

The European 1+MGenomes Initiative

Reunião Anual
Sociedade Portuguesa de Genética Humana

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Personalised Medicine - concept

*Characterisation of individuals' phenotypes and genotypes (e.g. **molecular profiling, medical imaging, lifestyle data**) for tailoring the right **therapeutic strategy** for the right person at the right time, and/or to determine the **predisposition to disease** and/or to deliver timely and **targeted prevention**".*

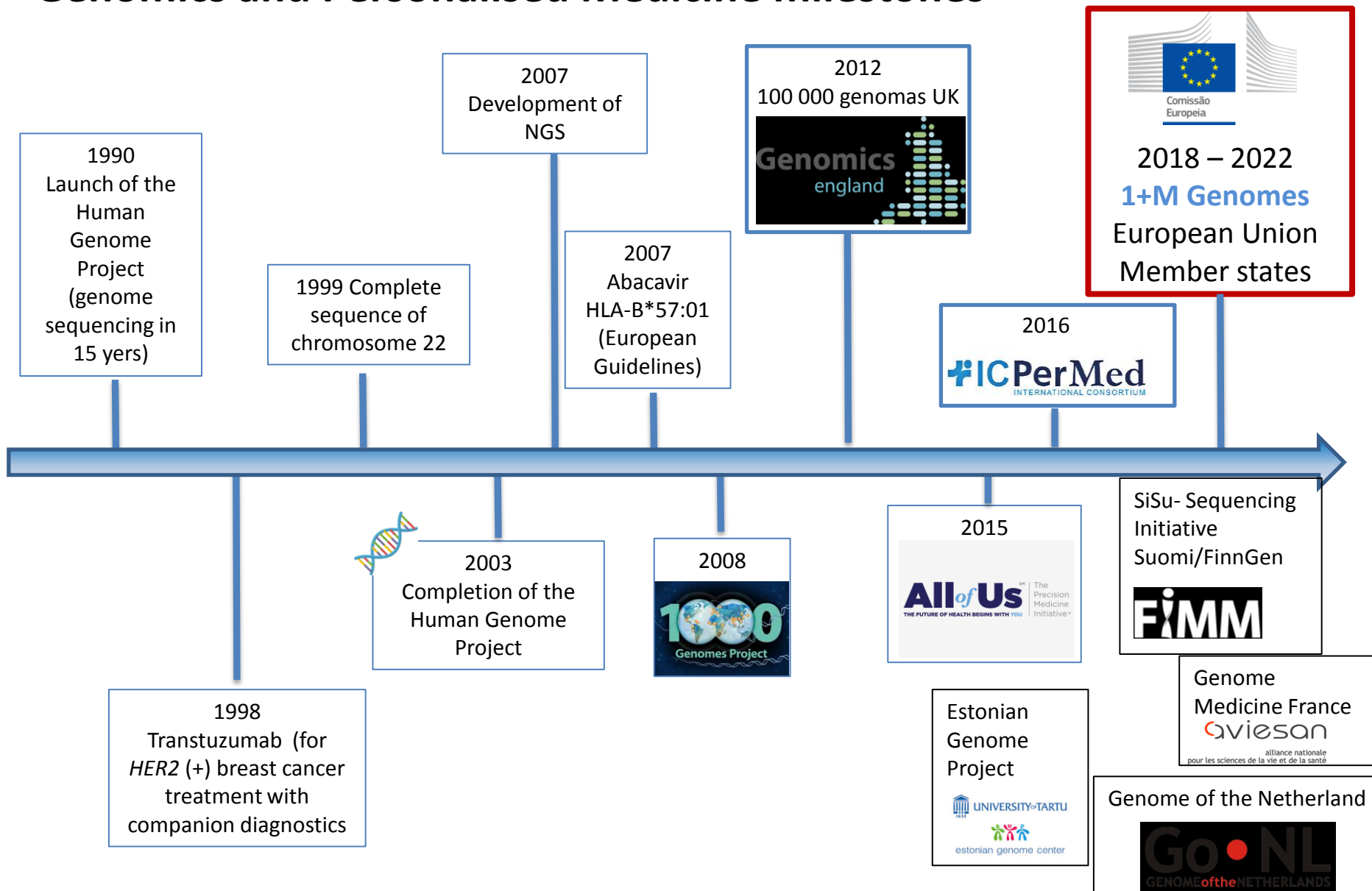
According to: Horizon2020 and the European Council Conclusions on personalised medicine for patients (2015/C 421/03)

Personalised Medicine and genomic profiling

The future is now?



