Description for general public

The aim of the study is to present free circulating miRNAs in peripheral blood serum of patients who are genetically confirmed of having amyotrophic lateral sclerosis (ALS) and hereditary spastic paraplegia (HSP). Determination of miRNA levels relative to disease progression as well as determination of their predictive role in symptomatic patients. The next step will be to check the progression of the disease, what changes occur within the miRNA during the course of the disease and whether miRNAs can be considered as biomarkers of neurodegenerative diseases. This is important because in previous studies on mouse models, the effect of miRNA on the occurrence of neurodegeneration, for example miR-206, has been demonstrated; the elevated level was detected in ALS mice. In contrast, many miRNAs have been detected in humans (eg ALS: miR-338-3p, miR-451, miR-1275, HSP: miR-140, miR-691, miR-96, miR-182 and many others) whose expression is associated with changes in muscle tissue, in the mitochondrial genome and development of axons.

Research in this project will cover microRNA issues found in peripheral blood serum. Because current analyzes come down to monitoring the level of small-molecular-RNA expression, but in the cerebrospinal fluid. This method is invasive and requires specialized preparatory procedures. However, the proposed project comes down to the simplest form, which is peripheral blood collection. This is a minimally invasive procedure for the patient and does not require complicated executive procedures. The first stage is collecting material from patients at the Institute of Psychiatry and Neurology in Warsaw. Next, miRNA profiling will be carried out using microarrays, which aims to identify the most informative microRNAs involved in the pathology. The next step will be to check the level of miRNA expression using Real-Time PCR. The last activity will be a bioinformatic study of the results obtained.

The reason for undertaking a given research topic is the fact that previous research has focused largely on miRNA profiling within a given tissue and cerebrospinal fluid. The study that we want to perform includes miRNA profiling from peripheral blood serum, is minimally invasive and has great diagnostic potential in the form of test speed, with relatively small financial outlays compared to tests carried out on cerebrospinal fluid.