

Estonian Biobank

Data, trust, and the future of prosperity: lessons from genetics

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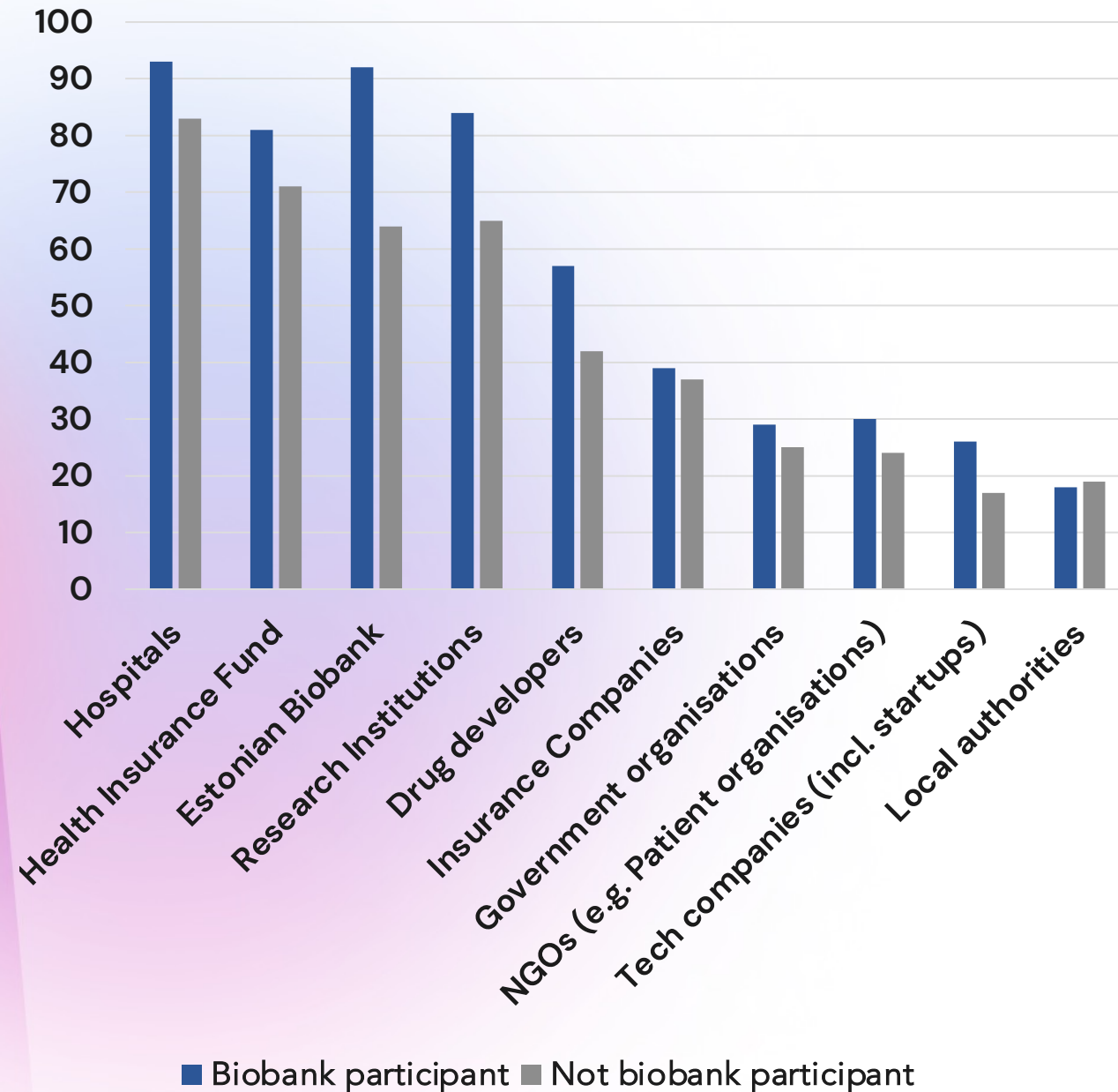
@lilimilani.bsky.social

Which organisations would you be willing to share your health and genetic data with?

