

ORIGINAL ARTICLE

Study and Clinical Evaluation of 25 cases of Primary Amenorrhea

Dr Tushar M Shah*, Dr Foram Lalcheta**, Dr Rini Buddhdev***

*Associate Professor, ** 2nd year OBGY resident, ***1st year OBGY resident in Obstetrics & Gynecology Department B.J Medical College, Civil Hospital, Ahmedabad.

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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

AIMS : The objective of this study is to note the various causes, complete clinical picture and the management in 25 such cases of Primary Amenorrhea. **SUBJECTS AND METHODS :** This is A Prospective study done in 25 cases. They were investigated, managed and patients were called up for follow up for their response to treatment.

RESULTS AND CONCLUSION : Out of 25 cases studied, maximum cases presented at 14-16 years of age, with chief complaint of primary amenorrhea, out of which 8% were married, 13 cases were of MRKH syndrome which was the most common cause of Primary Amenorrhea.

INTRODUCTION

Primary Amenorrhea is absence of menstruation and secondary sexual characters by age of 14 years or absence of menstruation regardless of secondary sexual characters by age of 16 years. Menstruation, also known as a 'period' or 'monthly', is a regular discharge of blood and tissue from the inner lining of the uterus through vagina⁽¹⁾. For a normal menstruation to occur the genetic make up, neuroendocrinological, embryological development is must. Any abnormality in above leads to amenorrhea. Menstruation marks the transition from a girl to a woman. It is the physical manifestation noticed of all pubertal changes and last to occur⁽²⁾. Incidence of Primary Amenorrhea is <1%. Development of female genital organs takes place from MULLERIAN DUCT (Paramesonephric Duct). Sexual development starts after 8 weeks. In absence of Y chromosome, functional testes and lack of AMH, Mullerian Duct develops and form uterus, fallopian tubes and upper vagina and at the same time in absence of testosterone, Formed from coelomic Epithelium. Hymen is formed by invagination of posterior wall of urogenital sinus and usually ruptures-spontaneously in perinatal period. Menarche is the end of cascade of events occurring in puberty.⁽⁴⁾

Causes of primary amenorrhea can be classified according to WHO as:

1. HYPOGONADOTROPIC
HYPOGONADISM (Group I)

[decreased estrogen, normal or low FSH, and no lesion in hypothalamic-pituitary region]

- Physiologic Delay.
 - Kallman syndrome
 - CNS tumors
2. HYPERGONADOTROPIC
HYPOGONADISM (Group II)
[decreased estrogen, but raised FSH]
 3. DEVELOPMENT DEFECT OF
GENITALTRACT (Group III)

OBJECTIVES

The objective of this study is to evaluate the age of presentation, presenting symptom, clinical findings of general and local examination, causes, outcome and management of 25 cases of primary Amenorrhea presented in Tertiary care hospital for a duration of 1 year from November 2016 to November 2017.

MATERIALS AND METHODS

This is a prospective study. A methodical, systemic approach was developed to achieve best diagnosis. Evaluation of primary Amenorrhea is designed to separate there productive system into its distinct structural components such as Genital outflow track, Uterus, Ovaries, Pituitary, Hypothalamus and to test functional integrity of each beginning at lowest level and

Correspondence Address : Dr Tushar M Shah
A-902, Status Tower, Opp. Doordarshan, Nr. SAL Hospital, Drive-in-Road, Thaltaj,
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progressing systemically to higher levels until the cause is determined. 25 Cases of primary Amenorrhea were studied in detail for following parameters-

- Age of presentation
- Presenting complaint
- Marital Status
- Past history of any medical illness
- Family History
- General Built (Height, weight, BMI and arm span)
- Presence of secondary sexual characters (according to Tanner's staging)
- Local examination (P/A, P/vulva, P/S, P/V)
- Hormonal profile (FSH, LH, Estrogen, AMH, TSH)
- Radiological investigations (USG, MRI)
- Special investigations (CT-IVP)
- Karyotyping

RESULTS

- Presenting age was in range from 11 to 25 years.

