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

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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.

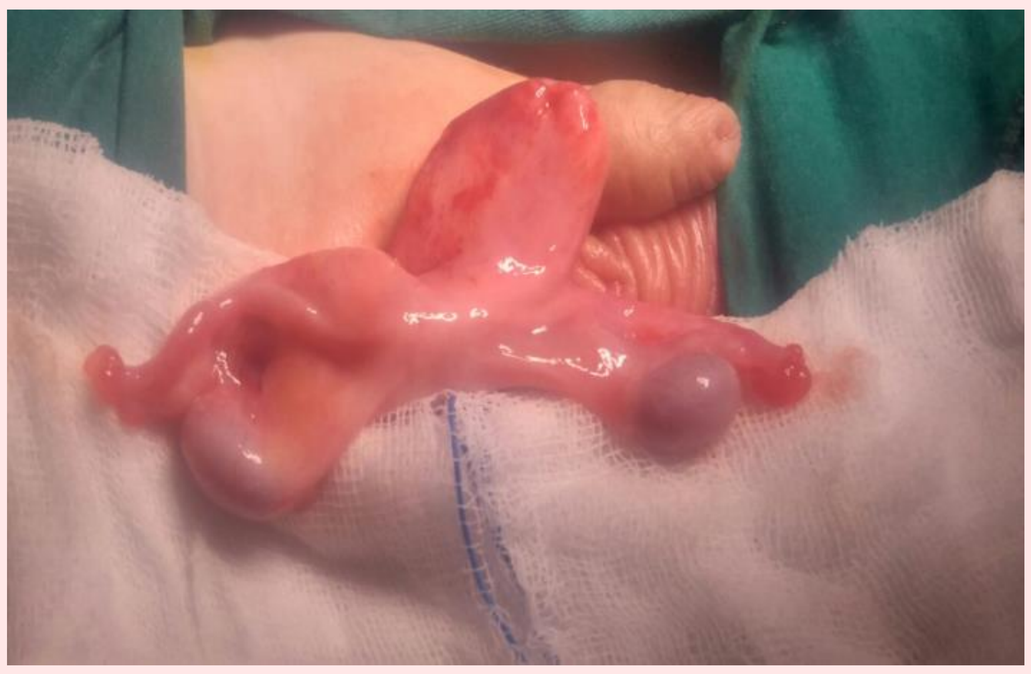

Figure1

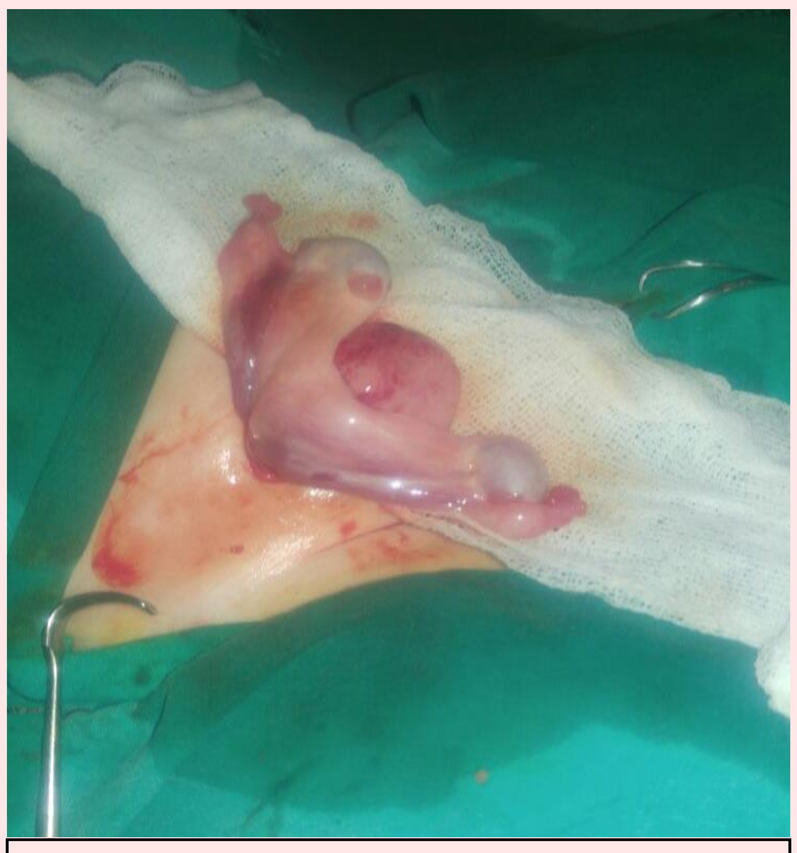

Figure2

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.