‘I AM A BOY SINCE 8-YEARS-OLD’ FEMALE DURING CHILDHOOD, VIRILIZATION AT PUBERTY

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Background:

5α-reductase-deficiency is an autosomal recessive disorder with clinical spectrum ranges from a male phenotype with hypospadias to a female phenotype with normal wolffian structures. Many different mutations of SRD5A2 gene has been described in affected patients and clinical signs can vary depending on the degree of enzyme deficiency.

Case Presentation:

Fourteen-years-old girl admitted to our clinic because of feeling as a boy since 8-years-old. Parents were first degree cousins. She was born at home. Physical examination showed remarkable facial acne and thick voice. In genital examination phallus was 4x1.5 cm and bilaterally gonads were palpable in labia major with volume of 15 ml. Breast development was Tanner stage 1, pubic hair development was Tanner stage 4. In pelvic ultrasound there was no uterus and testes were observed in distal inguinal canal. Standard dose ACTH test was normal. In laboratory analysis; basal LH was 5.62 mIU / ml (N = 1.24-8.62), FSH was 10.2 mIU / mL (N: 1:27 to 19:26), total testosterone was 4.31 ng / ml (N = 1.75-7.81) and anti-müllerian hormone was 9.81 ng / ml (N: 1.3-14.8). After βhCG stimulation test; testosterone:androstenedione ratio was 3.27 (N> 0.8), testosterone:dihydrotestosterone ratio was 433.7 (N <10). Karyotype was 46XY. SRD5A2 gene analysis revealed a homozygous mutation in exon 1 (c.193g> C, p.ala65pro) confirming the diagnosis of 5-alpha-reductase deficiency. Psychiatric evaluation was consistent with male sexual identity. Patient eventually underwent to sex-change hormonal therapy and gender reassignment surgery by the decision of Gender Committee.

Conclusion:

5α-reductase-deficiency, although rare, should be suspected in any girl presenting with pubertal virilization. In patients with 5α-reductase-deficiency diagnosed so late, the management is highly problematic and requires extensive psychological evaluation and support of the patient and his family for the final decision of gender assignment. Experienced multidisciplinary approach is extremely important for good clinical management.

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