Biobank registers in Finland

TMF Workshop: Sharing experiences and lessons learned while operating National Biobank Registers

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Topics

- The Finnish Biobank Law & Finnish Biobanks
- BBMRI.fi: the Finnish Biobank Network
- Biobank cataloging efforts:
  - Epidemiological sample collections
  - Clinical sample collections
- Challenges of standardizing & cataloging

Ministry of Social Affairs and Health, Finland
N.B. Unofficial translation. Legally valid only in Finnish and Swedish

688/2012

Biobank Act

Chapter 1

General provisions

Section 1

Objectives

The objectives of this act are to support research that utilises human biological samples, to promote openness in the use of these samples and to secure the protection of privacy and self-determination when processing these samples.

The law came into force on September 1, 2013

Main objectives:

- Support research that utilizes human biological samples
- Promote openness in the use of the samples
- Secure the protection of privacy and self-determination when processing these samples

http://www.finlex.fi/fi/laki/kaannokset/2012/en20120688.pdf

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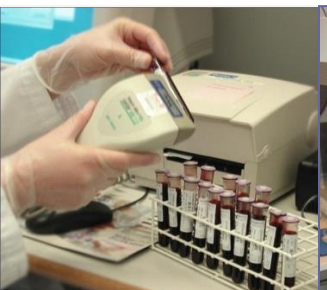
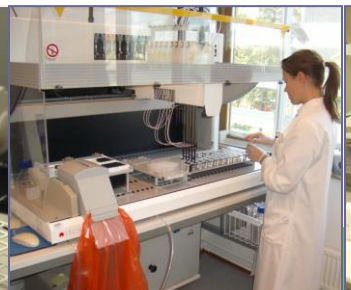
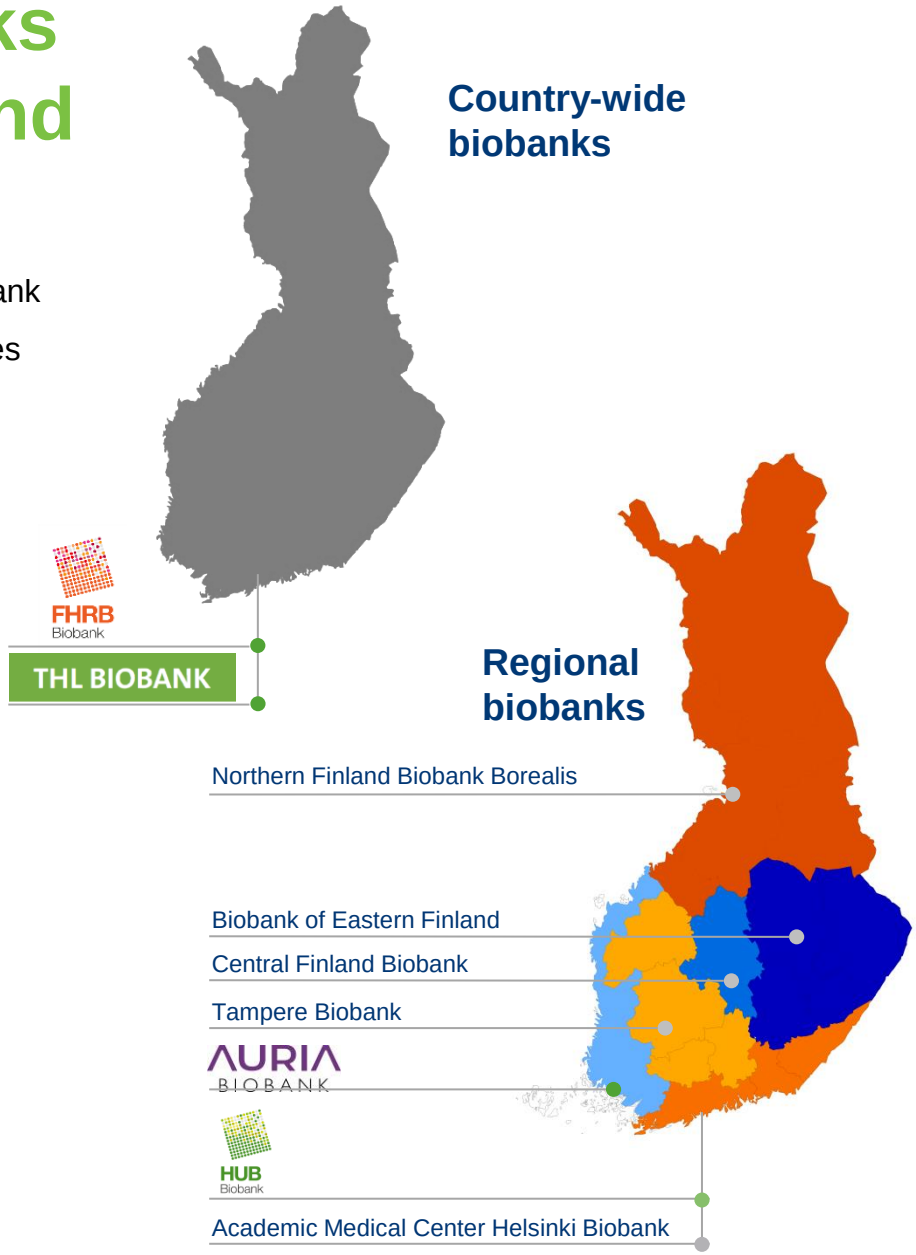
Features of the Biobank Law

- Biobanks have to register in a national Biobank Register (maintained by the National Supervisory Authority for Welfare and Health)
- Allows wide consent. Example:
  - ”The aim is to develop new methods for diagnosis, treatment and prognosis of urological cancers… ” (HUB project)
- Allows transfer of old sample collections and hospital’s clinical collections to a biobank with new wide consent
- New samples (clinical and research) can be collected directly to a biobank for future undefined research


Biobanks in Finland

- Registered biobank
- Biobank initiatives



THL Biobank:

- Belongs to the National Institute for Health and Welfare
- Hosts country-wide population- and family-based research sample collections

HUB &FHRB:

- Samples from hospital patients with specific diseases

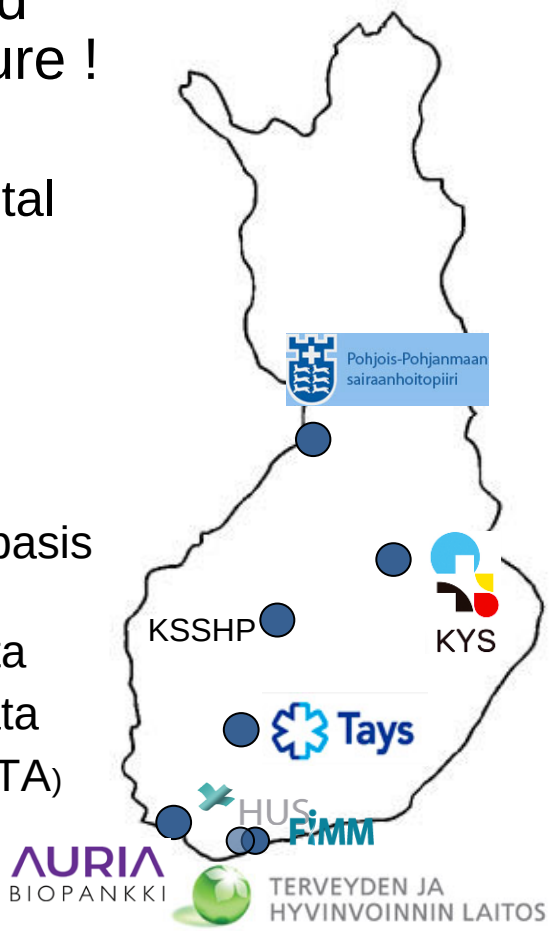
Regional biobanks:

- Operated through collaboration between hospital districts and universities
- Host mainly hospital sample and data collections