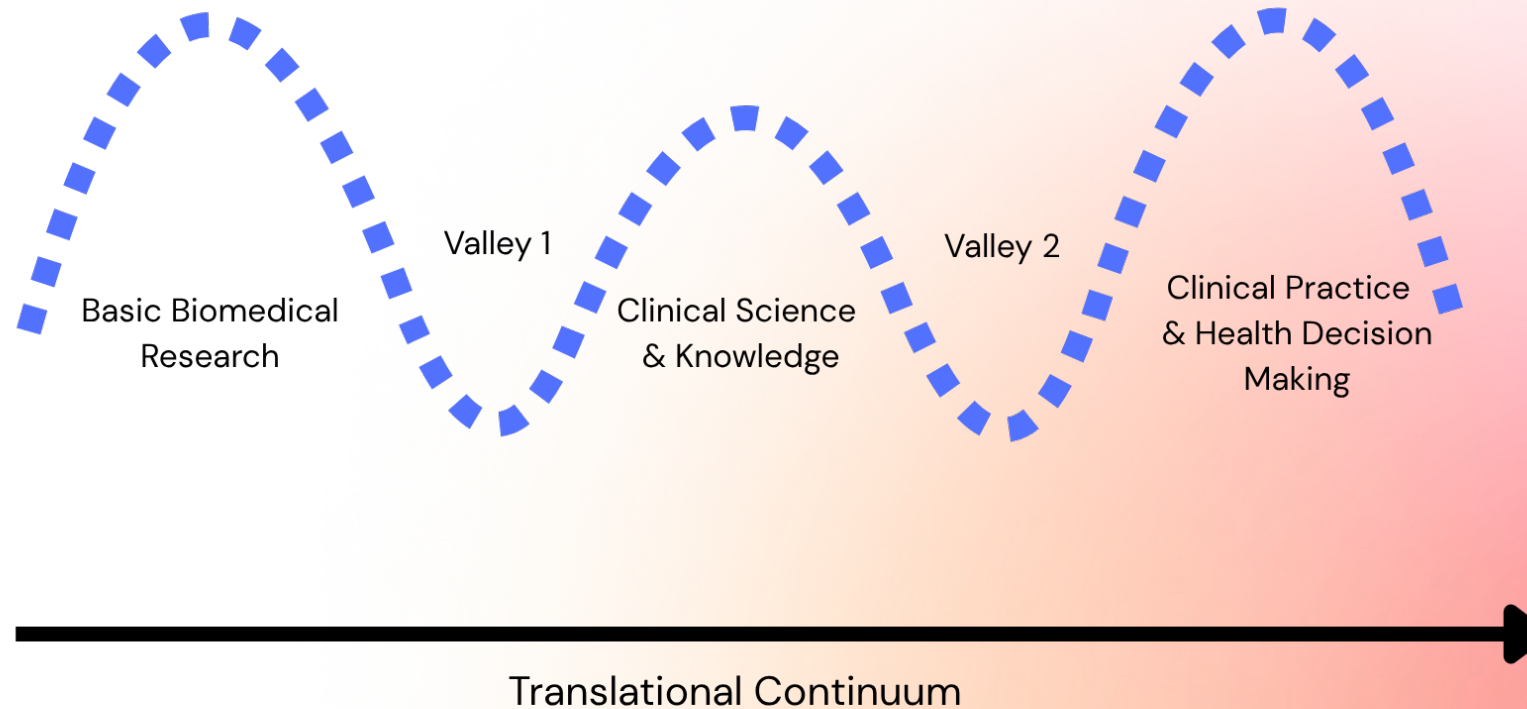
Piret Hirv et al. 2025

Survey of 1466 individuals (31% Biobank participants)

