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RESEARCH ARTICLE

MAYER ROKITANSKY SYNDROM: A CASE REPORT AND REVIEW OF LITTERATURE

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Abstract

The Mayer-Rokitansky-Küster-Hauser syndrome is a rare disorder affecting women with normal karyotype and ovarian function. It is characterized by partial or total aplasia of the uterus and two thirds of the vagina. The main symptom is a primary amenorrhea and absence of the uterus which are diagnosed during examination and imagery. This paper reports the case of a 20 year-old woman diagnosed with the MRKH syndrome and discusses its psychological, fertility and sexual intercourse impact on patients. A large number of studies have been conducted to improve the management of patients. This article presents these studies and treatment options like a neovagina or a human uterine allotransplantation.

Introduction:-

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare disorder affecting women with normal karyotype and ovarian function (1,2). It is characterized by partial or total aplasia of the uterus and two thirds of the vagina. Examination and imagery are the main tools used for diagnosis. The main symptom is a primary amenorrhea and absence of the uterus which are diagnosed during examination and imagery. This paper reports the case of a 20 year-old woman diagnosed with the MRKH syndrome and discusses its psychological, fertility and sexual intercourse impact on patients. A large number of studies have been conducted to improve the management of patients. This article presents these studies and treatment options like a neovagina or a human uterine allotransplantation.

Case Report:

A 20-year-old nulliparous woman without any medical history or family history. The patient doesn't have any congenital abnormalities or family cases that are similar. Her mother's obstetric history was unremarkable, no teratogen medication or radiation during pregnancy. The patient has been married for a year andreported experiencing painfulsexual intercourse. Moreover, she presented a primary amenorrhea without any cyclic pelvic pain. A review of her pubertal development revealed pubarche at age 13. She has a normal height, weight, development of breasts, distribution of axillary, pubic hair (Tanner Stage 5) and external genital female. A bimanual pelvic examination revealed a narrow vaginal canal with a blind end. The ultrasonography didn't reveal any structure between the bladder and the rectum. The uterus and ovaries could not be visualized. MRI of the pelvis confirmed the diagnosis of MRKH with a unilateral renal aplasia. No other abnormalities were found.

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Discussion:-

The Mayer-Rokitansky-Küster-Hauser syndrome is a rare female condition that causes a total or a partial aplasia of the uterus and two thirds of the vagina with normal karyotype and ovarian function (1,2)

The Mayer-Rokitansky (MRKH) syndrome is a disorder that happens in females with no history intheirfamily and affects the reproductive system.It is present in 1 in every 4500 women (1-5) .It's a genital aplasia of the uterus and the proximal two thirds of the vagina with other müllerian duct abnormalities in women who have a normal ovarian function, a normal external genital and a normal female chromosome pattern: 46,XX (1,2).In type 1, the proximal two thirds of the vagina is absent. In type 2 or MURCS (Müllerian duct aplasia, renal aplasia, cervicothoracic, somite dysplasia), there are other malformations like :

1. vertebral or skeletal abnormalities: spinal bones in 20 to 26% of cases like scoliosis, hemivertebra, vertebral fusion,agenesis or costal malformation, spina bifida, sacralization of L5…
2. abnormalities at the extremities and the face: brachymesophalangia, ectrodactylia, supernumerary thumb, radial agenesis, Holt-Oram syndrome (atriodigital dysplasia), facial asymmetrysyndactyly, malformation of forearm bones) (1-3)
3. cardiac abnormalities (heart defects, rhythm disorders (right branch block), left-right shunt, conotroncal heart disease)
4. upper tract urologic abnormalities in 50% (kidney abnormality like unilateral renal agnesis (23-28%) and/or kidney in pelvic position (17%), renal hypoplasia (3) mega-ureter, horseshoe kidney)
5. and otologic abnormalities(less well-known like hearing loss in 10 to 25% of cases documented by Strubbe and al.) (1).

Type 2 is more frequent than type 1.Some authors suggested that the name MRKH syndrome should be reserved for pure genital abnormality, and that pathological associations should be referred to as GRES (Genital renal ear skeletal syndrome). The abnormalities of the pituitary axis are not classical, and ovarian function is usually considered normal (3).

The MRKH syndrome is due to an incomplete development of the Müllerian duct during the fifth week of gestation. This structure in the embryo develops into the uterus, fallopian tubes, cervix, upper two thirds parts of the vagina and renal system. Ovarian function is normal because ovarian develops from the primitive ectoderm, independent of the mesoderm (2).

The etiology of MRKH syndrome is still unknown. There is no link between exposure to teratogenic substances during pregnancy and the syndrome. Nongenetic and genetic causes are incriminated (4). Before, we thought that it was a sporadic cause but some family cases support the hypothesis of genetic etiology. The most commonly proposed mode of transmission is autosomal dominant with incomplete penetrance and variable expressivity (3). The main genes that may be involved are the genus of fetal development and sexual differentiation, such as HOX, WNT and those coding for the anti-müllerian hormone (AMH) and its receptor (4).

