

Building genomics

genomDE :
National and
European initiatives

30th November 2020

DIGITAL EVENT

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30TH NOVEMBER 2020



Supported by the European Union's Structural Reform Support Programme (SRSP) and implemented in cooperation with the European Commission's Directorate-General for Structural Reform Support (DG REFORM)



Genomic Strategies in European Countries

Finland

Aarno Palotie, M.D., Ph.D.
University of Helsinki (Institute for Molecular
Medicine, FIMM)
The Broad Institute of MIT and Harvard

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hello

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FINLAND IN FACTS

5.4 million



1.4 million
inhabitants in
metropolitan
area

Life expectancy:
year



Official languages
are **FINNISH**...
(spoken by 88.9%)

...and **SWEDISH**
(spoken by 5.3%)

SÁMI is the mother
tongue of about 1,900 people.



Christianity:
73.8% **LUTHERAN**
and about
1.1% **ORTHODOX**



GDP per capita (2014)

37,559 €



REPUBLIC, parliamentary democracy,
200 members in one chamber

Independence Day
DECEMBER 6

1917



1995



1955

Why Finland?



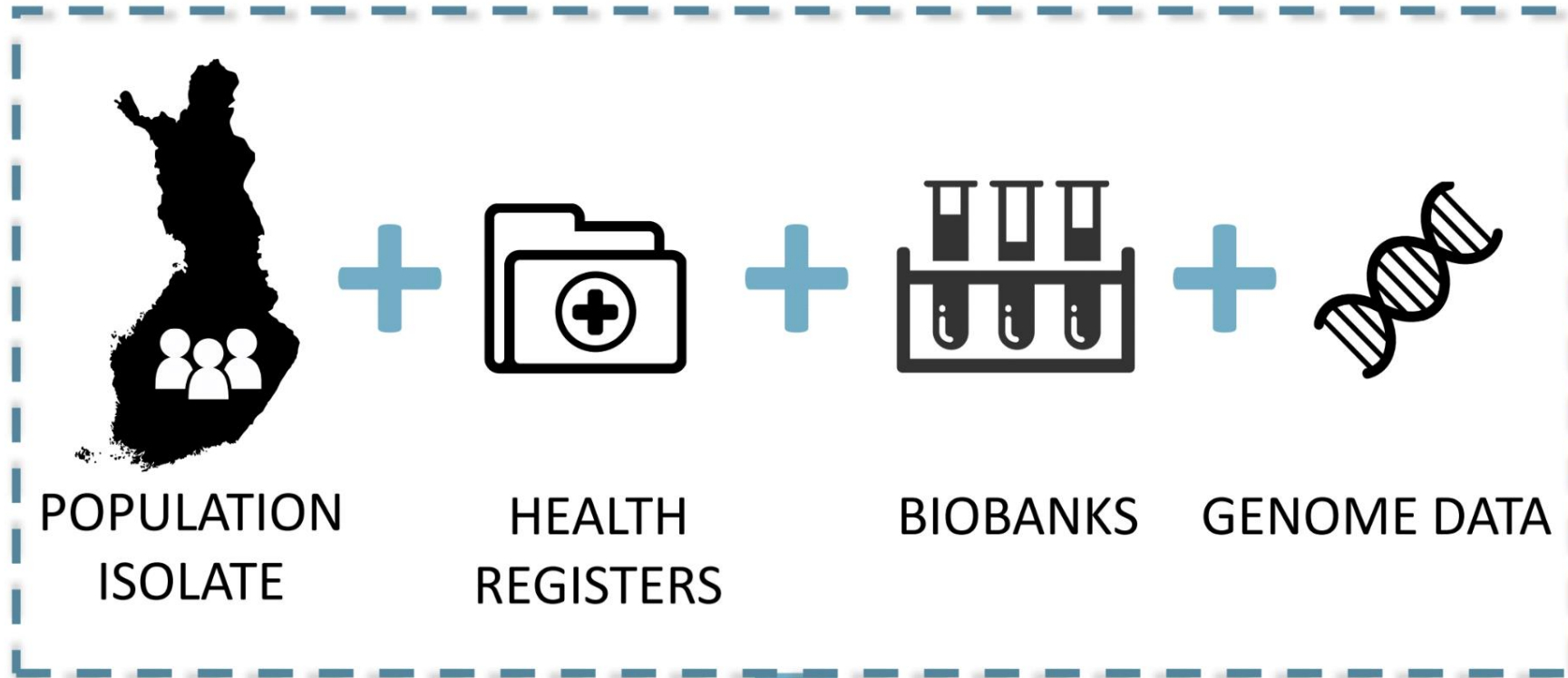
- ✓ Countries with Biobanks
- ✓ Universal Healthcare
- ✓ Unique personal identity code
- ✓ Isolated population
- ✓ Recalling made easy
(biobank act + social security number)

From Discovery to treatment and prevention



10 – 20 years





POPULATION
ISOLATE

HEALTH
REGISTERS

BIOBANKS

GENOME DATA



Innovative Study Designs

National registers



Hospital discharge
Hospital procedure
Outpatient visit
Outpatient procedure

151167-123J

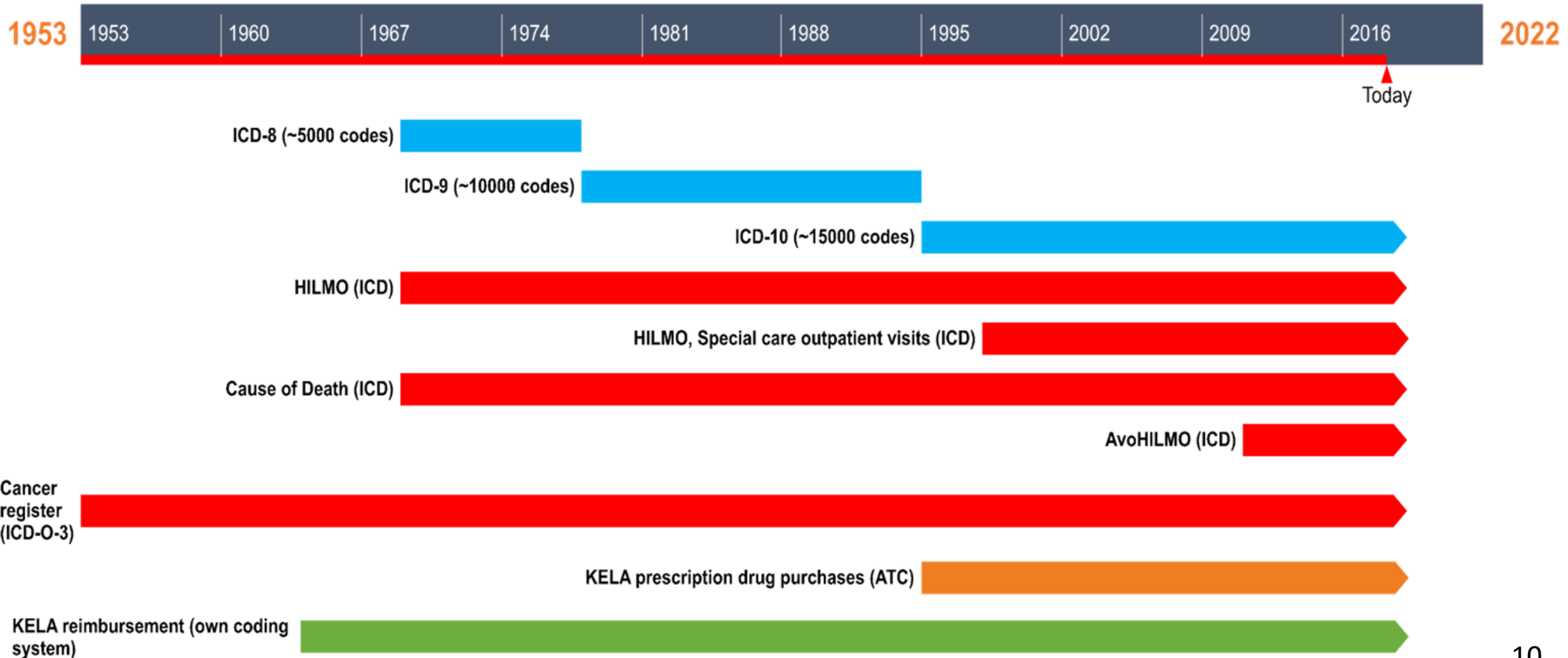
Cause of death
Drug purchase
Drug reimbursement

