LIPOID ADRENAL HYPERPLASIA DIAGNOSED WITH SEVERE CHOLESTASIS IN NEWBORN

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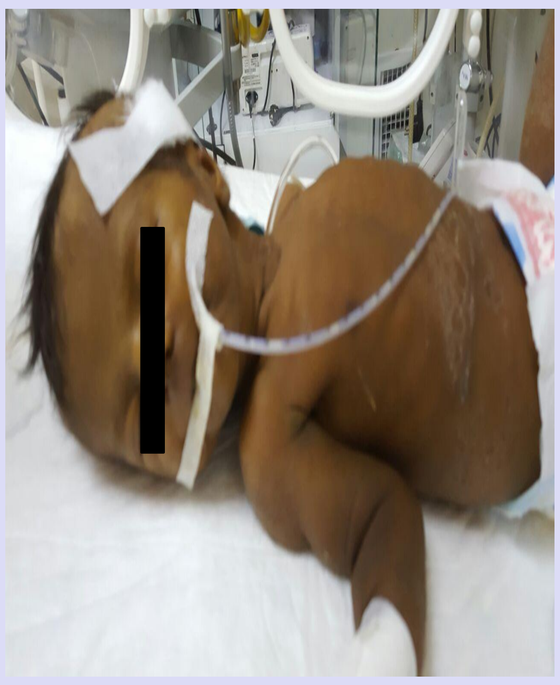
INTRODUCTION

Congenital lipoid adrenal hyperplasia is the most severe form of congenital adrenal hyperplasia and is rarely seen. Steroid synthesis cannot be done in the adrenal gonads. Adrenal glands have hyperplasia and lipid accumulation. Male babies are born to girls. Most of the cases are lost with severe adrenal insufficiency. Patients diagnosed and treated at supraphysiological doses during neonatal period

Here we present a case of lipoid adrenal hyperplasia diagnosed with severe cholestasis, hyponatremia and hyperpotassaemia. In this case phenotype was female and the testis was palpated in the inguinal canal.



CASE

A 26-day-old baby was transferred to our clinic with the doubt of adrenal insufficiency. Her parents were first-degree cousins. She was born in term. There was nutritional intolerance. Phenotype was girl's appearance and two-handed testicles in the inguinal region. There was extensive hyperpigmentation throughout the body. A complete blood count was normal. **Direct bilirubin: 18 mg / dl, total cholesterol: 371 mg / dl, LDL cholesterol: 12 mg / dl**, **Na: 126 meq / l**, **Triglyceride: 230 mg / dl**, **ACTH: >1250 pg/ml**, **Cortisol: 1,63 ug / dl**, LH: 1,49 mU / ml, Progesterone 1,15 ng / dl, Testosterone 24 ng / dl, Aldosterone 49 pg / ml. Pelvic ultrasonography revealed no uterus or over. Karyotype analysis was 46 XY. Congenital lipoid adrenal hyperplasia was suspected. Hydrocortisone and fludrocortisone treatment were administered at high doses. The cholestasis of the patient was also recovered when the adrenal crisis resolved. The homozygous mutation in the star gene was detected in the genetic screening. The patient was diagnosed with congenital lipoid adrenal hyperplasia.

DISCUSSION

STAR protein regulates the passage of the cholesterol in the mitochondria. In the lack of this protein there is no flow of steroidogenesis because of insufficient cholesterol transmission. Cholesterol accumulates in adrenals and gonads. The phenotype of 46 XY genotype patients is girl. Severe salt loss is accompanied by this disease. Late onset forms will also accumulate lipid over time, resulting in salt loss. There was severe salt loss in our case. Steroid treatment at high doses improved the patient's salt loss. This case is unique presentation because of lipoid adrenal hyperplasia associated with cholestasis. We have improved cholestasis with steroid therapy, and severe salt loss clinic has been corrected with long-term high-dose hydrocortisone and fludrocortisone treatment.

CONCLUSION

46 XY congenital lipoid adrenal hyperplasia is a disease that should be considered in the newborn with signs of sexual dysfunction and adrenal insufficiency. In these cases cholestasis may be problem. These patients can survive at supraphysiological doses with hydrocortisone and fludrocortisone treatment.

References

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