

Abstract Guidelines

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SEVERE HYPERNATREMIC DEHYDRATION IN AN INFANT WITH NETHERTON SYNDROME

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Autosomal recessive congenital ichthyosis (ARCI) is a group of inherited disease of cornification in which progress has recently been made in the identification of pathogenic mechanisms causing the disorder. Transglutaminase 1 (TGM 1) has been found as a defective gene in a large fraction of patients with lamellar ichthyosis. More recently the mutation of SPINK 5 was described in the Netherton syndrome. Netherton syndrome is a rare ARCI characterized by ichthyosis and the characteristic hair abnormality trichorrhexis invaginata. We report a patient with the severe hypernatremic dehydration form of the Netherton syndrome.

Infant G. was the first child of consanguineous parents. Ichthyosis was present at birth. He was admitted to intensive care at the age of 4 days for an important loss of weight and dehydration. Severe hypernatremia and convulsions occurred. Despite intensive care the baby died at the age of 11 days. The diagnostic of Netherton syndrome was confirmed by the pathognomonic hair shaft anomaly, trichorrhexis invaginata (bamboo hair). Skin biopsy showed premature lamellar body secretion and foci of electron-dense material in the intercellular spaces of stratum corneum which are relatively specific markers for Netherton syndrome. These abnormalities could explain the impaired permeability barrier in Netherton syndrome, and account for hypernatremia and dehydration in infants with the syndrome. Netherton syndrome is characterized by a large variability of phenotype expressivity. The major neonatal complication is the hypernatremic dehydration, which can be fatal as in this patient or complicated by neurologic signs (intracranial hemorrhage) and secondary sequellae.

Title: Capitals/boldface

Authors: Initials + surname in capitals/italics

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