Register data
for
administration

Register data
for
administration

Register data
for
administration

Nationwide registries



Data from every healthcare visit over a lifetime, nationwide





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GOVERNMENT BACKING



**Ministry of Health
and Welfare**



**Ministry of Education
and Culture**



**Ministry of Employment
and Economy**



HEALTH SECTOR GROWTH STRATEGY

Government backing

- National Genome Strategy
- The Biobank act
- Secondary usage of register data
- National Genome Center

Health Sector Growth Strategy for Research and Innovation Activities

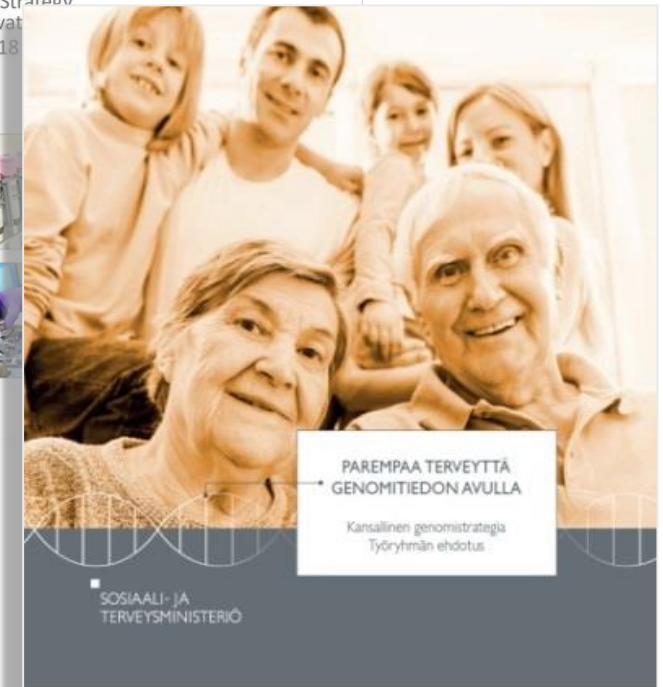
MEE reports 16/2014



Health sector
GROWTH STRATEGY

Innovating together

Health Sector Growth Strategy
for Research and Innovation
Roadmap for 2016–2018



PAREMPAA TERVEYTTÄ
GENOMITIEDON AVULLA

Kansallinen genomistrategia
Työryhmän ehdotus

SOSIAALI- JA
TERVEYSMINISTERIÖ

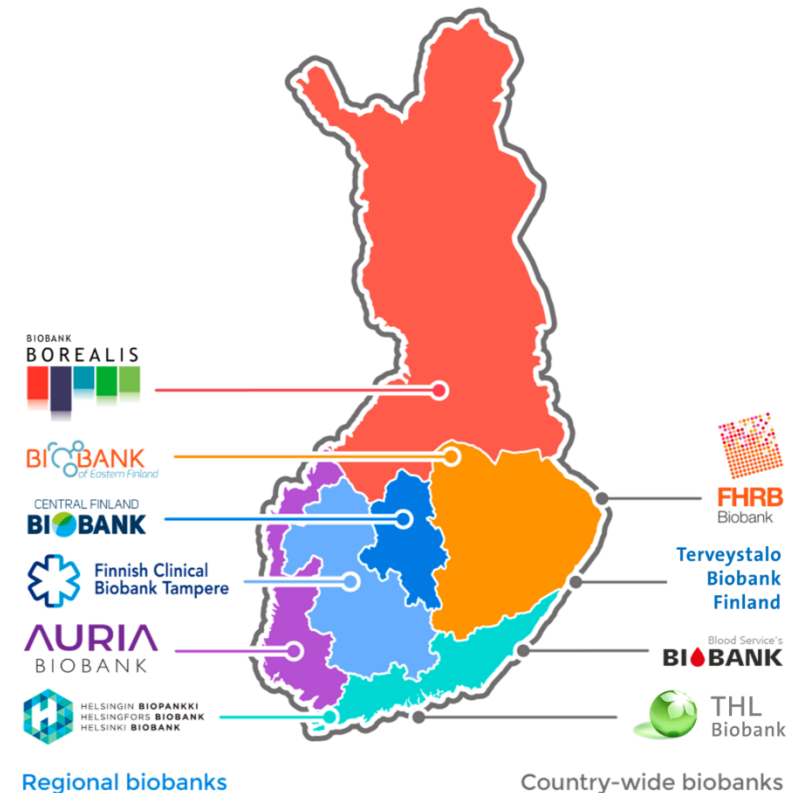
**GENOME
CANCER
NEURO
DRUG
DEVELOPMENT
BIOBANK** | **C
E
N
T
R
E**



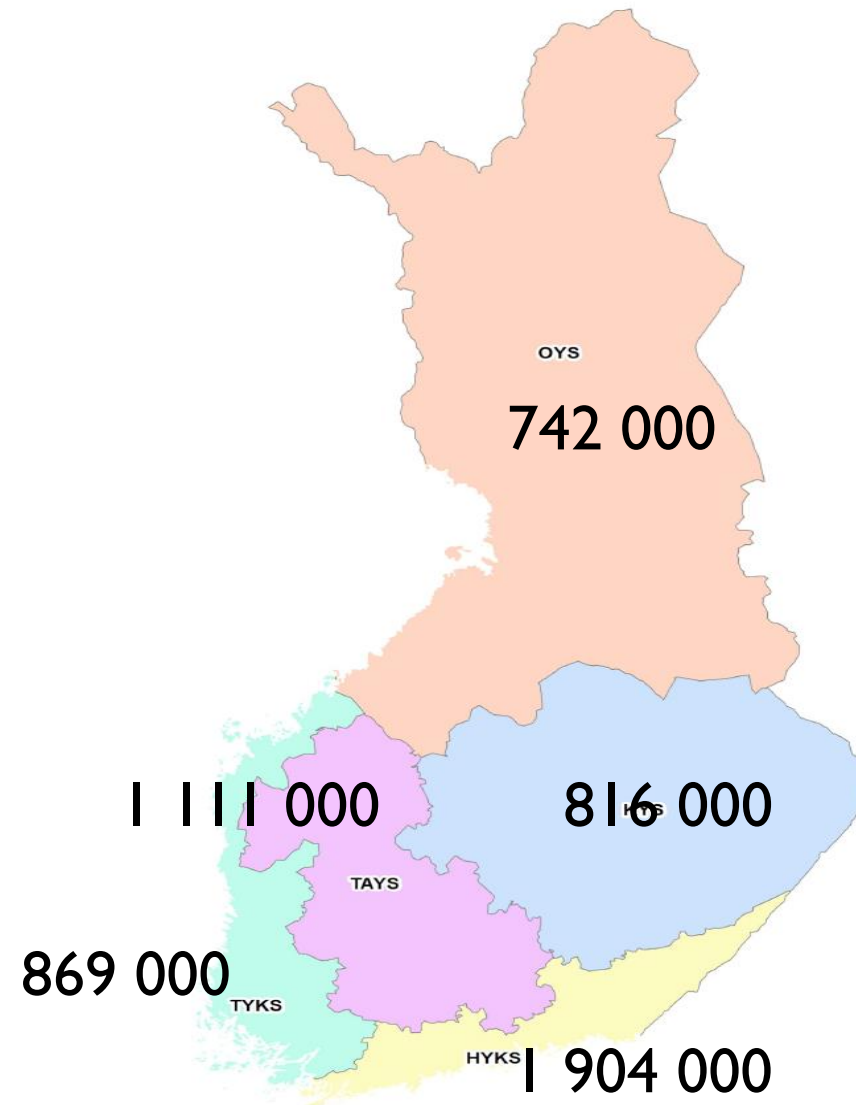
The Finnish Biobank Act is Unique

- Registration of biobanks, wide consent and protection of participants
- Transfer of existing sample and data collections to biobanks
- Possibility to recontact
- Possibility to collect samples and data from the health care
- Covers the whole country
- **Collaboration with industry**

