Integrated Genomics Platform:
Putting Patients and Their Genomes into the Focus of our Research.

The IGP Team[1,2,3,4,*]
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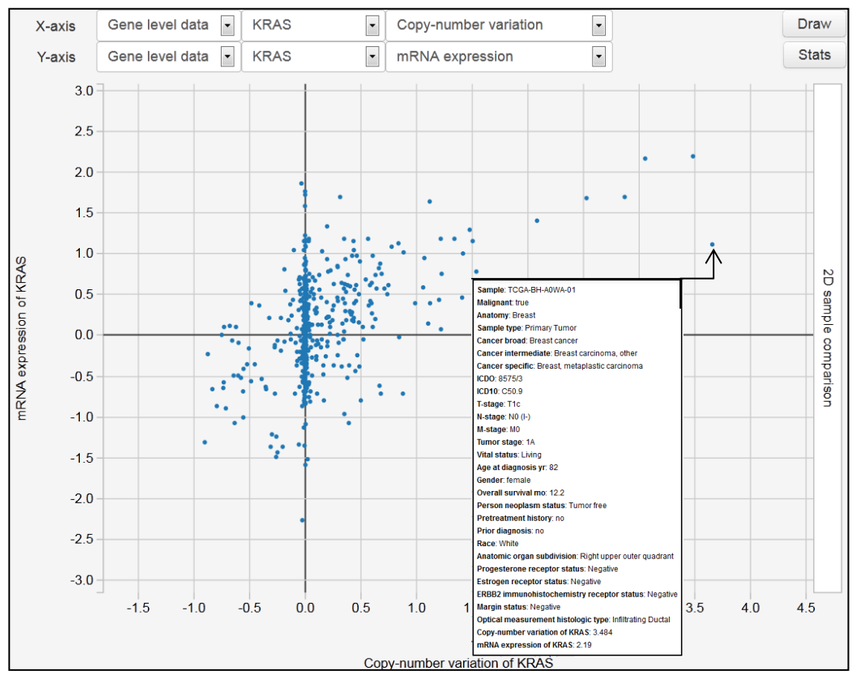
Summary

The fast progress in the generation of genomic data has reached the patient. Especially the advent of next generation sequencing and high resolution microarrays enable accurate descriptions of diseases with a strong genetic component ultimately leading to novel therapeutic approaches. Application of these technologies, however, lead to large amounts of data in need of effective storage and analysis. As now several data types (mutation, expression, microRNAs) become available for each patient, patient-centric views and analyses become mandatory. Consistent data handling and storage is a scientific and technological challenge towards both the research organization and the IT infrastructure.
We are therefore establishing the Integrated Genomics Platform (IGP) as a strategic initiative to develop a new central platform for research genomics data from Cardiology, Oncology and Clinical Sciences. This platform supports advanced data analysis capabilities and is intended to simplify discovery processes, e.g. for novel therapeutic targets and genetic biomarkers.
We aim to overcome current bottlenecks to enable true translational research by establishing a global mandatory repository and toolbox for storage and analysis of genomics data as well as common standards for data annotation, privacy & security.



Project Set-up & Use Cases

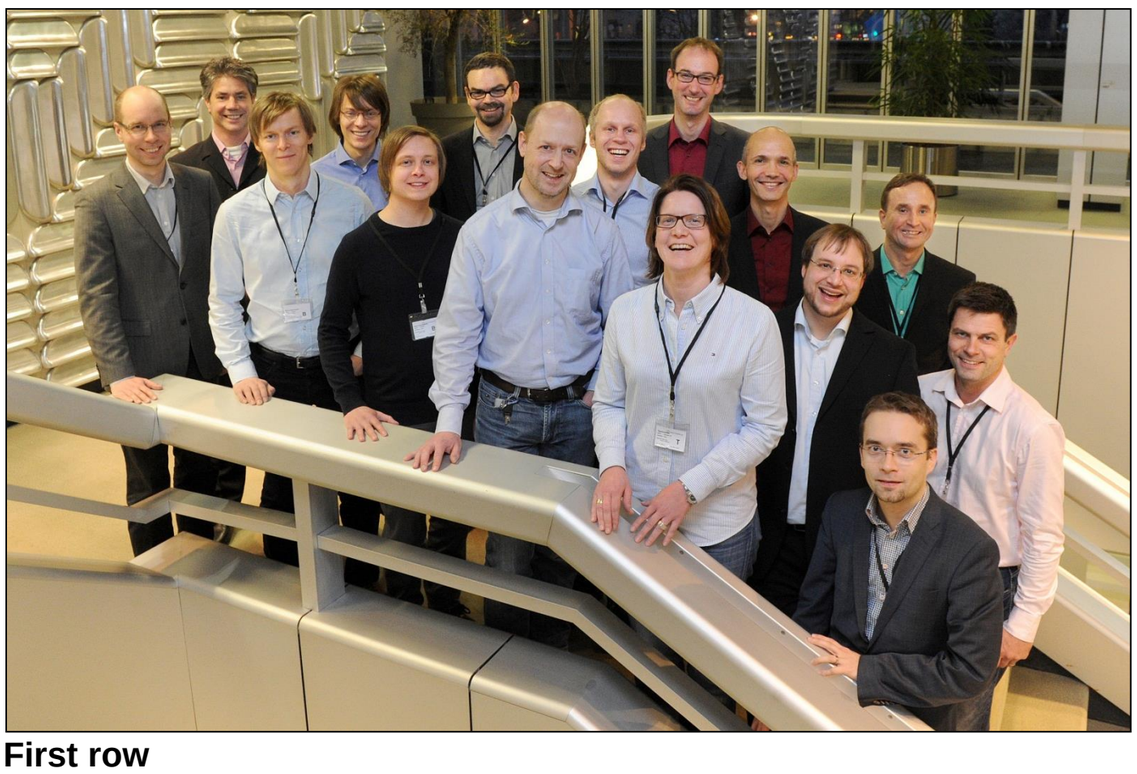
IGP can address many scientific questions …

