Atypical Mayer–Rokitansky–Kuster–Hauser syndrome – report of a rare case

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Mayer–Rokitansky–Kuster–Hauser syndrome is a rare disorder affecting 1 in 5000 women. It is characterized by congenital absence of the uterus and the vagina in a phenotypically and karyotypically normal female. Here, we report a case of atypical Mayer–Rokitansky–Kuster–Hauser in a 13-year-old girl who also had abnormalities in the ovaries.

Keywords:
atypical, Mayer–Rokitansky–Kuster–Hauser syndrome, mullerian ageanesis

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Introduction

Mayer–Rokitansky–Kuster–Hauser (MRKH) is a rare disorder. Its incidence is ~1 in 5000 new born girls [1]. It is characterized by absent or hypoplastic uterus and upper two-third of the vagina in a phenotypically and karyotypically normal female. There are two main types:

(1) Typical type: with normal ovaries and fallopian tubes.
(2) Atypical type: with associated abnormalities of the ovaries, fallopian tubes, kidneys, or skeletal system. Teratogenic drugs (Thalidomides), some genes (GALT gene, CFTR gene, different candidate genes), and familial background (diabetes mellitus) have all been proposed as etiological factors, but the role of none is proved with certainty.

Our case is an atypical MRKH with bilateral ectopic ovaries, the exact incidence of which is unknown.

Case history

A 13-year-old girl presented with intense cyclic lower abdominal pain and both iliac fossa swellings for 3 months. General examination was within normal limits. Local examination indicated poorly developed secondary sexual characteristics. On palpation, there were two soft to firm round swellings about 2.5×1.5 cm each in both iliac fossa. On examination of the genitalia, labia majoras and minoras were normal (Fig. 1a), but the vagina was shallow and ended blindly. The urethral opening was normally placed and the clitoris was normal. Ultrasound (USG) examination showed absent uterus and ovaries and the iliac fossa swellings contained multicystic structures likely to be ovaries (Fig. 2b). All the other organs including the kidneys were within normal limits. The karyotype was 46, XX (Fig. 2a). Hormonal levels were normal. Fine-needle aspiration cytology from both the swellings showed germ cells and supporting cells (Fig. 1b).

A diagnosis of atypical MRKH with ectopic ovaries was made. The patient was placed on low-dose oral hormones to prevent cyclical pain and the option of surgical repositioning of gonads into the pelvic cavity was chosen to prevent torsion mainly. Vaginal reconstruction will be considered once the patient is physically and emotionally mature to start a sexual life. The patient as well as her parents were counseled regarding her future marital life and fertility options as far as financially feasible.

Discussion

The condition, MRKH syndrome, was first described by Mayer in 1829. Patients have a genetically normal karyotype (46, XX). The syndrome represents a developmental dysplasia of the mullerian duct system resulting in the absence of the uterus and the upper two thirds of the vagina [2]. Three different types of MRKH have been described. In the typical form (type 1), both the ovaries and the fallopian tubes are normal. Its incidence is 1 in 5000 new born girls [1].

