Summary of the roadmap document adopted on 4 February 2020 by the signatories of the Declaration

Towards access to at least 1 million sequenced genomes in the EU by 2022
and novel markets for services, products, and medicines.

Based on data-driven innovation, and the development of new
these scientific and medical advances are likely to form the
consequence of personalised medicine. For the economy,
result from the greater efficiencies and cost savings that are
...outcomes will include: better health, better quality of life, and
socio-economic benefits to Europe. It will result in faster and
Personalised medicine

Away from “one size fits all” treatments towards individually
individual’s genomic characteristics. It can initiate a shift
disease diagnosis, treatment, and prevention based on each
individual’s genomic characteristics. It can initiate a shift
environments, personalised medicine can support improved

Understanding an individual’s characteristics,
including their genetics, is key to delivering the right
care to the right person at the right time.

Massive population-based analysis and significant
advances in the early diagnosis and prevention of disease
can only be achieved when a critical mass of collected data
has been reached. This will be based on data and knowledge
shared between organisations and across borders. Indeed,
the larger the available sample collections and associated
datasets, the more robust the analysis. Among future
activities will be interpreting common yet complex disorders
such as diabetes; understanding cancer in its many varieties;
recognising individuals’ susceptibilities to infectious diseases;
and diagnosing and finding leads for the treatment of
patients with very rare diseases. In all of these domains, it
will be important to generate large enough cohorts of people,
with their genetic information, to investigate diseases and
conditions bearing in mind the great diversity of Europe’s
population.

Collecting and analysing genomes will also help experts
understand and leverage pharmacogenomics – the branch
of genetics concerned with determining an individual’s
response to a particular medication – more effectively. As a
result, doctors will be able to choose the right treatment for
the right patient by predicting reliably the side-effects of
medicines and identifying who would be a non-responder to
a specific therapy.

Today, the key is to enable sharing of genomic and other
data. Research and clinical data need to be used together by
linking the information rather than bringing it all together
physically in a central place. This needs to happen without
breaching individuals’ privacy or compromising data security.

Data-sharing is the future for Europe’s health and care. It will
support and help in implementing a number of European
Union initiatives and activities, such as Europe’s Beating
Cancer Plan, European Data Strategy including a European
Health Data Space, and the European Open Science Cloud.

Tackling cancers, common complex,
infectious, and rare diseases

Genome sequencing – the ability to analyse the unique
sequence of an individual’s DNA – helps doctors and
scientists understand diseases better (for example, how to
predict, prevent, diagnose and treat diseases). Genomics can
help determine the future risks of various debilitating
diseases, such as cancer and many common complex
diseases, as well as supporting the diagnosis of rare diseases.

Tackling cancer is of fundamental importance for Europe’s
future. It has been estimated that, by 2035 globally, the
number of cases of cancer will double. However, many
cancers can be prevented, by implementing steps already
made clear through research and investigation. Genomics will
help to tackle the cancer challenge by delivering the
additional information needed for doctors and policy-makers
to make appropriate decisions: actions that are core to the
implementation of Europe’s Beating Cancer Plan.

Rare diseases affect no more than one person in 2,000.
Nevertheless, in Europe alone, there are millions of such
patients who have between 6,000 and 8,000 forms of
rare diseases. It can take up to 5 or 6 years to diagnose the
precise condition of a person with a rare disease. It is clearly
important to reduce this timespan. Understanding and using
genomics offers great potential to do so.

Pressing, new imperatives have arisen. Throughout spring
and summer 2020, a new coronavirus disease (COVID-19)
spread in Europe and worldwide. To face the growth of the
disease, rapid action has been needed. Understanding the
susceptibility of individual citizens to COVID-19 infection, and
the great variety of morbidities the virus may cause, is
therefore another major challenge that can be supported by
genomics.
Louise has a long family history of breast cancer. One day, she heard on TV that mutations in the BRCA genes increase the risk of breast cancer by up to 80%. Although it turned out that she doesn’t have mutations in these genes, her risk of developing the condition is still high due to her family history. In order to detect early potential breast cancer, she started breast cancer screening at age 35, much earlier than the usual recommended routine screening. A few years later, Louise indeed developed breast cancer. To determine the most suitable type of treatment, she underwent a genomic test assessing the utility of chemotherapy versus other treatments in her case. Test results showed that Louise would not benefit much from chemotherapy, so doctors opted for a more effective personalised treatment, which also prevented her from experiencing the unpleasant side effects of chemotherapy.

