

**Bavaria joins forces
with the
National Infrastructure for
Genomic Medicine**

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In cooperation with



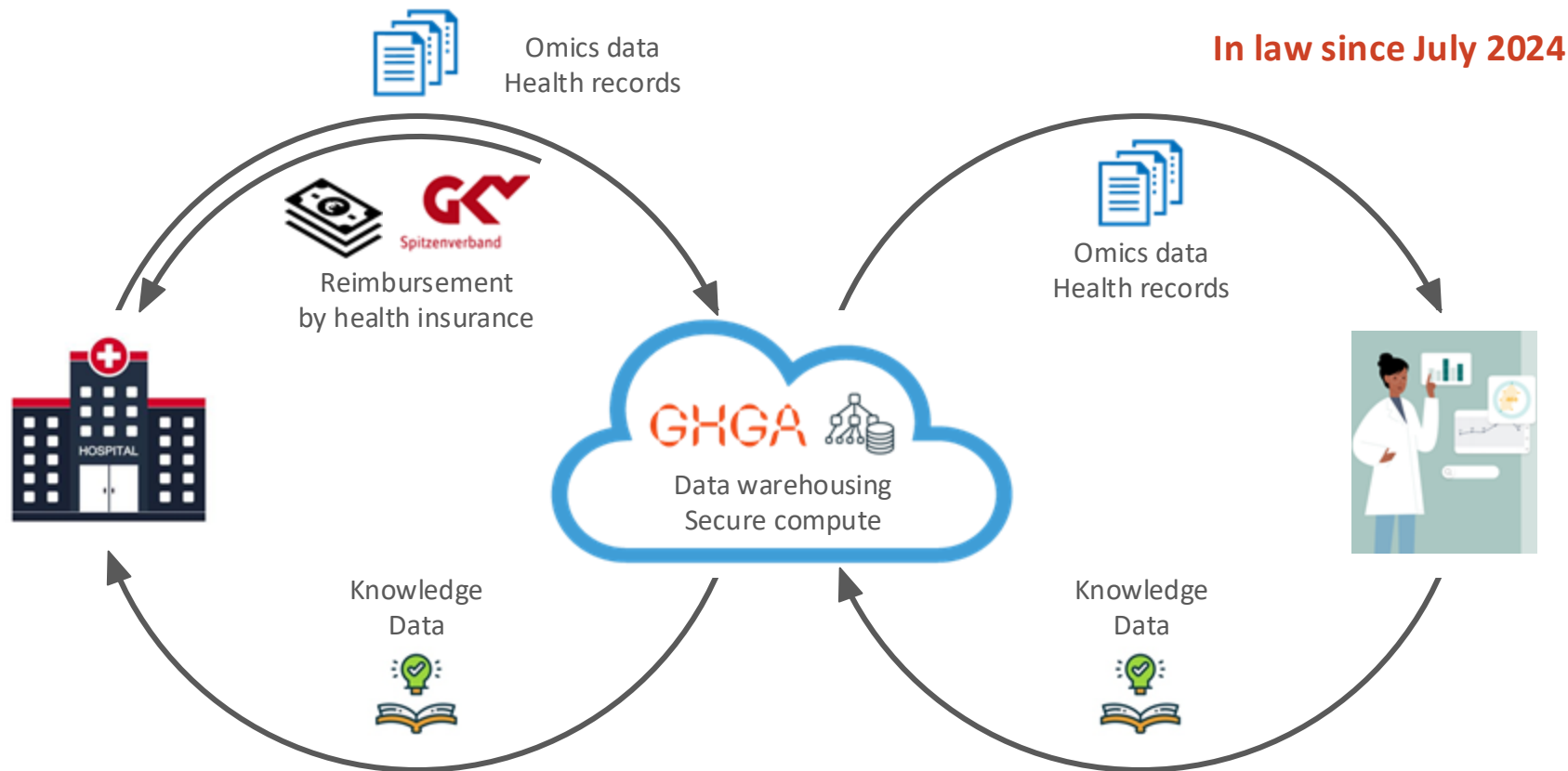
From N=1 to population-level

What is needed to scale genetic diagnostics?

