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

Dienstag, 19. Juli 2022, 15:00 Uhr

Ort: Seminarraum 108/109
(Fachbereich Mathematik und Informatik, Arnimallee 6, 14195 Berlin)

Disputation über die Doktorarbeit von

Herrn Sebastian Niehus

Thema der Dissertation:
Multi-Sample Approaches and Applications for Structural Variant Detection

Thema der Disputation:
Analysis of the expanded 1000 Genomes Project cohort using svtools

Die Arbeit wurde unter der Betreuung von **Prof. Dr. K. Reinert** durchgeführt.

Abstract: The 1000 Genomes Project (G1k) has enabled numerous important discoveries since its initial release in 2012. One limiting factor of the G1k cohort has since been the low coverage at which most genomes of the cohort had been sequenced. Now, the G1k cohort has been re-sequenced, allowing for promising follow up analyses.
In this talk, I will present the recent expansion of the G1k cohort by the deep sequencing of 3,202 genomes, with a focus on the population-scale structural variant (SV) calling on this expanded cohort via the svtools pipeline. Svtools enables the SV calling on thousands of samples simultaneously by smart management of its subprocesses and data. I will discuss the strategies underlying svtools as well as its algorithmic approaches for the detection, merging and genotyping of SVs with high accuracy.

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. K. Reinert