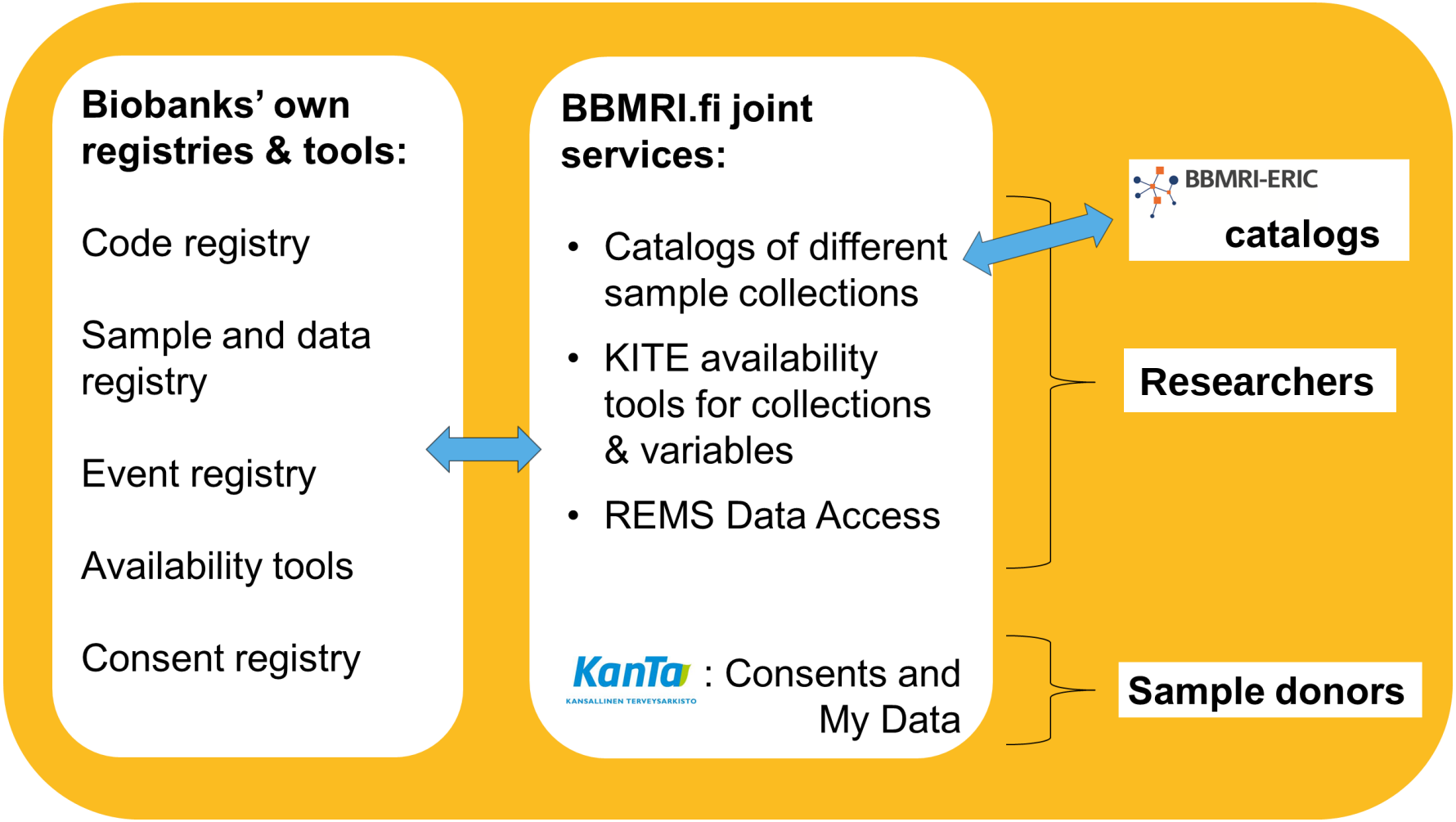
A Unique National Collaboration

- Large National Biobanks are being established **simultaneously** with the BBMRI.fi infrastructure !
  - Coordination by THL
  - Partner biobanks cover 6 universities and all hospital districts

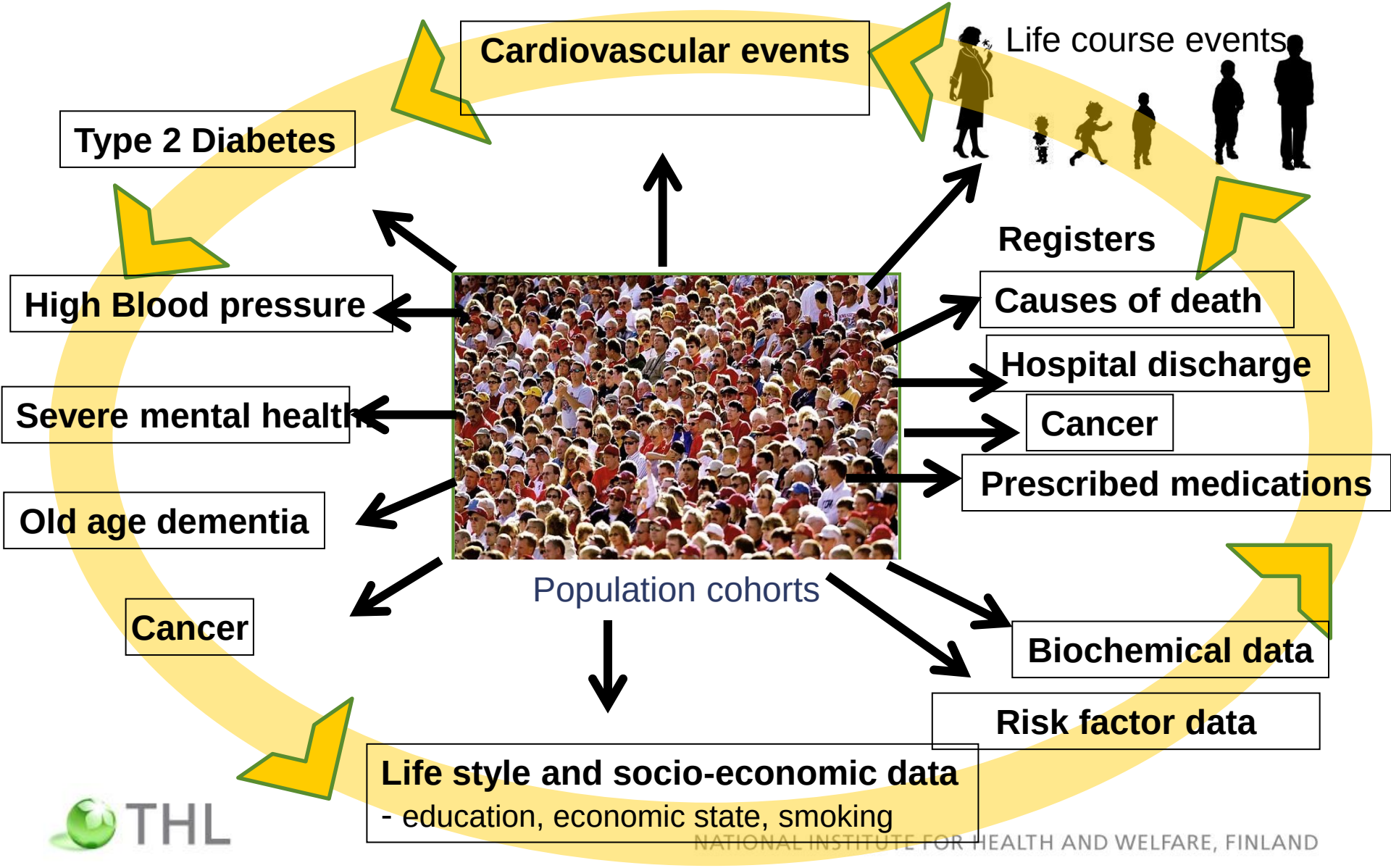
- Finland is a Member Country of BBMRI-ERIC
- BBMRI.fi general aims are in line with BBMRI-ERIC:
  - Ensure access to high quality samples and data for the basis of high quality research
  - Ensure utilization of BBMRI standards for samples & data
  - Build a common access portal for biobank samples & data
  - Organize nationally unified, legal access principles (& MTA)