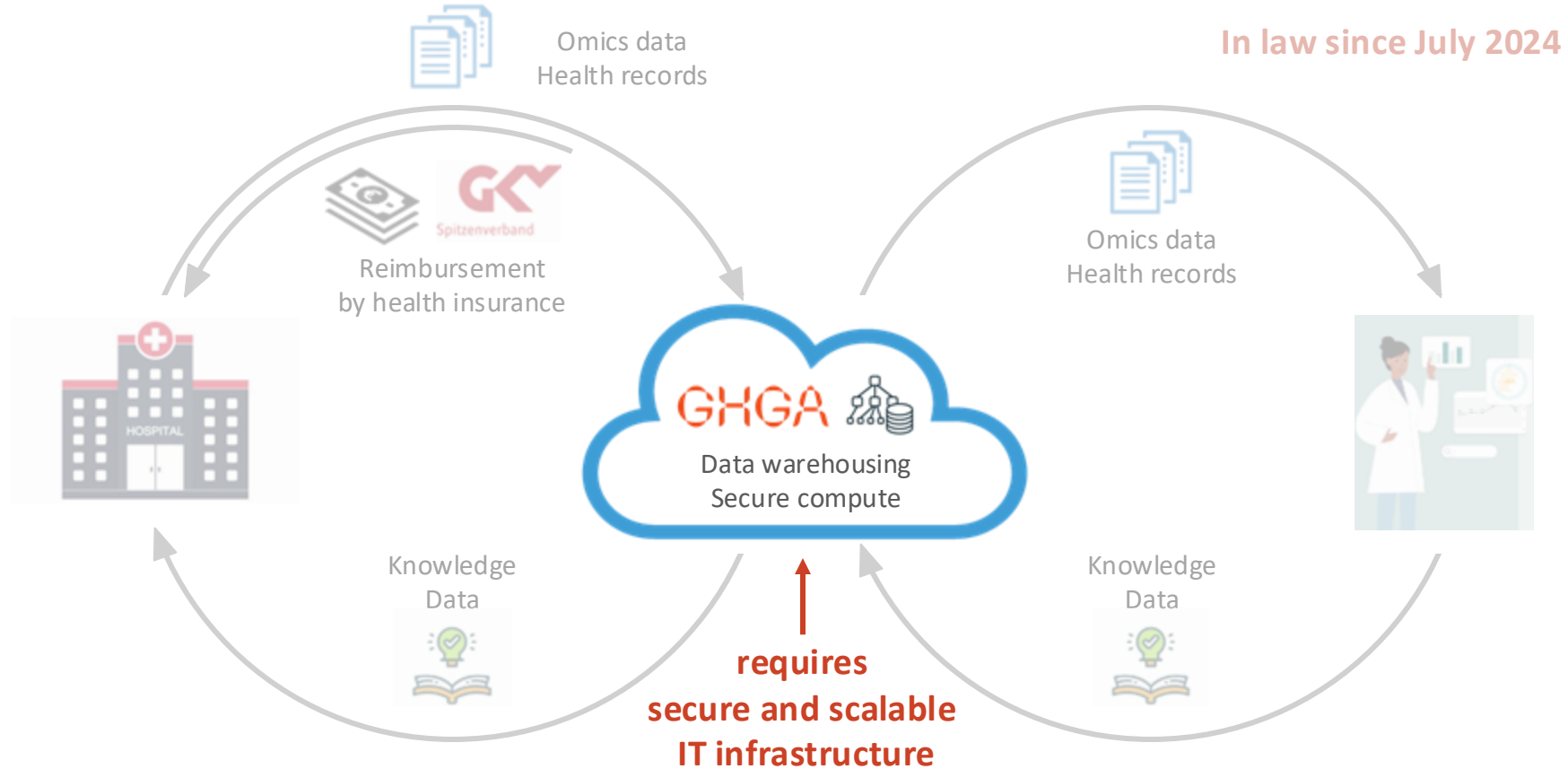


1. Legal framework
2. IT infrastructure
3. Data and Workflow standardization
4. Scalable algorithms

Genomic Medicine: a new German law to bring whole genome sequencing to routine care



Building a national learning healthcare system requires a scalable IT Infrastructure



MV GenomSeq: The German Clinical Genomics Initiative

Mission: Bring genome sequencing to routine care in **rare disease and oncology**

