Key updates from 1+MG/B1MG

1+MG Trust Framework
B1MG Maturity Level Model
1+MG and industry engagement

Chair: Ruben Kok

01

1+MG Trust Framework

What is it, what will it look like?

Serena Scollen

ELIXIR, 1+MG Coordination Group, B1MG Coordinator

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1+MG Trust Framework

Recommendations and guidelines, best practices
Stakeholder input

To realise the 1+MG vision

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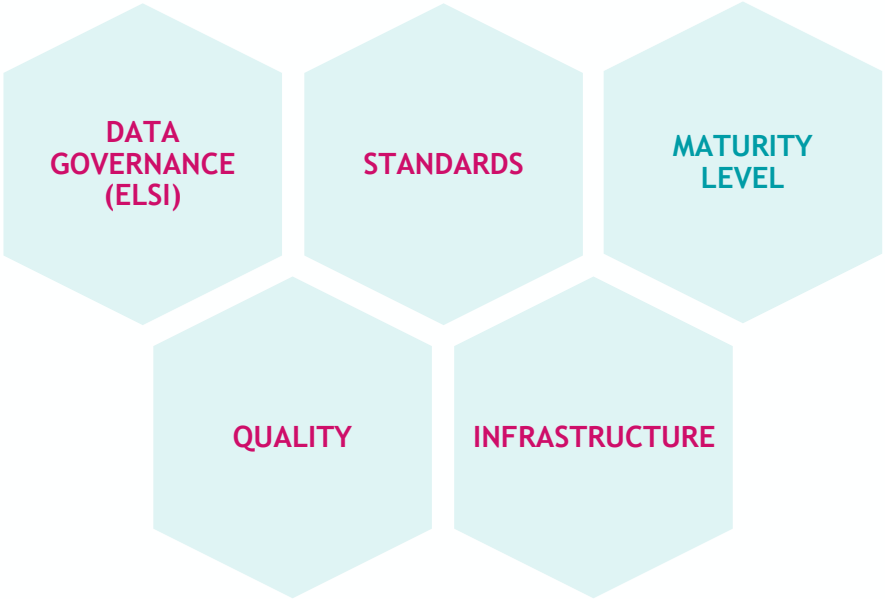
1+MG Trust Framework

Genomics Data



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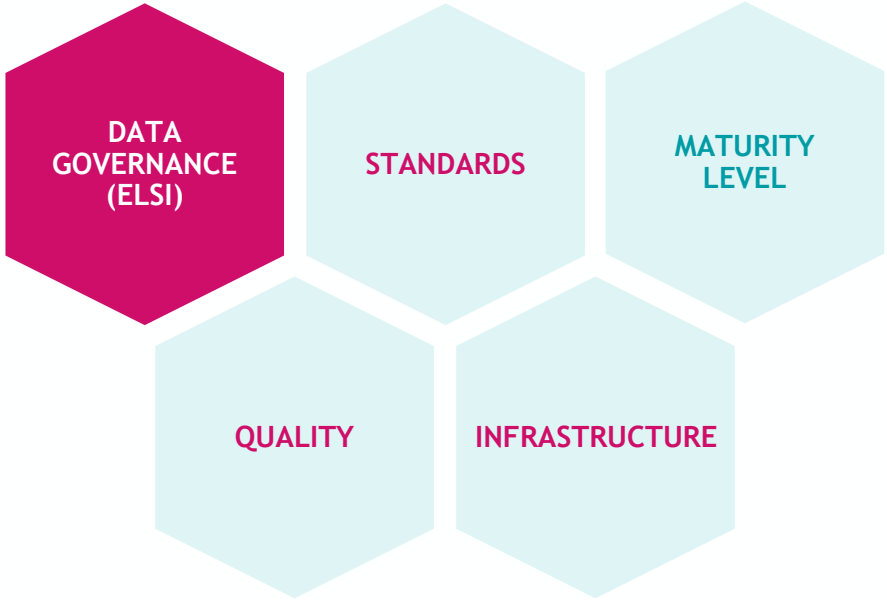
1+MG Trust Framework

Genomics Data



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DATA GOVERNANCE (ELSI)

