

Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKH)

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At Vydehi Institute of Nursing, we came across two cases with rare Mayer-Rokitansky-Kuster-Hauser Syndrome. One was a 19 years old female girl got admitted to the Vydehi Hospital, Bangalore with the complaints of primary amenorrhea. On examination the patient's secondary sexual characteristics were found to be normal. After the investigation, we noticed the absence of uterus and upper vagina with presence of normal ovaries. No other abnormalities were identified. She has two elder sisters and a younger brother. There was no history of delayed menarche in the mother and sisters. Hormonal assays were normal. The patient and parents were informed about the rare clinical entity and the treatment option. Patient underwent the counselling session on this rare disorder and also about her procreation.

The second case was that of a 34 year old female who got admitted to the Vydehi Hospital, Bangalore, for vaginoplasty. She had the history of primary amenorrhoea which was diagnosed 15 years back. Patient was suggested to undergo vaginoplasty six month before her marital life starts. She underwent McIndoe-Reed procedure and educated to use dilator for 3 months. So which condition led her to go for vaginoplasty?

Menstruation has long been an important societal marker of female sexual development and one of the most tangible sign of female endocrine and reproductive tract maturation. The mullerian ducts are the primordial anlage of the female reproductive system from where uterus develops. It differentiates to form the uterus, fallopian tubes, cervix and superior aspect of the vagina. A wide variety of malformation can occur when this system is disrupted.

What is MRKH Syndrome?

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare disorder which is characterised by congenital absence of uterus and the upper two-third of the vagina with normal ovarian function.

Mayer (1829) had first described a partial and complete duplication of vagina due to abnormal development of the Mullerian ducts in 4 stillborns along with other anomalies like cleft lip, limp, cardiac defects and urinary tract anomalies. Subsequently Rokitansky (1838) reported 19 cases of uterovaginal agenesis and 3 cases of renal agenesis. Kuster (1910) described several cases of similar anomalies with various musculoskeletal defects. Hauser (1961) emphasised the importance of distinguishing this syndrome from that of testicular feminisation in both of which vaginal development is defective.

Incidence

About 1 in every 5,000 female newborn babies have

this condition.

Types of MRKH Syndrome

Type I MRKH syndrome is characterised by an isolated absence of the proximal two-third of the vagina, whereas **type II** is marked by other malformations, including vertebral, cardiac, urologic (unilateral renal agenesis), and otologic anomalies (hearing loss).

Aetiology

The exact cause of MRKH syndrome is unknown. The reproductive abnormality of MRKH syndrome is due to incomplete development of the Mullerian duct.

Research is ongoing to determine the exact underlying causes of MRKH syndrome including identifying the gene or genes involved in the development of the disorder. The abnormal gene can be inherited from either parent, or can be the result of a new mutation (gene change) in the affected individual. The risk of passing the abnormal gene from affected parent to offspring is 50 percent for each pregnancy regardless of the sex of the resulting child.

Chromosomes, which are present in the nucleus of human cells, carry the genetic information for each individual. Each chromosome has a short arm designated "p" and a long arm designated "q". Chromosomes are further sub-divided into many bands that are numbered. For example, "chromosome 1q21.13" refers to band 21.1 on the long arm of chromosome 1. The numbered bands specify the location of the thousands of genes that are present on each chromosome.

To date, seven deletions and a duplication of chromosomal segments have been identified in several persons affected by MRKH syndrome and the seven

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segmental deletions likely to be involved in MRKH syndrome have been identified in chromosomes 1 (1q21.1), 4 (4q34-qter), 8 (8p23.1), 10 (10p14-15), 16 (16p11.2), 17 (17q12) and 22 (22q11.21), and the duplication was found on the chromosome X (Xpter-p22.32). This has led researchers to define several candidate genes: HNF1B (formerly TCF2), LHX1, TBX6, ITIH5 and SHOX, which are currently under investigation.

Clinical Manifestations

- ❑ The patient undergoes puberty with normal thelarche (breast development) and adrenarche (pubic hair); however, menses do not begin (i.e., primary amenorrhea).
- ❑ Patients may report cyclic abdominal pain due to cyclic endometrial shedding without a patent drainage pathway.
- ❑ Ovarian function is normal, patients experience all bodily changes associated with menstruation.
- ❑ Infertility.
- ❑ Difficulty with intercourse.
- ❑ Voiding difficulties, urinary incontinence, or recurrent UTIs.
- ❑ Vertebral anomalies (most commonly scoliosis).

Investigations

The most common age for MRKH to be diagnosed is when a young girl is between 15 and 18 years old with the complaint of not attaining menarche.

Physical examination findings are as follows:

- ❑ Normal secondary female sexual characteristics are present after puberty.
- ❑ Speculum examination of the vagina may be impossible or difficult because of the degree of vaginal agenesis.
- ❑ The vulva, labia majora, labia minora, and clitoris are normal.
- ❑ Chromosomal analysis to exclude karyotypic abnormalities.
- ❑ Circulating levels of luteinising hormone (LH) and follicle-stimulating hormone (FSH), which are normal in MRKH syndrome, indicating appropriate ovarian function.
- ❑ Testosterone levels can be assayed and are in the normal female range.

Imaging modalities used for MRKH syndrome include the following:

Ultrasonography

- ❑ Easily depicts the upper level of the vagina and

the length of its obstruction.

- ❑ Can identify uterine duplications and tubal obstruction.
- ❑ Allows simultaneous assessment of the kidneys and bladder for abnormalities and visualisation of some vertebral anomalies.

