PAEDIATRIC DERMATOLOGY

BULLOUS ERUPTION IN A 1-YEAR-OLD FEMALE IN THE PHILIPPINES: A DIAGNOSTIC CHALLENGE

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Background: Epidermolysis Bullosa, Generalized-Severe, (EBS-gen sev) is a rare genodermatosis, resulting from multiple gene mutations, including KRT5 and KRT14, and is characterized by marked mechanical fragility of epithelial tissues. It presents at birth with disseminated trauma or friction-induced blistering. Grouped blisters with a “herpetiform” arrangement may appear spontaneously on the trunk, upper limbs or neck, associated with hyperkeratosis of the palms and soles. Other clinical features include nail dystrophy, nail shedding and hair loss. Erosions heal without scarring, and lesions tend to improve with age. There is no specific treatment. Most therapies consist of supportive therapy, which include wound care, nutritional support, infection control and prevention of complications.

Observation: A 1-year old Filipino female presented at birth with red bumps and blisters on the distal extremities, which later spread to the rest of the body during infancy. There was no evidence of trauma or viral infection. Hyperkeratosis of the palms and soles, and nail thickening eventually developed. Skin punch biopsy revealed Subepidermal Vesicobullous Dermatitis, inflammatory predominantly eosinophils, pointing towards Infantile Bullous Pemphigoid, which was highly unlikely due to the early onset and clinical presentation. ELISA for anti-BP180 was equivocal. Direct immunofluorescence revealed negative results, indicating a diagnosis of Epidermolysis Bullosa. Genetic Mutation Testing was done in Israel, still with pending results. The patient was treated symptomatically, and during the months of follow-up, the child continued to have blister formation but was otherwise healthy.

Key Message: Epidermolysis Bullosa Simplex, the Generalized-Severe type, is a rare inherited disorder and our case is unique to previously reported cases of EBS-gen sev, as our patient presented with pathognomonic cutaneous lesions but with inflammatory histopathologic findings. The case highlights that optimal skin management is cornerstone in therapy. Thus, treatment of this condition is an active area of investigation.