GENETICS AND GENODERMATOSES

KINDLER'S SYNDROME, A CLINICAL, GENETIC AND LABORATORY STUDY OF 210 CASES IN PANAMA, 1986-2017.

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Introduction: we presented in Costa Rica at the Central American Dermatological Congress (1988) the first 4 cases of KS in Panama. To date we have studied 210 cases.

Objective: to present the clinical, genetic, laboratory and natural history of KS.

Materials and methods: patients were recruited in the Hospital of Bocas del Toro, the Chiriquí Hospital and the Ngabe Bugle Comarca, between May 1986 and December 2017. The studies were carried out in the Children's National Hospital (Costa Rica), University of California at San Francisco, Bioepithelial Research Unit (Spain), and Hospitals of Panama.

Results: all patients were members of the Ngabe bugle tribe and resided in isolated villages in rural Panama. The patients were 6 months to 80 years old when they were first seen. 51% were men and 49% women. We have seen congenital blisters (97%), skin fragility (100%), early photosensitivity (94%), severe skin atrophy (96%), severe periodontal disease (70%), phimosis (90% male patients), anus imperforate (1 female patient), and squamous skin cancer (3 cases between 25-35 years). In 33 patients, linkage studies and homogeneity analysis were carried out, mapping the gene to 20p12.3. Several alterations were demonstrated in the redox mode of fibroblasts culture of patients with SK.

Conclusions: this study includes the largest cohort of patients with KS worldwide. We need more genetic studies in our patients to try to develop a useful understanding of the genotype-phenotype relationship and obtain a new treatment for them.