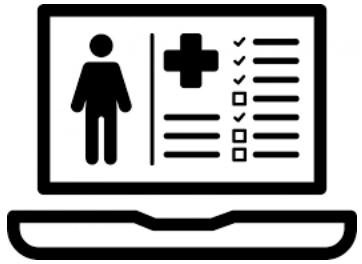
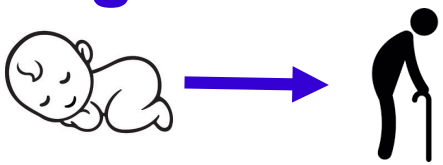
FinnGen

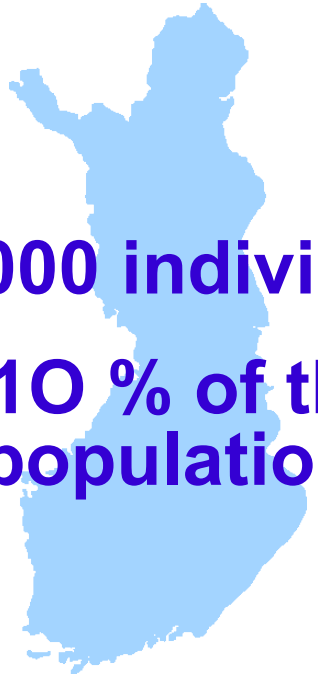


National Health Register data



FINNGEN

1



500 000 individuals

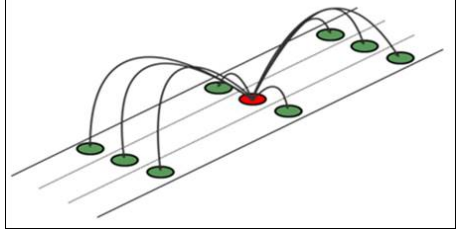
~10 % of the population

500 000 individuals Combined genotype and register data

Association analyses

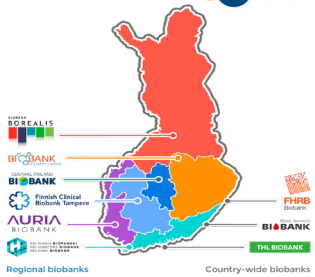
Axiom GWA array

Imputation



BUSINESS FINLAND

BBMRI.fi



Regional biobanks

Country-wide biobanks

gsk GlaxoSmithKline

SANOFI

abbvie

AstraZeneca

Biogen

Celgene

