# ‘1+ Million Genomes’

## MG-NL + 1+MG initiative + B1MG project

Ruben Kok, DTL – Health-RI

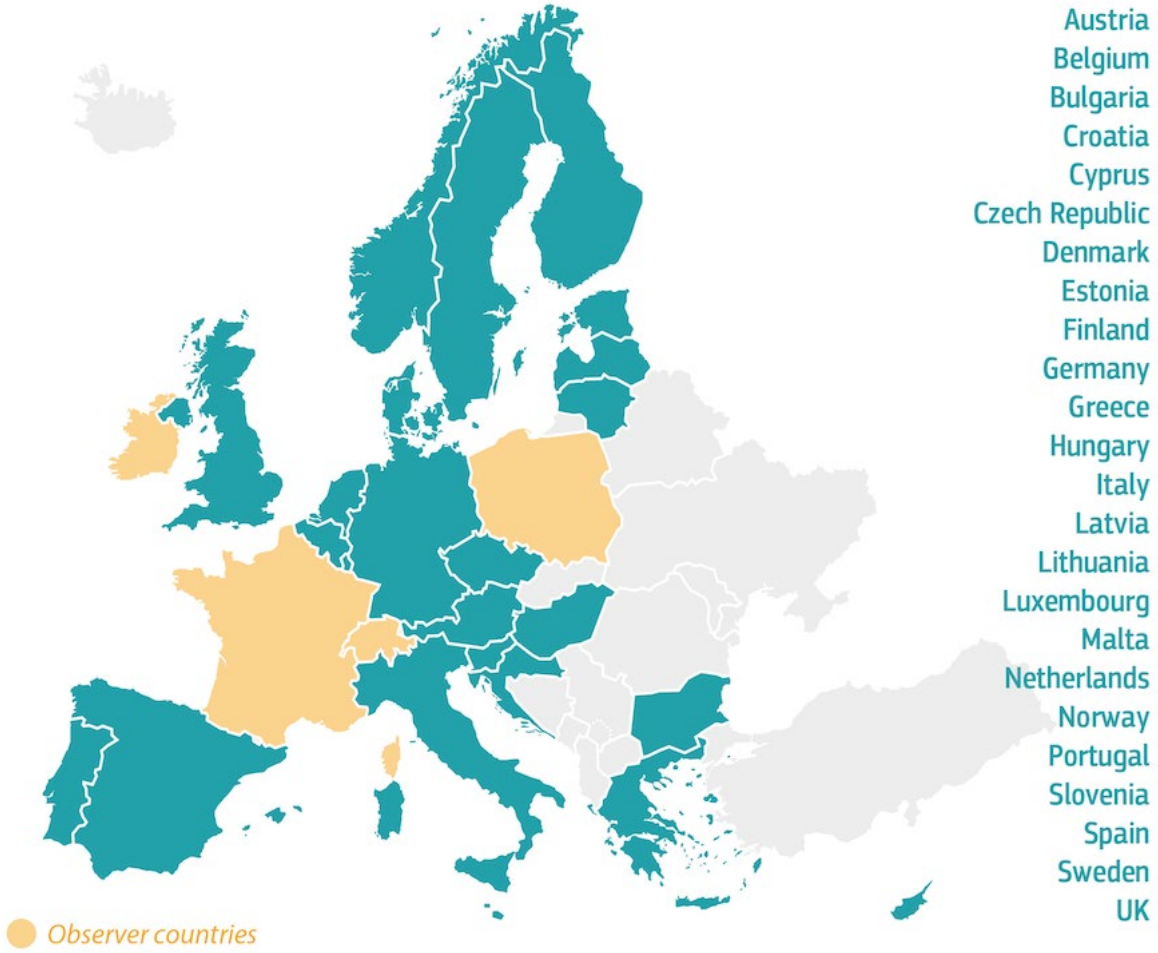
**ZonMw GGG meeting**
**March 31 2022**

health RI
enabling data driven health

DTL
DIGITAL LIFE SCIENCES

https://b1mg-project.eu

# Build cohort of 1+MG and infrastructure for federated cross-border access to genomic health datasets




## 24 countries have signed the 1+MG declaration

https://digital-strategy.ec.europa.eu/en/policies/1-million-genomes

## DECLARATION OF COOPERATION

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

Česká republika

and

Eesti Vabariik

and

Reino de España

and

Repubblica italiana

and

Κυπριακή Δημοκρατία

and

Lietuvos Respublika

and

Grand-Duché de Luxembourg

and

Repubblika ta' Malta

and

República Portuguesa

and

## 1+MG Roadmap 2020-2022

*This document has been elaborated by a drafting team consisting of the 1+MG coordination team and WG leaders. Preparatory work included several written exchanges, teleconferences and one physical meeting (10 December 2019). All experts from the relevant Working Groups had the opportunity to contribute with their inputs via WG leaders (as indicated in the table of content below). The representatives of the 1+MG signatory countries were consulted on the draft roadmap by way of a written procedure (with the deadline of 17 January 2020). The present version includes the outcome of that consultation and implements the feedback received.*

