

Diffuse Hyperpigmentation of the Skin in a Newborn

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Familial glucocorticoid deficiency is a rare cause of primary adrenal insufficiency characterized by glucocorticoid deficiency in the absence of mineralocorticoid deficiency. Patients with familial glucocorticoid deficiency are usually diagnosed during the neonatal period when they present with generalized hyperpigmentation of the skin and/or hypoglycemia. They also may present with infections, failure to thrive or collapse, and coma.^{1,2}

Neonatal hyperpigmentation can be categorized as localized, or more rarely generalized.³ Diffuse or generalized hyperpigmentation in the neonatal period is rare. Differential diagnoses of generalized hyperpigmentation in newborns include metabolic causes (hyperthyroidism, hypothyroidism, acromegaly, and Addison's or Cushing's syndrome) and chronic hepatic or renal disease.³ The combination of hyperpigmentation, low cortisol, and high adrenocorticotrophic hormone levels with normal mineralocorticoid production is characterized by isolated glucocorticoid deficiency. Here, we present a newborn with isolated glucocorticoid deficiency presenting with generalized hyperpigmentation.

A male baby was born at 31 weeks with a birth weight of 970 g to non-consanguineous parents. Gradual hyperpigmentation of the skin was noticed during follow-up. He had normal male genitalia. Unlike his fair-skinned parents, he had generalized hyperpigmentation of the entire body and the oral mucosa (Figure 1A). Laboratory studies showed a glucose level of 52 mg/dL (N, 50–100 mg/dL), sodium level of 129 mEq/L (N, 135–145 mEq/L), potassium level of 5.4 mEq/L (N, 3.5–5.5 mEq/L), cortisol level of 2.7 mcg/dL (N, 3–19 mcg/dL), ACTH level of >1250 pg/mL (N, 15–60 pg/mL). Arterial blood gas, serum 17-OH progesterone, plasma renin activity, and aldosterone levels were in the normal range. Magnetic resonance imaging of the adrenals showed normal anatomy. Based on this data, a diagnosis of familial glucocorticoid deficiency (molecularly not confirmed) was made, and oral hydrocortisone treatment (12 mg/m²/day) was started (1). The hyperpigmentation was resolved after 2 months (Figure 1B).

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Figure 1. (A) Diffuse hyperpigmentation of the skin. (B) Resolution of hyperpigmentation.

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