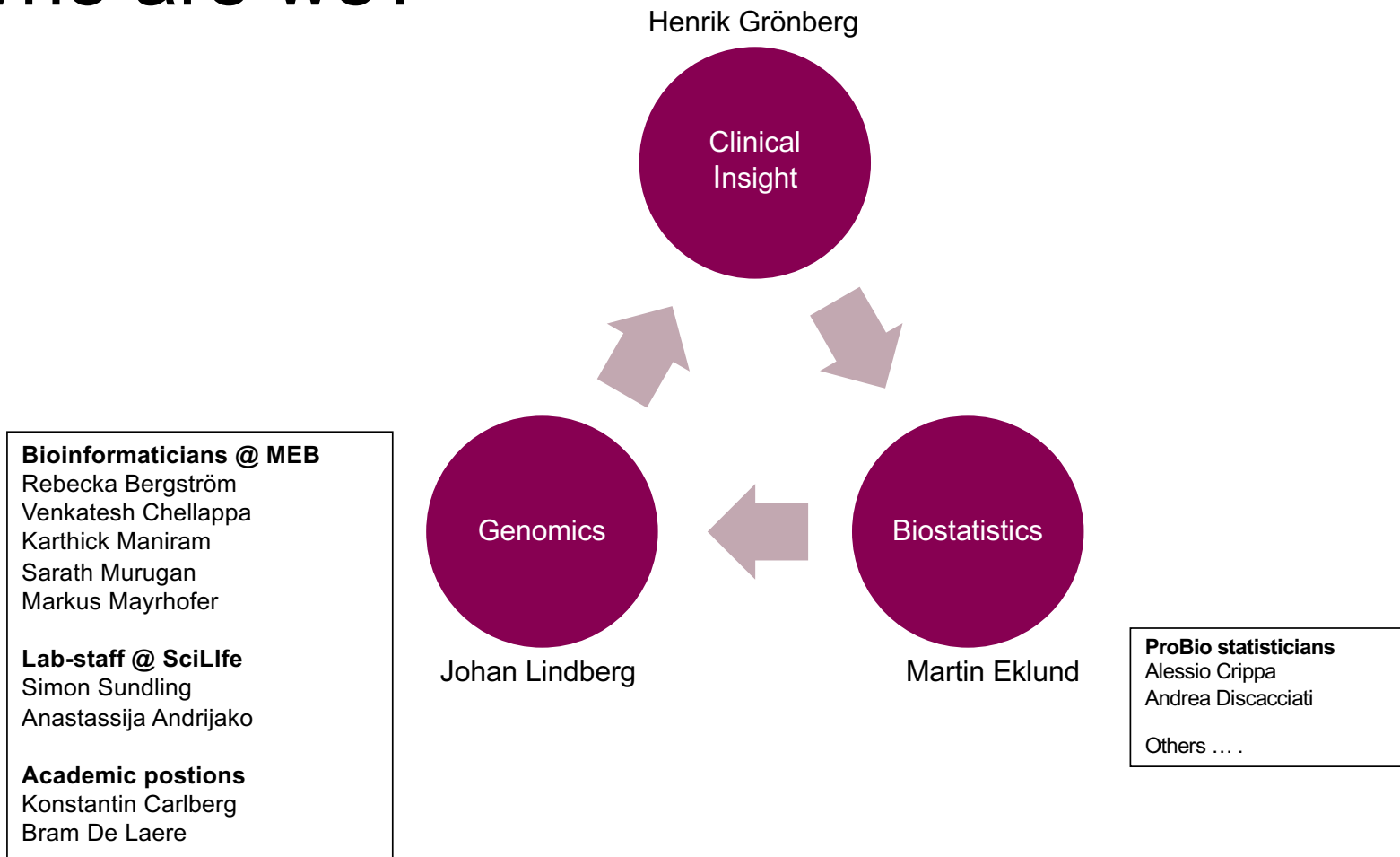


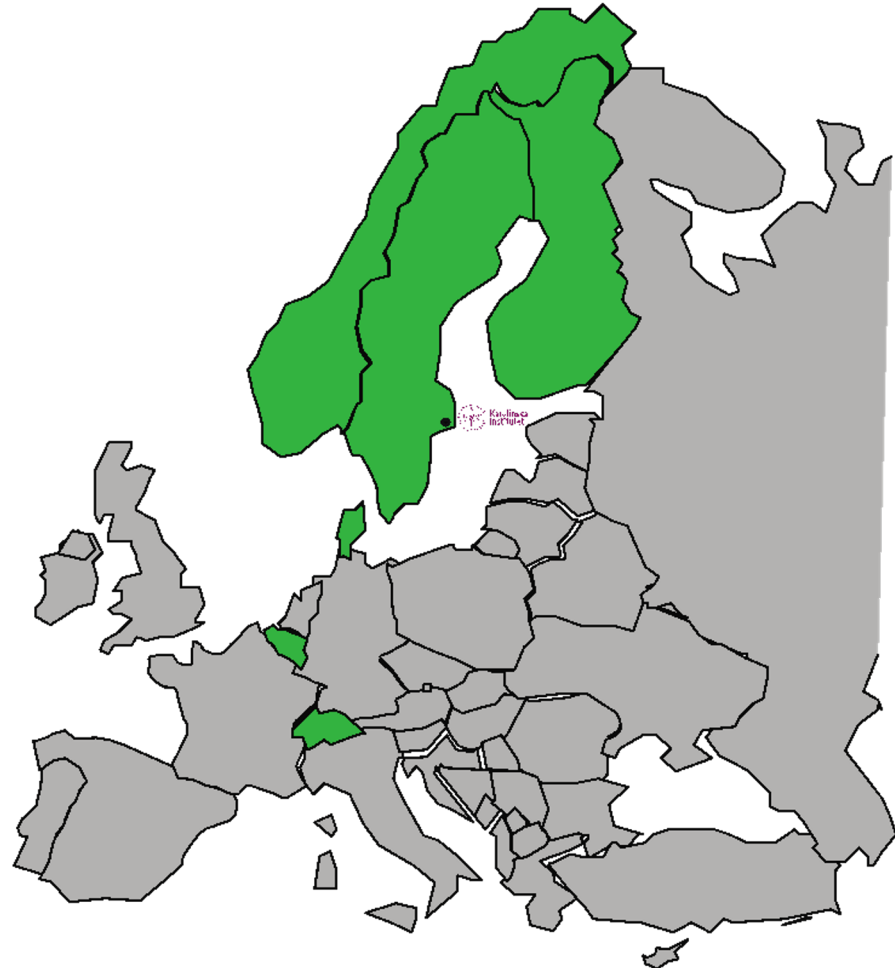
Welcome to Clinical Cancer Genomics, vt 2022

Who are we?



Prospective clinical trials using genomics

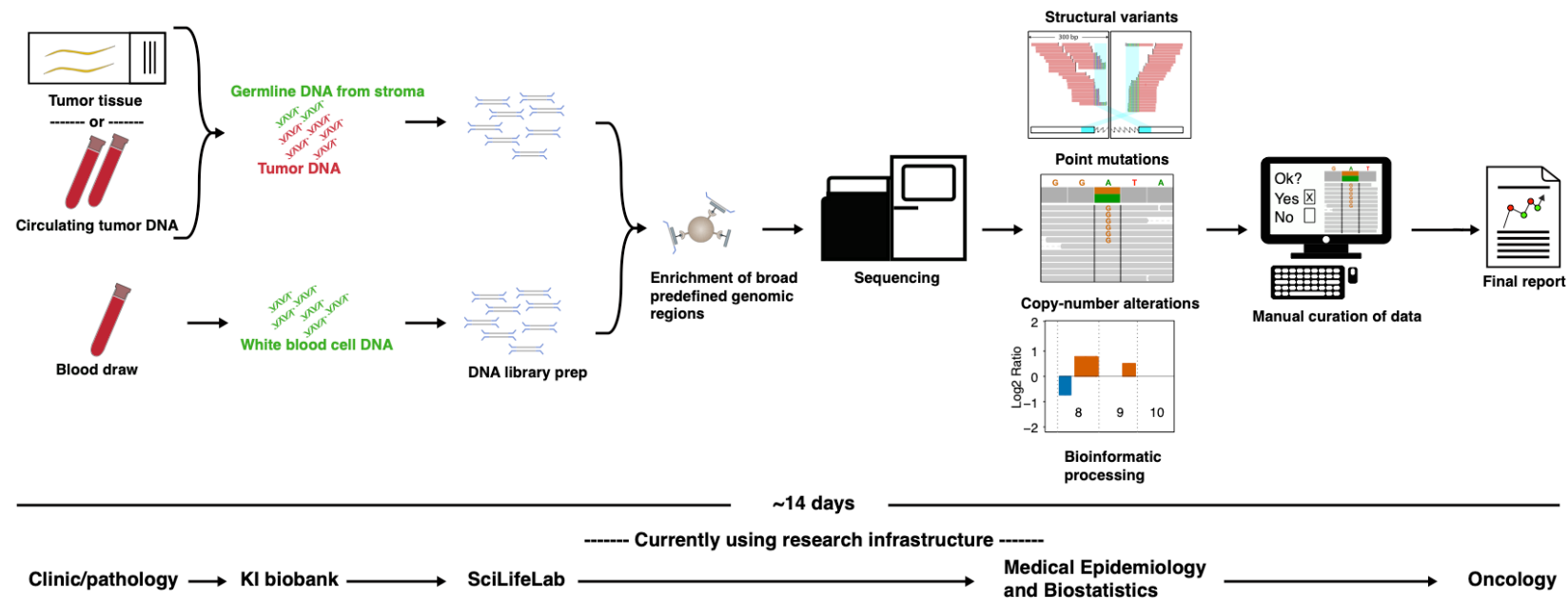
- Established infrastructure to enable the trials
- ~50 sites in 6 European countries ship biomaterial to KI



Genomics infrastructure

Experience

- > 4500 cases profiled using a validated research infrastructure/process
 - Research projects (DNA and RNA analysis)
 - Prospective clinical trials (DNA analysis)
 - Work towards accreditation and clinical implementation

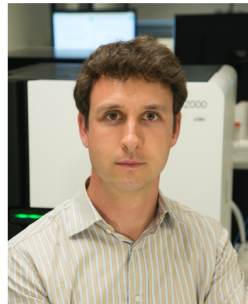


Modified version of an existing course

- <https://pmbio.org/course/>
- Course content under MIT license and Creative Commons.