Funded by Enterprise Estonia



Clinical and Translational Research: Valleys of Death



Estonian Biobank

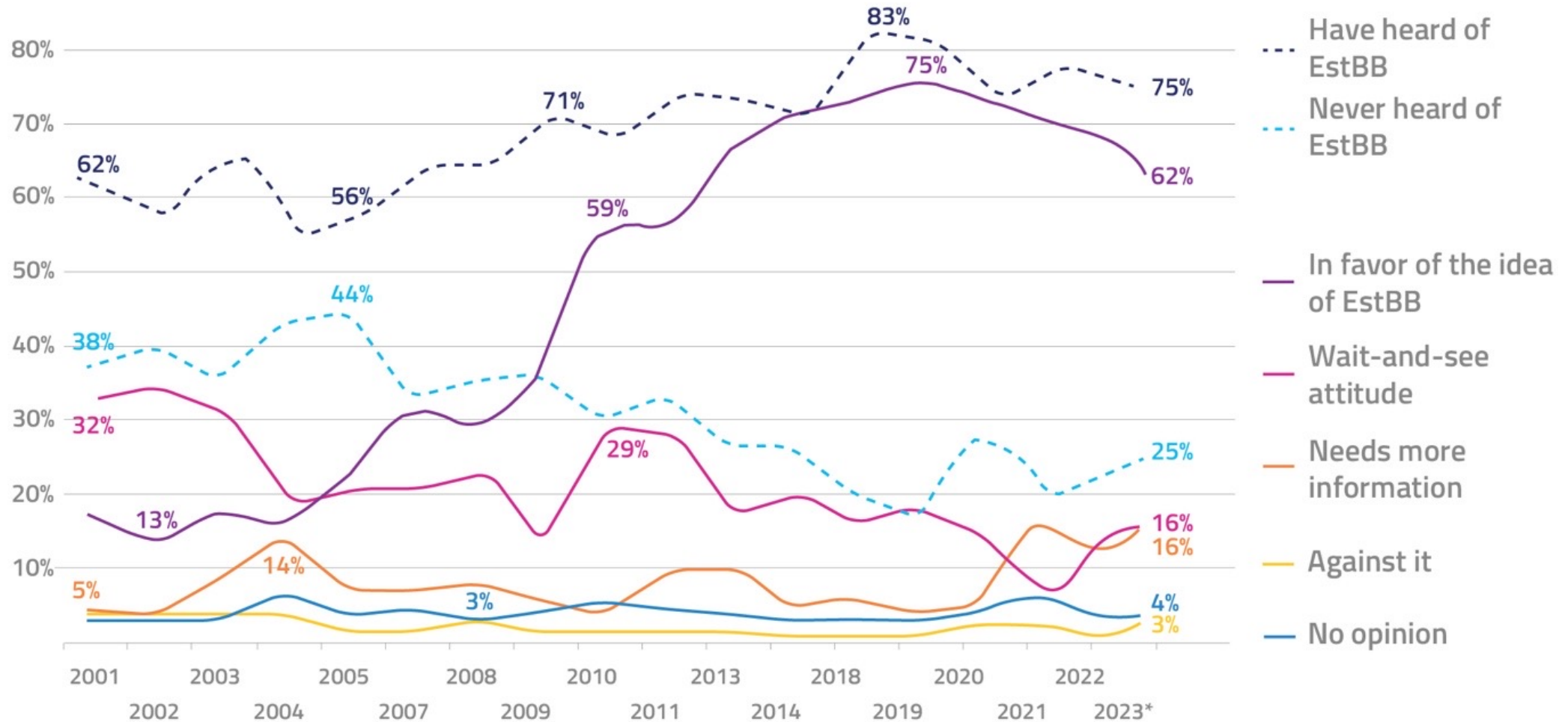
- 212,000 participants (20% of adult population)
- Genotype & metabolomics data for all
- Whole genome sequences for 5,000
- Electronic health records, questionnaire data
- Recall studies based on genetic findings



