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Etiological factors of primary amenorrhoea: A prospective study

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Abstract
Amenorrhoea is absence of menses in women of reproductive age. Primary amenorrhea is defined either as absence of menarche by 14 years of age in the absence of secondary sexual characteristics or absence of menses by 16 years in the presence of normal growth and secondary sexual characteristics. Secondary amenorrhea is characterized as the cessation of previously regular menses for three months or previously irregular menses for six months. According to World Health Organization estimates, amenorrhea stands as sixth largest major cause of female infertility and affects 2–5% of all women in the child bearing age.[1,2] In the present study, 3 patients presented before 15yrs of age, 12 patients presented in the age group of 16 -20 years, 5 patients of 21-25yrs age group, 1 patients of 26-30 years age group, 1 patients of 30-40 years age group and past 40yrs was seen in 2 patients. The complaint of not having attained menarche is seen in only 11 cases, dyspareunia in 4 cases, cyclical pain abdomen in 3 cases, dysuria were seen in 2 cases, infertility and short stature was seen in 2 cases each and malnutrition was in one case. Our study found MRKH syndrome in 10 cases and HGH in 8 cases. Next most important cause is cryptomenorrhoea in 3 cases. AIS was seen in 2 cases followed by PCOS and imperforate hymen each in one case. Treatment for primary amenorrhea may start with watchful waiting, depending on the person's age and the result of the ovary function test. If there is a family history of late menstruation, periods may start in time. If there are genetic or physical problems that involve the reproductive organs, surgery may be necessary. This will not guarantee, however, that normal menstrual cycles will occur.

Keywords: Primary amenorrhea, aetiology, symptoms

Introduction
Amenorrhoea is absence of menses in women of reproductive age. Primary amenorrhea is defined either as absence of menarche by 14 years of age in the absence of secondary sexual characteristics or absence of menses by 16 years in the presence of normal growth and secondary sexual characteristics. Secondary amenorrhea is characterized as the cessation of previously regular menses for three months or previously irregular menses for six months. According to World Health Organization estimates, amenorrhea stands as sixth largest major cause of female infertility and affects 2-5% of all women in the child bearing age [1, 2].

About 2–5% of adolescent girls present with primary amenorrhea. The incidence is increasing because of increased reporting, better utilisation of healthcare, declining trend in child marriage and increased awareness due to social media. 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. 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 [3, 4].

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. We conducted this retrospective study to evaluate the etiology of primary amenorrhoea in women presenting at tertiary care centre in India [5, 6]

Objective
Etiological aspects of Primary Amenorrhoea

Methodology
Source of data: Sambhram Medical College and Research Institute Sample size: 25 Cases
Period of study: July 2018 to December 2018

Inclusion criteria
1. Female of age >14yrs with primary amenorrhoea with absence of secondary sexual characteristics
2. Female with age >16yrs with primary amenorrhoea with normal growth and development and appearance of secondary sexual characteristics.

Exclusion criteria
Secondary amenorrhoea

1. Female <14yrs age

Results

Table 1: Age wise distribution of Primary Amenorrhoea

Age Group	No. of Cases
14-15 Years	03
16-20 Years	12
21-25 Years	05
26-30 Years	01
30-40 Years	01
>40 Years	02
Total	25

In the present study, 3 patients presented before 15yrs of age, 12 patients presented in the age group of 16 -20 years, 5 patients of 21-25yrs age group, 1 patients of 26-30 years age group, 1 patients of 30-40 years age group and past 40yrs was seen in 2 patients.

Table 2: Symptoms at Presentation

Symptoms	No. of Cases
Not Attined Menarche	11
Dyspareunia	04
Cyclic Abdominal Pain	03
Dysuria	02
Infertility	02
Short Stature	02
Malnutrition	01
Total	25

The complaint of not having attained menarche is seen in only 11 cases, dyspareunia in 4 cases, cyclical pain abdomen in 3 cases, dysuria were seen in 2 cases, infertility and short stature was seen in 2 cases each and malnutrition was in one case.

