1+MG NMG meeting
Launch of the survey : Collecting data on genomic medicine implementation in European healthcare systems

December 11, 2025

Frédérique Nowak, on behalf of the B1MGplus project and the 1+MG WG7

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Journey to an infrastructure for genomic data

B1MG plus

Cross-border access to genomic data, implementation of genomics-based health

2018 2020 2022 2024 2026 2028

Design & Testing

Scale-up & Sustainability

B1MG Beyond one Million Genomes

European Genomic Data Infrastructure

Population Genomics

GOE GENOME OF EUROPE

EDIC, healthcare

B1MG plus

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

B1MG has received funding from the European Union's Horizon 2020 Research and Innovation programme under grant agreement No 951724

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

GOE receives funding from the European Union's Digital Europe Programme

B1MGplus Summary information

3-year CSA project

EUR 2M across 4 WPs

Start date 1st Feb 2025

36 project partners

20 Beneficiaries

16 Associated Partners

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

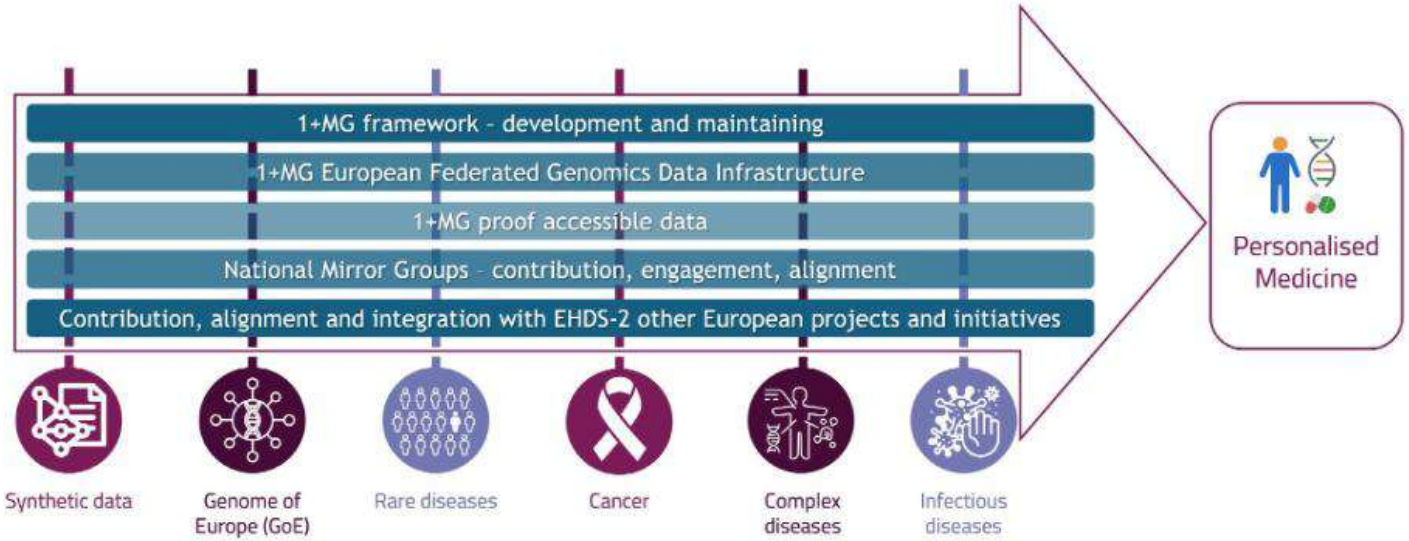
Beyond 1 Million Genomes Plus

Call: DIGITAL-2024-CLOUD-DATA-AI-06
(Cloud, data and artificial intelligence)
Topic: DIGITAL-2024-CLOUD-DATA-AI-06-GENOME
Type of Action: DIGITAL-CSA
(DIGITAL Coordination and Support Actions)
Proposal number: 101194865
Proposal acronym: B1MGplus

