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A Novel Mutation in a Patient with 5-α Reductase Deficiency Reared as Girl

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5-α reductase deficiency is one of 46,XY disorders of sexual differentiation characterized by androgen metabolism disorder. In the literature, there are few cases with 5-α reductase deficiency reared as girl.

A 12-year and 8-month-old female patient presented with primary amenorrhea, absent breast development, axillary and pubic hair. Physical examination revealed a weight of 53.9 kg [75-90p, 0.87 standard deviation score (SDS)], a height of 167 cm (95p, 1.72 SDS), and normal vital signs. Genital examination disclosed female external genitalia with no cliteromegaly, hirsutism, or acne. The target height was 160.2 cm, and bone age revealed 12 years.

Follicle-stimulating hormone, luteinizing hormone, estradiol, and total testosterone levels were 3.68 mIU/mL, 3.04 mIU/mL, < 10 pg/mL, and 202.76 ng/mL. Adrenocorticotropic hormone and serum cortisol levels were normal in terms of adrenal insufficiency. Ultrasound imaging revealed no uterus and ovary. Karyotype analysis revealed 46,XY and SRY + was detected by quantitative fluorescent polymerase chain reaction. 5-α reductase deficiency was diagnosed with homozygous IVS3 + 1G > T (c.547 + 1G > T) mutation. Prophylactic bilateral gonadectomy was planned.

We emphasize the importance of karyotype analysis in patients with delayed puberty and primary amenorrhea. Prophylactic bilateral gonadectomy should be kept in mind for 5-α reductase deficiency in patients reared as girl to prevent the development of gonadal malignancy.

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Two Cases of Klinefelter Syndrome

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Klinefelter syndrome is the most commonly seen sex chromosomal disorder in males. The typical clinical features of this syndrome are symptoms of hypogonadism in different degrees. Up to 80% of patients with Klinefelter syndrome have 47,XXY karyotype, which is the prevalent type.

Case 1: A 36-year-old male patient applied to our clinic due to the complaint of erectile dysfunction. On physical examination, height was 183 cm, weight 80 kg, and BMI was 23.9 kg/m^2. Axillary and pubic hair were present. He had bilateral gynecomastia. Testis volume measured with orchidometer was 20 mL. Karyotype was 47,XXY. In the laboratory examination, follicle-stimulating hormone was 36 mIU/mL (1.27-19.26), luteinizing hormone (LH) 25 mIU/mL (1.24-8.62), and total testosterone was 2.15 ng/mL (2.8-8). Spermiogram demonstrated azoospermia. Intramuscular testosterone treatment was initiated once in three weeks.

Case 2: A 43-year-old male patient applied to our clinic due to the complaints of libido loss and infertility. On physical examination, height was 185, weight was 95 kg, BMI was 27.8 kg/m^2, axillary and pubic hair were present, the penis was small, and he had truncal obesity. Karyotype was 47,XXY. Follicle-stimulating hormone was 61 mIU/mL (1.27-19.26), LH 25 mIU/mL (1.24-8.62), total testosterone 0.24 ng/mL (2.8-8), and free testosterone was 3.15 pg/mL (57-178). In bone densitometry, L1-4 Z was -3.5. Intramuscular testosterone treatment was initiated once in three weeks.

In Klinefelter syndrome, testosterone replacement treatment eliminates all negative effects related to androgen deficiency; however, it has no effect on fertility. It should be remembered that Klinefelter syndrome may be detected in infertile males.

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Major Depression and Fabry Disease: A Case Report

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Fabry disease is a genetic lysosomal storage disease which affects several organs. The main defect is absence of alpha galactosidase enzyme activity. Kidney, central nervous system, cardiovascular system, and ocular system are the main influenced systems, but neuropsychiatric symptoms may develop in some cases. Current studies showed that psychiatric symptoms may be seen in both genders apart from neurological ones.

A 22-year-old female patient with history of two suicide attempts was consulted from psychiatry clinic. Her father had Fabry disease. He had renal transplantation and his enzyme level is low 3.2 nmol/mg/h (normal range > 30). GLA gene mutation analysis revealed that he had p.G261D (c.782G > A) heterozygote mutation. Enzyme replacement treatment was administered