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A Case of 46,XX DSD Due to a Novel Mutation in P450 Oxidoreductase Gene

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P450 oxidoreductase (*POR*) enzyme deficiency is a rare form of congenital adrenal hyperplasia, characterized by combined and partial impairments in steroidogenic enzymes. It may be associated with Antley-Bixler syndrome.

Here we report a newborn with ambiguous genitalia, skeletal malformations, and adrenal insufficiency who was diagnosed with Antley-Bixler syndrome.

A 12-day-old newborn presented with ambiguous genitalia. She was born small for gestation age with a birth weight of 2 400 g at 38 weeks of gestation to a non-consanguineous couple. The pregnancy was uneventful except for maternal voice deepening. Her weight was 2 350 g (-1.99 SDS), her length was 49 cm (-0.12 SDS), and head circumference was 32.5 cm

(-1.47 SDS). She had prominent eyeballs, frontal bossing, dysplastic ears, bilateral upper extremity contractures, left choanal stenosis, and genital virilization (Prader stage 3) with 1 cm phallus and bilaterally non-palpable gonads.

Adrenocorticotrophic hormone test showed adrenal insufficiency with a low cortisol peak (6 mcg/dL) and high 17-OH progesterone peak (50 ng/mL). Her karyotype was 46,XX. Bilateral ovarian cysts were detected on ultrasound imaging. These findings suggested *POR* deficiency and Antley-Bixler syndrome. The molecular genetic analysis of *POR* gene revealed a novel compound heterozygous mutation (IVS3-1 G>A (c.238-1 G>A)/c.929_937delTCTCGACT). Both parents were heterozygous for these mutations.

POR deficiency should be considered in patients with congenital adrenal hyperplasia with a history of maternal virilization during pregnancy and these patients should be evaluated for the presence of skeletal malformations.

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New Chromosomal ins(6;7)(Q13:P22) Anomaly in Klinefelter Syndrome Detected Coincidentally in Patient with Signs of Primary Hypogonadism

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We described our case to show that different genetic disorders can accompany Klinefelter syndrome.

An 18-year-old male patient referred to our clinic with complaints of aggressive behavior, learning difficulties, inability to gain weight, tall stature, lack of facial hair, and erectile dysfunction. He has been using valproic acid for epilepsy. On physical examination, the height was 187 cm, body weight 73.4 kg, BMI 21 kg/m², arm span 192 cm, upper body 91 cm, lower body 96 cm, and ratio of pubis-vertex/pubis-floor was < 1. Vital signs were normal, but he had slight mental retardation. Testes were small and in the scrotum; penis length was 2.5cm. No beard was present; axillary and pubic hairs were scarce. Gynecomastia was absent. Systemic examination was normal except for midsystolic murmur. Height of mother and father were 162 and 175 cm, respectively. With these clinical findings, pre-diagnosis of hypogonadism was established and workup was performed.

Blood count and biochemical analysis were normal; follicle-stimulating hormone was measured as 55.95 mIU/mL, luteinizing hormone 8.64 mIU/mL, total testosterone 2.42 ng/mL, and IGF-1 436 ng/mL. Scrotal sonography showed small right (15*10 mm) and left (15*15) testes. Karyotype analysis demonstrated an extra X chromosome (47,XXY) and ins(6;7)(q13:p22). By sequence analysis of exon 1 region of androgen receptor, we detected 22 CAG repeat (normally, 12-30). We diagnosed the patient as having Klinefelter syndrome. Echocardiography performed for chest pain revealed mitral valve prolapse. The patient was informed about genetic counselling and fertility. Testosterone was given for treatment of secondary sexual characteristics.

To our knowledge, our case is the first Klinefelter patient having ins(6;7)(q13:p22); however, its clinical implication is not precise yet.

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PRO1-Related Combined Pituitary Hormone Deficiency: Case Report

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Mutations of *PRO1* are the most frequent genetic defect in non-syndromic combined pituitary hormone insufficiency and are characterized by growth hormone (GH), prolactin, TSH, and gonadotropin deficiency.

A 3-year-and-3-month-old girl was referred with growth retardation and abnormal thyroid function tests. She was born