In 2015 Martin was born in Dublin, Ireland. Doctors recorded an abnormally small head and face, as well as a slow development rate. Genome sequencing identified many possible candidates for the genes that were causing his slow development. However, a precise diagnosis would be needed to start treatment. Genome sequencing identified many possible candidates for the genes that were causing his slow development. In this case this worked not only for Martin’s condition but also for the Spanish patient. Development in sequencing of cancer genomes are rapidly improving insights and predictive power of genomic tests on risk, prognosis and treatment of cancer. The 1+MG network will greatly increase the availability of this knowledge to oncologists, improving the choices for patients and the reducing over-treatment.

Justyna heard about the ongoing biobanking project in her country in the media, so she read more about the genetic risks of certain diseases. Her healthcare provider recommended a genomic analysis to estimate her Polygenic Risk Score (PRS), a test for most common diseases which was just introduced as a new clinical trial in the university hospital. The genomic analysis also showed that one specific statin could increase her risk of muscle inflammation and should be avoided. Justyna is now more aware of how to prevent CAD and make adjustments in her lifestyle, as well as to receive the right treatment and regular check-ups, if needed. Europe is currently developing PRS tests for early identification of risks factors for common diseases. The 1+MG project will yield genome sequences for many European citizens, in combination with phenotype information, all collected with informed consent in biobanks across Europe. This will further the implementation of PRS testing in regular health care and will boost the availability of cheap prevention throughout Europe and elsewhere.

Pablo (27 years old) develops severe COVID-19 symptoms. Shortly after, his brother Pedro (30 years old) is also tested positive. Both need to be treated in intensive care. The two brothers do not belong to any of the known risk groups, but their close family relationship suggests a genetic risk factor. Imagine we would have the EU federated genomic data network of 1+MG in place offering European clinicians the possibility to receive alerts when they encounter patients with similar characteristics. Patients’ increased susceptibility to life-threatening conditions caused by an infectious disease such as COVID-19 could be rapidly discovered and the necessary treatment adapted and personalised.
Cooperating Europe-wide on genomics

By 2018, several countries in Europe had established national initiatives that aim to generate genomic data and link it to phenotypic data (the observable health properties of a person). To help transform health and care in Europe, genomic and linked phenotypic data need to be accessed not only within but also across national boundaries.

Moving towards federated secure access to genomic data

Making collective use of Europe’s genomic data is a considerable challenge. Data security aspects play a critical role: trust needs to be ensured along several dimensions. This initiative pursues a federated approach which will bring together the strengths and qualities of national programmes, while drawing on their synergies and ensuring full compliance with data protection rules and standards. The initiative will not pool genomic data in a central repository. Instead, accredited users will be able to send approved algorithms to the many local databases involved.

Collaborating on the 1+ Million Genomes Initiative

The 1+ Million Genomes initiative, also referred to as ‘1+MG’, was launched on 10 April 2018, the European Union’s second Digital Day, when 13 European Union countries signed the declaration: “Towards access to at least 1 million sequenced genomes in the European Union by 2022.” The countries agreed to work collectively on a cross-border network of national genome cohorts associated with other relevant data. The aim is to advance data-driven research and healthcare solutions to benefit European citizens. By autumn 2020, more than 20 European countries have joined the initiative, a few others have observer status. The initiative is open for more Member States of the European Union, the European Economic Area (EEA) and European Free Trade Association (EFTA) to join it.

The 1+ Million Genomes initiative is a huge European-wide undertaking. In its endeavour to link genomic data throughout Europe, concrete preparatory work needs to take place between now and the end of 2022. Europe will be working on these solutions for the next three years.
Which are the main areas of cooperation?

To create this framework with its focus on more than one million genomes, new ways of collaborating are needed. This is why the 1+ Million Genomes initiative was founded. The signatory countries are keen to move forward on accelerating developments in genomic science into enhanced healthcare. Various areas of activity are at the core of the initiative’s work: organisation; implementing personalised healthcare in health systems in cost-effective and efficient ways; ethical, legal, and social issues; health and phenotypic data; good genomic practice and standards; and infrastructure and interoperability.

Organisation of work

Strong coordination and collaboration of all the initiative’s efforts have been put in place. This is happening through cooperation among the signatory countries, supported by expert working groups. One expert group focuses on governance and ensures overall coordination. It covers several aspects of activity, including stakeholder engagement, industry engagement, a learning framework, and education and communication.

