



# 1+ Million Genomes

## Roadmap 2020-2022

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**

## Why genomics for health?

Interpreting people's health characteristics – including their genomes – is key to delivering effective health and care. Genome sequencing and data linking can help doctors predict, prevent, and diagnose diseases and treat people and patients better based on their individual genomic characteristics. Many more people could benefit from sharing data about their genomes and from doctors' access to the wealth of linked genomic databases. People with four areas of disease – cancers, common complex diseases, infectious diseases, and rare diseases – may especially benefit. The potential advantages to health systems through enhanced prevention policies are enormous, thanks to available reference genomic data for screening and personalised medicine. Europe's health systems are likely to gain from the savings that result from higher disease prediction rates, earlier diagnoses, and the specific tailoring of therapies to individuals. Benefits will come to the economy too: many data-driven innovations and the opening-up of new and novel markets to health and care-related services and products lie ahead.

### Understanding the role of genomics and personalised medicine

**Every individual is unique.** So too is every genome. Combined with an understanding of people's lifestyles and environments, personalised medicine can support improved disease diagnosis, treatment, and prevention based on each individual's genomic characteristics. It can initiate a shift away from "one size fits all" treatments towards individually designed therapies and treatments.

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

**Personalised medicine** is expected to bring significant socio-economic benefits to Europe. It will result in faster and more accurate diagnosis of diseases, better identification of the likely responses of individuals to medication, and advances in preventive medicine. For people, the beneficial outcomes will include: better health, better quality of life, and increased life expectancy. For health systems, savings will result from the greater efficiencies and cost savings that are the consequence of personalised medicine. For the economy, these scientific and medical advances are likely to form the basis of data-driven innovation, and the development of new and novel markets for services, products, and medicines.

*There is huge promise contained in genomic information for Europe's people, healthcare systems, and innovators.*

**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 to 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.

*Researchers are learning that nearly all conditions and diseases have a genetic component. Today, more than 30 million Europeans are affected by rare diseases, of which over 80% have a genetic origin. Inherited genetic mutations play a major role in some cancers.*

**It is vital to understand a person's genetic predisposition to disease.** Besides cancers, other conditions needing attention include all types of diabetes, cardiovascular diseases, asthma, and depression as well as all common diseases with a complex, partly genetic, background. Individuals may not be born with such diseases but may be at high risk of acquiring them.



**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**  
**IMPROVING  
CANCER TREATMENT**

**BREAST CANCER**

Louise has a long family history of breast cancer. One day, she heard on TV that mutations in the BRCA1 and BRCA2 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.

**CANCER SCREENING**

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.

**TREATMENT**

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.

Developments in the 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.

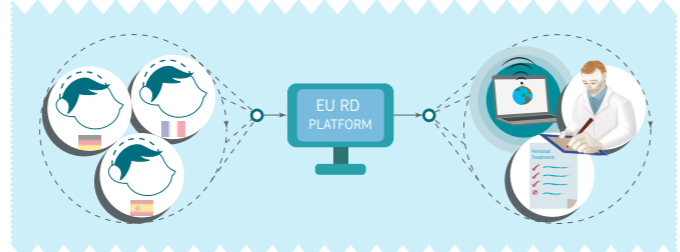


**MARTIN**  
**IMPROVING PROGNOSIS  
FOR RARE DISEASES**

**MICROCEPHALY**

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.

**EUROPEAN PLATFORM ON  
RARE DISEASES REGISTRATION**



Launching a query through an EU federated platform that facilitates matching of cases with similar phenotypic and genotypic profiles allowed his doctors to find a second case in Spain with similar symptoms. Comparison of the sequencing results of the Irish and Spanish patients highlighted one mutation both had in common, which pointed to a defect that could be counteracted by supplying a specific metabolite.



Matching of cases with similar phenotypic and genotypic characteristics through an EU federated platform facilitates reaching an accurate diagnosis and treatment for rare disease patients with different backgrounds in separate countries. In this case this worked not only for Martin's condition but also for the Spanish patient.



