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CASE SERIES OF PRIMARY AMENORRHEA DUE TO MULLERIAN AGENESIS ENCOUNTERED IN BSMMU HOSPITAL

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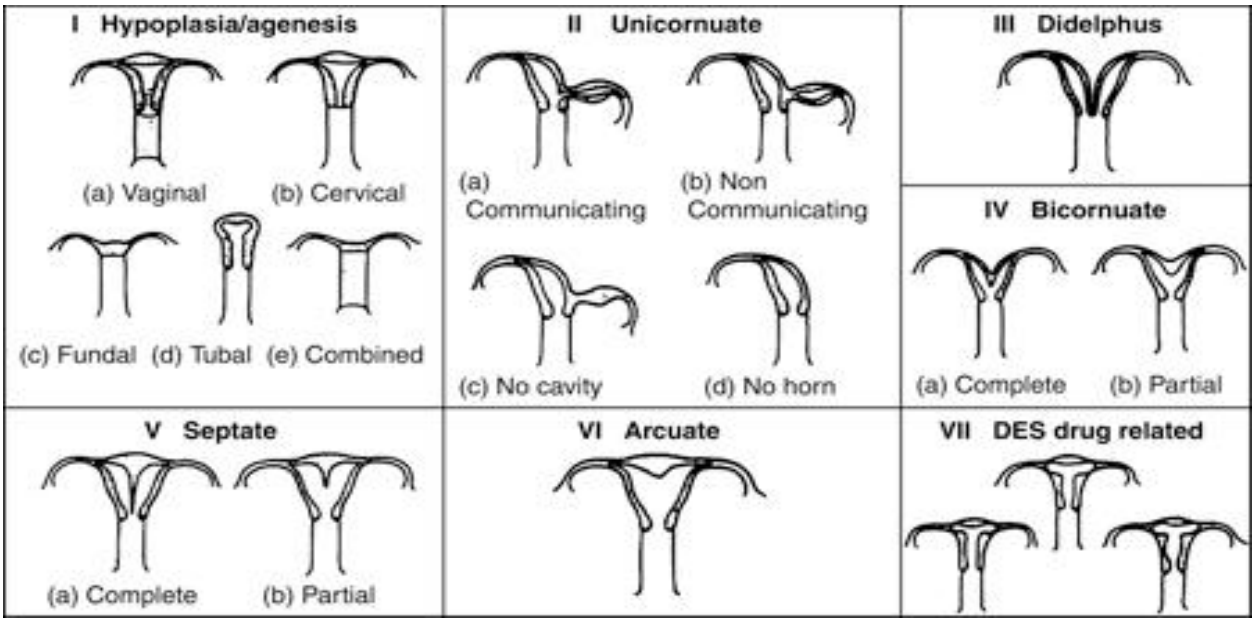
ABSTRACT

Congenital anomalies of the female reproductive tract may involve the cervix, uterus, fallopian tubes or vagina. Woman's obstetric and gynecologic health may be inimically affected, depending upon the type of congenital defect. In this case series patients presenting in the outpatient department of BSMMU hospital with Primary amenorrhea due to Mullerian agenesis were studied. Mullerian agenesis is found to occur in 1 out of every 4,000–10,000 females.[1] Patients with Mullerian agenesis commonly present with congenital absence of the vagina, uterus, or both, which also is referred to as Mullerian aplasia, Mayer–Rokitansky–Küster–Hauser syndrome or vaginal agenesis. In BSMMU these patients were offered optimium management and were followed up. Satisfactory vaginal creation was nonsurgically managed with successive vaginal dilation and surgically by neovagina creation. After one year of study period it was clear that most of these patients with few exceptions were treated optimally and they could live a healthy and better life.

KEYWORDS: Mullerian agenesis, Primary amenorrhea, BSMMU (Bangabandhu Sheikh Mujib Medical University).

INTRODUCTION

Epithelial lining of the female reproductive organs is formed by paired ducts known as Mullerian ducts. These ducts are also called as paramesonephric ducts due to their location besides the mesonephric (Wolffian) ducts. Mullerian ducts are the fundamental and primordial anlage of the female reproductive tract which differentiates into fallopian tubes, cervix, body of uterus and the upper part of vagina. Mullerian developmental anomalies occur due to embryological disruption of the normal process that affects the system.[2] According to the site & type of the defect, the AFS has classified the Mullerian developmental defects into following types and subtypes.[3]



Embryologic growth failure of the Mullerian duct results in Mullerian agenesis, with resultant agenesis or underdevelopment of the uterus, vagina or both. The vaginal canal is absent or noticeably shortened.[4] A single midline uterine remnant may be present or uterine horns (with or without an endometrial cavity) can exist. The ovaries are normal in structure and function due to their separate embryologic source.

Primary amenorrhea is defined either as the absence of menses in 14-year-old girls without the development of secondary sexual characteristics or the absence of menses in 16-year-old girls with normal development of secondary sexual characteristics.

