



Roadmap for genomics in healthcare

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B1MG WP5

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A Roadmap for Genomics in Healthcare

- **Output of B1MG (WP5) & purposes to stay useful and relevant for the future**
 - **Aims to guide & support** countries and HCS in establishing their own path towards implementing genomics in clinical care
 - It is supported by the **1+MG MLM**
 - **Gathers knowledge** (1+MG & B1MG and also other PM initiatives) and the goal is to keep it as a **live document**, regularly updated
- Will be available soon (1+MG trust framework)



A Roadmap for Genomics in Healthcare



Implementation of genomic medicine in healthcare

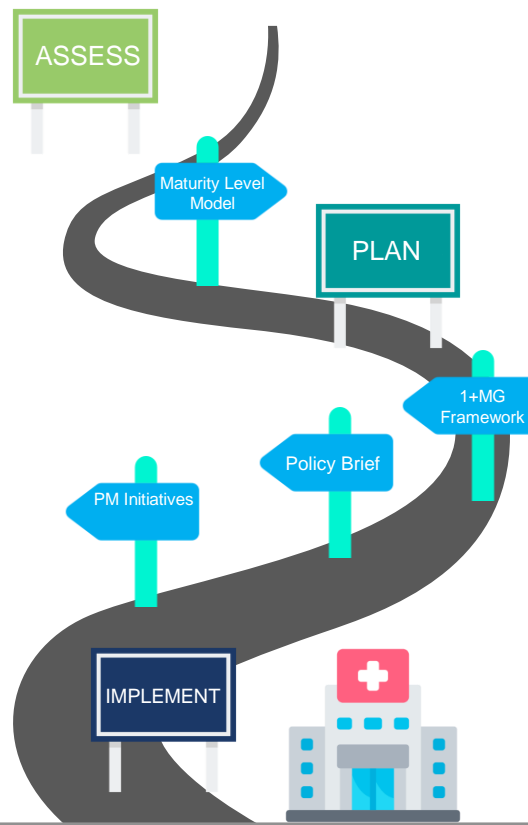
The implementation of genomic medicine in healthcare systems will bring us closer to making personalised medicine a reality, with major socioeconomic benefits.

Through genomic medicine, citizens and patients will be able to widely benefit from genomic data analysis for accurate and timely diagnosis, more effective treatments with fewer adverse events and, in the near future, accurate profiling for disease prevention.

However, implementation of genomics in healthcare is complex and requires adjustments in the governance, structure and organisation of health services, as well as dedicated investments. Moreover, the progress of implementation varies with country context.

Facilitating dialogue and cooperation among countries for capacity building and sharing of best practices is therefore key to promoting equity in access to Personalised Medicine (PM) across Europe.