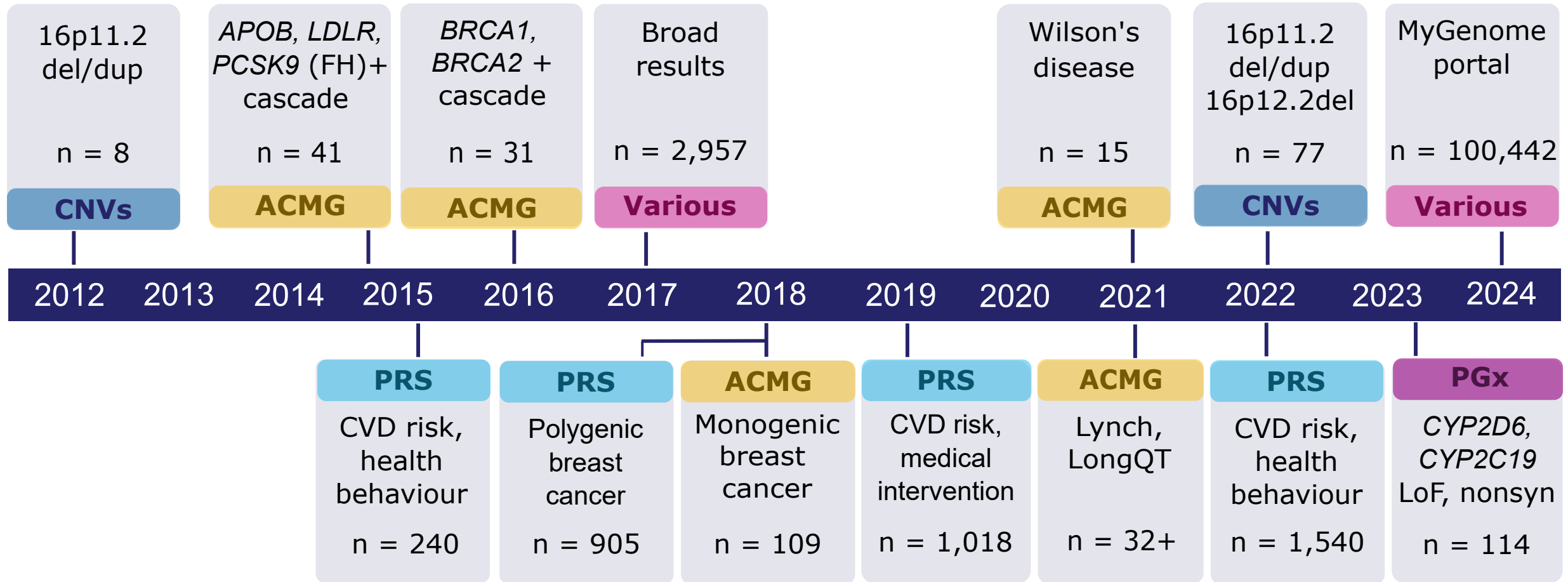
Review Article | [Open access](#) | Published: 05 April 2025

