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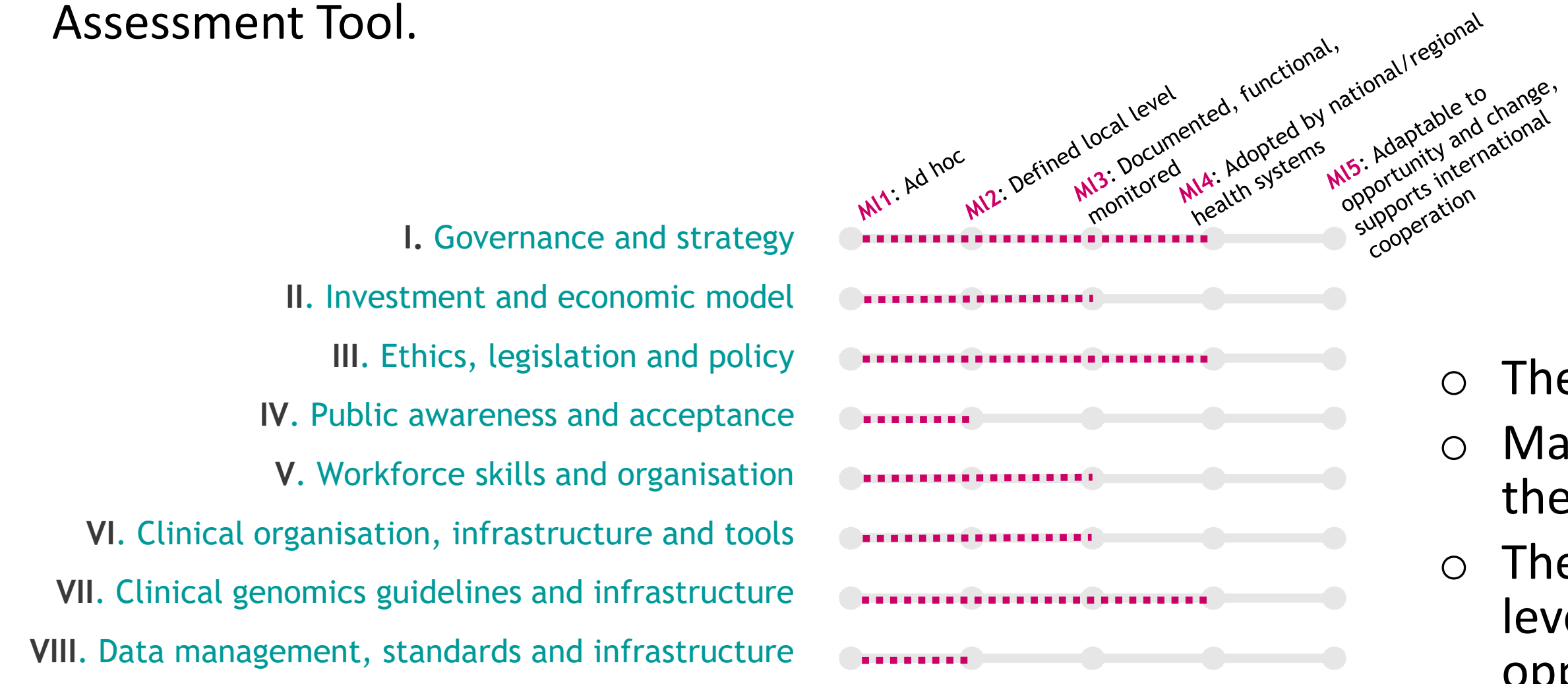
