Case report

Mayer-Rokitansky-Kuster-Hauser syndrome Type 2 or MURCS co-occurrence with Benign Ovarian Tumor – a tremendous case report

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Abstract

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKHS) is a congenital condition characterized by aplasia of the vagina with or without concurrent uterine and/or cervical aplasia.Between the two types ,type 2 or MURCS is a rare form including Mullerian agenesis,Renal agenesis and Cervicothoracic somite anomalies.A 17 years old virgin presented with feeling of heaviness and lump in abdomen for 6 months with history of primary amenorrhoea till the date and a diagnosed case of MRKH with normal female karyotype 46XX also having unilateral left sided renal agenesis and history of multiple surgery for congenital heart disease (TOF), repair of inguinal hernia and laparotomy for pelvic mass with biopsy taking. Evaluation of the patient suggests a tremendous presentation of MURCS association with benign ovarian tumor. A review of the literature reveals no other cases of MRKH with these tremendous anomalies having history of corrected TOF and inguinal hernia as well as with co occurrence of a huge benign ovarian tumor.

Key words: Mayer-Rokitansky-Kuster-Hauser syndrome, MURCS, Mullerian agenesis, Benign ovarian tumor.

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Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by congenital aplasia of the vagina with possible concurrence absence of the uterus and / or fallopian tubes but having normal female 46XX karyotype. There are two types, type 1 which has no other abnormality, type 2 comprises of Mullerian agenesis, Renal agenesis and Cervicothoracic somite anomalies (MURCS)- which is associated with renal and skeletal anomalies and less commonly auditory and cardiac defect.[1] MRKH is a congenital disorder of yet unknown etiology, the occurrence of ovarian tumor with it, is rare.[2] The incidence of MRKH has been recorded 1 in 4500 women.[1] Pelvic mass in MRKH are about uterine remnants, adenomyosis or fibroids in most publications but ovarian tumor could not be ignored as these patients do have ovaries.[3]

The inheritance mode appears to be of the autosomal dominant type[4] though it becomes sporadic in nature mostly.[5] MRKH syndrome is usually present in the form of primary amenorrhea with internal genitalia abnormalities.[3] There is normal secondary sexual characteristics and normal external female genitalia with about renal agenesis (25-50%)[1] The incidence of abnormally located ovary in MRKH is significantly increasing.[7] The second most common cause of primary amenorrhea after gonadal dysgenesis is MRKH syndrome.[6]

Case description

A 17 years old unmarried girl with a known case of MRKH syndrome associated with absence of left kidney and with history of multiple surgery for correction of Fallot's tetralogy that is for congenital heart disease at her 5 years of age ,repair of inguinal hernia on left side at her 7 years of age and at her 14 years of age laparotomy and biopsy taking from pelvic mass, presented to us at

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BSMMU in department of Gynaecological Oncology with the complaint of feeling of lump and heaviness in abdomen for 6 months duration and with all these history above mentioned with no other significant associated symptoms.

On examination, patient appeared well looking, no anemia or edema found, she was not icteric, she had no dehydration, had a normal neck appearance and size, BMI was within normal limit ,the Tanner staging for breast development was stage IV .

On chest examination revealed normal S1 and S2 heart sound and the lung Fields were clear.

Her abdomen was distended by a huge mass of 26cm in length and 22cm in breadth occupying hypogastric, both iliac, both lumbar, umbilical and partly epigastric region with mostly cystic in nature having soft to firm consistency and mobility in all direction where lower limit could be reached.

Right kidney was ballotable, no other organomegaly, no inguinal, axillary, supraclavicular or submandibular lymphadenopathy. Her external genitalia was grossly normal with normal pubic hair distribution.

Vaginal inspection was done, vagina was blind.

On per rectally, there was a mass felt with firm consistency but mass was high up, no deposit was found, rectal mucosa was free of any growth. There were no musculoskeletal or neurological abnormalities. Ultrasound showed huge cystic SOL with absence of left kidney. CT urogram showed left kidney not in usual position, possible ectopic left kidney. DTPA renogram showed left kidney nonvisualized with dilated right kidney.

Case management

The patient was counselled and was done MRI of whole abdomen with contrast, showed Large complex abdomino-pelvic mass (about 23cm×21cm×13cm) with possible ovarian origin, malrotated right kidney, non-visualized left kidney. All the tumor markers done were normal except slight raising of CA 125 (41 U/ml).

Pre-operative ureteric stenting was performed before planning of laparotomy with keeping facility for frozen section biopsy. She was also counselled for vaginoplasty but it was refused by them along with her legal gurdian on that time.

During surgery, after opening of abdomen with whole midline longitudinal incision, a huge mass of 26cm×22cm accordingly length and breadth was found on left ovary, right ovary was enlarged and cystic in nature, uterus was absent. Left sided salpingo oophorectomy was performed and sent for frozen section, report was negative for malignancy.

During through exploration appendix was found inflammed, appendectomy was performed.

