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Pharmacogenomics implementation in Germany

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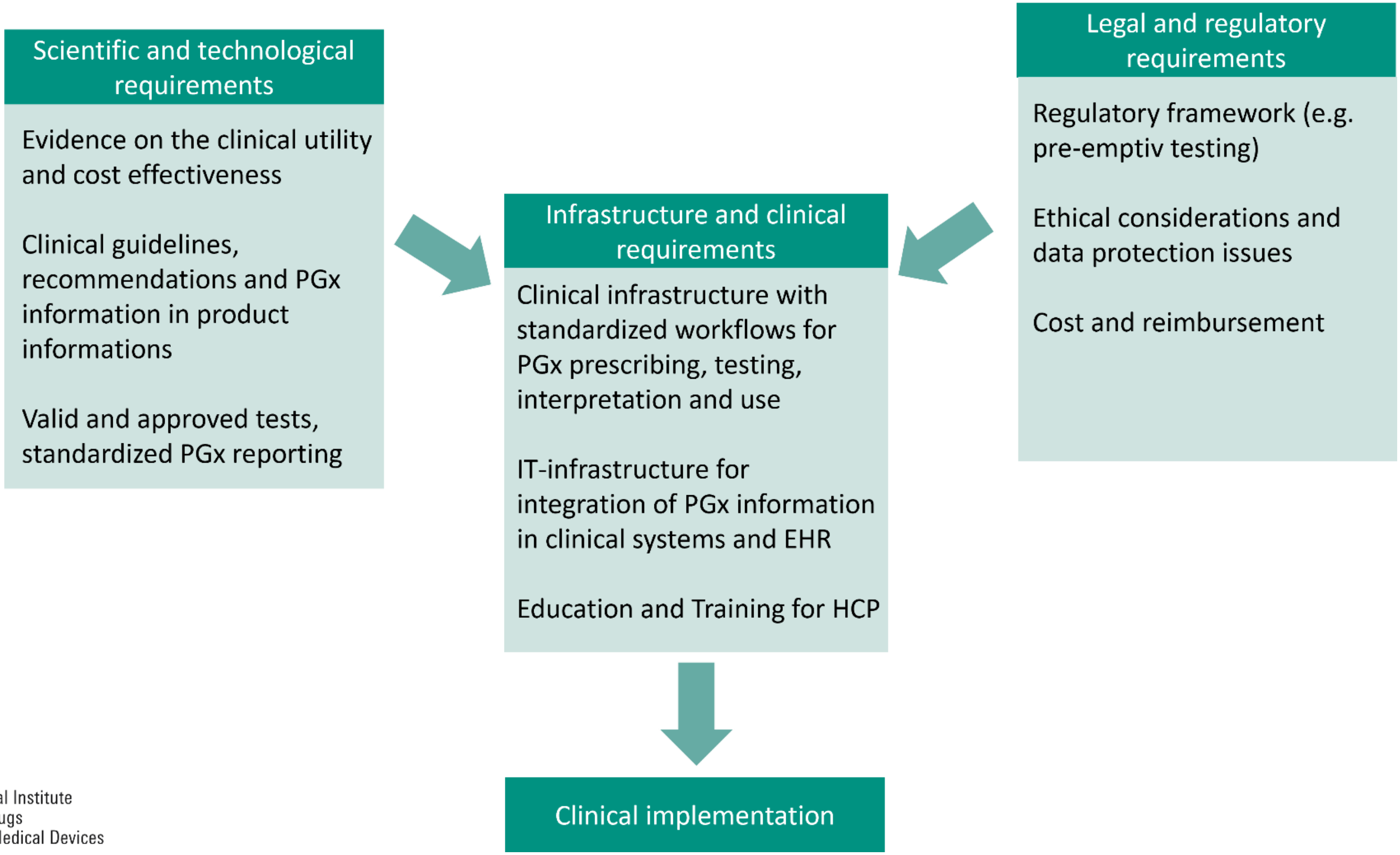
Pharmacogenomics implementation

Evidence on the utility of PGx to improve drug therapy is obvious. Nevertheless, translation in the clinical practice and implementation of PGx in healthcare is still suboptimal.



