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A RARE CASE REPORT OF PRIMARY AMENORRHEA IN GONADAL DYSGENESIS

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ABSTRACT Primary amenorrhea is a common gynecological presentation with diverse etiologies, among which gonadal dysgenesis is an important but rare cause. Gonadal dysgenesis results from defective gonadal development due to abnormal germ cell migration or differentiation and may be associated with normal or abnormal karyotypes. We report a rare case of an 18-year-old female presenting with primary amenorrhea and poorly developed secondary sexual characteristics. Clinical examination revealed Tanner stage II breast development with absent pubic and axillary hair and infantile external genitalia. Evaluation showed hypergonadotropic hypogonadism. Diagnostic laparoscopy was done, confirmed the presence of a hypoplastic uterus with normal fallopian tubes and bilateral streak ovaries. Karyotyping demonstrated a normal 46,XX pattern, leading to the diagnosis of pure 46,XX gonadal dysgenesis. Early diagnosis of gonadal dysgenesis is crucial for timely initiation of hormone replacement therapy, development of secondary sexual characteristics, prevention of osteoporosis, and psychological support. A multidisciplinary approach is essential for optimal management and long-term care of such patients.

KEYWORDS : Primary amenorrhea, 46XX gonadal dysgenesis, hypergonadotropic hypogonadism

INTRODUCTION

Primary amenorrhea is defined as the absence of menarche by the age of 13 years in the absence of secondary sexual characteristics, or by 15 years of age in the presence of normal growth and secondary sexual development. It represents a significant diagnostic challenge, as it encompasses a wide spectrum of congenital, genetic, endocrine, and anatomical abnormalities. The common causes include disorders of sex development, hypothalamic–pituitary dysfunction, gonadal failure, and Müllerian anomalies.[1]

Gonadal dysgenesis is a major cause of hypergonadotropic hypogonadism and primary amenorrhea, resulting from defective development of the gonads due to abnormal germ cell migration or differentiation. Based on karyotype, gonadal dysgenesis is classified into two broad categories: those with a normal karyotype (46,XX or 46,XY—Swyer syndrome), referred to as pure gonadal dysgenesis, and those with abnormal karyotypes, most commonly Turner syndrome (45,X) and mosaic variants (45,X/46,XX or 45,X/46,XY).[2]

Pure 46,XX gonadal dysgenesis is a rare condition characterized by a normal female phenotype, presence of Müllerian structures, streak or small ovaries, lack of spontaneous puberty, and primary amenorrhea. Affected individuals typically present with absent or delayed secondary sexual characteristics and biochemical evidence of hypergonadotropic hypogonadism with elevated follicle-stimulating hormone and luteinizing hormone levels and low estrogen levels. The estimated incidence of 46,XX gonadal dysgenesis is less than 1 in 100,000 females.[3]

Imaging modalities such as ultrasonography and magnetic resonance imaging play a crucial role in demonstrating the presence of the uterus and gonadal morphology, thereby differentiating gonadal dysgenesis from Müllerian agenesis and androgen insensitivity syndrome. Karyotyping is essential for definitive diagnosis and classification. Early identification is vital to initiate hormone replacement therapy for induction of secondary sexual characteristics, maintenance of bone health, and psychological well-being.[4]

CASE STUDY

An 18-year-old female from Manvi, Raichur, presented to the Obstetrics and Gynecology outpatient department with a complaint of primary amenorrhea. She was born out of a non-consanguineous marriage. There was no history of cyclical abdominal pain. Her siblings had attained menarche at an average age of 14 years. There was no significant past medical or surgical history.

On general physical examination, her vital parameters were within normal limits. She had a height of 152 cm, weight of 42 kg, and body mass index of 18.18 kg/m². She had normal intelligence and a female phenotype. There were no dysmorphic features suggestive of Turner syndrome such as webbed neck, low hairline, shield chest, skeletal deformities, or thyroid enlargement.

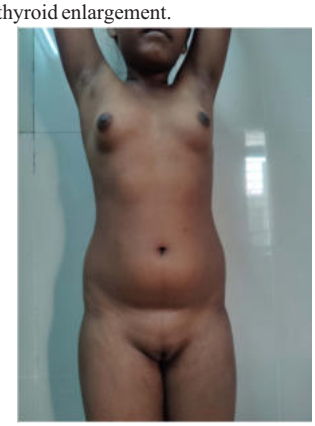

Figure -1 Breast-Tanners stage II; Pubic and axillary hair Tanners stage I

Assessment of secondary sexual characteristics revealed Tanner stage II breast development, with absence of axillary and pubic hair (Tanner stage I). External genital examination showed infantile hypoplastic labia majora, labia minora, and clitoris. Systemic examination including cardiovascular, respiratory, abdominal, and neurological systems was unremarkable.

