Mayer-Rokitansky-Kuster-Hauser syndrome associated with serous papillary cystadenocarcinoma of the ovary

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Introduction

The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome occurs in 1 out of 4500 female births. It 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 with a normal 46XX karyotype. Their ovaries and fallopian tubes are normal. The coelomic epithelium which creates the ovaries develops independently of the Mullerian duct which creates the uterus, cervix and 2/3 of the upper vagina. Therefore, individuals with MRKH syndrome have normal ovaries and fallopian tubes and they are at a normal risk of developing ovarian malignancy.

Case report

A 47-year old unmarried woman was admitted to the gynaecological casualty unit at de Zoysa Maternity Hospital in November 2010 with complaints of abdominal pain for 2 weeks. She stated that she has never had menstrual periods. As she has not sought medical help, she was not investigated for amenorrhoea before. She had neither fever nor associated bowel or urinary symptoms. There was no family history of genital tract malignancies. On examination she was 150 cm tall and weighing 55 kg. Her cardiovascular system was normal. Gynaecologic examination revealed complete vaginal agenesis. Phenotypic sex was female; pubic hair and breast development were Tanner stage 5. Abdominal and rectal examination revealed a cystic non tender, mobile mass extending from pelvis corresponding to about 20 weeks size gravid uterus. Serum CA-125 level was elevated (2930 U/ml). Ultrasound scan showed 15 × 15 × 12 cm cystic mass with solid areas and multiple septae. Uterus could not be identified. There was no ascitic fluid. Both kidneys were normal in echogenicity. Intravenous urographic examination (IVU) showed a normal urinary tract. At laparotomy, partly cystic well-shaped mass was found arising from left adnexae. Uterus was formed of bilaterally rudimentary uterine bulbs. Left side was larger than

right. Right fallopian tube and right ovary appeared normal. Left fallopian tube was swollen and adhered to ovarian mass arising from left ovary. Tumor and the rudimentary uterine bulb was excised and sent for histology. Ascitic fluid was sent for cytology. There were no enlarged lymph nodes or tumor deposits in the abdomen or in the pelvis. Histopathological evaluation of the specimen revealed a rudimentary uterus with moderately differentiated serous papillary cystadenocarcinoma of the ovary. Further surgery is planned for a pelvic clearance prior to chemotherapy. Treatment MRKH syndrome depends on the symptoms, associated anomalies and the age of the patient. If the patient suffers pelvic pain due to a rudimentary horn, resection by laparotomy or laparoscopy may resolve the problem. If the diagnosis is made on an amenorrhoeic young girl, vaginoplasty could be done.

Discussion

Common presentations of MRKH syndrome are primary amenorrhoea, sterility and pelvic pain. Developing an ovarian neoplasm together with the syndrome is an unexpected rare state. For a long time the syndrome has been considered as a sporadic anomaly¹, but increasing number of familial cases now support the hypothesis of a genetic cause². But the etiology of MRKH syndrome still remains unclear. MRKH syndrome is subdivided into two types: type I (isolated) or Rokitansky sequence, and type II or MURCS association (Mullerian duct aplasia, renal dysplasia and cervical somite anomalies). MURCS association is more frequently associated with renal, vertebral, and, to a lesser extent, auditory, cardiac defects and digital anomalies (syndactyly, polydactyly). Type I MRKH syndrome is less frequent than MURCS association³. Complete aplasia of the uterus in the presence of two rudimentary horns linked by a peritoneal fold and normal fallopian tubes correspond to isolated MRKH syndrome type I. Type II MRKH syndrome is characterized by symmetric or asymmetric uterine hypoplasia, accompanied by aplasia of one of the two horns or by a size difference between the two horn rudiments, coupled with tubular malformations such as hypoplasia or aplasia of one or both tubes. Cases of polycystic ovaries⁴ and ovarian tumors⁵ have been described in women presenting with MRKH syndrome type II.

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Figure 1. Cyst and rudimentary horn.

Figure 2. Other ovary, tube and rudimentary horn.

Figure 3. Removed specimen.

Conclusion

Ovarian neoplasms can arise in patients with MRKH syndrome although this is rare. Their risk of developing gynaecological malignancies is no different from that of normal female population⁶.

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