AIMS AND OBJECTIVES

1) To account clinical presentations of the Mullerian duct anomalies.
3) To note the mode of management implied in the different diagnosed cases.

MATERIALS AND METHODS

An observational study was conducted at BSMMU Hospital Dhaka Bangladesh. In 1 year of our study more than 30 patients reported with primary amenorrhea, 9 cases were found to be suffering from Mullerian agenesis [diagnosed through previous clinical history, physical examination, radiological scans (mainly magnetic resonance imaging) and diagnostic laparoscopy] and hence were included in the study. Study focused on multiple parameters of every individual case. The study focused on the chief complaint of the cases when they presented themselves at the outpatients department. The age at presentation was also put into record for the purpose of age level stratification. The positive clinical findings and detailed investigation reports were taken into account with the aim of formulating the final diagnosis for the case. Individual cases were finally provided an optimum mode of management designed after immense case study and their outcome were also evaluated in the serial follow up at outpatient department for a period of 1 year.

RESULTS AND ANALYSIS

- Among the 9 cases which presented in the outpatient department, the most common complaint among them was absence of menarche & inability to do coitus.
- The patients who were enrolled in the study mostly belonged to the age group of
  14 - 20years.

Presentation of these patients

Typically, patients with Mullerian agenesis present with primary amenorrhea in adolescence with normal growth and development. Clinical examination reveal, patients have normal height, secondary sexual characteristics, body hair, and external genitalia. Vagina is either absent or present as a short blind-ended structure without a cervix at the vaginal apex. Patients with mullerian agenesis have a normal 46XX karyotype and a normal hormonal profile.

Evaluation of these Patients

Investigations for evaluation of patients with Mullerian agenesis include conventional trans-abdominal, translabial, or transrectal ultrasonography; three-dimensional ultrasonography; and magnetic resonance imaging. These tests can be used to evaluate the mullerian structures and are helpful in definitively characterizing anatomy. Magnetic resonance imaging has been suggested to assess the reproductive anatomy, although it rarely is needed in the initial evaluation. Although laparoscopy is not necessary to diagnose mullerian agenesis, it may be useful in the treatment of patients with functional rudimentary uterine horns.

Evaluation for other associated anomalies (congenital, renal, or others) is essential because up to 53% of patients with mullerian agenesis have concomitant congenital malformations, especially of the abdominal wall, urinary tract, and skeleton. Ultrasonography is a better screening tool to find associated abnormalities like renal agenesis or a pelvic kidney. Common skeletal abnormality associated with Müllerian agenesis is scoliosis. Varying rate of hearing impairment has also been noted in patients with Müllerian agenesis.

Management of Patients

The goal of treatment was to provide the patient with an unscarred vagina that allows sexual functioning. Excision of uterine anlage can also prevent endometriosis and resultant ovarian function impairment.[5,6] Psychosocial counseling has a pivotal role in management of patients with Mullerian agenesis. It addresses the functional and emotional effects of genital anomalies as well as correction of the anatomical defect. After the diagnosis of mullerian agenesis, the adolescent should be offered counseling to emphasize that healthy sexual relationships are possible. Future fertility options should be addressed with adolescents and their parents or guardians. Assisted reproductive techniques and surrogacy should be discussed. The success of treatment is predicted by good emotional outcome after diagnosis and vaginal creation. Good relationship between the patient and her parent(s) or guardian(s) and the ability to share feelings with family and friends has an immense impact and is the best predictor of good treatment. Contact with a support group of young women with the same diagnosis is helpful. Referral to a mental health professional for ongoing support is worthwhile.

***Nonsurgical Creation of a Neovagina*:** Timing for nonsurgical or surgical creation of a neovagina is elective; however, it is best planned when the patient is emotionally mature and expresses the desire for correction. Nonsurgical creation of the vagina is the appropriate first-line approach in most patients. Patient is convinced for successful self-dilation requiring them to

manually place successive dilators on the vaginal dimple for 30 minutes to 2 hours per day.

Surgical approach of Creating a Neovagina: Surgical intervention for creating a neovagina is for patients who are unsuccessful with dilators or for patients who prefer surgery after a thorough informed consent discussion with their health care providers and their respective parents or guardians, if appropriate. Surgical approach of creating a new vagina requires ongoing postoperative dilation or vaginal intercourse to maintain adequate vaginal length and diameter.

Patients Info

Patient 1: Miss Sabeha 17 years of age, hailing from Barisal, presented with chief complaint of non-establishment of menstruation. Her general examination revealed normal findings, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 1cm and on per rectal examination uterus could not be felt. Her ultrasonography revealed absent uterus, hormone profile was normal, Karyotyping was 46XX. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of normal saline 20cc daily in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 5cm. The typical age of presentation was mostly found in between 15 - 17 yrs. In one notable study by Mane *et al.*, the mean age of presentation was typically at 17 years.[7]

Patient 2: Miss Rupkali was 20 years old hailing from Madaripur presented with chief complaint of non-establishment of menstruation. Her general examination revealed normal findings, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 3cm and on per rectal examination uterus could not be felt. Her hormone profile was within normal limits; ultrasonography revealed absent uterus with normal ovaries, her karyotyping was normal 46XX. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of normal saline 20cc daily in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 6cm.