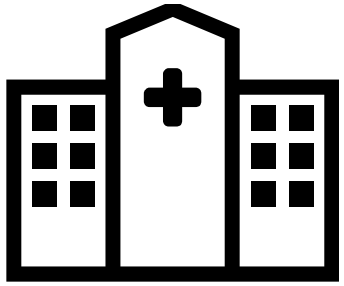
MSD

Pfizer

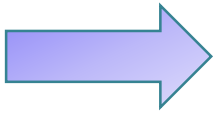
Genentech

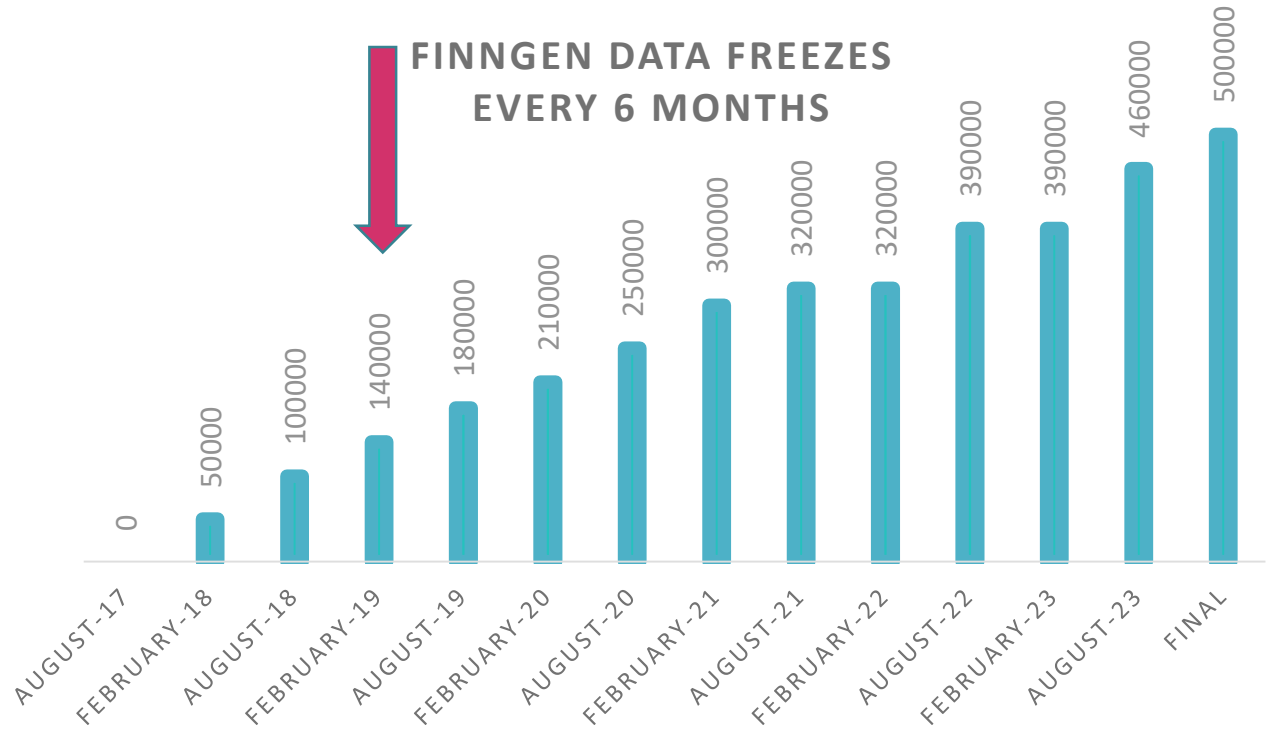
A Member of the Roche Group

FINNGEN

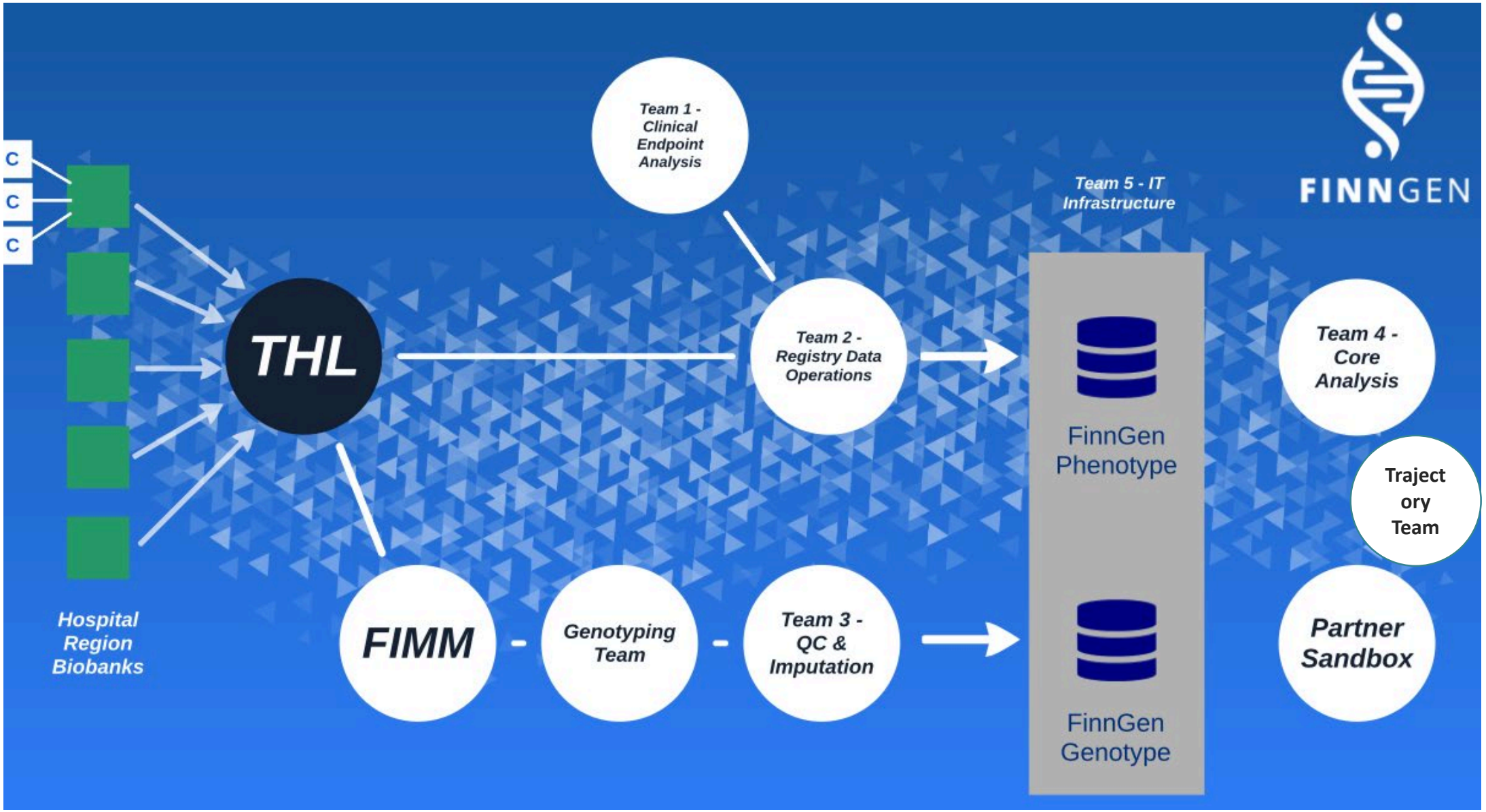


THL





- March 1 data freeze 146 000 individuals


Team 1 - Clinical Endpoint Analysis
Team 5 - IT Infrastructure
FINNGEN
C
C
C
THL
Team 2 - Registry Data Operations
FinnGen Phenotype
Team 4 - Core Analysis
Traject ory Team
Hospital Region Biobanks
FIMM
Genotyping Team
Team 3 - QC & Imputation
Partner Sandbox
FinnGen Genotype

