GENETICS AND GENODERMATOSES

SHOULD WE RECOMMEND A PERIPHERAL BLOOD SMEAR TO PATIENTS WITH CONGENITAL ICHTHYOSIFORM ERYTHRODERMA?

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Background: Dorfman Chanarin syndrome is a rare syndromic ichthyosis which could mimic non bullous eythrodermic congenital ichthyosis. This diagnosis should be suspected in every patient with a history of congenital ichthyosis by performing a simple peripheral blood smear showing the Jordan's anomaly.

Observation: A patient aged 4 years born of a consanguineous marriage was referred to our department for a congenital ichthyosis. He was initially classified to have non bullous eythrodermic congenital ichthyosis. Physical examination revealed a generalized ichthyosis with fine white grayish scales on an erythematous background. Palms and soles showed hyperlinearity. He has a mild bilateral lower lid ectropion. Abdominal examination revealed hepatomegaly. Neurological examination was normal. There was no muscular weakness. Complete cell blood count was normal but the peripheral blood smear showed cytoplasmatic vacuoles in leukocytes. Liver function tests revealed elevated transaminases and mildly elevated serum alkaline phosphatase. Electromyogram was normal. In our patient, the diagnosis of Dorfman Chanarin syndrome was confirmed by a genetic analysis. A mutation analysis of the ABDH5 gene was performed revealing a mutation in intron 5 of ABDH5 gene in homozygous state in the patient.

Key message: This case is being reported to increase awareness of dermatologists to this rare syndromic ichthyosis since the prognosis of this entity and the non syndromic ichthyosis represented by erythrodemic congenital ichthyosis is not the same. A complete and careful history taking of similar cases of ichthyosis in the family, physical examination and the laboratory findings especially the peripheral blood smear are important as in our case to assess an appropriate diagnosis.