

A U S H A N G

FREIE UNIVERSITÄT BERLIN

Fachbereich Mathematik und Informatik

Promotionsbüro, Arnimallee 14, 14195 Berlin

D I S P U T A T I O N

Freitag, 05.05.2023, 16:00 Uhr

Ort: Seminarraum 046

(Fachbereich Mathematik und Informatik, Takustr. 9, 14195 Berlin)

Disputation über die Doktorarbeit von

Herrn Jakob Konrad Hertzberg

Thema der Dissertation:

Identification and Prioritization of Putative Pathogenic Structural Variants based on Functional Annotation

Thema der Disputation:

Transformer Neural Networks and their Application in Bioinformatics

Die Arbeit wurde unter der Betreuung von **Prof. Dr. M. Vingron** durchgeführt.

Abstract: The transformer neural network architecture is designed for processing sequential data by utilizing the concept of “self-attention” to focus on relevant parts of the input sequence and computing weighted representations for each element. This allows for capturing long-range dependencies in the data. Originally developed for neural machine translation, transformers have since been applied to a wide range of sequence processing tasks including bioinformatic applications such as predicting protein structures, tissue-specific expression patterns, or quantifying the effects of genomic variants. In my first talk, I will provide an overview of the machine-learning concepts leading up to the development of transformers, describe the model architecture in detail and discuss recent examples of transformer models for variant prioritization. The second talk will be a summary of my dissertation in which I describe a novel pipeline for the detection and prioritization of potentially disease-causing structural variants in patients with limb malformations.

Die Disputation besteht aus dem o. g. Vortrag, danach der Vorstellung der Dissertation einschließlich jeweils anschließenden Aussprachen.

Interessierte werden hiermit herzlich eingeladen

Der Vorsitzende der Promotionskommission

Prof. Dr. M. Vingron