B1MG plus

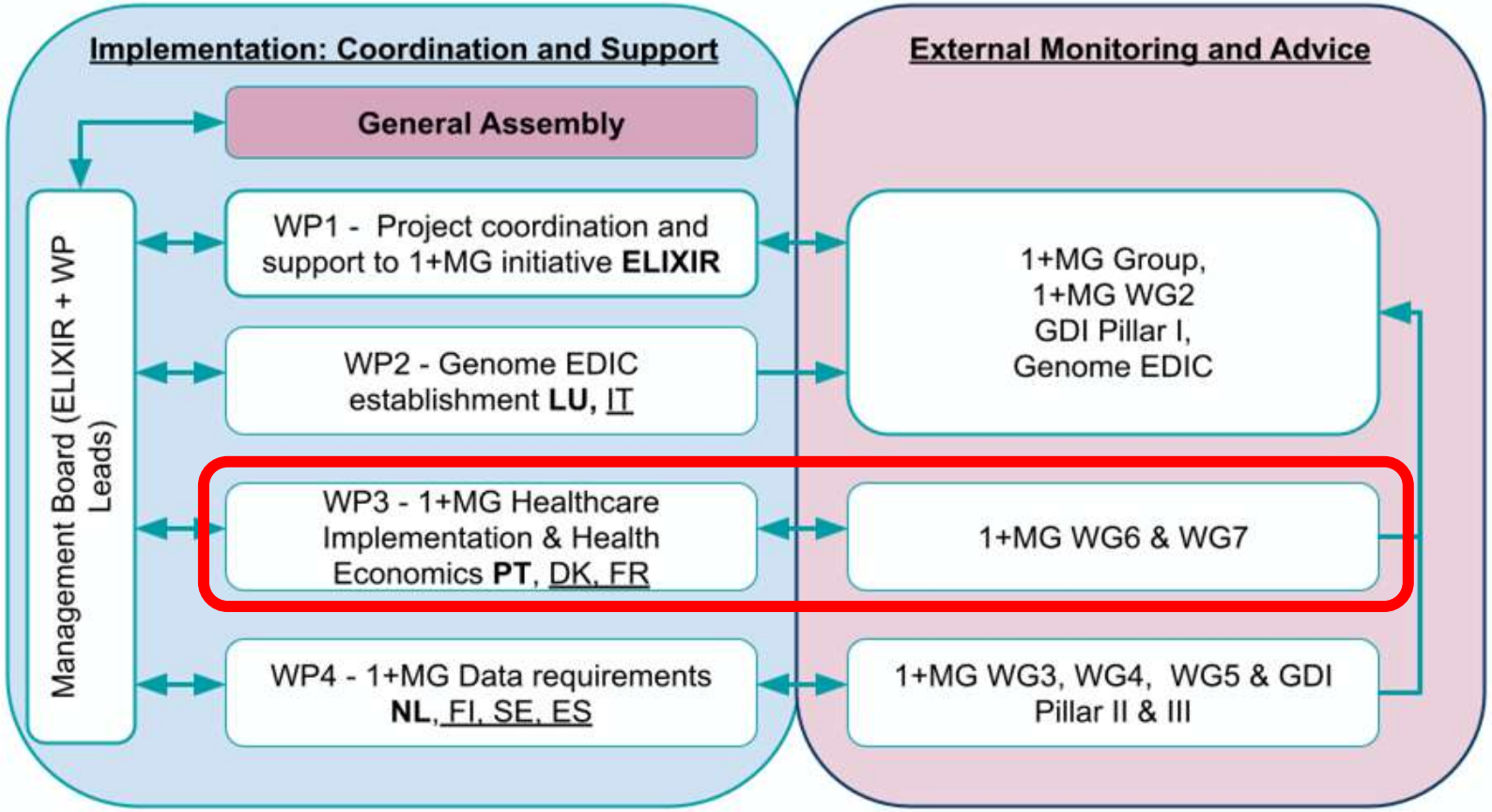
Objectives :

