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RESEARCH ARTICLE

CASE SERIES OF PRIMARY AMENORRHOEA

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ABSTRACT

Background: Amenorrhoea is absence of menstrual bleeding . Primary Amenorrhoea 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. For a normal menstruation to occur the normal genetic makeup, neuro-endocrinological, embryological development is must. Any abnormality in above leads to amenorrhea. Incidence of Primary Amenorrhea is less than 1%. **Objective:** The objective of this study was to note the various causes, complete clinical picture and the management in 25 such cases of Primary Amenorrhoea. **Materials and Methods:** This is a Prospective study done in 25 cases .They were thoroughly investigated (i/e clinical examination,radiological examination and hormonal study) to get the exact cause of Primary Amenorrhoea. **Results:** Out of 25 cases studied, maximum cases presented at 14-16 years of age, with chief complaint of primary amenorrhoea, out of which 8% of them were married, 44% cases were of MRKH (Mayer-Rokitansky-Kuster-Hauser syndrome) which was the most common cause of Primary Amenorrhoea in our study. **Conclusion:** Most common cause of primary amenorrhoea in our study was MRKH Mayer-Rokitansky-Kuster-Hauser syndrome with normal secondary sexual characterstics.

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INTRODUCTION

Primary Amenorrhoea 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 linning of the uterus through vagina. According to World Health Organization estimates, amenorrhoea stands as sixth largest major cause of female infertility and affects 25% of all women in the child bearing age (Dutta *et al*., 2013). Amenorrhoea is a symptom that reflects some underlying disease anywhere in the hypothalamic-pituitary-ovarian-uterine axis. There are different causes of primary amenorrhoea. It includes anomalies of mullerian development, gonadal dysgenesis, constitutional delayed puberty, tuberculosis, CNS tumors, idiopathic etc (Speroff *et al*., 1999). For a normal menstruation to occur the genetic makeup, neuroendocrinological, embryological development is must Any abnormality in above leads to amenorrhea.

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Mentruation marks the transition from a girl to a woman. It is the physical manifestation noticed of all pubertal changes and last to occur. About 2-5% of adolescent girls present with primary amenorrhea (Wachtell, 1979). A case of primary amenorrhoea should be evaluated thoroughly as it has impact on both physical and psychological well being of the patient. Early diagnosis and timely intervention is necessary to prevent long term health and social consequences. As soon as the etiology is established in a particular case, the patient can be counselled regarding the prognosis and future fertility options.
There are studies from various regions of the world on etiology of primary amenorrhoea. The two main causes are mullerian anomalies and gonadal dysgenesis with different frequencies in different parts, some have shown anatomic abnormalities as the most common cause while others have reported gonadal failure as the commonest one (Schorge, ?; Cunningham *et al*., 2008; Tanmahasamut *et al*., 2012). 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, Wolffian duct regresses. The ovaries are developed from genital ridge. This ridge is formed at 5th week by multiplication of coelomic Epithelium.

Hymen is formed by invagination of posterial wall of urogenital sinus and usually ruptures spontaneously in perinatal period. Transverse vaginal septum results when vaginal plate formed by fused sinovaginal bulb fails to breakdown. Menarche is the end of cascade of events occurring in puberty.

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

Hypogonagotropic Hypogonadism (Group I): Decreased estrogen, normal or low FSH, and no lesion in hypothalamic-pituitary region. Fig-1

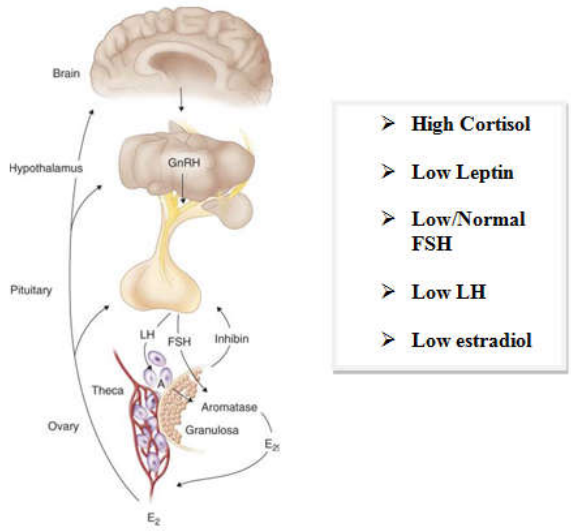


Fig- 1

- Physiologic delay.
- Kallman syndrome (Hypothalamic Failure).
- CNS tumors.
- Hypothalamic /pituitary dysfunction.

Hypergonadotrophic hypogonadism (Group II): Decreased estrogen but increased FSH. Fig-2

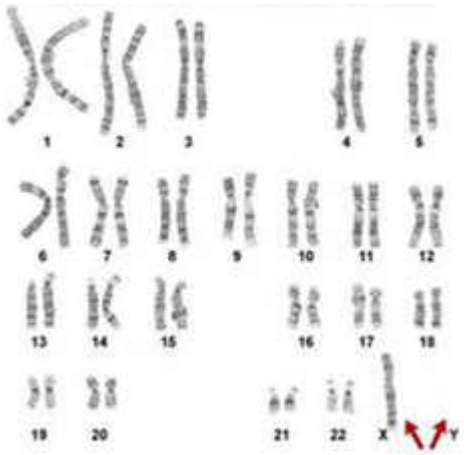


Fig. 2. Karyotype of a female with Turner's Syndrome

- Gonadal Dysgenesis.
- Turner Syndrome.

Development defect of genital tract (group iii) fig-3

Fig. 3. Imperforate Hymen

- Transverse vaginal septum.
- Imperforated Hymen.

We have conducted this study to evaluate the etiology of primary amenorrhoea in women presenting at tertiary care centre in India.