Development of a Roadmap for Genomics in Healthcare

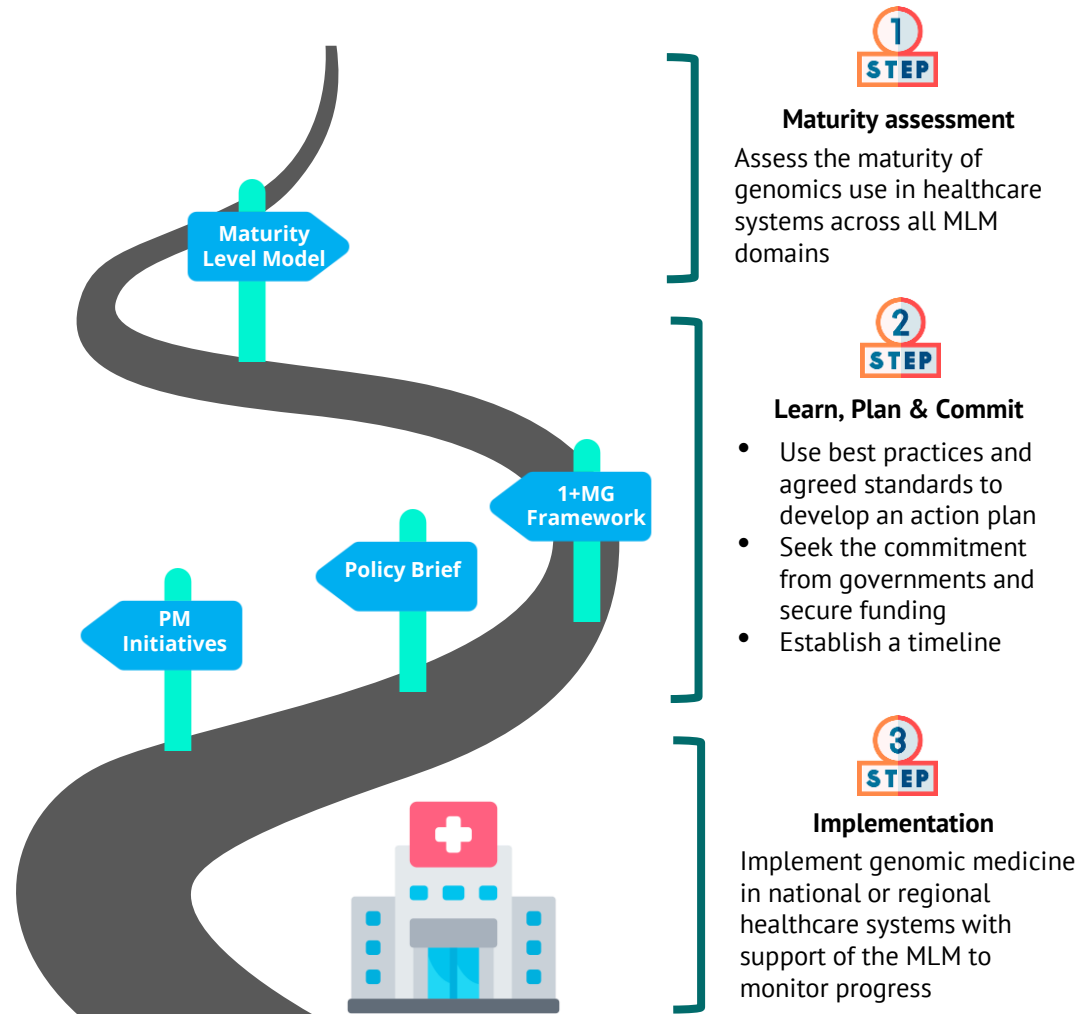


This roadmap was developed in the context of the 1+Million Genomes (1+MG) initiative activities to promote the societal impact of genomic medicine. It is based on shared knowledge and experience from experts, in various crucial areas of genomic medicine, across Europe and beyond.

The roadmap is supported by the 1+MG Maturity Level Model (MLM) framework. This MLM is a tool for health systems to self-assess their maturity level regarding the implementation of genomics in healthcare, and to define a path to optimization according to a common matrix.

As such, the MLM aims to promote and facilitate the adoption of genomics in clinical practice, close equity gaps in access across Europe, and make personalised medicine accessible to all citizens and patients in Europe.

Roadmap steps



ASSESS - the 1+MG Maturity Level Model



What is it?

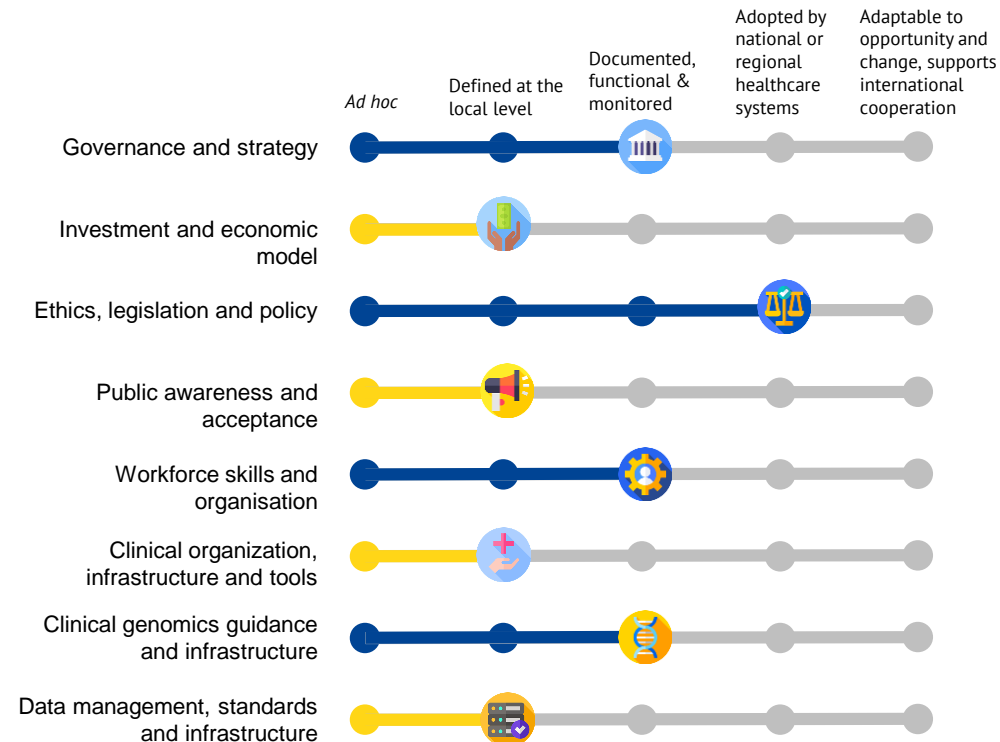
The 1+MG MLM is a tool for healthcare systems to self-evaluate the level of maturity of their genomic medicine practices according to a common matrix, and to define a path to optimization.