10 Biobanks



Five University Hospital Districts



Finnish Biobanks – FINBB



FOUNDING UNIVERSITIES



FOUNDING HOSPITALS



FINNISH HOSPITAL DISTRICTS



BIOBANKS



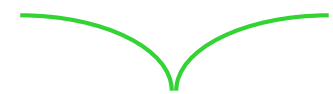
STRATEGIC COLLABORATION



+ Patient, data and sample finding

+ Study set-up & single point of contract

+ Data and sample delivery & analytics



**ACADEMIC
and
INDUSTRY
RESEARCHERS**



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ACCESS TO DIGITAL HEALTH DATA

Photo (c) Jussi Hellsten/VisitHelsinki

Portal to "all" health care information

Kanta

User groups

Healthcare service providers

Primary care organizations
Hospital districts
Private healthcare providers

Providers of social services

Pharmacies

Healthcare professionals

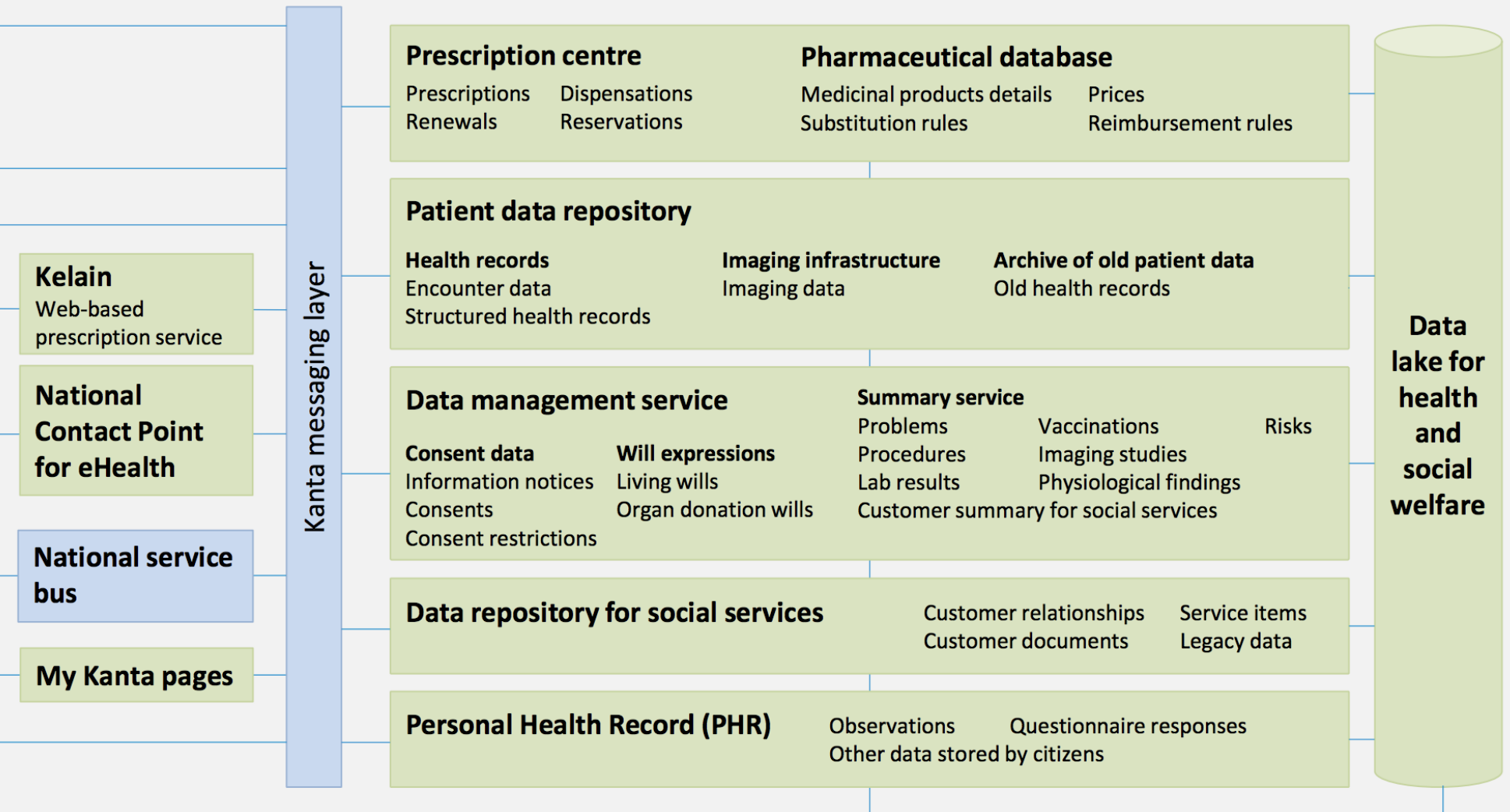
eHealth Digital Service Infrastructure

Receivers of medical certificates

Citizens

and apps used by them

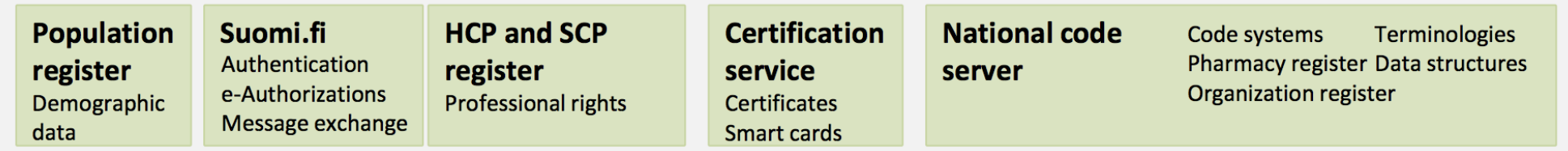
Kanta services



Main standards

HL7 V3: CDA R2 L3 and Medical Records
HL7 FHIR (Kanta Personal Health Record)
JSON, XHTML (PHR and social services)
PDF/A (old patient data and social services)
DICOM IT-I Profiles (imaging and eHDSI)
W3C XML DSig
WS Addressing, WS-I
SAML, X.509

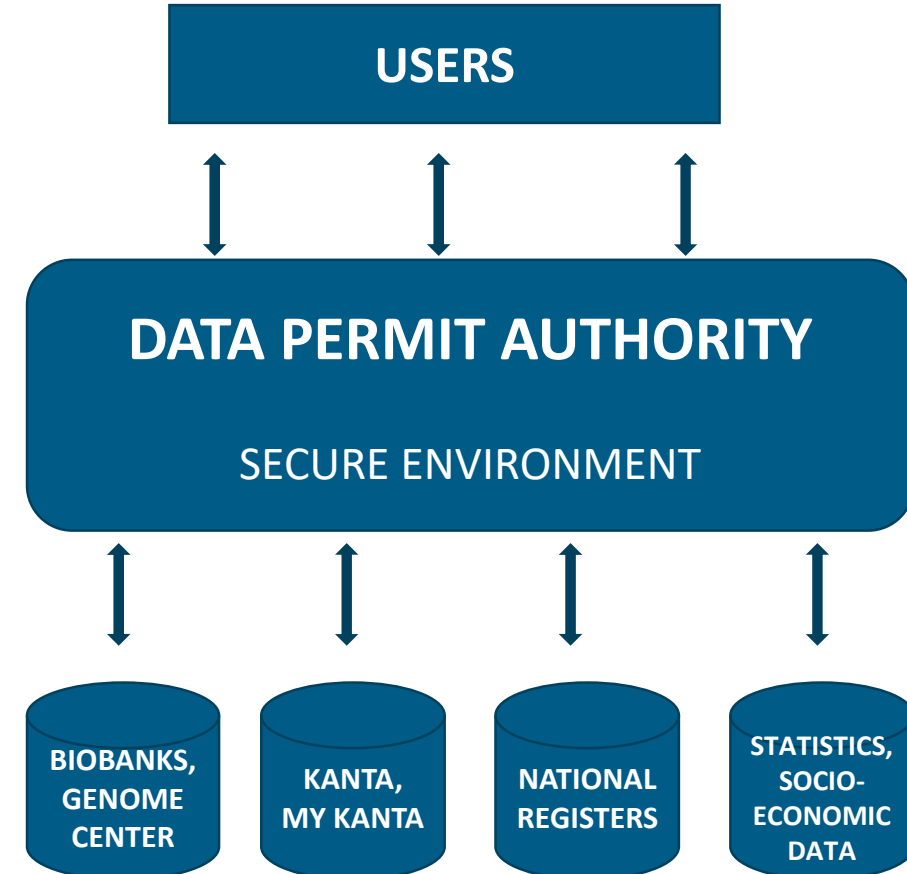
Other national services



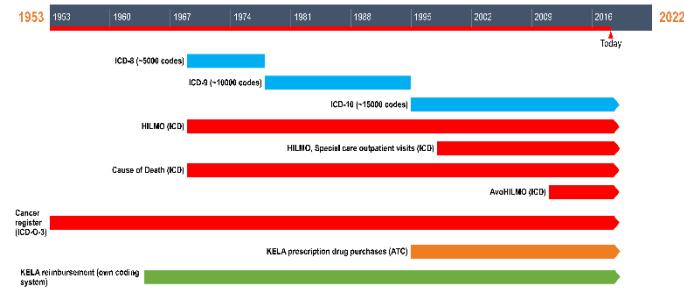
Secondary use of data legislation

Act on Secondary use of Health and Social Data

- To ensure authorities, institutes and companies access to health and social data in Finland
- To provide efficient and secure procedures to utilize the data in research, development and innovation activities, education and knowledge management duties