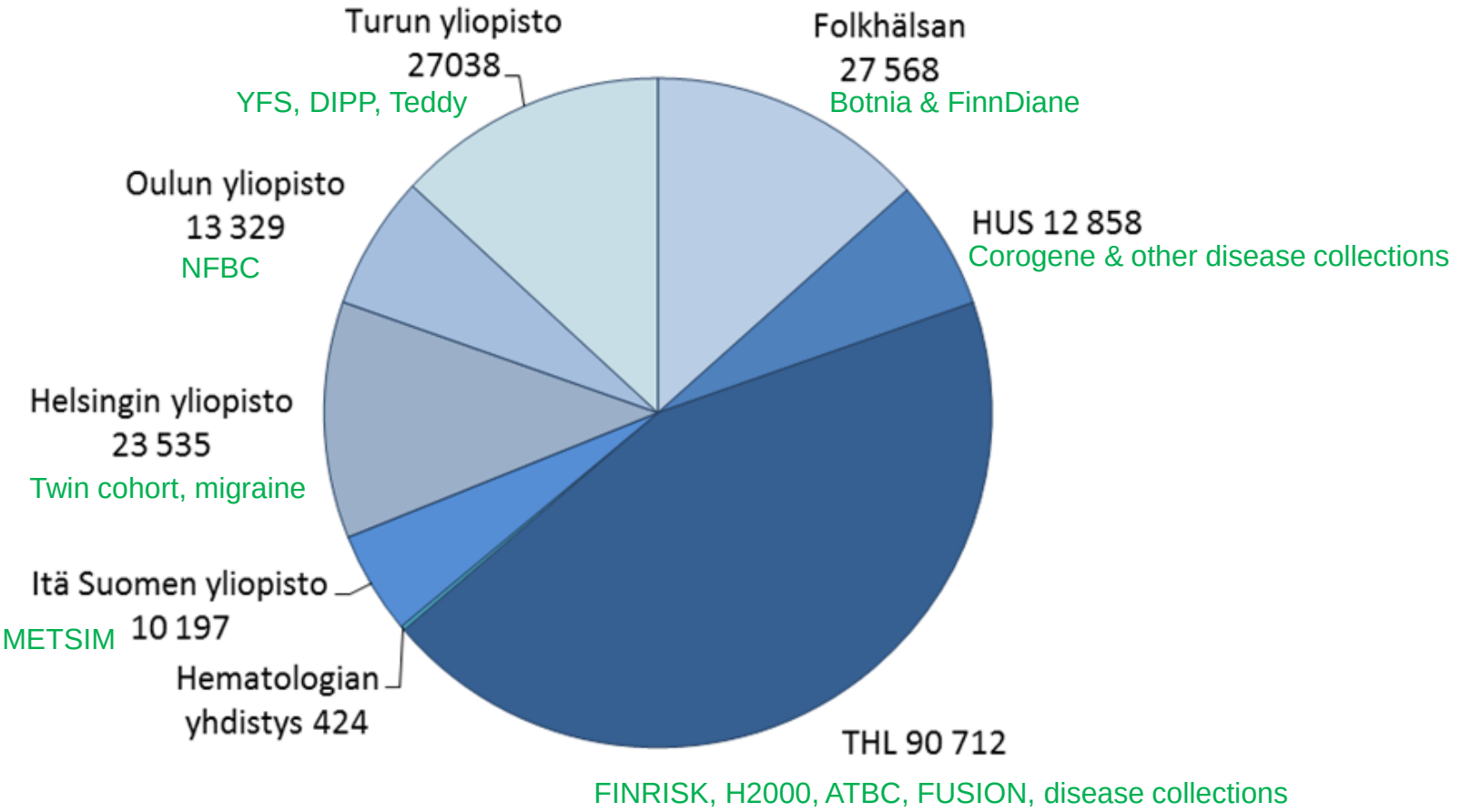
A plan for National ICT-infrastructure



Population cohorts



Existing DNA samples collections



Tools aiding researchers in finding suitable samples

Searchable catalog:

- Type of sample collection
- Basic information (target population, number of samples, regions, age groups)
- Data categories & Standardized variables

Nationalbiobanks.fi

KITE

Find suitable collections

Availability service of biobank:

- based on individual-level phenotype data
- in collaboration with cohort researchers
- only aggregated results

REMS

Electronic application system

Obtain permission, MTA & DTA

Raw data database:

SamWise

- Specific secure access to data
- Selection & downloading data for analysis
  - Variable values and metadata
  - Omics data

bbmri-lpc

aims to promote the access to European population based cohorts

Nationalbiobanks.fi

Pages currently focus on DNA sample collections

Nationalbiobanks.fi: study description

FINRISK

Published on Tuesday, 12 June 2012 21:50 | Written by Super User | | Hits: 7558

The National FINRISK Study (BRIF 1640)

The FINRISK cohorts comprise the respondents of representative, cross-sectional population surveys that are carried out every 5 years since 1972, to assess the risk factors of chronic diseases (e.g. CVD, diabetes, obesity, cancer) and health behavior in the working age population, in 3-5 large study areas of Finland. DNA samples were collected in the following survey years: 1987, 1992, 1997, 2002, 2007, and 2012. The MONICA and EHES (EU) procedures were applied in phenotype collection (cf. MORGAM) and a wide spectrum of laboratory tests was carried out from serum and plasma samples. Background information on socioeconomic status, medical history, diet, exercise, measured anthropometric measures, etc. was collected by questionnaires and during a clinical visit. Plasma/serum samples are still available for the 2002-2012 cohorts. The cohort sizes are 6000-8800 per survey. The cohorts have been followed up by linking them to the national hospital discharge register, causes-of-death register and cancer register. Genome-wide SNP data is available for several subsets of FINRISK participants (see GWAS Table).

Link to homepage [in Finnish]

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Contact person:
Satu Männistö, firstname.lastname (at) thl.fi

Key reference:
Thirty-five-year trends in cardiovascular risk factors in Finland. Vartiainen E, Laatikainen T, Peltonen M, Juolevi A, Männistö S, Sundvall J, Jousilahti P, Salomaa V, Valsta L, Puska P. Int J Epidemiol. 2010 Apr;39(2):504-18

FINRISK 1992, 1997, 2002, 2007, 2012 (1972-): Detailed information

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KITE availability tools:

- Catalog information on biobanks and sample collections based on MIABIS
- Browse & search tools for each collection's standardized variables
- First implemented by THL Biobank

- https://kite.fimm.fi

FIMM
Institute for Molecular Medicine Finland
Nordic EMBL Partnership for Molecular Medicine

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KITE: MIABIS attributes for describing sample collections

The National FINRISK Study 1992

Acronym: FINRISK 1992

Description

The FINRISK cohorts comprise the respondents of representative, cross-sectional population surveys that are carried out every 5 years since 1972, to assess the risk factors of chronic diseases (e.g. CVD, diabetes, obesity, cancer) and health behavior in the working age population. FR1992 studied the following regions: North Karelia, North Savonia, Turku/Loimaa and Helsinki/Vantaa.

Age range / gender: 25 - 64 year / male and female

Data categories

biological samples, survey data, national registries, Physiological/Biochemical measures

Survey data
Health information, sociodemographic characteristics, socioeconomic characteristics, physical environment, mental health, familial disease history, individual disease history, individual history of injuries, medication use, perception of health/quality of life, woman's health, reproductive history, Sleep, Alcohol usage, Smoking, Diet, Exercise, physical activity

Medical data

Inclusion Criteria
Population representative sampling

Collection Type
Cohort, Cross-sectional, Population-based

Consent
broad consent

Rules for access
cooperation, co-authorship, return data

Donor / sample counts
6024 /

Sample type
DNA, plasma, serum

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Steps in retrospective variable harmonization of epidemiological cohorts