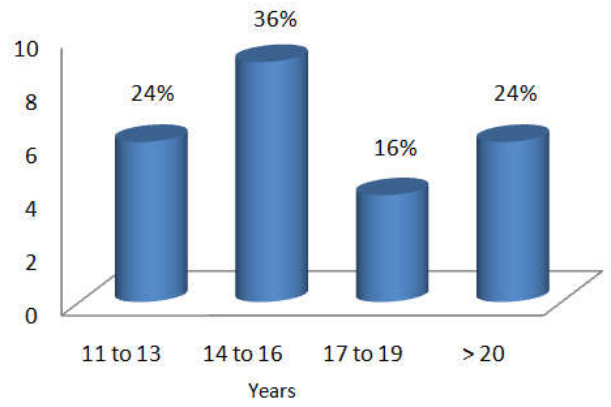
MATERIALS AND METHODS

This is a prospective study. A methodical, systemic approach was developed to achieve best diagnosis. Evaluation of primary Amenorrhea is designed to separate the reproductive 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 progressing systemically to higher levels until the cause is determined. 25 Case of primary Amenorrhea were studied in detail for following parameters-

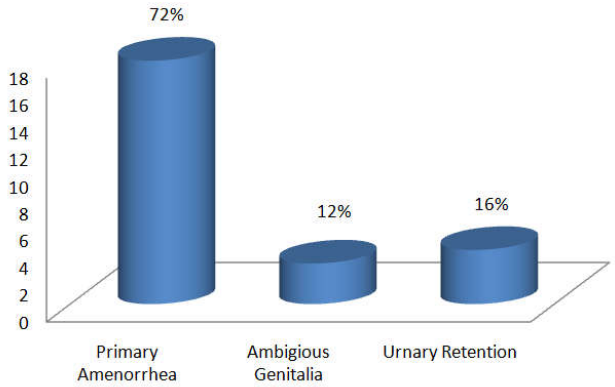
- Age of presentation.
- Presenting Complaints (a/w cyclical pain or galactorrhoea).
- Eating habits and exercise pattern.
- Marital Status.
- History of vasomotor instability.
- Past history of any medical illness.
- Family History.
- History of tuberculosis.
- General Built (Height, weight, BMI and arm span)
- Presence of secondary sexual characters (according to tanners staging)
- Local examination (PV / PR and examination of external genitals).
- Hormonal profile (FSH, LH, Estrogen, AMH, TSH).
- Radiological investigations (USG, MRI).
- Karyotyping.

RESULTS

- Presenting age for the cases were in range from 11 years to 25 years. (Graph 1)



- Presenting Symptoms: (Graph 2)



- Out of 25 cases, 5 had complaint of cyclical pain associated with primary Amenorrhea.
- Out of 25 cases, 3 of the patients were married and 22 were unmarried.
- Causes of Primary Amenorrhea: (Table 1)

CAUSES	NUMBER (%)
MRKH Syndrome	11 (44%)
Cervical Agenesis with vaginal septum	3 (12%)
Turner syndrome	2 (8%)
Disorder Sex development	2 (8%)
Imperforate Hymen	5 (20%)
Pure Gonadal Agenesis	2 (8%)

DISCUSSION

Adolescent girls with primary amenorrhoea are brought to the Gynecologists by their mothers with major concern regarding their reproductive life. The defects have been compartmentalised and may lie within the uterus, ovaries, pituitary or hypothalamus. Genetic and chromosomal anomalies also contribute to a major portion of primary amenorrhoea especially in cases of gonadal failure. The workup of primary amenorrhoea should be very meticulous including history, physical examination, hormone evaluation, pelvic imaging (either ultrasound or MRI). The importance of cytogenetic studies and karyotype cannot be overemphasized in establishing the diagnosis. It should be done in all cases of hypergonadotropic hypogonadism and patients with androgenic features.

Previous studies have been reported from all parts of the world indicating the frequency of various etiologies, cytogenetic abnormalities in cases of primary amenorrhoea. Gonadal dysfunction has been considered as the commonest factor for primary amenorrhoea worldwide followed by pituitary/hypothalamic disorder and outflow tract anomalies.[3] While in our study the most common cause of Primary Amennoerhoea was MRKH Syndrome followed by outflow tract anomalies. Literature shows greater prevalence of gonadal dysfunction leading to primary amenorrhoea in western countries while that of outflow tract anomalies in Asian-African countries. Most of the studies from United States have mentioned gonadal dysgenesis as the most common cause of amenorrhoea while a large study from Thailand of 295 cases has shown Mullerian anomaly as the commonest cause in Thai population (Schorge, ?; Cunningham *et al*., 2008; Reindollar, 1989; Tanmahasamut *et al*., 2012;) which was in correlation to our present study. In a study done in AIIMS New Delhi, 54.4% of patients of Primary Amenoerrhoea presented with Mullerian Agenesis(Alka Kriplani2017) which was in correlation to our present study i.e 44%.The proposed reason for this difference might be the environmental and racial or genetic influence. All of them had normal symmetrical secondary sexual characters (Tanner stage V) and normal breast development indicating normal ovarian function. Patient with turner syndrome presented with complaint of 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.

Conclusion

Amenorrhea has got multifactorial etiology. For patients with Amenorrhea physical examination should focus on pubertal development and possible genital outflow obstruction. MRKH syndrome was the most common cause of primary amennorhoea. Research on MRKH is limited and further studies are needed. Out of 25 cases of Primary Amenorrhoea, 44% had MRKH syndrome, 20% had imperforate Hymen, 12% had cervical agenesis with transverse vaginal septum, 8% had turner syndrome, 8% had disorder sex development and 8% had pure gonadal agenesis.

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