The Estonian Biobank's journey from biobanking to personalized medicine

Population survey (n=1,000): Public Awareness and Opinion



Return of Results to Biobank Participants



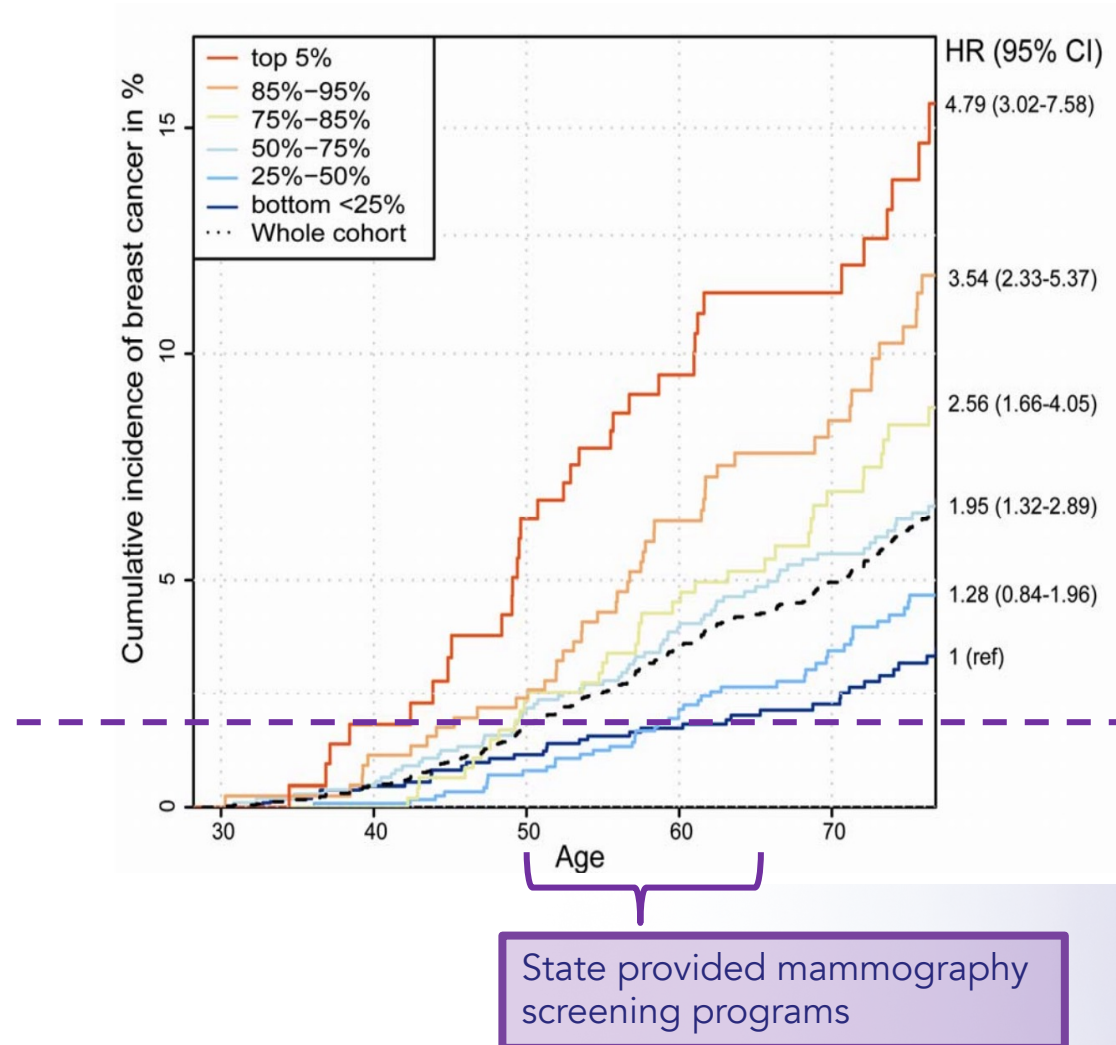
Clinically actionable genetic findings

- **Familial hypercholesterolemia: 1 in 180**
Alver et al., 2019
- **Hereditary breast and ovarian cancer (BRCA1/2): 1 in 125**
Leitsalu et al., 2020
- **Lynch syndrome: 1 in 800**
Roht et al., 2024
- **Wilson's disease: 1 in 20,000**
Nurm et al., 2024
- **Lysosomal storage disease: 1 in 20,000**
unpublished

Polygenic prediction of breast cancer: comparison of genetic predictors and implications for screening

Kristi Läll, Maarja Lepamets, Marili Palover, Tõnu Esko, Andres Metspalu, Neeme Tõnisson, Peeter Padrik, Reedik Mägi, Krista Fischer

doi: <https://doi.org/10.1101/448597>



Estonian Biobank MyGenome portal

An ethical and legally compliant solution to fulfill the responsibilities of the Estonian biobank to its participants:

1. **Return of results:** responsible and ethical return of research findings to biobank participants based on dynamic consent.
2. **Data transparency:** effortless access and overview of the data stored and processed within the biobank.
3. **Scientific study platform:** robust and user-friendly platform for conducting scientific research studies, enabling streamlined collaboration between biobank participants and researchers.

Geenidoonorite portaal



- Saa teaduspõhist personaalset geeniinfot
- Värskenda teadmisi geneetikast
- Osale uuringutes ja panusta teadusesse

Logi sisse

Content available at release

1. Personalized reports across five domains:

- Cumulative risk for type II diabetes
- Cumulative risk for coronary artery disease
- Pharmacogenetics reports
- Caffeine metabolism profile
- Genetic ancestry reports

2. Research:

- “Evaluating the clarity and impact of provided genetic results”, L. Leitsalu et al.