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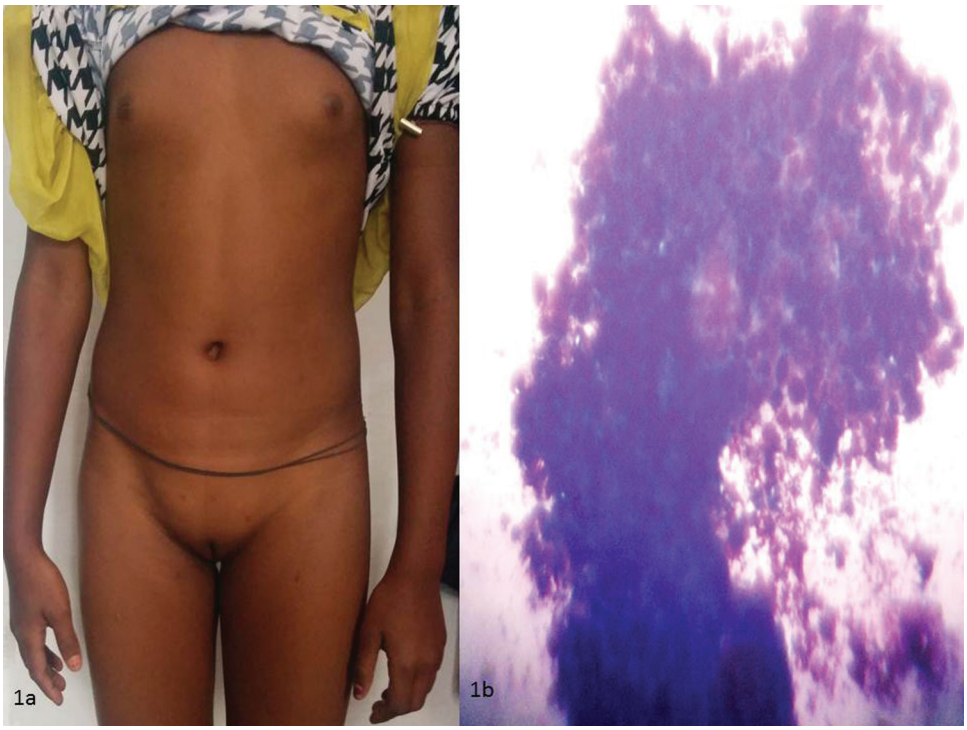
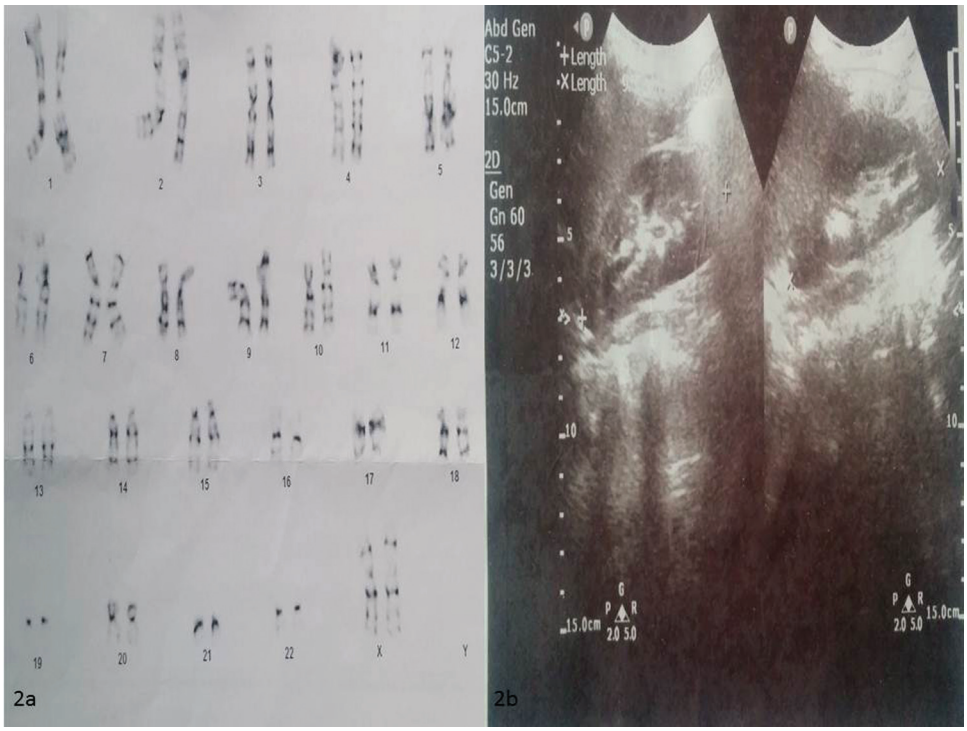
Figure 1



(a) Normal external genitalia and poorly developed breasts. (b) Cluster of germ cells on fine-needle aspiration cytology smear.

Figure 2



(a) Normal karyogram of the patient. (b) Ultrasonography image of cystic inguinal ovaries.

The atypical variant (type 2) may show hypoplasia or aplasia of one or both fallopian tubes. There may be the presence of cystic ovaries or other abnormal form or position of ovaries too [3]. Dysplasia of the urinary system may be present in type 2. The third type in addition to mullerian duct and renal dysplasia also has skeletal or cardiac malformations. The exact incidence of type 2 and 3 is unknown. The most common

presenting feature is primary amenorrhea with normally developed secondary sexual characteristics and normal external genitalia [4]. However, our patient had the atypical form, with ill-developed secondary sexual characteristics, along with primary amenorrhea. In 6–10% of MRKH cases, patients present with cyclic abdominal pain [3]. The etiology of this condition remains unknown [5]. Teratogenic drugs, some genes, and familial background have all been proposed as etiological factors, but the exact role of none is proved. It is not a life-threatening disease. With treatments, sexual relationships are possible and fertility options are also available. Ideal management consists of surgical reduction of the gonads into the pelvic cavity as soon as discovered to reduce the risk of torsion and loss of function [6]. Vaginal dilators or surgical reconstruction of the vagina should be considered when the patient is physically and emotionally mature to start a sexual life. Using assisted reproductive techniques, women with MRKH syndrome can have oocytes harvested, fertilized, and implanted in a surrogate. Adoption is another feasible option. Counseling of the patient as well as the parents is very important (Figs 1 and 2).

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Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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