### Table of content



Adopted Feb 2020

Prof. dr. Gertjan van Ommen
28 sept 1947 - 7 nov 2020

# Roadmap and objectives

Aligned to the 1+MG roadmap

2020

**Engaging** local, regional, national and European stakeholders

… to define requirements for accessing genomics and personalised health data.

2021

**Translating** requirements for data quality, standards, technical infrastructure, and ELSI

… into technical specifications and implementation guidelines for genome-based health, based upon European best practices: 1+MG trust framework.

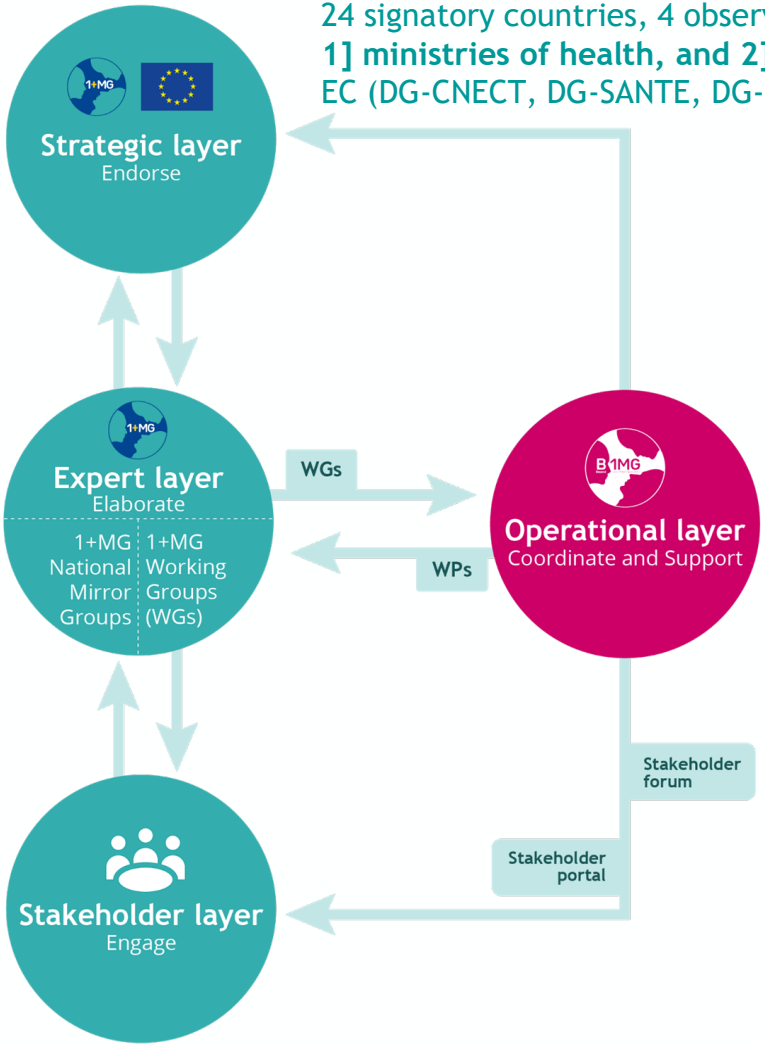
2022

**Driving** adoption and supporting long-term operations

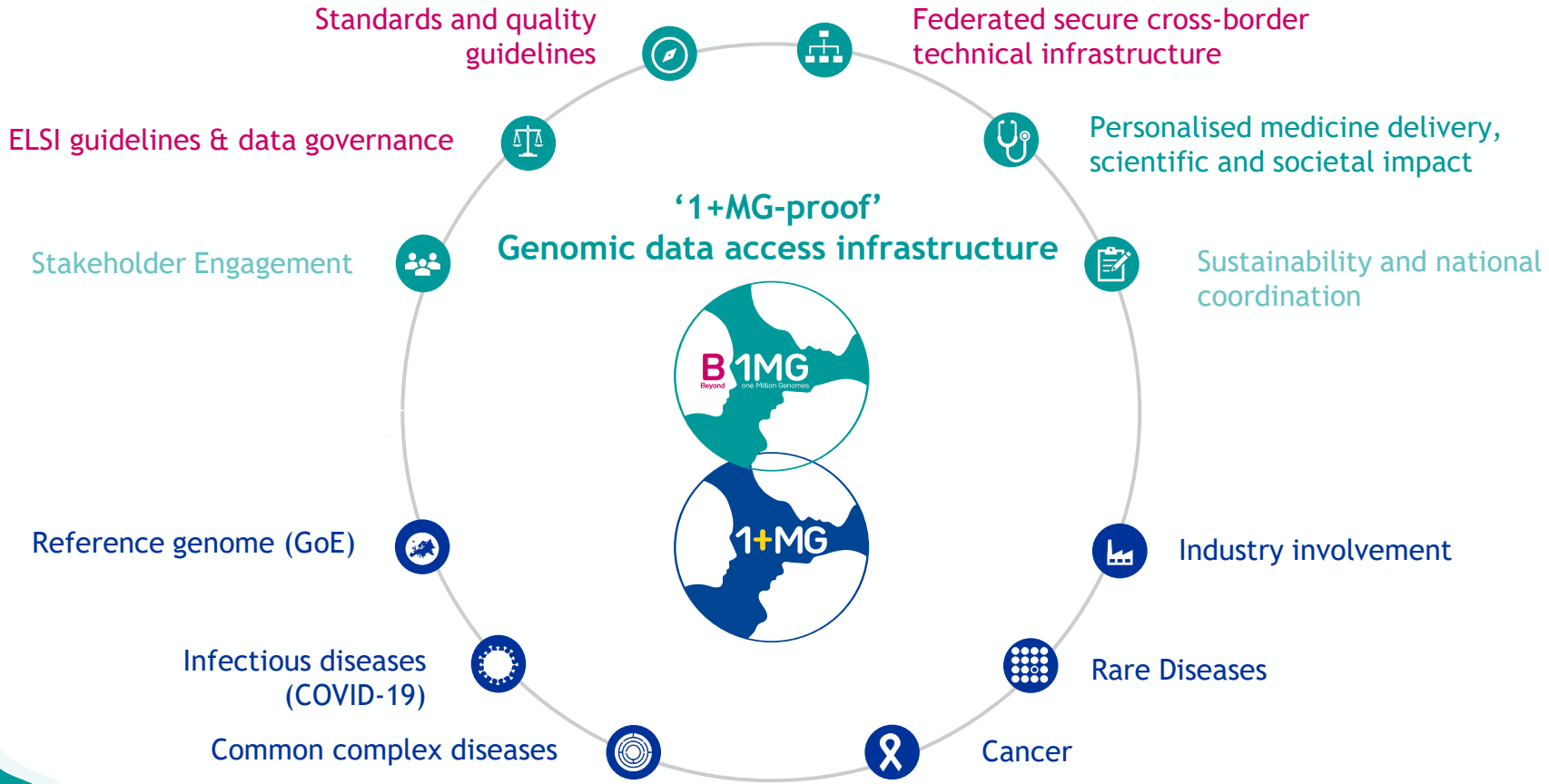
… by providing guidance on phased development via the B1MG maturity level model and a methodology for economic evaluation.

# Organisation & governance structure

24 signatory countries, 4 observer countries:
**1] ministries of health, and 2] science representatives**
EC (DG-CNECT, DG-SANTE, DG-RTD)