Scientific and technological requirements

Evidence on the clinical utility and cost effectiveness

Clinical guidelines, recommendations and PGx information in product informations

Valid and approved tests, standardized PGx reporting

Legal and regulatory requirements

Regulatory framework (e.g. pre-emptiv testing)

Ethical considerations and data protection issues

Cost and reimbursement

Infrastructure and clinical requirements

Clinical infrastructure with standardized workflows for PGx prescribing, testing, interpretation and use

IT-infrastructure for integration of PGx information in clinical systems and EHR

Education and Training for HCP

Clinical implementation

Current situation in Germany

Overall, implementation of PGx in the health care system is weak

PGx is mainly used in specialized centers (e.g. university hospitals) and in the framework of research studies.

Implementation in the outpatient sector is very low.

Clinical disciplines with best practice in implementation of PGx:
Psychiatry and Oncology

Major barriers for clinical implementation:

- Suboptimal integration in clinical workflows
- Lack of education and knowledge among HCP about PGx
- Ethical and data protection consideration
- Cost and lack of reimbursement

Suboptimal integration in clinical workflows

Absence of regulations, guidelines and local workflows in respect to PGx testing

Low availability of PGx testing in hospital setting and outpatient sector
- Lack of testing infrastructure
- Time delay until reporting of test results

Heterogenous IT-infrastructure in hospitals and outpatient sector
- No integration of PGx result in clinical information systems and EHR
- Low interoperability between healthcare sectors

Lack of knowledge and low experience among HCP in respect to genomic medicine and PGx

- ➢ **Engagement of different stakeholders in the implementation process required:**
  - ➢ **Hospital leadership and administration**
  - ➢ **Physicians**
  - ➢ **Clinical Pharmacist**
  - ➢ **Laboratory, Bioinformatics, IT**

Ethical consideration and data protection issues

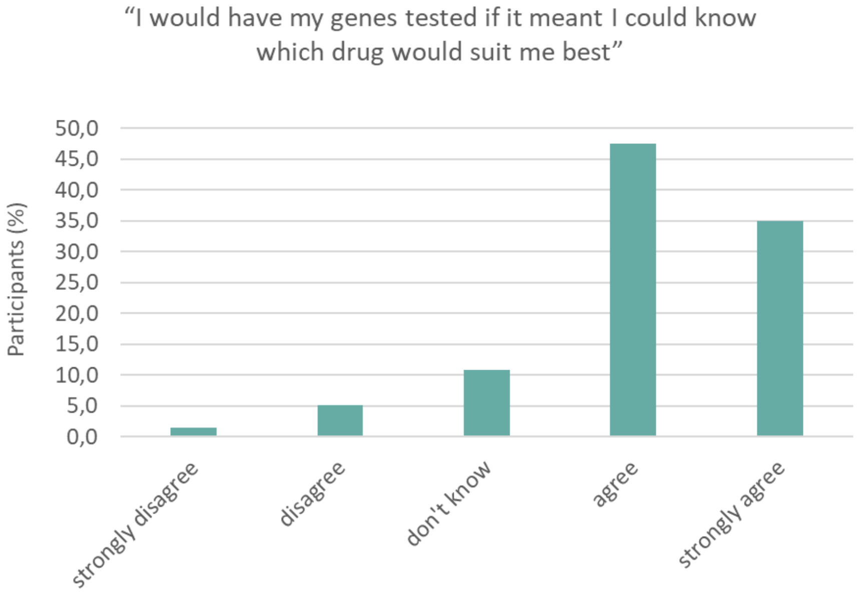
Comparatively strict regulation for genetic diagnostics

- Genetic diagnostics law
- Requirement of informed consent
- Genetic counseling by human geneticists

Concerns about the potential misuse of "genetic" data

- Narrow interpretation of GDPR
- Opt-in for genetic testing
- Pre-emptive PGx testing?

Acceptance of PGx among patients is high, even if patients lack knowledge about PGx.



EMPAR study: 10.000 patients with cholesterol lowering drugs, anticoagulant therapy and patients who suffered ADR (Huebner et al. 2024, unpublished data)

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Cost and reimbursement

No general reimbursement of PGx by the public health insurance system*.