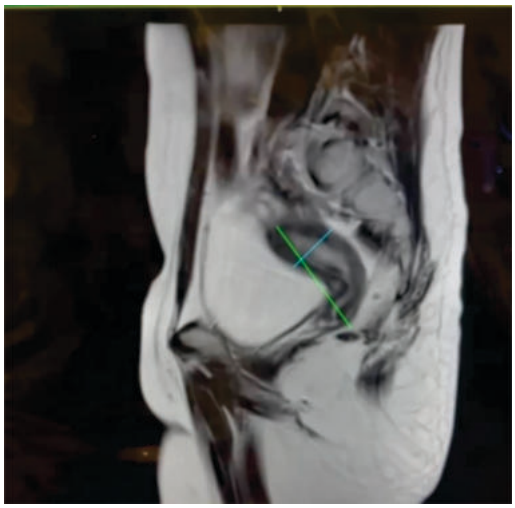

Figure-2 MRI PELVIS showing hypoplastic arcuate uterus measuring 3.5x1.5x1cms

Laboratory investigations revealed hemoglobin of 12.4 g/dL, total leukocyte count of 9,700 cells/mm³, and normal renal and liver function tests. Thyroid function tests were within normal limits. Hormonal evaluation demonstrated hypergonadotropic hypogonadism, with elevated follicle-stimulating hormone (68.80 mIU/mL) and luteinizing hormone (52.71 mIU/mL), low estradiol levels (24 pg/mL), normal prolactin levels, and reduced anti-Müllerian hormone (0.4 ng/mL), suggestive of ovarian insufficiency.

Ultrasonography of the abdomen and pelvis revealed an arcuate uterus with bilateral small ovaries. Magnetic resonance imaging of the pelvis confirmed a hypoplastic arcuate uterus measuring 3.5 × 1.5 × 1 cm with bilateral small ovaries of reduced volume and few follicles, raising the possibility of gonadal dysgenesis.

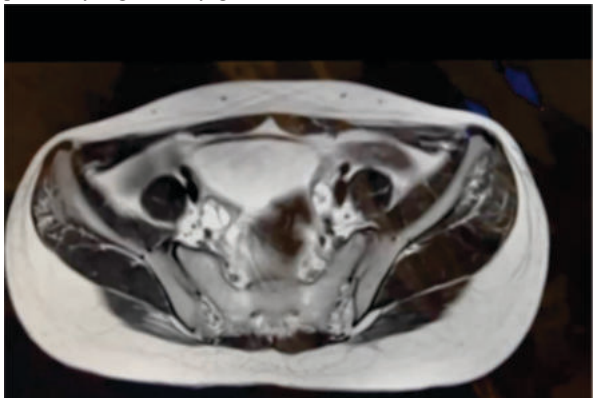

Figure 3 - MRI PELVIS bilateral streak ovaries right ovary measuring 1.1x0.8x0.4 cms volume 1.5cc and left ovary measuring 1x0.8x0.5cms volume 1 cc with few follicles

Examination under anesthesia showed normal vaginal length, a normal cervix with a pinpoint external os, and hypoplastic external genitalia.

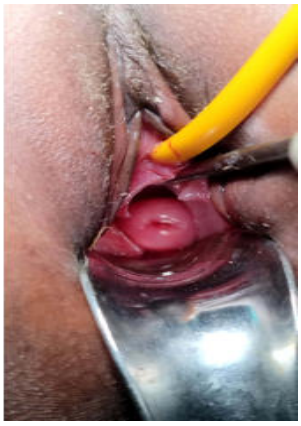

Figure-4 Per speculum examination-vaginal orifice normal and cervix normal with pinpoint os.

Diagnostic laparoscopy revealed a hypoplastic arcuate uterus with normal bilateral fallopian tubes and bilateral small ovaries, consistent with streak gonads.

