



Beyond One Million Genomes



1 Million Genomes

1+MG Maturity Level Model

Glossary

Acceptance

Perceived usefulness of genomic medicine to patients. Recognition from citizens, patients and patients' associations of a positive impact of the use of genomic medicine on patients levels of satisfaction

API

Application Programming Interface. A software intermediary that allows two applications to talk to each other.

Awareness

Public's level of understanding about the importance and implications of genomic medicine.

Centrally

Based within a national or regional node.

Clinical interpretation of genomic results

Translation of the technical output of a clinical genetic or genomic test into potentially clinically actionable information.

Cost-effectiveness assessment

Cost-effectiveness analysis is a form of economic analysis that compares the relative costs and outcomes of different courses of action.

Costed implementation plan

A multi-year roadmap that enables governments to prioritise interventions, engage stakeholders around one strategy, forecast costs and mobilise resources to meet identified gaps, namely to implement genomics in healthcare systems.

Data protection

Certainty that personal data is used fairly, lawfully and transparently - for specified, explicit purposes - in a way that is adequate, relevant and limited to only what is necessary, accurate and, where necessary, kept up-to-date, for no longer than is



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necessary, and handled in a way that ensures appropriate security, including protection against unlawful or unauthorised processing, access, loss, destruction or damage.

Data reuse

Reuse, or secondary use, of health data for purposes other than the primary reason for which they were originally saved. Other purposes may include scientific research, development and innovation activities, teaching and statistics.

Data reception

Uniform processes (such as quality control and standardisation) to receive (download) or access (through API) both data and metadata in a consistent way, enabling infrastructures to adhere to global standards and principles for genotypic and phenotypic data. It includes logically describing datasets to an extent that they can become actionable on the infrastructure, even if they are stored nationally or locally. (Adapted from the 1+MG Scoping paper)

Dataset structure

The dataset is formatted in a standard way to support interoperability, i.e. via use of international standards.

Dedicated governance

The process by which decisions are made and implemented. Governance is the process by which public institutions conduct public affairs and manage public resources.

Economic model

A structured approach to help decision-makers choose between alternative ways of using resources, by weighting the cost of an action against the benefits that it provides. It is frequently used to anticipate the costs and benefits of new health care technologies, policies and regulations.

Federated model

A distributed network of repositories for sharing genomic information.

Further processing

The processing of personal data for a different purpose(s) than the initially collected.

Genetic data

Personal data related to the inherited or acquired genetic characteristics of an individual, which give unique information about his/her physiology or health, that result from an analysis of a biological sample from the individual in question. [ref. Art. 4(13) GDPR]

Guidelines for clinical interpretation of genomic results

Guidelines for translating the technical output of a genetic or genomic test into potentially clinically actionable information.

Guidelines for clinical reporting of genomic results

Guidelines for reporting the actionable results of a genetic or genomic test to the attending clinician and/or patient.

Health data



Personal data related to the physical or mental health of an individual independent of its origin (e.g. healthcare context, research, clinical trials, the data subject directly, smart devices). [ref. Art. 4 GDPR]

HTA framework

Health Technology Assessment framework. A multidisciplinary process that uses explicit methods to determine the value of health technology at different points in its lifecycle to help decision-makers make informed decisions.

ICT (clinical) tools

Information and communication technology, such as electronic health records, telehealth or online resources.

ISO

The International Organisation for Standardisation

Locally

Within a single institution, i.e. not beyond a lab, department or hospital.

Metadata

Data that provides information about other data. For example, the origin of the data, the processing details or the sharing permissions.

Multidisciplinary teams

Teams comprised of individuals who span across different areas of expertise to cover all knowledge areas required for genomic medicine.

No-cost access plan

Detailed set of rules that determines rights, duties and procedures to benefit from access to genomic tests at no cost

Norms

A set of principles of right action binding upon group members and serving to guide, control or regulate appropriate and acceptable behaviour. E.g. legislation, policies, professional regulations, codes of conduct.

Personal data

Data related to a living individual, who is likely to be identified by the data directly or combined with other data (e.g. through a pseudonym). [ref. Art. 4 GDPR]

Primary bioinformatics analysis

The initial analysis that turns the machine output of genomic sequencing into genomic information for clinical/research interpretation or other contexts.

Reception and interfaces

This consists of two areas.

(1) Reception. Uniform processes (such as quality control and standardisation) to receive (download) or access (through API) both data and metadata in a consistent way, enabling infrastructures to adhere to global standards and principles for genotypic and phenotypic data. It includes logically describing datasets to the extent that they can become actionable on the infrastructure, even if they are stored nationally or locally.

(2) Interfaces. Organisations offer interfaces (APIs) following international standards



that form the technically interoperable infrastructure backbone.
[Adapted from the 1+MG Scoping paper]

Record

A dataset record is a collection of fields of information about the same person, item or object in a database. It can be thought of as a row of information within a database table.

Secondary data analysis

The use of existing data, collected for a prior study, to pursue a research interest that is different to that of the original work. [ref: <https://sru.soc.surrey.ac.uk/SRU22.html>]

Sequence-associated metadata

Data that provides information about other data, specifically about genomic-sequence data.

Societal benefits

Any advantages, gains or improvements as a result of employing a genomic approach to a group of people (e.g. patients, citizens).

Structured dataset metadata

Metadata (data that provides information about other data) for datasets that supports data discoverability using international standards.

Vulnerable groups

Vulnerable groups of population include children, adults with diminished capacities, the elderly, racial or ethnic minorities, the socioeconomically disadvantaged, underinsured or those with certain medical conditions who are at risk for unequal healthcare access, outcomes and exploitation.

