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46 XY GONADAL DYSGENESIS MIMICKING TURNER SYNDROME: DIAGNOSTIC CHALLENGES AND CLINICAL IMPLICATIONS

Suseelah Bala Subramaniam[1], Rabeah Md Zuki[2], Loh Hoong Heng[1]
[1]*Department of Internal Medicine, Hospital Kulim*
[2]*Department of Endocrinology, Hospital Sultan Abdul Halim*

INTRODUCTION

Swyer syndrome (46, XY gonadal dysgenesis) is a rare disorder of sex development, characterized by a phenotypic female with streak gonads and hypergonadotropic hypogonadism. It presents with primary amenorrhea and delayed puberty. Overlapping clinical features with Turner syndrome may cause diagnostic uncertainty, highlighting the role of karyotypic evaluation in diagnosis.

CASE

A 35-year-old phenotypic female was first evaluated at age 27 during admission for an acute viral illness, when incidental findings of primary amenorrhea, short stature (height 131 cm), webbed neck, pectus excavatum, and absent secondary sexual characteristics raised suspicion of Turner syndrome. She had hypertension, with initial imaging suggesting coarctation of the thoracic aorta. CT angiography showed focal narrowing of the descending aorta; multidisciplinary review favored aortic folding, and she was managed conservatively. Endocrine evaluation demonstrated hypergonadotropic hypogonadism, with markedly elevated follicle-stimulating hormone (FSH 108.6 IU/L) and luteinizing hormone (LH 23.16 IU/L) in the presence of low estradiol levels. Pelvic imaging revealed an atrophic uterus with absence of bilateral ovaries, consistent with gonadal dysgenesis. Karyotypic analysis was performed, demonstrating a 46, XY genotype with confirmed SRY gene presence, establishing the diagnosis of Swyer syndrome. DEXA scan demonstrated osteoporosis, in keeping with prolonged hypogonadism. She was commenced on hormone replacement therapy, resulting in the development of secondary sexual characteristics and regular withdrawal bleeding. She remains under structured multidisciplinary follow-up, with endocrine-led management of hypothyroidism and osteoporosis, alongside cardiology follow-up and gynecological surveillance.

CONCLUSION

This case highlights the diagnostic complexity of disorders of sex development with overlapping phenotypes and the need for re-evaluation when clinical progression is atypical. Diagnosis enables hormone replacement therapy for induction of secondary sexual characteristics and preservation of bone health. Integration of clinical, biochemical, and genetic data within a multidisciplinary framework is essential to achieve diagnostic accuracy and optimize outcomes.