- Create a federated genomics infrastructure based on a EDIC
- Develop guidelines for metadata, data and software standards and quality assurance
- Provide policy and legal frameworks to ensure secure, ethical data sharing
- Facilitate the uptake of genomics in healthcare delivery and public health policy



B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

B1MGplus project and governance structure



1+MG WG

- **WG1 - Governance, NMGs**
- **WG2 - ELSI**
- **WG3 - Clinical data**
- **WG4 - Sequencing quality**
- **WG5 - Infrastructure**
- **WG 6 - Health Economics**
- **WG 7 - Implementation in Healthcare**
- **WG8 - Rare Diseases**
- **WG9 - Cancer**
- **WG10 - Common complex diseases**
- **WG11 - Infectious diseases**
- **WG12 - Pharmacogenetics**

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

WP3 - 1+MG Healthcare Implementation & Health Economics

Objectives in articulation with 1+MG WG6 & WG7 :

- O3.1 To bridge **implementation gaps on genomics in healthcare systems across Europe**, through mapping, self-assessment and benchmarking using the 1+MG Maturity Level Model (MLM) structure.
- O3.2 To **document the implementation of genomic medicine in national healthcare systems**, facilitating sharing of experiences across countries and informing policy makers.
- O3.3 To **promote the awareness, literacy level and education of citizens, health professionals and policy makers** on genomic medicine across Europe .
- O3.4 To contribute to implementation of WGS and other genetic tests in healthcare, by informing policy makers and decision-makers on **how to measure and model its economic impact**, supporting the right decisions and legitimising budget allocation.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

WP3 - 1+MG Healthcare Implementation & Health Economics

O3.2 To **document the implementation of genomic medicine in national healthcare systems**, facilitating sharing of experiences across countries and informing policy makers

- Genomic medicine maturity varies across European countries
- Solutions for specific challenges which have been previously developed by a country can serve as best practices for the other countries
- Mapping and disseminating these best practices will facilitate faster development and implementation of genomics in healthcare in a consistent manner across Europe

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

WP3 - 1+MG Healthcare Implementation & Health Economics

Task 3.3. Mapping and sharing successful practices for genomic medicine in healthcare systems

⇒ Conduct a mapping exercise to:

- **document the organisational frameworks** put in place for the implementation of genomic medicine in healthcare systems
- **identify best practices**

⇒ Launch a survey that addresses the provision of genetic/ genomic tests **in clinical setting**, both for diagnosis, treatment and prevention.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Method

- 3 online meetings with 1+MG WG7 members (experts from 17 countries)
- A face to face session organised at the B1MGplus Bucharest workshop in March 2025
- Draft of the questionnaire sent to WG7 for review
- Creation of an online questionnaire

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Who are the respondents?

- The survey is sent to 1+MG National Mirror Groups (NMGs).
- **A collective answer is expected by country**, including countries with a regional healthcare system.
- NMG will be the survey respondents.
- For the survey follow-up, we will ask to provide us one contact person per respondent NMG. Contact details will not be made public, but may be made available to other experts within the 1+MG initiative and B1MGplus project if asked by experts.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

What will be the outcomes of the survey?

- A webinar will present the aggregated results (October 2026).
- A more in-depth analysis of some answers will be performed by experts from other 1+MG WG or B1MGplus
- A synthesis of the results, with good practice identifications and gaps analysis, will contribute to recommendations for Genome EDIC on implementation and sustainability of genomic medicine in healthcare systems (B1MGplus Deliverable 3.5)

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Timeline

- The responses to the questionnaire will provide **a snapshot** of the organisational frameworks implemented for genomic medicine **at the end of 2025**
- Launch date: **December 18, 2025**
- Countries will have **a 3-month response time (March 18, 2026)**

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Questionnaire structure

Questions aim to describe the organisational framework implemented for genomic medicine in European countries.