Table 1: Age distribution of Patients

Age	Percentage
11- 13 Years	6 (24%)
14- 16 Years	9 (36%)
17- 19 Years	4 (16%)
19- 21 Years	3 (12%)
> than 21 Years	3 (12%)

- Presenting Symptoms:

Table 2 : Presenting Symptoms

Symptom	Percentage
Primary Amenorrhea	13 (52%)
Cyclical abdominal pain	5 (20%)
Ambiguous Genitalia	3 (12%)
Urinary Retention	4 (16%)

- Out of 25 cases, 3 of the patients were married and 22 were unmarried.
- Causes of Primary Amenorrhea:

Table 3 : Causes

Causes	Percentage
MRKH Syndrome	13(52%)
Imperforate Hymen	10(40%)
Turner syndrome	1 (4%)
Uterus didelphus blind vagina blind vagina	1(4%)

- MRKH syndrome is the most common cause of Primary Amenorrhea in our Study i.e 52% . These patients had normal secondary sexual characters that was symmetric with age. They had normal Development of breasts which indicates normal ovarian function(6). All had normal karyotyping (46XX). MRI is gold standard to diagnose any congenital anomalies⁽³⁾. These patients were explained about their pathology and counseled regarding their menstrual and reproductive function. Primary goal of management in MRKH syndrome is creation of functional vagina. They were explained about vaginoplasty and were called for follow-up before 6 months of their marriage. MRKH syndrome is associated with renal, cardiac and skeletal malformations⁽⁷⁾. Thus all the cases of MRKH syndrome underwent USG KUB, 2D echo and X-ray spine. 16% of them were associated with abnormal USG KUB.

USG KUB	Percentage
Normal	7 (53%)
Horse shoe kidney	1 (0.7 %)
Fused Crossed Ectopic Kidney	1 (0.7%)
Absent Kidney	2 (0.15%)

- There were 7 cases of imperforate hymen, 5 presented with complaint of cyclical lower abdominal pain and 2 case with urinary retention. All of them had normal symmetrical secondary sexual characters and normal breast development indicating normal ovarian function. All the cases were managed surgically and 1 case with recurrent Hematometra and Hematocolpos ,total hysterectomy was done, Ovaries were preserved
- Of 3 patients with transverse vaginal septum, both had resumed menses regularly post resection of septum and vaginoplasty.

- Patient with Turner syndrome presented with c/o primary amenorrhea with short stature, shield chest (widely spaced nipples) with underdeveloped breast and absent pubic and axillary hairs (Tanner stage I). MRI pelvis suggested of 6x4 mm of hypoplastic uterus with absent ovaries. Karyotyping s/o mosaicism 45,x,(92)/46,x,i(x)(910)(8) , 92% of cell with XO and 8% of cells with XX. She was given withdrawal with Oral contraceptive pills (MALA-N) and since then patient is menstruating normally. 2D ECHO and USG KUB was normal .
- One patient had uterus didelphis with 2 separate cervix with two localized collection in vaginal fornix, right ovary 6x3 cm complex cyst, and blind vagina In this patient laprotomy was done to remove both uterus and vaginoplasty was done



CONCLUSION

Amenorrhea has got multifactorial etiology. For patients with Amenorrhea physical examination should focus on pubertal development and possible genital outflow obstruction. Mullerianagenesis (MRKH syndrome) was the most common cause of primary amenorrhea. Early surgery offered to patients may reduce patients suffering, help restore patients outflow tract and may preserve fertility in some cases. Research on mullerian anomalies is limited and further studies are needed. Out of 25 cases of Primary Amenorrhea, 52% had MRKH syndrome which was the most common cause of primary amenorrhea, 28% imperforate Hymen, 12% had transverse vaginal septum.

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