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

Donnerstag, 20. September 2018, 9:00

Ort: Seminarraum 031
(Fachbereich Mathematik und Informatik, Arnimallee 7, 14195 Berlin)

Disputation über die Doktorarbeit von

Herrn Max Schubach

Thema der Dissertation:
Learning the Non-Coding Genome

Thema der Disputation:
Estimating the relative pathogenicity of variants from the whole genome

Die Arbeit wurde unter der Betreuung von **Prof. Dr. P. N. Robinson** durchgeführt.

Abstract:
The use of high-throughput sequencing approaches, like whole exome or whole genome sequencing, for the identification of disease causing mutations has seen massive growth over the last few years. These efforts have produced extensive catalogues of human genetic variation. However, pinpointing the phenotypically causal variants among the millions of detected variants remains a major challenge. In 2014, Kircher et al. proposed the genome wide score, Combined Annotation–Dependent Depletion (CADD), to asses the functional deleteriousness of variants from coding and non-coding regions. CADD integrates many diverse annotations into a single measurement using machine-learning methods. To ensure that the score is not limited to a small set of genetically or experimentally well-characterized mutations, it is trained on variants derived from an evolutionary model. The disputation talk will cover the ideas behind whole-genome variant pathogenicity prediction using machine learning techniques on the example of the CADD approach.

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. R. M. Piro