GHGA

THE GERMAN HUMAN GENOME-PHENOME ARCHIVE

A Brief Introduction

The German Human Genome-Phenome Archive (GHGA)

Flashlight Talk, CSAMA 2023

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Training Coordinator GHGA

In cooperation with

nfdi

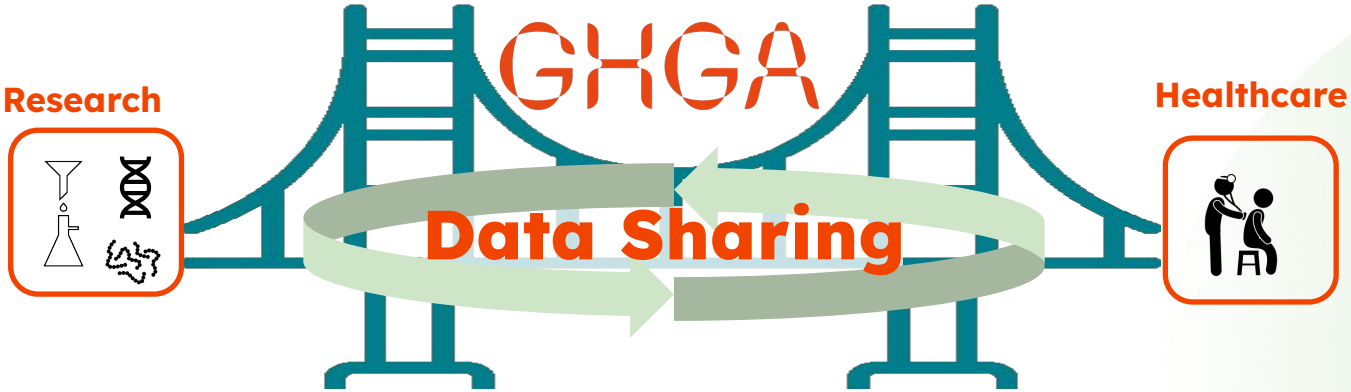
Omics as Driver for Research & Healthcare

- Technological revolution made genome sequencing affordable
- Omics data will revolutionise healthcare - improving the prevention, diagnosis and treatment of certain diseases
- Genomics is increasingly part of the becoming standard care

→ More data is being produced, but locally stored
→ missing exchange between research and health care (data/ knowledge)

GHGA

The GHGA Vision: Enabling Genomic Medicine

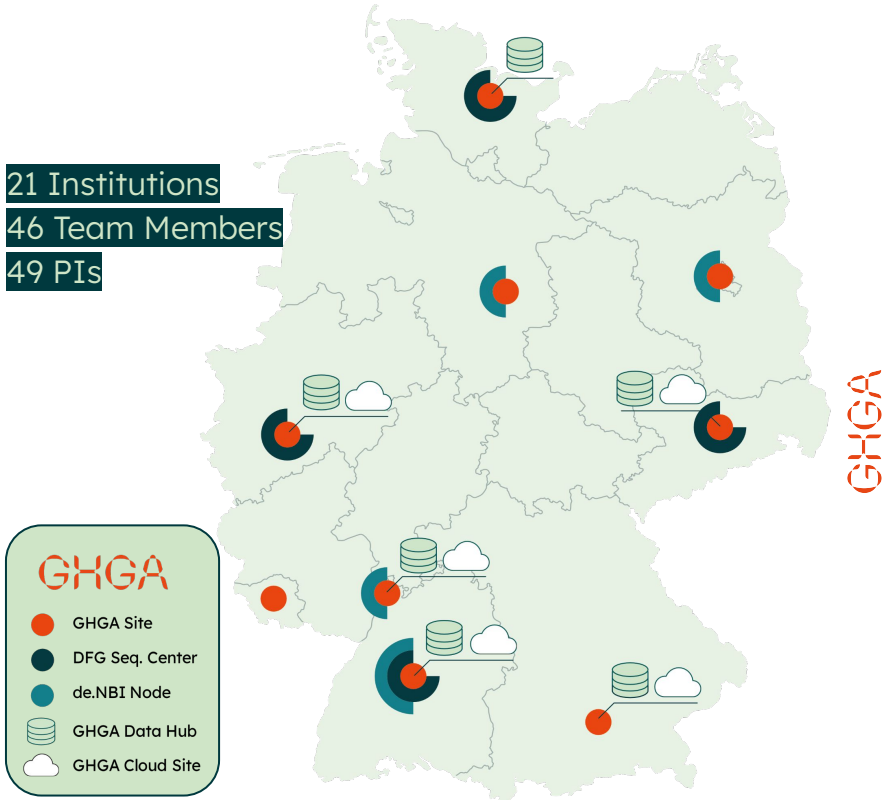


Main Aims:
Establishment of a nationally coordinated, interdisciplinary infrastructure integrating genome research and healthcare

