

Chair Prof. Böttinger Digital Health & Personalized Medicine

Machine learning to predict functional effects of genetic variants

There are currently two open Master theses in the area of genetic variant prediction with machine learning, that will be supervised by Henrike O. Heyne at HPI, Potsdam.

Background:

Ion channel diseases are at the forefront of precision therapy, where different functional entities are correlated with different disease outcomes and response to pharmacotherapy. Thus, categorizing ion channel variants' functional consequences are a prerequisite to choosing the optimal therapy for individuals suffering from channel-related diseases. However, standard functional tests with electrophysiology experiments are difficult to scale. The motivation of the Master thesis projects are thus to develop methods that predict such variant effects *computationally*.

Project Summary:

These projects are doing machine learning on genetic, biological and patient data to predict the functional effects of genetic mutations.

The first project will optimize an already existing tool

<https://www.science.org/doi/10.1126/scitranslmed.aay6848> after the initial dataset has been largely updated.

The second project aims to develop a completely new prediction method of genetic variants in NMDA receptors. The motivation is to better understand these channels and to predict optimal treatment for a patient. For example, only individuals with diseases due to loss-of-function mutations would profit from NMDA enhancers such as

Serine <https://pubmed.ncbi.nlm.nih.gov/34997442/>

Multiple Collaborators, including:

Rikke Moeller - clinical genetics, sodium/calcium channels (Denmark)

Sumaiya Iqbal - computational structural biology, machine learning (Broad Institute of MIT and Harvard, US)

Johannes Lemke - clinical genetics, NMDA receptors (Leipzig, Germany)

Please reach out to Henrike O. Heyne to learn more.

<https://hpi.de/boettinger/team/henrike-heyne.html>