**JUSTYNA**  
**PREVENTING COMMON  
AND COMPLEX DISEASES**

**POLYGENIC RISK SCORE**  
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.

**CORONARY ARTERY DISEASE**

This genomic analysis showed that she has a PRS in the top 5% for Coronary Heart Disease (CAD). Drugs such as statins and other preventive measures lower the cholesterol levels in the blood and reduce the CAD risk, so her doctor suggested to initiate statin treatment and make some lifestyle changes.

**PREVENTION**

Now Justyna has to take statins. 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 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 phenotypic 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 & PEDRO**  
**TACKLING HIGH  
SENSITIVITIES TO  
INFECTIOUS DISEASES**

**COVID-19**

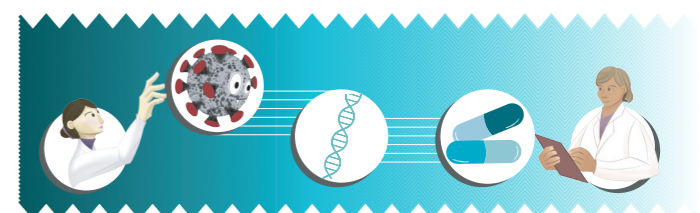
Pablo (27 years old) develops severe COVID-19 symptoms. Shortly after, his brother Pedro (30 years old) is also tested positive. Both need to be treated in intensive care. The two brothers do not belong to any of the known risk groups, but their close family relationship suggests a genetic risk factor.

**INFLUENCES**

**GENETIC**  
Genetic profiling of the brothers is undertaken and compared to other disease cohorts and population biobank resources. It turns out that Pablo and Pedro are carriers of a rare genetic mutation in TLR7 (Toll-Like Receptor 7). This gene on the X chromosome regulates the interferon level as a defensive response to viruses. Based on this finding, Pablo and Pedro receive a specific treatment to restore the natural capacity of their immune system to fight the virus.

**DATABANK NETWORK**

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.





## What is the 1+ Million Genomes initiative?

Pooling the knowledge encoded in genomic data is an area of great promise for Europe's citizens, healthcare systems, and innovators. One country or one research institute working on its own cannot reach the scale of genomes needed to understand and improve the health of Europe's people. Research, policy and care practice need to be brought together to focus on translating research outcomes into clinical applications and informing policy. This is why so many European countries are enrolled in the move forward on the 1+ Million Genomes initiative. Joining efforts means multiplying opportunities and will result in benefits for health and economies. Until 2023, Europe will be working on solutions to six key challenges that affect this pan-European initiative. Among them are governance, infrastructure, and interoperability challenges.

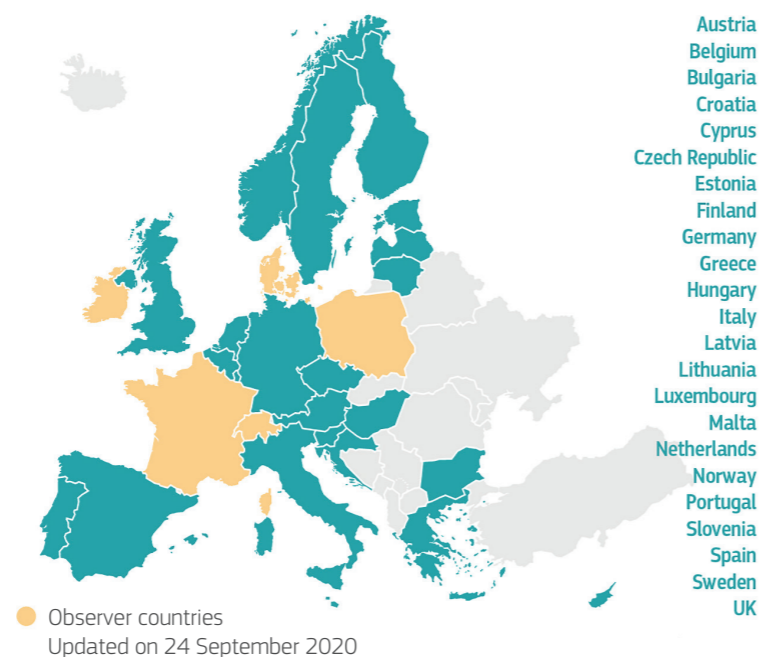
### Cooperating Europe-wide on genomics

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.