– From Researchers for Research–

Who we are

- NFDI consortium
- Network of data hubs co-located with major academic sequencing centers
- Connected to national cloud infrastructure (de.NBI cloud) for large-scale analyses
- German national node within the federated European Genome-Phenome Archive (EGA)



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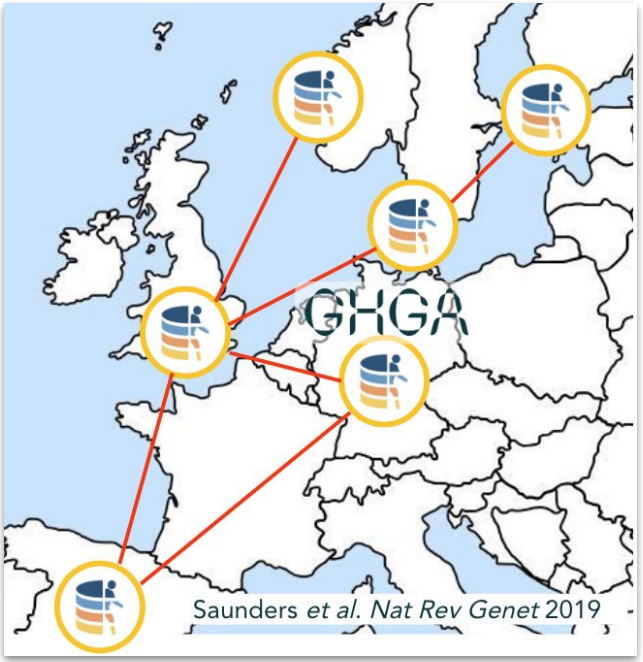

GHGA as national node within a European data sharing infrastructure:
federated EGA and GDI

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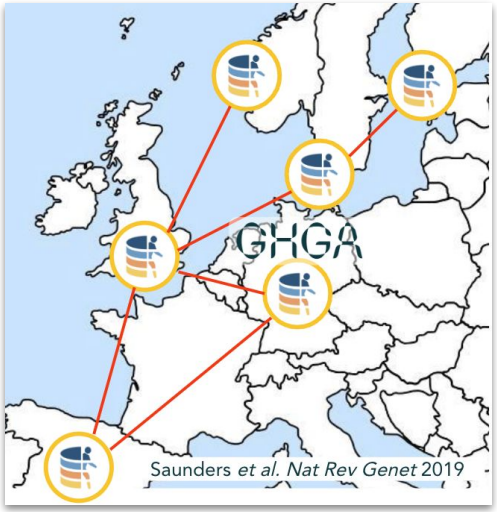
GHGA as national node within a European data sharing infrastructure: federated EGA and GDI

EUROPEAN GENOME-PHENOME ARCHIVE

European Genomic Data Infrastructure

Federated EGA Network



- European research community promotes EGA federation to enable international data sharing.
- Exchange of technologies, ‚standards, methods and best-practice approaches
- **Linking national datasets via common metadata schemes**
- GHGA is integrated into the new “**GDI - The European Genomic Data Infrastructure”** project ⇒1+MG Initiative and EHDS

GHGA

Deutschland ist jetzt offizieller Partner des europäischen Großprojekts "1+Million Genomes Initiative". Bundesforschungsministerin Anja Karliczek und Bundesgesundheitsminister Jens Spahn unterzeichneten heute in Berlin gemeinsam die Deklaration „Towards access of at least 1 million sequenced Genomes in the EU by 2022“. 16.01.2020

Connection to other international key activities

European Commission

elixir

B1MG Beyond one Million Genomes

Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.

Projects that are planning to use GHGA

- In general: GHGA will be open for data submissions for all German research institutions
- Development with “Pioneer Projects” to develop infrastructure according to needs of the communities

Data Types in GHGA

- HUMAN Genome, Exome, Epigenome, Transcriptome (including bulk and single cell data), …

➔ **Connection to clinical metadata** (“Phenome”) is key!