Data governance policy *How to translate ethical norms and legal requirements into an efficient and scalable procedural framework for data inclusion and use?*	Under governmental review
Legally compliant legal framework for operations *How to interpret and apply the various legislations for secondary use of health and genetic data?*	Under country expert review
Transparency and Consent Guidance *How to comply with ethics norms and legal requirements for the inclusion of data into 1+MG?*	Under stakeholder review
Data protection by design and default *How to translate the 1+MG ELSI trust framework into the information management and IT infrastructure of 1+MG?*	Under B1MG internal review

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DATA GOVERNANCE (ELSI)

Data Governance Policy

Make data governance responsible, flexible, scalable and feasible

Countries can use the policy to:

- Offer appropriate protection for data subjects
- Ensure ethical norms such as incidental findings, consideration of vulnerable subjects and groups are considered
- Compatibility with national and EU legal frameworks
- Have a homogeneous appearance as “one” resource towards users
- Support differing national organisational and legal landscapes

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DATA GOVERNANCE (ELSI)

Legally compliant legal framework for operations

Give recommendations to Member Countries on legal instruments for the implementation

In-depth legal analyses to:

- Interpret GDPR for secondary use: understanding purposes and further processing, requirements of consent, identification of controllers and processors…
- Analysis of national legal situation
- Identification of possible legal bases for the operation
- Requirements for secondary use from the Data Governance Act
- Compatibility with the European Health Data Space

DATA GOVERNANCE (ELSI)

Transparency and consent guidance

For drafting information and consent forms

- Ensure data subjects are informed
- Implement GDPR requirements and ethical norms
- Give the right level of choice to data subjects
- Provide “checklists” to validate consent forms
- Provide 1+MG specific phrases to be used and example text for context specific elements

All data subjects will have to receive 1+MG specific information and the opportunity to opt-in or out of 1+MG (“re-consenting”)

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DATA GOVERNANCE (ELSI)

Data Protection / ELSI by Design and Default

Offer protection to all stakeholders

- Recommendation of organisational and technical safeguards to ensure secure processing at each step
- Translation of data governance into information management and IT infrastructure
- Definition of ELSI metadata along the data life cycle
- Plan workflows and tools with data protection / ELSI in mind

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1+MG Trust Framework

Genomics Data

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QUALITY STANDARDS

Documented best practices in sharing and linking phenotypic and genetic data (updated version)	Ready for review (Oct/Nov-22)
Phenotypic and clinical metadata framework 1v0	Endorsed by 1+MG Special Group
Phenotypic and clinical metadata framework 2v0	Ongoing (Dec-22)
Phenotypic and clinical metadata framework	Ongoing (May-23)
Quality metrics for sequencing	available
Best practices for Next Generation Sequencing (NGS)	Ongoing (May-23)

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STANDARDS

Data standards

Promote data interoperability

- Advice on which standards are of preference or mandatory
- An approach for working with common standards
- Documented best practices in sharing and linking phenotypic and genetic data
- Minimal data sets (where relevant) for 1+MG use cases

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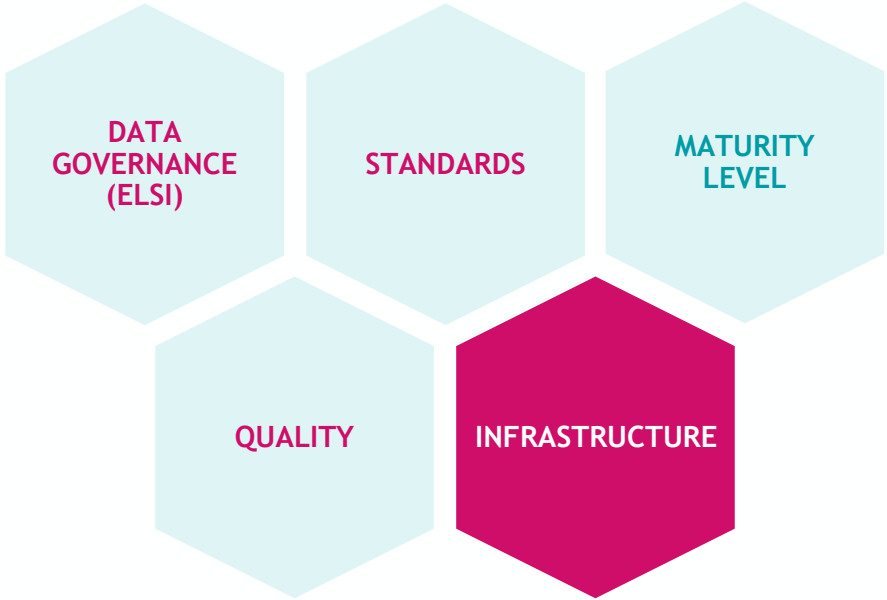
1+MG Trust Framework

