



Torsion of Ovarian Dermoid Cyst with MRKH Syndrome - A Rare Presentation

Authors

Dr Eram Ali, Dr Tamkin Khan, Dr Dalia Rafat

Department of Obstetrics & Gynaecology, JN Medical College
Aligarh Muslim University Aligarh

ABSTRACT

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and the upper part (2/3) of the vagina in women showing normal development of secondary sexual characteristics and a normal 46, XX karyotype. Women with this syndrome usually present at young age with primary amenorrhoea as first sign. Increasing number of familial cases of MRKH syndrome support the hypothesis of a genetic cause and the syndrome appears to be transmitted as an autosomal dominant trait with incomplete penetrance and variable expressivity. However, the etiology of MRKH syndrome still remains unclear. Association of MRKH syndrome with various malignant ovarian masses (sertoli and epithelial cell tumour) have been reported, however very few cases of MRKH syndrome with dermoid cyst of ovary has been reported till date. At present, ovarian pathologies are not considered to be part of the MRKH or MURCS clinical spectrum, as no association between these pathologies and utero-vaginal aplasia have been reported so far. Hence we are reporting this rare case of torsion of dermoid cyst ovary with MRKH syndrome with late and uncommon presentation.

Key Words: MRKH-Mayer-Rokitansky-Küster-Hauser, MURCS - Mullerian duct aplasia, renal dysplasia and cervical somite anomalies

INTRODUCTION

Mayer Rokitansky Kuster Hauser syndrome is second most common cause of primary amenorrhea after gonadal dysgenesis. Incidence of MRKH syndrome has been estimated as 1 in 4500 female live births. In 1829 Mayer described congenital absence of vagina as one of the abnormalities found in still born infants. Rokitansky in 1838 and Kustnerin 1910 described an identity in which vagina was absent and a small bipartite uterus was present with normal ovaries and anomalies of renal and skeletal system was frequently observed. Hauser and associates further

described the spectrum of anomalies associated with the syndrome hence over the years this disorder came to be known as Mayer Rokitansky Kuster Hauser syndrome. MRKH syndrome can be classified into two types, Type I MRKH, isolated MRKH or Rokitansky sequence and Type II MRKH or MURCS association (Mullerian duct aplasia, renal dysplasia and cervical somite anomalies). MRKH type I syndrome is characterized by complete uterus aplasia in the presence of two rudimentary horns linked by a peritoneal fold and normal fallopian tubes while Type II MRKH is characterized by uterine

hypoplasia accompanied by aplasia of one of the two horns or by a size difference between the two horn rudiments, with tubal malformation of one or both tubes Association of MRKH syndrome with various malignant ovarian masses (sertoli and epithelial cell tumour) have been reported. However vary few cases of MRKH syndrome with dermoid cyst of ovary has been reported till date ^[1]. In our case, the chief complaint of patient was pain lower abdomen and not coital difficulty, primary amenorrhoea or primary infertility for which she reported to outpatient of our hospital at age of 40 years. Hence we are reporting this rare case of twisted dermoid ovary with MRKH syndrome with late and uncommon presentation.

CASE SUMMARY

40 year old lady came to our outpatient department with complains of dull aching pain in right lower abdomen since 4 months which was gradual in onset progressive in nature non-radiating not associated with bladder and bowel complains. There was no history of cyclical abdominal pain, bleeding or discharge per vaginum. Her appetite was normal and she had normal sleep pattern and bowel habits. Patient married at the age of 27 years and she got divorced after 4 years of marriage because of inability to conceive. Patient again got remarried 1 year back. There was no history of coital difficulty, dyspyruria or bleeding while coitus. On general

physical examination her height was 155 cm, weight was 56 kg. Breast showed Tanner stage 5 development, axillary and pubic hair were normally developed. Respiratory and cardiovascular systems were normal.

On per abdominal examination there was tenderness present in right iliac fossa on deep palpation, however, there was no guarding or rigidity.

Per speculum examination showed blind vagina, with vaginal length of 6cm. On per vaginal examination, the length of vagina was confirmed to be 6 cm, fullness was felt in right adenexa, there was no palpable uterus, while of per-rectal examination, firm to cystic mass felt in right adenexa measuring about 5*5 cm, uterus could not palpated, left fornix was free and rectal mucosa free.

Differential diagnosis of MRKH syndrome, testicular feminization syndrome with right adenexal mass was made.

Her hormonal profile – serum FSH, LH, Prolactin, Thyroid was in normal range reflecting normal ovarian function.

On investigation, her karyotype was XX, USG and CT scan (Figure 1 & 2) findings suggested an ill-defined mass of 6*5.7 cm in right adenexa solid to cystic in consistency with areas of calcification suggestive of dermoid, however bilateral ovaries were normal in size and shape and uterus was not visualized.



Figure 1: Transverse section of pelvis CECT showing dermoid cyst ovary with absent uterus

