High-throughput DNA sequencing data obtained from testing patients with hereditary retinal diseases reveal a large number of genetic variants of unknown clinical significance. There is an urgent need for further molecular studies aimed at verifying the pathogenicity of these variants. It has intersected with inaccessibility of retina, a disease relevant tissue, from the affected individuals. With the novel discoveries indicating that human retinal genes may be also expressed in the skin, we hypothesize that the dermal cells may serve as a good model for studying the processing of mutated genes responsible for the development of retinal degenerations.

The aim of the study is to focus on *ABCA4*, one of the most important genes involved in the development of several retinal dystrophies. Our main aim is to investigate the composition of *ABCA4* transcripts and study *ABCA4* expression at mRNA and protein levels in hair follicles, keratinocytes and fibroblasts. For this purpose primary keratinocyte and fibroblast cell cultures will be established from collected skin biopsies. Our next aim is to compare the experimental data to choose and implement the most adequate cell type for studying molecular processing of the mutated *ABCA4* gene in patients.

To accomplish the presented aims qualitative analysis of the *ABCA4* transcripts will be performed using the Rapid Amplification of cDNA Ends (RACE) technique and cDNA sequencing. Quantitative *ABCA4* expression at mRNA and protein levels will be measured by real-time and Western blot analysis, respectively.

The expected result of the study will be the identification how *ABCA4* mutations affect processing of the *ABCA4* gene and verify their pathogenicity in an intracellular environment. Moreover, for the first time we will identify the sequences of *ABCA4* transcripts in different cell types of the skin. We are convinced that the model proposed in this study may be successfully implemented for testing other retinal gene mutations causing a broad spectrum of dystrophies of the posterior part of the eye and may become a starting point for discovering the role that *ABCA4* plays in the skin.