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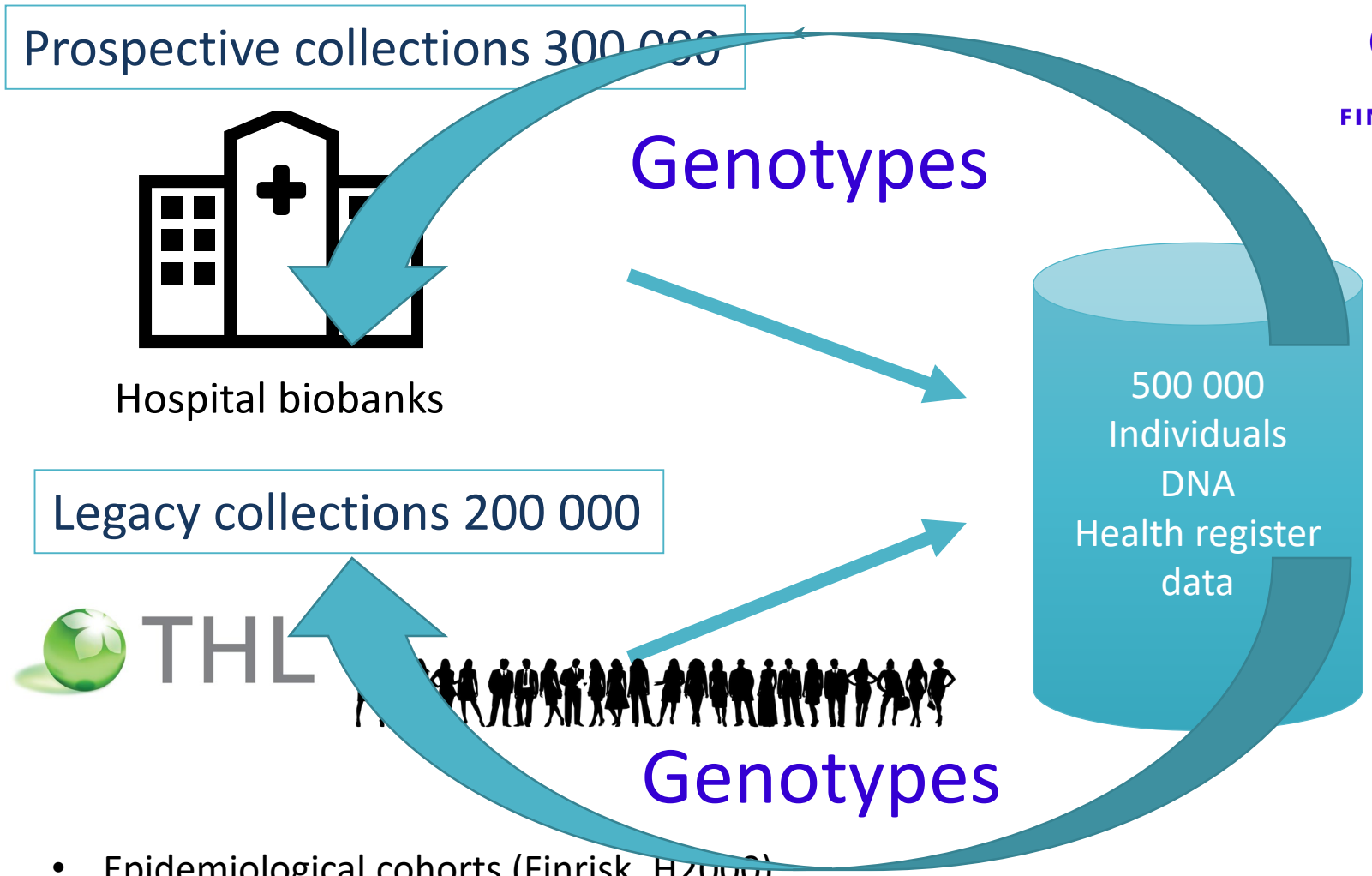
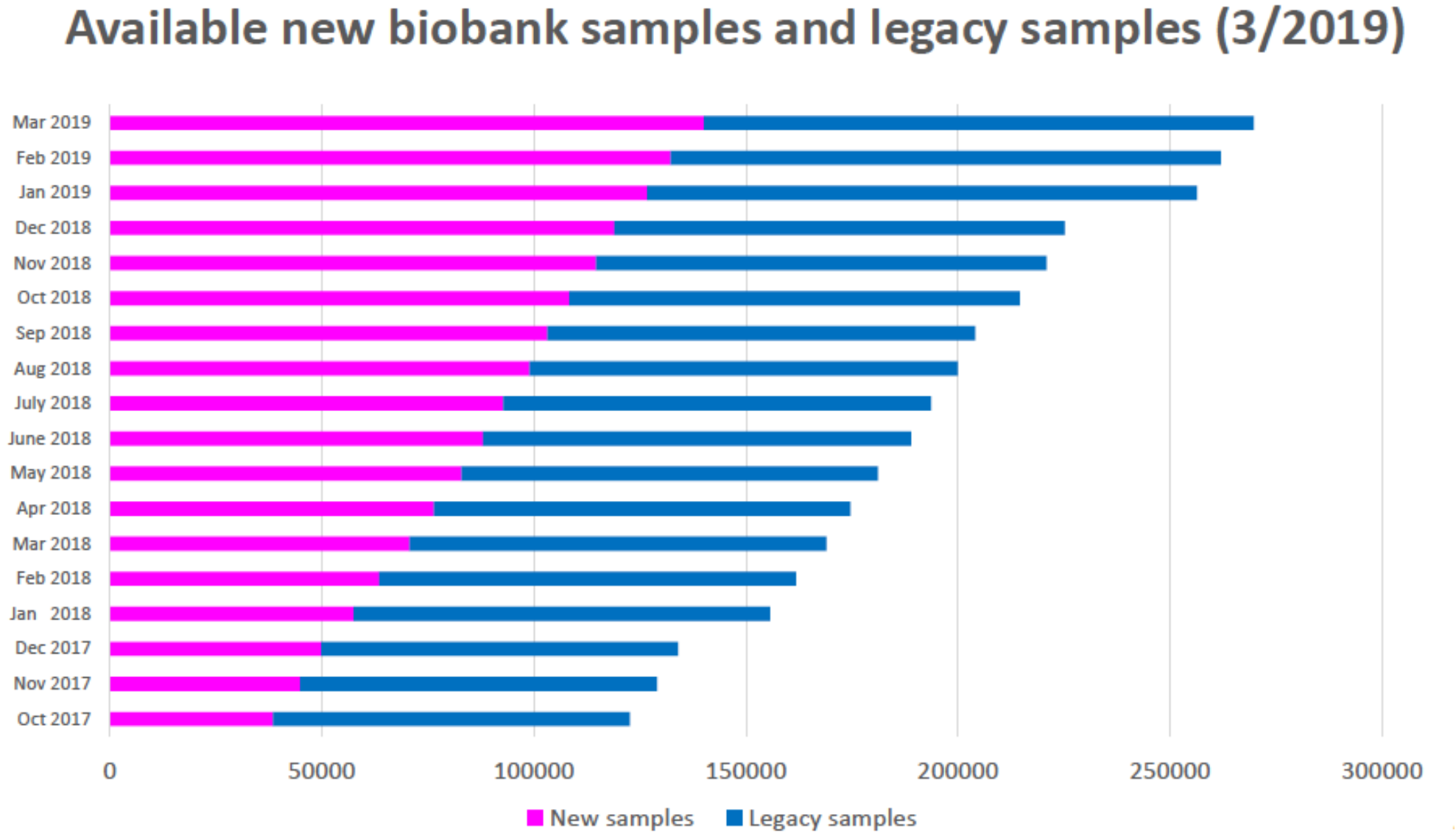