- They **cover key topics** regarding implementation of genomic medicine in healthcare systems
- They must take into account :
  - ✓ The **clinical applications** for genomic medicine
  - ✓ The **level of territorial organisation** of the country's healthcare system
  - ✓ The **technical approaches** used for genetic/genomic testing

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Questionnaire outline

- The questionnaire outline follows the structure of the Maturity Level Model for Genomics in Healthcare developed in the context of 1+MG

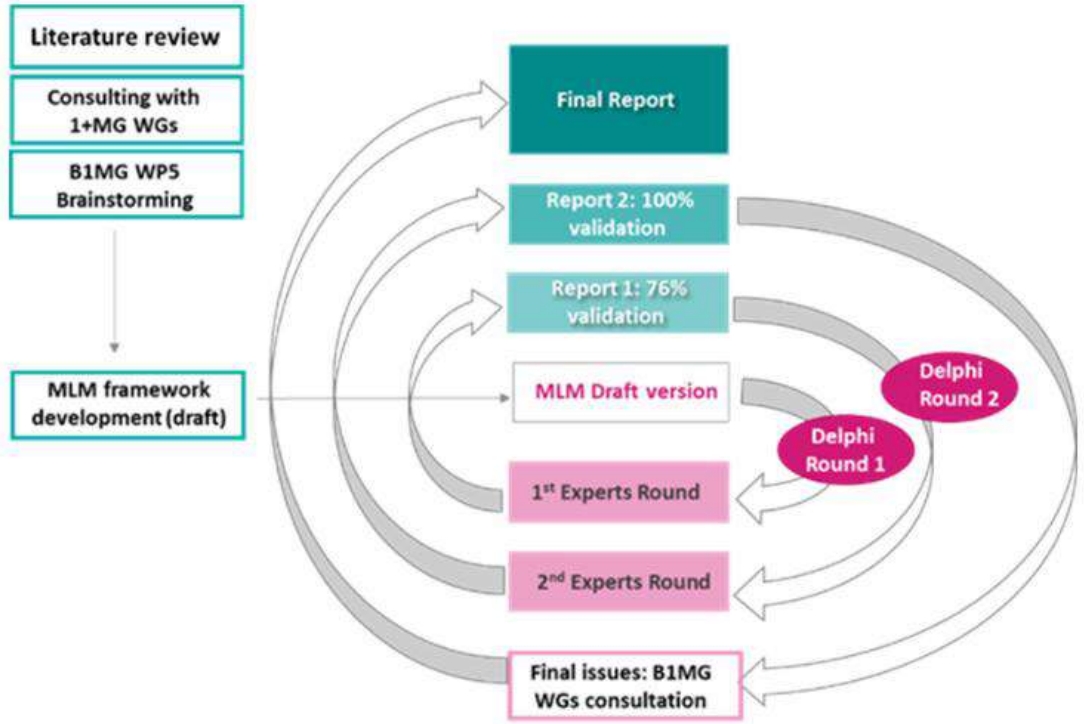
- Questions encompass the MLM domains and sub-domains to ensure the accurate covering of key topics

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

MLM domains

MLM domain cover key topics for the implementation of genomics in healthcare which have been validated by experts.

- **8 MLM domains**
  I. Governance and Strategy
  II. Investment and economic models
  III. Ethics, legislation and policy
  IV. Public awareness and acceptance
  V. Workforce skills and organisation
  VI. Clinical organisation, infrastructure and tools
  VII. Clinical genomics guidelines and infrastructure
  VIII. Data management, standards and infrastructure
- **33 MLM subdomains**
- **49 MLM indicators**