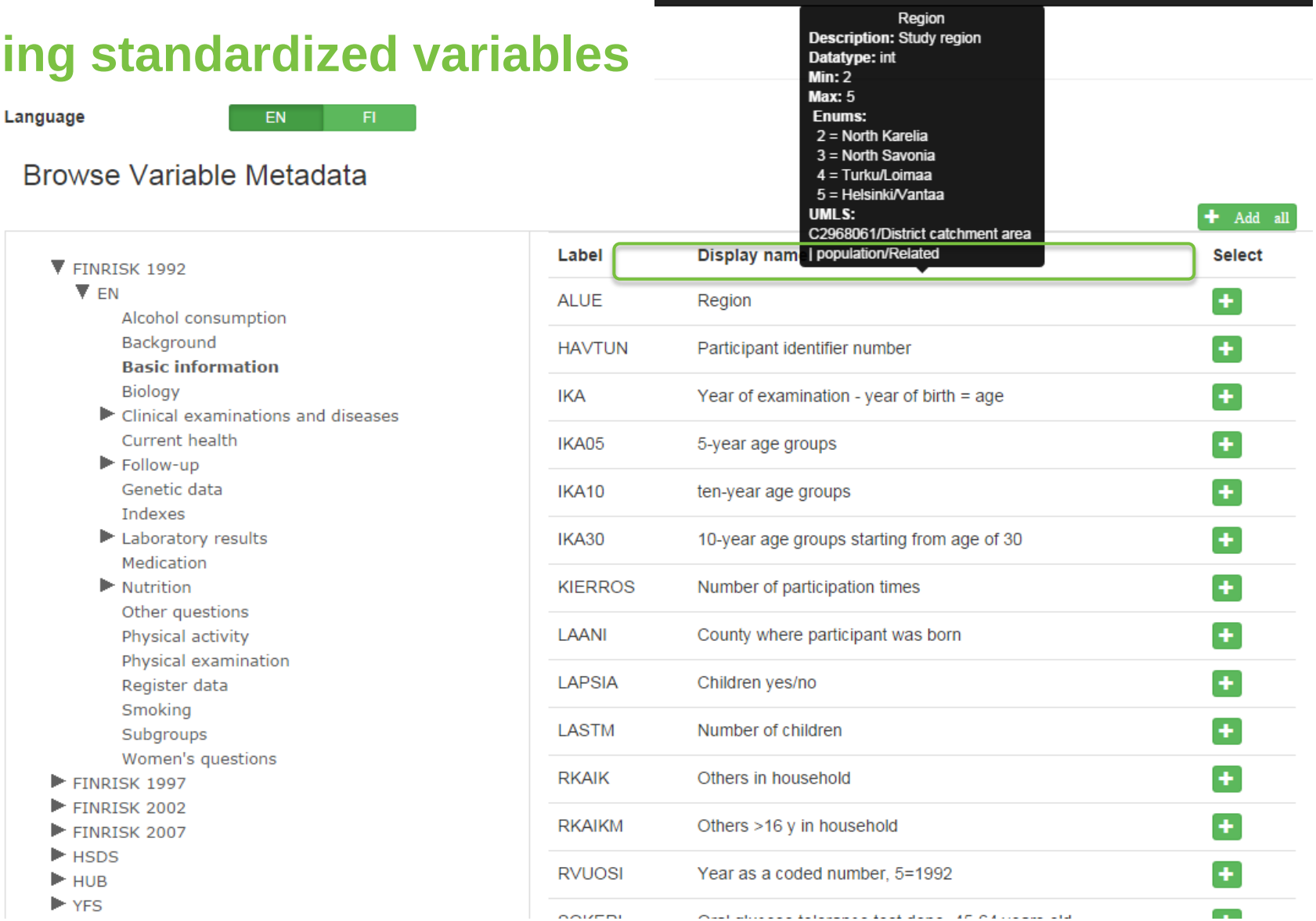
Manual

- 1. Tabulating all variable metadata
- 2. Translating variable metadata to English
- 3. Adding reference terms: UMLS-metathesaurus
- 4. Creating an ontology (=structure), adding keywords
- 5. Importing the datasets to SamWise/KITE

Tools needed:

- 6. Cross-referencing variables with other datasets
- 7. Building up reference dictionaries (UMLS and keywords)

KITE:
Browsing standardized variables



https://kite.fimm.fi

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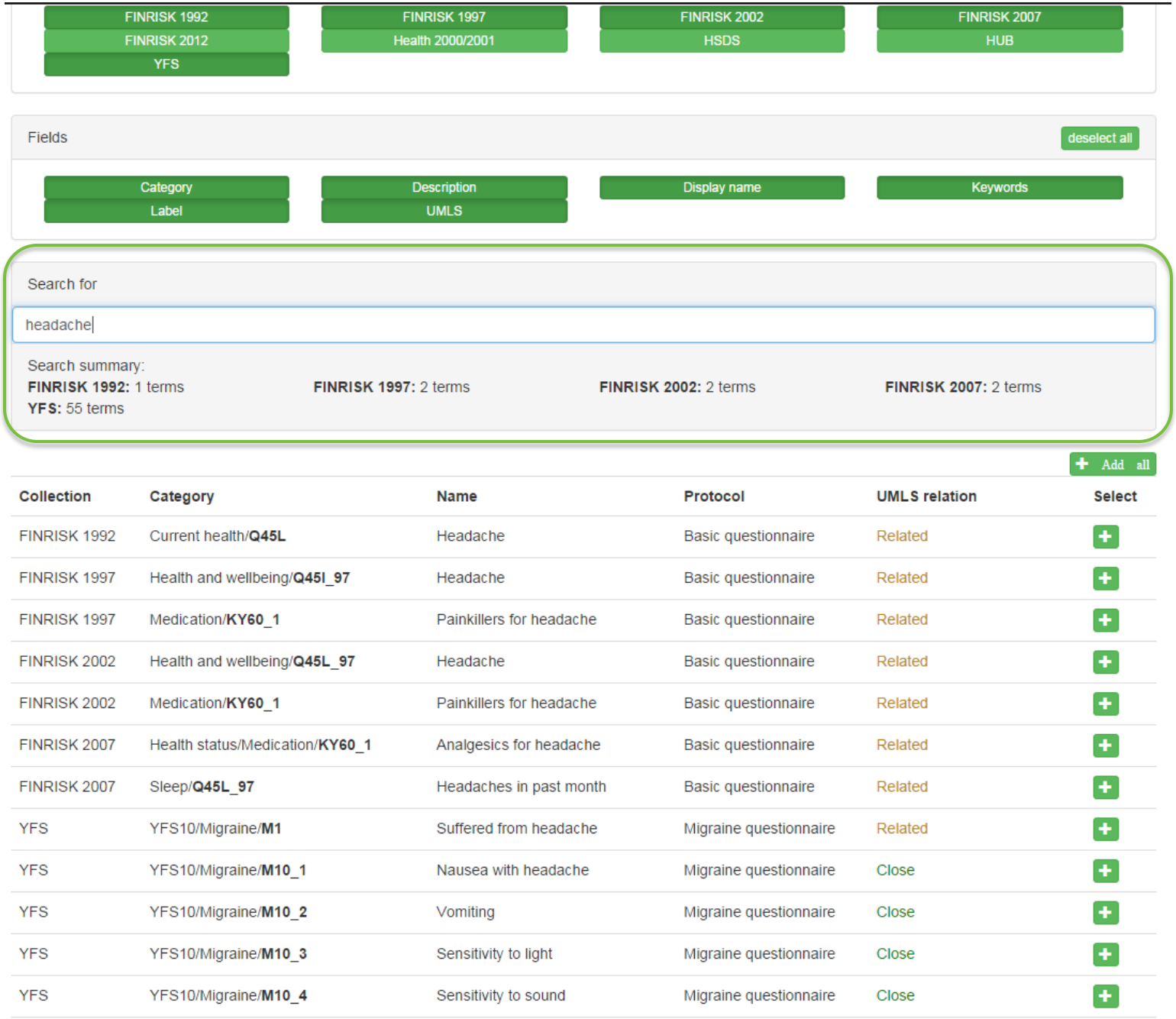
KITE: Searching variables



