Bilateral Ovarian Agenesis and Bone Modeling

Disease in Pre-puberty Girl With Primary

Amenorrhea

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Abstract

Ovarian agenesis (OA) is a rare congenital condition characterized by the absence of one or both ovaries,often associated with chromosomal abnormalities, hormonal imbalances, and structural deformities. The condition is frequently diagnosed in females presenting with primary amenorrhea and delayed sexual development. This case report highlights a unique presentation of bilateral ovarian agenesis in a patient with chromosome X translocation, bone modeling disease, and primary amenorrhea.

A 17-year-old female with primary amenorrhea and a Madelung deformity presented with wrist pain, paresthesia, and limited range of motion. Imaging revealed delayed joint growth, a fragility fracture, and osteoporosis. Further evaluation uncovered a small uterus and absent ovaries on ultrasound and magnetic resonance imaging (MRI). Hormonal analysis showed elevated gonadotropins, follicle stimulation hormone and luteinizing hormone (FSH and LH), low estradiol, and low anti-mullerian hormone (AMH) levels. Laparoscopy confirmed rudimentary bilateral ovaries and chromosomal analysis revealed 46,X,der(X)t(X;3) (p11;p11), indicating an X chromosome translocation with an SRY gene microdeletion. The patient was diagnosed with bilateral ovarian agenesis and referred for multidisciplinary care. Treatment included hormonal therapy with progyluton and estrofem, transitioning to marvilon, alongside physical therapy, nutritional support, and psychological counseling. After six months, the patient showed

improvements in Tanner's score, weight, mood, and bone density (transition from osteoporosis to osteopenia). Menstruation was restored, reflecting the success of the combined hormonal therapy and supportive treatments.

This case underscores the importance of integrating cytogenetic, hormonal, and clinical evaluations in diagnosing and managing rare presentations of ovarian agenesis. Early hormonal therapy and multidisciplinary care can significantly improve physical and psychological outcomes, including restoring menstruation and bone density. This is the first reported case of bilateral ovarian agenesis with chromosome X translocation presenting with phenotypic amenorrhea and bone deformities, demonstrating the value of

tailored therapeutic approaches. Ongoing monitoring remains essential to ensure continued progress and mitigate long-term risks.