Post-operative period was insignificant. Final histopathology report was 'Mucinous Cystadenoma'

Discussion

MRKH should be distinguished from hymen atresia, vaginal atresia and Complete Androgen Insensitivity Syndrome (CAIS).[8-10] It was first described by the German anatomist and physiologist Mayer in 1829, rather reported by Rokitansky (1839), Kuster (1910), Hauser and Scheniner (1961) then named MRKH (14).

MRKH syndrome is generally classified into two types, type 1 being seen in isolated cases of uterovaginal agenesis, while type 2 is seen in cases of uterovaginal agenesis associated with extragenital anomalies including urologic (e.g. renal agenesis, pelvic kidney, horseshoe kidneys), skeletal, auditory and cardiac anomalies, so called Mullerian hypoplasia, renal agenesis, cervicothoracic somite dysplasia (MURCS), it is the most severe form.[10]

Meyer-Rokitansky-Kuster- Hauser Syndrome: 46 XX

- Female with varying degrees of absence of mullerian structures
- Rectal exam and Ultrasound exam may show absence of uterus
- Usually have bilateral rudimentary uterine tissues, fallopian tubes and ovaries
- Neovagina by Frank dilators or McIndoe vaginoplasty procedure
- May have renal abnormalities (IVP needed) (18)

MRKH can confirm the presence of ovaries with absence of uterus.[15] MRI is an important imaging for diagnosis.[11,12] In regards to the management, the choice is of surgery.[13] considering the patient's clinical condition and the fact of huge ovarian mass to be finally confirmed for its nature, not in accordance with vaginoplasty.

The diagnosis of MRKH syndrome imposes a significant psychological burden on patients because of associated infertility.[15] The distress can be alleviated by psychological counselling and support. Treatment include progressive vaginal dilator or surgical neovagina creation by vaginoplasty. Assisted reproductive technique and surrogacy may be options with regard to fertility.[16,17]

To conclude, although MRKH with co existing congenital cardiac defects or dysplastic thumb have been reported but none have been observed simultaneously, in this case the tremendous presentation of corrected Fallot's tetralogy as congenital heart disease as well as

with associated benign ovarian tumor, thus this case is separating our patient from any previous reported case in the literature.

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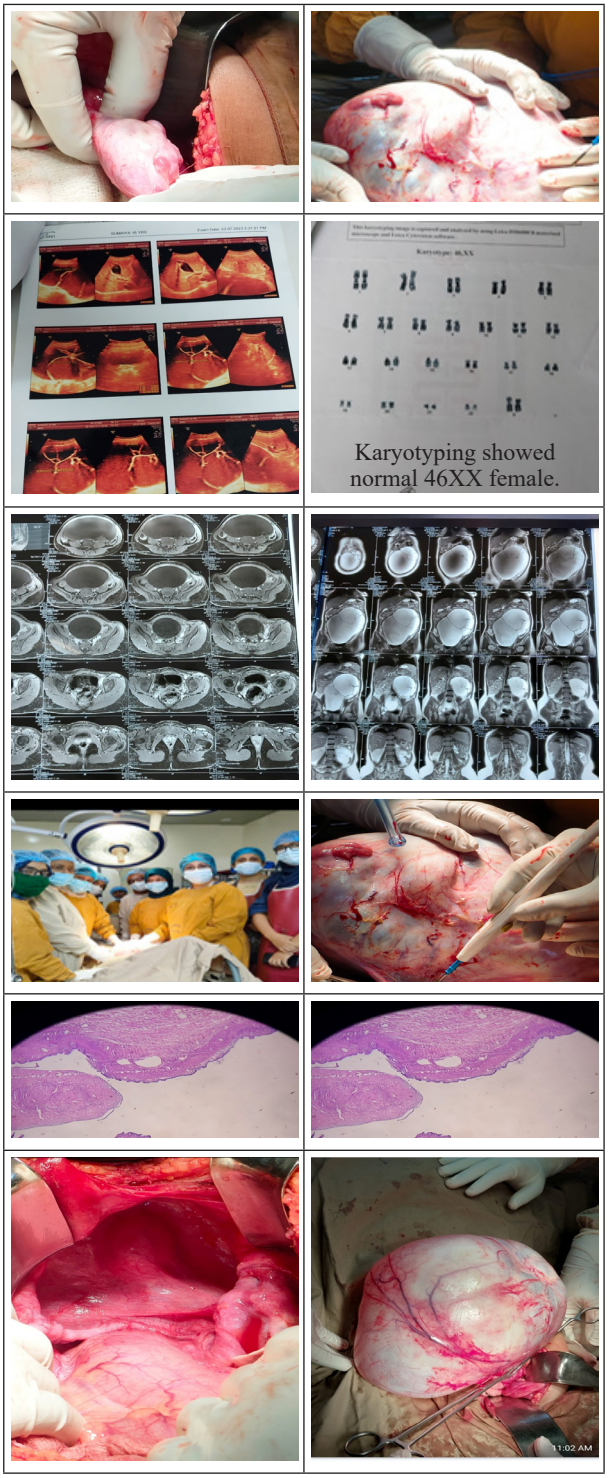

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