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Case Report

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A Rare Case of Secondary Amenorrhoea Due to a Congenital Anomaly in a a Young Female Patient: A Case Report

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ABSTRACT

Unilateral ovarian agenesis affects approximately 1 in 11,240 women while bilateral agenesis is even rarer. A 22-year-old Sri Lankan single woman and also a university undergraduate presented with a five-year absence of menstruation. She denied sexual activity, contraceptive use, or significant medical history. Physical examination revealed Tanner Stage III breasts and scanty hair. Investigations showed an atrophic uterus, congenital bilateral ovarian anomaly, and elevated FSH levels. Karyotyping performed on follow-up revealed a 46, XX genotype. The management involved a diagnostic laparoscopy, confirming intra-abdominal findings without complications. The post-operative follow-up was unremarkable. This case underscores the rarity of congenital bilateral ovarian anomaly in young women, presenting challenges in diagnosis and management. Despite extensive evaluation, including diagnostic laparoscopy and karyotyping, the exact aetiology remains elusive. Further research is needed to elucidate the underlying mechanisms of this rare condition and guide future management strategies.

Keywords: Amenorrhoea, congenital bilateral ovarian anomaly, karyotyping

INTRODUCTION

Unilateral ovarian agenesis affects approximately 1 in 11,240 women, while bilateral agenesis is even rarer (1). We present a rare instance of congenital bilateral ovarian anomaly (BOA) accompanied by uterine atrophy and narrowed fallopian tubes, manifesting as amenorrhoea for five years. The aim is to shed light on this uncommon anomaly and explore avenues for determining its aetiology. This case report follows the CARE Guidelines (2).

CASE REPORT

The patient is a 22-year-old single woman, and also a Sri Lankan university undergraduate who presented to the Obstetrics and Gynaecology Unit of National Hospital Kandy with a chief complaint of absence of menstruation for the past five years. She reported experiencing only two menstrual episodes, one at menarche and another following a norethisterone trial two years post-menarche. The patient denied any sexual activity, contraceptive use, or significant medical history. There was no

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family history of menstrual disorders, PCOS, or subfertility. Socially, she lived with her parents until moving to a university hostel, where she experienced some adjustment issues and academic stress.

On physical examination, the patient appeared thinly built with a BMI of 17 kg/m^2. There were no signs of pallor, goitre, or voice changes. Her breasts were compatible with Tanner Stage III development, and there was scanty pubic and axillary hair. Abdominal examination revealed no abnormalities and genital examination showed age-appropriate labia, clitoris, and prepuce, with an annular hymen observed.

Transabdominal ultrasound scan (TAS) revealed an atrophic uterus measuring 2.7×1.8 cm with BOA. The uterine length was measured in the midsagittal plane from the outer fundal serosal surface of the fundus to the external cervical os. The uterine height was measured in the midsagittal plane antero-posteriorly between outer serosal surfaces. The uterine width was measured in the transverse plane. Serum hormone profile showed an elevated FSH level and normal serum prolactin. The patient's presentation and diagnostic findings align with a diagnosis of congenital BOA, contributing to secondary amenorrhoea.

The patient underwent diagnostic laparoscopy under general anaesthesia, following shared decision-making. During laparoscopy, findings included an atrophied uterus (Figure 1), small fallopian tubes (Figure 2 and Figure 3), BOA (Figure 2 and Figure 3), and no retroperitoneal masses or free fluid. Post-operatively, the patient's recovery was uneventful.

Karyotyping performed on follow-up revealed a 46, XX genotype, which rules out the suspicion of congenital ovarian absence. The ultrasound kidney-ureter-bladder (KUB) scheduled prior to discharge was equivocal.

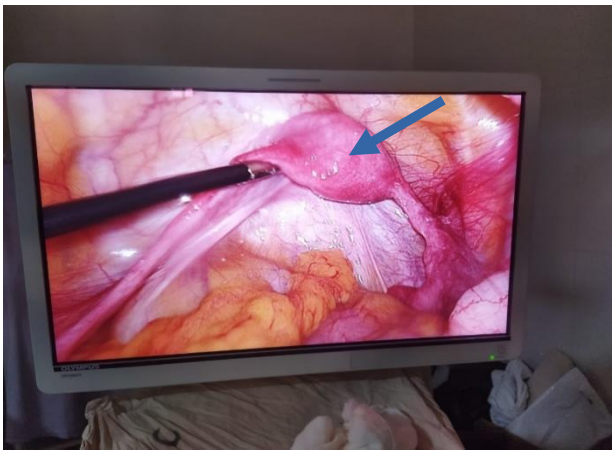

Figure 1: Diagnostic Laparoscopy - Central View: Laparoscopy revealed an atrophied uterus indicated by the blue arrow

