Turkish Archives of Pediatrics

SCIENTIFIC LETTER
DOI: 10.5152/TurkArchPediatr.2022.22031

Diffuse Hyperpigmentation of the Skin in a Newborn

Ayşe Anık[1], Reyhan Deveci Sevim[2], Ahmet Anık[2]

[1]Division of Neonatology, Department of Pediatrics, Aydın Adnan Menderes University, Faculty of Medicine, Aydın, Turkey
[2]Division of Pediatric Endocrinology, Department of Pediatrics, Aydın Adnan Menderes University, Faculty of Medicine, Aydın, Turkey

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 adrenocorticotropic 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/m^2/day) was started (1). The hyperpigmentation was resolved after 2 months (Figure 1B).

Corresponding author:
Ahmet Anık
✉ahmet.anik@yahoo.com
Received: February 3, 2022
Accepted: February 21, 2022

Cite this article as: Anık A, Deveci Sevim R, Anık A. Diffuse hyperpigmentation of the skin in a newborn. *Turk Arch Pediatr.* 2022;57(3):370-371.

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

Informed Consent: Written and verbal informed consent was obtained from the parents.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – A.A., R.D.S., A.A.; Design – A.A., R.D.S., A.A.; Supervision – A.A., R.D.S., A.A.; Resources – A.A., R.D.S., A.A.; Materials – A.A., R.D.S., A.A.; Data Collection and/or Processing – A.A., R.D.S., A.A.; Analysis and/or Interpretation – A.A., R.D.S., A.A.; Literature Review – A.A., R.D.S., A.A.; Writing Manuscript – A.A., R.D.S.; Critical Review – A.A.

Declaration of Interests: The authors have no conflict of interest to declare.

Funding: The authors declared that this study has received no financial support.

REFERENCES

1. Chan LF, Clark AJ, Metherell LA. Familial glucocorticoid deficiency: advances in the molecular understanding of ACTH action. *Horm Res.* 2008;69(2):75-82. [CrossRef]
2. Kirkgoz T, Guran T. Primary adrenal insufficiency in children: diagnosis and management. *Best Pract Res Clin Endocrinol Metab.* 2018;32(4):397-424. [CrossRef]
3. Jacoby E, Barzilai A, Laufer J, et al. Neonatal hyperpigmentation: diagnosis of familial glucocorticoid deficiency with a novel mutation in the melanocortin-2 receptor gene. *Pediatr Dermatol.* 2014;31(1):e13-e17. [CrossRef]