- Lack of PGx information in drug labels
  → PGx test is required, recommended, actionable or informative?
- Lack of evidence about the cost effectiveness of PGx in different indications

➢ Directive or regulation on reimbursement

PGx tests according to the catalogue of billable services:

- **CYP2C9: Siponimod** - secondary progressive multiple sclerosis (CYP2C9*1, CYP2C9*2 und CYP2C9*3)
- **CYP2C19: Mavacamten** - symptomatic hypertrophic obstructive cardiomyopathy (CYP2C19*2 und CYP2C19*3)
- **CYP2D6**: **inhibitors of glucocerebroside synthase** - Morbus Gaucher Type 1
- **DPYD**: prior to systemic therapy with **5-fluoroΥracil** (c.1905+1G>A, c.1679T>G, c.2846A>T und c.1236G>A/HapB3)
- **UGT1A1:** before systemic therapy with an **irinotecan-containing drug** (UGT1A1*6 und UGT1A1*28)

**exceptional cases: Individual application for reimbursement possible*

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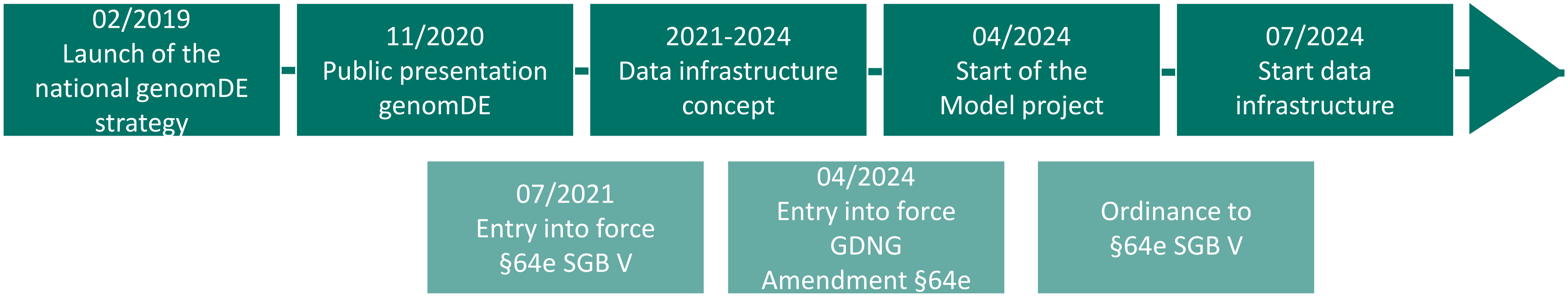
National strategy genomic medicine: genomDE

The aim of the genomDE strategy is to introduce genome medicine into standard care to establish genetically confirmed diagnoses and enable preventive measures and tailored therapies



02/2019
Launch of the national genomDE strategy

11/2020
Public presentation genomDE

2021-2024
Data infrastructure concept

04/2024
Start of the Model project

07/2024
Start data infrastructure

07/2021
Entry into force §64e SGB V

04/2024
Entry into force GDNG
Amendment §64e

Ordinance to §64e SGB V

Model project §64e SGB V: Model project for comprehensive diagnostics and therapy identification using genome sequencing for rare and oncological diseases

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Model project §64e SGB V

Aim: Improve the care of patients suffering from **rare diseases and oncological diseases** by establishing genome sequencing in **routine healthcare**

Timeframe: 2024 – 2029

Indications: Rare diseases, oncological diseases, hereditary predisposition syndromes

Service providers: 25 university hospitals and centers with expertise in genomic medicine in the fields of oncology and rare disease

Reimbursement: By health insurance companies (public and private)

