

A U S H A N G

FREIE UNIVERSITÄT BERLIN

Fachbereich Mathematik und Informatik

Promotionsbüro, Arnimallee 14, 14195 Berlin

DISPUTATION

Montag, 1. Juli 2024, 15:00 Uhr

Ort: Seminarraum 006

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

Disputation über die Doktorarbeit von

Gaojanyong Wang

Thema der Dissertation:

Copy number variation characterization using nanopore sequencing

Thema der Disputation:

Application of hidden Markov model for segmentation in copy number variation detection

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

Abstract: Copy number variation (CNV) refers to the variation in the number of copies of a specific DNA stretch in the genome. CNVs are important for genetic diversity and evolution, gene dosage effects, and disease susceptibility. Therefore, various techniques, such as array comparative genomic hybridization, single nucleotide polymorphism arrays, and next-generation sequencing (NGS), have been employed to characterize CNVs. However, the data generated by these techniques can be noisy. Segmentation is a crucial step in CNV detection that addresses signal fluctuations and divides the genome into regions with the same copy numbers. Segmentation is vital for effective CNV analysis, as it allows for the examination of these uniformly segmented regions rather than millions of individual genome loci. In this talk, I will first introduce the fundamental mathematics of the hidden Markov model (HMM). Then, I will discuss how HMMs are applied to segmentation in CNV detection using arrays and NGS. In the end, I will conclude by summarizing the advantages and disadvantages of using HMM in the segmentation process.

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