Ethical, legal, and social issues

A number of ethical, legal, and social issues need to be brought into focus and resolved. Examples include the values and ethics underpinning this pan-European framework; the underlying legal framework and challenges of finding ways to process genomic data across borders despite legal differences; the contractual arrangements related to data use and data inclusion; and the societal implications, transparency, policies, and processes to govern data access.

Genomics innovation in national healthcare systems

Embedding genomics expertise and research outcomes into personalised healthcare will lead to greater efficiencies and, thus, cost savings. Accelerating the transfer of research knowledge into improved and cost-effective healthcare is key (including improving disease prediction rates, diagnosis, and the tailoring of therapies and treatments to individuals). This requires innovation in healthcare systems across Europe: the 1+MG initiative will help the countries involved to learn from each other in this respect. Analysing the potential costs and benefits of the adoption of genomics in healthcare will be part of these steps. The 1+MG framework will thus assist in the analysis of economic viability, and in the harmonisation of cost assessment and economic evaluation models. This will help towards generating comparable evidence across European healthcare systems and facilitate countries to make informed policy choices about the implementation of genomics-based personalised medicine.

Clinical and phenotypic data

Agreeing on a minimal set of phenotypic and clinical data to be collected in each genomics study is crucial. This agreement will enable comparative studies across data collections that may have different backgrounds. The initiative will build consensus to implement this minimal dataset across European laboratories and clinics.

Good genomic practice and standards

When sequencing genomic data, it is important to draw on good practice. As a result, guidelines and appropriate standards and forms of accreditation will need to be drawn up. Genome sequencing standards, benchmarking and metrics, as well as good quality control of data, are needed.

Infrastructure and interoperability

The initiative’s proposed infrastructure uses a federated approach. Each country in Europe is invited to contribute to the endeavour, by bringing to the enterprise the strengths and qualities of its own national programme. The initiative is not centralised and will not pool genomic data in one place. Rather, through interoperability, it will bring data warehouses up to agreed standards and link them with a secure infrastructure. Users, like accredited research institutions, healthcare professionals, and health policy agencies, will be able to access this secure network to execute approved algorithms and analyse available data remotely.
What is the timeline and next steps?

The 1+ Million Genomes initiative has set up an intensive agenda of activities to reach its goals by 2022. Its signatory countries and expert groups are working together on many stepwise, concrete actions. By aligning the investments and advances expected in 2020-2022, the aim of creating a cohort of at least 1 million sequenced genomes can be achieved.

Laying out the 1+MG Roadmap 2020-2022

Between 2020-2022, three phases of activity will concentrate on engaging, translating, and driving the 1+MG initiative forward.

2020 – Engage – Engaging countries and other stakeholders in governance and collaboration

- Assemble national, regional, and local public authorities as well as a broad range of international stakeholders in the initiative. They will link together different genomic medicine resources in line with the initiative’s declaration.
- Define a governance model of coordination and cooperation. The cooperation model will cover the terms and conditions needed for secure access to genomic data and associated health data across borders, and the use of national and regional data collections for research and improving healthcare.

2021 – Translate – Translating the mission into concrete infrastructure, guidelines, and pilots

- Pilot the technical infrastructure for distributed, authorised and secure access to national and regional genetic data, and facilitate the interoperability of registries and databases.
- Support the development of an ethical and legal framework for cross-border access to genomic datasets to advance research across Europe, while taking appropriate measures to protect the privacy of individuals.

2022 – Drive – Moving forward on sharing, scaling, and sustaining the initiative

- Embed the coordinated data governance framework and infrastructure for Europe-wide large-scale processing of genome and health data in national and regional science and healthcare systems.
- Scale activities to enable distributed and secure access to research cohorts of at least 1 million sequenced genomes.

The underpinning aim of this three-year initiative will be to support a European data strategy that reinforces the creation of data spaces in key strategic sectors and domains of public interest. In the health sector, this will translate into a European health data space, essential for making advances in preventing, detecting, and curing diseases and for making informed decisions that will improve the accessibility, effectiveness, and sustainability of health-care systems.

A Horizon 2020 project “Beyond 1 Million Genomes” (B1MG) will provide support and coordination to the implementation of the roadmap.

For more general information, the 1+ Million Genomes initiative encourages people to visit its website: https://ec.europa.eu/digital-single-market/en/european-1-million-genomes-initiative
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