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.

Table 1. Clinical findings

	Case 1	Case 2	Case 3*	Case 4*
Age at diagnosis, year	1.24	0.08	5.16	2.64
Karyotype	XX	XY, t(4;9)(p16.6?;p13.3)	XY	XY
Birthweight, g/gestational weeks	3600/39	1750/33	2200/39	2800/39
Parents	1. cousin	1. cousin	Same region	Same region
Presentation	Adrenal crisis	Adrenal crisis	No symptom	Adrenal crisis
Length/Height, cm (SDS??)	72 (-1.83)	44 (-6.05)	105 (-1.10)	95 (0.96)
Weight, g (SDS??)	8000 (-2.65)	1675 (-4.67)	18.6 (-0.10)	11.5 (-1.55)
External genitalia	Labial synechiae	Normal female	Penis 6x1.8 cm	Penis 5x2 cm
Adrenal imaging	Normal (MRI)	Hyperplasia (MRI)	Normal	Normal
Basal cortisol, µg/dL	< 1	8.15	7.6	9.2
Stimulated cortisol, µg/dL	< 1	8.03	7.8	9.4
Adrenocorticotropic hormone, pg/mL	259	1250	>1250	>1250
Progesterone, (ng/mL, N: < 30)	1.4	0.03	< 0.1	< 0.1
DHEAS, (µg/dL, N: 50-500)	4.2	16.41	48.5	30.7
17-OHP, ng/mL	0.7	0.56	0.34	
1.4 Androstenedione, ng/mL	0.18	1.2	0.33	0.33
Testosterone, (ng/mL)	0.3	0.02	< 0.13	< 0.13
Aldosterone, (ng/mL, N: 35-410)	< 1	33	1.3	0.16
Renin, pg/mL (N: 5.2-33.4) PRA (N:0.98-4.18)	> 500 -	> 520 -	-	- 19.43
CYP11A1 mutation	p.R451W	p.W152X	p.R451W	p.R451W

(P-07)

The Role of Adenovirus Serotype 36 in Childhood Obesity

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This study aimed to determine the role of Adenovirus 36 (Adv 36) in childhood obesity and to evaluate the obesity-triggering effect of its latent infection on adipose tissue.

The study group was composed of 31 obese children who were admitted to the pediatric endocrinology outpatient clinic, while the control group comprised 30 non-obese children without any chronic disease. In obese children, both an adipose tissue sample and blood samples were obtained, while only blood samples were obtained in control subjects. The adipose tissue samples were taken by a needle aspiration procedure from the subcutaneous tissue of abdomen in obese children. Besides biochemical tests, Adv 36 specific antibody and viral DNA in blood samples were investigated in all subjects, while viral nucleic acid with real-time PCR from adipose tissue was investigated only in obese subjects.

SGPT, triglyceride, and insulin levels were higher in the obese group. There was no case with a positive result of Adv 36 antibody in the control group, while the seropositivity rate for Adv 36 was 13% among the obese children. Regarding the latent Adv 36 infection, there was no positive PCR result from the adipose tissue samples in obese children.

There was a high serological evidence of Adv 36 infection in obese individuals. However, the results of PCR in adipose tissue could not show the presence of latent infection among obese children in the current study. Thus, further studies are needed to evaluate the possible associations between Adv 36 and development of childhood obesity.