CASE REPORT

Mullerian, Renal and Skeletal Anomalies A Rare Association

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Abstarct

The MURCS syndrome is a very rare complex association of mullerian, renal tract and cervicothoracic somite anomalies. A case of a 17 years old girl with these associations and a management pitfall is presented and the embryology and management options discussed.

Key Words

Mullerian, Renal, Congenital Anomalies

Introduction

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKHS) is a congenital malformation characterized by an absence of the vagina associated with a variable abnormality of the uterus and the urinary tract but with functional ovaries (1). We present a rare case of this syndrome in association with a cavitated and another non-cavitated rudimentary horn, tubal luminal atresia, unilateral renal agenesis and cervical vertebral fusion and trace the embryologic origins and management options of this anomalous conglomeration.

Case Report

A 17 year old girl presented to the outpatient department with primary amenorrhoea. There was no history of abdominal pain. She was 125 cm in height, weighed 37 kgs and had a short broad neck with restriction in neck movements and normal secondary sex characters. Both breasts were in Tanner stage III of development. Respiratory and cardiovascular examinations did not reveal any abnormality. The abdomen was scaphoid, with no evidence of lump or organomegaly. Pubic hair were coarse, sparse and of feminine distribution. The external urinary meatus was normally situated but the vaginal introitus was blind and replaced by a dimple only. The perineal length was 4cm and uterus could not be felt on rectal examination.She had a normal echocardiogram and female karyotype. X-ray of the cervical spine revealed fusion of 5th and 6th cervical vertebrae (*Fig. I*). Abdominal ultrasonography showed a small (2cm x 2cm) sized uterus, normal ovaries and absence of right kidney, which was later confirmed on intravenous pyelogram. The serum thyroid profile, prolactin, follicle stimulating hormone and leutinising hormone levels were within normal range. After a complete preoperative workup, the patient was taken up for laparoscopy which revealed a small muscular nodule in place of uterus and normal ovaries. Reproductive prognosis was explained at the time of discharge from the hospital and vaginoplasty was advised before initiation of coital function. The girl returned after 6 months with acute cyclical abdominal pain for the past 3 months. Ultrasonography of pelvis detected a 6cm x 6cm sized mass with echogenic contents on the left side of pelvis. Only the right ovary could be visualized and appeared normal. Serum CA-125, alpha fetoprotein and beta-hCG levels were in the normal range. Magnetic Resonance imaging (MRI) of lower abdomen and pelvis suggested a thick walled cystic mass of 6 x 6cm and normal ovaries on both sides. Keeping the possibilities of a functional uterine horn or a twisted ovarian tumor, a laparotomy was carried out. A rounded 7 x 7cm sized uterine horn was visible towards the left side of pelvis with very thin 7cm long fallopian tubes attach to its left cornu (*Fig. 2*). A fold of peritoneum replaced

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the cervix. The right horn was a 2cm sized muscular nodule, located 3cm away from the lower part of the larger horn, with no evidence of right fallopian tube or round ligament. Both ovaries were normal in appearance. Excision of both horns of the uterus and left salpinx was done. The girl fared well postoperatively and was discharged after 7 days. Histology of the functional left horn revealed adenomyosis while luminal atresia was evident in the attached hypoplastic fallopian tube. The rudimentary right horn showed muscular elements, with no evidence of lumen / endometrium.

Discussion

When the dynamic processes of differentiation, migration, fusion and canalization are interrupted, a wide spectrum of mullerian duct anomalies (MDA) can result. The developing kidney and urinary system are closely related to the reproductive tract and their abnormalities are frequently associated. Asymmetric and obstructive fusion defects of the Mullerian system often coexist with unilateral mesonephric agenesis and manifest as ipsilateral renal agenesis, ureteric agenesis, or both (1). Disruption of the developing local mesoderm and its contiguous somites accounts for some associated axial skeletal abnormalities.Mayer Rokitansky Kuster Hauser syndrome (MRKHS) is characterized by congenital absence of vagina along with uterine aplasia or hypoplasia. According to the American Fertility Society (AFS) classification of the anomalies of mullerian ducts, MRKHS is the most common type I defect (2). There are wide variations in clinical presentations which makes the MDA's difficult to diagnose. Most of the abnormalities are associated with functioning ovaries and age-appropriate external genitalia. Primary amenorrhoea is the commonest presentation in cases of absence of a functioning uterus. In the event of a functional uterus or uterine horn, cyclic pain abdomen adds to the amenorrhoea. Adenomyosis and endometriosis externa

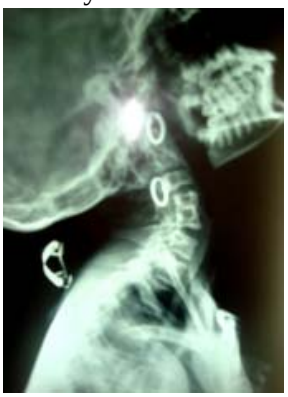

Figure 1: X ray Picture of Cervical Spine (lateral view) Showing Fusion of 5th and 6th Cervical Vertebrae.