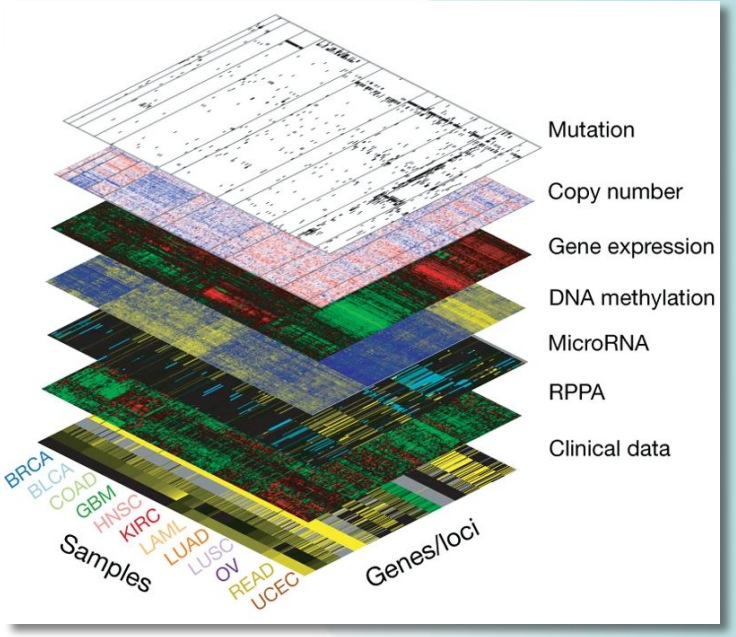
➔ **Access to raw data** is often essential for research

Disease Communities:

- Initial Focus on **Rare Diseases** and **Cancer**

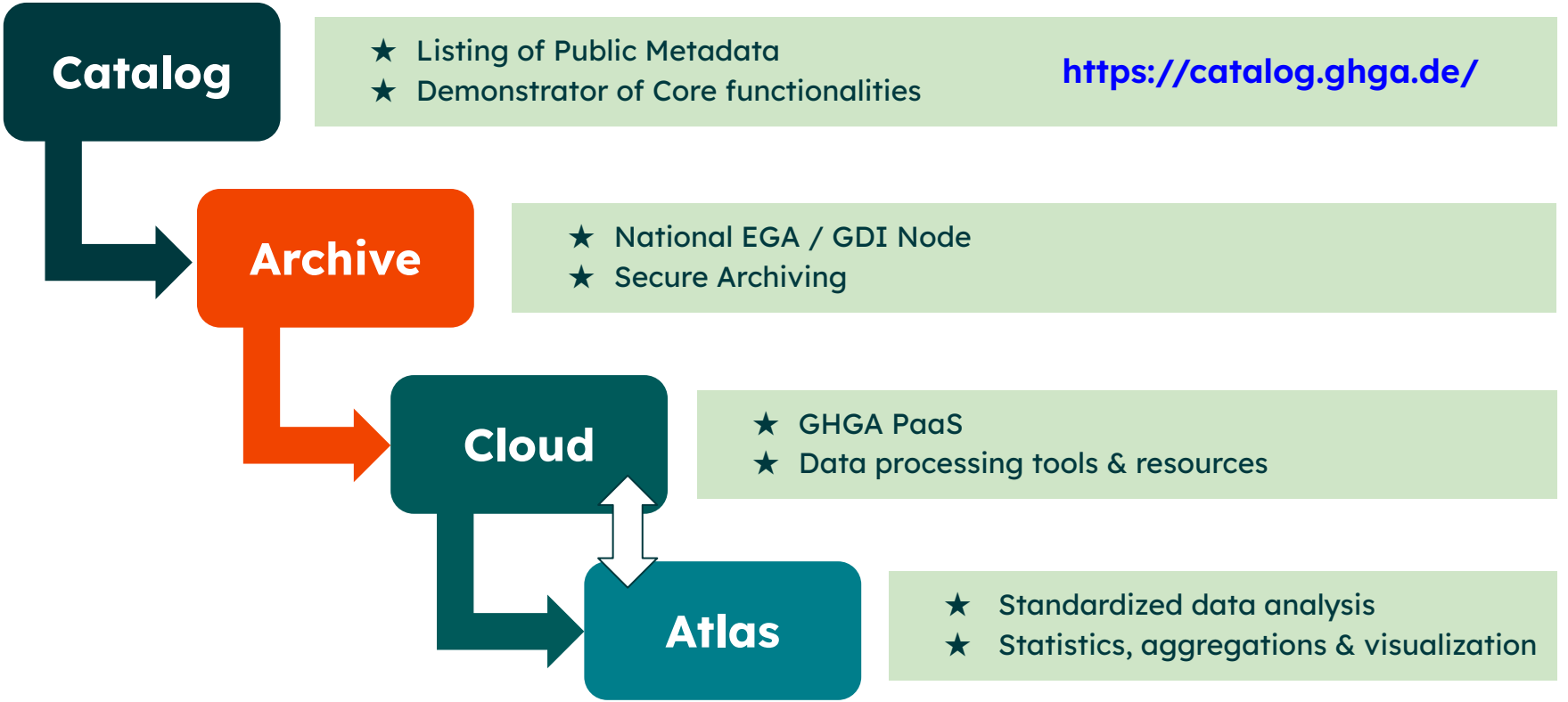
Regulated Data Access:

