English summary of the project - liquid biopsy as a complementary tool in diagnostics of early-stage mycosis fungoides – a pilot study

Introduction. Primary cutaneous lymphomas (PCLs) are a heterogenous and rare group of lymphoproliferative diseases. Due to a wide differential diagnosis and non-characteristic symptoms, they are often difficult to diagnose. Mycosis fungoides (MF) is the most common PCL. Importantly, the difference in years of survival of these patients is significantly different between early and advanced stages of the disease. The median time from the onset of the symptoms to diagnosis ranges from 13 up to 41 months.
Liquid biopsy is a novel method, which may in the future allow us to diagnose neoplasms basing on the peripheral blood sampling. This technique bases on peripheral blood sampling in order to analyze the circulating tumor DNA (ctDNA), which is a subset of circulating cell-free DNA (ccfDNA). Several studies have utilized liquid biopsy to successfully identify characteristic variants in ctDNA, which have been also found in the tumorous DNA.

Aim of the project. Aim of this project is to address whether it is possible to diagnose early MF using liquid biopsy.

Description of the study. In this project we are planning to collect peripheral blood samples and fragments of skin collected from the skin involved in the neoplastic process. Patients included in this study will have a diagnosis of MF of early stage. Next, the DNA is going to be isolated and several genes (which have previously been selected basing on the review of the literature) will be sequenced with the help of innovative methods such as next generation sequencing. Subsequently, using the sequences from our patients, the analysis of the sequenced data will be performed.

The reason for tackling the researched subjects. We believe that results of our research would facilitate and improve the process of diagnosing and managing patients with MF, especially concerning personalized therapy based on the concrete variants and their response to the treatment (such as in the advanced stages of melanoma immunotherapy personalized treatment) in the future.

The most important expected effects. The most important expected effect is to successfully identify the same genetic variants in ctDNA and in neoplastic lesions DNA. We hope that it will facilitate our theory that a characteristic molecular signature found in ctDNA may allow to diagnose MF.