GENETICS AND GENODERMATOSES

TYPE I LEUCOCYTE ADHESION DEFICIENCY (LAD I): A FAMILIAR CASE.

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Backgroung: Primary immunodeficiencies(PIDS)are rare,inherited diseases,characterized by altered function or absence of immune cells.Among them is leukocyte adhesion deficiency type I (LAD-I),a rare autosomal recessive disorder characterized by primary immunodeficiency,caused by mutations in the ITGB2 gene and characterized by inability of leucocytes to migrate from the circulation towards the area of inflammation and associated recurrent and life-threatening bacterial infections. Pyoderma gangrenosum (PG) is an uncommon noninfectious neutrophilic dermatosis characterized by recurrent,sterile, necrotic skin ulcers.It is commonly associated with underlying systemic. Pathogenesis of PG remains unclear though aberrant immune responses have been implicated. The diagnosis of PG can be challenging and is of exclusion and management is empirical with local or systemic immunosuppressive therapy.Though skin ulcerations are common, predominant clinical presentation as PG can often mimic other disease.It is unusual in children even more in LAD-I.

Observation: Here we present a yemenian family with LAD-I from consanguineous relatives.All patients had history of chronic recurrent skin ulcerations, without bleeding tendency, requiring steroids and antibiotics, associated with persistent neutrophilia, without history of delayed cord separation, initially diagnosed as epidermolysis bullosa, successively as PG. LAD-I should be kept in mind while evaluating patients with PG especially in children with persistent neutrophilia in the absence of other rheumatological disorders.

Keynote: Diagnosis of LAD-I is extremely important for management, as treating these patients without adequate antibiotic cover may be fatal, as happened to one of them, and these patients often require hematopoietic stem cell transplantation for permanent cure, therefore genetic counseling especially on consanguineous parents is mandatory.

Key words: adhesion defects- bacterial infections- exome sequencing- leucocyte adhesion deficiency- primary immunodeficiencies.