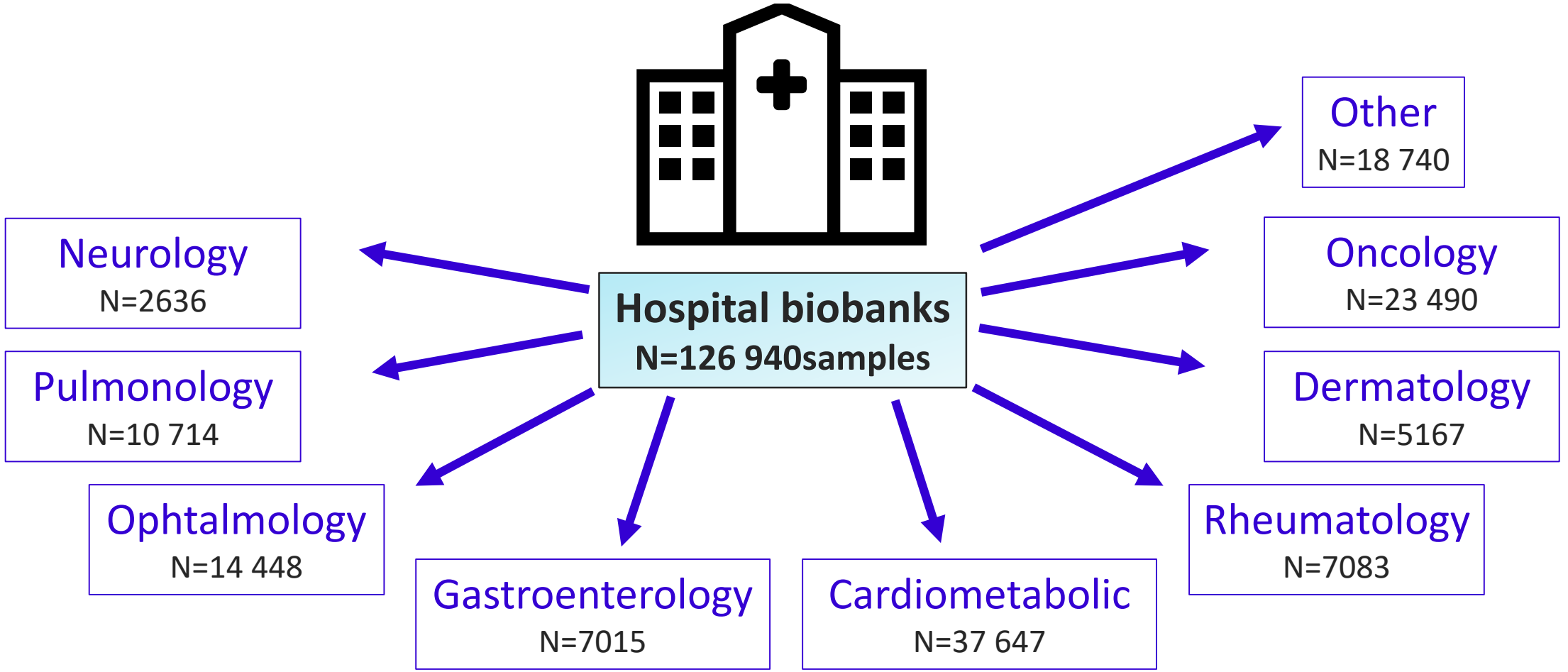