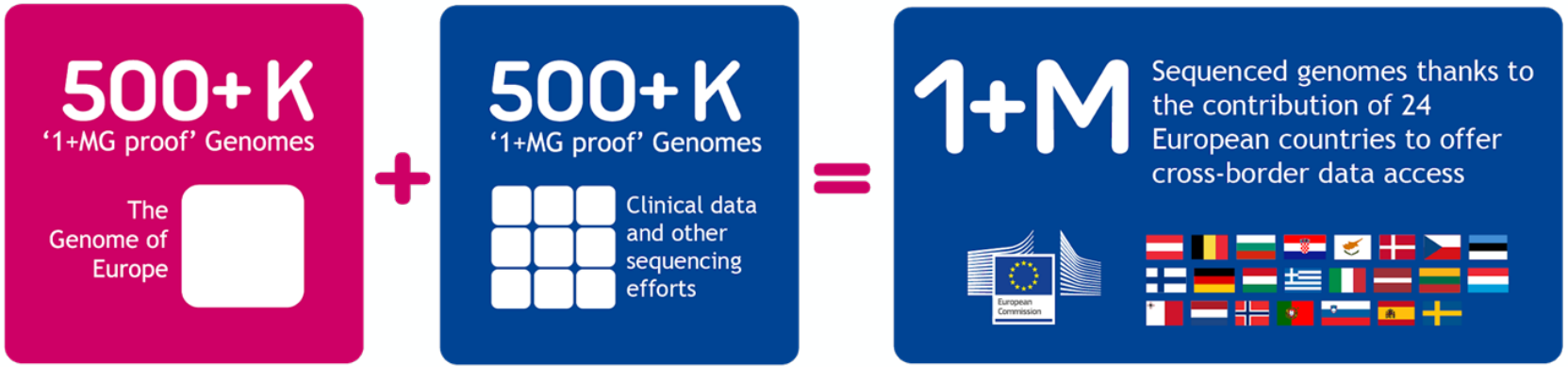
# 1+MG & B1MG teams



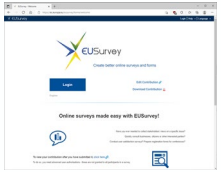
# European reference genome collection (A. Uitterlinden cs)

European commitment to achieve access to at least 1 million sequenced genomes by 2022

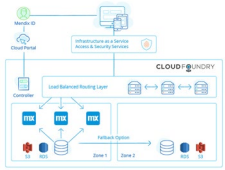
B1MG is creating legal guidance, best practices, recommendations and an infrastructure to achieve the goal



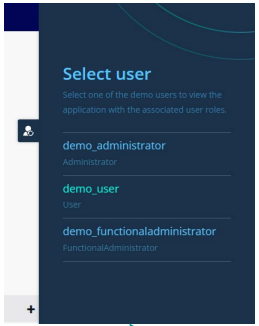
# Accessible Genomes Dashboard (J. Belien cs)

