Fachhochschule Dortmund
University of Applied Sciences and Arts

WisPerMed
Wissens- und datenbasierte Personalisierung von Medizin am Point of Care

UNIVERSITÄT DUISBURG ESSEN
Offen im Denken

RP6 - Analysis of genome data

Research question

Personalized representation of the reliability of genome variants

Diagnosis and therapeutic decisions in oncology are based on gene variants or tumor mutational burden [1,2].

Technology: Currently gene panels (few genes, deep sequencing); soon genome sequencing (WGS)

WGS advantage: More options for therapy decisions

WGS disadvantage: low and heterogeneous sequencing depth; observed variants of varying reliability

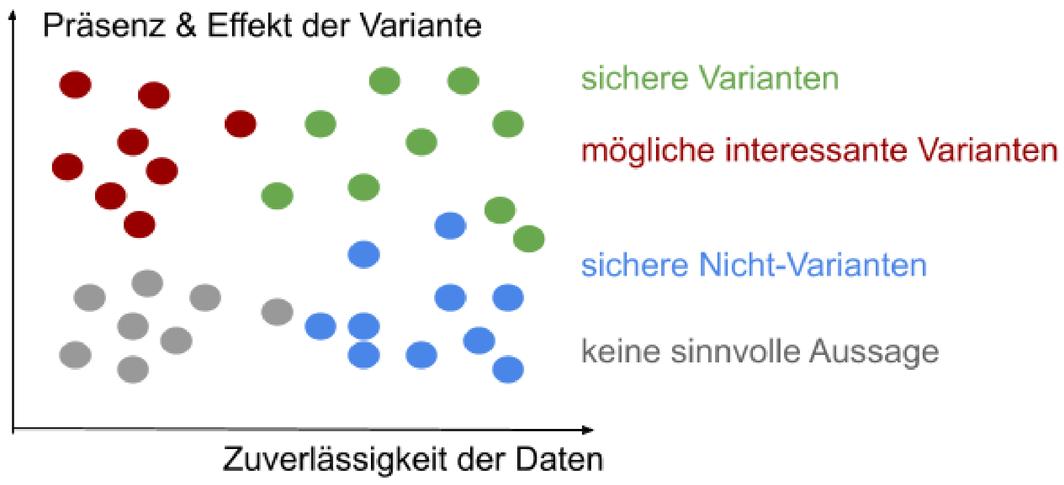
Question: How can we distinguish between

- „safe non-variants“ und
- „non-safe variants“ ?

Objective: Doctors at the point of care can prioritise relevant reliable variants and probably exclude ineffective therapy options.

Solution approach

One-dimensional representation (p-value or a posterior probability) does not do justice to the problem,
equally non-high-dimensional representation from many quality indicators (information overload).
→ Development of a 2D to 3D representation



Program: Development of intuitively understandable, visualisable quantitative measures for both dimensions; development of communication and presentation at the PoC; support on questions of acceptance by FP 11

State of the art

Numerous methods and tools exist for variant identification from genome data [3,4,5,6].

However: No method reports "safe non-variants". No method distinguishes between "safe non-variants" and "non-safe variants”:

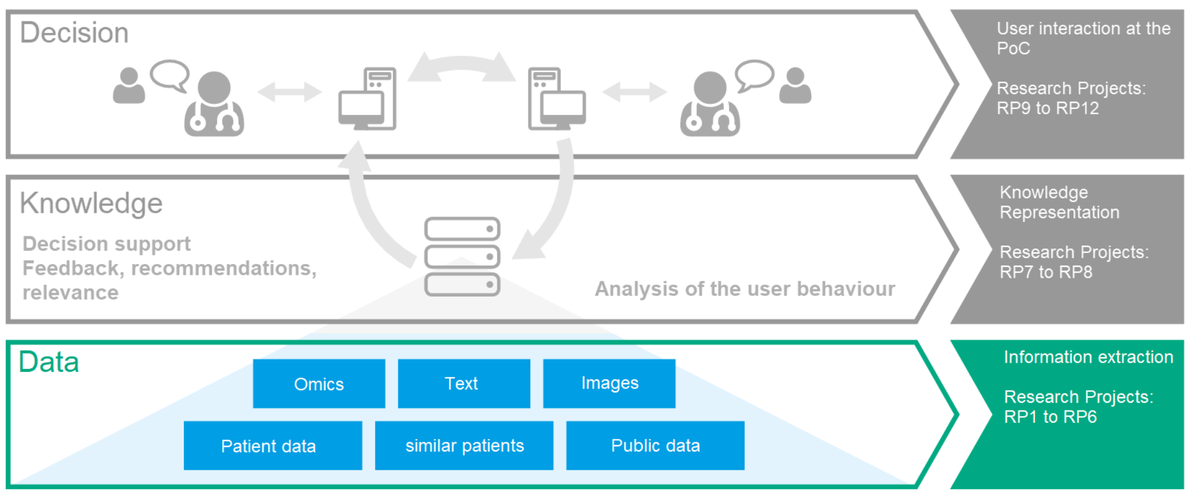
- GATK (Broad Institute): many different quality indicators, several threshold values such as for coverage, beach bias lead to information overload [6].
- Bayesian method (e.g. FreeBayes [4], Varlociraptor [5]):

  a-posteriori probability for the presence of the variant; low for little data and indistinguishable from a non-variant.

We have extensive experience in the presentation and interpretation of variants [1,2,5].

Integration

- Primary domain: Information extraction from omics data
- Communication of information via the Smart Hospital Information Platform (SHIP) at the point of care.
- Prospective testing of the use of information in the Molecular Tumor Board in melanoma patients* with existing exome or WGS data
- Integration of the representation in OpenEHR model for the patient file



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