- Epidemiological cohorts (Finrisk, H2000)
- Disease collections:

Available new and legacy samples (02/2019)



FinnGen Disease Groups in new samples



Abbvie

Astra Zeneca

Biogen Celgene

Merck/MSD Pfizer

Genentech GSK

Sanofi

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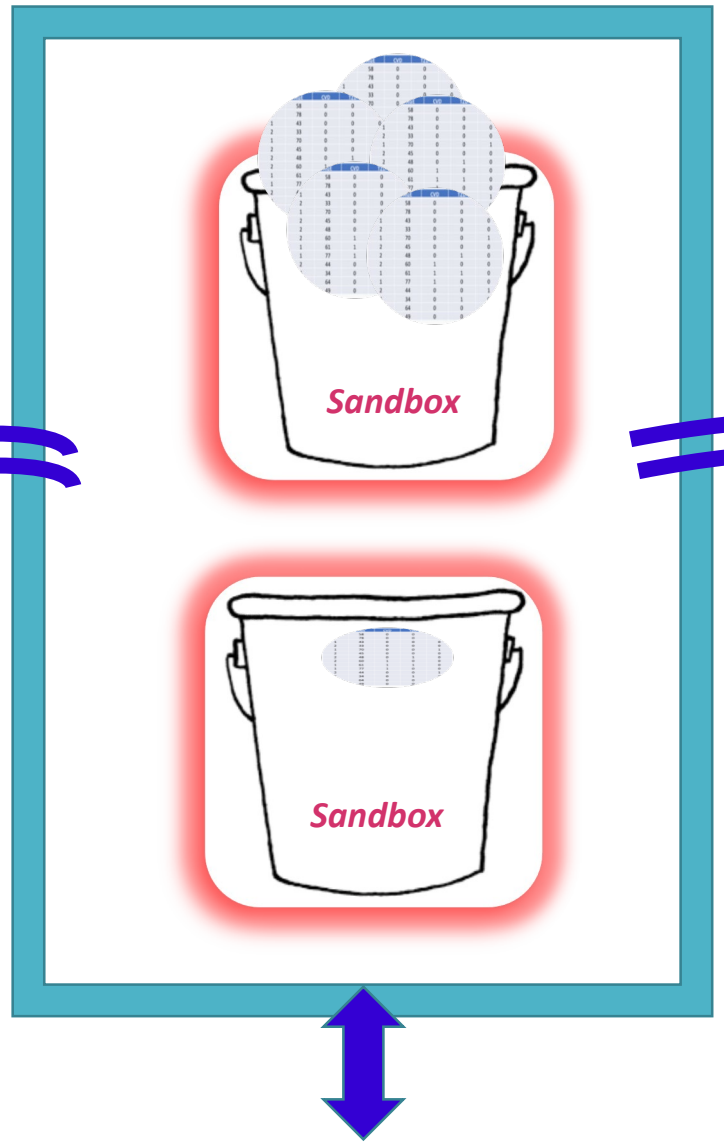