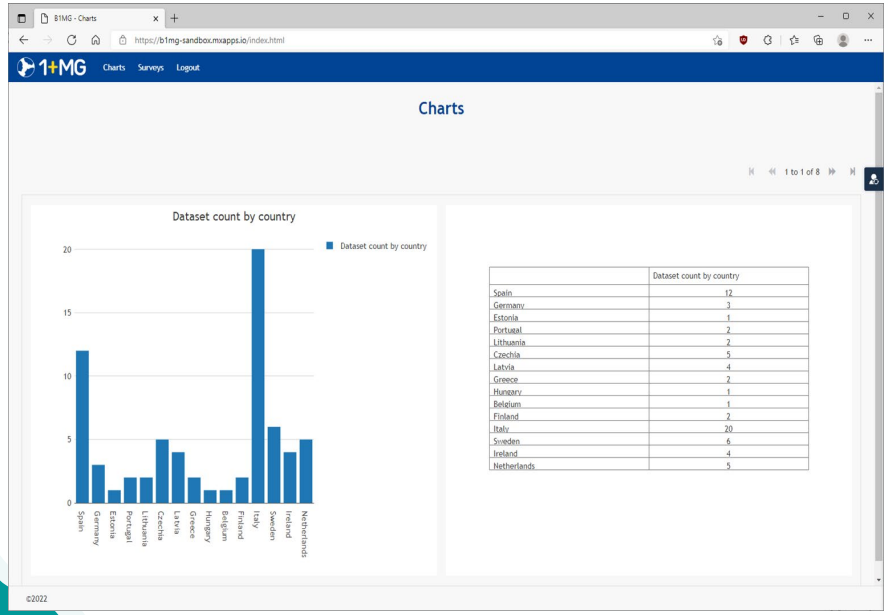


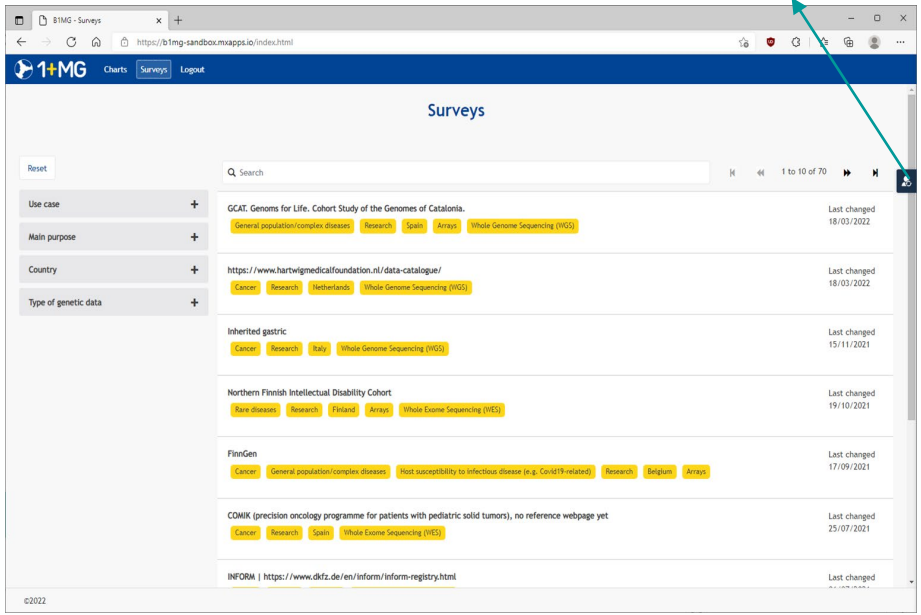

API



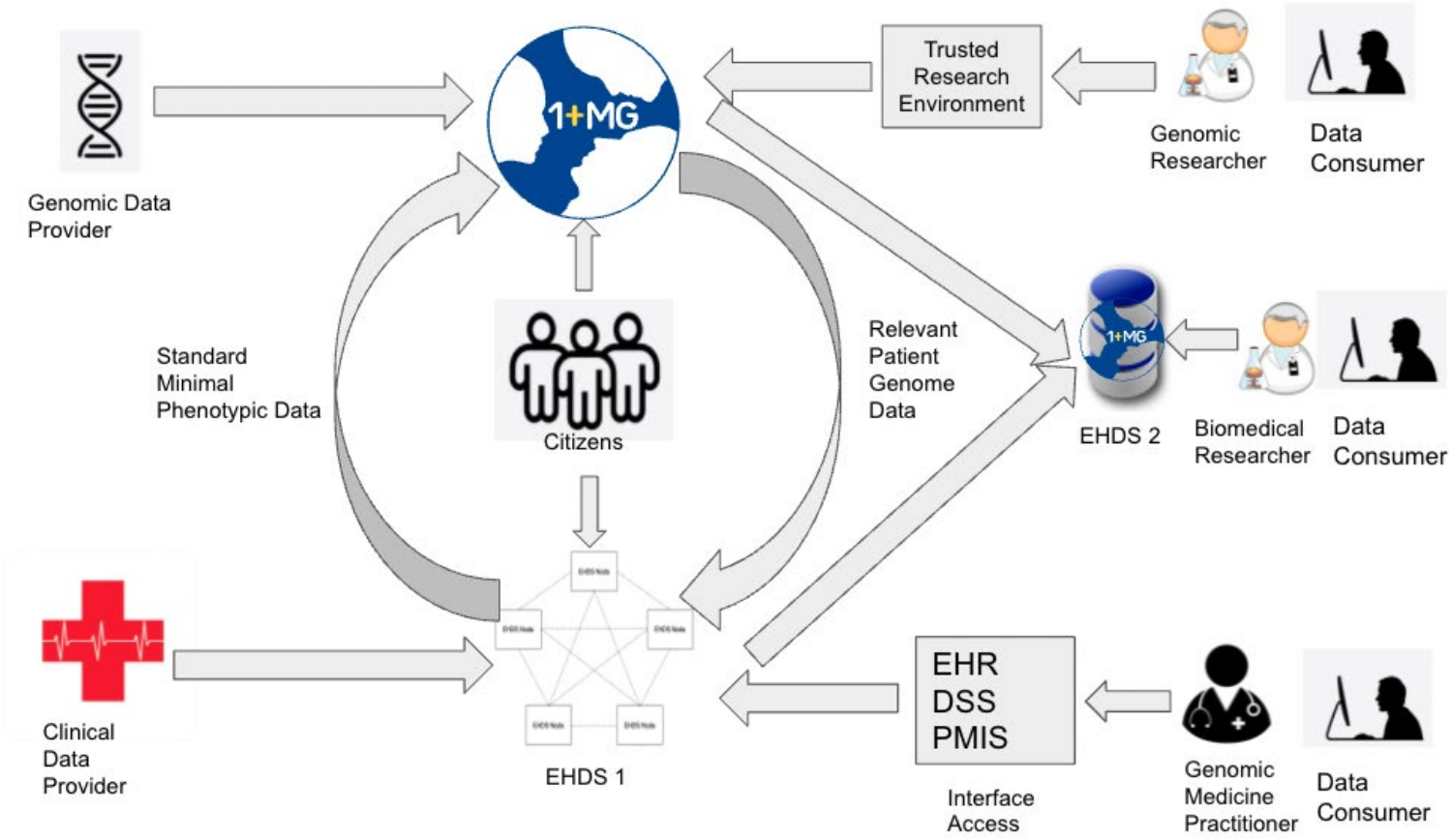

Mendix cloud
Frankfurt



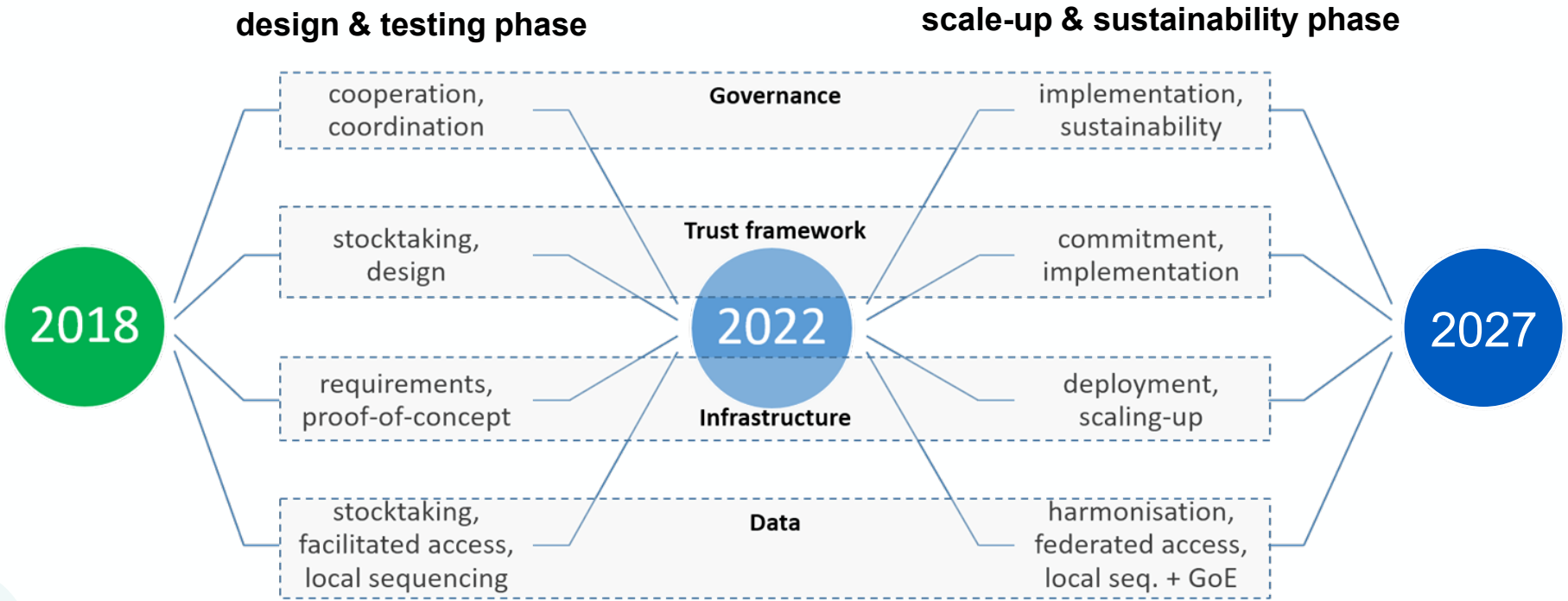



# Data landscape



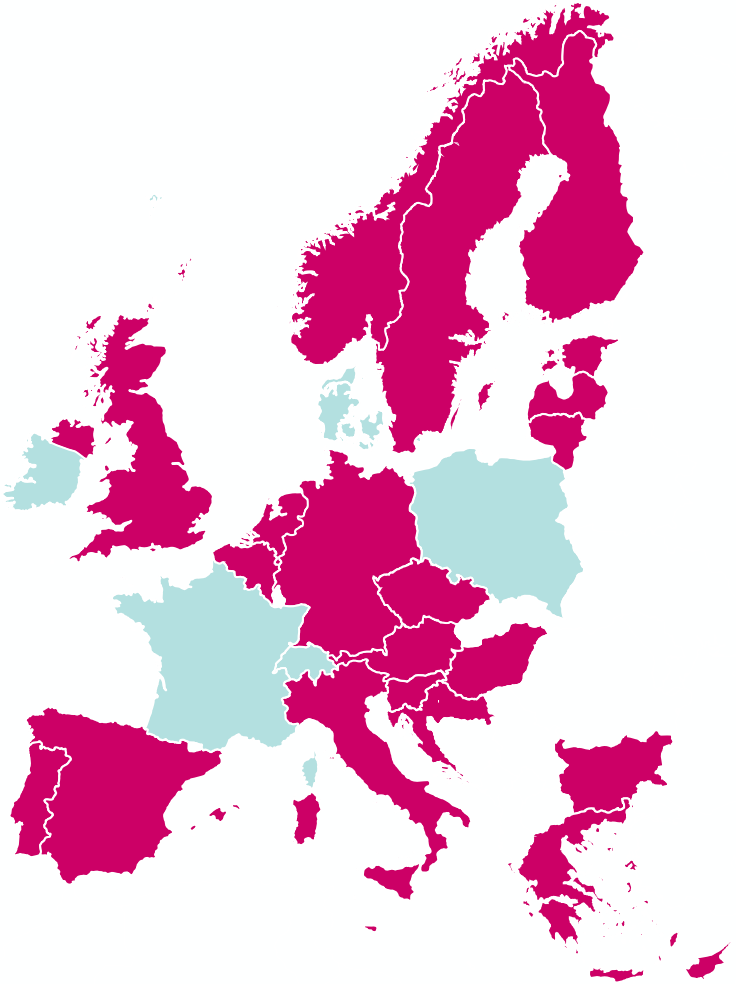
# 1+MG Roadmap 2018-2027



# Learning network of 1+MG National Mirror Groups

- Ministries of Health and Science
- National Stakeholders
- Guiding national implementation along 1+MG recommendations

1+MG Working groups and National Mirror Groups

# 1+MG National Mirror Group-NL?

- Most participating countries have a NMG with link to 1+MG WGs
- NL experts (MG-NL) have been closely involved since 2018 in developing 1+MG;
  - Assembled input from the major initiatives and expert communities