How?

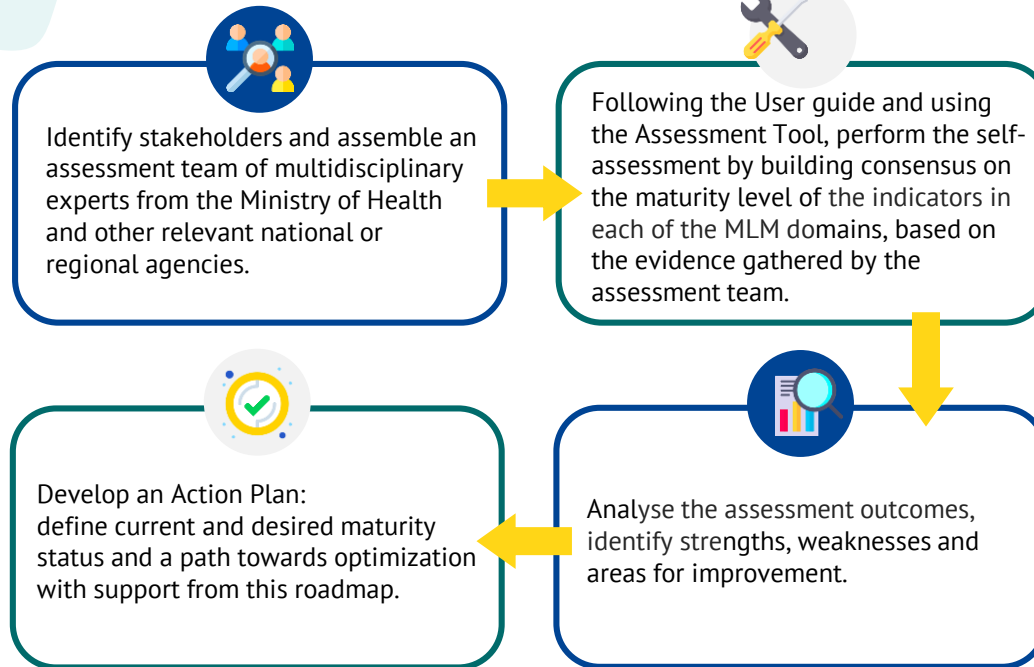
The MLM assesses indicators in eight key domains for the implementation of genomics in healthcare. Maturity of indicators in each domain is assessed by selecting one of five pre-defined levels for each indicator.

The five maturity levels are indicative of a maturity progression, from a non-existent or *Ad hoc* level of implementation to a high level of maturity, characterized by a system fully adopted by the healthcare system, adaptable to opportunity and change, and in support of international cooperation.