Secure access to:

- Individual level data
- Analysis results
- Data cannot be copied

Google Cloud

Health & Genome Data

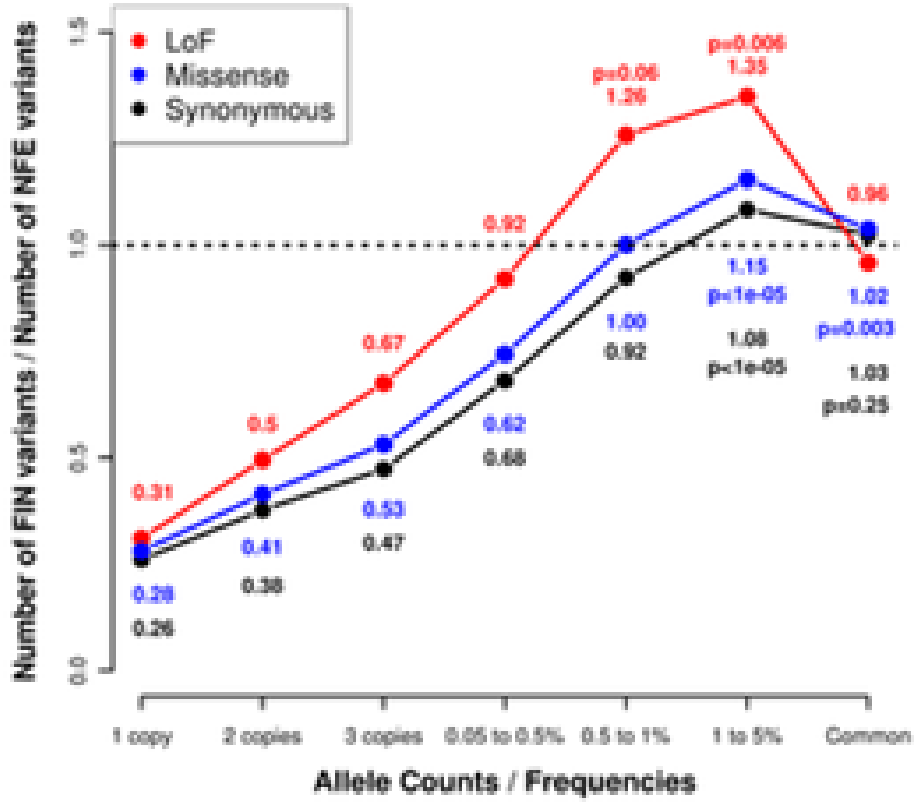
Tools

RARE  COMMON

Finnish enriched variants in common diseases

Cancer

High impact variants
”reversing Mendelian discovery”



