

## **WHEN A 13 YEAR-OLD BOY IS NOT A BOY? A LATE CLINICAL PRESENTATION OF CONGENITAL ADRENAL HYPERPLASIA**

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Congenital adrenal hyperplasia (CAH) is the most common cause of genital ambiguity in the newborn. 90-95 % of CAH cases are caused by 21-hydroxylase deficiency. These infants can survive despite lack of treatment. However, both girls and boys are subjected to progressive masculinization.

A 13 year-old patient who reared as male presented with cryptorchidism. The patient had been diagnosed as hypospadias and cryptorchidism by a urologist and referred us for hormonal evaluation. On clinical examination; axillary and pubic hair corresponding to Tanner Stage 5, bilateral cryptorchidism, chordee, labiascrotalization, external urogenital orifice at penoscrotal region, penil stretched length: 7cm. Laboratory data; high level of serum 17-hydroxyprogesterone, testosterone, dehydroepiandrosterone sulphate, adrenocorticotrophic hormone and low level of serum cortisol. Bone age was 16.5 years, pelvic ultrasonography revealed hypoplastic uterus and ovaries. Urogenitography showed urogenital sinus and vagina. Karyotyping: 46, XX. Simple virilizing form 21-hydroxylase deficiency was diagnosed and hydrocortisone therapy was started. The family was informed and recommended virilizing genitoplasty by the multidisiplinary team. The patient and his parents approved the decision, male sex assignment was maintained. Two years after hydrocortisone therapy, he is underwent testosterone therapy.

Patients with CAH have a risk of being diagnosed late. The role of social structure may be important (in some rural area of our country, there is a tendency appreciating male children). Still, there are major problems and uncertainties concerning gender assignment of the patients' families and physicians.

**Presentation Type:** Poster Presentation

**Authorize Publishing:** Yes

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