Data Permit Authority



FINDATA



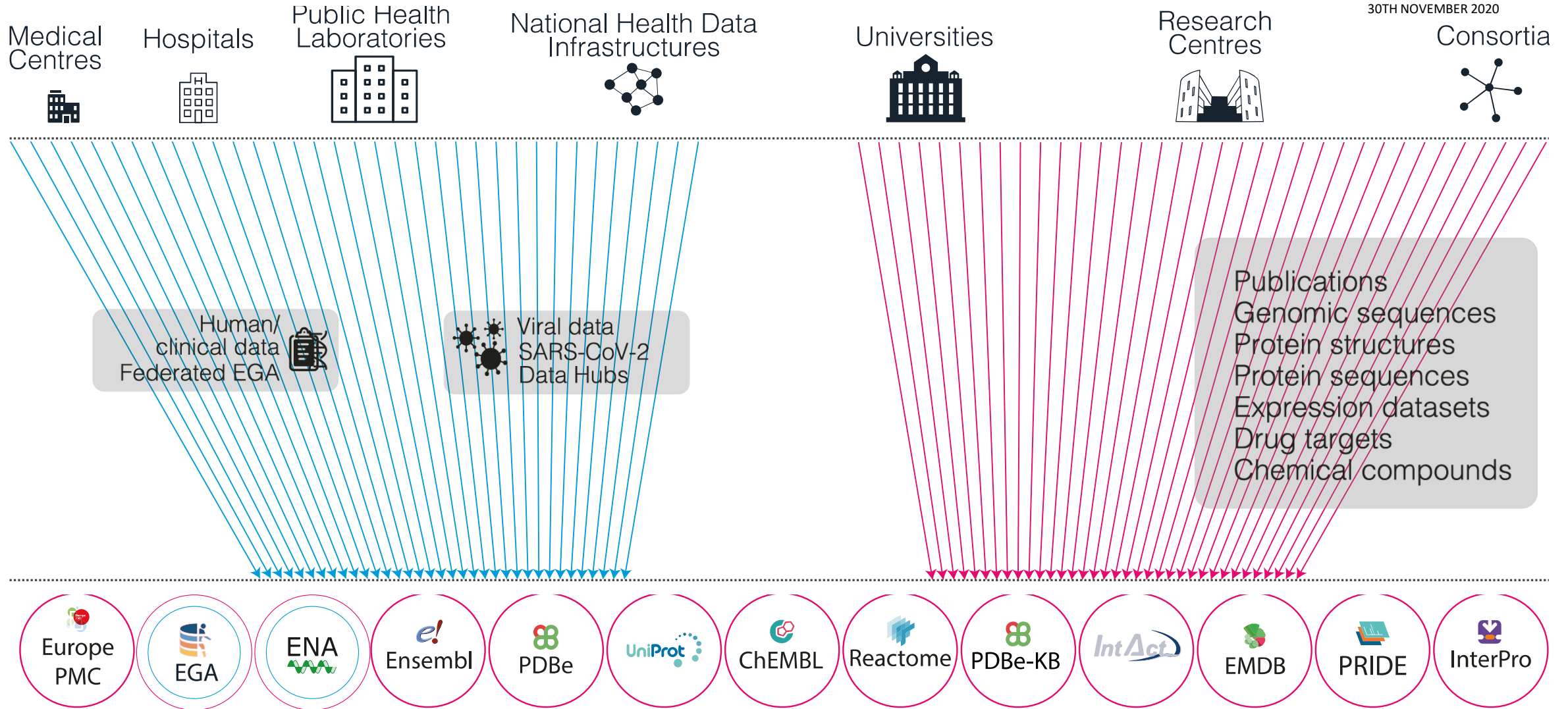


Sensitive health and bio data & HPC in Finland



Building genomics

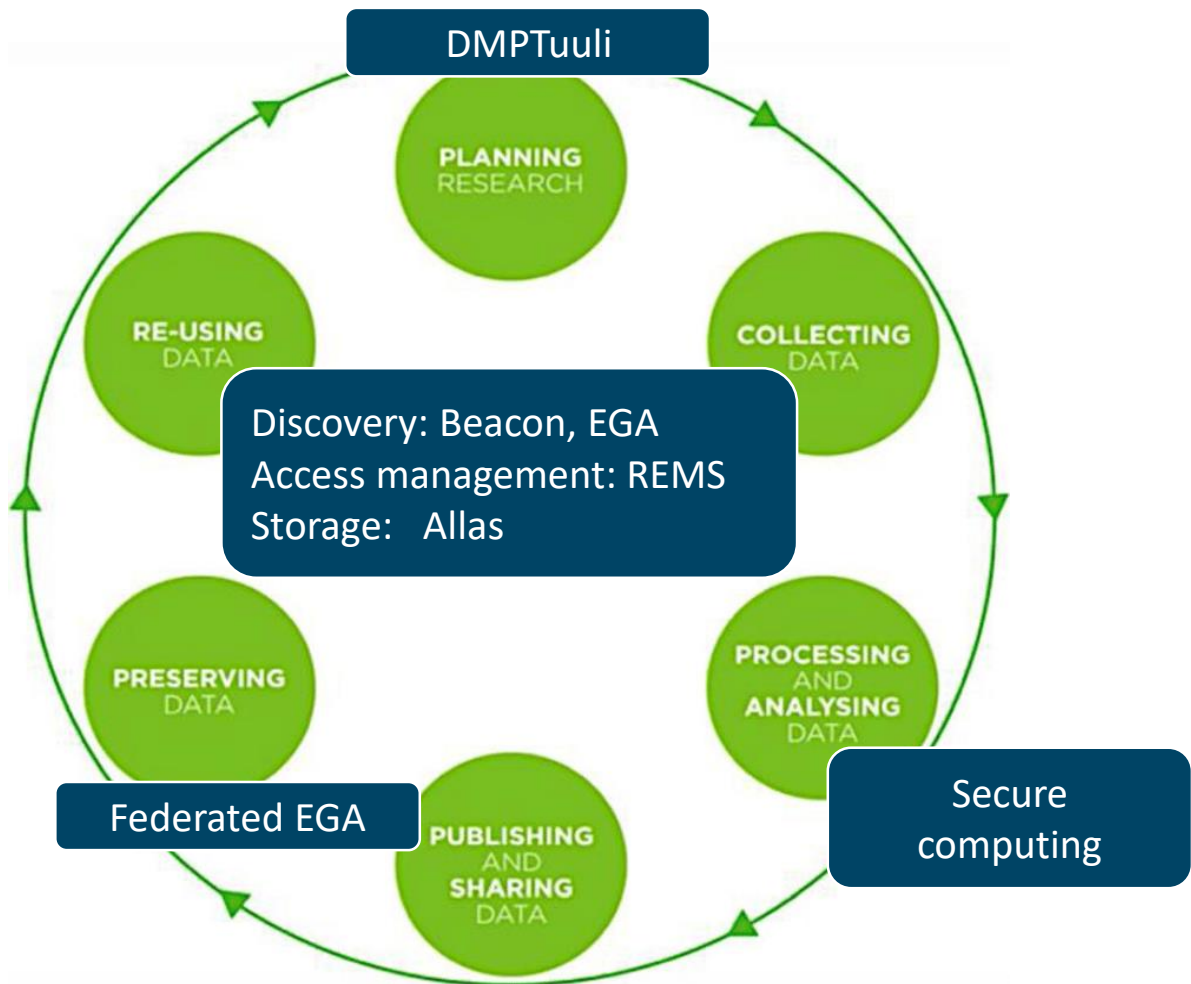
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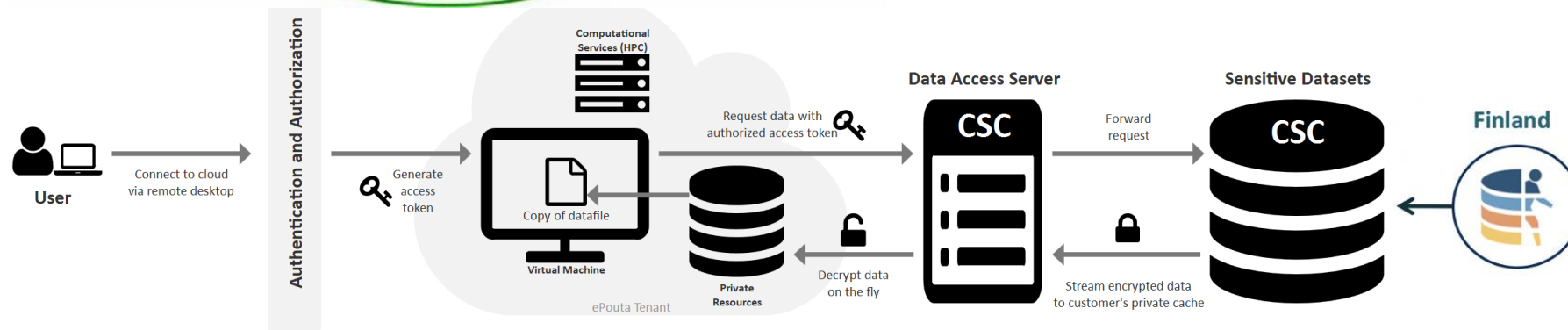
➔ Access-controlled data ●