Genomics Data



Assessment for Healthcare

INFRASTRUCTURE

Secure cross-border data access roadmap (Deliverable)	Available
Secure data access demonstrator (PoC) (Video / Slides)	Ongoing (May-22)
Secure cross-border data access roadmap updated	Ongoing (May-23)

Data discovery

Access management tools

Data processing

Data reception

Storage and interfaces

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INFRASTRUCTURE

Infrastructure

Enable real crossborder use cases

- Recommendations for the standards that connect infrastructure components, as well as implementations to support the five functionalities
  - Interoperability with other projects / data spaces
- A proof of concept demonstrator for cross border data access
  - Rare Disease use case
  - Now working on Cancer use case
- Enabled phenotypic queries using Beacon V2 (data discovery service)

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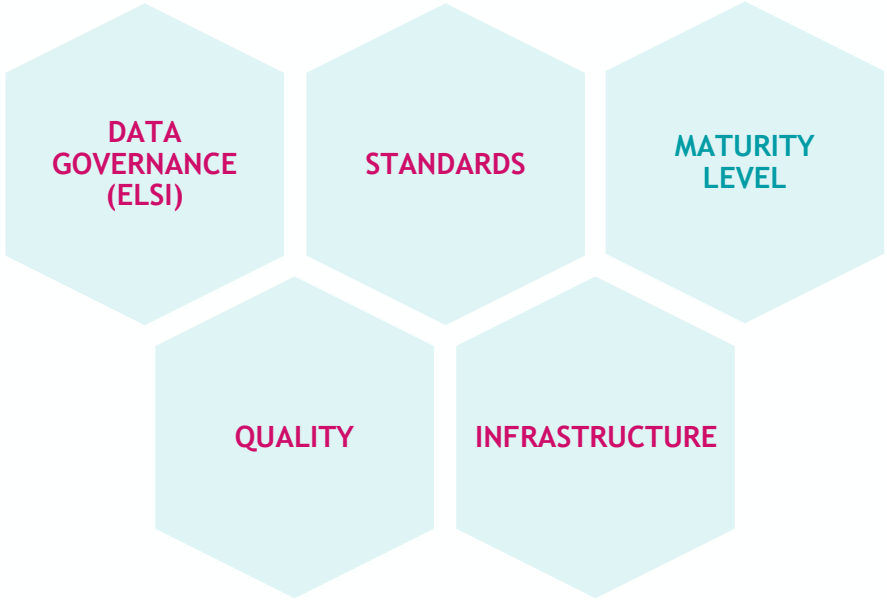
1+MG Trust Framework

Genomics Data



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Audience Q&A Session

ⓘ Start presenting to display the audience questions on this slide.

Infrastructural components of trust framework (in summary)

- Recommendations for the standards that connect infrastructure components, as well as implementations to support the five functionalities
  - Interoperability with other projects / data spaces
- A proof of concept demonstrator for cross border data access
  - Rare Disease use case
  - Now working on Cancer use case
- Discovery now via Beacon V2 - Rare Disease discovery query replicated using V2 functionality
- Processing functionality demoed via GPAP for rare disease - proposal on cBioPortal for cancer
  - Processing also extended to discovery - e.g. variant calling and annotation in a federated context

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Recommendations from SF2021 - Technical infrastructure (WP4)