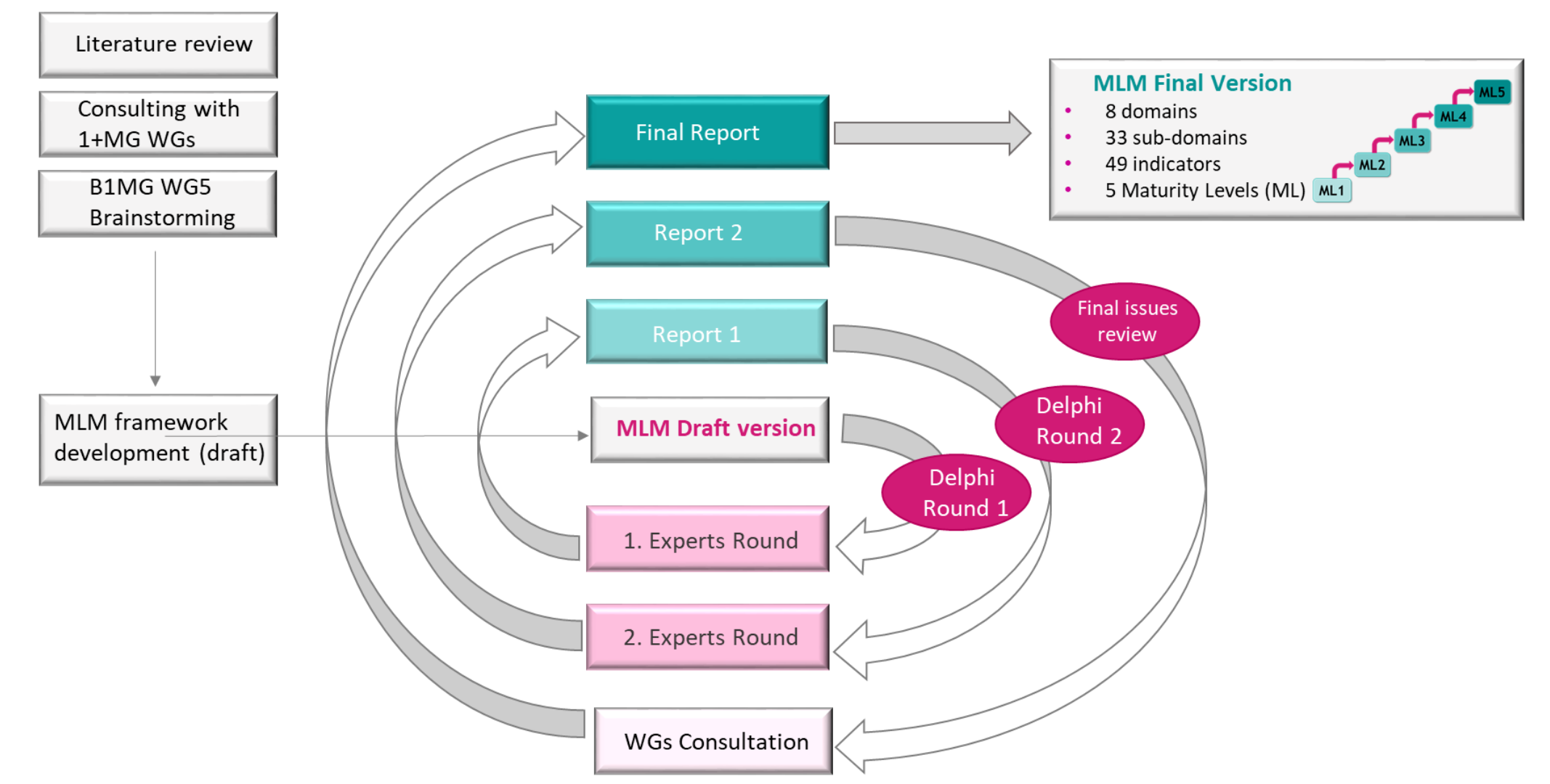
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Background

