

patients were diagnosed in the newborn period. Median follow-up was 5 years (1-19 years). Follow-up period was longer than five years in 56%. About half of the families admitted knowing the terms DSD, ambiguous genitalia, indeterminate genitals, and intersex; however, only 2% preferred using DSD, 6% intersex, and 14% ambiguous genitalia. Fifty-two percent of the parents used a disease name in Latin addressing the disorder. Sixty-nine percent who were familiar with the name indeterminate genitals were diagnosed in the neonatal period ($p=0.046$). The clinic mostly involved in the management was related to referring the disease with a name in Latin ($p=0.024$) or as chromosomal abnormality ($p=0.048$).

Parents of DSD patients avoid using any word containing “sex” and prefer disease names in Latin instead. Direct translation and usage of new terminology may not achieve the desired result. Each country has its own social norms, local committees should be employed to develop proper terminology.

(P-05)

A Male Case of Aromatase Deficiency with a Novel *CYP19A1* Mutation

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Aromatase deficiency (AD) is a rare autosomal recessive disorder caused by *CYP19A1* gene mutations and is characterized by lack of conversion of androgens to estrogens. Men usually present with continuing linear growth after puberty, tall stature, unfused epiphyses, delayed bone age, genu valgum, decreased bone mineral density, obesity, dyslipidemia, liver steatosis, insulin resistance, and impaired fertility. We here report a male case of aromatase deficiency with a novel *CYP19A1* mutation.

A 30-year-old man with a tall stature (192 cm) presented with genu valgum. He complained to grow continuously. X-ray revealed incompletely fused epiphyses. Bone age was compatible with 14 years. Follicle-stimulating hormone and luteinizing hormone and testosterone were in normal ranges, but estradiol was undetectable. Insulin resistance as well as elevated serum alanine aminotransferase, aspartate aminotransferase and

gamma-glutamyl transferase levels were found. Abdominal ultrasonography revealed steatohepatitis. In bone mineral density analysis, Z score was normal. The sperm count and vitality were normal. Sequencing of the *CYP19A1* gene revealed a novel 6-base homozygote deletion in exon 10 (c.1465_1470del GAAATG). The parents and sister were heterozygous for the same mutation. Estrogen replacement therapy was started.

We report a male patient with AD who had a novel deletion in *CYP19A1* gene. AD is an extremely rare condition. Till recently, all mutations have been in coding exons, mostly in exons 9 and 10. Estrogen replacement in AD has great impact on the recovery of dysplastic bone, lipid, liver, and glucose metabolism, but fails to improve insulin resistance. This will hopefully clarify the link between the deletion and the phenotype.

(P-06)

CYP11A1 Mutations Result in Various Clinical Phenotypes

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Cytochrome P450 side-chain cleavage enzyme (*CYP11A1*) is the first enzyme and catalyzes the rate-limiting step of steroidogenesis. *CYP11A1* deficiency is associated with adrenal insufficiency (AI) and commonly with a disorder of sex development (DSD) in 46,XY individuals. Our objective was to define the clinical presentation of our patients with *CYP11A1* mutations, one of whom had a novel *CYP11A1* mutation.

Four patients were presented. Case 2 has been reared as a girl and she has a novel *CYP11A1* mutation. Cases 3 and 4 are siblings. Clinical findings are given in Table 1.

These cases demonstrate that *CYP11A1* deficiency can be seen in the newborn period or in early childhood as classical or non-classical forms. Normal genital appearance can found in 46,XY patients in non-classic form and this does not exclude life-threatening AI risk.