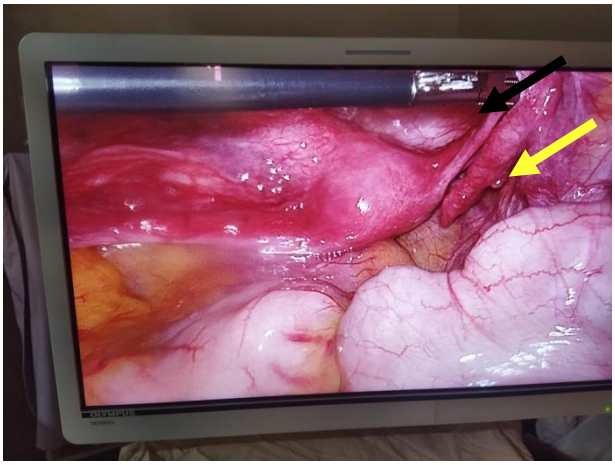

Figure 2: Diagnostic Laparoscopy - Left View: Laparoscopy revealed an absent left ovary (yellow arrow) and a small ipsilateral fallopian tube (black arrow)

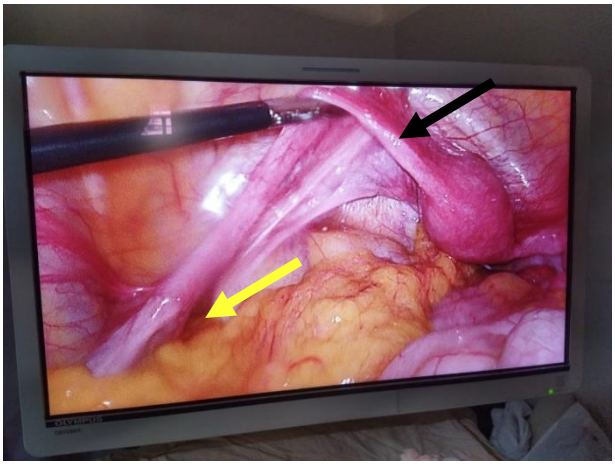

Figure 3. Diagnostic Laparoscopy - Right View: Laparoscopy revealed no visible right ovary macroscopically (yellow arrow) and a small ipsilateral fallopian tube (black arrow)

DISCUSSION

The findings of this case, presenting as congenital BOA in a young female patient, raise intriguing questions regarding the underlying pathophysiology and potential contributing factors. The absence of menstruation for five years in the absence of any known medical conditions or interventions is a rare and perplexing presentation.

Embryologically, the development of the genitourinary (GU) system from the intermediate mesoderm and the subsequent colonization of genital ridges by primordial germ cells are well-established processes. However, deviations from this norm, such as bilateral ovarian absence, are uncommon and often poorly understood. While various hypotheses exist in the literature, including vascular, embryological, and torsion-related causes (3), the exact mechanism in this case remains elusive.

The embryological hypothesis posits abnormalities in gonadal development as a potential explanation for unilateral ovarian absence (UOA). However, the presentation of congenital BOA challenges this theory, suggesting a more complex aetiology. The absence of vascular accidents or torsion-related events in the patient's history further complicates the picture, leaving the exact cause open to speculation.

One potential explanation could have been genetic factors influencing gonadal development but, the patient's karyotype was 46, XX (4). Genetic abnormalities or mutations affecting the genes involved in gonadal differentiation may disrupt normal ovarian development, leading to bilateral absence. However, without further genetic testing or familial history, karyotyping alone would not value in arriving at a precise diagnosis.

Although no ovaries were observed during laparoscopy and the patient had elevated FSH levels, the possibility of residual or rudimentary ovarian tissue secreting oestrogen cannot be excluded, as the patient presents with secondary amenorrhoea and a 46, XX karyotype.

Furthermore, an 18-year-old female with secondary amenorrhoea, hypergonadotropic hypogonadism, bilateral gonadal absence, a primitive uterus and right fallopian tube, and normal kidneys and vagina was described by Mutchinik et al (5). This elucidates the fact that congenital BOA can also occur even in a normal female karyotype.

In conclusion, while this case provides valuable insights into the presentation and management of bilateral ovarian absence, the underlying mechanisms remain poorly understood. Further research, including genetic studies and larger case series, is warranted to elucidate the pathophysiology of this rare condition and guide future management strategies.

CONCLUSION

In summary, we present a rare case of congenital bilateral ovarian anomaly leading to a secondary amenorrhoea in a 22-year-old Sri Lankan woman. Despite extensive evaluation, including diagnostic laparoscopy and karyotyping, the exact cause of BOA remains elusive. The absence of menstruation for five years in the absence of any known medical conditions or interventions underscores the complexity of this presentation. While various hypotheses, including vascular, embryological, and genetic factors, have been proposed, further research is needed to elucidate the underlying pathophysiology of BOA. This case highlights the importance of thorough evaluation and shared decision-making in the management of cases of secondary amenorrhoea with suspected ovarian anomalies. Further investigation and collaboration are essential to advance our understanding and improve the care of patients with rare reproductive disorders.

Author declaration

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None.

Authors' contributions:
Conceptualization and Design: Ranatunga RMDBR; Data Collection and Patient Care: WA, RMDBR; Manuscript Writing: NPH, NG; Critical Review and Editing: RMDBR.

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The authors declare that there is no financial or non-financial conflict of interest.

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Written informed consent was obtained from the patient before they participated in the case, ensuring their understanding of the purpose, procedures, and potential implications involved.

Statement on data availability:
All data generated during this study are included in this published article.

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