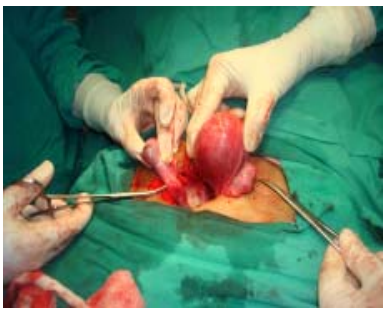

Figure 2: Intraoperative Photograph Showing the Enlarged Left Rudimentary Horn with Attached Hypoplastic Fallo pian Tube and Right Nonfunctional

(in cases of patent fallopian tubes) may be the sequelae of the obstructed menstrual outflow. The MRKHS may also be associated with various renal and skeletal anomalies, in view of the embryologic association of these systems. Klippel Feil syndrome, characterized by congenital fusion of cervical spine, short neck, a low posterior hair line and limited range of motion in cervical spine may coexist with MRKHS (3). An association of mullerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia called MURCS has also been documented (4).

Although the definite cause of vaginal/uterine agenesis in MRKHS is not known, a familial tendency is apparent in 4% cases (5). Mutations in either the antimullerian hormone or mullerian inhibitory substance (MIS) or its receptor gene have been proposed as the other possible etiologies for this defect (6). The presence of a hypoplastic fallopian tube and cavitated rudimentary horn on one side and a noncavitated rudimentary horn with absence of fallopian tube on the other side would appear to suggest the unequal development of paramesonephric (PMN) ducts of the two sides. Also the hypothesis of caudo-cranial direction of fusion of PMN ducts fails to explain the presence of rudimentary uterine horns and the absence of tube, cervix and vagina in the present case. A bidirectional fusion of PMN ducts during embryogenesis was later suggested by Duffy in 2004 (7). It is probable that a segmental fusion of the PMN ducts occurs which accounts for presence of varied stages of development of these ducts in one individual. This case appears to support the earlier theory that canalization of these ducts follows fusion and that it can proceed in any direction (8).Management needs to be individualized for each case. In the case of complete absence of uterus, vaginal agenesis is amenable to vaginoplasty for provision of coital function. However, the presence of a functional but occluded rudimentary horn compounds the problem and would require either an anatomic continuity with the exterior or excision of

the horn. Most cases of rudimentary horn of uterus are associated with a unicornuate uterus and are removed due to risk of ectopic pregnancy in case of patent ipsilateral tube or obstruction to the collected menstrual blood within the rudimentary horn if the tube attached to it is absent or occluded (9,10).

The creation of a neovagina with establishment of uterovaginal continuity or excision of the functional rudimentary horn by laparotomy along with vaginoplasty have been the two conventional surgical options for these patients and successful pregnancies have been reported after uterovaginal anastomosis in MRKHS (11).Creation of a neovagina by bilateral pudendal thigh fascicutaneous flap procedure and laparotomy to establish uterovaginal continuity has also been tried in a few cases (12). The main advantages of this modification are maintenance of sensation, no need for dilatation and an inconspicuous scar. Total laparoscopic reestablishment of uterovaginal continuity in MRKH syndrome with a functional horn, has been proposed as an alternative to the option of laparotomy and radical excision of the rudimentary uterus. Although this technique has the attendant advantages of minimal access surgery, the stenosis of the track is a risk here too. (13).Operative laparoscopy may be an alternative for the management of symptomatic rudimentary horns too. Laparoscopic ultrasonography may be of some value in picking up the thin endometrial echo in these cases (14).

In the present case, although the rudimentary uterine horns were diagnosed on the laparoscopic examination, they were not removed as the girl had presented only with primary amenorohoea at 17 years of age with no abdominal pain. Moreover, both the horns appeared to be tiny muscular bands, giving the impression of noncavitatory and nonfunctional horns on either side. However, subsequently, one of the horns which had a cavity and endometrium lead to onset of cyclic pain and enlargement of that horn. The subsequent surgery was required on account of this delayed onset of menstruation into one of the horns. Nevertheless, the commonly accepted benefits of minimally invasive surgery such as enhanced visualization of the cul-de-sac, decreased adhesion formation, smaller incisions, reduced postoperative pain, and shorter hospital stay are in favour of the laparoscopic approach. Surgical expertise, experience, and availability of instrumentation must also be considered.

Conclusion

It can thus be concluded that rudimentary uterine horns, irrespective of whether they exist in association with a patent uterovaginal tract or in isolation, should be removed. The assisted reproductive techniques and maternal surrogacy enable a woman without a uterus to have her own genetic children.

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