Case Report

Familial primary ovarian insufficiency (POI) - A unique familial association

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Abstract Amenorrhea means absence of menses and is broadly divided in to two types: primary and secondary. Primary ovarian insufficiency is attainment of cessation of menses before the age of menopause with clinically elevated levels of follicle-stimulating hormone and low reserves of estrogen. The present case report takes into account a familial pattern of primary ovarian insufficiency with hypoplastic uterus in two generations of a family.

Keywords: 46 XX, family, primary ovarian insufficiency, siblings

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INTRODUCTION

Amenorrhea refers to absence of menses and is broadly divided as primary and secondary. Primary amenorrhea is defined as no menses by age 13 in the absence of growth or development of secondary sexual characteristics or no menses by age 15 regardless of the presence of normal growth and development of secondary sexual characteristics; on the contrary, a woman who has menstruated previously, but had no menses for an interval of time equivalent to a total of at least three previous cycles or no menses over a 6-month period is termed as secondary amenorrhoea.[1,2] Out of the two, secondary being the most common one.[2,3]

A woman, younger than age 40 years, with evidence of persisting hypergonadotropic hypogonadism in form of low level of estradiol and elevated levels of follicle-stimulating hormone (FSH) (>20U/L) is diagnosed with premature ovarian failure, a condition that in recent years is preferentially referred to as primary ovarian insufficiency (POI).[1,2] POI leads to premature follicular depletion in almost all of the cases leading to low ovarian estradiol reserve.[2] Due to this low estradiol reserve, the females fail to attain the normal stages of puberty and present primarily as primary amenorrhoea or in some cases with irregular cycles with scanty flow or primary infertility. We report a familial association of POI associated with hypoplastic uterus in two generations.

CASE REPORT

A 27-year-old female presented to gynecology outpatient department with complaints of scanty flow during peroids and history of bleeding only upon taking oral contraceptive pills. On further history evaluation, it was found out that patient had already been evaluated for the same and was diagnosed with POI at some other hospital.

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The patient had undergone a chromosomal analysis which was XX and an ultrasonography suggestive of hypoplastic uterus. The patient had also undergone diagnostic laparoscopy which revealed a hypoplastic uterus with atrophic ovaries. However, no ovarian biopsy had been performed. On evaluating the family history, it was found that patient's younger sister (23 years) and her third-degree paternal cousin (30 years) had same complaints and were similarly evaluated and subsequently diagnosed with POI. Patient's maternal aunt (37 years) also visited the OPD with complaints of irregular cycles with scanty flow and primary infertility. On her hormonal examination, similar finding of high FSH with a chromosomal analysis of 46 XX with a hypoplastic uterus was found. She too was subsequently diagnosed with POI.

DISCUSSION

The case reports a familial inheritance of POI in two successive generations, all being associated with hypoplastic uteri with major complaints of irregular and scanty flow of periods and primary infertility. The patient presenting with these complaints needs a thorough history and clinical examination. History focusing on the primary complaint as well as any similar complaints amongst the family members should be taken. The clinical examination comprises of head-to-toe examination with evaluation of thyroid and tanner staging of breast and pubic hair and examination of external genitalia. The patient should also be evaluated for the presence of axillary hair along with a per abdominal examination to check for any palpable mass in search of any undescendent testes.[4] In our cases, on examination all the females had underdeveloped breast and pubic hair (tanner stage 1-2), with absence of any axillary hair and no mass palpable per abdominally. The chromosomal analysis of all the females was 46 XX and ultrasonography of all was suggestive of hypoplastic uterus. The FSH levels of all the females were more than 100 U/L with low estradiol values and a normal thyroid function.

Laboratory evaluation involved determination of the level of FSH, thyroid-stimulating hormone, estradiol, and prolactin.[5] As the differential diagnosis is an exhaustive process, compartmentalizing the causes into hypothalamic, pituitary, ovarian, and lower organs disorders would be of benefit in such patients.[6]

The patients are currently on hormonal replacement therapy and under regular follow-up.

CONCLUSION

We report a familial inheritance of POI in two subsequent generations, all being associated with hypoplastic uteri with the major complaint of irregular and scanty flow of periods and primary infertility. This familial inheritance is rare and on extensive analysis can show any undiagnosed chromosomal defect and can be a good scope of research.[5]

Declarations

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Ethics committee approval
Not applicable

Contributions
SS and VS were involved in conception, literature search, analysis of data, and drafting the manuscript.

Authorship
All authors had access to the data and a role in writing this manuscript.

Consent for publication
The authors certify that they have obtained written informed consent of the patient for publishing the case details in a journal.

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