Patient 3: Mrs. Sanya 20 years old hailing from Mymensingh presented with complaints of non-establishment of menstruation with dyspareunia. Her general examination revealed normal findings, normal height and weight, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 3cm and on per rectal examination uterus could not be felt. Ultrasonography revealed small rudimentary mullerian bulbs, hormone analysis was normal; X-ray revealed spinal deformity .She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of vitamin C 10 ml mixed with 10 ml of distilled water daily in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 5cm and she was advised to use vaginal dilators. On discharge she was advised use of vaginal dilators and regular intercourse.

Patient 4: Mrs. Laila 19 years old hailing from Keryanigong, presented with the complaints of non-establishment of menstruation and dyspareunia. Her general examination revealed normal findings, normal height and weight, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 4cm and on per rectal examination uterus could not be felt. Her investigations profile revealed normal findings except ultrasonogram which showed hypoplastic uterus. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of normal saline 10ml in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 5cm. On discharge she was advised use of vaginal dilators and regular intercourse.

Patient 5: Miss Polin 20 years old hailing from Narsindhi, presented with complaints of non-establishment of menstruation. Her general examination revealed normal findings, normal height and weight, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 4cm and on per rectal examination uterus could not be felt. Basic investigations were normal, hormone profile was within normal range, ultrasonography revealed rudimentary mullerian ducts and absent uterus. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of normal saline 10 ml in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 5cm. On discharge she was advised use of vaginal dilators and regular intercourse.

Patient 6: Mrs. Lucky bano 19 years old hailing from Gopalgunj, presented with non-establishment of menstruation and dyspareunia. Her general examination revealed normal findings, normal height and weight, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 2cm and on per rectal examination uterus could not be felt. Her investigation profile showed normal hormone profile, absent uterus on ultrasonogram and pelvic kidney. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. A reconstructive surgery for vagina performed.

Patient 7: Miss Rubina 17 years old hailing from Barguna, presented with chief complaint of non-establishment of menstruation. Her general examination revealed normal findings, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 1cm and on per rectal examination uterus could not be felt. Hormone profile was within normal limits, ultrasonography showed hypoplastic uterus with normal ovaries, karyotyping was 46XX. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of normal saline 20cc daily in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 5cm.

Patient 8: Miss Jahan 18 years old hailing from Gornudhj, presented with non-establishment of menstruation. Her general examination revealed normal findings, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 2cm and on per rectal examination uterus could not be felt. Basic investigations were within normal range, ultrasonography showed absent uterus, karyotyping was normal, hormone profile was normal. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. We managed her by giving injection of normal saline 20cc daily in potential vaginal space for 14 days with digital dilatation. After 14 days her vaginal length got increased to 5cm.

Patient 9: Miss Jesmine Bano 14 years old hailing from Sawar, presented with the complaints of non-establishment of menstruation. Her general examination revealed normal findings, her secondary sexual characteristics were developed, her per abdominal examination revealed no abnormality. On pelvic examination we found her vulva, perineum normal, length of vagina was 2cm and on per rectal examination uterus could not be felt. Her hormone analysis was normal, ultrasonography showed rudimentary mullerian bulbs with absent uterus and pelvic kidney. She was diagnosed as a case of Primary amenorrhea with Mullerian agenesis. Her counseling was done and was asked to come back when her marriage is fixed for the surgical management.

CONCLUSIONS

- Out of approximately 30 primary amenorrhoea cases seen at the out patients department 9 cases were found to be suffering from agenesis.
- Most of them presented at an early age group of 14 - 20 yrs.
- Commonest presenting complaint was non-establishment of menstruation and or dyspareunia.
- Treatment modality was mostly individualized according to the case diagnosed.

DISCUSSION

The actual prevalence rate for Mullerian anomalies varies from 1 in 4000 to 1 in 10,000[8] and we found in our study a prevalence rate of around 10 in 8000 as we had this data from the tertiary care. The treatment of amenorrhea requires first determining its cause, so a thorough history and physical examination, accompanied by imaging studies and measurements of hormone levels are important to narrow the differential diagnosis. The most significant steps in the effective management of mullerian agenesis are correct diagnosis of the underlying condition, evaluation for other associated congenital anomalies and psychosocial counseling before any treatment or intervention to address the emotional and functional effects of genital anomalies. Laparoscopy in diagnosed cases of Mullerian agenesis is seldom required but may be appropriate in the patient presenting with pelvic pain. Nonsurgical creation of the neovagina should be the first-line approach. In patients where surgical intervention is required, referrals to centers with expertise in this area should be considered because of availability of surgeons having extensive experience in construction of the neovagina. In general, the treatment of amenorrhea must be patient-tailored according to the causative factor.

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