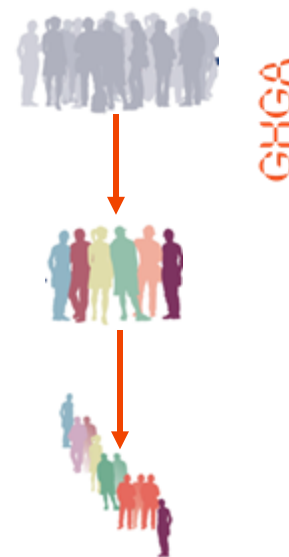
- Based on national law (§64e SGB V, 2024 – 2028)
- Funded for by health insurance and Federal Ministry of Health
- Designed to be *research compatible*
- **Generation of 90,000 whole exomes / genomes, corresponding to ~15 PB of raw and processed data**

GHGA's role

- Conception of the data platform
- **GHGA will provide genome data management ⇒ GHGA Data Hubs forsen to operate Genome Data Centers**
- Research-consented data (expected 90+%) to be shared in GHGA
- Strong financial incentive for data deposition
- Connect to international activities such as GDI / 1+MG and GA4GH



Fed. Minister of Health Prof. K. Lauterbach launches MV GenomSeq, July 2024



We are GHGA

- Institutions: 13 (Co-)Applicants, 15 Participating Institutions
- PIs: 9 (Co-)Spokespersons, 26 Participants
- Team: >40 Team members spread across 15 institutions



**A Research Data Infrastructure from
Researchers for Researchers!**

Mission Statement: ghga.de



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FOR PATIENTS

ABOUT US

NEWS & EVENTS

RESOURCES

DATA PORTAL

FAQ

We are building a **secure national omics data infrastructure** that provides a framework for the use of human genome data for **research** purposes while preventing data misuse. GHGA bridges the gap between research and medical care, and creates the opportunity to effectively use both data and technology to develop new therapies and diagnostic tools.

No Research without Data.

GHGA - The German Human Genome-Phenome Archive

We care about genomic data.

We make it usable.

We protect it.

GHGA

Closing the gap: connected healthcare in Europe

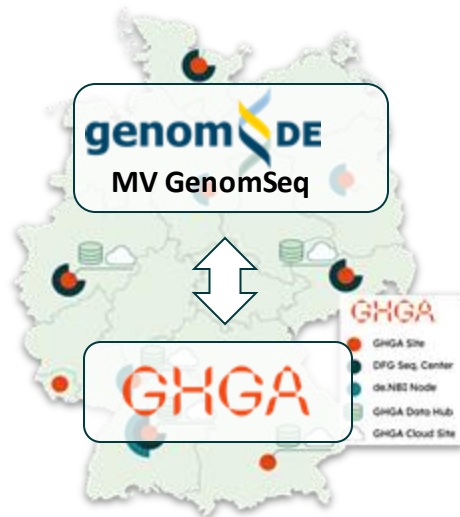
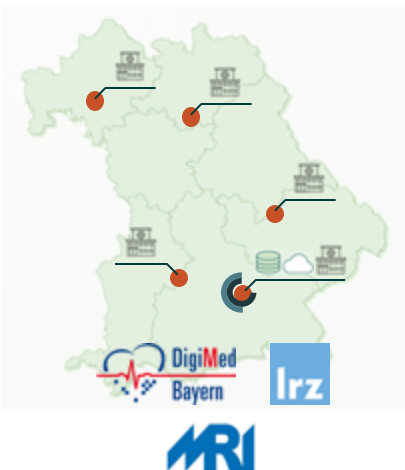
Bavaria
IT Infrastructure



Germany
Legal Framework



Europe
Interconnections

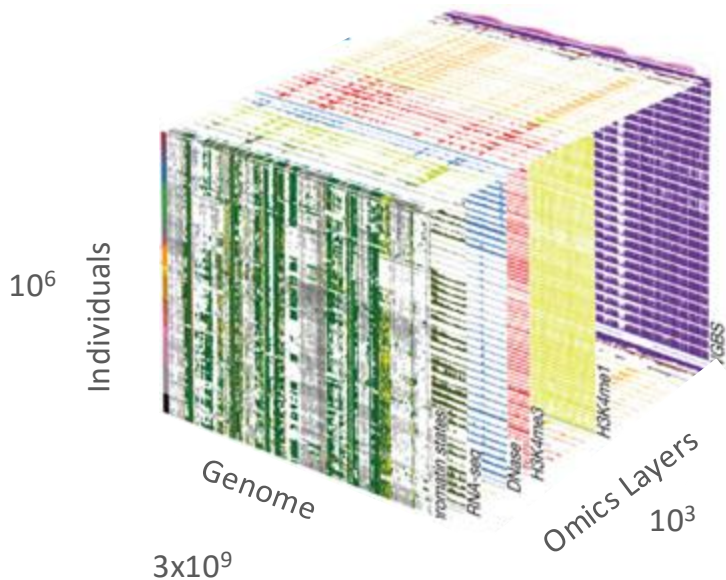


The Bavarian Genomic Data Center connects *local, national, and European* health data spaces.