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Assistant Professor of Genetics
Assistant Director, MGI



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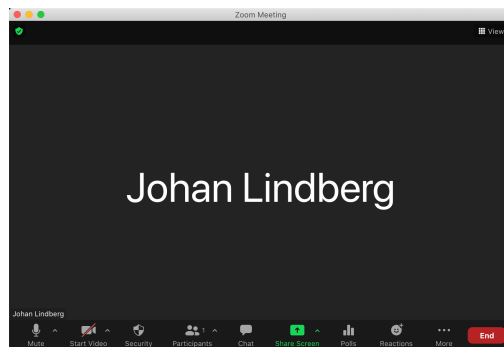
McDonnell Genome Institute, Washington University School of Medicine

Online set-up

Course webpage, info, lectures and practical sessions.

The screenshot shows the 'Clinical Cancer Genomics Course' webpage. The header is blue with a search bar. The main content area is white and contains a welcome message, a table of contents, and a description of the course. The table of contents includes links for Learning Outcomes, Contents of the course, Literature and other teaching material, and Additional Information. The course description states that it aims to provide an introduction to cancer genomics and to support the acquisition of practical knowledge regarding how to apply state-of-the-art methodology to interrogate the cancer genome in a routine clinical setting or a clinical trial setting. The course will include lectures covering the technology advancements that have enabled high-throughput analysis of cancer genomes and the knowledge that can be obtained by applying these technologies. This encompasses both laboratory sample processing and downstream bioinformatics. Lectures will be held in the mornings with computer-based exercises in the afternoon. The exercises will include processing and analysis of DNA- and RNA-sequencing data covering file formats, quality control aspects, identification of somatic variation, curation of identified somatic- and germline variants for clinical use, clonality estimation and annotation of variants. The main objective of the course is to facilitate that students get an understanding of basic theory and obtain practical knowledge that will enable course participants to apply the covered methodologies in their own research- or clinical laboratory.

Lectures, labwork supervision and breakout sessions via Zoom.




Student

AWS server



Hands-on exercises

 Clinical Cancer Genomics Course

Search

[↑ Back to top](#)

Clinical Cancer Genomics Course

- Course Details
- Schedule
- Lectures
- Pre-course setup**
 - Installation
 - Download Files >
 - Lab Session**
 - Day 1 >
 - Day 2 ▾
 - [Human Genome](#)
 - Annotation
 - Processing of DNA sequencing data
 - Introduction to IGV
 - Day 3 >
 - Day 4 >

HUMAN GENOME REFERENCE FILES

```
# Make sure CHRS environment variable is set.
echo $CHRS

# Create a directory for reference genome files and enter this dir.
#Command below already run
#mkdir -p ~/workspace/inputs/references/genome
cd ~/workspace/inputs/references/genome

# Download human reference genome files.
#Command below already run
#wget http://genomedata.org/pmbio-workshop/references/genome/$CHRS/ref_genome.tar

# Unpack the archive using 'tar -xvf' ('x' for extract, 'v' for verbose,
# 'f' for file).
tar -xvf ref_genome.tar

# View contents.
tree

# Remove the archive.
rm -f ref_genome.tar

# Uncompress the reference genome FASTA file.
gunzip ref_genome.fa.gz

# View contents.
tree

# Check the chromosome headers in the fasta file.
cat ref_genome.fa | grep -P "^>"
```

Table of contents

- Split the long fasta by chromosome
- Explore the contents of the reference genome file
- Index the fasta files

Schedule

	Mon	Tues	Wed	Thur	Fri
Lecture 1	Lectures about the cancer genome and how to interrogate it.		Bioinformatics pipelines and high-throughput computing environments	Copy number alterations	How to curate somatic- and germline data in a clinical trial setting
Lecture 2		Guest lecture, Clinical Genomics	QC and somatic and germline (small) mutation variant callers.	Structural variation	Clinical trials
Lecture 3		Lab introduction	Lab introduction	RNA-sequencing and lab introduction	Annotating, interpreting and reporting somatic- and germline variation.
Labwork	Tutorials, basic tools for cancer genomics	Files, tools and running a basic bioinformatic pipeline.	Calling and QC of somatic/germline variants	Analysis of copy number data, structural rearrangements and RNAseq data.	Investigating databases and working with data interpretation. Finishing off any remaining labwork from previous days

AM

PM

- Examination – show for a course instructor each day that you have completed and understood the labwork.
 - During the exercises you get questions ...
- This is our first time – have patience!
 - Any ideas/things you want to discuss for your own projects – let us know during the practical sessions, we can have a project brainstorm.

Questions?