

FREIE UNIVERSITÄT BERLIN
Fachbereich Mathematik und Informatik

Promotionsbüro, Arnimallee 14, 14195 Berlin

DISPUTATION

Donnerstag, 21. Juli 2016, 10.00 Uhr

Ort: 1.1.53, Arnimallee 14, 14195 Berlin

Disputation über die Doktorarbeit von

Herrn Vikas Bansal

Thema der Dissertation:

**Computational Analysis of High-Throughput Sequencing Data
in Cardiac Disease and Skeletal Muscle Development**

Thema der Disputation:

Detection of copy number variations using high-throughput sequencing

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

Abstract: High-throughput sequencing (HTS) technology is rapidly evolving and revolutionizing the research in life sciences. Due to its low cost and high throughput, HTS is commonly used by various labs to answer biological questions. Using this technology, it is now possible to sequence an entire genome in less than one day. Besides whole genome sequencing, HTS has various other applications like targeted resequencing, mRNA abundance and various properties of chromatin. Using HTS, we can investigate the differences between the sequenced reads and the reference sequence. Genetic mutations can alter the single base (Single Nucleotide Variations), can delete or duplicate a DNA sequence (Copy Number Variations) or can invert the DNA sequence (Inversion) or can insert or delete a small sequence of 2 to 50bp (indels). Copy Number Variations (CNVs) are regions of a genome present in varying number in reference to another genome or population. In the last years, several computational strategies have been developed for detecting CNVs from DNA-seq data. In my PhD defense, I will introduce CNVs in general, technologies to detect CNVs and then focus on a particular method – CoNIFER (Copy Number Inference From Exome Reads). In the end, I will present a novel copy number variation calling method, which I developed, to identify individual disease-relevant copy number variations (CNVs) using exome or targeted resequencing data of small sets of samples.

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