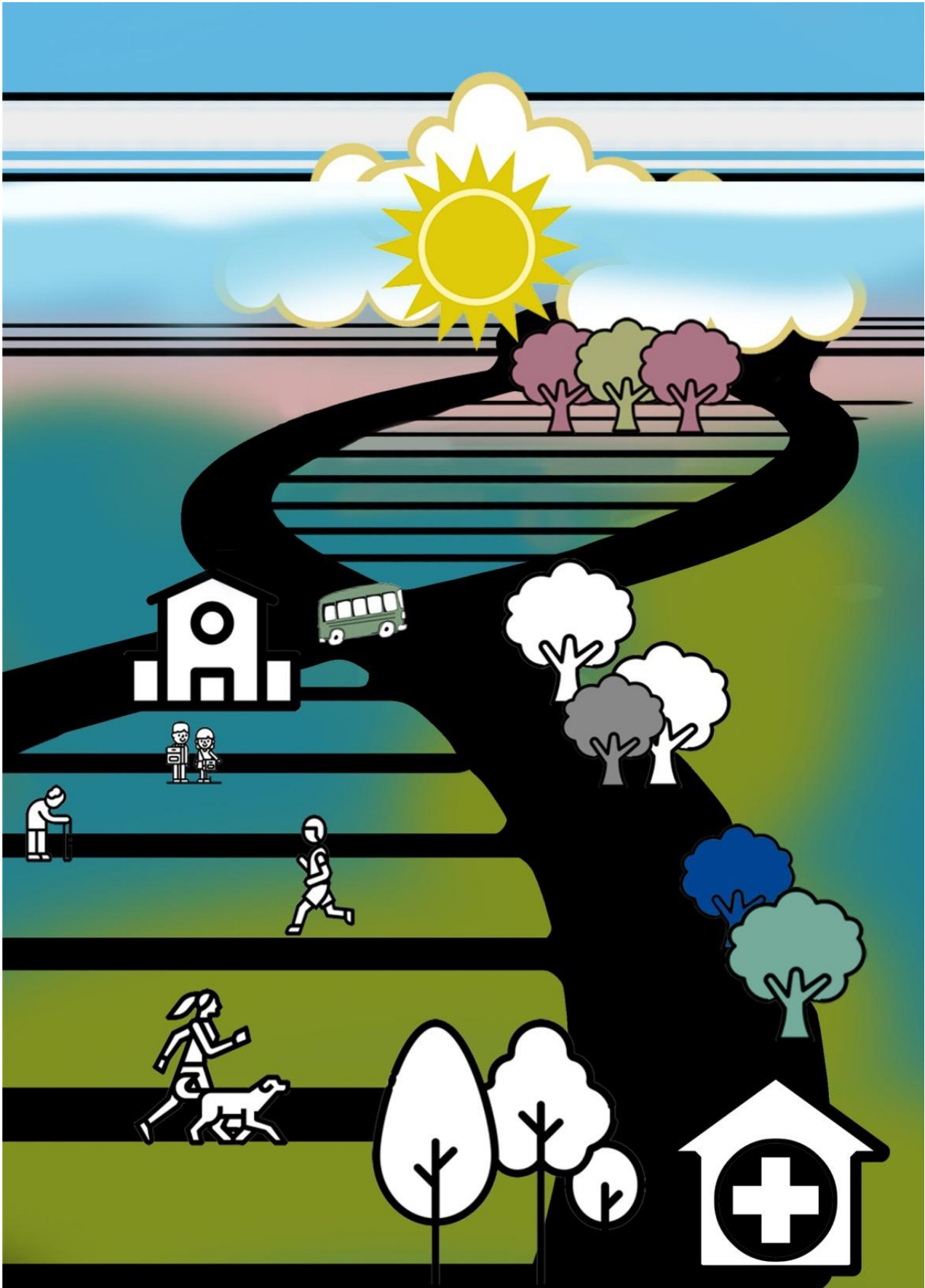# A Roadmap for Genomics in Healthcare

# 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