  - Health-RI coordination (health data infrastructure); ZonMw support
- MG-NL team established, with experts across domains
  - Representation from health research, health care and societal stakeholders in NL

ZonMw

health RI
enabling data driven health

# NL participation in 1+MG

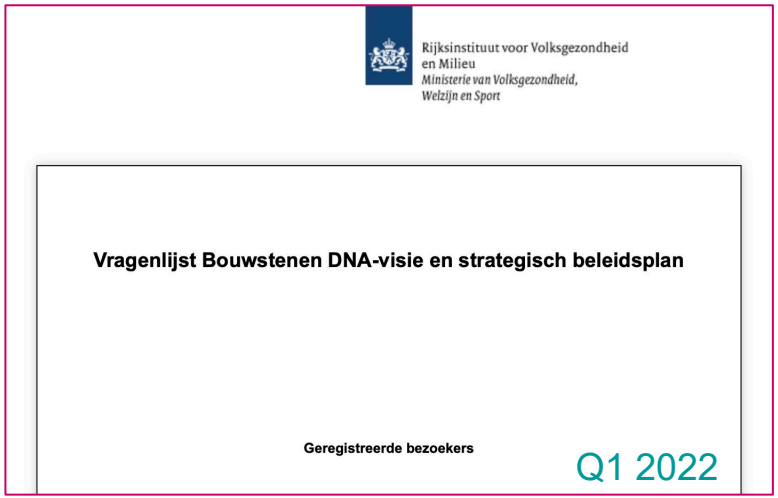
| Working group | Representative |
|---|---|
| 1. Scope, stakeholders and governance | Ruben Kok (DTL/Health-RI), co-lead |
| 2. Ethical, Legal, and Societal Issues (ELSI) | Jasper Bovenberg (BBMRI-NL), Marjanka Schmidt (NKI), Susanne Rebers (NKI) |
| 3. Common standards for capturing clinical and phenotypic data requirements incl. FAIR implementatie | Jeroen Beliën (AmsterdamUMC), co-lead, Gijs Santen (LUMC) |
