Epidemiological, clinical, and paraclinical characterization of patients with genodermatoses

ABSTRACT

The habilitation thesis entitled "Epidemiological, Clinical, and Paraclinical Characterization of Patients with Genodermatoses" presents the scientific and academic results obtained by the candidate Assoc. Prof. Dr. Ana Sorina Danescu after her doctoral and postdoctoral studies, with a specific focus on the study of genetic skin disorders, particularly hereditary epidermolysis bullosa and incontinentia pigmenti.

The research field is complex, and most scientific results were achieved through collaboration with international and national research groups, the most significant collaboration being with the University of Freiburg, Germany, initiated in 2005 and continued to the present day.

The habilitation thesis contains two main parts: the first part, containing seven chapters, is dedicated to scientific activities, showcasing the primary results obtained from studying genetic disorders, followed by chapters that include professional and academic achievements. The second part presents perspectives on the development of scientific, academic, and professional careers.

In the first chapter, epidemiological aspects of genodermatoses are studied, representing the first study conducted in Romania to evaluate the incidence and prevalence of these conditions. The second study assesses the correlation between the phenotype and genotype of Epidermolysis Bullosa, being the first study conducted in Romania to perform genetic analysis on a patient cohort.

The third study focuses on evaluating the quality of life of patients with Epidermolysis Bullosa, and the correlation between quality of life and disease severity. The fourth study evaluates a specific type of Epidermolysis Bullosa, namely Kindler Syndrome, requiring a complex genetic analysis, including mRNA analysis to identify mutations that could not be identified through usual genetic analyses.

The fifth study addresses a condition initially diagnosed as Epidermolysis Bullosa; however, the clinical evolution with the appearance of new clinical manifestations necessitated a genetic analysis for precise diagnosis. The sixth chapter presents a study on another genetic condition, namely incontinentia pigmenti, with the presented cohort including a significant number of patients, describing their phenotypic and genotypic aspects. The emphasis is on the multisystemic nature of the condition, highlighting the importance of genetic analysis for diagnosis, and the fact that periodic evaluation of patients can reveal certain severe extracutaneous manifestations.

The final chapter includes a detailed analysis of a case of incontinentia pigmenti, where cutaneous manifestations were associated with severe hepatic impairment, requiring liver transplantation. The study aimed to identify a pathogenetic mechanism explaining this association.

Results from these studies have been published in reputable journals such as the Journal of the European Academy of Dermatology and Venereology and the Journal of Investigative Dermatology. Other important research directions included autoimmune bullous diseases and dermatologic oncology.

The study results have been published in 32 articles, with the candidate being the primary author in the majority of them.