Genomics and Personalised Medicine milestones



Declaration of Cooperation

"Towards access to at least 1 million sequenced genomes in the European Union by 2022"

*"This is a **game changer** for European **health research and clinical practice**: sharing more genomic data will improve understanding and **prevention of disease**, allowing for more **personalised treatments** and targeted drug prescription"*

April 10, 2018 - 13 European signatory countries



COUNTRIES AGREED TO COOPERATE IN LINKING GENOMIC DATA ACROSS BORDERS:

Signatories agree to work together to **provide cross-border, data-driven health and care solutions to benefit citizens of the Union**. As part of this goal, the signatories will work towards **building a research cohort of at least 1 million sequenced genomes accessible in the EU by 2022**.



● Observer countries

The initiative is open to Member States of the European Union, the European Economic Area (EEA) and the European Free Trade Association (EFTA) and it is facilitated by the European Commission.

Main objective:

Declaration for delivering cross-border access to **genomic databases**



1 million **genomes accessible** in the EU by 2022



Linking access to existing and future genomic databases across the EU



Providing **proper scale** for research with clinical impact



The 1+M Genome Initiative - Scope

- **Federated framework** allowing secure and authorized **cross-border access** to genomic and other health data across the EU, supporting research and healthcare
- Allow users to **search and access the data** through a user friendly and effective **data governance structure** building on existing national and European initiatives
- Ensure that **citizens, researchers and healthcare systems in Europe can benefit** from the **full potential of genomics** to advance targeted health care interventions leading to better **prevention, early diagnosis and treatment of diseases**

Aims - signatories will cooperate to:

1. Define a **coordination mechanism** of national, regional and local public authorities to **link ongoing genomic medicine initiatives**;
2. Ensure distributed, authorised, secure **access to national and regional banks of genetic and other relevant data** for the advancement of science and innovation;
3. Define a **governance model of cooperation** for distributed **access to genomic data across borders**, and usage of data;
4. Develop **technical specifications** for secure **access and cross-border exchange of genomic datasets** and facilitate **interoperability** of registries and databases to support research;

Aims - signatories will cooperate to:

5. Develop a secure **infrastructure and tools** to enable **cross-border sharing and analysis of genetic and other datasets**, building upon existing infrastructure;
6. Develop a coordinated **data governance framework** to facilitate **large-scale processing of health and related data** to support shared health policy goals, achieve **better health** for the citizens, **sustainability of health systems**, boost **large-scale data-driven** biomedical/clinical R&D;
7. Promote the use of **open standards and data management** systems to ensure **interoperability** of genomic and other health data for research on personalised medicine and genetic diseases;
8. Strengthen cooperation on the **implementation of the GDPR** on processing of personal data concerning health.

Benefiting the citizen/patient?



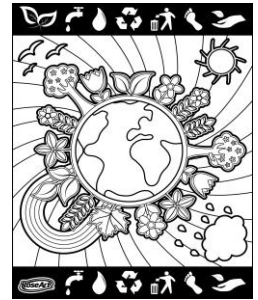
Genome sequence



Health data - EHR



Life-style information

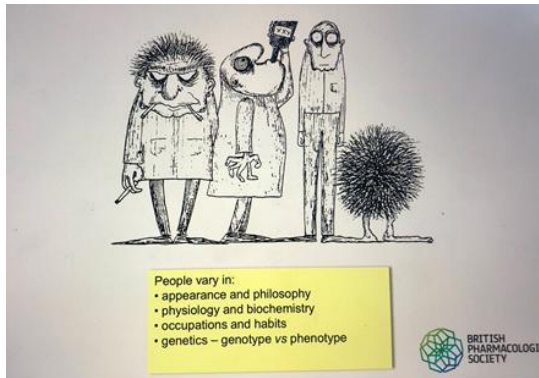


Environmental exposure



Individual health profiling

Why 1 M genomes (or more)?



