Connecting Endocrinology Across the Life Course
Joint Congress of ESPE and ESE 2025
Copenhagen, Denmark, 10-13 May 2025
ESPE European Society for Paediatric Endocrinology
European Society of Endocrinology

**Abstract N°: JOINT4357**

**Persistent Müllerian Duct Syndrome (PMDS) with Transverse Testicular Ectopia: A Case Report**

**Azad Akberzade*[1], Nabat Aghayeva[1], Laman Sultanova[1, 2]**

*[1]META Pediatric Endocrinology Center, BAKU, Azerbaijan, [2]Azerbaijan Medical University, META Pediatric Endocrinoloogy Center, Baku, Azerbaijan*

Introduction:

Persistent Müllerian Duct Syndrome (PMDS) is characterized by the presence of Müllerian remnants (fallopian tubes, uterus, and the upper one-third of the vagina) in individuals with a male phenotype. It is a rare cause of 46,XY disorders of sex development. In some cases, PMDS may be associated with transverse testicular ectopia (TTE), and approximately 120 cases have been reported in the literature. Patients typically present with cryptorchidism, which is the most common disorder of sex development in male infants, occurring in about 3% of newborn males.

Case Presentation:

A 10-month-old male infant presented with bilateral cryptorchidism and unilateral inguinal hernia. His parents were first-degree relatives. Pubertal examination was consistent with Tanner stage 1, and the testes were not palpable. Hormonal analysis showed FSH: 1.2 mIU/mL, LH: <0.69 mIU/mL, Testosterone: <0.23 ng/mL, and ACTH: 15.9 pg/mL. Ultrasound (USG) revealed no left testis, while a mass was detected adjacent to the right testis. Diagnostic laparoscopy confirmed the presence of both testes on the right side, with no testis on the left. Müllerian remnants, including fimbriae, a fallopian tube, and a rudimentary uterus behind the bladder, were identified. A biopsy from the abnormal-looking testis (ectopically located left testis, larger in size with cystic structures) revealed immature testicular tissue. Anti-Müllerian Hormone (AMH) level was 183 ng/mL (reference range: 33–60.2 ng/mL). Karyotype analysis confirmed 46,XY. Due to the potential risk of tumor development in the context of PMDS, surgical removal of the abnormal testis and Müllerian remnants was planned while preserving the healthy testis and vas deferens. Orchidopexy for the healthy testis and inguinal hernia repair were performed, and further genetic analysis was planned.

Conclusion:

Anti-Müllerian Hormone (AMH) is secreted by Sertoli cells from the 8th gestational week. PMDS results from either decreased AMH secretion due to an autosomal recessive mutation or mutations in the AMH receptor gene (AMHR-II). Mutations in AMH and AMHR-II genes lead to similar phenotypes. The characteristic clinical features of PMDS include cryptorchidism and testicular ectopia associated with an inguinal hernia. Since testicular differentiation is normal, testosterone production and external genitalia remain unaffected. Although rare, TTE should be considered when

a testis is not found in its usual location, and the contralateral side should be examined. In cases of cryptorchidism with an inguinal hernia, PMDS should always be considered in the differential diagnosis.

**Disclosure of interest:** None declared