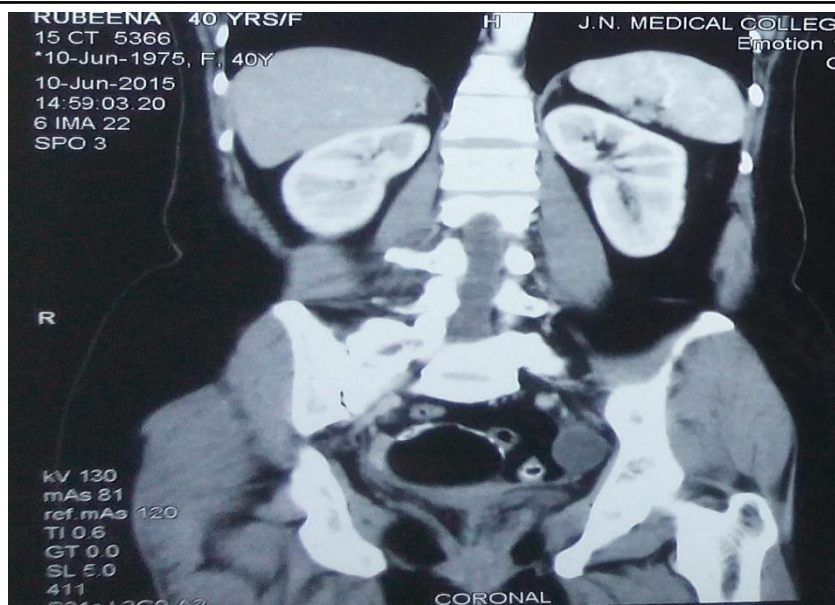


Figure 2: Saggital section of pelvis CECT showing dermoid cyst ovary with absent uterus

Examination under anaesthesia revealed blind vagina of 6 cm and absent uterus. Patient underwent laparotomy for the same condition, per op it was found that a mass 6*6 cm regular with smooth surface with three twist (Figure 3) at its pedicle arising next to right ovary on untwisting

the pedicle the length of the pedicle was found to be 3.5 cm. The pedicle was clamped cut and ligated, haemostasis was achieved. Bilateral ovaries were normal in size shape and texture. There was rudimentary stump at the site of uterus, suggestive of MRKH syndrome.



Figure 3: Per op finding of laprotomy- three twist at the pedicle of dermoid cyst

Cut section of ovary (Figure 4) showed predominately sebaceous material with hairs and histopathology showed tissue elements of all three layers endoderm, mesoderm and ectoderm, features consistent with dermoid cyst ovary.



Figure 4: Cut section of dermoid cyst showing hair and sebaceous material with calcific foci

DISCUSSION

According to the intra-operative findings, our patient fitted into category of MRKH type 1 syndrome. Since patient got remarried to a widower with two children, and there was no coital difficulty due to adequate vaginal length, patient did not report to hospital for treatment of primary amenorrhoea and infertility. It was torsion of dermoid cyst which gave rise to pain lower abdomen for which patient reported to hospital.

In literature association of malignant ovarian tumor have been described with pure gonadal dysgenesis, mixed gonadal dysgenesis and testicular feminization syndrome. Tsaur et al^[2] described the combination of MRKH syndrome and immature teratoma of the ovary in a 4-year-old girl while Hyo et al^[3] reported the case of serous papillary carcinoma of supernumerary ovary in a patient with MRKH syndrome. Very few cases of MRKH syndrome with Dermoid Cyst of ovary has been reported till date. At present, ovarian pathologies are not considered to be part of the MRKH or MURCS clinical spectrum, as no association between these pathologies and utero-vaginal aplasia have been reported so far.

The etiology of MRKH syndrome has remained quite unclear until now^[4, 5] although the spectrum of malformations encountered suggests a developmental field defect^[6, 7], involving organ systems which are closely related during embryogenesis. More precisely, MRKH syndrome may be attributed to an initial affection of the intermediate mesoderm, consequently leading (by

the end of the fourth week of fetal life) to an alteration of the blastema of the cervicothoracic-somites and the pronephric ducts^[7]. These latter subsequently induce the differentiation of the mesonephric and then the Wolffian and Müllerian ducts. Exact etiology of MRKH with dermoid is not known but atypical division of germ cell tissue is most commonly accepted theory.

Women with MRKH syndrome suffer extreme depression and anxiety, so the patient and the relative should be properly counselled with empathetic attitude.

With advancement in the treatment, invitro fertilization with own oocyte and surrogate pregnancy can be offered to patient of MRKH desirous of pregnancy.

CONCLUSION

Aplasia or absence of Müllerian derivatives suggestive of MRKH syndrome have been described in cases of gonadal dysgenesis or agenesis in XY or XO patients presenting with female phenotypes. At present, these types of ovarian pathologies are not considered to be part of the MRKH or MURCS clinical spectrum, since no single group of patients showing a random association between any of these pathologies and utero-vaginal aplasia has been reported so far. However, such studies should be undertaken on large cohorts of women with MRKH, to confirm this assumption.

REFERENCES

1. Mayur D, Tasneem D., Bava A.K., Michelle F. Mayer RokitanskyKuster Hauser syndrome with dermoid cyst. Bombay Hospital Journal 2010 ;52:502-505.
2. Tsaun GT1, Lee MH, Su SL, Wu MJ, Huang TW Mayer-Rokitansky-Kuster-Hauser syndrome with immature teratoma of the ovary at age 4 years. GynecolOncol. 1995 Mar ; 56(3):456-9
3. HyoSook Bae,1 Min Ji Ryu,1 In Sun Kixm,2 Sun Haeng Kim,1 And Jae Yun Song1 Cancer of the supernumerary ovary in Mayer-Rokitansky-Küster-Hauser Syndrome: A case reportOncolLett. 2013 Feb; 5(2): 598–600
4. Guerrier D, Mouchel T, Pasquier L, Pellerin I: The Mayer-Rokitansky-Kuster-Hauser syndrome (congenital absence of uterus and vagina)--phenotypic manifestations and genetic approaches. J Negat Results Biomed 2006 ; 5:1.
5. Kobayashi A, Behringer RR: Developmental genetics of the female reproductive tract in mammals. Nat Rev Genet 2003, 4:969-980.
6. Braun-Quentin C, Billes C, Bowing B, Kotzot D: MURCS association: case report and review. J Med Genet 1996 ; 33:618-620.
7. Duncan PA, Shapiro LR, Stangel JJ, Klein RM, Addonizio JC: The MURCS association: Mullerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia. J Pediatr1979 ; 95:399-402