1.980 CSC computing projects by PIs in health & bio area
One of the largest consumer of CSC billing units (30%)

● Open research data

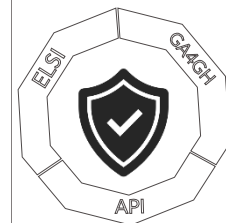
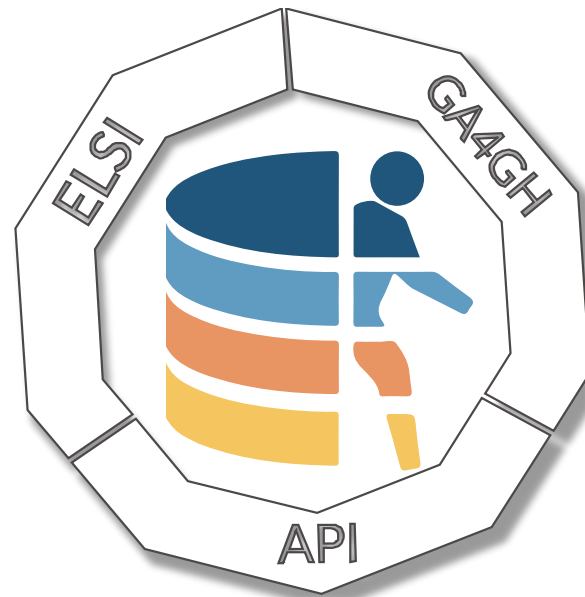


- DMPTuuli : data management planning tool
- Beacon, EGA : openly searchable indexes
- REMS : secondary use access management tool
- Allas : encrypted, local cloud storage
- secure HPC, FEGA : secure desktop to HPC
- FEGA : national storage, international discoverability



FEGA

Federated European Genome Archive



Consistent
Framework

Implementing the GA4GH
suite of products

Compliant with Ethical,
Legal and Social
Implications (ELSI)