*One country or one institute working in isolation will not manage to achieve the scale needed.*

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 health-care 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.

23 countries have signed the 1+MG Declaration since 2018



### 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.

This joint effort among European countries will not start from scratch. It will connect already existing experience and data resources and will involve a wide diversity of stakeholders, including many from related ongoing programmes and initiatives.

### Collaborating on the 1+ Million Genomes Initiative

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.

Among the six activities needed are to: set up a mechanism for cross-border collaboration and data governance; examine the underlying ethical, legal, and social issues around expanding work on the collection and sharing of genomes; determine the minimal set of health data to be included in every dataset and develop good genomics practice; design the infrastructure needed and determine how to implement it; and apply the work to the main use cases of rare diseases, cancers, complex common diseases, and infectious diseases (e.g. COVID-19).

Today, Europe's countries are at very different stages of development in the field of genomics. The aim is therefore to set up and achieve a programme to upgrade and connect already existing national and regional genomic initiatives or support their establishment where this is needed.

*The aim is to create a framework that will enable linking, access, and analysis of genomic and health data in Europe, both inside and across national boundaries.*

## 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: 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.

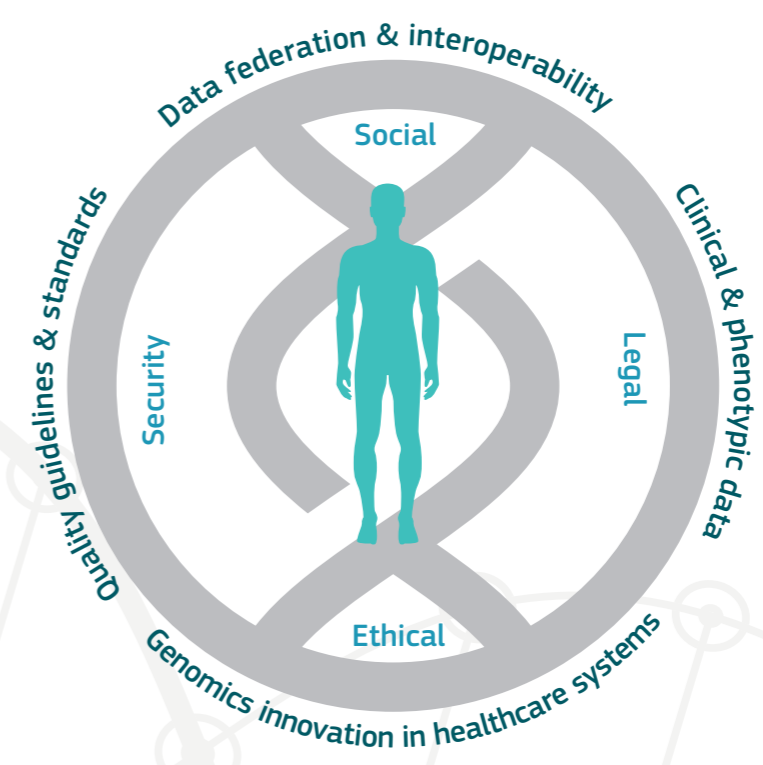
**An important priority** is to address the implications of the 2018 [General Data Protection Regulation](#) (GDPR) in relation to questions about consent, privacy, and research uses of genomic data in Europe.