Four phases to maturity level assessment

1
STEP



Using the Maturity Level Model

Visit the MLM here



Maturity assessment example – Domain 1: Governance and Strategy

Indicator	ML1	ML2	ML3	ML4	ML5
Governance: Country/region has dedicated governance for genomics in healthcare	No dedicated governance	Elements of governance exist but are not fully functional	Scope has been defined but are still under development	A fully operating governance body has been established and monitored based on a work plan	An institutionalised governance body open for international collaboration
Priority: Genomics in healthcare is established as a priority at national/regional level	Not included in health plans	Inclusion in relevant health plans under discussion	Inclusion in relevant health plans	Implemented as part of health and other plans (e.g. education, research, ...)	Implemented in health and other plans, and is periodically evaluated
Strategy: There is a national/regional strategy for genomics in healthcare with an implementation plan	No strategy	A strategy for implementation plan is under discussion	A strategy for implementation plan is developed and approved	The strategy for genomics in healthcare is implemented	The strategy is implemented and aligned with European and international strategies

ASSESS - Benchmarking



MLM in real-life settings

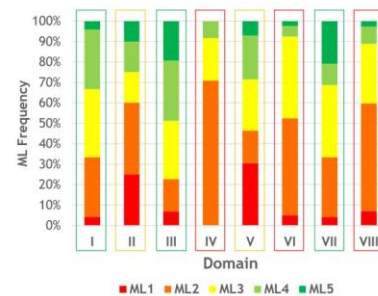
The 1+MG MLM has been used by eight European countries in a pilot exercise, to establish the maturity levels of genomic medicine in their healthcare systems: Belgium, Denmark, Finland, Ireland, Italy, Lithuania, Portugal, and Spain.

The maturity level assessment of these healthcare systems was fundamental for identifying areas of investment, exchanging best practices and harmonising procedures, towards bridging development gaps across Europe and achieving equity in access to Personalised Medicine for all citizens.



Mapping maturity in Europe

The pilot exercise allowed the identification of common European strengths and weaknesses in the MLM domains, as well as maturity asymmetries across Europe.