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- **Statistical power to understand complex phenotypes;**
- **Population dimension to identify rare disease cases/cancer variability**

Benefits for the citizen/patient:

- Biomarkers for early diagnosis and therapeutic intervention (cancer/rare diseases)
- Genomics of ADR/Drug efficacy
- Prevention of common disorders by estimating disease risk through genomic and other profiling



Improve health outcomes

Personalised Medicine

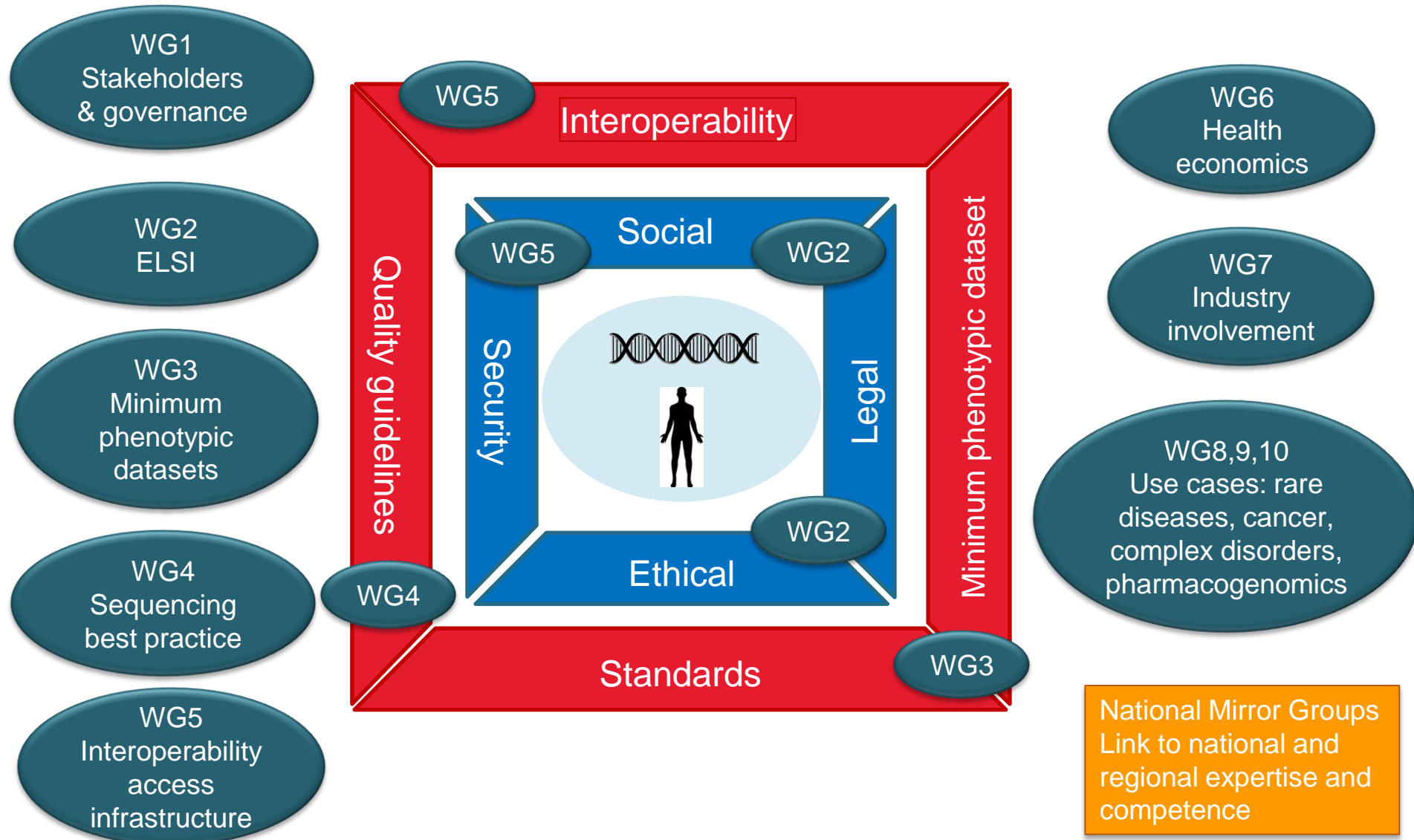
Benefits for healthcare, innovation and society:

- **Accelerate research** – data availability for joint analysis, generalize across population datasets
- **Sharing resources and expertise** – improved infrastructure, knowledge expertise, guidelines, technology
- **Common solutions** for ELSI issues
- **Data sharing** – improved interoperability, less data fragmentation, less data silos and better access
- **Synergies** for innovation, I&D translation to clinical practice, better healthcare and more prevention
- **Economic evaluation with societal perspective** – new models and approaches for healthcare systems, better sustainability of healthcare systems
- **European competitiveness** – benefits for healthcare, economy development, investment, jobs