.

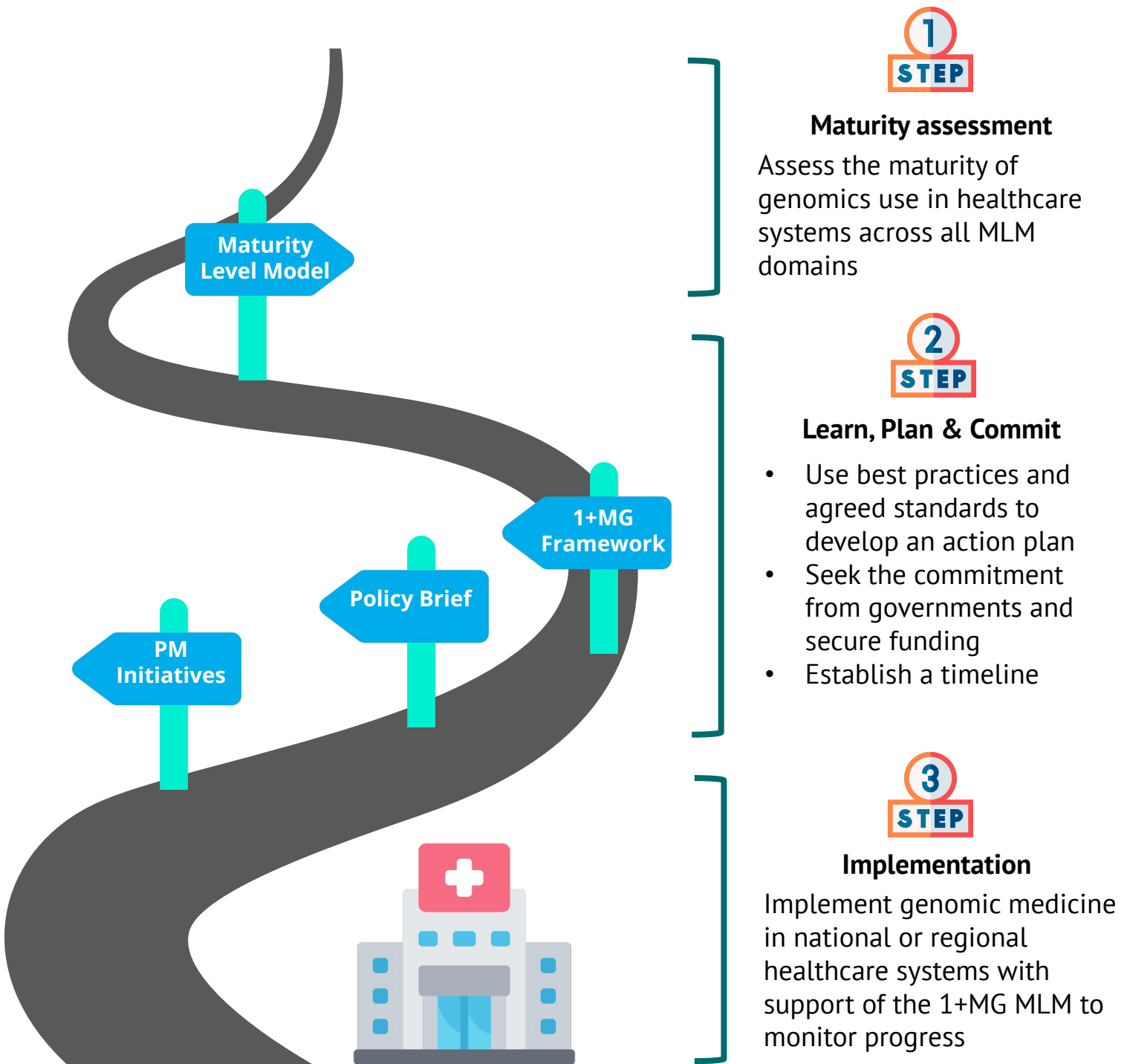# Development of a Roadmap for Genomics in Healthcare

