is accompanied by decreasing BTM concentrations after 24 months of CSHT. Therefore, the added value of evaluating BTMs seems to be limited and DEXA-scans remain important in follow-up of transgender adolescents.

KEYWORDS: transgender adolescents, gonadotropin-releasing hormone analogues, bone mineral density.

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DIFFERENT FORMS OF CONGENITAL ADRENAL HYPERPLASIA IN TWO SIBLINGS IN A FAMILY: A CASE REPORT

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Introduction. CAH — is a group of diseases with autosomal recessive type of inheritance. 21-hydroxylase deficiency is responsible for 95% of all cases of CAH. Depending on the severity of 21-hydroxylase deficiency the disease is divided into three forms: salt wasting, simple virile, and nonclassic. If both parents are known to be heterozygous carriers of pathogenic genes, each sib has only 25% chance of being affected. **Clinical case.** A 7-day-old female girl was referred to our hospital with ambiguous genitalia. According to the medical history, she was born at term to a 28-year old healthy mother from her second gestation with a spontaneous delivery without any complications. Birth weight was 3290 g. Genital examination revealed clitoromegaly, single urogenital onfice, posterior labial fusion. Karyotype analysis showed normal female karyotype 46XX. Biochemistry revealed hyponatriemia (Na 131 mmol/l), hyperpotassemia: (K 6,55 mmol/l). Blood hormone analysis showed increased levels of 17-hydroxyprogesterone (811 ng/mL) and dehydroepiandrosterone (989,8 mcmol/l), hypocortisolemia (69,6 nmol/l).These results suggested a salt wasting form of CAH. In the gene analysis of CYP21 heterozygous mutations IVS2-13A/C>G and 30-kb deletion were detected.Replacement treatment, including hydrocortisone at the dose of 41 mg/m^2/day and fludrocortisone at the dose of 0,15 mg/day was initiated. The dose of hydrocortisone was gradually decreased to 24 mg/m^2/day. On the therapy the child showed positive dynamics of electrolytes levels, the general status was compensated. Weight at the age of 37 days was 4060 g (meant weight 4090 g.).During collection of the family history the baby's mother marked special features of her older son. By the time of sister's birth the boy was 2 years 8 months old. The parents reported high velocity of growth since birth and acne after 2 years of age. Laboratory investigation showed high level of 17-hydroxyprogesterone (299,8 ng/l) and testosterone (12,6 nmol/l). The boy was admitted to the hospital. Physical examination revealed acne on the face and upper back, penile enlargement, pigmentation of the scrotum, though both testis were prepubertal in size. Height was 106 cm (> 97th percentile). Bone age was 6 years 10 months. His predicted height (159 cm) was significantly lower than genetic one (177 cm). Levels of blood electrolytes were normal. A diagnosis of virile form of CAH was considered. Hydrocortisone treatment at the dose of 13,3 mg/m^2/day was initiated. The boy showed a compound heterozygous mutation (IVS2-13 A/C>G and 30-kb deletion). **Conclusion.** Although the sibs had similar mutations, they exhibited different phenotypes. According to the literature, presence of IVS-2 mutation may determine both salt wasting and simple virile forms. It might result from the variable splicing of this mutation due to variation in RNA splicing factors.

KEYWORDS: congenital adrenal hyperplasia, 17-hydroxyprogesterone, hydrocortisone treatment.

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INDICATORS OF RESPIRATORY MITOCHONDRIAL FUNCTION IN DIABETES MELLITUS

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Changes in the level of mitochondrial dehydrogenases in the respiratory cells in patients with diabetes mellitus had been found in the earlier studies. However, the diagnostic criteria for verification of energy disturbances in the respiratory system in subjects with type 1 diabetes are not established. **The aim** of the study was to perform the cytochemical analysis of mitochondrial function in patients with diabetes. A total of 116 Caucasian subjects were recruited and studied: 57 person with type 1 diabetes, aged 54.2±1.3 years and 59 participants without diabetes, aged 47.8±3.5 years. Those with the history of respiratory disease and smoking history were specifically excluded. Cytochemical analysis was performed by analyzing the activity of succinate dehydrogenase (SDH) and lactate dehydrogenase (LDH) using computer morphometry. The substrate for the study was the bronchoalveolar secret. The viability of epithelial cells and alveolar macrophages of the bronchi was significantly decreased in patients with type 1 diabetes compared to control group — 49.6±1.5% and 73.2±2.8% vs 57.6±1.9% and 85.3±2.7%, respectively ($p<0.001$). Phagocytic number and phagocytic index was also decreased in those with type 1 diabetes compared to controls — 39.4±1.7% and 7.1±0.4% vs 48.8±1.3% and 8.7±0.3%, respectively ($p<0.05$). The levels of mitochondrial activity SDH and LDH in patients with type 1 diabetes were 12.4±0.9 and 11.5±0.9 standard units and in the control group — 19.8±0.7 and 23.6±1.1 standard units ($p<0,01$). In subjects with diabetes it was the negative correlation between the activity of SDH and LDH of the cellular elements of the respiratory system and hyperglycemia and of index endobronchitis activity, with $r = -0.39$ ($p=0.003$) and $r=-0.29$ ($p=0.03$) and $r=-0.53$ ($p=0.02$) and $r = -0.39$ ($p=0.01$), respectively. We can speculate that the level of dehydrogenase activity may serve as a diagnos-