Case Letter

# Consider hypertriglyceridemia in congenital ichthyosis

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Sir,

Chanarin–Dorfman syndrome (DCS) is a highly rare autosomal recessive inherited disorder caused by mutations in the ABHD5 gene (3p21) [1]. This results in the inhibition of the enzyme triglyceride lipase leading to lipid overload in various tissues, including the skin, muscle, liver, central nervous system, and blood leukocytes. This metabolic abnormality results in variable systemic manifestations such as congenital ichthyosiform non-bullous erythroderma, growth retardation, splenomegaly, steatohepatitis, cardiomyopathy, renal failure, sensorineural hearing loss, various ocular disorders such as cataract, nystagmus, retinal changes, and neurological impairment with proximal muscle weakness, areflexia, hypotonia, and mental retardation [1-3]. The muscle deficit remains subclinical in most cases, despite the elevation of muscle enzymes in more than 50% of cases. The diagnosis is based on the demonstration of Jordans bodies in leukocytes and muscles [1,2]. This syndrome is not too severe in the absence of neurological involvement [4]. The treatment of DCS is symptomatic and consists of the local application of emollients. A diet low in fatty acids and high in medium-chain triglycerides improves the cutaneous and hepatic manifestations [5]. Acitretin is useful in the treatment of skin and muscle manifestations with close monitoring of the lipid profile [3,5-7].

Herein, we report the case of a six-month-old infant from a consanguineous marriage presenting ichthyosis since birth and a history of a collodion baby. A general examination revealed a delay in the growth and development of the child. A dermatological examination revealed large, dark scales, not especially thick, over the entire body, sparing the large folds and face, with erythematous skin on the upper part of the trunk and the upper limbs (Fig. 1), associated with palmoplantar keratoderma (Figs. 2a and 2b), flattened ears, and ectropion. Biology revealed severe hypertriglyceridemia (3 g/L) rechecked on two occasions, creatine phosphokinase mb (CPK mb) five times the normal value, and CPK three times the normal value.

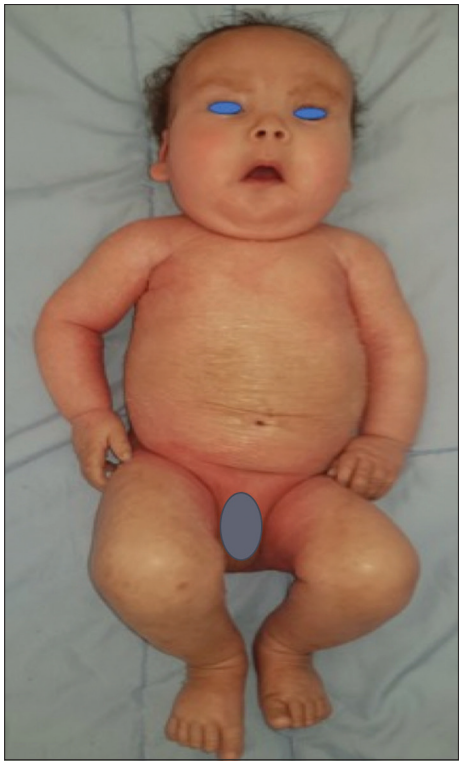

**Figure 1:** Ichthyosis with ectropion.

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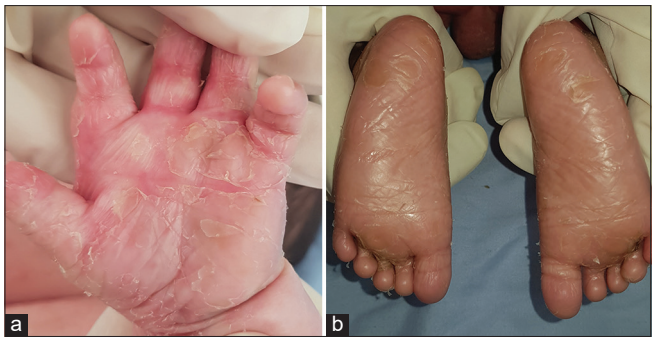


**Figure 2:** (a and b) Palmoplantar keratoderma.

Liver workup and renal function were normal, and a search for Jordans bodies in leukocytes was negative. An abdominal ultrasound was conducted, which revealed no evidence of hepatosplenomegaly or hepatic steatosis. However, due to insufficient resources, genetic tests could not be performed. Emollients were prescribed for the patient, who was subsequently lost to follow-up.

Our patient was diagnosed as a case of CDS based on the history of a baby collodion, congenital non-bullous ichthyosiform erythroderma with the elevation of muscle enzymes. Jordans bodies in leukocytes were negative, as was the case in an eight-year-old child reported by Arora [3]. Unfortunately, a mutation search could not be performed due to insufficient resources.

## Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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