Challenges:

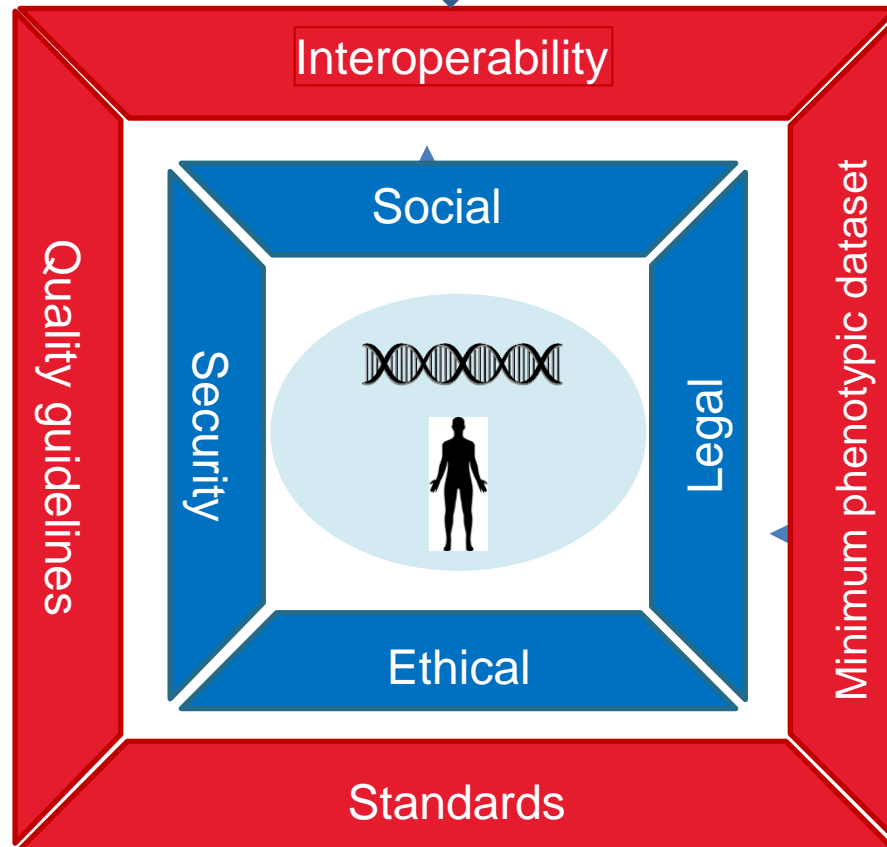
- **Citizen participation** with full understanding of benefits and implications of genome sequencing and sharing;
- **GDPR** applications vs national laws ;
- **Cross-border** data sharing;
- **Infrastructure for sharing** - sequencing, bioinformatic analysis and data sharing;
- Selection of **phenotypic datasets**;
- **Case studies** – Rare disease, cancer, complex common disorders, pharmacogenomics;
- **Health economics**;
- **Private sector involvement** - IP, contribution, access to data;

A common framework for federated access



Join forces:

**National clinical
genomics initiatives**



Governance

Advisory Board

1+M Genomes Board – Member States and the European commission

Stakeholder forum -
Scientists, Industry, European projects and initiatives, Patient groups
A stakeholder representation to provide input from a wider range of actors

National mirror groups
- ensuring alignment and input from national and regional initiatives and expertise

Executive Board
Implementation and accountability



Secretariat - Coordination structure

Coordination and Support Action



Developing the Roadmap 2020-2022

In Portugal – mirror groups:

- Populational samples, biobanks 
- National genomic IS and expertise 
- Health data 

SPMS_{EPE}
Serviços Partilhados do
Ministério da Saúde
- Data sharing IS 

elixir
PORTUGAL
- Health Cluster, Universities, R&D Centers, private sector
- Other governmental entities (eg DGS, INFARMED)

***Mirror Groups - Roadmap for Portuguese genomes
Strategy for the adoption by healthcare systems***

Conclusions - *The future is now!*

The 1+M Genomes initiative will have an enormous impact in:

- 1) **Knowledge**
- 2) **Technology**
- 3) **Diagnosis, prevention and care**
- 4) **Healthcare system models**
- 5) **Health professional backgrounds**
- 6) **Citizen literacy**
- 7) **Economic development**



THANK YOU!