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CASE REPORT

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Large leiomyomas in Mayer-Rokitansky-Küster-Hauser syndrome

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Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome (mullerian agenesis) is a cause of primary amenorrhoea; second in frequency only to gonadal dysgenesis. Women with this syndrome are characterized by presence of 46 XX karyotype, female secondary sex characters, normal ovarian functions including cyclic ovulation and undeveloped vagina. The uterus usually consists of rudimentary bicornuate cords [1,2]. Occurrence of leiomyoma in a patient with mullerian agenesis has not been described in the literature. We present a patient with mullerian agenesis who was found to have leiomyomas arising from both of the rudimentary horns of the uterus.

Case report

A 30 year-old married nullipara presented with a complaint of primary amenorrhoea. She had noticed a lump in the abdomen for the past 2 years. There was no history of cyclical abdominal pain, dyspareunia or hormone withdrawal bleeding. Her appetite remained normal and there was no weight loss. She did not have symptoms related to the bladder or the bowel. She neither had excessive growth of body hair nor a change in her voice.

Her physical examination showed a female body contour and normal hair pattern. Her height was 152 cm and weight 48 kg. Thyroid was not enlarged. There was no galactorrhea. On abdominal examination, she was found to have a 13 x 10 cm mass arising from the pelvis. The mass was irregular, mobile, non-tender and was occupying most of the right iliac fossa and hypogastric region. There was no free fluid in the abdomen. She did not have hepatosplenomegaly. The external genitalia were normal. Speculum examination revealed a blind 4 cm deep vaginal pouch and absent cervix. On bimanual examination, a firm nodule (2 x 2 cm) was felt in place of the uterus. The abdominally palpated mass was felt anteriorly on the right side of the nodule and a smaller mass (5 x 4 cm) of the same nature was palpable in the left fornix. Neither of these masses could be separated from the uterine nodule. Rectal examination confirmed the findings of bimanual examination.

The investigations revealed a normal intravenous pyelogram and a chromatin positive buccal smear. Hormonal profile showed serum follicle stimulating hormone (FSH) 5.4 mIU/mL; serum leutenizing hormone (LH) 13.2 mIU/mL; serum prolactin 15.9 ng/mL and CA-125 19.3 IU/mL. Trans-abdominal ultrasonography showed a hypoplastic 2.2 x 1.6 cm size uterus without an endometrial cavity. Two masses of mixed echogenicity measuring 10.3 x 6.3 cm and 4.1 x 3.9 cm and extending to both the adnexas were found superior to the uterus. Ovaries could not be seen. On MRI, two hypointense masses were found measuring 11.5 x 7.0 cm and 4.5 x 4.0 cm. Their lower poles were not seen separate from the uterus. Right ovary were normal and had normal follicles. The left ovary however, could not be identified.

With a provisional diagnosis of MRKH syndrome associated with two pelvic masses, she was taken up for a laparotomy. On opening the abdomen through a midline vertical incision, a rudimentary bicornuate uterus (1.5 x 1.5 cm) with a small cervix (1 x 1cm) was found. Two large leiomyomas, measuring 12 x 10 cm and 5 x 5 cm were seen arising from the right and the left horns of the uterus, respectively (Figure 1). A total abdominal hysterectomy was performed in view of the presence of large leiomyomas in a non-functional uterus. She had an uneventful recovery and was discharged

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on the 4th postoperative day. She was well at 6 months follow up.

On cut section of the specimen, uterine horns were found non-communicating and they did not have endometrial lining. Both the leiomyomas had whorled appearance and they did not show features of degeneration. Histological examination revealed features of leiomyoma with a hypoplastic bicormate uterus.

Figure 1. Large leiomyomas arising from horns of rudimentary uterus.

Discussion

MRKH syndrome is characterized by absent or undeveloped uterus and upper part of the vagina. Failure of fusion and development of mullerian ducts results in muscular thickening (analge) at the proximal end of each tube that are joined in the midline by a visible and palpable cord resembling hypoplastic bicornuate uterus without an endometrial lining [1]. Rarely, an active endometrium can exist with uterine analge, which becomes active in the presence of well estrogenised state. Reports have described patients with functioning endometrial tissue or even a hematometra in one or both of the rudimentary uterine horns [1]. Parikh [3] in his review of MRKH syndrome states that fibroids and adenomyosis rarely develop in the rudimentary non-functioning uterus. Enatsu et al [4] reported the first case of adenomyosis in MRKH syndrome.

The exact pathogenesis of neoplastic transformation of uterine smooth muscle in a patient with normal uterus is not known. Cytogenetic abnormalities in the form of spontaneous chromosomal rearrangements are known to occur in uterine leiomyomas. These chromosomal arrangements may be responsible for the initiation and progressive growth of the leiomyomas [2]. The leiomyomas are found to have higher concentraton of estrogen receptors as compared to normal myometrium. Therefore, they have more potential to proliferate in the presence of exogenous and endogenous estrogen [5]. Apart from that, insulin like growth factor 1 and 2 have also been implicated in the growth of leiomyomas [6].

As the proximal ends of mullerian ducts have smooth muscles, the presence of myoma in a case of mullerian agenesis is a theoretical possibility [1,2]. However, occurrence of large leiomyoma in a rudimentary uterine bulb has not been reported earlier. The possible reason for this uncommon occurrence could be a decreased concentration or sensitivity of the estrogen receptors or a lesser genetic predisposition for the clonal chromosomal abnormalities that are observed in women with normal uterus with leiomyomas [2].

In our patient, we could not perform cytogenetic and receptor studies to point out the exact etiopathogenesis of this unique occurrence of large leiomyomas in the rudimentary uterine bulbs. Hysterectomy was performed in our case in view of non-functioning uterus and risk of recurrence after myomectomy.

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