3. Educational resources on genetics

My results



Predispositions



The influence of genes on the effects of drugs



Origin and relatives



Discover what science says about your genetic uniqueness and origins.

What kinds of health concerns and medications is your body more sensitive to? How many genetic relatives do you have in different regions of Estonia?



Scientific research

Which studies can you participate in?

[look closer >](#)



What are genes

What is the role of genes?

[look closer >](#)



Check out the gene lab

How are your genes studied?

[look closer >](#)



My contribution to science

What is the role of gene donors in personalized medicine?

[look closer >](#)

Data transparency

1. Participants can view the **source** of their current data.
2. Participants are encouraged to **update their health and life-style data** to improve risk prediction accuracy.
3. **Data integration:** updated information is securely imported into the biobank database with participant consent.

The screenshot displays a user interface for viewing health data. At the top, the title 'Sinu terviseandmed' (Your health data) is followed by a toggle switch labeled 'Näita, kust minu andmed pärinevad' (Show where my data comes from), which is currently turned on. Below this, several data cards are shown, each with a metric name, a value, and a unit. Each card also includes a link to 'Sinu varasemalt esitatud andmed' (Your previously submitted data) with a date. The metrics include weight (59 KG), height (170 CM), pulse (67 CM), systolic blood pressure (123 MM/HG), diastolic blood pressure (79 MM/HG), total cholesterol (4.1 MMOL/L), and HDL cholesterol (2.1 MMOL/L). At the bottom, there is a section titled 'Kas suitsetad?' (Do you smoke?) with radio button options for 'Jah' (Yes), 'Ei' (No), and 'Olen varem suitsetanud, kuid praegu ei suitseta' (I have smoked before, but I do not smoke now). The 'Ei' option is selected. A link to 'Sinu varasemalt esitatud andmed' (Your previously submitted data) with a date is also present at the bottom.

Sinu terviseandmed Näita, kust minu andmed pärinevad

KAAL
59 KG
Sinu varasemalt esitatud andmed. 11.10.2023

PIKKUS
170 CM
Sinu varasemalt esitatud andmed. 11.10.2023

VOOÜBERMÖÖT
67 CM
Sinu varasemalt esitatud andmed. 11.10.2023

SÜSTOOLNE VERERÖHK 123 MM/HG / **DIASTOOLNE VERERÖHK** 79 MM/HG
Sinu varasemalt esitatud andmed. 26.01.2024 Sinu varasemalt esitatud andmed. 26.01.2024

ÜLDKOLESTEROOL 4.1 MMOL/L
Sinu varasemalt esitatud andmed. 11.10.2023

HDL KOLESTEROOL 2.1 MMOL/L
Sinu koeproovi analüüsist saadud andmed. 08.10.2021

Kas suitsetad?