Recommendation	Achievement	Next steps
1. Synthetic data set for cancer	Working with WG9 on synthetic data - example mutation and phenotypic data	Minimal metadata model being mapped to phenopackets
2. Standardised cancer data analysis software	Identification of cBioportal with WG-9. Functions for data collection, visualisation. *Analysis, portability?*	Data analysis functions and cBioportal portability (with EOSC4CANCER)
3. Generate PRS through distributed analysis across data hubs	In discussion with CINECA WP4 PRS use case - based on GA4GH standards	Initiate contact with WG-10 (Common and Complex Diseases)
4. Collect and link host + pathogen data and communities	In discussion with WG-12. Virus sequence data analysis workflow development project with EuroHPC, ELIXIR, https://permedcoe.eu (https://Covid19dataportal.org). Discussions with COVID-19 Host Genetics Initiative https://www.covid19hg.org.	E.g. Federated EGA technology-based services are needed to manage sensitive host data, biosample services to link pathogen and host data
5. Requirements for locating data beyond projects	TEHDAS (WP7 infrastructure) discussions with https://tehdas.eu Joint Undertaking	Continue alignment between 1+MG, TEHDAS, EHDS2Pilot and EMA/Darwin infrastructure alignment
6. Multiple use cases different infrastructure requirements	Extended WG-5 proof-of-concept work from rare diseases (WG-8) into cancer (WG-9). Presented PoC to infectious disease (WG-12) and planning work.	Complete cancer PoC with WG-9. Start the GDI project with more use cases (Pillar III).
7. Enable data-visiting approaches to enable sharing data with other stakeholders without data leaving the organisation	Use of containerisation (e.g. Singularity) to allow repeatable and benchmark analyses to be sent to the data - can support data generation and harmonisation	Biohackathon to extend containers into secure data, as well as GA4GH workflow standards where containerisation is not ideal

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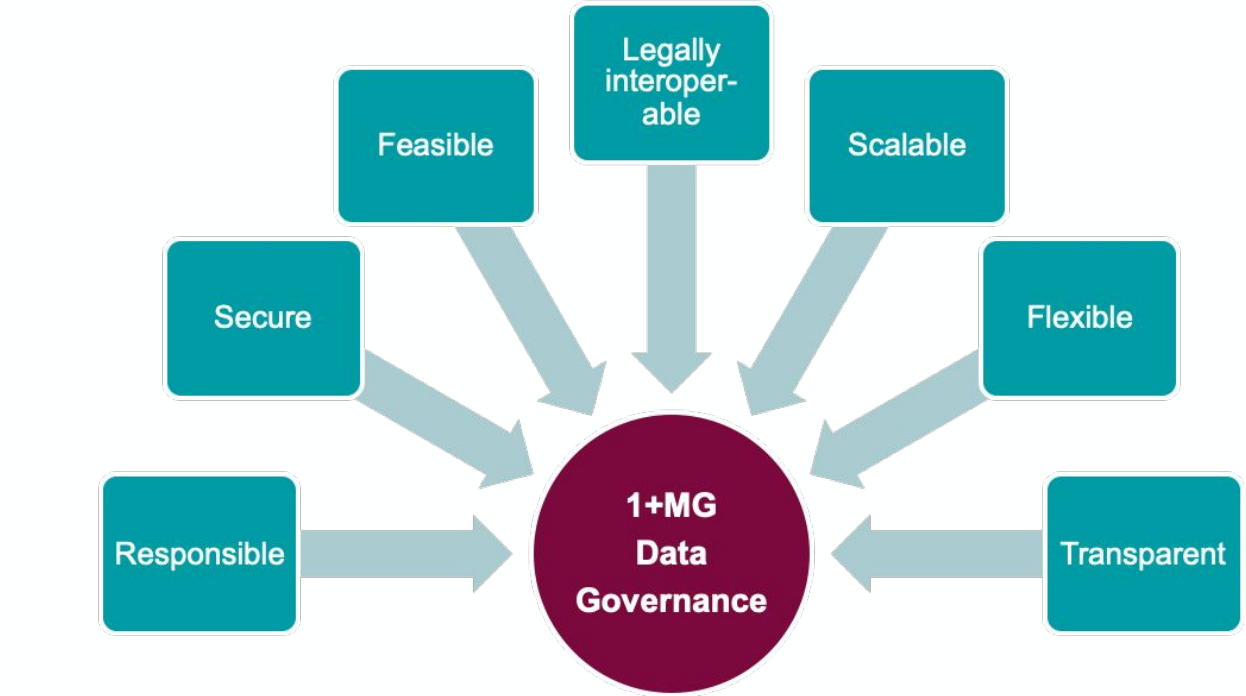
Data Governance components of the trust framework (in summary)