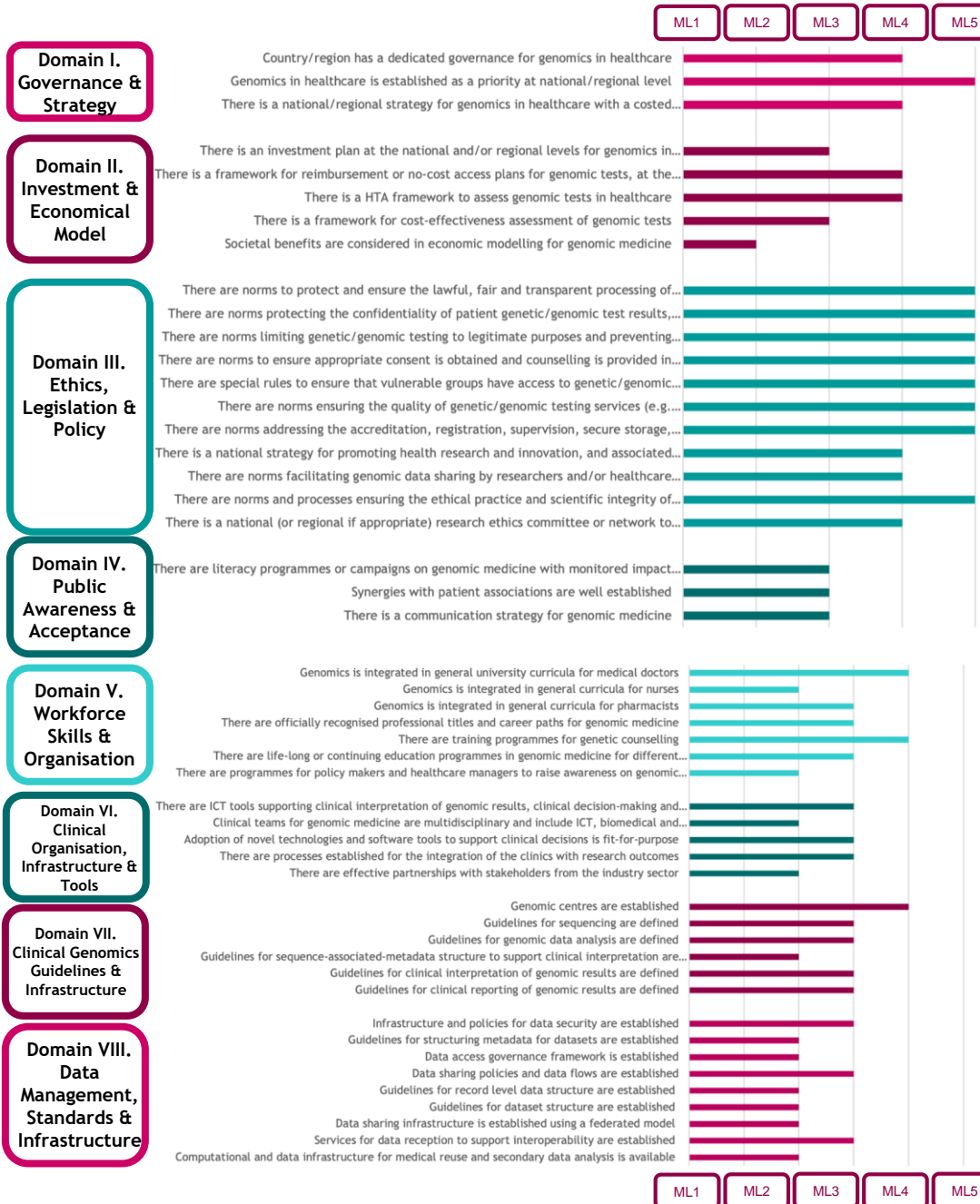


Benchmarking maturity

Benchmarking the MLM allows healthcare systems to understand how they are performing compared to the median maturity of other systems, for each indicator in the eight MLM domains. The benchmarking of maturity will promote sharing of best practices in a continuous improvement effort.

ASSESS - Benchmarking from 8 healthcare systems

1
STEP



LEARN, PLAN & COMMIT

- Learn from country exchange visits



- Exchange of best practices and experiences is fundamental for equitable access to genomic medicine for every citizen across Europe.
- Three Country Exchange Visits (CEV) carried out by the B1MG project led to Policy Recommendations on Key Issues for Genomics in Healthcare, providing guidance to build efficient and sustainable genomic medicine strategies:
 1. Patient and citizens trust and engagement are critical aspects for a fair and valuable implementation of genomic medicine
 2. Infrastructure for implementation of genomics in clinical practice requires a safe and trusted environment for collection and use of genomic and health data
 3. Ethical and legal frameworks are needed to ensure secure and transparent data collection, analysis and use, protecting citizens' and patients' rights
 4. Training of healthcare professionals guarantees a competent and knowledgeable workforce
 5. Synergies among healthcare, research and industry benefit citizens and patients, healthcare services, health economy and society at large

Country Exchange Visits

– United Kingdom recommendations



- Political support is a powerful enabler for the implementation of genomics in healthcare.
- The concept of a learning healthcare system was implemented to ensure that results from routine genomic testing done within the NHS would feed back into research.
- A well informed and engaged workforce requires investment in developing professionals, such as genetic counsellors, and new professions, like clinical scientists, and medical informaticians.

[Watch the UK Country Visit](#)

Country Exchange Visits – Finland recommendations

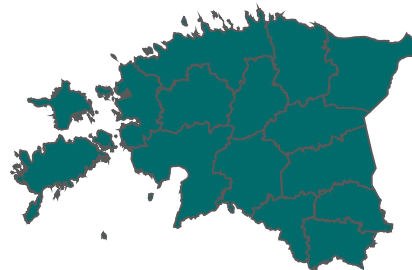
2
STEP

- The National Genome Centre is the public authority responsible for the Finnish population genome database, promoting equity and the responsible use of genomic data.
- The Finnish Health Sector Growth Strategy has a political commitment of ministries from diverse sectors.
- A strong legal and ethical framework to ensure citizens' trust is a priority.
- A stable collaboration between healthcare, research and industry, recognises the private sector as a valuable partner.



[Watch the Finland Country Visit](#)

Country Exchange Visits – Estonia recommendations



[Watch the Estonia Country Visit](#)

- A health portal links multiple electronic databases, including clinical diagnosis, prescription and billing, in a central technical infrastructure.
- Long-term investment in communication campaigns resulted in widespread public support, with a significant percentage of the Estonian population represented in the national Biobank and expecting benefits from genomic data use.
- The Human Genes Research Act provides a legal framework dating back to 2000 when its national Biobank was established.