ASSESS

Maturity Level Model

PLAN

1+MG Framework

Policy Brief

PM Initiatives

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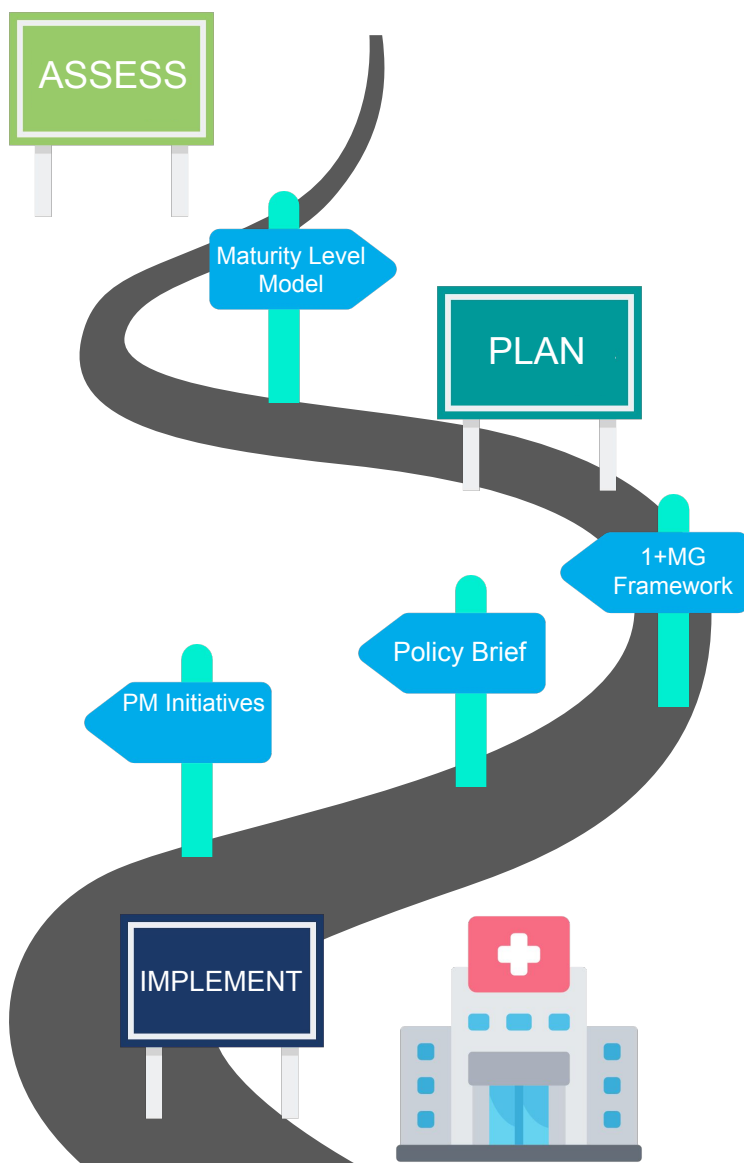
This roadmap was developed in the context of the 1+Million Genomes (1+MG) initiative activities to promote the societal impact of genomic medicine. It is based on shared knowledge and experience from experts, in various crucial areas of genomic medicine, across Europe and beyond.

