Research Paper Medical Science

Study of cases of primary amenorrhea due to developmental defects in adolescent girls

Dr. Krishna V. Rathod	M.S. Obs & Gynec
Dr. Harita Desai	M.S. Obs & Gynec
Dr. Nilesh A. Shah	M.S. Obs & Gynec

ABSTRACT

Menstruation is the endpoint of a series of events which begin in the cerebral cortex and terminates at the uterine tissues in the HPO axis. Primary amenorrhea can be due to disorder of the outflow tract or uterine target organs. Study was carried out to study the presentation, clinical and radiological findings, management and outcome of the patients with primary amenorrhea. A prospective study of 33 cases was carried out during one year period from July 2012 to June 2013. Of all cases, 15 were of imperforate hymen, 13 of MRKH syndrome, 3 had vaginal septum and 2 had uterus didelphys with blind vagina. 9 patients had renal anomalies. For the patient with primary amenorrhea, the physical examination should focus on pubertal development and possible genital outflow obstruction. Early surgery of the patient may reduce suffering, restore outflow tract and preserve fertility in some cases.

KEYWORDS primary amenorrhea, adolescent, MRKH syndrome, imperforate hymen

Introduction:
Attainment of menarche is important life event for a woman.

Primary Amenorrhea (PA) is clinically defined as the absence of menses by age 14 in the absence of normal growth and secondary sexual development; or the absence of menses by the age of 16 years in the setting of normal growth and secondary sexual development.

Menstruation is the endpoint of a series of events which begin in the cerebral cortex and hypothalamus and terminates at the uterine and ovarian tissues in the hypothalamic-pituitary-ovarian axis (HPO axis).

Material and Method:
This was prospective study of 33 patients presented with primary amenorrhea at department of Obstetrics and Gynecology, Civil Hospital, Ahmedabad. Cases were subjected to clinical examination, radiological studies (USG, MRI, 2-D Echo, X-rays, IVP etc.), karyotype examination. All cases were studied for following parameters: Age of the patient, Marital status, Presenting symptoms, Radiological findings, Management, Complications, follow up.

Results and observations:
In the study age of presentation of all cases was between 13 to 19 years.

Table no. I: age distribution of the patients

Age(years)	No. of patients
13	1(3%)
14	3(9%)
15	2(6%)
16	6(18%)
17	4(12%)
18	6(18%)
19	11 (33%)

27 (82%) patients were unmarried and 6 (18%) were married.

All patients presented with primary amenorrhea and of them 19 patients had associated cyclical abdominal pain of varying severity.

On examination:
All patients had well developed secondary sexual characters which ruled out sex chromosomal abnormalities.

Per abdomen, per vulval and per rectal examination were carried out and findings were noted.

16 patients had blind vagina on examination and 15 patients were diagnosed having an imperforate hymen with typical findings suggestive of hematocolpos.

On radiological studies (USG/MRI),

In 10 patients uterus was not seen and 3 patients had hypoplastic uterus. They were diagnosed having MRKH Syndrome.

Of them 9 patients underwent Mclode's vaginoplasty[1] , in which split thickness skin graft was taken and vaginoplasty was done. 3 patients vaginal pouch was deep so conservative management with vaginal dilator was done and 1 patient was not willing for surgery. Vaginoplaty was planned 6-8 months before the marriage was planned.

15 patients had complaint of primary amenorrhea associated with cyclical abdominal pain of varying duration.

On radiological studies (USG/MRI), they were having varying degree of hematometra & hematocolpos. They were diagnosed as cases of imperforated hymen. Incision and drainage was done in 14 patients.[2]

One patient was having associated Down's syndrome, diagnosed by karyotype, hysterectomy was done.

One patient had hematometra with normal adenexa on USG and had history of vaginoplaty 2 years back. Vaginal dilation f/b excision of transverse vaginal septum was done and mould was kept. Other 2 patients had hematometra and hematocolpos on USG suggestive of transverse vaginal septum. Septum resection, hematometra drainage, and vaginoplasty were done.

One patient had bilateral hypoplastic horns of the uterus sep-

arate from each other and blind vagina. Laprotomy was done & both horns were excised. Patient was explained about vaginoplasty in future.

One had uterus didelphis with 2 separate cervix with two localized collection in vaginal fornix, right ovary 6×3 cm complex cyst. In this patient laprotomy was done to remove both uterus and vaginoplasty was done.

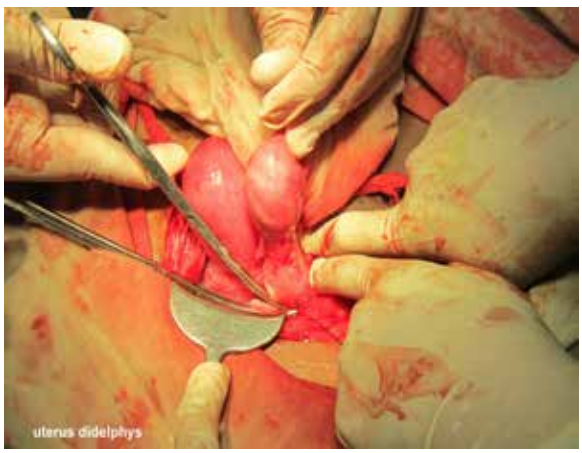

Figure 1: uterus didelphis [per operative]

Figure 2: uterus didelphis with two separate cervices [after removal]

Table no. II: findings of USG KUB

USG KUB	No of patients
Normal	24(72%)
Absent one kidney	4 (12%)
One ectopic kidney and other normal	3 (9%)
One kidney absent and other ectopic with Duplication of ureteric system	1 (3%)
Bilateral ectopic kidney	1(3%)

Findings of USG KUB was confirmed by IVP or CT- IVP.