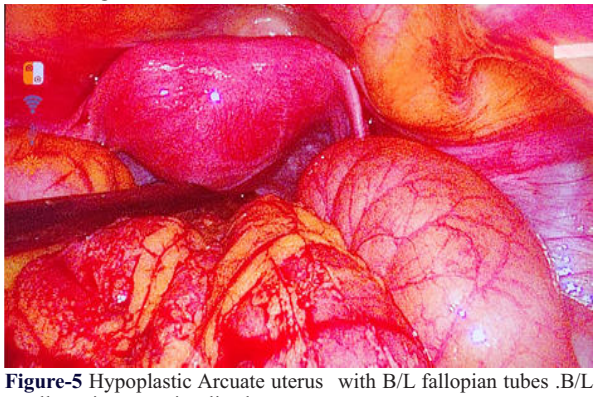

Figure-5 Hypoplastic Arcuate uterus with B/L fallopian tubes .B/L small ovaries were visualized.

Peripheral blood karyotyping demonstrated a 46,XX chromosomal pattern, confirming the diagnosis of pure 46,XX gonadal dysgenesis. The patient was started on estrogen replacement therapy and counseled regarding the condition, need for long-term hormone therapy, bone health, and regular follow-up.

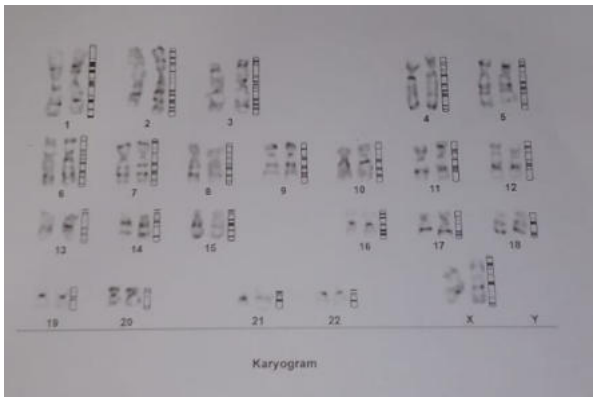

Figure-8 karyotype showed 46XX

Primary amenorrhea due to gonadal dysgenesis encompasses a heterogeneous group of disorders characterized by failure of ovarian development and resultant hypergonadotropic hypogonadism. Pure 46,XX gonadal dysgenesis is a rare condition in which affected individuals have a normal female phenotype, normally formed Müllerian structures, and

streak or hypoplastic ovaries[5] Unlike Turner syndrome, these patients lack somatic abnormalities and have a normal chromosomal complement, making diagnosis challenging without detailed hormonal and imaging evaluation[6]

In the present case, the patient demonstrated classical clinical features including primary amenorrhea, incomplete pubertal development, and absent pubic and axillary hair. Biochemical evaluation revealed markedly elevated gonadotropins with low estradiol and reduced anti-Müllerian hormone levels, consistent with primary ovarian insufficiency. Imaging studies confirmed the presence of a hypoplastic uterus and small ovaries, effectively excluding Müllerian agenesis and androgen insensitivity syndrome, which are important differential diagnoses in primary amenorrhea.

Diagnostic laparoscopy remains valuable in selected cases to directly visualize gonadal morphology and internal genital organs, especially when imaging findings are inconclusive. The identification of bilateral streak ovaries in this patient supported the diagnosis of gonadal dysgenesis[7] Although karyotyping revealed a normal 46,XX pattern, mutations in genes involved in ovarian development such as FOXL2, BMP15, FSHR, and NR5A1 have been implicated in the pathogenesis of pure gonadal dysgenesis, though advanced genetic testing may not be routinely available[8]

Early recognition of this condition is crucial, as delayed diagnosis may result in compromised bone mineral density, adverse cardiovascular effects, and significant psychological distress[9] Hormone replacement therapy is the cornerstone of management, aimed at inducing secondary sexual characteristics, achieving uterine growth, and preventing long-term complications[10]

CONCLUSIONS

Pure 46,XX gonadal dysgenesis is an uncommon etiology of primary amenorrhea characterized by hypergonadotropic hypogonadism, streak or hypoplastic ovaries, and preserved Müllerian structures in individuals with a normal female phenotype. This case emphasizes the importance of a structured diagnostic evaluation incorporating clinical assessment of pubertal development, endocrine profiling, pelvic imaging, diagnostic laparoscopy, and cytogenetic analysis to achieve an accurate diagnosis and exclude other disorders of sex development[11] Early initiation of hormone replacement therapy is essential for induction and maintenance of secondary sexual characteristics, optimization of uterine development, preservation of bone mineral density, and mitigation of long-term metabolic and cardiovascular risks. Comprehensive management necessitates a multidisciplinary approach with long-term follow-up, genetic

counseling, and psychosocial support to improve overall health outcomes and quality of life in affected individuals. [12]

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