### 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**.

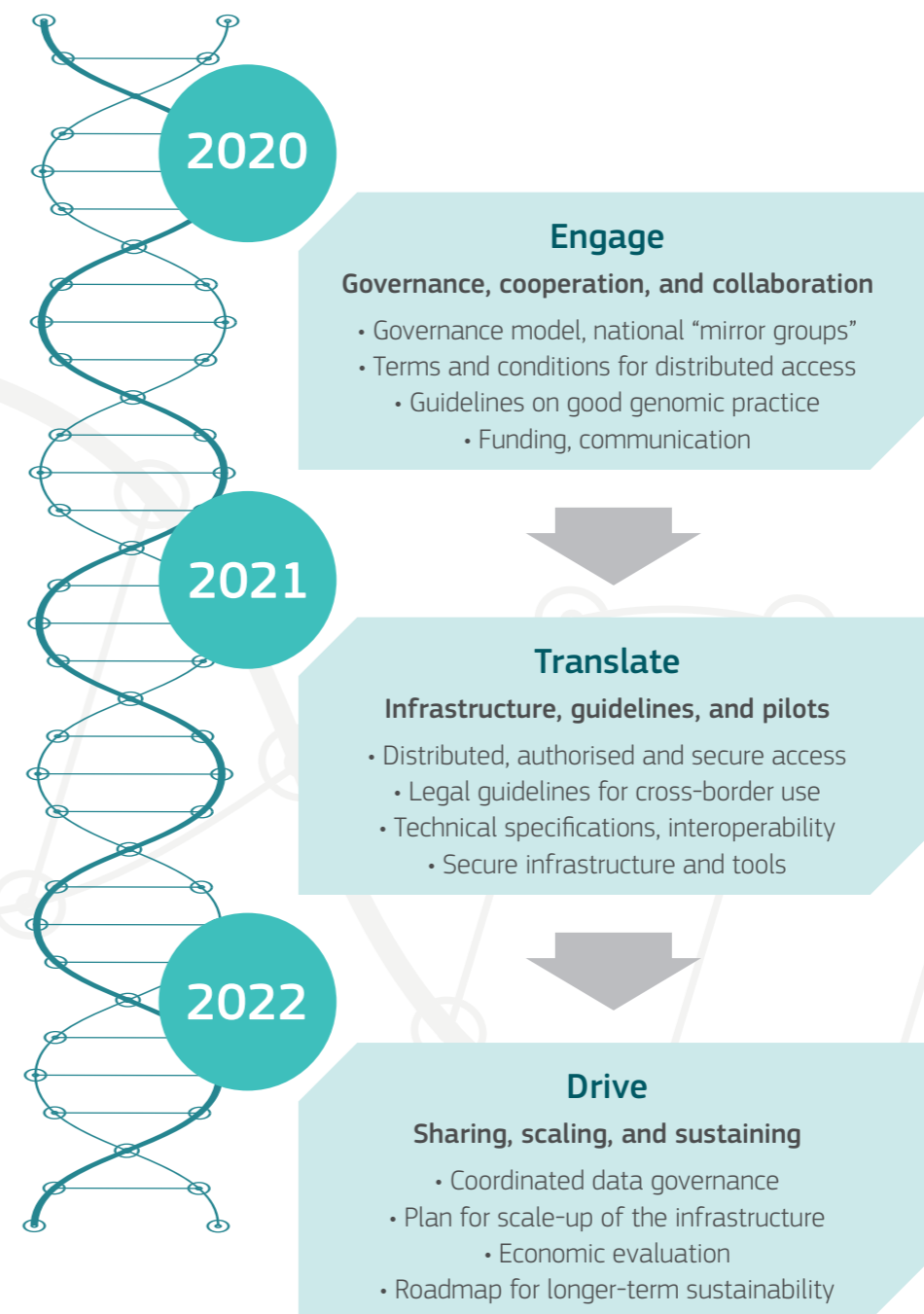
- Test-run the secure infrastructure and legal framework to enable **cross-border sharing and analysis of genetic data and associated health datasets**, anonymised as appropriate. The datasets will come from multiple European countries, build on existing **initiatives in genomics and personalised medicine**, and focus on **four use cases**.

#### 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>



# 1+MillionGenomes

Roadmap 2020-2022



More information  
**European Commission  
DG CONNECT**  
Unit H3 - eHealth,  
Well-Being and Ageing  
EUFO 02/161  
L-2557 Luxembourg  
CNECT-H3@ec.europa.eu  
<http://bit.ly/ECeHealth>



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[www.digitalhealtheurope.eu](http://www.digitalhealtheurope.eu)



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Oxfordstr. 2 · 53111 Bonn, Germany  
<https://www.empirica.com>