The roadmap is supported by the 1+MG Maturity Level Model (MLM) framework. This 1+MG 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 1+MG 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

Maturity Level Model

1+MG Framework

Policy Brief

PM Initiatives

1 STEP

Maturity assessment
Assess the maturity of genomics use in healthcare systems across all MLM domains

2 STEP

Learn, Plan & Commit

- Use best practices and agreed standards to develop an action plan
- Seek the commitment from governments and secure funding
- Establish a timeline

3 STEP

Implementation
Implement genomic medicine in national or regional healthcare systems with support of the 1+MG MLM to monitor progress

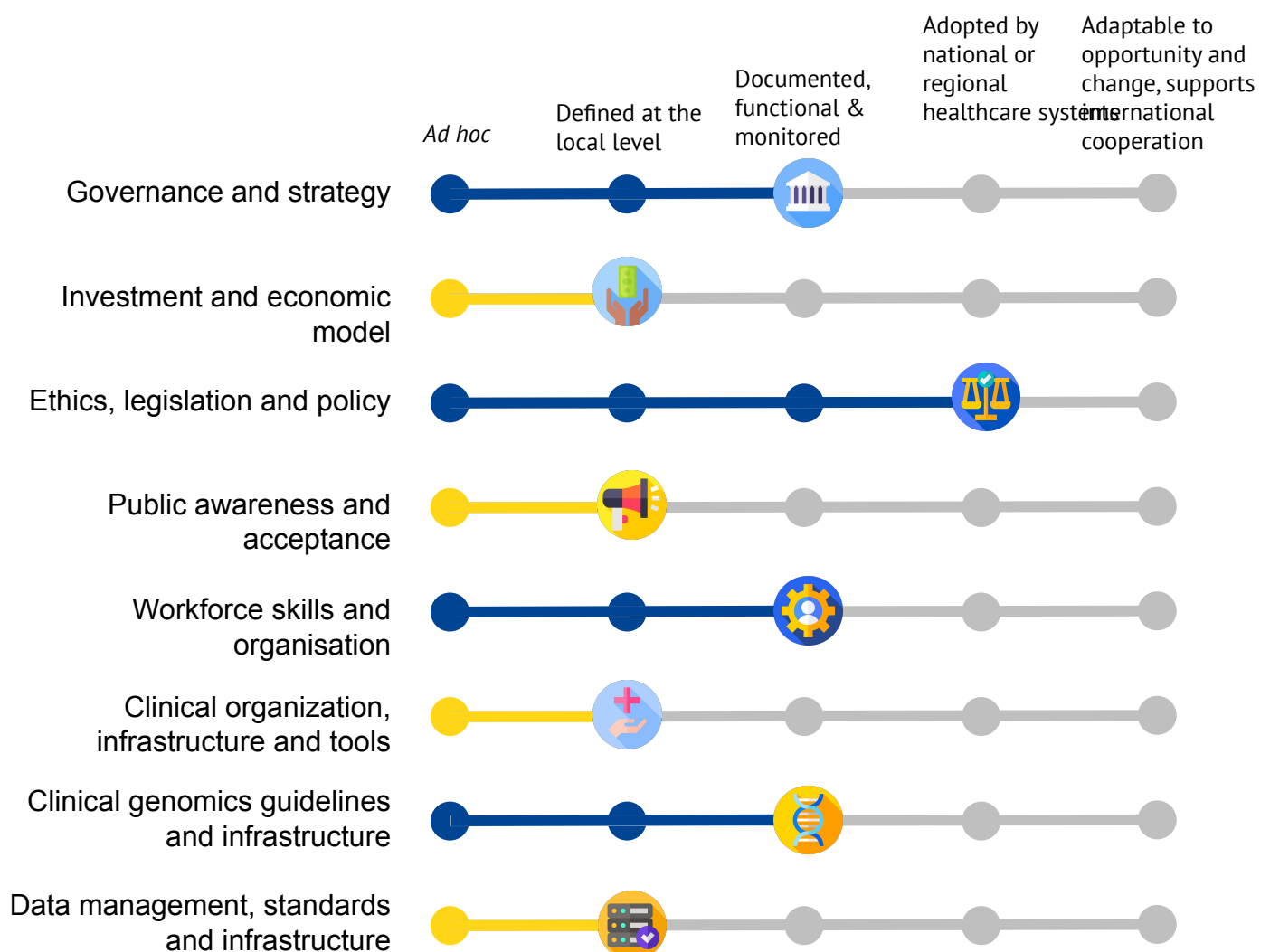# ASSESS - the 1+MG Maturity Level Model

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 predefined 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.