LEARN, PLAN & COMMIT



- Use available guidelines and best practices to optimise the use of genomic medicine in healthcare systems

Domain I. Governance & Strategy

- Policy Brief: [Genomics in healthcare: key issues for implementation](#)
- Country Exchange Visit (CEV) Report
- CEV to the UK: [Genomics in the UK - a tale of research and healthcare](#)
- CEV to Estonia: [Governance of personalised medicine in Estonia](#)
- CEV to the UK: [Implementing genomics in the NHS](#)



Guidance from other initiatives related to Personalised Medicine

- [ICPerMed](#)
- [ICPerMed Vision Paper](#)
- [Example from Estonia \(ICPerMed\)](#)
- [Example from France \(ICPerMed\)](#)
- [Example from Sweden \(ICPerMed\)](#)

Domain II. Investment and economic models

- CEV to the UK: [Genomics England, a strong collaboration with the NHS](#)
- 1+MG policy brief: [Recommendations from the 1+MG HEOR workshop](#)
- 1+MG workshop summary paper: [Review of EU initiatives related to the implementation of whole genome sequencing into clinical practice](#)
- B1MG Deliverable D5.3: [Economic models methodology and case studies](#)



Guidance from other initiatives related to Personalised Medicine

- [HEcoPerMed](#)
- [Short scenarios for future PM \(HEcoPerMed\)](#)
- [Health economic perspective on PM \(HEcoPerMed\)](#)
- [Case studies on health economic modelling \(HEcoPerMed\)](#)
- [SRIA on PM \(EP PerMed\)](#)



Domain III. Ethics, legislation and policy



- 1+MG [Consent recommendations: For stakeholder interaction and discussion](#)
- B1MG policy document: [Recommendations for 1+MG Minimal Standards for Inclusion of Special Subjects](#)
- B1MG policy document: [Policy document on minimal standards for feedback provision of results of research](#)
- CEV to Finland: [Legislation-authorisations for companies/research groups to use genomics data](#) & [Regulating the unknown, a guide to regulating genomics for health policy-makers](#)

Guidance from other initiatives related to Personalised Medicine



- [Consent Policy GA4GH](#)
- [Consent toolkit GA4GH](#)
- [Data privacy policy GA4GH](#)
- [Ethics policy GA4GH](#)
- [Data sharing GA4GH](#)
- [SRIA on PM \(EP PerMed\)](#)
- [NAGEN 1000 \(ICPerMed\)](#)

Domain IV. Public awareness and acceptance

- CEV to the UK: [Patient engagement-every data point has a face](#)
- B1MG Deliverable 1.6: [Citizen engagement and public trust in genomic data sharing](#)
- B1MG poster: [Recommendations for trustworthiness from an experts workshop organised by B1MG](#)

Guidance from other initiatives related to Personalised Medicine



- [Engaging patients and the public GA4GH](#)
- [Involving patients with CV diseases \(ICPerMed\)](#)
- [NAGEN 1000 \(ICPerMed\)](#)



Domain V. Workforce skills and organisation

- CEV to the UK: [Training and tools in genomics for the entire healthcare workforce](#)

Guidance from other initiatives related to Personalised Medicine



- [SRIA on PM \(EP PerMed\)](#)
- [Education in pharmacogenomics \(ICPerMed\)](#)
- [NAGEN 1000 \(ICPerMed\)](#)



Domain VI. Clinical organization infrastructure and tools



- CEV to the UK: [Genomics England, a strong collaboration with the NHS](#)
- CEV to Finland: [The FinnGen project: opportunities for discoveries and translation](#)
- CEV to Finland: [1+MG-the role and perspectives of industry in genomics](#)
- CEV to Estonia: [Private sector in personalized medicine in Estonia, government's view](#)



Guidance from other initiatives related to Personalised Medicine

- [PERMIT Project \(ICPerMed\)](#)
- [PERMIT recommendations \(ICPerMed\)](#)
- [SRIA on PM \(EP PerMed\)](#)
- [Pharmacogenetics-based clinical decision \(ICPerMed\)](#)
- [Adoption of EMR \(ICPerMed\)](#)
- [PERSEPHONE project and multidisciplinary teams \(ICPerMed\)](#)
- [Institutional synergies for lung cancer \(ICPerMed\)](#)
- [NAGEN 1000 \(ICPerMed\)](#)