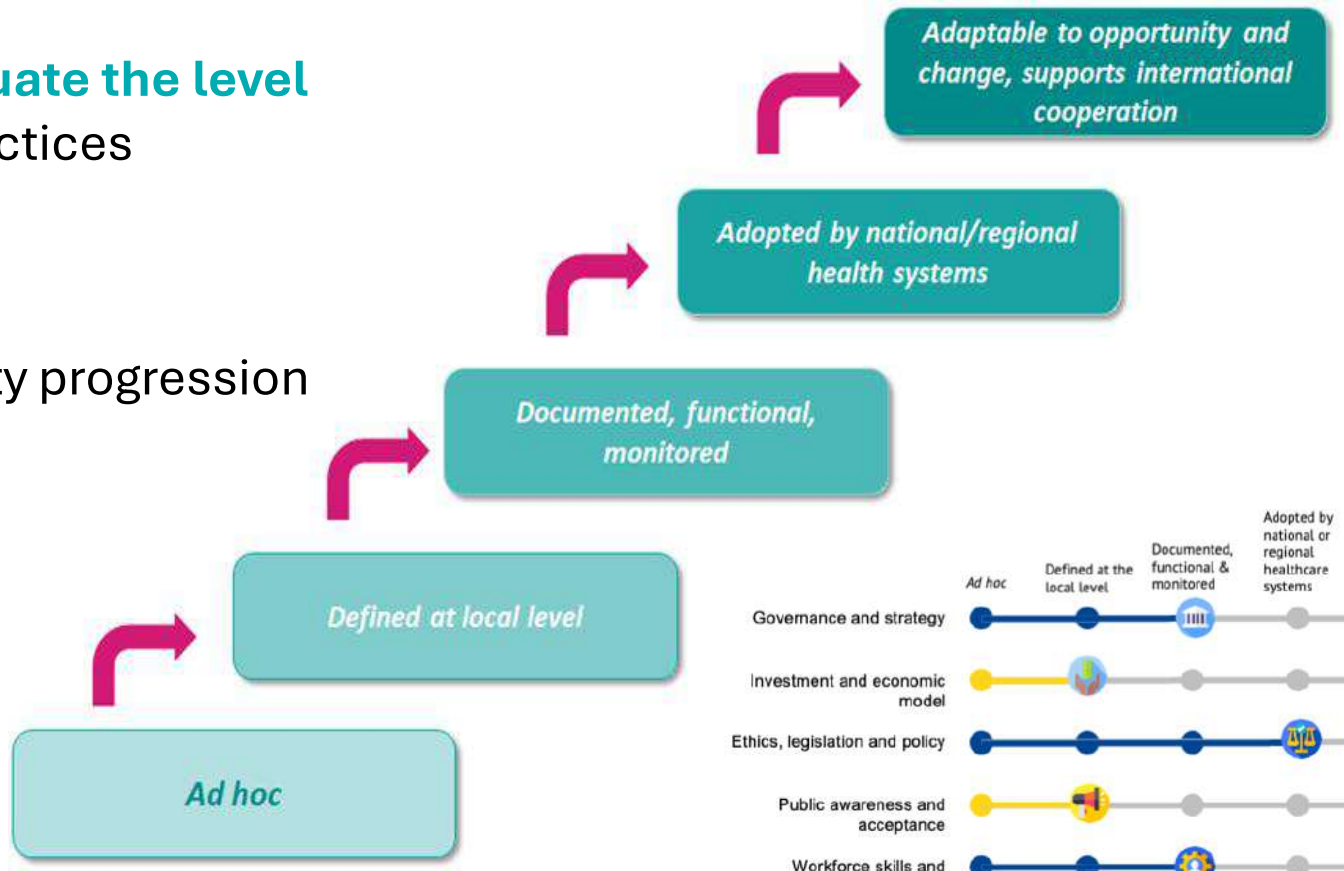
https://framework.onemilliongenomes.eu/genomics-into-healthcare#maturity-level-model

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Maturity Level Model for Genomics in Healthcare

A tool for healthcare systems **to self-evaluate the level of maturity** of their genomic medicine practices according to a **common matrix**.