- Genomic medicine implementation in healthcare systems can bring us one step closer to making personalised medicine a reality, with major socioeconomic benefits. Citizens and patients can nowadays widely benefit from genomic data analysis for accurate and timely diagnosis, effective treatments with less adverse events, and accurate profiling for disease prevention;
- Implementation of genomics in healthcare is complex, and requires adjustments in the governance, structure and organization of health services, as well as dedicated investments. Implementation is also dependent on the country context;
- In the context of the 1+Million Genomes (1+MG) initiative, we developed a Maturity Level Model (MLM) for health systems to self-evaluate the maturity of their genomic medicine practices, and define a path to optimization; a MLM is 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.
- A MLM pilot in eight European countries provided important information regarding common strengths, weaknesses and asymmetries across Europe.

MLM development & DELPHI validation

- The initial MLM framework was developed based on literature review and input from the 1+MG experts;
- For MLM validation, a DELPHI survey sought consensus from an expert panel of leaders of major national and international genomic initiatives.
- To support MLM's users on the assessment process a Toolkit was prepared including an User Guide, Glossary, and an Assessment Tool.

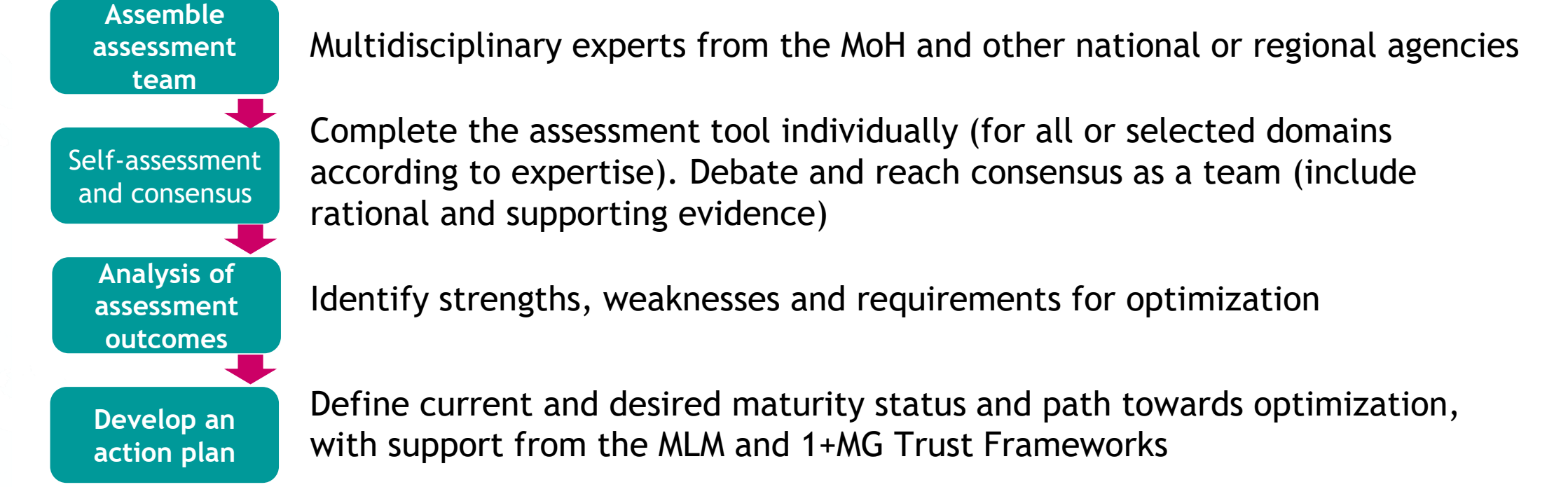
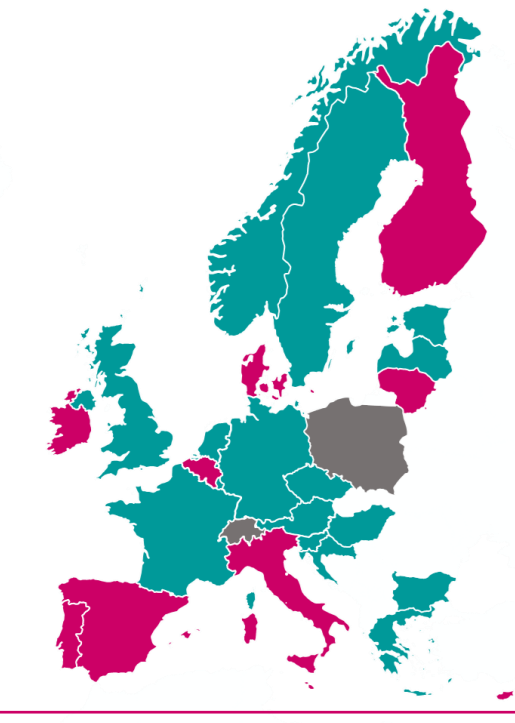


- The MLM assesses indicators in eight key domains for implementation of genomics in healthcare;
- Maturity in each domain is assessed by selecting one of five pre-defined maturity levels regarding the specified indicators.
- The five maturity levels are indicative of a maturity progression, from a non-existent or *Ad hoc* level of implementation to an optimized level of maturity characterized by a system adaptable to opportunity and change, and in support of international cooperation