Domain VII. Clinical genomics guidelines and infrastructure



- B1MG Deliverable 3.1: [Quality metrics for sequencing](#)



Guidance from other initiatives related to Personalised Medicine

- [Returning results GA4GH](#)
- [Sequence annotation GA4GH](#)
- [Genomic centers, example from Germany \(ICPerMed\)](#)
- [CPIC guidelines](#)
- [DPWG guidelines](#)
- [PharmGKB resources](#)
- [ACMG guidelines](#)



Domain VIII. Data management, standards and infrastructure

- B1MG Legal Taxonomy status: [Taxonomy of member state legal requirements applicable to cross-border exchange of health and genetic data](#)
- B1MG Deliverable 3.7: [Documented best practices in sharing and linking phenotypic and genetic data](#)
- 1+MG recommendation: [Recommendations for a Data Access Governance Framework](#)
- B1MG Proof of Concept: [Federated data access rare diseases proof of concept](#)



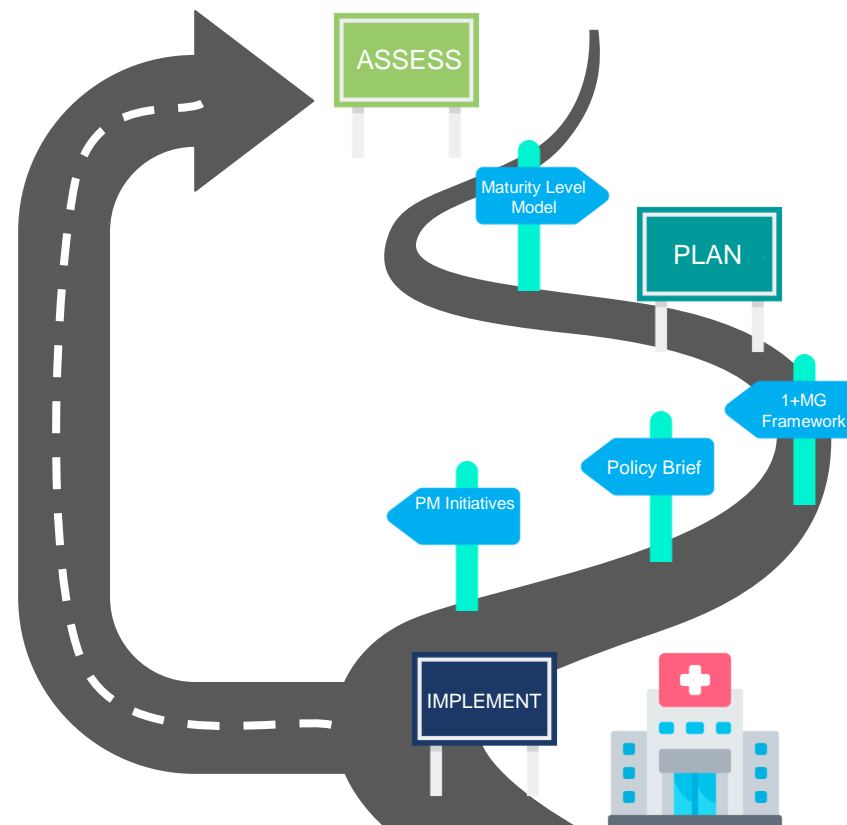
Guidance from other initiatives related to Personalised Medicine

- [Data connect API \(GA4GH\)](#)
- [Interoperable data, the Swiss example \(ICPerMed\)](#)
- [Resource Entitlement Management System REMS](#)
- [Life Science AAI](#)
- [Federated EGA \(FEGA\)](#)
- [htsget](#)

Implementation

This roadmap, with assessment and suggested guidance documents, will help health systems define the roadmap towards implementation of genomics into healthcare. The MLM Domain indicators and corresponding maturity levels will also provide support to monitor progress along the path to higher maturity.

This process can be an iterative process. The MLM is designed to be performed at regular intervals, consistently assessing the advancement over time, helping to identify areas of strength and potential bottlenecks, as well as solutions. This roadmap is intended to be a living document, regularly updating sources of guidance overtime. It thus offers the opportunity to continually access up to date best practices from the international community.



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