GENETICS AND GENODERMATOSES

TREATMENT OF DOMINANT DYSTROPHIC EPIDERMOLYSIS BULLOSA WITH ORAL METHOTREXATE RESULTING IN SUSTAINED REMISSION.

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Background: Dominant dystrophic epidermolysis bullosa (DDEB) is an inherited disease resulting from a glycine substitution mutation in COL7A1 gene. Its prevalence is 2.4 cases per million in the US.
We report a case of DDEB responsive to oral methotrexate.

Observation: A 67-year-old Caucasian woman with a lifelong history of DDEB was treated with topical medications, various doses of systemic corticosteroids and multiple immunosuppressive agents with limited success and persistent disease. Her initial presentation in early childhood consisted of multiple blisters on knees, ankles, and other large joints. The diagnosis was made at the age of 3.5 years. She had multiple skin infections and constant pruritus. In the second decade, the blisters primarily involved the extremities.
Physical examination revealed multiple papules and excoriations on external aspect of the upper extremities and middle back, dystrophic nailbeds, and hyponychia. No mucous membranes were involved.
Patient's mother and sister have DDEB. Her sisters' two daughters have a mild form of DDEB.
The diagnosis was confirmed by immunofluorescent mapping at the age of 54. Treatment with oral methotrexate, at a dose of 15 mg weekly and 1 mg daily of folic acid was initiated in August 2011. She achieved dramatic clinical improvement in six weeks. Seven months after starting methotrexate, she was weaned off prednisone. During the following six years the patient had no skin lesions and no pruritus.

Key message: Dominant dystrophic epidermolysis bullosa can be treated with oral methotrexate. In this patient with DBEB a clinical remission was achieved after forty years of persistent active disease.