Potential of ML and AI in Genomic Medicine

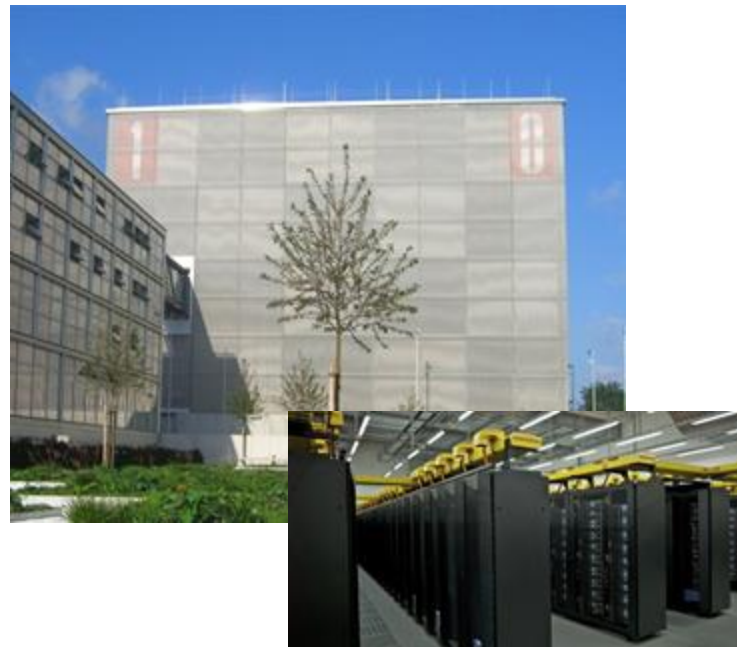
Data Cube

Harmonized and rich data



LRZ Cube

Secure compute power



In rare diseases, the outlier is the signal

>50% no causal mutation found from genome only

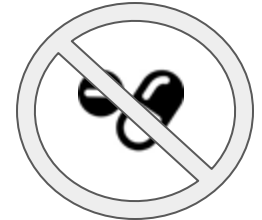
Screen for carriers



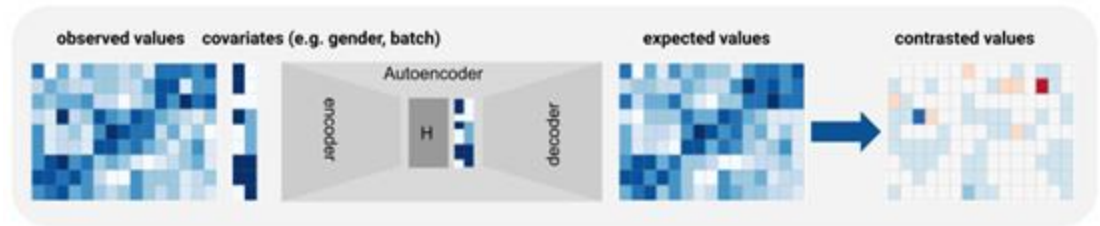
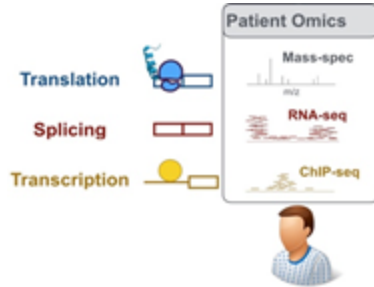
Preimplantation testing



Therapy development



Outlier detection in omics



Diagnostic rate **+15%**

US/Canada



EU



Thank you!

Data Hub Team



Christian Mertes



Florian
Hölzlwimmer



Jochen
Blesch



Juliane
Winkelmann



Julien
Gagneur



Drew
Behrens



Shounak
Chakraborty

Partners and Sponsors