5 maturity levels are indicative of a maturity progression



Aim

Promote and facilitate the adoption of genomics in healthcare systems and define a path to optimization

3plus project receives funding from the European Union's Digital Europe Programme under grant agreement r

Clinical applications of genomic medicine

8 clinical applications of genomic medicine (human genetics/ genomics) have been identified, covering prevention, screening, diagnosis and treatment:

- ✓ Rare diseases
- ✓ Prenatal diagnosis
- ✓ Cancers (tumor genetics/ genomics)
- ✓ Cancer genetic predisposition
- ✓ Prenatal genetic screening (Noninvasive prenatal testing - NIPT)
- ✓ Newborns genomic screening
- ✓ Pharmacogenetics
- ✓ Complex diseases

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Regional/ national organisations of the healthcare system

Questions address the **level of territorial organisation of the healthcare system** with possible answers:

- Yes, **at national level**
- Yes, **at regional level**
- Yes, **at local level**
- Planned or in development **at national level**
- Planned or in development **at regional level**
- Planned or in development **at local level**

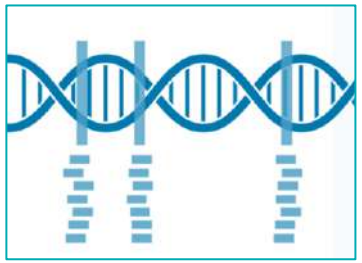
B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Technical approaches

Provision of genetic and genomic tests **in clinical setting** with different technical approaches :

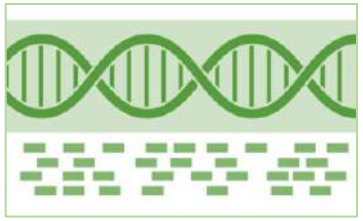
- **Gene panels sequencing**

  - small & **large** panels, other targeted approaches)

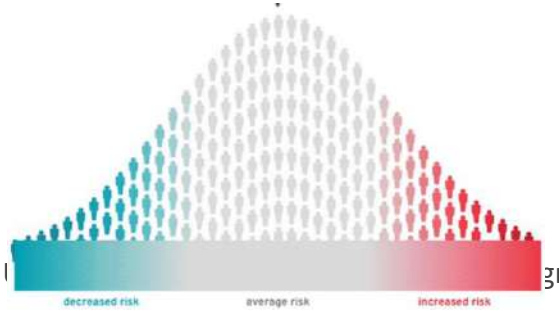


- **Comprehensive sequencing approaches**

  - Whole Exome Sequencing (WES)
  - Whole Genome Sequencing (WGS)
  - Long Read Whole Genome Sequencing (LR-WGS)
  - Whole Transcriptome Sequencing (WTS), Whole DNA methylome analysis…



- **Polygenic risk scores**



B1MGplus project receives funding from the European Union's Horizon Europe research and innovation programme under grant agreement number 101194865.

Question structure

- Collect the information in a structured way (**closed-ended questions**) to enable effective analysis of results.
- For some questions, the questionnaire opens sub-questions if the answer is yes/ planned/ in development/ in preparation, to explore the topic in more detail.
- Collect detailed information to highlight best practices (**links to documents and websites of interest**).

Questions

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

44 questions

B1MG

A.	**Governance and Strategy**	1 question
B.	**Investment and economic models**	
1.	Access and reimbursement	7 questions
2.	Health economics	4 questions
C.	**Ethics, legislation and policy**	
1.	Consent to genetic/genomic testing	5 questions
2.	Quality of patient care involving genetic/genomic testing	3 questions
3.	Health data sharing and reuse	3 questions
D.	**Public awareness and acceptance**	
1.	Acceptance/ Engagement and trust	2 questions
2.	Communication to the general public	1 question
E.	**Workforce skills and organization**	5 questions
F.	**Clinical organisation, infrastructure and tools**	
1.	ICT tools for clinical decision	1 question
2.	Multidisciplinary teams	1 question
3.	Uptake of novel tools and technologies for genomics	1 question
4.	Synergies with research	2 questions
5.	Partnership with industry	2 questions
G.	**Clinical genomics guidelines and infrastructure**	
1.	Sequencing/ genotyping infrastructure	1 question
2.	Sequencing guidelines	2 questions
3.	Clinical interpretation	1 question
H.	**Data management, standards and infrastructure**	2 questions

