GENETICS AND GENODERMATOSES

KINDLER SYNDROME

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BACKGROUND: Kindler syndrome is a genodermatosis, autosomal recessive; characterized by vesicles and skin trauma blisters appearing early the first days of life simulating an Dystrophic Congenital Epidermolysis Bullosa; later during childhood is associated with photosensitivity and progressive poikiloderma; additionally genitourinary malformations and other alterations. Diagnostic criteria are: blisters on neonatal period, vascularis, photosensitivity, poikilodermia, severe cutaneous atrophy and palm-sole hyperkeratosis, acral congenital blisters, severe periodontal disease and phimosis.
Is caused by mutation of FERMT1 or KIND1 gene, that codes for Kindlin-1 protein producing a change in actin-cytoskeleton-matrix extracellular interaction causing a disorder in cell adhesion and migration processes.

OBSERVATION: We present the case of a child of 20 months of age, parents of Italian origin, who have started disease at the moment of birth with acral vesicles and blisters clear content, anoniquia and congenital phimosis. At first time disease was associated with diagnosis of Congenital Epidermolysis Bullosa which was confirmed by biopsy. Lesions resolve spontaneously; then presented outbreaks of vesicular lesions and eczema that leave lesions pinpoint scar appearance with a tendency towards generalization, adds sensitivity to Sun (erythema on exposed areas), palm-sole hyperkeratosis with appearance of acral predominance actinic keratoses. Patient has periodontal disease since 1 year of age. Given clinical evolution is done a second biopsy reported poikiloderma, concluding like Kindler syndrome.

KEY MESSAGE: Kindler syndrome, Poikiloderma, photosensitivity, periodontal disease and phimosis.