Collection	Category	Name	Protocol	UMLS relation	Select
FINRISK 1992	Current health/**Q45L**	Headache	Basic questionnaire	Related	+
FINRISK 1997	Health and wellbeing/**Q45I_97**	Headache	Basic questionnaire	Related	+
FINRISK 1997	Medication/**KY60_1**	Painkillers for headache	Basic questionnaire	Related	+
FINRISK 2002	Health and wellbeing/**Q45L_97**	Headache	Basic questionnaire	Related	+
FINRISK 2002	Medication/**KY60_1**	Painkillers for headache	Basic questionnaire	Related	+
FINRISK 2007	Health status/Medication/**KY60_1**	Analgesics for headache	Basic questionnaire	Related	+
FINRISK 2007	Sleep/**Q45L_97**	Headaches in past month	Basic questionnaire	Related	+
YFS	YFS10/Migraine/**M1**	Suffered from headache	Migraine questionnaire	Related	+
YFS	YFS10/Migraine/**M10_1**	Nausea with headache	Migraine questionnaire	Close	+
YFS	YFS10/Migraine/**M10_2**	Vomiting	Migraine questionnaire	Close	+
YFS	YFS10/Migraine/**M10_3**	Sensitivity to light	Migraine questionnaire	Close	+
YFS	YFS10/Migraine/**M10_4**	Sensitivity to sound	Migraine questionnaire	Close	+

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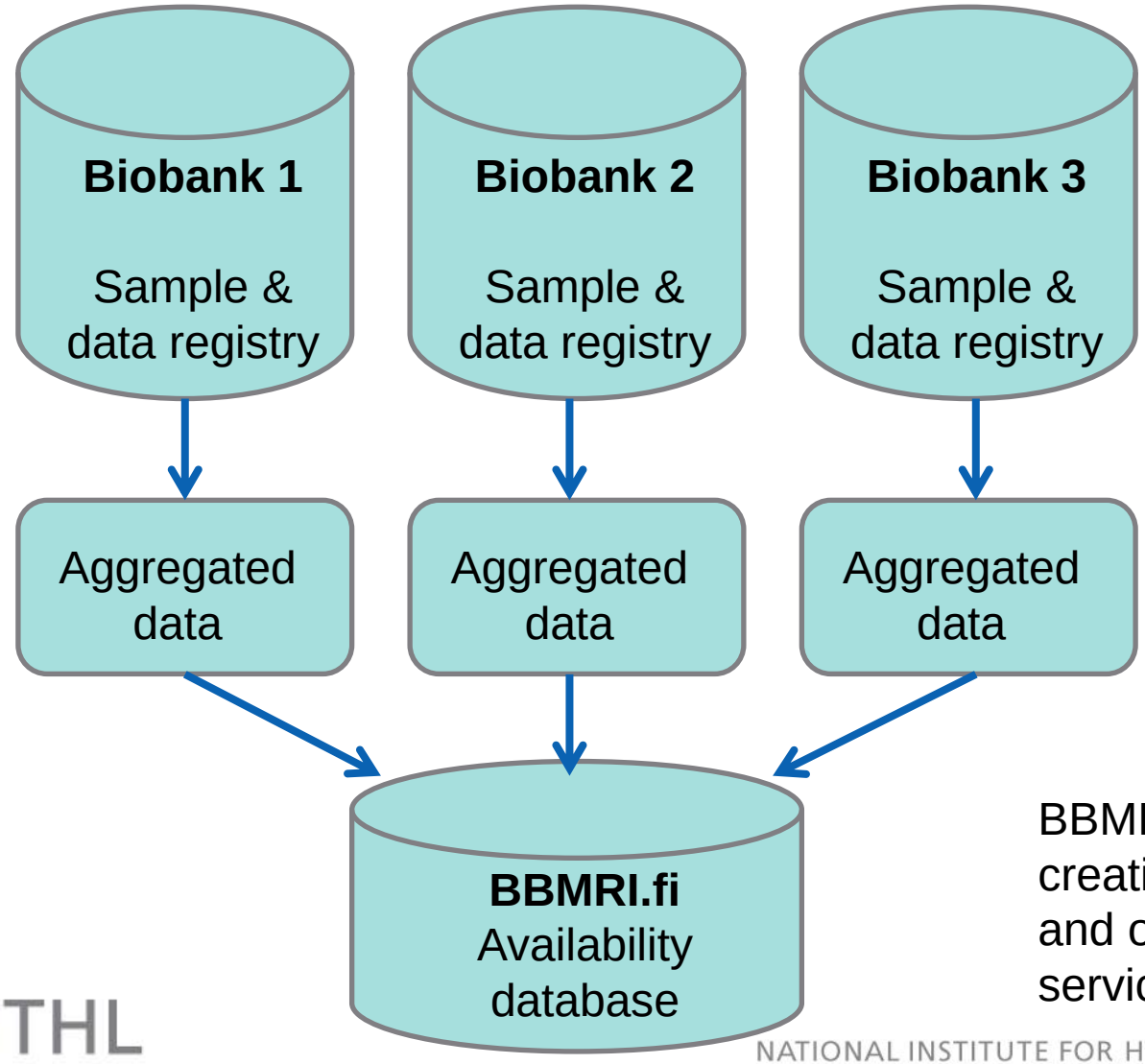
National Availability Database (based on KITE)



Each biobank decides what data is offered to the availabilty database

Each biobank anonymizes & aggregates the data selected

BBMRI.fi is responsible for creating the joint database and offering availability services

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REMS electronic data access for biobanked samples & data:

Resource Entitelement Management System

- Personal login and researcher profile
- Sample and data request
- Approval and material transfer agreement
- First implemented by THL Biobank