- confirmation token that proofs data submission is required

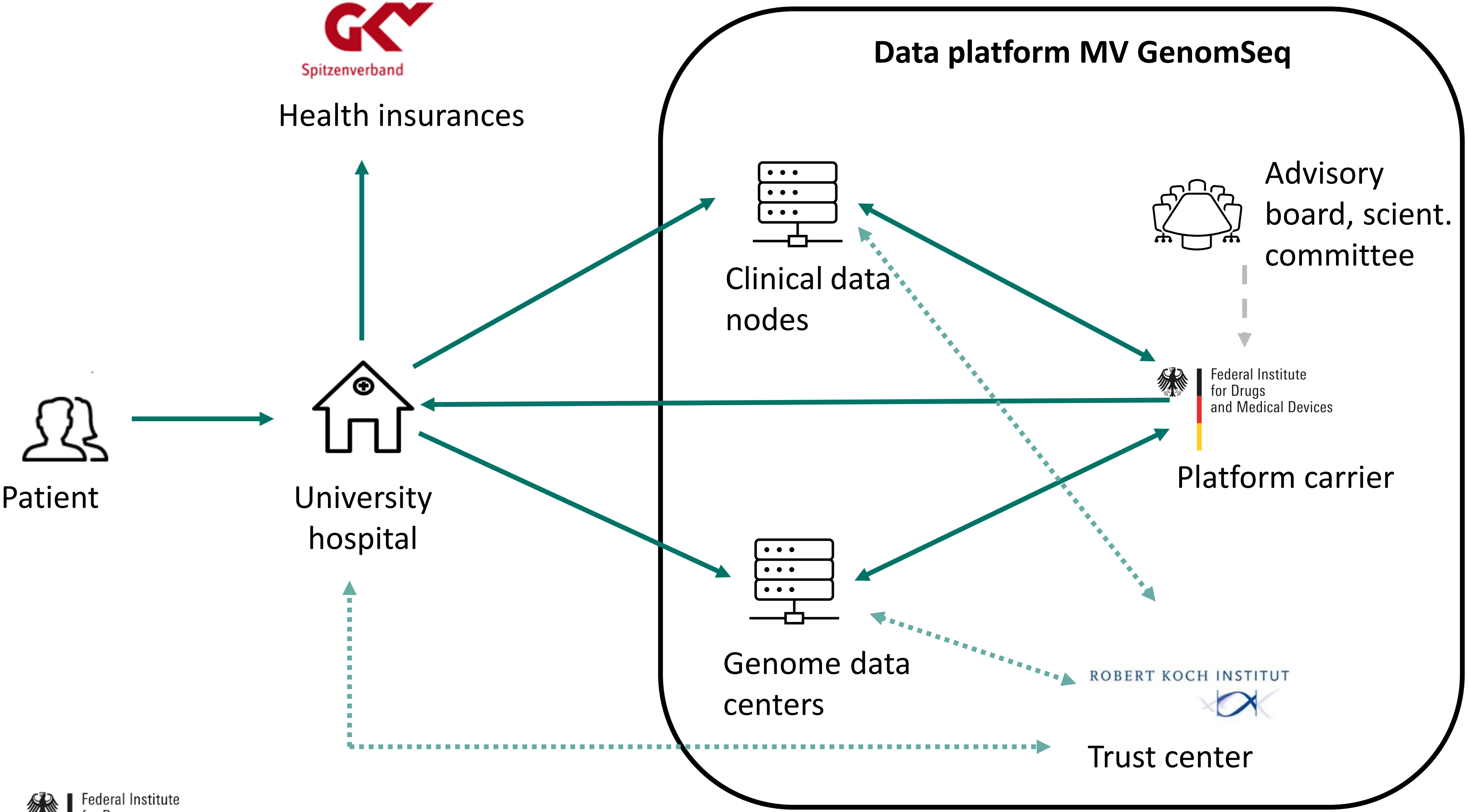
Model project §64e SGB V

Core element:
Establishment and operation of a data infrastructure in which genomic and clinical data from patients is stored and made available for healthcare and research
→**MV GenomSeq**

Data infrastructure:

- **Central** platform governance (BfArM), independent **central** trust center (RKI)
- Data storage is **decentralized** in clinical data nodes and genome data centers
- Builds on existing data infrastructures from clinical networks established at university hospitals and genomic data infrastructure projects.
- High quality, harmonized and interoperable data for patient care and research

