

**A U S H A N G**

**FREIE UNIVERSITÄT BERLIN**

**Fachbereich Mathematik und Informatik**

Promotionsbüro, Arnimallee 14, 14195 Berlin

**DISPUTATION**

**Donnerstag, 16. Mai 2013, 14.00 Uhr**

**Ort: Max-Planck-Institut  
14195 Berlin, Ihnestr. 63-73 (Dahlem),  
Hörsaal Erdgeschoss**

**Disputation über die Doktorarbeit von**

**Herrn Sebastian Köhler, M.Sc.**

**Thema der Dissertation:**

**Phenotype informatics:  
Network approaches towards understanding the diseasome**

**Thema der Disputation:**

**Integration of clinical information and molecular genetics analyses**

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

**Abstract:** High-throughput data on the associations and interactions between genes/proteins has been available for several years. In combination with links between human genes and human Mendelian diseases, this data is highly important for biomedical research, but the lack of a large scale classification of those diseases and a quantifiable similarity measure has hampered more sophisticated analyses.

In my presentation I will focus on an approach for quantifying the association between diseases. I will show how the construction of an integrated network consisting of diseases and genes can enhance knowledge discovery in biomedicine, and especially human genetics. Two approaches based on this integrated graph will be discussed, a regression model and a random walk model.

The second part of the presentation will cover the last chapters of my thesis entitled "Phenotype informatics: Network approaches towards understanding the diseasome".

I will present an approach towards an ontological, computer interpretable representation of phenotype information in humans and model organisms. I will show how an integrated phenotype ontology for human, mouse, and zebrafish can be applied to the interpretation of Copy-number variant disorders in humans.

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