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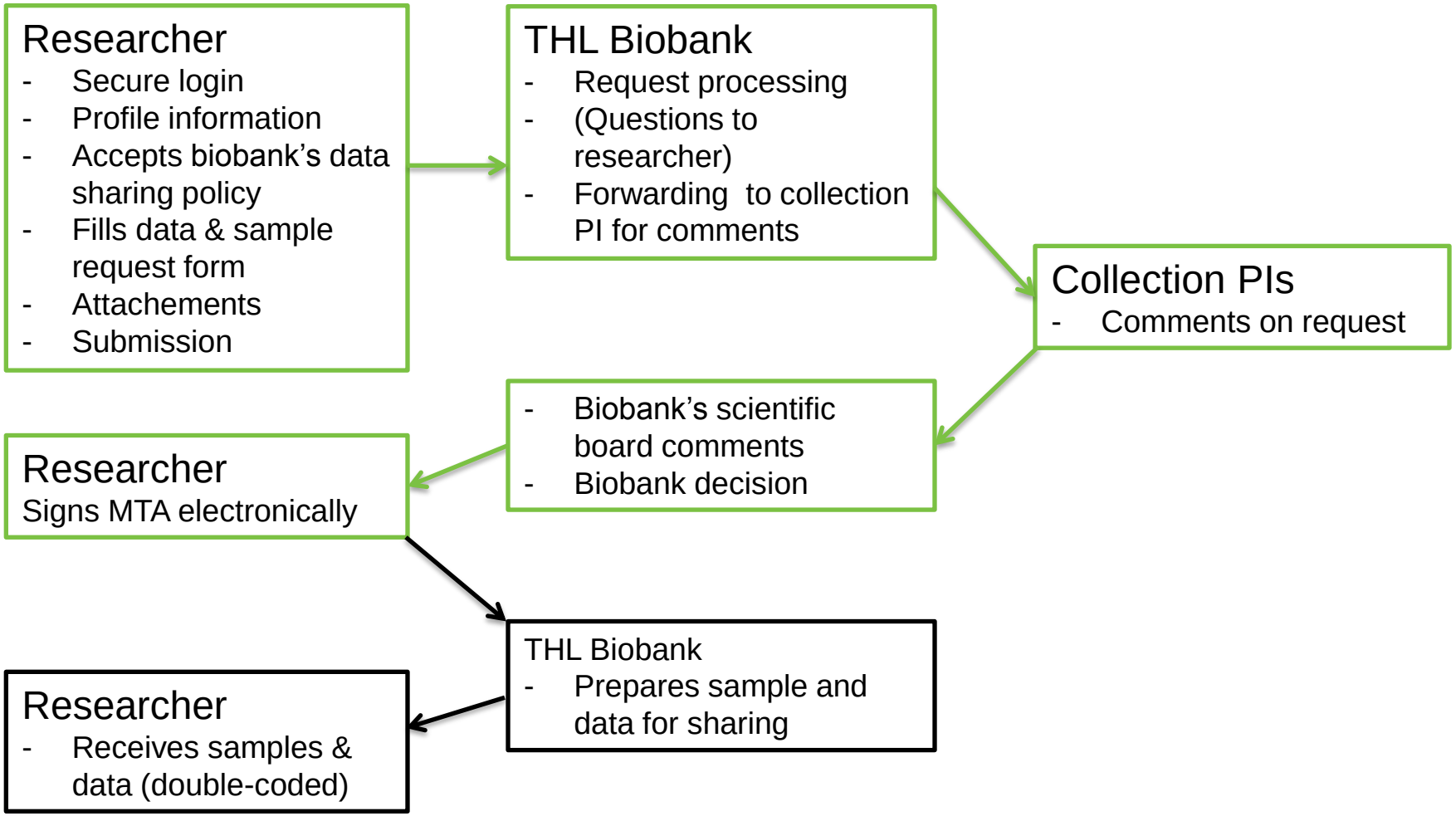
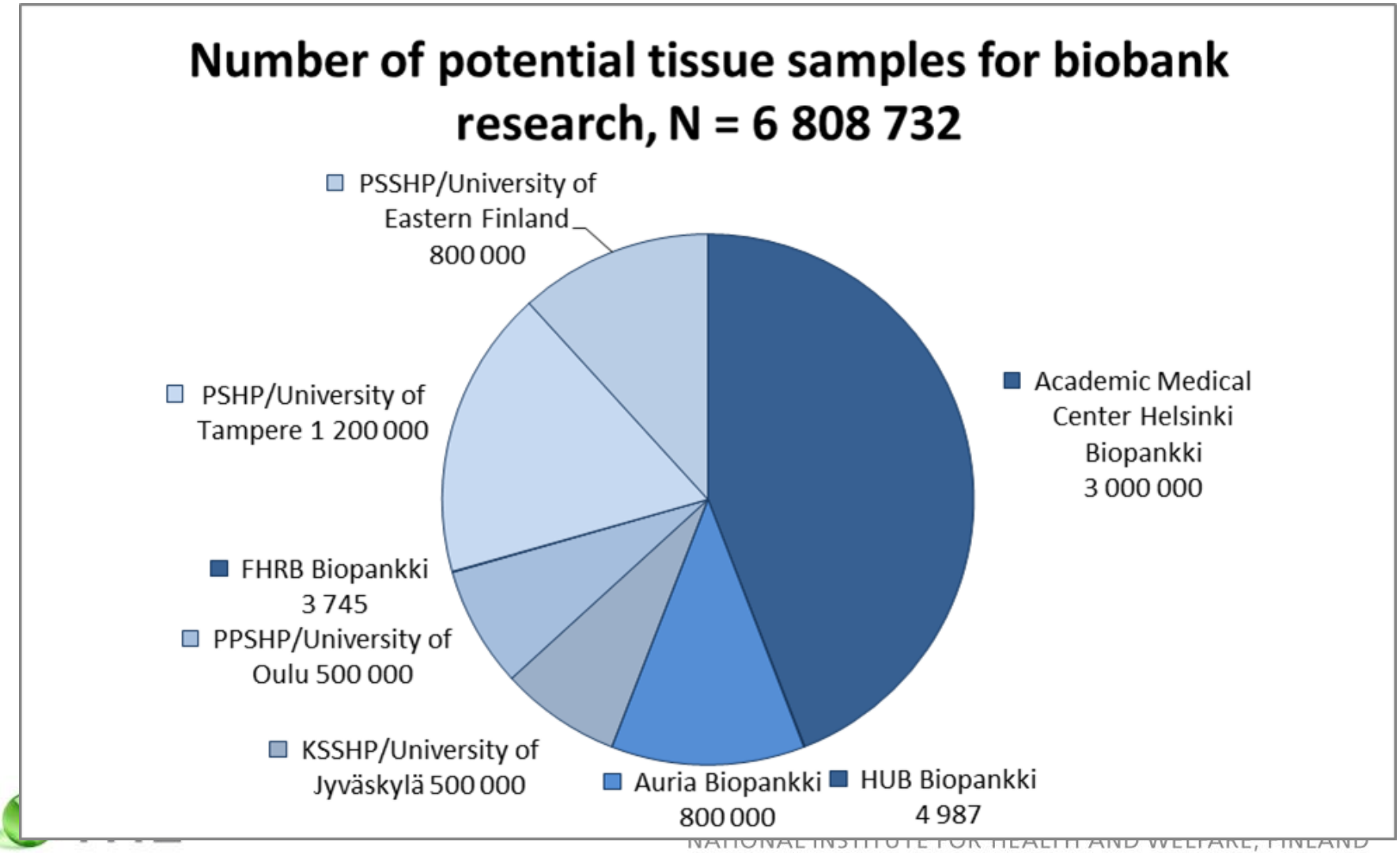
REMS: Resource Entitlement Management System

