

Abstract

This bachelor's thesis compares the reliability of predicting the pathogenicity of mutations in the γ chain of fibrinogen using various predictive algorithms. The work is designed as a "blind study," where we investigate the potential pathogenicity of 70 missense mutations described in the literature. To analyse the reliability of the individual programs, we used statistical metrics, specifically sensitivity, specificity, accuracy, and Matthews correlation coefficient. We tested the following programs PANTHER-PSEP, PMut, SNPs&GO, PhD-SNP, SIFT, Mutation Taster, PolyPhen2, and Provean. These are introduced in the introductory theoretical part, which also describes fibrinogen, its role in blood clotting, and diseases related to mutations in fibrinogen. The comparison showed that the quality of predictions by the various programs differs significantly. Programs more reliable predict pathogenic than benign mutations.