Annex A
Genome UK: 2022 to 2025 Implementation plan for England

High level metrics

The office for life sciences (OLS) has derived a set of high-level metrics to quantify long-term changes in the genomics environment and measure progress against Genome UK ambitions. The chosen metrics will track progress in implementing Genome UK commitments by measuring the impact of actions of delivery partners. There will also be external factors which will have some influence over the changes and trends seen in the chosen metrics. Although it will not be possible to quantify the extent to which these changes can be attributed to either the implementation of specific Genome UK commitments or external factors, the proposed metrics are closely aligned with expected impacts from Genome UK commitments and have been chosen as useful indicators of success.

The metric proposals in Table 1 are in draft, and OLS will refine or expand the list of metrics based on evolving data availability and feedback received following the publication of the implementation plan. We will seek to use publicly available data wherever possible.

We are working with the devolved governments to achieve a harmonised approach across the UK where possible.

We will publish further information via a metrics baseline report in due course.

Table 1: List of proposed metrics for measuring impacts of Genome UK implementation

Proposed metric	Data provider	Genome UK commitments for which metric measures impact	Further work needed ahead of baseline report
Genomic testing activity in the NHS over time	NHS England (when publicly available) National Disease Registration Service (including the National Congenital Anomaly and Rare Disease Registration Service and the National Cancer Registration and Analysis Service)	Offer all patients with a rare genetic disorder a definitive molecular diagnosis using tests that will support research into their condition wherever possible Offer genomic testing to all people with cancer where it would be clinically beneficial	OLS will explore scope for breakdown into data categories (including whole genomes, whole exomes and large cancer panels), as well as demographic and regional breakdown. A final list with definitions will be included in the baseline report

Proposed metric	Data provider	Genome UK commitments for which metric measures impact	Further work needed ahead of baseline report
Number of diagnoses returned to the NHS following diagnostic research by approved researchers	Genomics England	Offer all patients with a rare genetic disorder a definitive molecular diagnosis using tests that will support research into their condition wherever possible	To be confirmed
Number of people registered as having a genomically-confirmed rare disease, registered in the National Congenital Anomaly and Rare Disease Registration Service	National Congenital Anomaly and Rare Disease Registration Service	Offer all patients with a rare genetic disorder a definitive molecular diagnosis using tests that will support research into their condition wherever possible	To be confirmed
Number of genomes available for research in national trusted research environments / secure data environments	UK Biobank Genomics England NIHR BioResource Our Future Health	Ensure that clinical genomic testing and genomics research contribute to powerful national data resources Establish a clear set of standards for genomic and health data Support the join-up of the NHS and research community with scalable and secure informatics systems, both for clinical decision support and large-scale data processing and analytics	OLS will further explore scope for breakdown into data categories (including whole genomes, whole exomes, and genome-wide association studies (GWAS) arrays) and what trusted research environments will be in scope. A final list with definitions will be included in the baseline report

Proposed metric	Data provider	Genome UK commitments for which metric measures impact	Further work needed ahead of baseline report
Number of researchers accessing named trusted research environments / secure data environments	Genomics England UK Biobank Our Future Health	Develop consent and data standards that support innovation for the benefit of patients and the NHS, while maintaining trust in the safe, appropriate and responsible use of data Develop systems to enable federated access to data for research use to enable comparisons across multiple datasets Track the use of our datasets and maintain an upward trajectory of both numbers and user experience Incentivise the genomics research community to prioritise areas of high NHS unmet need	OLS will work with delivery partners to establish whether the number of organisations or number of individuals can be tracked over time. The established methodology will be published in the baseline report
Number of clinical trials recruiting participants on the basis of genomic data (genomically-informed clinical trials)	NIHR BioResource Our Future Health	Support hypothesis-driven identification, recruitment, phenotyping and biosampling of uniquely informative cohorts of patients Work at a UK level to ensure there is equitable access to opportunities to participate in clinical trials informed by genomic data commitments	OLS will establish what clinical trials this information relates to, and how many can be evaluated as part of this metric. Full details of the scope of the metric will be included in the baseline report and OLS will work further with delivery partners and data providers

Proposed metric	Data provider	Genome UK commitments for which metric measures impact	Further work needed ahead of baseline report
Number of participants enrolled in research studies with a genetic element (including polygenic risk scores)	Genomics England UK Biobank Our Future Health NIHR BioResource	Support hypothesis-driven identification, recruitment, phenotyping, and biosampling of uniquely informative cohorts of patients Work at a UK level to ensure there is equitable access to opportunities to participate in clinical trials informed by genomic data commitments	OLS will establish what research studies collect this information and how many can be evaluated as part of this metric. Full details of the scope of the metric will be included in the baseline report and OLS will work further with delivery partners and data providers