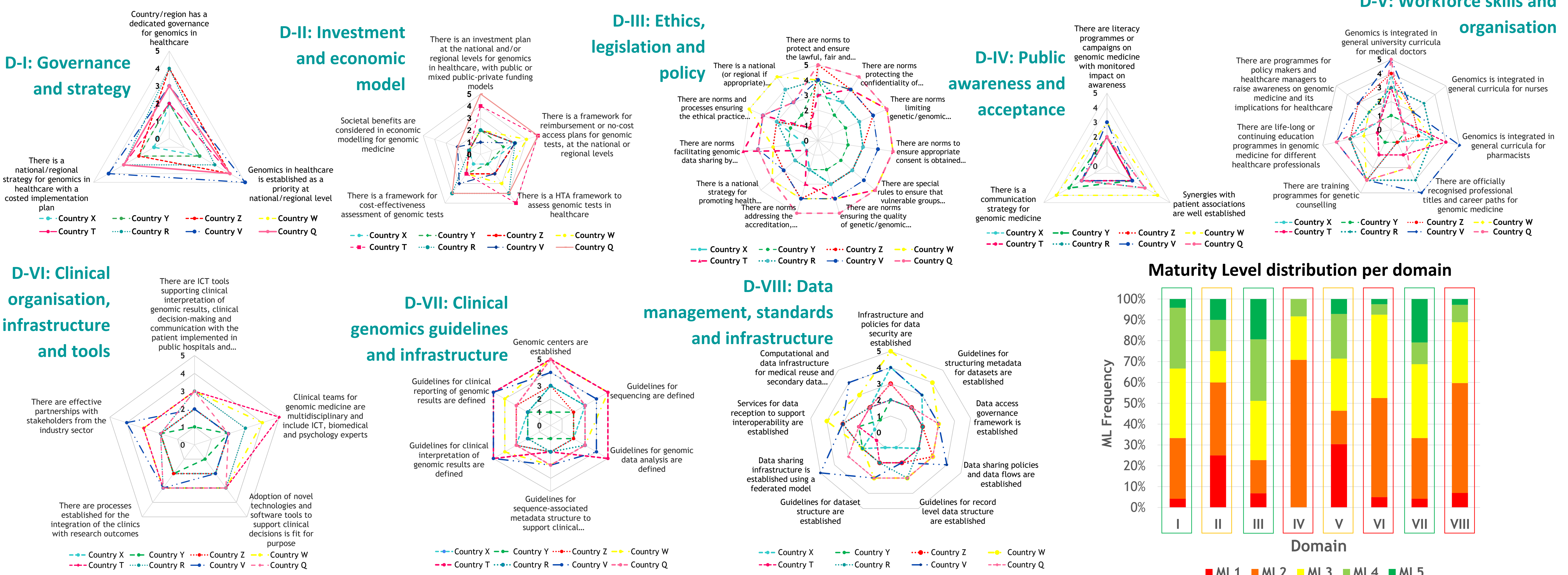
Piloting in real world settings

- The MLM Toolkit was piloted in real-life context in Belgium, Denmark, Finland, Ireland, Italy, Lithuania, Portugal and Spain (countries in pink);
- A pre-defined assessment protocol includes evidence-based assessment by a multidisciplinary team of organizations involved in healthcare, and implies reaching consensus for selection of maturity level for every indicator;



Pilot outcomes

- The pilot assessment using the MLM allow the identification of weaknesses, strengths and requirements for improvement, as a basis for the development of optimization plans in healthcare systems of each country;
- Overall, the highest MLs were found for D-III (ELSI) and D-VII (clinical genomics guidelines and infrastructures); MLs were consistently lower for D-IV (Public awareness and acceptance) and D-VIII (Fairification and security of data);
- D-I (Strategy and governance) showed mid to high MLs: Genomic medicine is a Priority, but still lacking strategy and governance; D-VI (Clinical organization, infrastructure and tools) show generally low MLs, indicating that ICT and other tools need investment; inter-sectorial partnerships need development;
- The widest asymmetries across these countries were found for D-II (Investment and economic models) and D-V (Workforce skills and organization).



Conclusion

- This work developed and validated a MLM for the assessment of the genomic maturity of healthcare systems; The pilot in eight European countries identified best practices, challenges and asymmetries across Europe, which may contribute to the development and monitoring of action plans for optimization;
- The MLM is a valuable tool to assess genomic practices in healthcare systems, identifying and prioritizing areas that need further investment nationally or at European level.