➔ Access only under “Controlled Access” with clear contractual regulations according **to GDPR and patient consent**

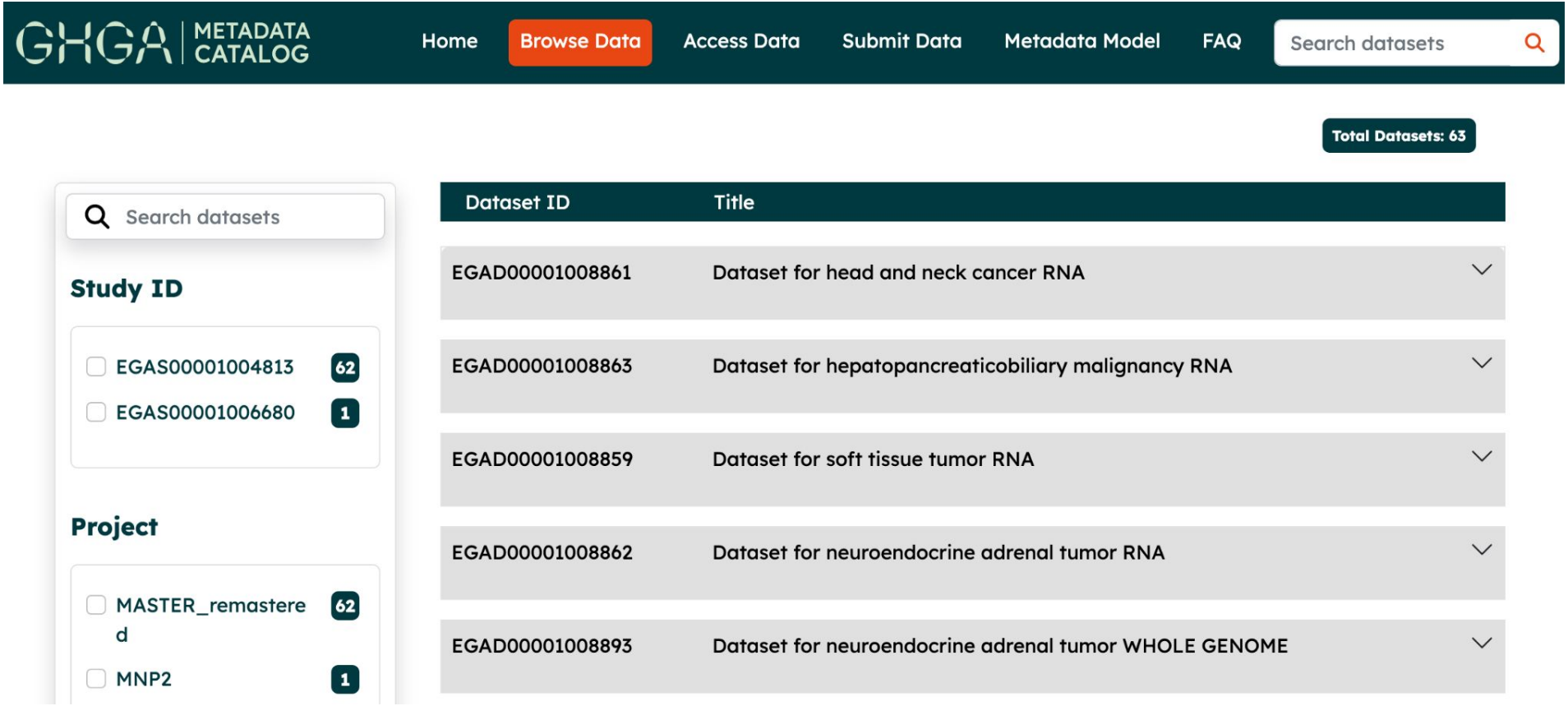


Source: ICGC/TCGA; The Cancer Genome Atlas Pan-Cancer analysis project. Nat Genet 45, 1113–1120 (2013)

