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Mutation p.Y572C in androgen receptor gene is associated with Sertoli cell only phenotype in a patient with complete androgen insensitivity

Ihtisham Bukhari[1,2], Guangyuan Li[5], Liu Wang[2], Xiaohua Jiang[2], Furhan Iqbal[2,3], Yuanwei Zhang[2], Zhang Huan[2], Howard J Cooke[4], Dexin Yu[5] and Qinghua Shi[2]
[1]Women University of Azad Jammu and Kashmir, Pakistan
[2]University of Science and Technology of China, China
[3]Bahauddin Zakariya University, Pakistan
[4]Western General Hospital, UK
[5]The Fourth Affiliated Hospital-Anhui Medical University, China

In current study, we enrolled a 46,XY female patient with a testis 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 CAIS. We focused on the effect of this mutation on the testicular histopathology of the patient. Surface spreading of testicular tissues showed an absence of spermatocytes while H&E staining showed that seminiferous tubules predominantly have only Sertoli cells with a few tubules containing spermatogonia. This meiotic failure is likely due to the effect of the AR mutation leading ultimately to Sertoli cell only syndrome. Tubules were stained with SOX9 and AMH which revealed Setoli cell maturation arrest. Western blot and real-time PCR data showed that the patient had high levels of expression of AMH, SOX9 and INNB in the testis. Therefore we suggest that dysfunctioning of AR enhances AMH though up-regulation of SOX9 which may serve to protect the testis from precocious Sertoli cell maturation.

bukhari5408@gmail.com

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