|	*Ad hoc*	Defined at the local level	Documented, functional & monitored	Adopted by national or regional healthcare systems	Adaptable to opportunity and change, supports international cooperation
Governance and strategy	●	●	●	○	○
Investment and economic model	●	●	○	○	○
Ethics, legislation and policy	●	●	●	●	○
Public awareness and acceptance	●	●	○	○	○
Workforce skills and organisation	●	●	●	○	○
Clinical organization, infrastructure and tools	●	●	○	○	○
Clinical genomics guidelines and infrastructure	●	●	●	○	○
Data management, standards and infrastructure	●	●	○	○	○

Using the Maturity Level Model

Identify stakeholders and assemble an assessment team of multidisciplinary experts from the Ministry of Health and other relevant national or regional agencies.

Following the User guide and Glossary, and using the Assessment Tool, perform the self-assessment by building consensus on the maturity level of the indicators in each of the MLM domains, based on the evidence gathered by the assessment team.

Analyse the assessment outcomes, identify strengths, weaknesses and areas for improvement.
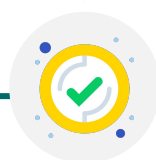
Develop an Action Plan:
define current and desired maturity status and a path towards optimization with support from this roadmap.

Using the Maturity Level Model

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 for genomics in healthcare	Elements of governance exist but are not fully functional	Scope of governance for genomics has been defined but elements are still under development	There is a governance body that is fully operating, led centrally, and activities are monitored based on a work plan	Governance body is institutionalised, recognised as the lead for genomics in healthcare, and is open to novel developments and supportive of international cooperation
Priority: Genomics in healthcare is established as a priority at national/regional level	Genomics in healthcare is not included in national/regional health plans	Inclusion of genomics in healthcare in relevant national/regional health plans is under discussion	Genomics in healthcare is included in relevant national/regional health plans	Genomics in healthcare is implemented as part of national/regional health and other relevant plans (e.g. education, research)	Genomics in healthcare is implemented in health and other relevant plans, and is periodically evaluated for optimisation, taking into account novel developments at the international level
Strategy: There is a national/regional strategy for genomics in healthcare with an implementation plan	No genomics in healthcare strategy with costed implementation plan	A strategy for genomics in healthcare with costed implementation plan is under discussion	A costed implementation plan for genomics in healthcare is developed and approved	The national/regional strategy for genomics in healthcare is under implementation	The national/regional strategy for genomics in healthcare is implemented, with monitoring and long term resources and aligned with European and international strategies

Visit the MLM here

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.
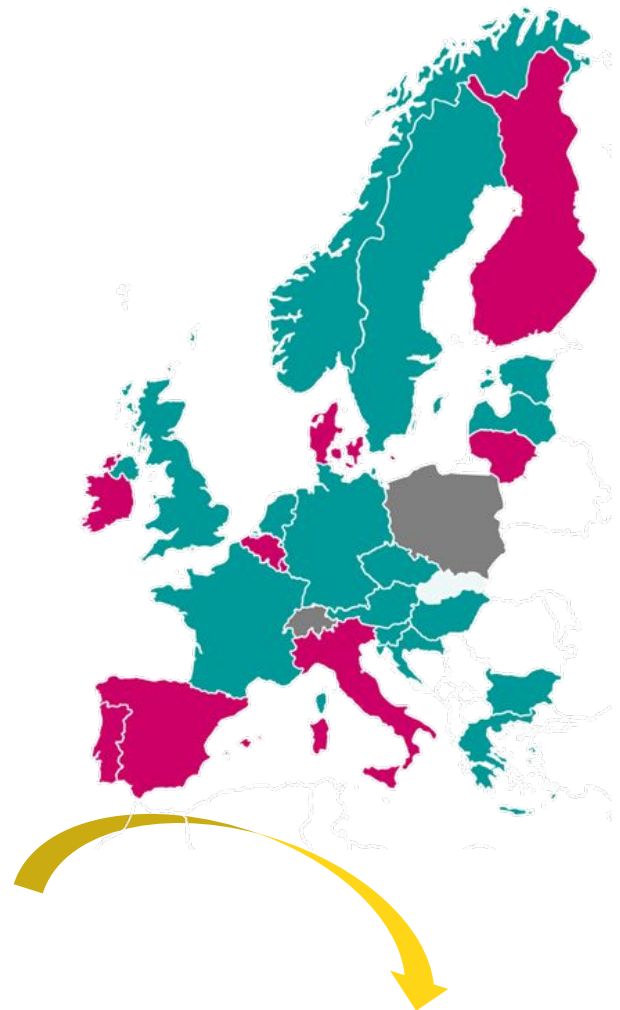
Mapping maturity in Europe