| 4. Good sequencing practice / development of standards for clinical interpretation | Edwin Cuppen (UMCU/Hartwig) |
| 5. Infrastructure: Interoperability, transfer between countries, local/federated system incl. systems development and deployment and data access governance | Morris Swertz (UMCG), Peter-Bram 't Hoen (Radboudumc), Rob Hooft (ELIXIR-NL, Health-RI) |
| 6. Health economics and outcome research | Ilse Custers (Lygature/Health-RI), co-lead, , Geert Frederiks (UMCU), Valesca Retel (NKI), Heleen Vellekoop (iMTA/HEcoPerMed) |
| 7. Involvement of the private sector (incentives, IP, contribution and access) | Jasper Bovenberg (BBMRI), co-lead |
| 8. Rare Diseases | Lisenka Vissers (Radboudumc), Gijs Santen (LUMC), |
| 9. Cancer | Edwin Cuppen (UMCU, Hartwig) |
| 10. Common and complex diseases | Andre Uitterlinden (Erasmus MC), Tessel Rigter (RIVM), Marianne Beekman (LUMC) |
| 11. Infectious diseases (host genomics aspects) | Nynke Rots (RIVM) |
| enome of Europe | André Uitterlinden (Erasmus MC), Co-Lead |

# (Attempts toward) funding NL DNA initiative

- Dec 2020: VWS requested proposal by MG-NL team:
  - Build national genome programme
    - ✓ Incl. national mirror group in 1+MG
  - Remove barriers for cross-border access to data
    - ✓ Secondary use in research & clinic
  - Build representative reference cohort of 50.000 genomes (WGS) of participating NL citizens
    - ✓ 50% from existing population cohorts, 50% from clinical genetics (RD) & oncology
    - ✓ Contribution to 1+MG collection/Genome of Europe