Common API interfaces



Ministry of Health
and Welfare



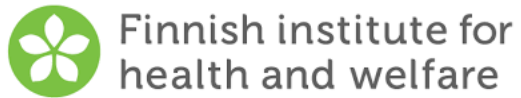
Ministry of Education
and Culture



Ministry of Employment
and Economy

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Hospitals

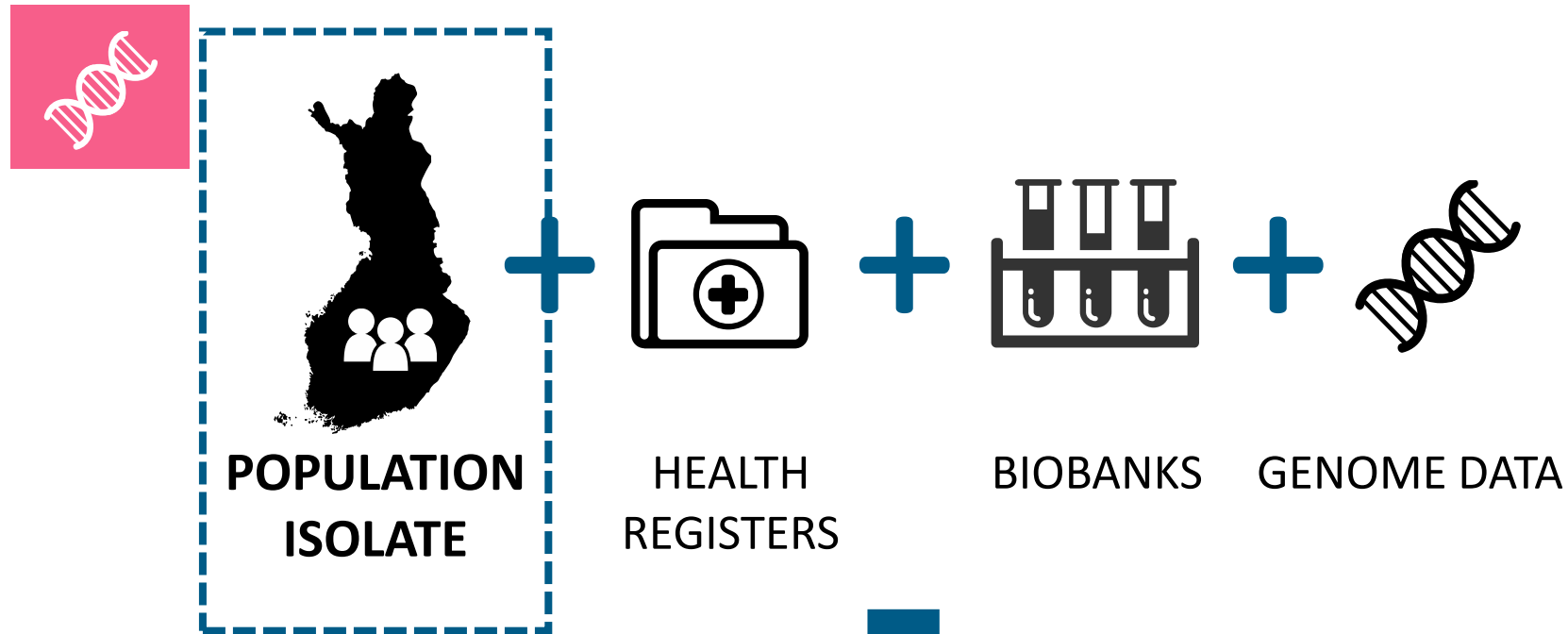


Universities



Business Finland





Innovative Study Designs

Finland, a population isolate

EARLY SETTLEMENT

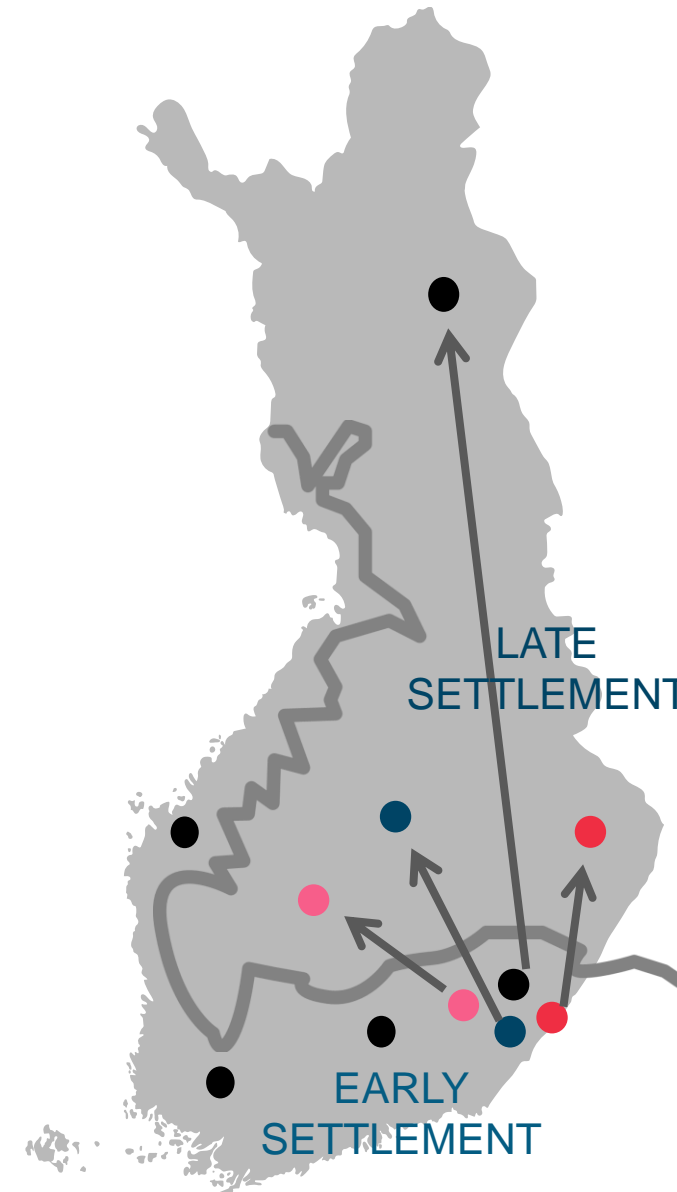
- 2000-10 000 years ago
- South and Coast

LATE SETTLEMENT

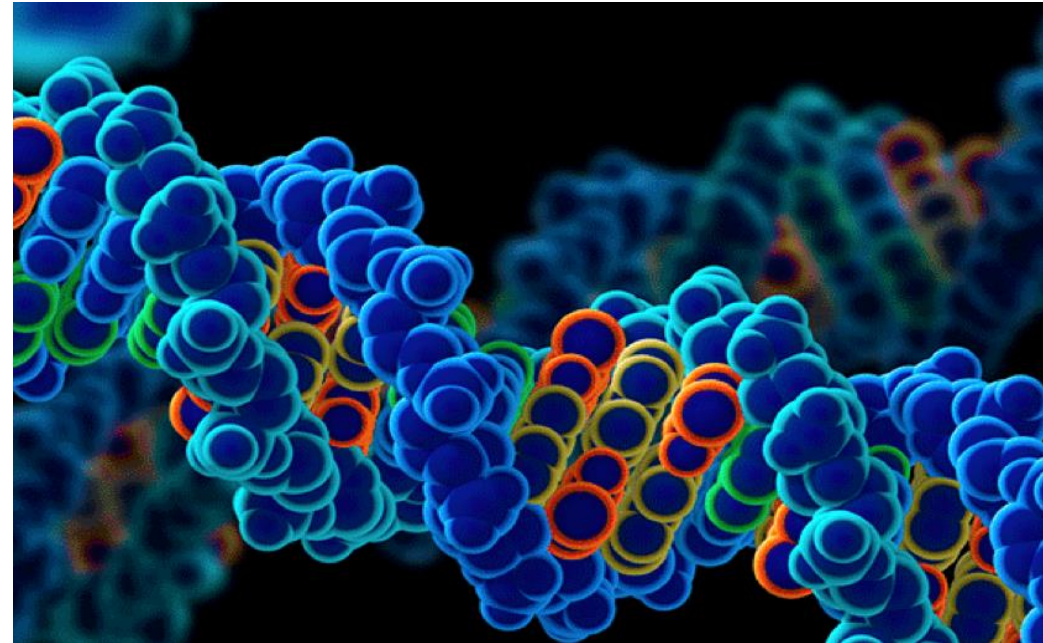
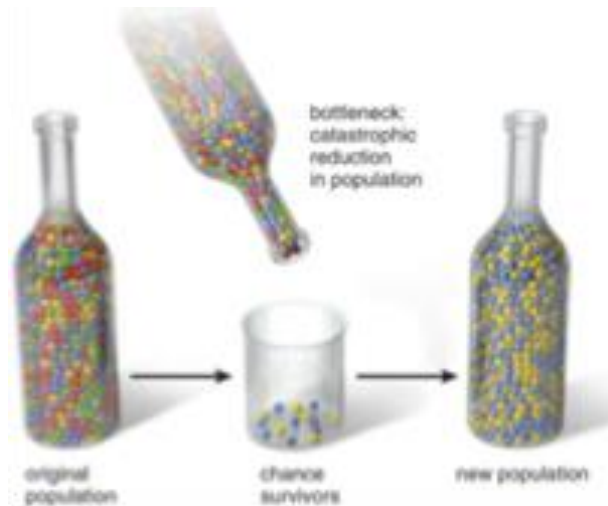
- 16th century
- multiple bottle necks

EXPANSION

- 18th century – *population 250 000*
- Today – ***population 5.5 million***



Power of a Genetic Isolate



Specific damaging genetic variants become enriched and easy to discover

Reconstructing genomes of Finns ('imputation') from inexpensive genotype data is much more accurate than in the rest of the world

Genetic isolate

Consequences for:

- Medical and genetic research
- Genetic diagnostics

Genetic isolate

Consequences for:

- Medical and genetic research
 - Easier to identify disease associated genetic variants
- Genetic diagnostics
 - Unique spectrum of diseases and genetic variants to test

FinnGen



500,000 individuals
10% of the population

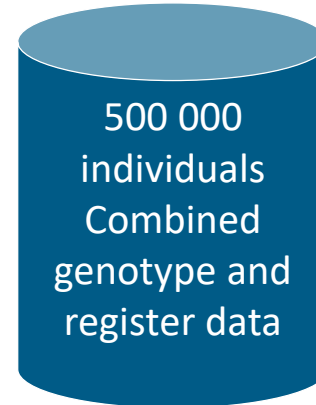
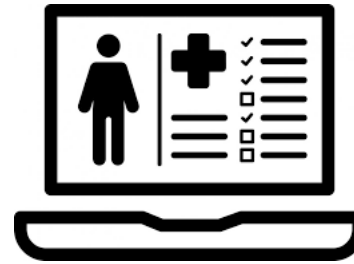
Existing sample collections:
200,000 Finns