A – Governance and strategy

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

A – Governance and strategy

Strategy and governance

- **Q1** - There is one or more strategies for implementing genomic medicine in the healthcare system.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

B - Investment and economic model

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

B 1- Access and reimbursement

B1MG plus

Investment plans	• **Q2** - There is one or more investment plans to set up clinical genetic/ genomic testing laboratories. • **Q3** - Operational costs for genetic/ genomic testing are funded by one or more investment plans.
Long term reimbursement	• **Q4** - Genetic/ genomic testing is fully or partially reimbursed by the healthcare system. • **Q5** - There is a potential funding gap between the end of investment funding and the start of reimbursement of genetic/ genomic testing.
Cost for patients	• **Q6** - Gene panels sequencing-based testing is at no cost for patients. • **Q7** - Comprehensive sequencing-based testing is at no cost for patients. • **Q8** - PRS-based testing is at no cost for patients.

B 2 - Health economics

HTA framework

- **Q9** - There is a Health Technology Assessment (HTA) body/ agency in the country.

Cost effectiveness framework

- **Q10** - Cost-effectiveness is part of a reimbursement/ implementation decision.
- **Q11** - Pharmaceutical reimbursement and diagnostic reimbursement are linked.
- **Q12** - There is a project or clinical evaluation framework assessing the cost-effectiveness of genetic/ genomic testing (either for gene panels testing, comprehensive genomic testing or PRS testing).

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

C – Ethics, legislation and politics

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

C 1 –Consent to genetic/ genomic testing

B1MG plus

Consent

- **Q13** - There are procedures or regulations for informed consent for genetic/ genomic testing (counselling and consent).
- **Q14** - There are information supports and templates of consent forms for genetic/ genomic testing.

Prescribers

- **Q15** - Medical specialties of doctors who can request genetic/ genomic tests.
- **Q16** - Medical doctors who request genetic/ genomic tests and who are not geneticists have to be trained in genomic medicine through continuing education programmes.

Incidental findings

- **Q17** - When individuals consent to genetic/ genomic testing, they also have the option to consent to the return of incidental or secondary findings.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

C 2 – Quality of care involving genetic/genomic testing

B1MG plus

Quality norms

- **Q18** - There are professional codes of conduct or regulatory bodies for quality of genetic/ genomic testing.
- **Q19** - Accreditation of clinical laboratories for genetic/ genomic testing is mandatory or recommended by national/ regional guidelines.

Patient evaluation

- **Q20** - There are procedures for patient evaluation of the quality of care for genetic/ genomic testing (patients perform evaluation).

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

C 3 –Health data sharing and reuse

Samples	• **Q21** - There is one or more organisations to enable the storage and the reuse of samples used for genetic/ genomic testing for research purposes.
Data	• **Q22** - When individuals consent to genetic/ genomic testing, they also have the option to consent to the reuse of their data for research purposes. • **Q23** - An information portal has been set up to inform patients and give them the opportunity to exercise their rights (according to GDPR).

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

D – Public awareness and acceptance

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

D – Public awareness and acceptance

Engagement and trust

- **Q24** - There are guidelines on the systematic inclusion of, or engagement with, patient associations in human genomics.
- **Q25 -** Citizen dialogues or consultations on genetic/ genomic testing are being developed or conducted *(e.g. The Belgian DNA Debate, the French Estates General on Bioethics…)*.

Communication to the general public

- **Q26** - General educational/information materials on genomic medicine (*e.g. leaflets, public media campaigns, etc*.) are developped towards the general population.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

E – Workforces skills and organisation

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

E – Workforces skills and organisation

B1MG plus

Genetic counsellors

- **Q27** - There are training programs for genetic counsellors.
- **Q28** - There are career paths for genetic counsellors.

Career paths in genomic medicine

- **Q29** - There are career paths for “clinical bioinformaticians” in genomic medicine.
- **Q30** - There are career paths for clinical laboratory scientists.
- **Q31** - There are career paths for other health professionals in genomic medicine.