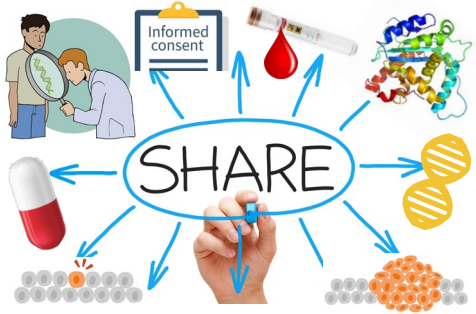
# Formal NMG and ‘DNA programme’ needed

- Prepare national policy to formalise a NMG and contribute to 1+MG through a national programme
  - VWS represented in 1+MG governance
  - Regular alignment with MG-NL team
  - VWS prepared ‘RRF-fiche’ to generate 50k WGS and set up a ‘DNA’ data infrastructure (aligned with Health-RI)
  - RIVM landscape analysis
  - RIVM: building blocks DNA vision
    - ✓ Working groups to be established to prepare for proposals how to address priority issues



# Health-RI partnership will help establish national base infrastructure

# FAIR GENOMES.org




Amsterdam University Medical Centers, location VUmc, NL
Netherlands Cancer Institute, Amsterdam, The Netherlands
Hartwig Medical Foundation, Amsterdam, The Netherlands
Leiden University Medical Center, The Netherlands
Leiden Institute for Advanced Computer Science, Leiden University, Leiden, NL
Erasmus Medical Center, Rotterdam, The Netherlands
Maastricht University Medical Center, The Netherlands


University Medical Center Groningen, The Netherlands
VSOP - Dutch Patient Alliance for Rare and Genetic Diseases
Durrer Center for Cardiovascular Research, Utrecht, The Netherlands
Princess Máxima Center for Pediatric Oncology, Utrecht, The Netherlands
University Medical Center Utrecht, The Netherlands
Dutch Techcentre for Life Sciences, Utrecht, The Netherlands
Radboud University Medical Center, Nijmegen, The Netherlands

**66 people** from **14 Dutch institutes** - F2F meetings, Zoom calls, workshops, interviews


# FAIR GENOMES: community development of guidelines, schema & systems for reuse of NGS data

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## GENOME DATA IS UNFAIR

Next-generation Sequencing (NGS) has revolutionized healthcare and research, but the data is fragmented across providers and institutes. By making NGS data more FAIR (Findable, Accessible, Interoperable, Reusable), we can unlock their full potential for everyone's health benefit.

## FINDING COMMON UNDERSTANDING

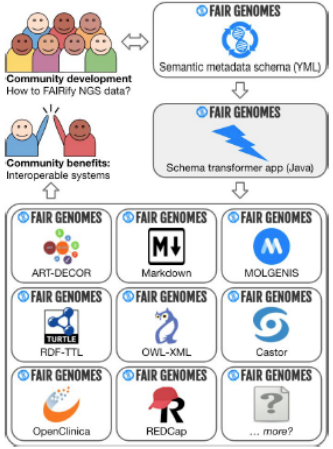
The FAIR Genomes consortium and friends (X-omics, EJP-RD, JRC CDE, 1+MG, SolveRD, GA4GH, MIABIS, ...) are creating a guideline and are defining data elements essential for sharing NGS data in a semantic schema. v0.2 has 107 elements in 9 modules, linked to common ontologies.

## TRY IT YOURSELF AND JOIN US

Open source development:
**https://github.com/fairgenomes**
Public demo available:
**https://fairgenomes-acc.gcc.rug.nl**

## SEAMLESS EDC INTEROPERABILITY

The schema is transformed into all formats for e.g. MOLGENIS. We plan to use iCRF Generator to also support Castor, OpenClinica and REDCap.

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4 = Amsterdam UMC, Vrije Universiteit Amsterdam, Amsterdam, NL
5 = VSOP - Dutch Patient Alliance for Rare and Genetic Diseases
6 = Leiden University Medical Center, The Netherlands
7 = Erasmus Medical Center, Rotterdam, The Netherlands
8 = Durrer Center for Cardiovascular Research, Utrecht, The Netherlands
9 = Maastricht University Medical Center, The Netherlands
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11= Dutch Techcentre for Life Sciences, Utrecht, The Netherlands
12 = Netherlands Cancer Institute, Amsterdam, The Netherlands
13 = Hartwig Medical Foundation, Amsterdam, The Netherlands
14 = Leiden Institute for Advanced Computer Science, Leiden University, Leiden, NL

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ZonMw

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