Magnetic Resonance Imaging (MRI)

- ❑ Provides excellent images of superficial and deep tissue planes.
- ❑ Can clarify inconclusive ultrasonography results concerning cavitation of the uterus.
- ❑ Improves assessment of sub peritoneal structures and detects the presence of a cervix.
- ❑ Can be used to image the spine if vertebral anomalies are suspected.
- ❑ MR urography (MRU) is an excellent imaging modality for visualisation of both the reproductive and the urinary anatomy and its function.

Laparoscopy

Laparoscopy is used in patients who have abdominal pain to evaluate and possibly resects (therapeutic laparoscopic surgery) the mullerian horn.

Pyelography

- ❑ It is performed to assess renal structure.
- ❑ Retrograde pyelography can be used to assess the renal collecting system, and it does not require intravenous contrast injection but does require cystoscopy.

Management

Treatment of such patient needs repeated psychological counselling. Often they are depressed concerning their sexual and reproductive life. Treatment options are: 1. Non-surgical method, and 2. Surgical methods.

1. Non-surgical Method

Frank technique

- ❑ Repeated use of graduated vaginal dilators for a period of 6-12 months. Presence of vaginal dimple (1 cm) is often seen.
- ❑ The technique is self-administered and requires time and patient motivation.
- ❑ Compliance may be poor in patients with a vaginal dimple or no vagina, because these patients may experience discomfort and abandon the di-

lator.

2. Surgical Methods

Various techniques of vaginal reconstruction (vaginoplasty) are done.

i. *McIndoe-reed technique (1938)*

- ❑ The most common surgical procedure used for vaginal reconstruction.
- ❑ A split-thickness skin graft is the most popular tissue for vaginal replacement, with the thigh or buttocks preferable as a graft donor site.
- ❑ The surgeon uses blunt dissection to create a pocket between the urethra and rectum; a cylindrical stent covered with the skin graft is placed into the potential space, and the graft is fixed into place by attaching cut edges of the skin incision to recreate the introitus; the labia majora are then sutured loosely together to hold in the mold.
- ❑ The stent is removed about 1 week later, and the patient uses a mold or dilator in the neovagina every day and night for 3 months to prevent contraction.
- ❑ Disadvantages of this procedure include scarring at the donor site, neovaginal stenosis, and the need for long-term dilation.

ii. *Williams vaginoplasty*

- ❑ It is particularly useful for patients with previously failed vaginoplasty.
- ❑ Vaginal pouch is created from skin flaps of labia majora in the midline.
- ❑ Although this simple procedure does not damage the urethra or rectum, dilation is needed for a lengthy period. The vagina created by this method is not anatomically similar to a normal vagina. Instead the vaginal pouch axis is directly posterior and horizontal to the perineum.

iii. *Rotational flap procedures*

- ❑ Use the pudendal thigh, gracilis myocutaneous, labia minora, and other fasciocutaneous flaps.
- ❑ Disadvantages of these techniques include extensive skin scarring at the donor graft site and the need for patient diligence in postsurgical dilation.

iv. *Intestinal neovagina*

- ❑ This is a major procedure uses an isolated segment of bowel for vagina.
- ❑ The recovery involves 4-6 weeks of healing from

major surgery.

- ❑ Sigmoid is generally the preferred bowel segment, as it can most easily be mobilised to the perineum in a tension-free manner.
- ❑ Patients who have undergone this reconstructive technique report a high degree of satisfaction.
- ❑ Young women who undergone this procedure usually experience chronic vaginal discharge requiring the need to wear a pad all the time.

v. *Vecchietti technique*

- ❑ Exerts continuous progressive pressure by an acrylic olive passed through the potential neovaginal space and the abdominal wall.
- ❑ A traction device is placed into the peritoneal cavity and gradually draws the olive upward over a period of days to weeks; this gradually lengthens the vaginal vault.
- ❑ This technique is now performed laparoscopically.

Nurses' role in patient education after vaginoplasty

The nurse should be educated about:

- ❑ The importance of continuous prolonged dilatation as well as stent care during the healing phase need to be emphasised.
- ❑ The foam is worn continuously for 6 weeks and is removed only during urination and defaecation.
- ❑ The foam is cleaned with the povidone-iodine solution covered with a fresh condom, lubricated and reintroduced into the neovagina.
- ❑ Low-pressure douche with warm water should be performed regularly.
- ❑ Genetic offspring are possible through use of a gestational carrier as the ovaries are normal.
- ❑ People with this condition can have genetic children through IVF with embryo transfer to a gestational carrier. Some also choose to adopt. Treatment by uterine transplantation is still in infancy stage in India.

Uterine Transplantation in Sweden

In October 2014 it was reported that a 36-year-old Swedish woman became the first person with a transplanted uterus to give birth to a healthy baby. She was born without a uterus, but had functioning ovaries. She and the father went through IVF to produce 11 embryos, which were then frozen. Doctors at the University of Gothenburg then performed the uterus transplant, the donor being a 61-year-old family friend. One of the frozen embryos

was implanted a year after the transplant, and a healthy baby boy was born prematurely at 31 weeks due to preeclampsia (Boggs, 2015).

Conclusion

MRKH syndrome is a rare developmental failure of a part or whole of the Mullerian duct. Patients with MRKH suffer from deep psychological problem and emotional trauma when they find that they have got no uterus and vagina. The person may suffer from depression, anger and isolation. At this stage, the role of a nurse is important as a counsellor and psychologist for the management of these patients.

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