- Genome-wide association studies in CardioGenomics
- Detection & Analysis of Driver Mutations in OncoGenomics
- Comparison of similarities or differences across research indications
- Patient stratification and biomarker detection with genomic features
- Distinction of survival times for groups of patients with disease sub-types, e.g. mutation-positive vs mutation-negative cancers
- Measuring correlations between different types of data, e.g. copy number vs point mutation or age vs tumor grade

… and tackle a variety of technological challenges

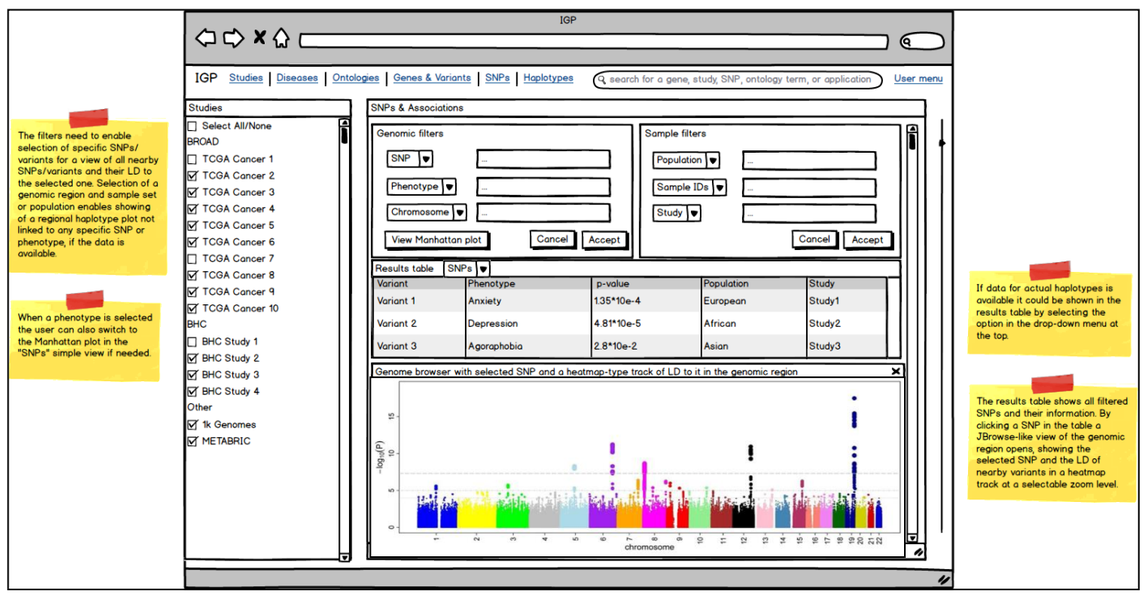
- Inreasing complexity of scientific questions towards data
  - Patient-centric combination of different genomics data types including clinical data across medical indications
- Size and amount of data sets
  - Genomic and clinical data of 20,000+ patients available inhouse
  - Cross-plattform integration from sequencing, microarrays, lab results and clinical parameters
- Adaptable to changing research areas and topics
- Replacement of silos by integration into scientific IT landscape

The IGP Team*



First row
Kalle Ojala, Henrik Edgren, Timo Mäkelä, Olaf Walter, Nora Manstein, Philip Groth, Sami Kilpinen
Second row
Robert Gomara, Carsten Jahn, Henrik Seidel, Martin Heger, Florian Sohler, Bertram Weiss, Henri Kajava, Rami Käkönen
(From left to right)

Advanced visualisations and analyses across studies and data types



Contribution to Breakthrough Innovation

- Cross-functional discoveries
  - From CardioGenomics and OncoGenomics to Clinical Sciences
- Single central One-Stop-Shop for Phenotypic and Genomic data
  - Medical history, patient information, mutations, gene expression, methylation, enriched with reference data from public sources
  - Mandate to collect and integrate research-related genomic data
- Discover novel relationships between:
  - Genotypes, Phenotypes, diseases, mutations, expression, copy number, external & internal data, cell lines, xenografts, humans

Functionalities

- Integration: Combine diverse data types across studies & indications
- Analysis: Enable simple and advanced statistical analyses
- Visualizations: Easy-to-use dashboards as well as interactive graphs

Project set-up

- Diverse team from Research areas, Bioinformatics and IT
- New technologies to enable Big Data, IT security and Data Privacy
- Prototype with over 10,000 samples from public sources established

Goals

Bringing our patients closer to our science

- Faster path to decision in our projects
- More meaningful discoveries from our data
- Ad-hoc check of ideas & questions to (de-)validate hypotheses
- Translational research: Cross bridges from early to clinical research
- Strenghtening Genomics research by strategic investments in R&D IT