☐ Jah

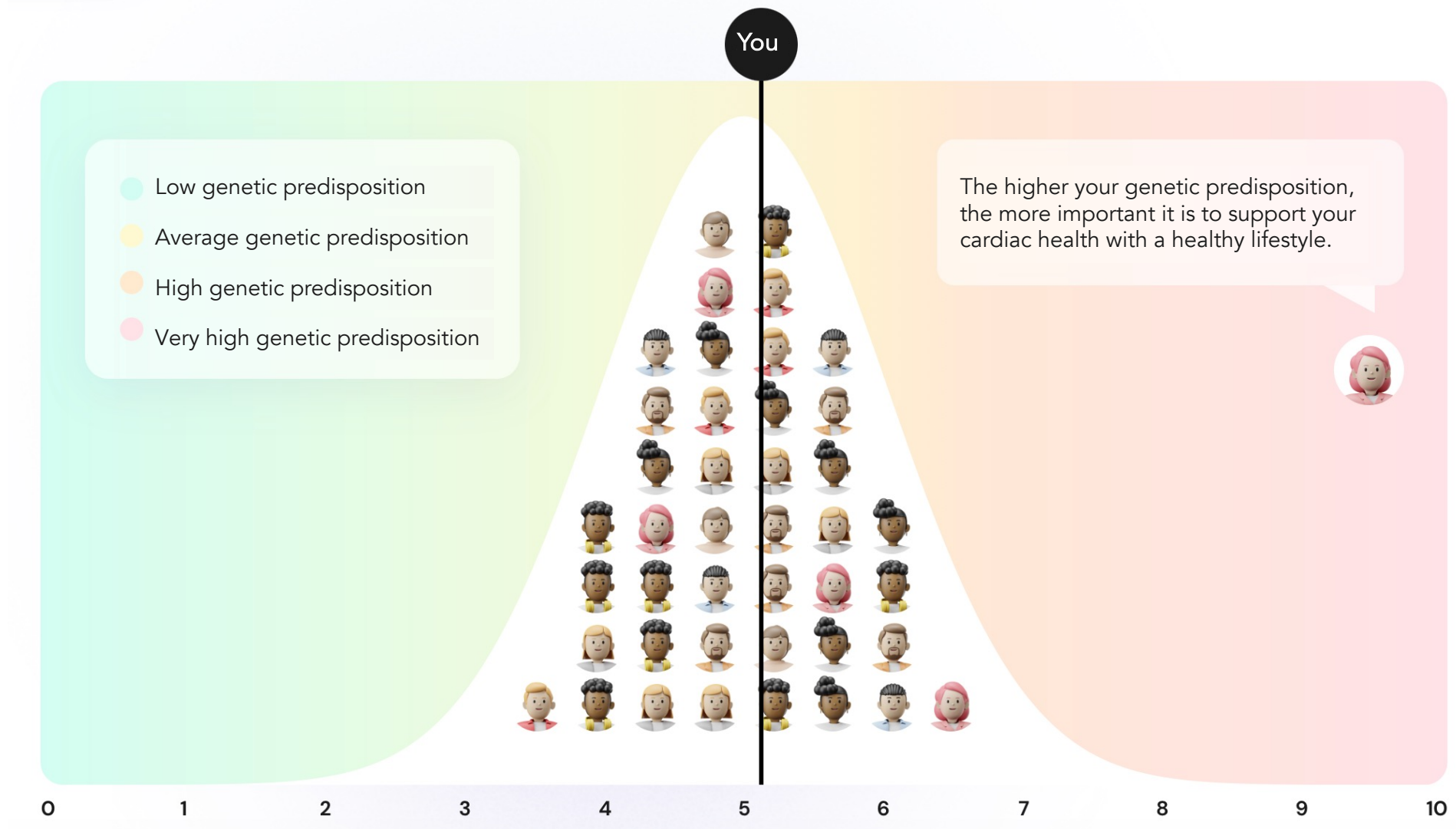
☒ Ei

☐ Olen varem suitsetanud, kuid praegu ei suitseta

Sinu varasemalt esitatud andmed. 11.10.2023



Disease predispositions



See how your lifestyle influences your total risk

Average genetic risk

5,7

Weight

90 (102)

KG

Other health related data

Systolic blood pressure

127

MM/HG

Total cholesterol

4.0 (6.5)

MMOL/L

HDL cholesterol

1.2 (1.1)