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 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.
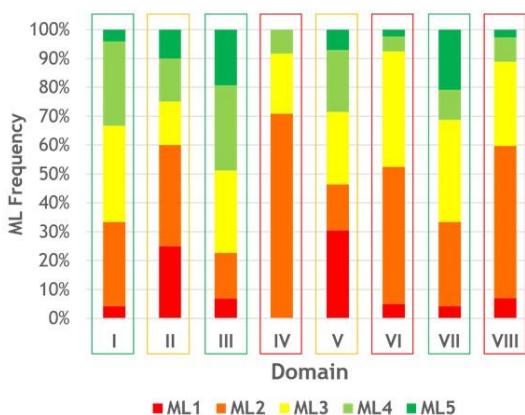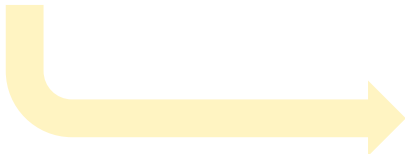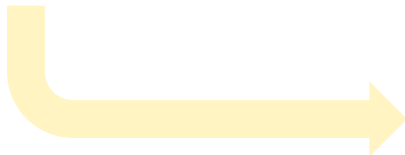
ASSESS – Benchmarking: an example
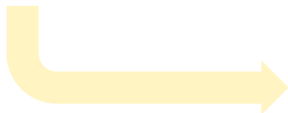

STEP 1

Benchmarking maturity

The results from the maturity level assessment of a healthcare system can be submitted online for benchmarking, using the Benchmarking Tool, as exemplified here for the indicators of Domain I.

Submission of maturity assessment

Domain I - Governance and strategy

I1 - Country/region has a dedicated governance for genomics in healthcare

I2 - Genomics in healthcare is established as a priority at national/regional level

I3 - There is a national/regional strategy for genomics in healthcare with a costed implementation plan

Submit — For each Domain I indicator, select and submit the attributed score, from a scale of 1 to 5.

For each indicator, the Benchmarking Tool will provide users with the percentage distance between their maturity level score (**pink**) and the highest maturity score (**blue**) among all submitted health systems, for that indicator. This information is accessible for each domain, exclusively by the submitters, using a private link.

Distance to top score

Distance to top score (%) is calculated (for each indicator) as Top score – Score submitted)/(Top score – Minimum score) x 100, minimum score being 1.

The overall relative frequency of maturity levels from all contributing health systems, for each indicator in every Domain, will be available publicly as dashboard. This will provide a graphical interface with at-a-glance view of maturity levels across Europe. Health system contributions will be anonymous.