Table 3: Etiology of primary amenorrhoea

Etiology	No. of Cases
Mrkh	10
Hupergonadotropic Hypogonadism	08
Cryptomenorrhoea	03
Ais	02
Polycystic Ovarian Syndrome	01
Imperforate Hymen	01
Total	25

Our study found MRKH syndrome in 10 cases and HGH in 8 cases. Next most important cause is cryptomenorrhoea in 3 cases. AIS was seen in 2 cases followed by PCOS and imperforate hymen each in one case.

Discussion
In the present study, 3 patients presented before 15yrs of age, 12 patients presented in the age group of 16 -20 years, 5 patients of 21-25yrs age group, 1 patients of 26-30 years age group, 1 patients of 30-40 years age group and past 40yrs was seen in 2 patients. The complaint of not having attained menarche is seen in only 11 cases, dyspareunia in 4 cases, cyclical pain abdomen in 3 cases, dysuria were seen in 2 cases, infertility and short stature was seen in 2 cases each and malnutrition was in one case. Our study found MRKH syndrome in 10 cases and HGH in 8 cases. Next most important cause is cryptomenorrhoea in 3 cases. AIS was seen in 2 cases followed by PCOS and imperforate hymen each in one case.
According to a retrospective study performed using 102 complete medical records of women with primary amenorrhea who attended the Gynaecologic Endocrinology Clinic, Department of Obstetrics and Gynaecology, AIIMS, New Delhi from September 2012 to September 2015. Cases were analysed according to clinical profile, development of secondary sexual characteristics, physical examination, pelvic and rectal examination, X-ray of chest and lumbo-sacral spine, hormone profile, pelvic USG, MRI, and cytogenetic study including karyotype. The three most common causes of primary amenorrhea were Mullerian anomalies (47%), gonadal dysgenesis (20.5%), and hypogonadotropic hypogonadism (14.7%) in the present study. There were 3 cases of Turner syndrome (45, XO), 5 cases of Swyer's syndrome (46, XY) and 2 cases of Androgen insensitivity syndrome (46, XY). One case had pituitary macroadenoma and eight cases (7.8%) were of genital tuberculosis [7].
According to a study by Asoke KP, One hundred and seventy-four cases of primary amenorrhea were referred from the obstetrics and gynecology department to our cytogenetic laboratory for chromosomal analysis. G-banded chromosomes were karyotyped, and chromosomal analysis of all patients was done. Out of 174 patients, we observed 23 (13.22%) participants with abnormal karyotype. In 23 cases of chromosomal abnormalities, 10 cases were sex reversal female (46, XY) and Turner karyotype (45, X) in 6 females. Other numerical and structural abnormalities were also seen such as 47, XXX; 45, X/47, XXX; 45, X/46, X, dic (X); 46, XX, inv (9); 45, X/46, X, I (Xq); 46, X, mar (X); and 45, X/46, XY in the primary amenorrhea cases. This study definitely attests the importance of chromosomal analysis in the etiologic diagnosis of primary amenorrhea patients. Karyotyping will help to counsel and manage the cases of primary amenorrhea in a better way. This study reveals the frequencies and different types of chromosomal abnormalities found in primary amenorrhea individuals and that might help to make the national database on primary amenorrhea in relation to chromosomal aberrations [8].
A retrospective study was performed using 15 complete medical records of women with primary amenorrhea who attended Gynecology OPD, Department of Obstetrics & Gynecology, from June 2016 to June 2018. Cases were analyzed according to clinical profile, development of secondary sexual characters, physical examination, pelvic and rectal examination, hormone profile, pelvic USG, MRI and cytogenetic study including karyotype. The most common cause of primary amenorrhea was Mullerian anomalies (60%) in present study. There were 2 cases

of early gonadal failure, 1 case of pure gonadal dysgenesis, 1 case of Turner mosaic, 1 case of Androgen insensitivity syndrome and 1 case of congenital adrenal hyperplasia. In this study, Mullerian anomaly was the most prevalent etiological factor leading to amenorrhea followed by premature ovarian failure. However, this was a small scale study and larger study group study must be needed. Racial, genetic and environmental factors could play role in the cause of primary amenorrhea [9].

Conclusion

Treatment for primary amenorrhea may start with watchful waiting, depending on the person's age and the result of the ovary function test. If there is a family history of late menstruation, periods may start in time. If there are genetic or physical problems that involve the reproductive organs, surgery may be necessary. This will not guarantee, however, that normal menstrual cycles will occur.

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