Summary

The dissertation outlines the potential of machine learning-based diagnostic decision support systems, focusing on two areas: the explainability of spline neural network models in the classification of microscopic images of leukocytes from peripheral blood smears, and an Anderson-Fabry disease screening system using natural language processing methods to extract clinical features from unstructured medical data.

The research addresses the challenge of explaining the decision-making process of spline neural network models in the classification of leukocytes in microscopic images from peripheral blood smears. Using the Shapely Additive Explanations algorithm, important cell differentiation elements were identified and the models' decisions were found to be consistent with laboratory practice.

The opportunities presented by the implementation of transparent systems to support the diagnosis of the rare disease Anderson-Fabry are discussed. Using natural language processing methods to extract clinical features identified by medical specialists, a screening system for Anderson-Fabry disease was developed. The system was validated by screening the medical records of patients from a Polish hospital. Among patients with a high risk of the disease, doctors confirmed Anderson-Fabry disease in further diagnostic procedures.

In conclusion, the study highlights the importance of artificial intelligence in medical diagnosis. By developing comprehensible diagnostic decision support systems in collaboration with medical experts, we can speed up the diagnostic process, thereby influencing patient outcomes. As artificial intelligence is developing at a very fast pace, it is important to pay particular attention to the explainability of the tools, and the security of the data. Ensuring that the implementation of artificial intelligence in diagnostic decision support systems will benefit patients and healthcare professionals.