B1MGplus project receives funding from the European Union’s Digital Europe Programme under grant agreement number 101194865.

F – Clinical organisation, infrastructure and tools

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

F – Clinical organisation, infrastructure and tools

B1MG plus

ICT tools	• **Q32** - A variant database has been developed to support biological and clinical interpretation of human genomic data.
Multidisciplinary teams	• **Q33** - There are recommendations for the organization of multidisciplinary meetings to validate the request of genetic/ genomic tests and/ or to discuss the results of genetic/genomic testing.
Uptake of novel tools and technologies	• **Q34** - There is an infrastructure in charge of evaluating and transferring new techniques in clinical practice to the clinical genetic/genomic testing laboratories.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

F – Clinical organisation, infrastructure and tools

B1MG plus

Synergies with research

- **Q35** - Pilot projects are conducted to evaluate the feasability of genetic/ genomic testing before implementation in healthcare setting.
- **Q36** - There is an organisation for functional validation of Variants of Unknown Significance (VUS).

Partnership with industry

- **Q37** - There are established collaborations with industry partners (*e.g. biotechnology, pharmaceutical, diagnostics companies*) to co-develop, validate or implement genomic tools in healthcare.
- **Q 38** - There are formal mechanisms (*e.g. public-private partnerships, joint funding schemes, innovation hubs*) to promote industry involvement in genomic medicine initiatives.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

G – Clinical genomic guidelines and infrastructure

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

G – Clinical genomic guidelines and infrastructure

B1MG plus

Infrastructure	• **Q39** - There is one of more a networks of clinical genetic/ genomic testing laboratories for the benefit of all patients (equal access).
Sequencing guidelines	• **Q40** - There is an officially approved or validated directory of clinical indications for genetic/ genomic testing. • **Q41** - There is a national/ regional group or committee responsible for the development and/or validation of genetic/genomic testing guidelines.
Clinical interpretation	• **Q42** - Which healthcare professionals are authorised to perform clinical interpretation for genetic/ genomic testing ?

g from the European Union's Digital Europe Programme under grant agreement number 101194865.

H – Data management, standards and infrastructure

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

H – Data management, standards and infrastructure

Data reuse	• **Q 43** - A strategy has been defined to enable the secondary use of clinical genetic/ genomic data.
EHDS implementation	• **Q44** - Preparatory work has begun at national level to implement EHDS in the country.

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Documents to help with completion

- Survey completion guide

Download the documents here :

- Excel file with the list of questions and sub-questions to help with the collection of information from different sources

Q9 - There is a Health Technology Assessment (HTA) body/ agency in the country.	**Yes**	☐
	No	☐
	Don't know	☐
If yes:		
Name of the HTA body/ agency:		
Link to the webpage:		
The HTA body/ agency has defined a process to evaluate genetic/ genomic testing (either for gene panels testing, comprehensive genetic testing or PRS testing).	*Yes*	☐
	Planned	☐
	In preparation	☐
	No	☐
	Don't know	☐
If yes, planned or in preparation		
The HTA body/ agency follows the EMA HTA guidelines.	*Yes*	☐
	No	☐
	Don't know	☐
The HTA body has started the evaluation process of genetic/ genomic testing.	*Yes*	☐
	No	☐
	Don't know	☐
If yes		
For which clinical applications?	*Rare diseases*	☐
	Prenatal genetic diagnosis	☐
	Cancers	☐
	Cancer genetic predisposition	☐
	Prenatal genetic screening (NIPT)	☐
	Genomic newborn screening	☐
	Pharmacogenetics	☐
	Complex diseases	☐
Links to publications presenting the methodology and/or results of the evaluation process of genetic/ genomic testing:		

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agr

Link to questionnaire

- Questionnaire is accessible here :

Or click here to start

B1MGplus project receives funding from the European Union's Digital Europe Programme under grant agreement number 101194865.

Thank you

/company/b1mgplus-project

B1MGplus website

B1MGplus project receives funding from the European Union’s Digital Europe Programme under grant agreement number 101194865.