

46XX gonadal dysgenesis mimicking coexistent mullerian agenesis

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Abstract

Background: 46, XX gonadal dysgenesis is a rare genetic heterogenous disorder characterized by sexual infantilism, primary amenorrhea and hypergonadotropic hypogonadism in 46, XX female. Whereas Müllerian agenesis or MRKH (Mayer-Rokitansky-Küster-Hauser) syndrome is characterized by a congenital hypoplastic uterus and the upper two-thirds of the vagina, causing primary amenorrhea but normal secondary sexual characteristics. Due to estrogen deficiency during the developmental period, gonadal dysgenesis may result in a hypoplastic uterus that is reversible with adequate estrogen replacement in postnatal life. Here we present a case of 46,XX gonadal dysgenesis with a hypoplastic uterus, which later developed to adult size and shape with 1 year of estrogen replacement, refuting coexistent Müllerian agenesis.

Case summary: A 15-year-old female came to us with primary amenorrhea and sexual infantilism. On examination, she has normal body habitus and vertical growth but absent secondary sexual characteristics (Tanner stage 1) with normal female-type genitalia. Investigations revealed hypergonadotropic hypogonadism (high FSH, LH and low estrogen). Karyotyping showed 46XX, pelvic ultrasonogram showed hypoplastic uterus with small gonads. We diagnosed the case as 46, XX Gonadal dysgenesis with Müllerian agenesis. We started low-dose estrogen as priming and continued for a year. After one year of estrogen therapy, a repeat ultrasonogram revealed a normally developed uterus and gonad. Full development of the uterus with estrogen therapy proved there was no Müllerian agenesis. We therefore confirmed the diagnosis of 46,XX Gonadal Dysgenesis only.

Conclusion: Gonadal dysgenesis in female karyotyping can mimic coexistent Müllerian agenesis due to lack of estrogen in the intrauterine period, leading to a hypoplastic uterus, but it recovers with estrogen replacement in postnatal life. [*J Assoc Clin Endocrinol Diabetol Bangladesh*, 2026;5(Suppl 1): S70]

Keywords: Gonadal dysgenesis, Mullerian agenesis, Estrogen therapy

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