Model project §64e SGB V



GKV Spitzenverband

Health insurances

Data platform MV GenomSeq

Clinical data nodes

Advisory board, scient. committee

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Platform carrier

Patient

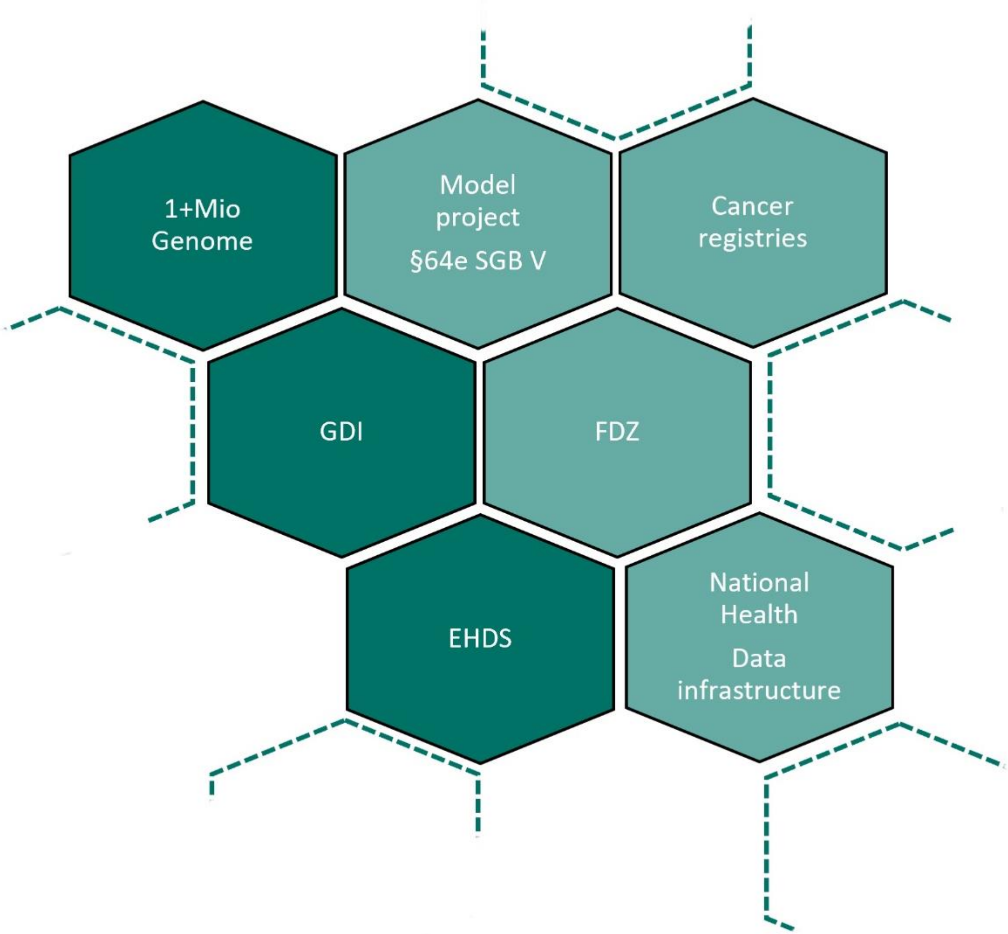
University hospital

Genome data centers

ROBERT KOCH INSTITUT

Trust center

Perspective: Model project §64e SGB V



- Positive evaluation → Transfer of genome sequencing to standard care
- Facilitate broader health care implementation of genome medicine (e.g. additional indications, PGx)
- Linkage of MV GenomSeq to cancer registries, Health Data Lab (FDZ Gesundheit), national research data infrastructures (e.g. GHGA), European genome initiatives (e.g. GDI) and the European Health Data Space

What can we learn from the model project §64e for clinical PGx implementation in Germany?

- Relevant national experts involved in discussions on standards
- For participation in-house expertise required (clinical, diagnostics, bio-informatics)
- Defined patient journey in all participating university hospitals
- Standardized IT- infrastructure
- Data for healthcare, research and evaluation

→ **Harmonization and standardization of clinical workflows**

→ **Evidence for the implementation in standard care will be generated**

- A legal framework regulating governance, data protection and data use
- Reimbursement enshrined by law, costs and provided service were negotiated between university hospitals and public health insurance companies

→ **Binding national regulation**

→ **Issues on data protection and reimbursement are solved**

Thank you very much for your attention!

Contact

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audit beruf**und**familie

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