

Contemporary issues in primary amenorrhea: An experience from a Tertiary Care Center

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ABSTRACT

Introduction: Amenorrhea is classified as primary if menstrual bleeding has never occurred in the absence of hormonal treatment. The clinical significance of a lack of regular menstrual cycles extends beyond reproductive concerns. Episodes of amenorrhea as short as 90 day may have implications for bone and cardiovascular health. **Aims and Objective:** To evaluate all patients presenting with primary amenorrhea in the Endocrinology OPD of Gauhati Medical College and Hospital. **Materials and Methods:** A total of 14 patients presenting to the Endocrinology OPD from March 2010 to May 2012 with a history of primary amenorrhea were included in the study. All patients were subjected to a detailed history, a thorough clinical examination, and relevant biochemical, hormonal, and radiological investigations. **Result:** In our study, the average age of presentation was 17.23 ± 4.2 years. Out of the 14 patients presenting with primary amenorrhea, 5 patients (35.71%) were found to have Turner's syndrome, 2 (14.28%) had XX (pure) gonadal dysgenesis, 2 (14.28%) patients had XY gonadal dysgenesis (Swyer syndrome), 2 (14.28%) patients had Müllerian agenesis, 2 (14.28%) patients had hypothalamic amenorrhea, and 1 (7.14%) patient was found to have multiple pituitary hormone deficiency. **Conclusion:** In concordance with other studies, Turner's syndrome, Müllerian agenesis, and gonadal dysgenesis are the commonest causes of primary amenorrhea in our study. However, in contrast to certain Western reports, primary amenorrhea rather than short stature remains the commonest cause for seeking medical evaluation in patients with Turner's syndrome.

INTRODUCTION

Amenorrhea refers to the absence of menstrual periods. Amenorrhea is classified as primary if menstrual bleeding has never occurred in the absence of hormonal treatment. The absence of menses by age 16 has been used traditionally to define primary amenorrhea. However, other factors such as growth, secondary sexual characteristics, and the secular trend to an earlier age of menarche, particularly in African-American girls also influence the age, at which primary amenorrhea should be investigated. Thus, an evaluation for amenorrhea should be initiated by age 15 or 16 in the presence of normal growth and secondary sexual characteristics; age

13 in the absence of secondary sexual characteristics or if height is less than the third percentile; age 12 or 13 in the presence of breast development and cyclic pelvic pain; or within 2 years of breast development if menarche has not occurred. It is a rare disorder occurring in $< 1\%$ of the female population.^[1] The clinical significance of a lack of regular menstrual cycles extends beyond reproductive concerns. Episodes of amenorrhea as short as 90 day may have implications for bone and cardiovascular health. Prolonged amenorrhea, depending upon its underlying cause, can be a harbinger of substantial cardiovascular risk.^[2]

AIMS AND OBJECTIVE

To evaluate all patients presenting with primary amenorrhea in the Endocrinology OPD of Gauhati Medical College and Hospital.

MATERIALS AND METHODS

A total of 14 patients presenting to the Endocrinology OPD from March 2010 to May 2012 with a history of

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primary amenorrhea were included in the study. We took those patients who presented with absence of onset of menses by 16 years in the presence of normal growth and secondary sexual characteristics; or by 13 years of age in the absence of secondary sexual characteristics. All patients were subjected to a detailed history, a thorough clinical examination, and relevant biochemical, hormonal, and radiological investigations.

RESULT

In our study, the average age of presentation was 17.23 ± 4.2 years. Out of the 14 patients presenting with primary amenorrhea, 5 patients (35.71%) were found to have Turner's syndrome, 2 (14.28%) had XX (pure) gonadal dysgenesis, 2 (14.28%) patients had XY gonadal dysgenesis (Swyer syndrome), 2 (14.28%) patients had Müllerian agenesis, 2 (14.28%) patients had hypothalamic amenorrhea, and 1 (7.14%) patient was found to have multiple pituitary hormone deficiency. Of the 5 patients who were diagnosed to have Turner's syndrome, their karyotyping revealed isochromosome in 1 patient, ring chromosome in 1 patient, 1 patient was having a 46XX/XY mosaic pattern, and 2 patients were having classical 45, XO karyotype. The patients with classic 45, XO karyotype were also having classical Turner's phenotype including short stature while in case of other 3 patients, none of them were having classical Turner's phenotype and the 1 with ring chromosome was having normal breast development (Tanner's stage 4). One patient with XY gonadal dysgenesis presented with primary amenorrhea along with ambiguous genitalia in the form of a microphallus, scrotal rugosity, and Müllerian structures and on laparoscopic biopsy was found to harbor a gonadoblastoma. Both the patients with pure gonadal dysgenesis were also found to have ill-formed labia minora along with primary amenorrhea and absent secondary sexual characters. Two patients in our study were diagnosed to have Müllerian agenesis, of which 1 patient fits into the phenotype of MURCS syndrome with Müllerian agenesis, unilateral renal agenesis, and anomalies of the cervico-thoracic somites in the form of Torticollis. The patient with combined pituitary hormone deficiency presented with short stature along with primary amenorrhea and was found to have growth hormone deficiency along with gonadotropin deficiencies.

DISCUSSION

Research in reproduction continues to provide critical insights into our knowledge of the mechanisms responsible for amenorrhea in women. Among various causes of primary amenorrhea, the breakdown of causes likely remains unchanged, with the 4 most common causes of primary amenorrhea being ovarian failure (48.5%), congenital absence of the uterus and vagina (16.2%), GnRH deficiency (8.3%), and constitutional delay of puberty (6.0%).^[3] In a study done in Thailand in 2009, the 3 most common causes of primary amenorrhea found were Müllerian agenesis (39.7%), gonadal dysgenesis (35.3%), and hypogonadotropic hypogonadism (9.2%). Amongst 88 cases of gonadal dysgenesis, 59 cases (67.0%) incurred abnormal karyotype including 45X (n = 21), mosaic (n = 31), and others (n = 7).^[4] In a study done in Mexico City in 1999, out of 19 patients presenting with Müllerian agenesis, 3 were found to have associated MURCS syndrome.^[5]

CONCLUSION

In concordance with other studies, Turner's syndrome, Müllerian agenesis, and gonadal dysgenesis are the commonest causes of primary amenorrhea in our study. The relatively later age of presentation as compared to Western world may reflect lack of awareness about primary amenorrhea prevailing in this region. In contrast to certain Western reports, primary amenorrhea rather than short stature remains the commonest cause for seeking medical evaluation in patients with Turner's syndrome.

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