A neonatal course of prenatally treated congenital adrenal hyperplasia

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Objective

Congenital adrenal hyperplasia is an autosomal recessive disorder due to steroid 21-hydroxylase deficiency (CAH) that affects about 1 in 14, 500 newborns. Affected females typically have virilized external genitalia. Maternal administration of dexamethasone in prenatal period can prevent or ameliorate the genital virilization of female infants but fetal therapy is controversial. The fetus is estimated that receives about 60 times its usual physiologic glucocorticoid exposure. Herein, we aimed to present a successfully treated case of CAH with maternal dexamethasone in fetal period.

Methods

The maternal and neonatal records of patient diagnosed with fetal congenital adrenal hyperplasia were reviewed retrospectively.

Results

A 34 year old mother, who had two girls with CAH previously, admitted to high risk pregnancy outpatient clinic. After comprehensive counselling of the possibilities about pregnancy and fetus, informed consent for the fetal therapy were obtained from the patient and her family. Prenatal treatment started during the 6th week of pregnancy. 20 microgram/kg/d dexamethasone was administered to the mother. Fetal karyotype was determined as 46, XX and the treatment was not interrupted until term. Dexamethasone was well tolerated during the pregnancy period, weight gain and blood pressure was in normal range. The pregnancy follow up visits were uneventful. A 2970 gr female newborn was delivered by vaginal delivery with 9/10 Apgar score. In physical examination, there was no finding of virilisation of external genitalia (Prader stage 1). Pelvic ultrasonography showed normal Mullerian structures. Hormonal findings confirmed the diagnosis of CAH. There was CYP21A2 mutation on exon 6 in genetic analysis. At postnatal 23th day, she was admitted to neonatal intensive care unit with acute adrenal crisis. She had vomiting and dehydratation. In biochemical studies, there were hypoglisemia and hyponatremia. After treatment with iv fluid, high dose hydrocortisone and fludrocortisone; she was discharged with oral maintenance medical treatment and scheduled follow up visit.

Conclusion

Prenatally administering dexamethasone to the mother, suppresses the overproduction of adrenal androgens in female fetuses. Success rate for ameliorating genital virilization is high and there is a significant reduction for genital reconstriction surgery requirement after fetal therapy. Long term outcomes of these neonates is unknown and teratogenic risks and neurocognitive impairment was demonstrated in animal models. Prenatal treatment decision should be implemented cautiously only after detailed counseling of parents about the possible neurocognitive and teratogenic effects and anatomic gaints of the therapy.