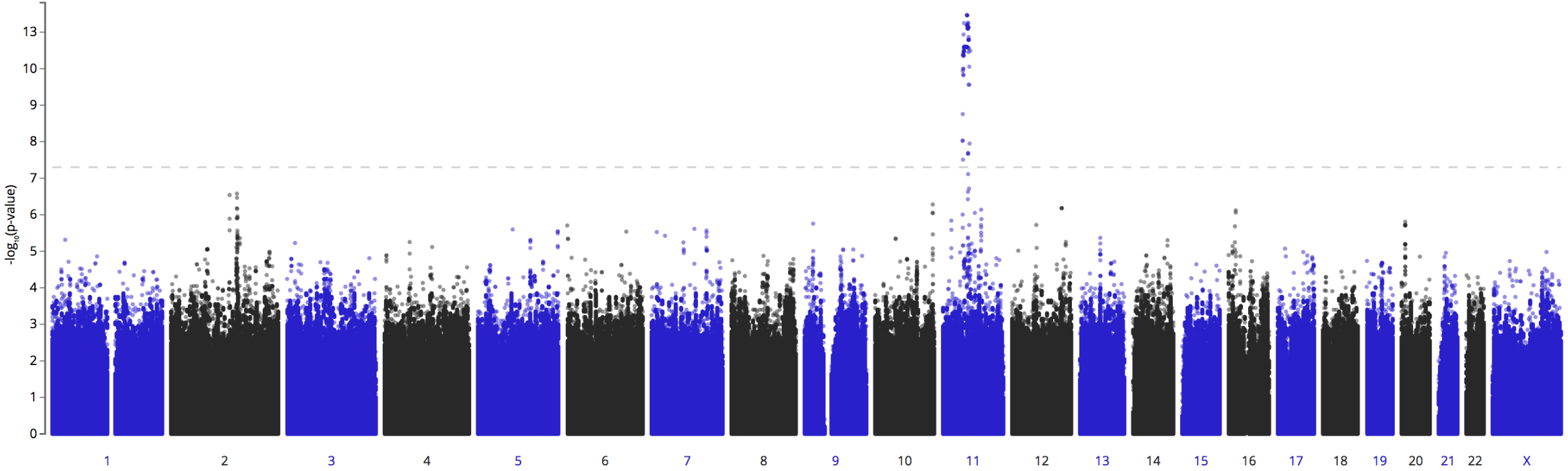
Cardiomyopathy, Hypertrophic obstructive

Category: Circulatory system

Includes ICD-10 codes: I422: Other hypertrophic cardiomyopathy
Control exclusion: Other heart diseases
Detailed info on endpoint definition

FINNGEN:**135** cases, **75137** controls
UKBB: **57** cases, **361137** controls
p-values smaller than 1e-10 are shown on a log-log scale

Truncation variant in MYBPC3
Carried by 6% of cases
<1 in 1000 controls
Enriched in Finland 4x



Towards Global Partnership

FINNGEN

GWAS/PheWAS

GWAS/PheWAS

Meta-analysis

FinnGen April 2019

FINNGEN

- Project has access to 240 000 biobank samples
- 170 000 genotyped
- GWAS and PheWAS analyses have been performed to 146 000 study participants
- First analysis results implicate new significant associations in both common and less common diseases

FinnGen 2019 tavoitteita

- Nopeuttaa näytekeräystä
- Kansainväliset Meta-analyysit 
- Valmistautuminen FinnGen2:een
- Demonstraatioprojekti, osoittamaan FinnGenin arvo
- Analyysityökalujen ja analyysiympäristön kehittäminen
- Löydösten jalostaminen kliinisiksi tutkimuksiksi

15

FinBB ja FinnGen

- Ensimmäisten add-on projektien käynnistäminen
- Olemassaolevat laboratorioanalyysit