Kindler Syndrome

Two siblings, a 13-year-old girl and an 8-year-old -boy (birth order 4 and 6) born out of a first degree consanguineous marriage, presented to us with recurrent trauma-induced clear or hemorrhagic acral blisters that healed without scarring, and cutaneous pigmentation. The blistering improved with age. There was history of similar acral blistering and anal stenosis in their first born sibling who died at the age of 12 days. Five other siblings were normal. Photosensitivity, oral ulcers, gum bleeding, dysphagia and constipation were present since birth. Cutaneous examination revealed poikiloderma mainly affecting face, neck and upper chest; multiple hyperpigmented macules over trunk and extremities; and cigarette paper like atrophy over abdomen and dorsae of hands and feet (***Fig.* 1**). Facial skin was shiny and stretched. Palmoplantar keratoderma, syndactyly, webbing of fingers and toes, sclerodermatous changes and reduced dermatoglyphics were noted (***Fig***. **2**). Poor dental hygiene, oral ulcers, gingivitis, periodontitis along with anal stenosis were also seen. Ectropion (both eyes), nail dystrophy, and thick, long cuticles over finger and toe nails were present. Patients lacked any neurodevelopmental delay. A diagnosis of Kindler syndrome was made due to typical clinical features. Symptomatic treatment was administered in the form of photoprotection, broad spectrum sunscreen, lubricant eye drops, dental care, laxatives and emollients. Kindler syndrome is a rare genodermatoses characterized by congenital or infantile acral blistering, photosensitivity, poikiloderma, diffuse cutaneous atrophy and mucosal involvement (urethral meatal, anal and esophageal stenosis). Ectropion, gingivitis, leukoplakia, squamous cell carcinoma, hypo – or anhidrosis, poor dentition, webbing between fingers and toes, pseudoainhum, syndactyly, sclerodermatous changes and nail dystrophy may be seen. It results from loss of function mutations in *FERMT1* (filopodin and ezrin/radixin/moesin) or *KIND1* (kindlin) gene encoding kindlin-1, a protein involved in actin-extracellular matrix linkage via focal contact that affects cell adhesion, signaling, morphogenesis, differentiation and migration. Immunostaining with anti-kindlin-1 antibody can be used as new diagnostic test. Early diagnosis of Kindler Syndrome aids in genetic counseling. Annual follow up for early detection of premalignant keratosis and malignancies is advocated due to risk of squamous cell carcinoma on acral areas or oral mucosa.

FIG. 1 *Poikiloderma affecting face, neck and upper chest.*

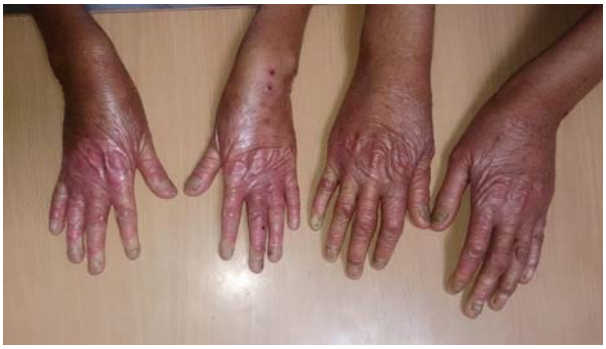

FIG. 2 *Sclerodermatous changes, and nail dystrophy with long cuticles.*

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