A NEONATAL CASE WITH FAMILIAL GLUCOCORTICOID DEFICIENCY TYPE 1 HAVING ADRENAL CRISIS IN EARLY PERIOD

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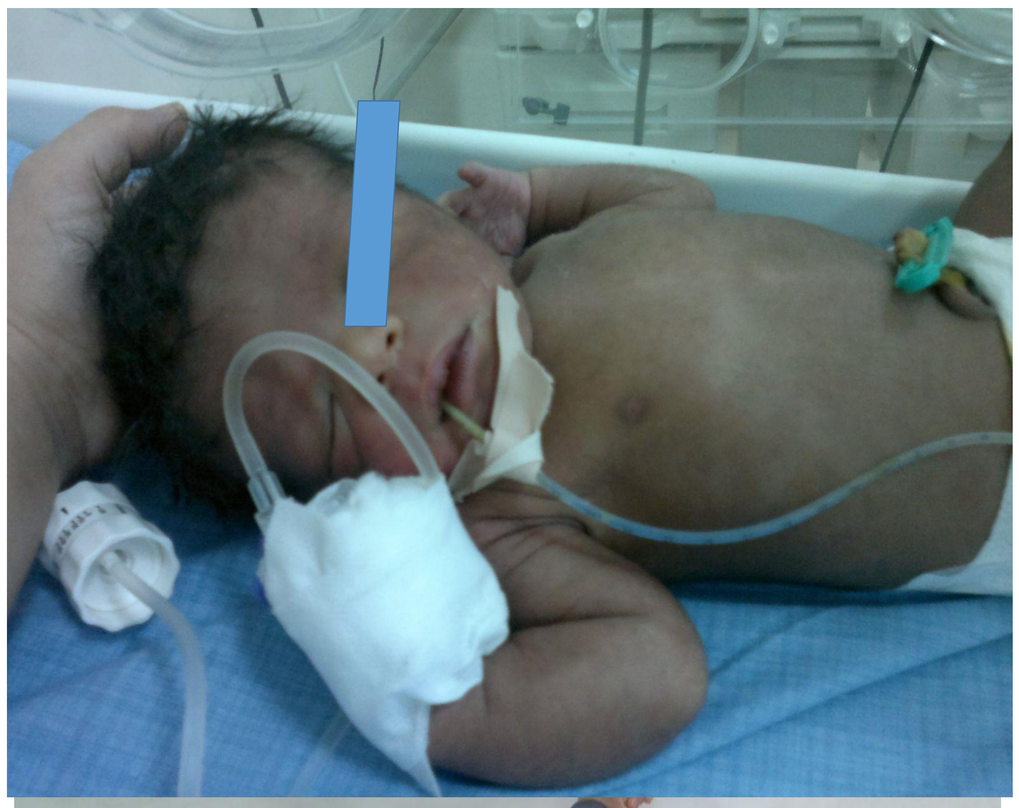
OBJECTIVE

Familial glucocorticoid deficiency (ACTH resistance) is a rare chronic adrenal insufficiency problem. Genetic tranmission is autosomal recessive. Glucocorticoid deficiency is characterized by increased ACTH levels and normal or partial incomplete aldosterone production. The familial glucocorticoid deficiency, which is a defect in the melanocortin receptor. Hypoglycemia, convulsions, increased pigmentation in the skin can be seen from the earliest stages of life.

CASE

Forty day old baby with postnatal hypoglycemia, hyponatremia, hyperkalemia, intensive hiperpigmentation, convulsions due to adrenal insufficiency was forwarded to starting treatment. Parents were relative and their first baby with similar findings was lost at the first day. On physical examination, there was severe hyperpigmentation. Hydrocortisone and fludrocortisone was continued in appropriate doses. Fludrocortisone was cut off on follow-up. The color began to opening. The genetic analysis showed that familial glucocorticoid deficiency type 1 with MC2R gene homozygous deletion of the entire gene.



CONCLUSION

In this study; we showed that treatment with familial glucocorticoid deficiency type 1 must be quickly and effectively because of adrenal insufficiency and hyperpigmentation develop very early and rapidly in neonatal period.

Authors declare no conflict of interest.