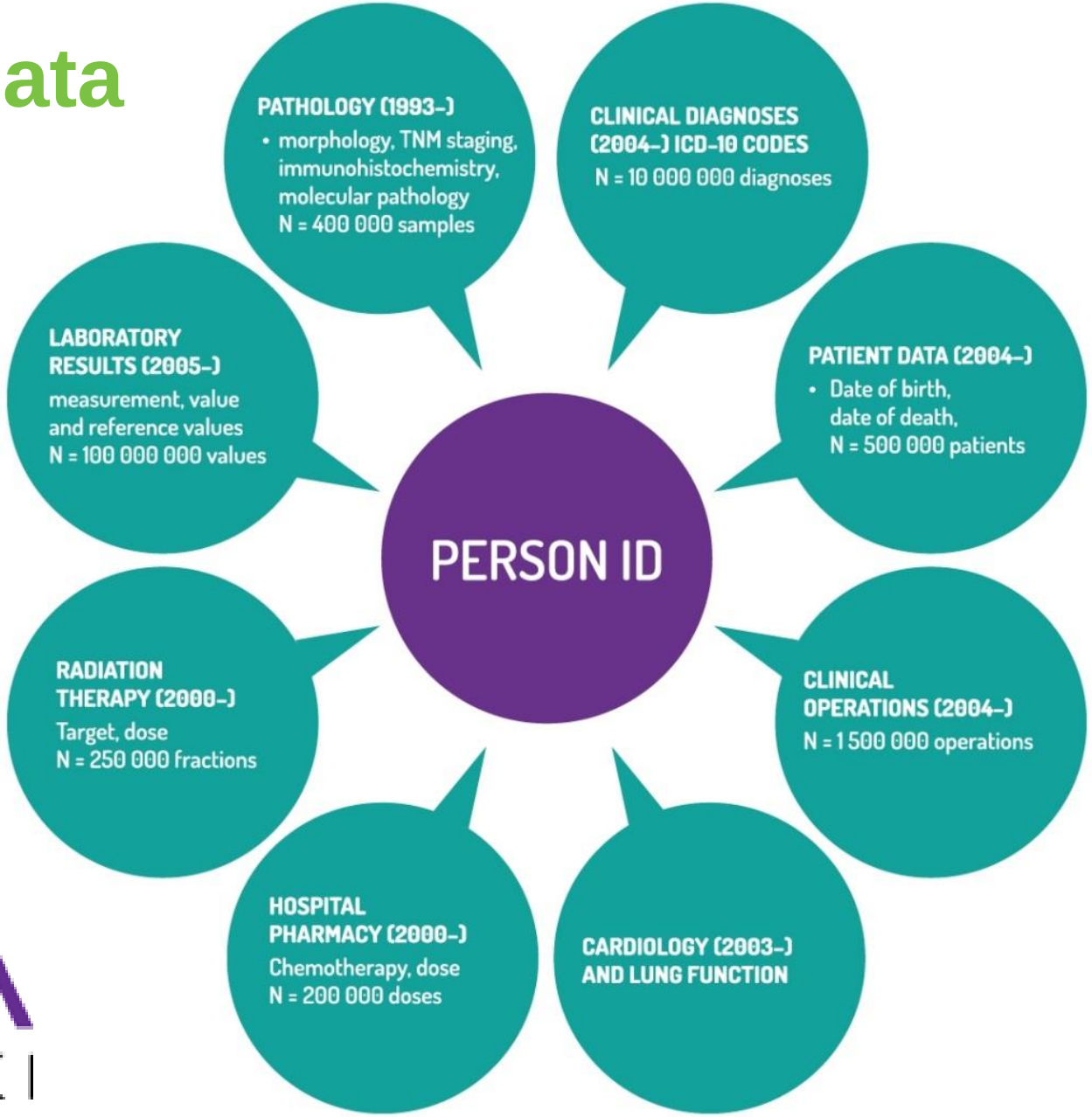


REMS vaiheet vihreällä

Old FFPE tissue samples in hospitals/clinical biobanks

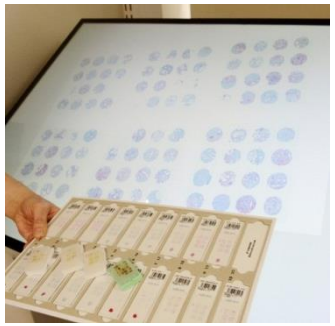


Clinical data



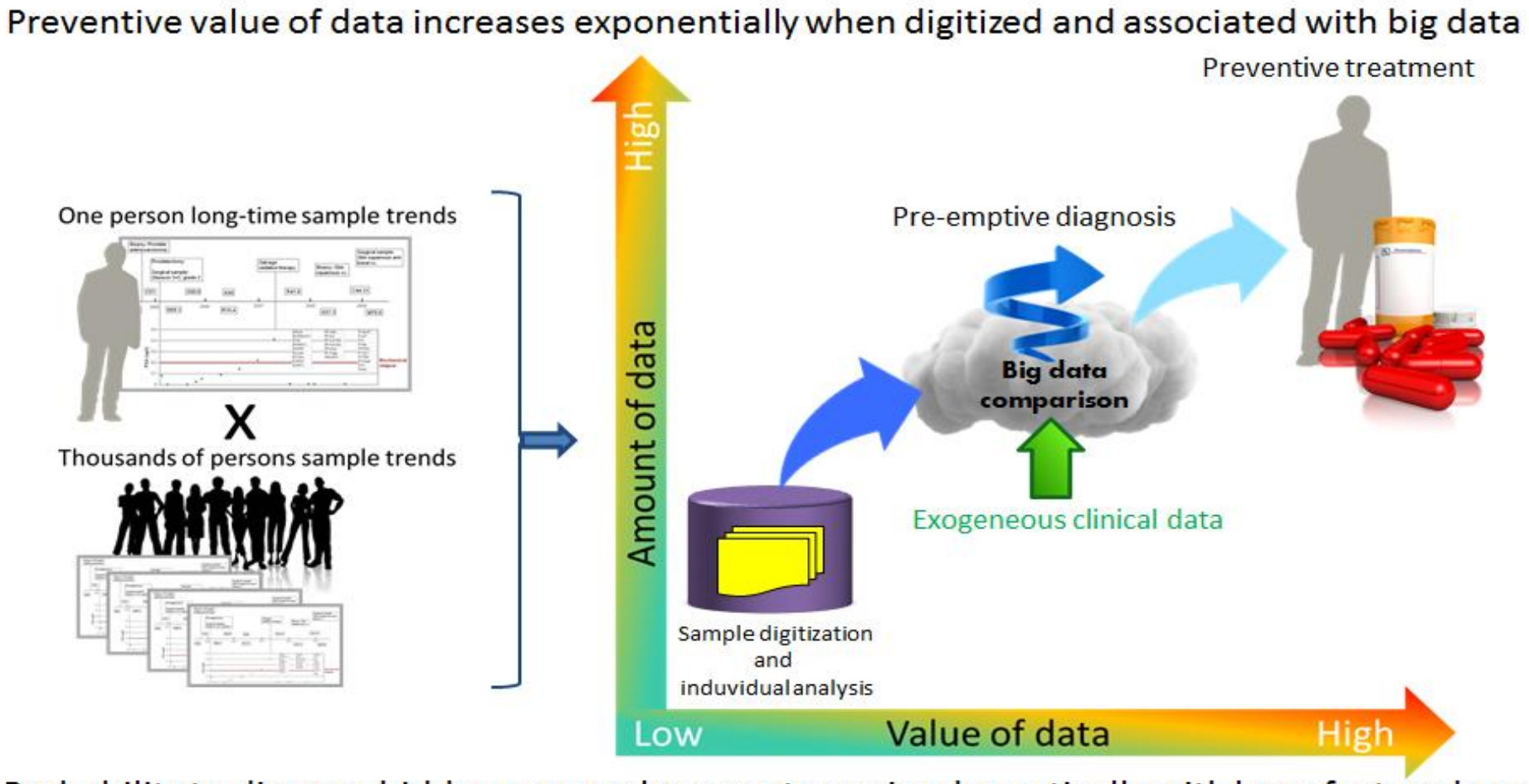
AURIA BIOPANKKI

Harmonizing Pathological Samples & Patient Data



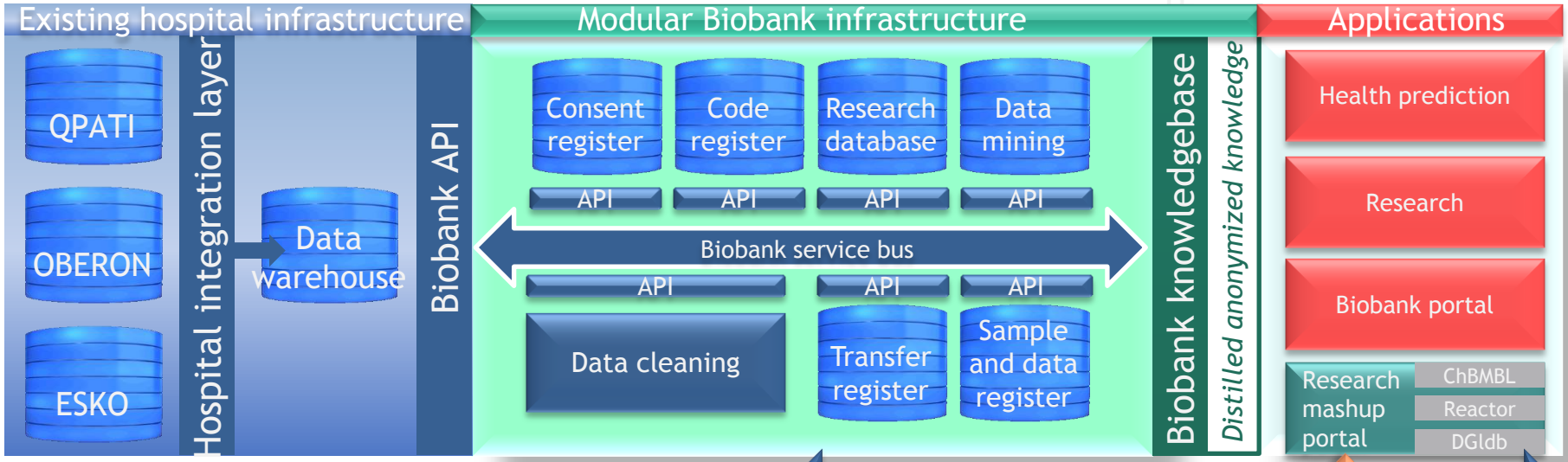
- Cataloging tissue samples: sample type, organ and histological diagnosis (snomed-code), donor age and sex, and name of biobank
- Standardization and harmonization of associated patient data
- Digital archive: delivery and storage of digital images of tissue specimens, annotation tools