Clinically, the main symptom is a primary amenorrhea (2,3). However, the patient may suffer from catamenial pelvic pain and infertility. They frequently report dyspareunia with difficulty in sexual intercourse. Women with the atypical form of the syndrome have cyclic abdominal pain associated with signs of endometriosis due to retrograde menstruation (4). The ovarian function is normal. The age of diagnosis is 19 years, which may seem like a late age, since the mainsymptom is a primary amenorrhea. This late onset of the disease is related to the age at which sexual intercourse is envisaged (4). Physical examination revealsnormal secondary female sexual characteristics after puberty, a normal breast and pubic hair development. The patient's height is normal or shorter in some studies (3).The external genitals are female. The urethral meatus is sometimes gaping and located lower. In speculum examination, the vagina can be impossible to find because of the degree of vaginal agenesis. The digital rectal exam doesn't find any medial uterine structure (3).

The laboratory studies, especially chromosomal analysis exclude karyotype abnormalities. The circulating level of LH and FSH is normal without hyperandrogenicity (2,3).

The ultrasonographyis the first-line exam. It doesn't find any organ between the bladder and the rectum (ie uterus).Sometimes a vestigial blade can be mistaken for a hypoplastic uterus.Since this structure is not hollowed out by a cavity, there is no evidence of a hyperechogenic line, normally corresponding to the uterine mucosa. (3). It can

identify uterine duplication or tubal obstruction. It also shows kidney, bladder and vertebral abnormalities. The MRI is more sensible and specific. It can diagnose without doubt the uterine and vaginal aplasia and the present or missing cervix (2). The uterine aplasia is best seen in sagittal section while vaginal aplasia will be best highlighted in cross section. Malformation should be investigated in both the ultrasound and MRI. Renal and skeletal malformations may not be symptomatic, therefore an abdominal ultrasound or even a uroscanner can be done, or an x-ray of the spine.In case of clinical signs of otological and/or cardiac abnormalities, this assessment will be supplemented by an audiogram and/or a cardiac ultrasound. The presence of malformation should also be investigated in family members (3). Intravenous pyelography completes the exploration of renal structure.

The laparoscopy can be used after inconclusive MRI results or in the case of laparoscopic surgery (especially neo-vaginal one). This procedure provides the patient with a neo-vagina which enhances sexual function and enables reproduction with assisted technique. It also deletes uterine anlage to prevent endometriosis (2) .

In some studies, three points are noted: obesity level, ovarian hyperstimulation with high LH peak and the association with ovarian masses.MRKH syndromeis likely due to inappropriate secretion of anti-müllerian hormone (AMH) in the antenatal period or an activating mutation of the AMH receptor gene. However, ovarian disorder does not appear to be linked to hypersecretion of AMH (1).

The MRKH syndrome has a psychological, fertility and sexual intercourse experience impact.

Regarding infertility, the options available to patients desiring children areadoption or gestational surrogacy. However, in recent years, new alternative have emerged such as human uterine allotransplantation. It allows patients to conceive, gestate, and give birthto their own offspring. The uterine allotransplantationwas carried out in Sweden in 2014 in a trial with 9 patients. After the procedure, the first birth happened within the same year. Approximatively 60surgeries have been performed worldwide and more than 18 births occured. Having recourse to this surgery has increased exponentially. 9 out of 10 women who had a human uterine allotransplantation were diagnosed with MRKH (5).

The creation of a neovagina can be proposed in the case of sexual discomfort. The method undertaken (surgical or not) must be discussed with the patient and the medical team.The patient must be fully informed of the details of the procedure and should consent to it. Indeed, preserving the functional vaginal opening after any type of reconstruction, requires frequent intercourse or passive dilation. The various techniques proposed are as follows (4):

1. Franck's method: instrumental dilation of the vaginal cup by mandrel or candles of increasing caliber. This treatment allows the acquisition of a vagina covered with a normal vaginal mucosa and which requires maintenance by regular dilations, as well as an important motivation on the part of the patient;
2. The creation of a neovagina by the technique of Vecchietti: surgery based on passive traction. An olive-shaped mould is inserted into the vaginal cul-de-sac and connects to the abdominal wall through traction wires through the intervesical space, allowing progressive dilation of the vaginal cavity by daily traction;
3. The creation of a neovagina byDavydov technique: surgery realising a vaginal plasty with peritoneal tissue.

The psychological impact is an important aspect of this diagnosis. Women with MRKH have a higher prevalence of mood disorderscompared to women in the general population. They are more vulnerable, they have more expenses and have to accept a risky treatment in order to give birth to a child. They suffer from feelings of personal grief, female identity crisis, and jealousy toward fertile women, which all ultimately lead to feelings of isolation (5).

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