

## CLINICAL CASE OF CONGENITAL ADRENAL HYPERPLASIA

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**Introduction.** Congenital adrenal hyperplasia (CAH) is the only one of all the various causes of intersex that can actually cause a medical emergency. In fact, before CAH was well understood, it was thought to occur much more frequently in girls. Now we know that this really reflects the fact that newborn girls with genital ambiguity often came quickly to medical attention, whereas CAH boys (who don't have an intersex appearance) often died without being diagnosed. Another problem that can happen is called "Salt wasting." Some people with CAH (whether intersexed or not) don't produce the right levels of hormones that control salt in the body. These people can get very sick, and even die, without medication to correct their body's salt level.

**Results of our research.** As a clinical example we show case history of a girl 4 mo. From anamnesis is known that the child was born from the second pregnancy. The child was born with birth weight –3350 g, body length –55cm. Hypertrophy of the clitoris (III stage of Prader ), hyperpigmentation of nipples, vulva were found at birth baby. In checking the child's looks stable electrolytes: potassium – 4gr./l, sodium-145 gr./l, chlorine – 111.6gr./l (the following parameters were observed on the fourth day). Levels of serum of 17OHP – 18.9 nmol/l (0.7–2.5–normal), testosterone more than 1600 ng/ml (normal–20.0–1148), cortisol-5.07nmol/l (normal–7.4–40.0). Clinically – shock and dehydration. On the second week of life condition of patient changed: there were observed electrolyte disorders – potassium – 6.0gr./l, sodium – 126.9gr./l, chlorine – 93.3gr./l Diagnosis : Congenital adrenal hyperplasia; Salt losing form; State of decompensation; Delayed physical development; Subnanism; Underweight (BMI – 14.7); Anemia deficiency I degree. Child was hospitalized into the hospital with following complaints: short stature and poor appetite. From 4 months of age the child is marked with physical retardation, inadequate weight gain, blood electrolytes: 17–OHP level is normal in the blood but besides of it being normal the child still has retardation (17–OHP is being inspected once in every two months).

Objectively: length – 67 cm (– 2.5 sigma), weight – 6500gr, BMI-14.7 (less than 3 percentile), surface of the body – 35cm<sup>2</sup>. Besides this the patient hyper pigmentation, virilization of the external genitalia, clitoral hypertrophy are marked. CBC: Hb – 104g/l, erythrocytes – 3.1T/l, leukocytes – 6.9G/l, ESR-2mm/hr, stab-0%, segment – 10%, eosinophils – 0% ,blood glucose-3.5mmol/l. Biochemical blood test : total protein – 55.1g/l, albumin – 47.6%, creatinine – 58mkmol/l, urea – 4.0mmol/l, total bilirubin – 8.6mcromol/l, direct-0mcromol/l, indirect-8.6mcromol/l, ALT-0.32, ACT-0.26, cholesterol – 4.9mmol/l. ECG-sinus rhythm, irregular heart rate – 187–130 BPM. US of abdominal cavity organs: liver without pathological lesions; gallbladder pear shaped ,sizes 1ml, the wall is not sealed , stones are not found. Pancreas – not increased echogenesity usually; spleen 44 – 18–12, usually echogenecity, homogeneous structure. Kidneys: the shape and position is normal. Adrenal glands – not increased in size, the structure is homogeneous presence of distinct border between the cortical and medullar layer is observed . Echogenesity is normal. US of the thyroid: right lobe – 0.2ml, left lobe – 0.2ml, echogenecity and structure is homogeneous, no pathological change on the brain. X-ray of hand bone : age corresponds to passport age. Treatment: individual diet (breastfeeding + required foods), hydrocortisone 5.0–2.5–2.5mg, kortynef 0.05–0.025mg.

**Conclusions:** 1:congenital adrenal hyperplasia is less common pathology of childhood. 2. Syndrome diagnosis requires a comprehensive evaluation of clinical, biochemical parameters.