CASE REPORT

Familial Glucocorticoid Deficiency Presenting with Skin Hyperpigmentation: A Case Report

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Abstract:

Familial Glucocorticoid Deficiency (FGD) has high morbidity and mortality, if not diagnosed and managed in time. The patient is liable to have hypoglycaemia which could be complicated by seizure and brain damage. Also these patients if not treated appropriately; will have high risk of infections and failure to thrive. We report a case of FGD baby of full term, male, birth weight 3 kg and born by uneventful normal delivery. On the second day of life, the baby had hypoglycaemia and later he developed mucosal membrane and skin hyperpigmentation. Critical sample during the hypoglycemic episode showed low serum cortisol, high adrenocorticotropic hormone level, normal serum electrolytes and normal kidney function. A rare familial glucocorticoid deficiency was diagnosed in time by doing appropriate investigations; includes critical sample during hypoglycaemia and the case was managed successfully by hydrocortisone 5 mg orally once daily.

Keywords: Familial Glucocorticoid Deficiency, Adrenocorticotropic Hormone, Cortisol

Introduction:

Newborn period is a very critical time, if the baby receives appropriate amount of fluid or feeding but still has hypoglycaemia, meaning there is a serious problem that should be investigated as soon as possible. One of the rarest disorders causing neonatal hypoglycaemia is FGD which is autosomal recessive disorder. The incidence of Familial Glucocorticoid Deficiency (FGD) [1-2] is unknown, limited number of FGD around the world is reported. FGD causes low level of cortisol and corticosterone, patients at risks for recurrent hypoglycaemia, infections and inappropriate weight gain. Patients could have blackish discoloration of skin and mucosal membranes [3] due to high Adrenocorticotropic Hormone (ACTH). MC2R, MRAP and NNT genes [4-6] are three known point genetic mutation responsible for the majority of cases.

Case Report:

Parents were first cousins and they had no family history of adrenal gland disorder. The baby was male, he was delivered at full term via normal vaginal delivery. The Apgar scores were seven and nine at one and five minutes, respectively. His weight at birth was three kilograms. Physical examination was normal. Baby was admitted to Shifa Neonatal Intensive Care Unit (NICU) at the age of 1 hour due to tachypnea; the oxygen saturation in room air was 95-98%. Septic workup was negative. At the age of 2 days, baby was fed by bottle and blood sugar was 40 mg/dl before the feeding. Critical sample was taken at 8 pm during hypoglycemic episode that showed: glucose 32 mg/dl, cortisol 100 ug/dl {normal range am (500-2500) ug/dl; pm (250-1250) ug/dl}, C-peptide 7.2

ng/ml (normal range 0.5-5.0 ng/ml), insulin level 59 mcIU/ml normal range (3-35) mcIU/ml. Baby maintained normoglycemia when kept to mother's milk and intravenous fluid 10% dextrose. Since the age of 4 days baby had hyperpigmentation of skin and mucosal membranes (Fig.1). Repeated endocrine investigation at 8 am showed: Cortisol 120 ug/dl{normal range am (500-2500) ug/dl; pm (250-1250 ug/dl}, 17-OH progesterone 2.1 ng/dl{normal range; male (0.7-3.6) ng/dl; female (0.1-4) ng/dl}, ACTH 3040 pg/ml (normal range 25-65 pg/ml), serum sodium:141 meq/l, serum potassium:4 meq/l. Abdominal and brain ultrasound were normal. Echocardiography showed small ASD, small VSD and small PDA. Baby was discharged home at the age of 10 days in good general condition and daily oral 5 mg Hydrocortisone was prescribed. (Fig. 2) showed the baby at the age of 4 months, he had appropriate growth for age and no more hyperpigmentation of mucosa or skin. At the age of 5 months serum electrolytes; serum sodium: 137 meq/l, serum potassium: 3.9 meq/l, serum calcium: 9.5 meq/l, serum ACTH:1572 pg/ml (normal range 25 - 65 pg/ml), C –peptide: 0.98 ng/ml (normal range 0.5-5.0 ng/ml).

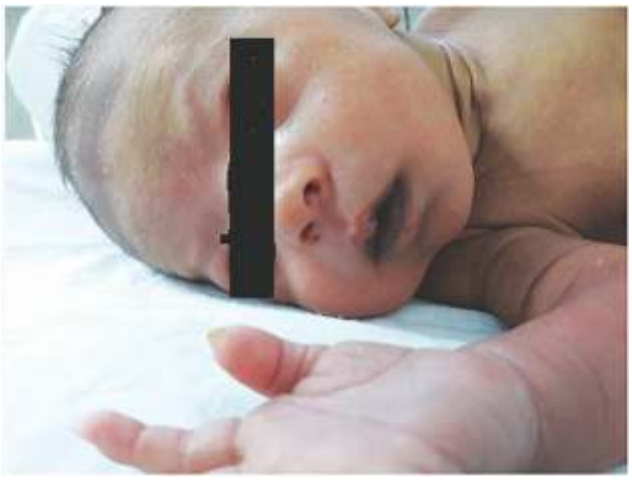

Fig. 1: At the Age of 4 days, Baby had Hyper-pigmentation of Skin and Mucosal Membranes

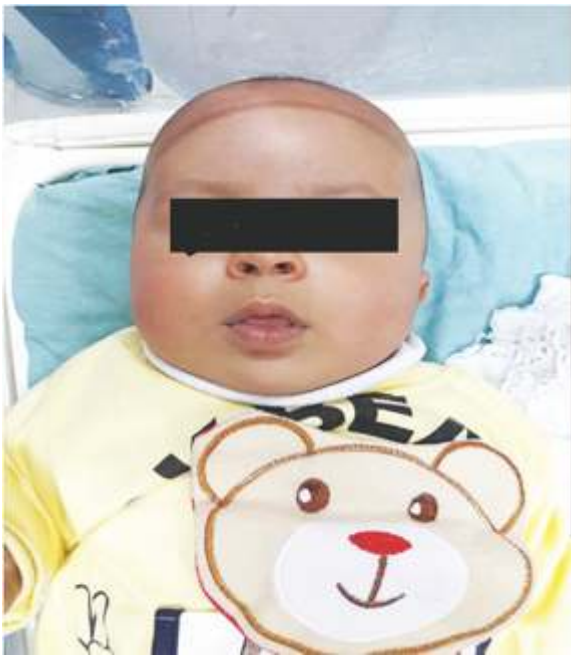

Fig. 2: Baby at the Age of 4 Months on Oral Hydrocotisone 5 mg Once Daily Growth Was Appropriate for Age and No More Hyperpigmentation

Discussion:

FGD has high morbidity and mortality if not diagnosed and treated early. Our case was diagnosed as early as the case was noticed to have hypoglycaemia and hyperpigmention of mucosal membranes and skin. Hormonal profile and serum electrolytes were diagnostic of FGD. Serum electrolytes including serum sodium and potassium were normal. ACTH hormone was so high and serum cortisol was low. These laboratory results excluded congenital adrenal hyperplasia and Addison's disease as serum electrolytes were normal. The baby was early diagnosed as a FGD case [7-9]; he had no manifestation other than mild hypoglycaemia and hyperpigmentation [10]. The baby was discharged home on oral hydrocortisone 5 mg orally once daily. Repeated investigations after 5 months of age showed significant drop of ACTH hormone, serum electrolytes were normal and baby had normoglycemia.

Conclusion:

A rare case familial glucocorticoid deficiency was diagnosed in time by doing appropriate investigations; includes critical sample during hypoglycaemia and the case was managed successfully by hydrocortisone 5 mg orally once daily

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