In patients with primary amenorrhea associated with müllerian duct anomaly, urogenital dysplasia was associated with. Total 9 (27%) patients had associated renal anomalies.

One patient had associated scoliotic deformity of the spine.

On follow up:
Patients underwent vaginoplasty had adequate vaginal space.

Out of 3 conservatively managed patient one patient had increased space with regular vaginal dilatation 2 were yet to come for follow up.

Of 3 patients with transverse vaginal septum, both had resumed menses regularly post resection of septum and vaginoplasty.

A patient with uterus didelphis with blind vagina was well on follow up but the space created by vaginoplasty was obliterated.

Table no. III: diagnosis

Diagnosis	No. of patients
Imperforate hymen	15(45%)
MRKH syndrome	13(39%)
Transeverse vaginal septum	3(9%)
Uterus didelphys with blind vagina	2(6%)

Discussion:
Amenorrhea is only a symptom and not a disease entity. Amenorrhea can be physiological or pathological.

Primary amenorrhea affects approximately 5% of amenorrheic women and it's commonly diagnosed in girls with normal pubertal development at the age of 16 years or as early as 14 years in girls with no pubertal development.

In this study, all patients were between 13-19 years of age, most common 19 years (33%).

27(82%) were unmarried adolescent girls.

Magnetic resonance imaging (MRI) is considered the gold standard for determining congenital anomalies.[3]

15(45%) were having imperforated hymen. Imperforate hymen occurs in 1 in 1,000 women. It may be diagnosed in childhood, but may also be missed and may present in adolescence with cyclic abdominal pain and primary amenorrhea. The typical physical finding is a bulging, bluish hymen, behind which is a blood-filled mass in the distended vagina (hematocolpos).

18(54%) patients were diagnosed having müllerian duct anomalies manifesting as primary amenorrhea. 13 patients had MRKH syndrome. Although MRKH syndrome is a rare condition with a reported incidence of 1:4000[4-6], it represents the second most common cause of primary amenorrhea only preceded by gonadal dysgenesis.[7]

3 patients had transverse vaginal septum. A complete transverse septum occurs in approximately 1 in 80,000 women and is due to incomplete fusion of the müllerian duct portion of the vagina and the urogential sinus component.[8]

And 2 patients had uterus didelphis with blind vagina.

Additionally, other malformations of the urological tract or rectum may be associated. In this study, of 18 cases of müllerian anomalies, 9 (50%) cases had associated renal malformations.[6]

Conclusion:
Amenorrhea (primary or secondary) has got multifactorial etiology.

For the patient with primary amenorrhea, the physical examination should focus on pubertal development and possible genital outflow obstruction which was most common finding in this study.

Müllerian agenesis, a congenital malformation of the genital tract is the second most common cause of primary amenorrhea. It is an uncommon but not rare anomaly.

Uterine abnormalities are frequently unrecognized at birth and are unreported.

Early surgery offered to the patient may reduce patients suf-

fering, help restore a patent outflow tract and may preserve fertility in some cases.

Research on müllerian anomalies is limited and future studies are needed with larger samples and improvements in research design.

Studies are also required regarding the various surgical managements done and improvement of quality of life with it.

REFERENCES

1. John A. Rock, Howard W. Jones. TeLinde' s Operative Gynaecology . 10th ed. New Delhi : Lipincott Williams and Wilkins; 2009 . p. 550-551 2. John A. Rock, Howard W. Jones. TeLinde' s Operative Gynaecology . 10th ed. New Delhi : Lipincott Williams and Wilkins; 2009 . p. 509-511 3. Troiano RN. Magnetic resonance imaging of mullerian duct anomalies of the uterus. Top Magn Reson Imaging 2003; 14:269–79. 4. Griffin JE, Edwards C, Madden JD, Harrod MJ, Wilson JD: Congenital absence of the vagina. The Mayer-Rokitansky-Kuster-Hauser syndrome. Ann Intern Med 1976, 85:224-236. PubMed Abstract 5. Folch M, Pigem I, Konje JC: Mullerian agenesis: etiology, diagnosis, and management. Obstet Gynecol Surv 2000, 55:644-649. PubMed Abstract | Publisher Full Text 6. Varner RE, Younger JB, Blackwell RE: Mullerian dysgenesis. J Reprod Med 1985, 30:443-450. PubMed Abstract 7. Strubbe EH, Willemsen WN, Lemmens JA, Thijn CJ, Rolland R: Mayer-Rokitansky-Kuster-Hauser syndrome: distinction between two forms based on excretory urographic, sonographic, and laparoscopic findings. Am J Roentgenol 1993, 160:331-334. 8. VARNER, R.E., J.B. YOUNGER & R.E. BLACKWELL.1985. 9. Mullerian dysgenesis. J. Reprod. Med. 30: 443– 450