



Beyond One Million Genomes



1 Million Genomes

1+MG MLM User Guide

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Beyond One Million Genomes

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Background

The [1+ Million Genomes Initiative](#) (1+MG) Maturity Level Model (MLM) framework for implementation of genomics into healthcare was developed as a tool to enable healthcare systems across Europe to self-assess the level of maturity of their genomic medicine

practices according to a common matrix, and define a path to optimization. As such, it aims to promote and facilitate the adoption of genomics in healthcare systems, and make personalised medicine accessible to citizens and patients across Europe.

The 1+MG MLM framework

The 1+MG MLM framework was designed to create a reference for the key domains necessary to enable genomic best practices in healthcare systems. The reference is provided by specific indicators for each domain (or subdomains). The maturity levels describe, specifically for each indicator, the possible current status relative to that indicator, and overall highlight the development path to increasingly more mature practices. This framework therefore allows the identification of the gaps in structures, processes and capacity, and the planning of future actions towards optimization. The indicators can further be used to monitor progress to maturity.

The 1+MG MLM was developed following a review of the literature and input from experts from the 1+MG Initiative Working Groups. Validation of the structure and contents of the MLM framework was carried out through a Delphi survey. It is structured in 8 Domains, covering key topics for the implementation of genomics in healthcare:

1. Governance and Strategy
2. Investment and economic models
3. Ethics, legislation and policy

4. Public awareness and acceptance
5. Workforce skills and organisation
6. Clinical organisation, infrastructure and tools
7. Clinical genomics guidelines and infrastructure
8. Data management, standards and infrastructure

Each Domain comprises several Subdomains (totalling 33). One or more Indicators (totalling 49) were defined for each Subdomain. For each indicator, 5 levels of maturity were specifically developed, overall representing a stepwise path that can support the identification and planning of measures for optimization. In general terms, the maturity levels describe indicators as:

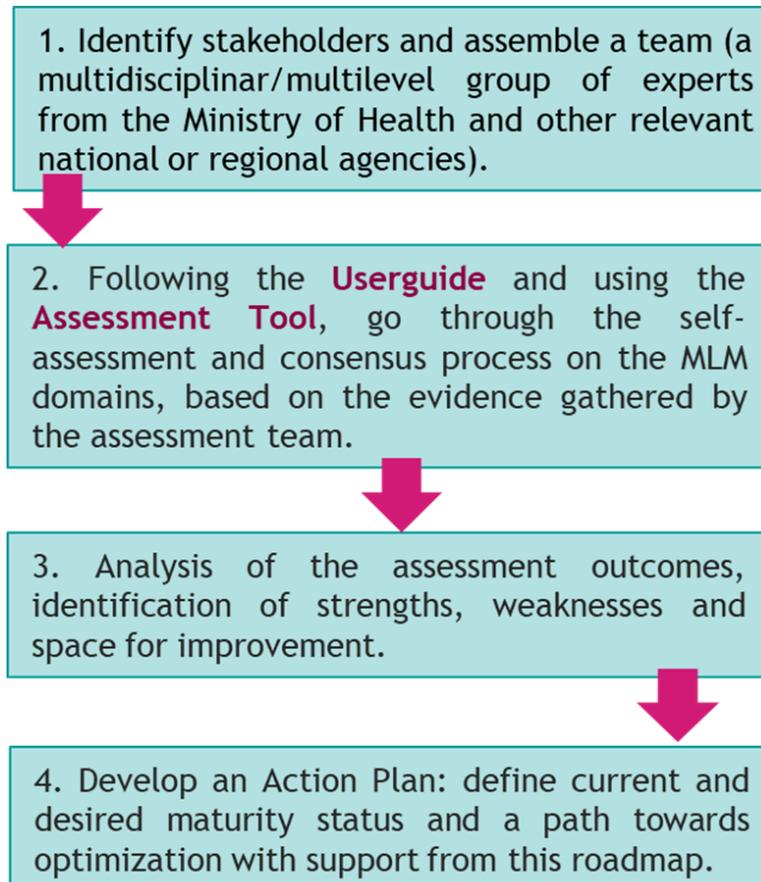
- Level 1: *Ad hoc*
- Level 2: defined at local level
- Level 3: documented, functional and monitored
- Level 4: adopted by national or regional healthcare systems
- Level 5: adaptable to opportunity and change and supporting international cooperation.



Using the 1+MG MLM

This User guide is intended to support healthcare systems in their self-assessment

exercise. The overall steps for this process are as follows:



1. Coordination team and identification of stakeholders

To facilitate the self-assessment process using the 1+MG MLM, a coordination team needs to be assembled, and a focal point assigned. The coordination team will review the country or regional landscape, and identify the right stakeholders from relevant national institutions and agencies that need to participate in the process.