GHGA Phases and Features



https://catalog.ghga.de/

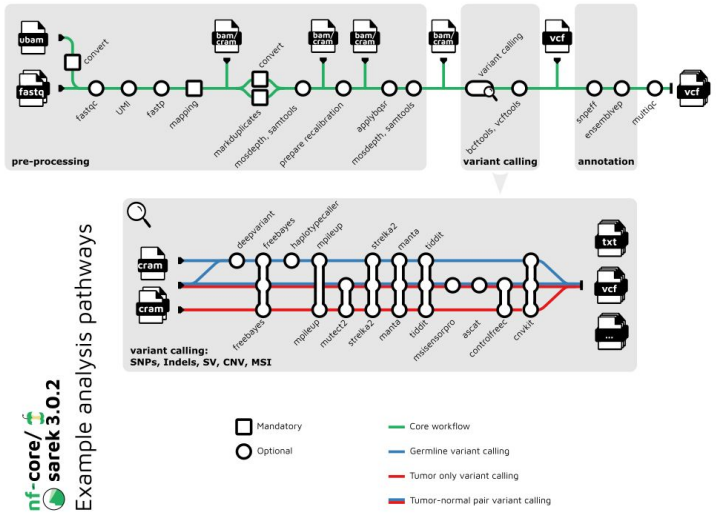


Workflow Standardization

More info: ghga.de/resources/data-analysis

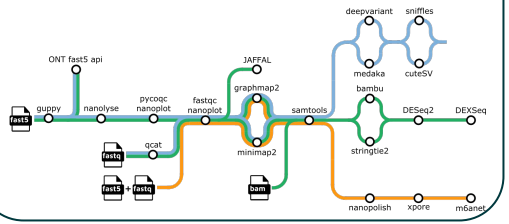
Short-read variant calling

- Germline (Nahnsen)
- Somatic (Hübschmann)



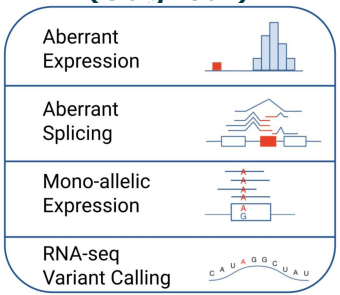
Long-read sequencing

(Ossowski)



RNA-seq for RD

(Gagneur)



Integration into communities

- Co-development with nf-core

nf-core

- Training (Stanford, Broad)
- Benchmarking (NGS-CN)

NGS-CN

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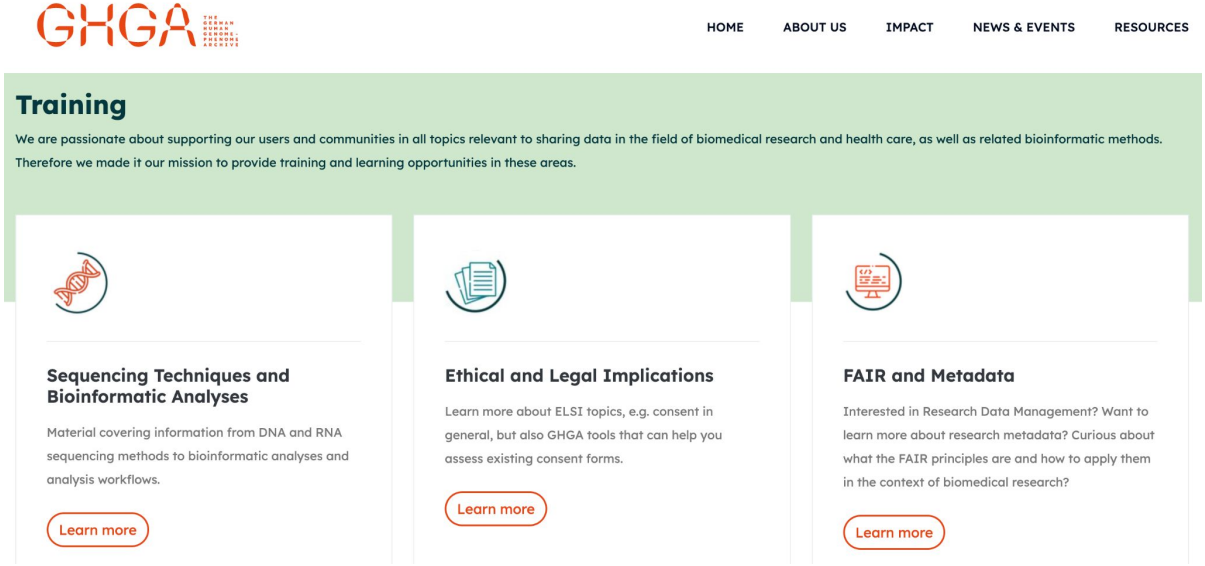
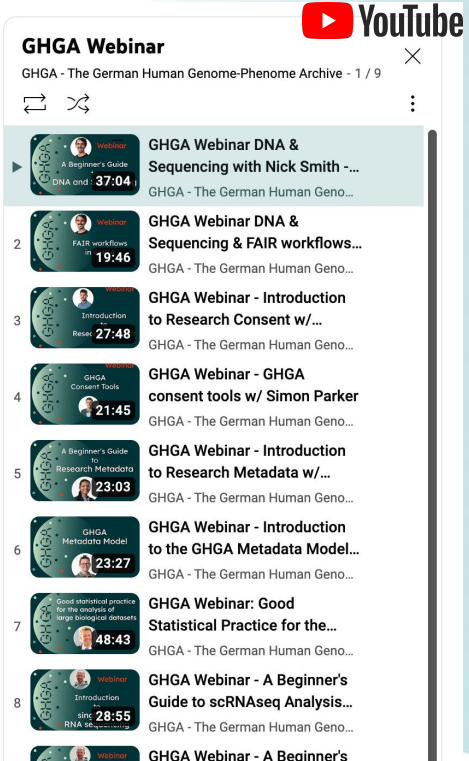
Training Resources

