GENETICS AND GENODERMATOSES

ANTISKIN AUTOANTIBODIES IN TWO NEWBORNS WITH INHERITED EPIDERMOLYSIS BULLOSA: MAY BE NOT ONLY AN EPIPHENOMENON?

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Background: It was supposed that, in EB, the skin and mucosal damage depends on genetic factors but is aggravated by the activation of an immune process that could take, in certain cases, a major role in defining the severity of the clinical manifestations.

Observation: We would like to report the case of two newborns with inherited epydermolisis bullosa and anti skin autoantibodies.

Key message: Has been debated if higher levels of anti-skin antibodies are the consequence of the deterioration of the skin condition and are produced as a secondary event contributing significantly to the worsening of the disease, but our cases demonstrate as auto antibodies appears also in the first weeks and months of age and can be an early trigger of autoinflammation. Moreover, our report demonstrate how anti skin autoantibodies can correlate with the depth of detachment of the dermal epidermal junction and therefore with the epidermolysis bullosa subtype. In fact the patient with both anti collagen VII and anti MB has a dystrophic EB form, instead the girl with only anti MB autoantibodies was diagnosed for a junctional form. This can be explained because in the junctional form only lamina lucida anti-bodies are exposed while in the recessive EB subtype both collagen antigen VII and lamina lucida antigens are exposed. In our opinion, the early detection and a larger analysis of a skin autoantibodies in infants with different forms of inherited epydermolisis bullosa can confirm our results and maybe can be used as an predictive exam of EB subtype.