Identification of a Novel Mutation in an Iranian Family with 17-β Hydroxysteroid Dehydrogenase Type 3 Deficiency; A case series

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Abstract

Background: To present the clinical and genetic features of a male ambiguity due to 17beta-hydroxysteroid dehydrogenase 3 (17B-HSD3) deficiency.

Case presentation: The proposita was an 11-year-old girl and the first child of a consanguineous family. The external genitalia were completely female and had a short vaginal pouch. She had palpable gonads in her inguinal area and underwent bilateral gonadectomy at the age of two. At age 10, she was referred to our clinic for more evaluation. In pelvic sonography, uterine and ovarian were not seen. Her karyotype was 46, XY and her LH and FSH levels were elevated, and three of the patient's aunts and one of the mother's aunts had similar signs.

Conclusion: We identified a novel homozygous missense variation (c.731T>A, p. Ile244Lys) in HSD17B3 gene. This alteration changes Isoleucine to Lysine in exon 10.

Keywords: 17-β-HSD3, Virilization, 46, XY, Iran