The stakeholders representatives will be assembled in an Assessment team, including experts in each of the MLM domains. The Assessment team will be responsible for the self-assessment, and will be briefed on the goals and procedures of the assessment task before starting the process, along with an explanation on how the tool was developed and validated. The coordination team will oversee the assessment process, including briefing, guiding, and surveying the data collection and coordinating the consensus



process. To improve the response rate and engagement of stakeholders, frequent and open communication between the Coordination team and the stakeholders representatives is important.

The self-assessment process may take several months to complete, and all those involved need to plan their contribution in advance.

2. The Assessment Toolkit

The MLM Toolkit includes a Glossary, the 1+MG MLM, the Assessment tool and this User guide.

The [1+MG MLM](#) is freely available, and describes the domains, subdomains, indicators and maturity levels addressed for self-assessment.

The MLM Assessment tool is used for the assessment and the collection of supporting

evidence. It is an Excel file with separate tabs for the assessment of each domain, covering every subdomain, indicator and maturity level. Each domain assessment tab also contains two columns to be filled for each indicator, one column for the maturity level assessment, and another column to provide the rationale for the self-assessment and to refer to the evidence that supports the maturity level selection. The MLM Assessment tool is illustrated in Figure 1.

Respondent Name:					
Affiliation:					
Domain	Subdomains	Indicators	Maturity levels	Maturity Level assessment	Rationale and evidence
Governance and strategy	Governance	Country/region has a dedicated governance for genomics in healthcare	1. No dedicated governance for genomics in healthcare 2. Elements of governance exist but they are not fully functional 3. Scope of governance for genomics has been defined but elements are still under development 4. There is a governance body that is fully operating, led centrally, and activities are monitored based on a work plan 5. 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	1. Genomics in healthcare is not included in national/regional health plans 2. Inclusion of genomics in healthcare in relevant national/regional health plans is under discussion 3. Genomics in healthcare is included in relevant national/regional health plans 4. Genomics in healthcare is implemented as part of national/regional health and other relevant plans (e.g. education, research) 5. Genomics in healthcare is implemented in health and other relevant plans, and is periodically evaluated for optimization, taking into account novel developments at the international level.		
	Strategy	There is a national/regional strategy for genomics in healthcare with a costed implementation plan	1. No genomics in healthcare strategy with costed implementation plan 2. A strategy for genomics in healthcare with costed implementation plan is under discussion 3. A costed implementation plan for genomics in healthcare is developed and approved 4. The national/regional strategy for genomics in healthcare is under implementation 5. The national/regional strategy for genomics in healthcare is implemented, with monitoring and long-term resources and aligned with European and international strategies		

Figure 1. 1+MG MLM Assessment tool for the “Governance and strategy” domain.



Each stakeholder organisation will address one or more domains, according to its expertise and scope of activities. For this purpose the coordination team can multiply each tab as needed. The respondent will be identified with name and affiliation on the top of each tab. The first tab of the Assessment tool is used to register the stakeholder representatives, as members of the Assessment team, and which domain(s) they will be assigned. The final tab of the Assessment tool is used to complete the overall self-assessment exercise, after consensus is reached regarding maturity levels for each indicator, documenting the rationale and the evidence provided.

The Assessment tool is used by the Coordination team as the file compiling all the Assessment team members individual assessments. The Coordination team will send only the tabs with the assigned domains to each Assessment team member.

To increase stakeholder engagement, the MLM-based assessment might benefit from being translated into the national language, along with a translated version of the Glossary. Similarly, the Assessment tool may be implemented as an online survey, facilitating collection of the evaluations.

3. The assessment process: data collection using the assessment tool and reaching consensus

The coordination team will oversee the Assessment process and manage the MLM Assessment tool. It is advisable that a shared folder is set up online, where the Assessment tool can be shared and any documents referred as evidence can be saved. Alternatively, the Assessment tool can be distributed by email, and the files with replies from each Assessment team member,

together with evidence documents, are sent back to a focal point within the coordination team.

For each assigned domain, Assessment team members will associate the maturity level that best characterises the current situation for each indicator. The rationale for this selection needs to be provided, with evidence in the form of reference to documents, URLs, records, etc, in the column “Rationale and evidence”.

Attribution of a maturity level to an indicator may not be immediately straightforward. For instance, it is possible that available evidence only partially supports a specific ML. In this case, it should be considered that the highest ML is not fully reached yet, and thus the ML attributed to that indicator should be the lower. When there is a national assessment but there are regional healthcare systems, the highest MLs should be attributed to indicators pertaining to services that are accessible at the national level; however, if there are clear asymmetries across regions, then the lower regional ML should be attributed nationwide, so that planning for development of all regions is promoted. Other ambiguous situations may arise, and need to be solved with good sense. Naturally, the need for consensus will be helpful in these situations.

