A CASE OF TRANSVERSE TESTICULAR ECTOPIA WITH PERSISTANT MULLERIAN DUCT SYNDROME: A NOVEL AMH GENE MUTATION

Suna KILINÇ, Tuğrul ÇİÇEK, Serdar MORALIOĞLU, Ayla GÜVEN

Saglik Bilimleri University Istanbul Bagcilar Education and Research Hospital, Department of 1Pediatric Endocrinology, 2Pediatric Surgery
Saglik Bilimleri University Istanbul Zeynep Kamil Education and Research Hospital, Department of 3Pediatric Surgery, 4Pediatric Endocrinology

INTRODUCTION:
The concurrence of transverse testicular ectopia (TTE) with persistent müllerian duct syndrome (PMDS) is extremely rare. Here, we report a case of TTE with PMDS in a 7-month-old male infant presenting with inguinal hernia and a novel homozygous mutation in the AMH gene.

CASE REPORT:
A 7-month-old male infant presented to the pediatric surgery department with a right-sided inguinal hernia and bilateral undescended testis. During herniotomy, tissues suggestive of a rudimentary uterus with fallopian tubes and testes like structures on both sides of uterus were found [Fig-1-2]. A biopsy of the gonads and structure located midline to the gonads was performed and patient was referred to our pediatric endocrinology department with a preliminary diagnosis of sexual development disorder. At physical examination, weight was 8.4 kg [standard deviation score (SDS): -0.09] and height was 71 cm (SDS: -0.51). Blood pressure was 98/60 mm/Hg. Both testes were not palpable. Penis size and appearance were normal. Parents were first-degree relatives.

Figure1

Figure2

Serum follicle-stimulating hormone (FSH) level was 0.92 IU/L (reference value 1.5-12.4 IU/L), luteinizing hormone (LH) was 1.17 IU/L (1.7-8.6 IU/L), total testosterone (TT) 0.025 ng/mL (0.12-0.21 ng/mL), estradiol (E2) was <5 pg/mL (<12 pg/mL), and AMH was <0.02 ng/mL (24.2-275.4 ng/mL). Karyotype analysis revealed a XY karyotype. Histopathological examination of the hernia contents was consistent with a rudimentary uterus with bilateral edematous tubal structures. Gonad biopsies showed immature seminiferous tubule. AMH gene sequence analysis performed with a preliminary diagnosis of AMH deficiency revealed a previously undescribed homozygous IVS2-3C>G (c.556-3C>G) mutation. The parents had the same mutation in heterozygous form. The patient was evaluated as a case of AMH deficiency and presented to our “Sex Development Disorders Council”. Upon their decision, orchiopexy was performed.

CONCLUSION:
If patients had a unilateral inguinal hernia and contralateral cryptorchidism, TTE with PMDS should be considered. The mutation detected in the AMH gene is associated with PMDS and its phenotype is variable.