

FREIE UNIVERSITÄT BERLIN

Fachbereich Mathematik und Informatik

Promotionsbüro, Arnimallee 14, 14195 Berlin

DISPUTATION

Dienstag, 12. Januar 2021, 15:00 Uhr

Ort: [WebEx](#)

Disputation über die Doktorarbeit von

Herrn David Heller

Thema der Dissertation:

Structural variant calling using third-generation sequencing data

Thema der Disputation:

Genome graphs and their application in pangenomics

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

Abstract:

Since its first publication in 2001, the human reference genome has transformed the biological and medical sciences. Acting as the baseline for most genetic analyses it has enabled numerous discoveries but at the same time imposed unique limitations and biases. In particular, analyses using a single linear reference genome are blind to the considerable amount of human genome sequence missing from the reference and its associated variants and phenotypes.

In this talk, I will present pangenomic approaches as an alternative that can capture the genetic diversity in human and other species more completely. Instead of choosing a single genome as the reference, pangenomes model the full set of genomic elements present in a population. After introducing different representations of pangenomes I will focus on genome graphs because they can model both simple and complex genomic variation. My talk will cover 1) the basic elements of genome graphs, 2) two different methods for genome graph construction and 3) the application of genome graphs for the genotyping of structural variants (SVs).

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