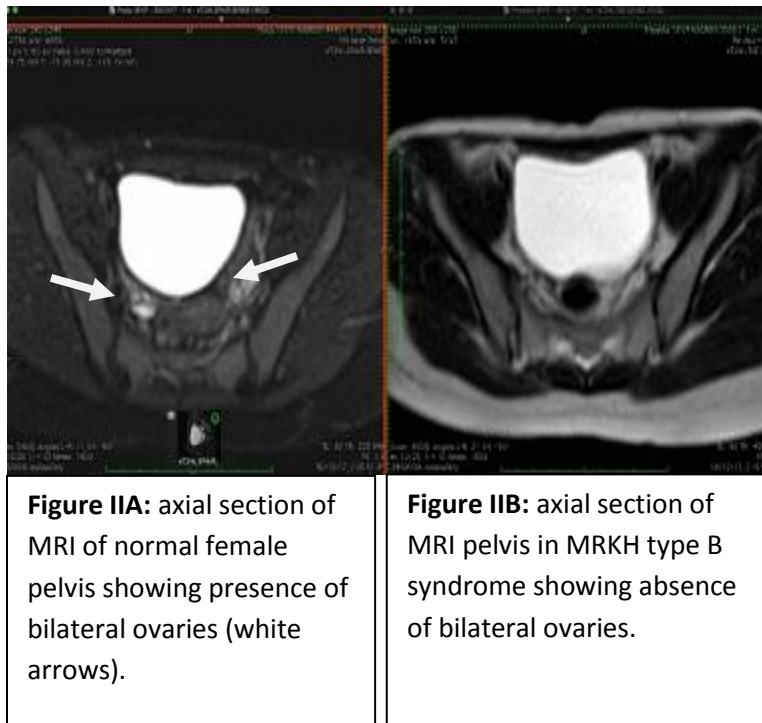


FIGURE II: Axial section of MRI pelvis in a normal female and a woman with MRKH type B syndrome.

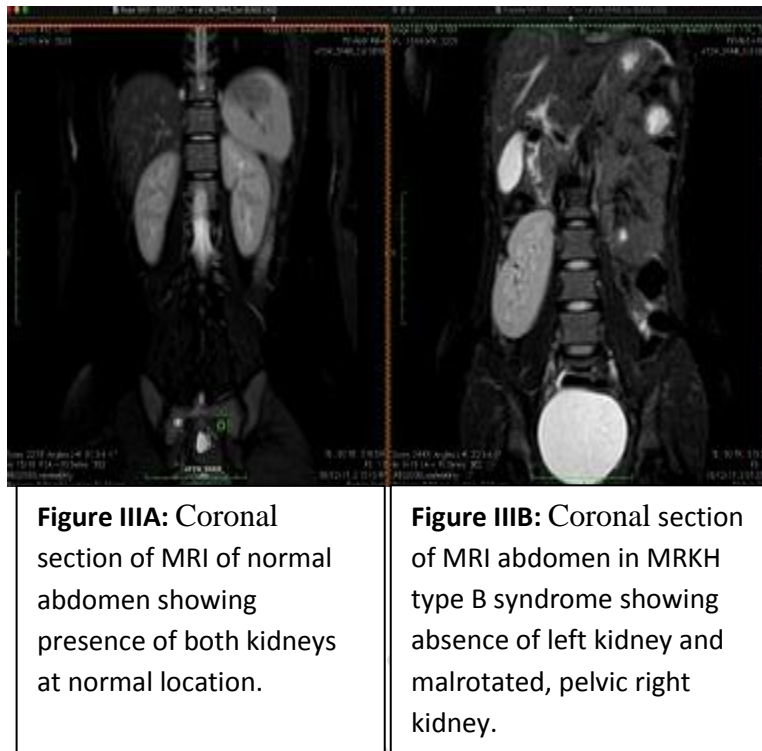


FIGURE III: Coronal section of MRI abdomen in a normal female and a woman with MRKH type B syndrome.

American Fertility Society Classification of Mullerian Anomalies			
I. Segmental müllerian hypoplasia or agenesis			
A. Vaginal			
B. Cervical			
C. Uterine fundus			
D. Tubal			
E. Combined anomalies			
II. Unicornuate uterus			
A. Communicating rudimentary horn			
B. Noncommunicating horn			
C. No endometrial cavity			
D. No rudimentary horn			
III. Uterine didelphys			
IV. Bicornuate uterus			
A. Complete (division to internal os)			
B. Partial			
V. Septate uterus			
A. Complete (septum to internal os)			
B. Partial			
VI. Arcuate			
VII. Diethylstilbestrol related			

Meckrage www.medicaps.com		I. Hypoplasia/Agenesis		II. Unicornuate		III. Didelphys	

Figure IV: Classification of Müllerian duct ano