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Realising a practice of personalised medicine & health

Long-term strategy: cross-border access to genomic data, implementation of genomics-based health
1+MG Group, National Mirror Groups and Thematic Working Groups
Use Cases Working Groups: cancer, infectious diseases, rare diseases, common complex diseases, industry
Genome of Europe (GoE)

Stakeholder feedback, recommendations and guidelines, design and testing

1+MG Trust Framework
- **Data governance**
- **Standards and Quality**
- **Infrastructure**
- **Maturity model**

Scale up and sustainability

Sustainable federated cross-border access to genomic health data

Genomic Data Infrastructure
European Health Data Space
National infrastructures & personalised medicine & health programmes

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1+MG Trust Framework

Recommendations and guidelines, best practices
Stakeholder feedback

GENOMICS DATA

Governance (ELSI)
Standards
Quality
Infrastructure

ASSESSMENT FOR HEALTHCARE

Maturity model

To realise the 1+MG vision

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Data Governance components of the trust framework: DATA GOVERNANCE POLICY

Key principles for design and implementation

- Offer appropriate protection for data subjects
- Ethical norms such as incidental findings, consideration of vulnerable subjects and groups
- Compatibility with national and EU legal frameworks
- Be as flexible as possible for Member Countries to allow for different national organisational and legal landscapes
- Have a homogeneous appearance as “one” resource towards users

→ **Make data governance responsible, flexible, scalable and feasible**

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Data Governance components of the trust framework: LEGAL FRAMEWORK

In-depth legal analyses

- Interpret of the GDPR for secondary use:
  Understanding purposes and further processing, requirements of consent, identification of controllers and processors…
- Analysis of national legal situation
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- Requirements for secondary use from the Data Governance Act
- Compatibility with the European Health Data Space

→ **Give recommendations to Member Countries on legal instruments for the implementation**

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Data Governance components of the trust framework: TRANSPARENCY & CONSENT

Guidance for drafting information and consent forms

- Ensure data subjects are informed
- Implement GDPR requirements and ethical norms
- Give the right level of choice to data subjects
- Provide “checklists” for mapping forms on for completeness
- Provide 1+MG specific phrasings to be used
- Provide example text for context specific elements

→ **All data subjects will have to receive 1+MG specific information and the opportunity to opt-in or out of 1+MG (“re-consenting”)**

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Data Governance components of the trust framework: DPbDD / ELSIbDD

Data Protection / ELSI by Design and Default

- Plan workflows and tools with data protection / ELSI in mind
- Translation of data governance into information management and IT infrastructure
- Recommendation of organisational and technical safeguards to ensure secure processing at each step
- Definition of ELSI metadata along the data life cycle

→ **DPbDD / ELSIbDD aims to offer protection to all stakeholders: data subject, data holder, infrastructure provider, 1+MG countries**

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Data standards and quality components of Trust Framework (in summary)

- Relevant quality measures for genome sequencing have been defined
- An Interlaboratory Comparison on germline whole genome sequencing and a benchmark on Tumor/Normal whole genome sequencing for somatic mutation detection are underway
- For the ILC 20 major European sequencing are participating - ethics are cleared, test items defined and acquired, signature of MTAs is being finalized and distribution of test items imminent
- For the benchmark 14 major European sequencing are participating - ethics are cleared, test items defined and acquired, signature of MTAs is being finalized and distribution of test items imminent
- Collection of participants results is scheduled for January 2023
- The conclusion of the two proficiency testing programme is due to be delivered in June 2023

1+MG Trust Framework

Genomics Data

Assessment for Healthcare

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Data quality
quality requirements

- Defined relevant quality measures for genome sequencing

- An Interlaboratory Comparison (ILC) is underway on:
  - Germline whole genome sequencing
  - Benchmarking on Tumor/Normal whole genome sequencing for somatic mutation detection
- Results due June 2023

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