Titel:

Filtering and assessment of NGS variants made reproducible

Abstract:

Due to NGS technology, it is possible to sequence increasingly larger DNA regions quickly and cheaply. The resulting lists of variants have to be filtered and evaluated according to different criteria in order to (a) exclude sequencing artifacts and (b) identify the mutations relevant for the development of a disease.
Up to now, the filtering process is typically carried out on an individual laboratory basis and the criteria and parameters used are rarely documented, which makes it difficult to reproduce published studies. However, especially when therapy decisions are affected, it is necessary to standardize the entire analysis process and to document the algorithms used in a reproducible manner.
In the talk, approaches to formalize and standardize the Variant Assessment process will be presented, as well as a specially developed language for platform-independent description of filter schemes (Scoring Scheme Description Language, SSDL). Filter schemes can be visualized in an understandable way, dynamically adapted, and the results can be inspected in real-time.