300,000 new sample donors
by the end of FinnGen2

FinnGen

National Health Register data

500 000 individuals
~10 % of the population

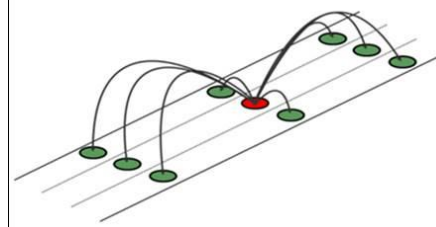


Association analyses

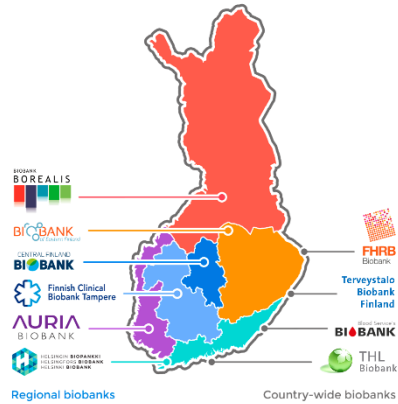


Axiom GWA array

Imputation



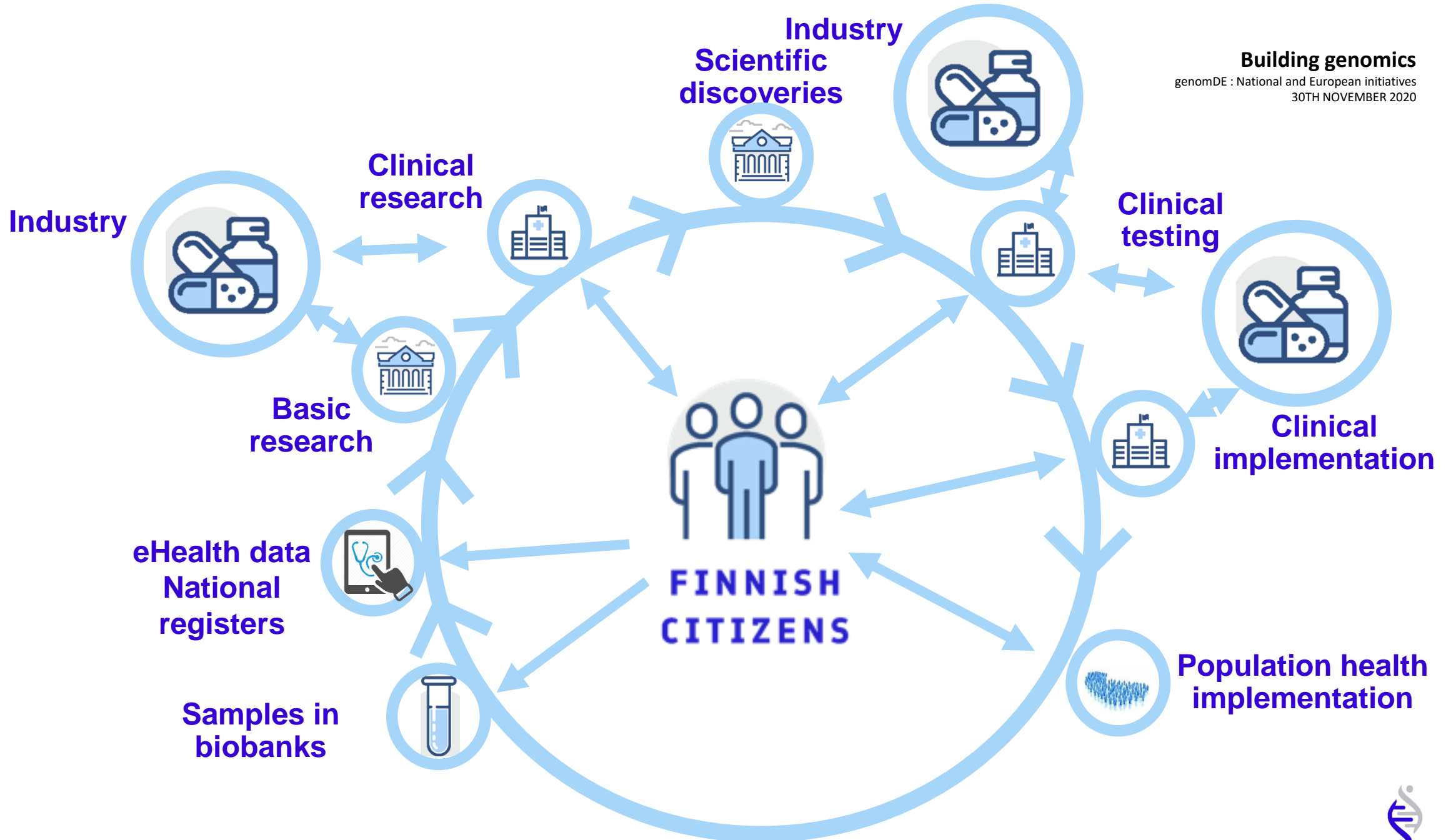
Public-private research project



Academic institutions
and public health care

Pharmaceutical
industry



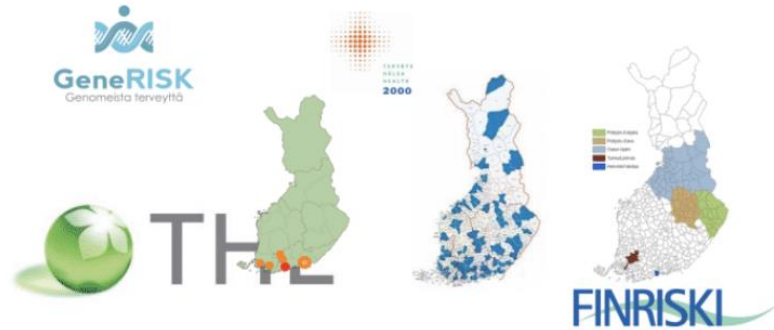
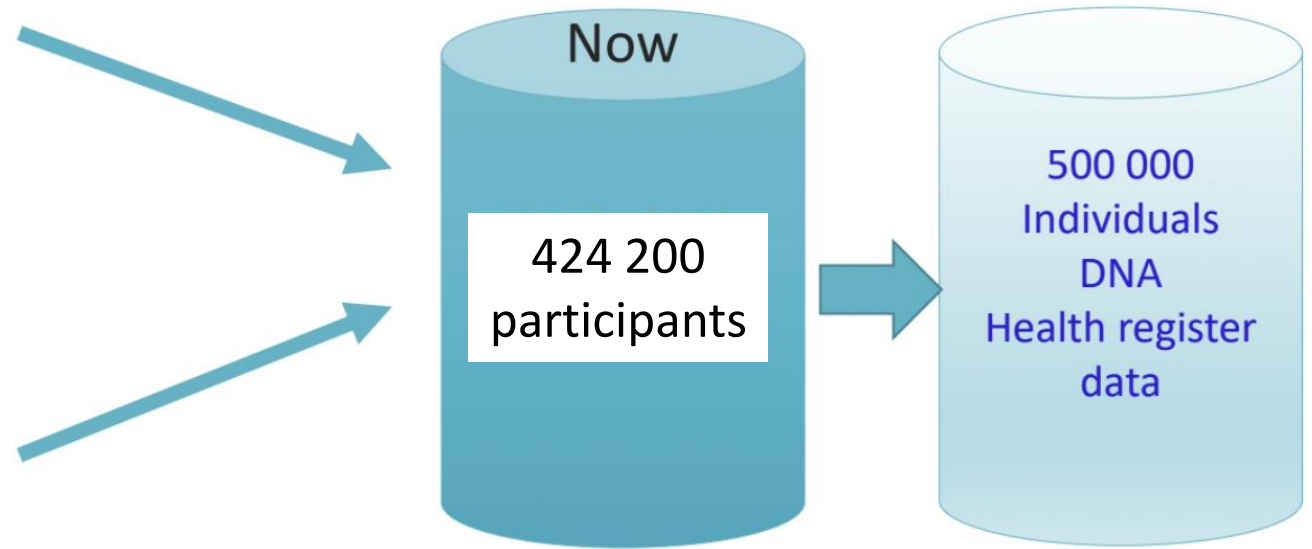