Added value from harmonizing patient data



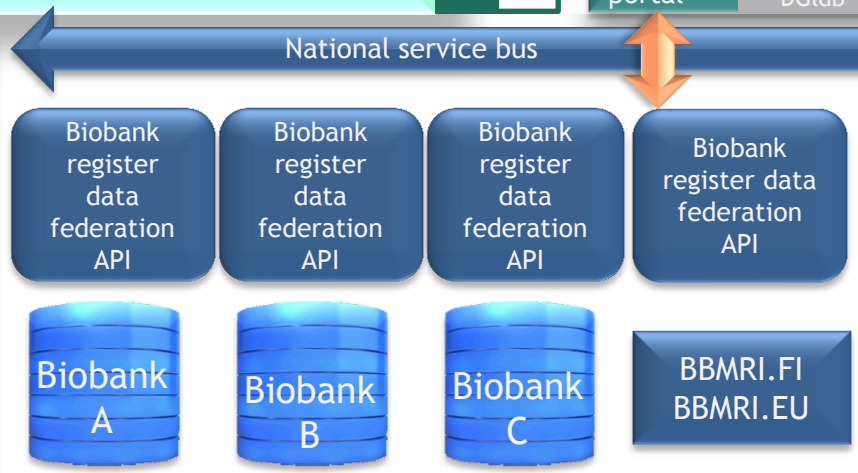
Slide from Pia Nyberg, Northern Finland Biobank Borealis

Clinical Biobanks: ICT Opportunities



ORGANIZING - SEARCHING - SERVICING - DISCOVERING

1. Constantly cumulative **information**
   - Samples and refined analyses
2. Content-based big data **mining**
   - Associated content from many sources
3. Application and discovery **tools**
4. Many areas and levels for **discoveries**
   - Prevention, treatments, care, knowledge
   - Nations, geographical, groups, individuals



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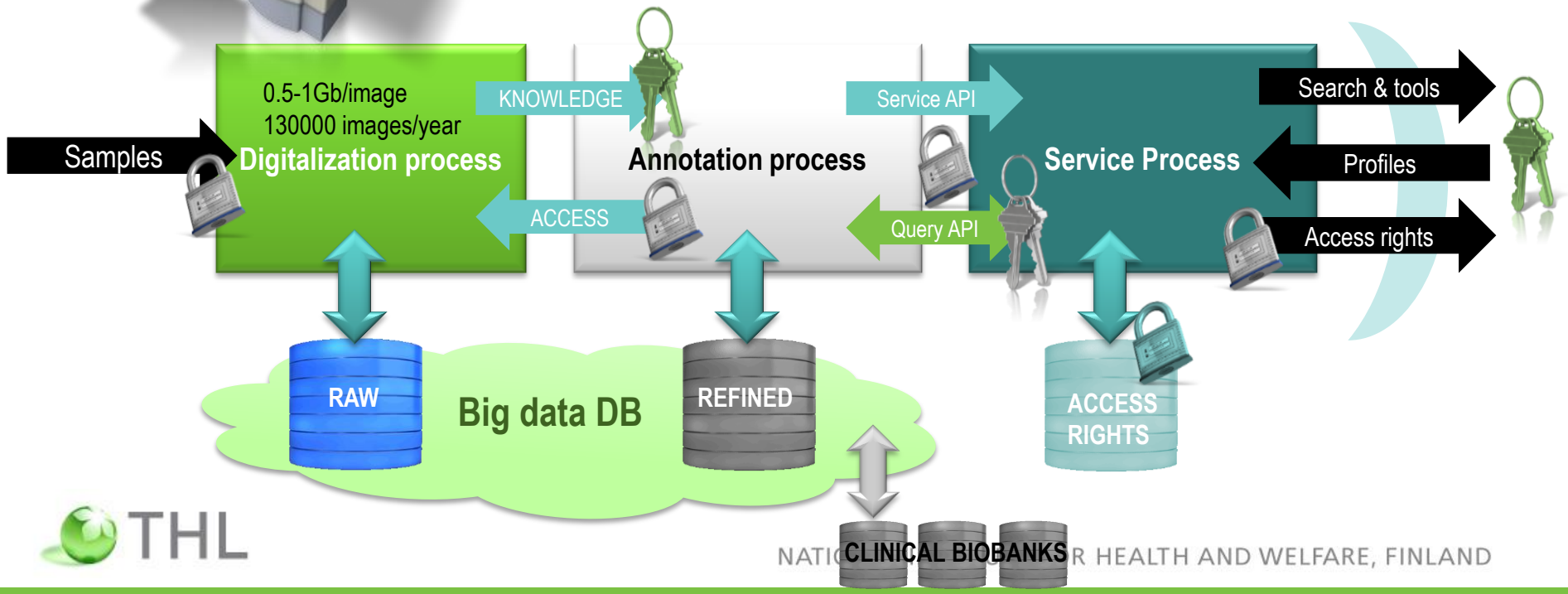
Slide from Pia Nyberg, Northern Finland Biobank Borealis

Digital biobank data is annotated & enriched: New opportunities for research, technology and citizens

General image analysis, specific analysis and associations

Service layer
Tools, processes & applications



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Slide from Pia Nyberg, Northern Finland Biobank Borealis

Contact information for the digitalization project

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Challenges in developing general catalogs & availability tools

- Different types of sample collections with different characteristics and different cataloging needs
- Differing levels of information about sample donors and different types of information
- How should one catalog hospital's pathological and other sample collections?
- Each hospital district has different database systems and uses different diagnosis vocabularies
- Different access policies for different collections – need to harmonize within a biobank and between biobanks
- Each epidemiological collection has its own database system

Different sample collections

FINRISK

- Prospective cohorts
- 5 regions in Finland
- Baseline questionnaire, clinical examination, laboratory measurements, follow-up register data
- DNA, RNA, cells, serum, plasma, urine, feces
- -omics data

HUCH pathological collections

- pathological samples collected for medical diagnostics
- District of Helsinki and Uusimaa
- Histological diagnosis and possibly associated medical records
- Tissues (frozen, paraffin)

Comparing variables between studies

FR87: marital status
1: married/cohabiting
2: single
3: living separately/separated
4: widow

FR97: marital status
1: married
2: cohabiting
3: single
4: separated/divorced
5: widow

FR02: marital status
1: married
2: cohabiting
3: single
4: separated/divorced
5: widow

Exact match

Exact match

EHES: MARIT_STATUS
1 = Single (never married)
2 = Married (including registered partnership)
3 = Widowed and not remarried
4 = Divorced and not remarried (incl. legally separated and dissolved registered partnership)

FR07: marital status
1: married
2: cohabiting
3: single
4: separated/divorced
5: widow

Close match

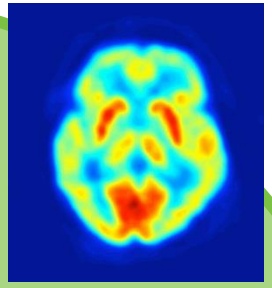
FR92: marital status
1: married/cohabiting
2: single
3: living separately/separated
4: widow

FR12: marital status
1: married
2: cohabiting
3: single
4: separated/divorced
5: widow
6: registered partnership

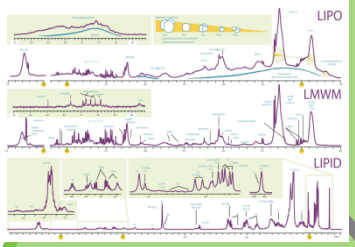
H2000: Are you currently
1: married
2: living with your partner
3: divorced or living apart
4: widowed
5: single

Big Data

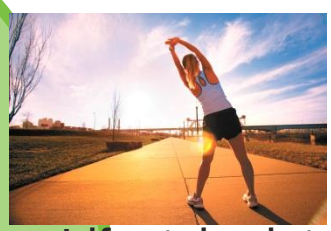
National health registries



Functional imaging



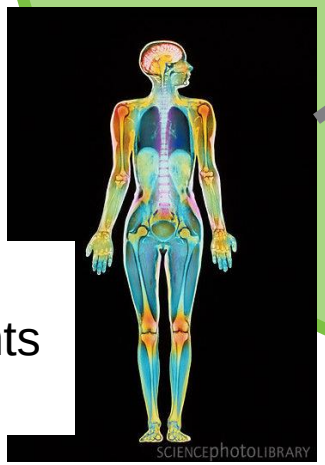
Metabolomics



Lifestyle data

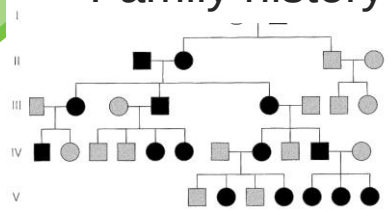
DNA and RNA Analyses

Reference database



Clinical measurements & imaging

Family history



Microbiome

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Thank you! Kiitos! Tack! תודה! Danke!