Once the Assessment team members have completed the assessment, results must be discussed among the Assessment team to reach a consensus on the maturity level per indicator. The Coordination team leads the consensus phase, facilitating the discussion, supported by the evidence provided, to reach an agreement for each indicator. The outcomes of the consensus discussion are registered in the last sheet of the Assessment tool, and rationale and evidence provided are documented. Sharing the completed



Assessment tool, containing all the individual assessments, rational and evidence, with all Assessment team members prior to the meeting will facilitate discussion and the process of reaching consensus.

Consensus means that a decision on a ML for a specific indicator must be agreed by the Assessment team as a whole. It is different from the “choice of the majority”. The process of reaching consensus may take several rounds of decision until the group agrees on a specific ML for the indicator under discussion. There are different levels of agreement, namely, (i) unqualified “yes”, (ii) “accept the decision”, (iii) “can live with the decision”, and (iv) “do not fully agree with the decision, however, will not block it and will support it”. Any blocks require a new round of discussion until there are no further blocks to the decision of the group.

4. Data analysis

Once consensus is reached for every domain, the coordination team can identify the areas of strengths and weaknesses in each domain,

The consensus has to ensure that all participants are equally involved. The facilitator(s) must be able to help the group make decisions and work together creatively and efficiently. Some skills required to perform the task of facilitator include being neutral, objective, patient, respectful, assertive, and a good listener. The facilitator(s) should ensure that, at the beginning of the meeting, all participants agree and feel comfortable with the set rules and guidelines of the meeting (e.g., levels and rules of consensus, number of rounds allowed, how to deal with impossible consensus situations), and that during the meeting everyone feels comfortable and is equally included, heard, respected, conflicts are resolved peacefully, and the discussion is kept on time.

as shown in Figure 2. This information can be used to set goals, define areas of investment and develop an action plan.