Relative frequency of maturity levels

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 [Beyond One Million Genomes](#) (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 National Health System (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

- 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

- 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.
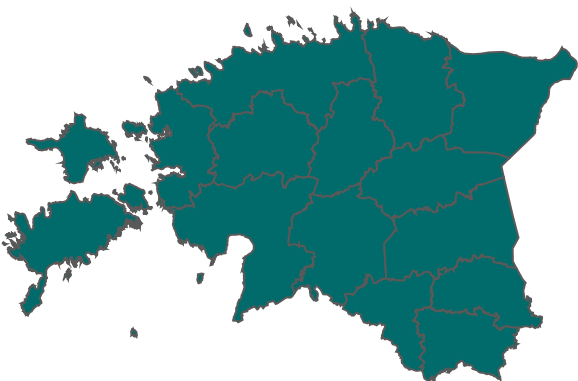
Watch the Estonia Country Visit

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

Domain I. Governance & Strategy

Guidance from 1+MG and other initiatives related to Personalised Medicine

- 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
- ICPerMed Publications
- ICPerMed Vision Paper
- ICPerMed Best Practices Implementation Examples
- SRIA on PM (EP PerMed)
- Danish strategy for personalised medicine
- Genomic Medicine France 2025
- Genome strategy Finland
- Genome UK implementation plan
- Genomics England
- Australia Genomics

Domain II. Investment and economic models

Guidance from 1+MG and other initiatives related to Personalised Medicine

- 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
- HEcoPerMed
- Short scenarios for future PM (HEcoPerMed)
- Health economic perspective on PM (HEcoPerMed)
- Case studies on health economic modelling (HEcoPerMed)

Domain III. Ethics, legislation and policy

Guidance from 1+MG and other initiatives related to Personalised Medicine

- 1+MG Framework on ELSI issues
- B1MG Resources
- 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
- GA4GH guidance products
- GDPR EU
- EHDS
- TEHDAS
- UK Biobank

Domain IV. Public awareness and acceptance

Guidance from 1+MG and other initiatives related to Personalised Medicine

- 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 Deliverable 1.5: Stakeholders trust in genomic data sharing landscape analysis
- Policy brief: key issues for implementation of genomics
- B1MG poster: Recommendations for trustworthiness from an experts workshop organised by B1MG
- Engaging patients and the public GA4GH
- Involving patients with CV diseases (ICPerMed)
- NAGEN 1000 (ICPerMed)

Domain V. Workforce skills and organisation

Guidance from 1+MG and other initiatives related to Personalised Medicine

- CEV to the UK: Training and tools in genomics for the entire healthcare workforce
- Education in pharmacogenomics (ICPerMed)
- NAGEN 1000 (ICPerMed)
- Genomic testing: a competency framework
- Genomics Education Program
- Instituto Roche: Competencies for healthcare professionals in personalised medicine (spanish)

Domain VI. Clinical organization infrastructure and tools

Guidance from 1+MG and other initiatives related to Personalised Medicine

- 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
- PERMIT Project (ICPerMed)
- PERMIT recommendations (ICPerMed)
- Phamacogenetics-based clinical decision (ICPerMed)
- Adoption of EMR (ICPerMed)
- PERSEPHONE project and multidisciplinary teams (ICPerMed)
- Institutional synergies for lung cancer (ICPerMed)

Domain VII. Clinical genomics guidelines and infrastructure

Guidance from 1+MG and other initiatives related to Personalised Medicine

- B1MG resources
- 1+MG Framework sequencing guidelines
- GA4GH products
- Genomic centers, example from Germany (ICPerMed)
- CPIC guidelines
- Dutch pharmacogenetics working group Guidelines
- PharmGKB resources
- ACMG guidelines
- ClinVar
- ClinGen

Domain VIII. Data management, standards and infrastructure

## Guidance from 1+MG and other initiatives related to Personalised Medicine

- 1+MG recommendation: Recommendations for a Data Access Governance Framework
- B1MG Proof of Concept: Federated data access rare diseases proof of concept
- 1+MG Framework technical infrastructure
- 1+MG Framework data standards
- Data connect API (GA4GH)
- Interoperable data, the Swiss example (ICPerMed)
- Resource Entitlement Management System REMS
- Life Science AAI
- Federated EGA (FEGA)
- Htsget protocol

