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The Effects of Androgen Receptor Mutation on Cryptorchid Testes in 46, XY Female

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Abstract : In the current study, we enrolled a 46, XY phenotypically female patient bearing testes in her inguinal canal. DNA sequencing of the AR gene detected a missense mutation C.1715A > G (p. Y572C) in exon 2 which is already known to cause Complete androgen insensitivity syndrome (CAIS). We further studied the effects of this mutation on the testicular histopathology of the patient. No spermatocytes were seen in the surface spreading of testicular tissues while H&E staining showed that seminiferous tubules predominantly have only Sertoli cells. To confirm this meiotic failure is likely due to the current AR mutation we performed mRNA expression of genes associated with AR pathway, expression and location of the associated proteins in testicular tissues. Western blot and real-time PCR data showed that the patient had high levels of expression of AMH, SOX9, and INNB in testis. Tubules were stained with SOX9 and AMH which revealed Sertoli cell maturation arrest. Therefore, we suggest that AR mutation enhances AMH expression which ultimately leads to failure in the maturation of Sertoli cells and failure in spermatogenesis.

Keywords : androgen receptor, spermatogenesis, infertility, Sertoli cell only syndrome
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