Live & On Training
ghga.de/resources/training



Sign up to our newsletter for news about future events like our monthly webinars!

Youtube
https://t1p.de/rgsse



Upcoming GHGA events

11 September 2023, Hamburg
NGS Harmonization & Standardization Workshop

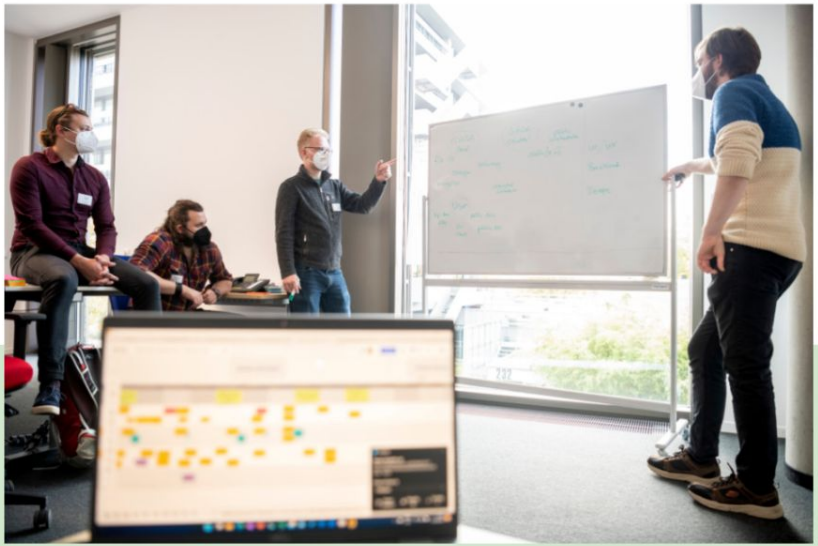
WS4) Standardizing and harmonizing NGS analysis workflows

Organizers: Dr. Florian Heyl (German Cancer Research Center, Workflow Coordinator for GHGA); Dr. Kübra Narci (German Cancer Research Center); Dr. Paul Menges (German Cancer Research Center); Dr. Christian Mertes (Technical University of Munich); Dr. Nicole Schatlowski (University of Tübingen); Dr. Julia Philipp (European Molecular Biology Laboratory, Training Coordinator for GHGA)

Description: With increasing numbers of human omics data, there is an urgent need for adequate resources for data sharing while also standardizing and harmonizing the processing of the data. Within the federated European Genome-Phenome Archive (EGA), the German Human Genome-Phenome Archive (GHGA) strives to provide (i) the necessary secure IT-

Upcoming GHGA events

ghga.de/events



Jobs

If you are interested in joining GHGA, please get in touch! We are constantly looking for talents in software development and cloud computing with an interest in setting up a state-of the art infrastructure for genome research. Please contact contact@ghga.de for further information.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