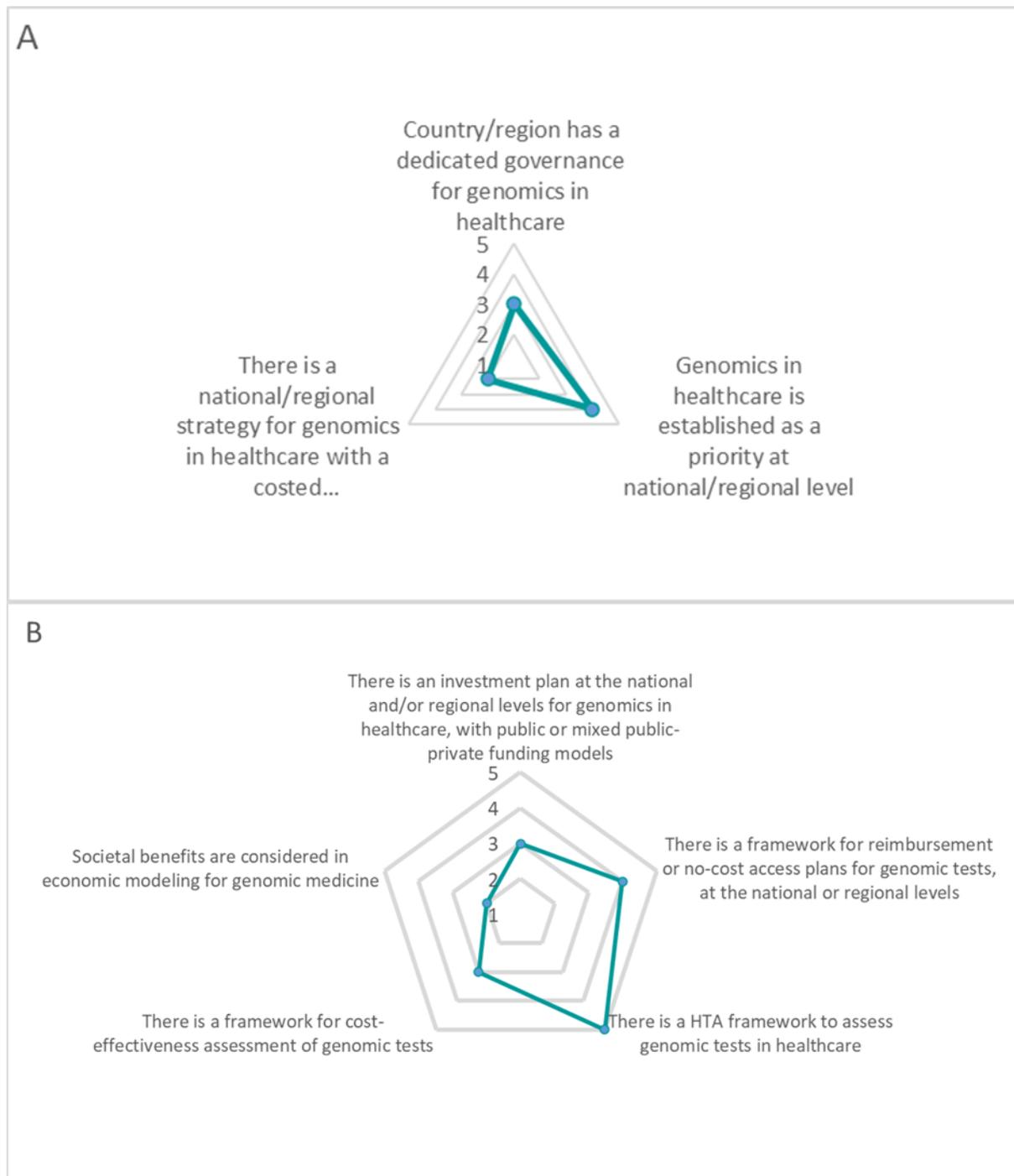


Figure 2: Examples of spider web diagrams illustrating the self-assessment outcomes, with hypothetical results for Domain I (A) and Domain II (B). Numbers 1 to 5 represent the maturity levels, for each of the indicators for Domain I (A) and Domain II (B).



Developing an Action Plan

Using the self-assessment results and the B1MG MLM framework, the coordination team can identify and plan the next steps towards optimising the use of genomics in healthcare. Planning further actions will feed on the analysis of the maturity assessment, but needs to consider additional issues, for instance the healthcare systems context, objectives and resources. At the stage of developing an action plan, it is important to consider engaging other stakeholders whose expertise can effectively help the implementation of the action plan.

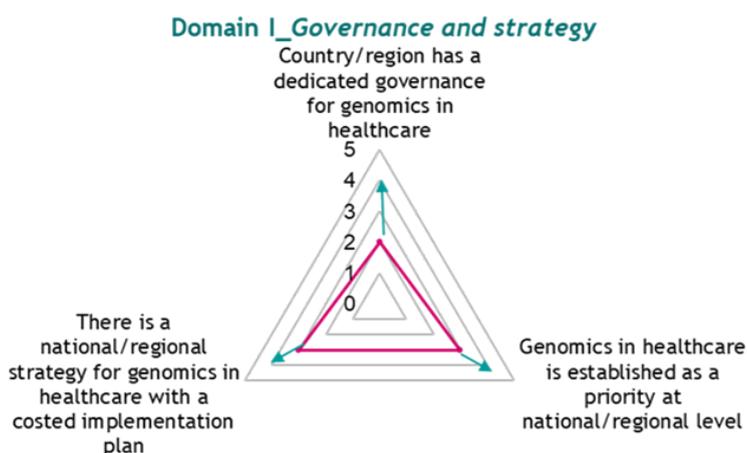
Some discussion topics can be addressed to construct the action plan:

- Where are the strengths and where are the challenges?
- What are the maturity goals for each domain?

- Are there contextual priorities?
- Which domains need more investment?
- In which domains is progress easier?
- Which issues are bottlenecks for progression?

The 1+MG MLM is an instrument for self-assessment of current status, but also offers a framework for progression. The indicators and maturity levels provide reference points to define the desired or possible maturity status, and also to determine the processes, structures and capacities that need to be built to reach higher maturity (see below, Figure 3, an example for Domain I). Once an action plan is implemented, the indicators can be used to monitor progress along the path for maturity.





Sub-domain: indicator	ML1	ML2	ML3	ML4	ML5
GOVERNANCE: Country/region has a dedicated governance for genomics in healthcare	No dedicated governance for genomics in healthcare	Elements of governance exist but they 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 optimization, taking into account novel developments at the international level.
STRATEGY: There is a national/regional strategy for genomics in healthcare with a costed 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

Figure 3: Example for domain “Governance and strategy”, illustrating a possible current scenario (pink line on the radar graph, and pink ML text on the table) and putative goals set (green arrows on the radar graph, and green ML text on the table) for each indicator. Numbers 1 to 5 represent the maturity levels, for each of the indicators.

The 1+MG MLM is intended to be a dynamic instrument. Over time, indicators and maturity levels may change, and other domains may be added. Following the

evolution of knowledge and practices will keep this MLM relevant, and a resource for healthcare systems maturity assessment for years to come.