Prospective collections 300 000



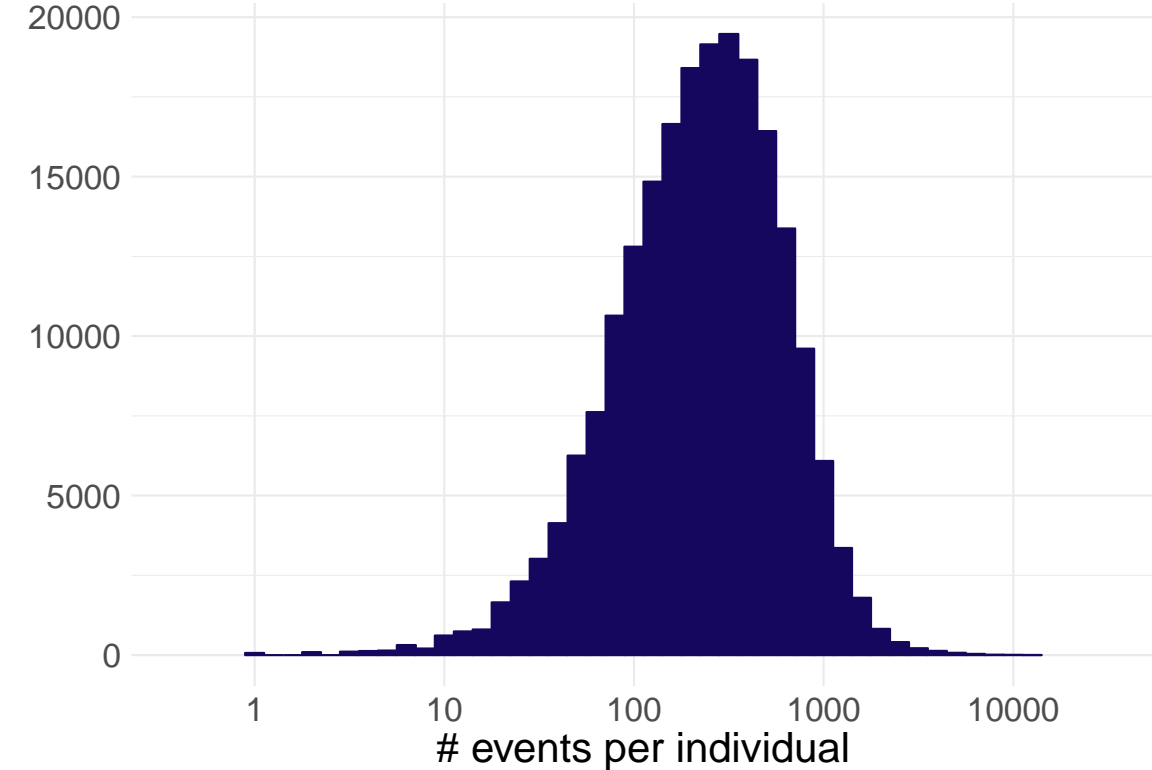
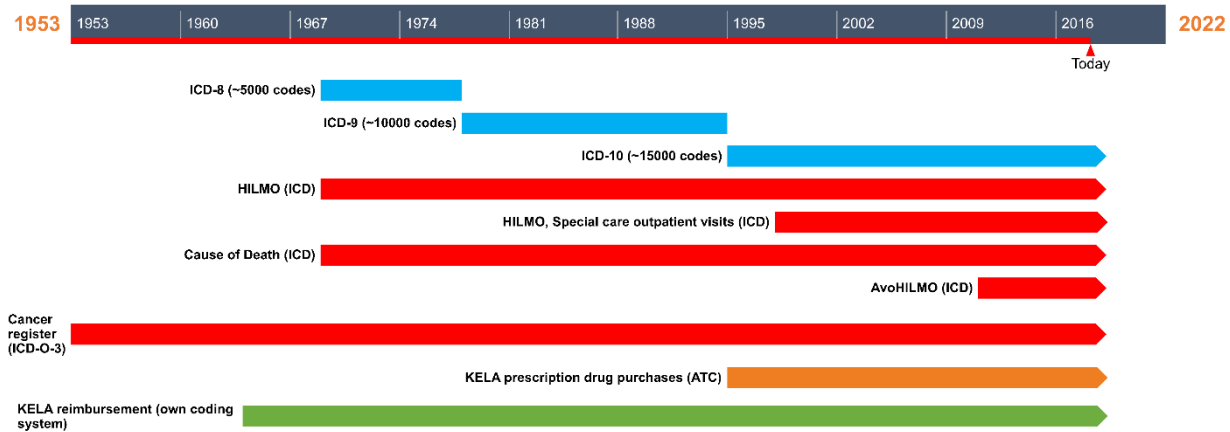
Hospital biobanks

Legacy collections 200 000



424 200 participants, so far
265 700 new samples
collected

Expanded Phenotype Potential



Mean 340 health events per individual
 (including 186 drug purchases)

Longitudinal analyses



Data protection



01

PARTICIPATION IS VOLUNTARY AND THE CONSENT CAN BE WITHDRAWN

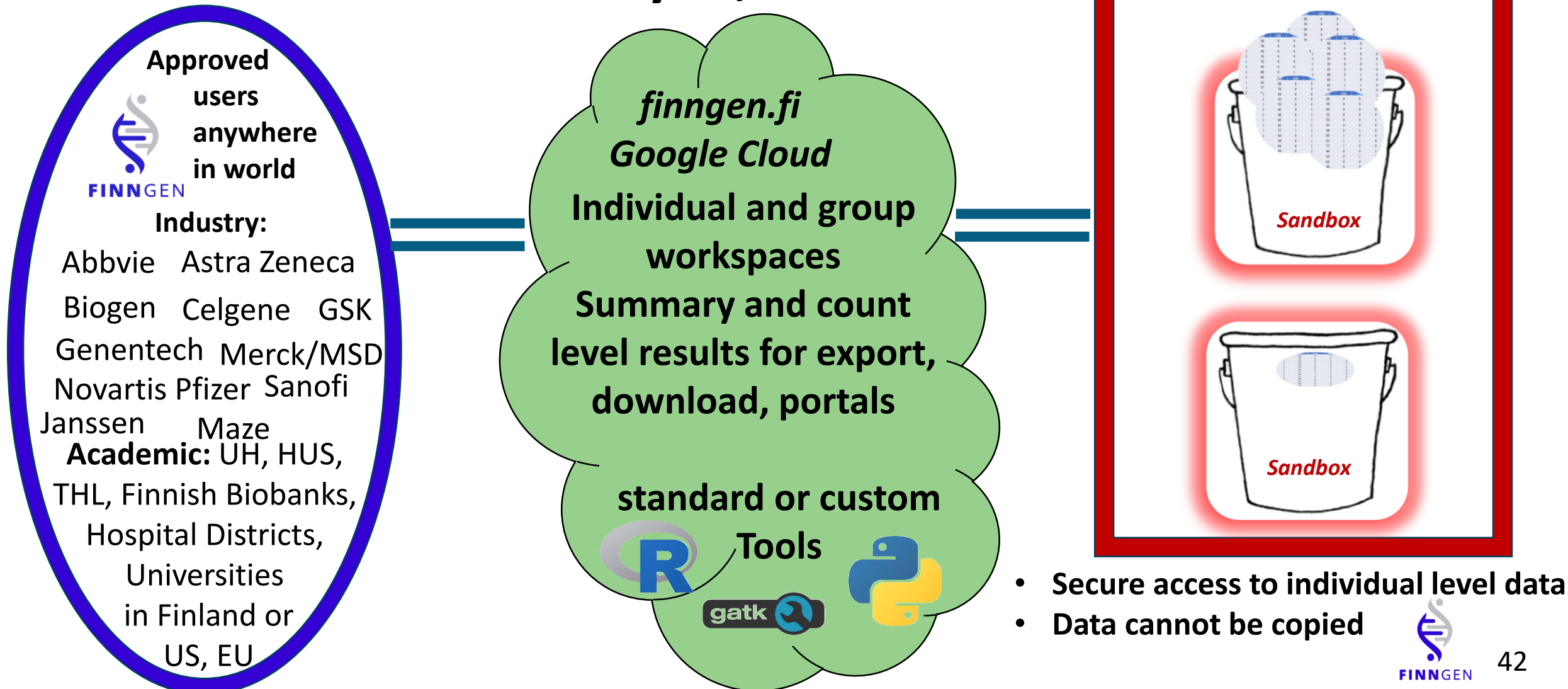
02

SAMPLES ARE CODED AND FINNGEN CAN NOT IDENTIFY PARTICIPANTS

03

GENOME DATA, ANALYZED AND PRODUCED FROM BIOBANK SAMPLES, IS OWNED BY FINNISH BIOBANKS

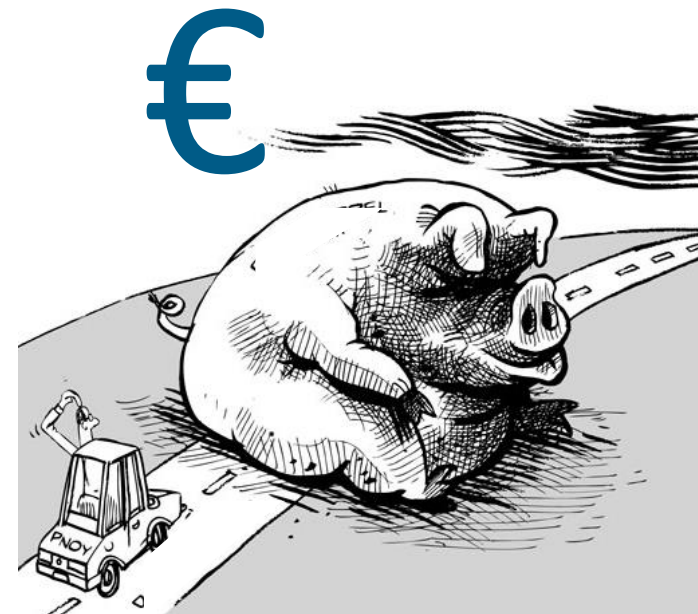
Sandbox for Data Analysis, version 2



- Secure access to individual level data
- Data cannot be copied

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Success with from analyzing first 269,000 participants

Thank you



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