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ASSESSMENT OF POLYMORPHIC VARIANTS, EXPRESSION OF SELECTED GENES AND PRESENCE OF AUTOANTIBODIES INVOLVED IN REGULATING AUTOIMMUNE RESPONSE IN PATIENTS WITH VITILIGO.

BACKGROUND

Vitiligo is an acquired depigmentation disorder. The etiopathogenesis of this disease has not been fully explained and its course is heterogeneous. To date, there are several hypotheses, among which the greatest importance is attributed to autoimmune processes. This theory is supported by the frequent association of vitiligo with other autoimmune disorders such as Hashimoto's disease, Graves' disease, alopecia areata, type 1 diabetes mellitus, psoriasis, Addison's disease, and pernicious anemia. Not all risk factors contributing to vitiligo and coexisting autoimmune disorders or severe disease progression have been identified. Researches also points to a significant genetic component with a polygenic mode of inheritance. Recent genome-wide association studies (GWAS) have identified over 50 loci associated with predisposition to vitiligo, including genes involved in immune regulation.

AIM OF THE STUDY

The aim of the study is to assess gene expression, polymorphic variants and the presence of autoantibodies (ANA, anti-TG, anti-TPO) in patients with vitiligo. The obtained results will be correlated in terms of the coexistence of other autoimmune diseases. Analysis of these parameters will allow for monitoring of patients at risk and potential detection of coexisting diseases at an earlier stage of their development. Additionally, the obtained data will be correlated with the course and activity of the disease process. This study will help identify factors influencing prognosis and disease dynamics. Understanding the molecular basis and mechanisms of the disease process may contribute to expanding therapeutic options as well as implementing personalized forms of treatment.

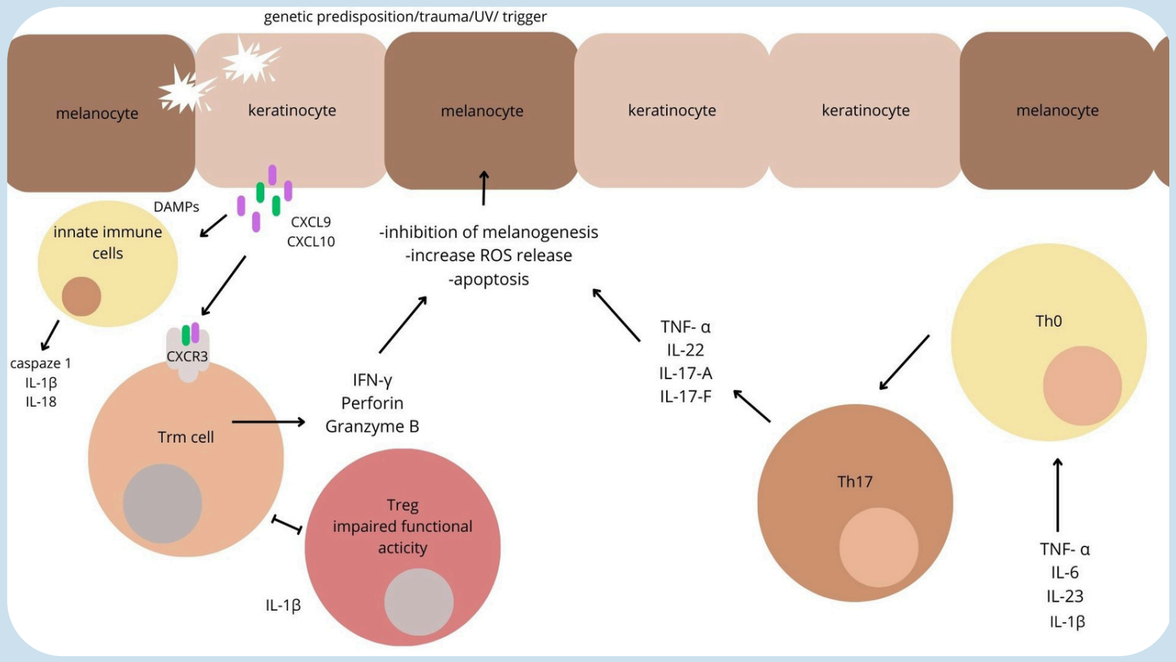

Figure 1. Pathogenesis of vitiligo lesions.

METHODOLOGY

The research material will consist of aproximately 100 patients with vitiligo aged 18 to 85 years. The obtained results will be compared with a reference group, which will consist of 50 healthy individuals appropriately matched for age and gender. Exclusion criteria from participation in the study will include: pregnancy, lactation, use of drugs prolonging bleeding and clotting time, hematological diseases, tumors during diagnosis or therapy. Clinical evaluation will include subjective and objective examination of the skin condition. The activity and degree of advancement of the disease will be assessed using selected scales: BSA (Body Surface Area), VIDA (Vitiligo Disease Activity Score), VES (Vitiligo Extent Score), and VASI (Vitiligo Area Scoring Index). Each patient will also complete a questionnaire containing questions about: age, gender, height, weight, use of substances, time of onset of the first skin changes and their distribution and location, coexistence of other autoimmune diseases, other autoimmune diseases in the family, and previous treatment. During the research, full venous blood will be collected. An assessment of the presence and specificity of autoantibodies in serum will be performed in all qualified individuals. Full venous blood will be used to assess the expression and single nucleotide polymorphisms (SNPs) of selected genes.

PATIENTS RECRUITMENT

In March 2023 consent from the bioethics committee number RNN/49/23/KE was obtained. Research material has been collected from patients and healthy controls since March 2023.

ACHIEVEMENTS

During this academic year, I took part in scientific conferences and workshops on the diagnosis and treatment of skin diseases.
I obtained a scholarship for an internship under the Erasmus + programme.
I published a work within the thematic scope of the doctoral dissertation (Kądziela M, Woźniacka A, Dziankowska-Zaborszczyk E, Kutwin M. Vitiligo: concomitant autoimmune and allergic diseases. Alergologia Polska - Polish Journal of Allergology. 2024;11(1):24-30. doi:10.5114/pja.2023.134244.).

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