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.
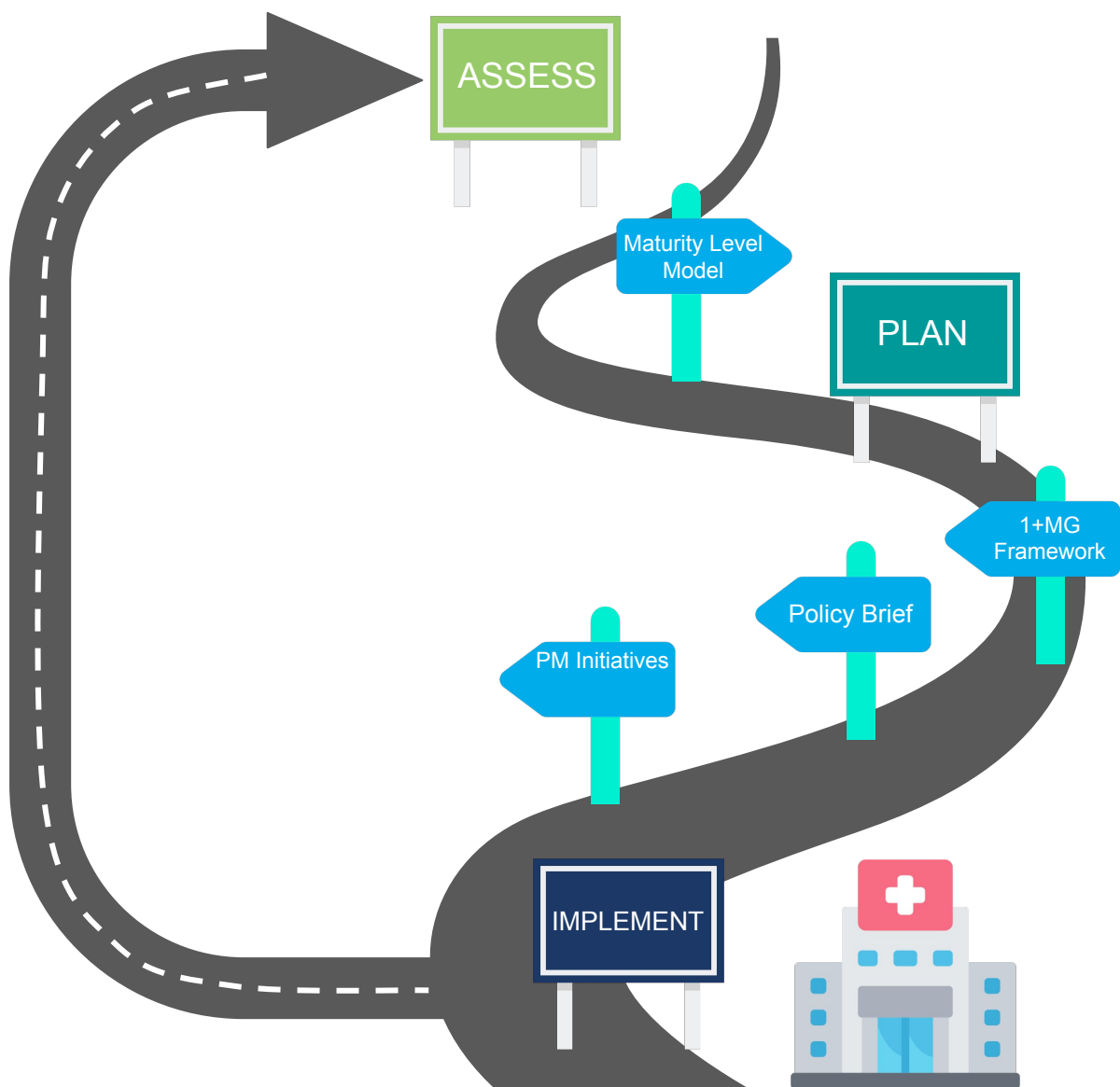
Authors

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