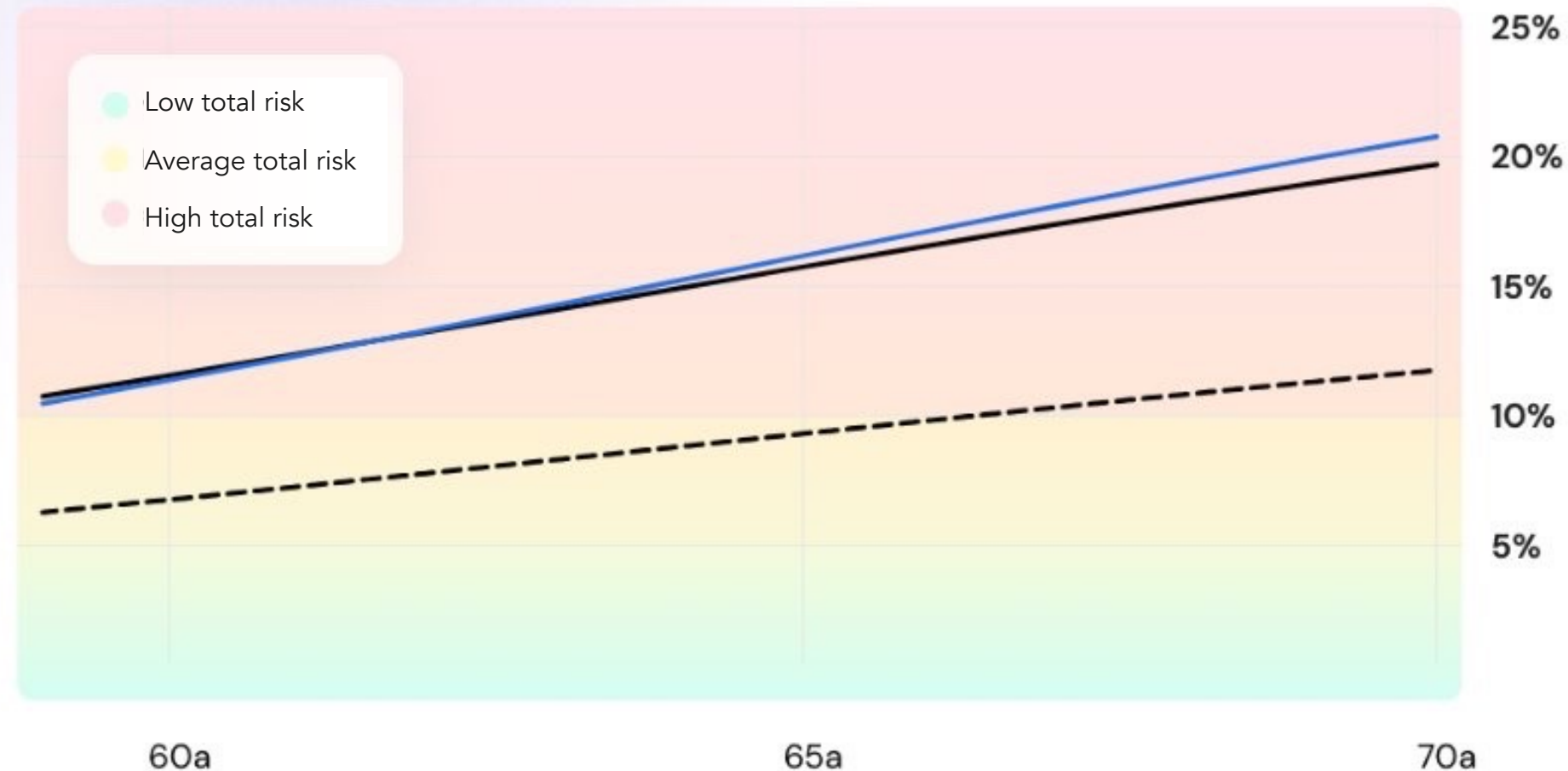
MMOL/L

Smoking

NO

YES

Genetic risk + lifestyle = cumulative risk



- Risk estimate based on your current health and lifestyle
- - - Risk estimate based on your modified health and lifestyle
- Average risk estimate of individuals with your age and gender

Pharmacogenetics

Recommendations for
37 active ingredients =
230 individual drugs

Escitalopram

Antidepressant, selective serotonin reuptake inhibitor (SSRI)

Medicines containing the active compound

Cipralex, Ciraset, Elicea, Elicea Q-Tab, Escitalopram Accord, Escitalopram Actavis, Escitalopram Grindeks, Escitalopram Orion, Escitalopram Teva, Eslorex, Estan

Recommendations in scientific literature

Reduced metabolism of escitalopram when compared to CYP2C19 normal metabolizers. Higher plasma concentrations may increase the probability of side effects. Initiate therapy with recommended starting dose. Consider a slower titration schedule and lower maintenance dose than CYP2C19 normal metabolizers.

Effect of gene on metabolism of the compound

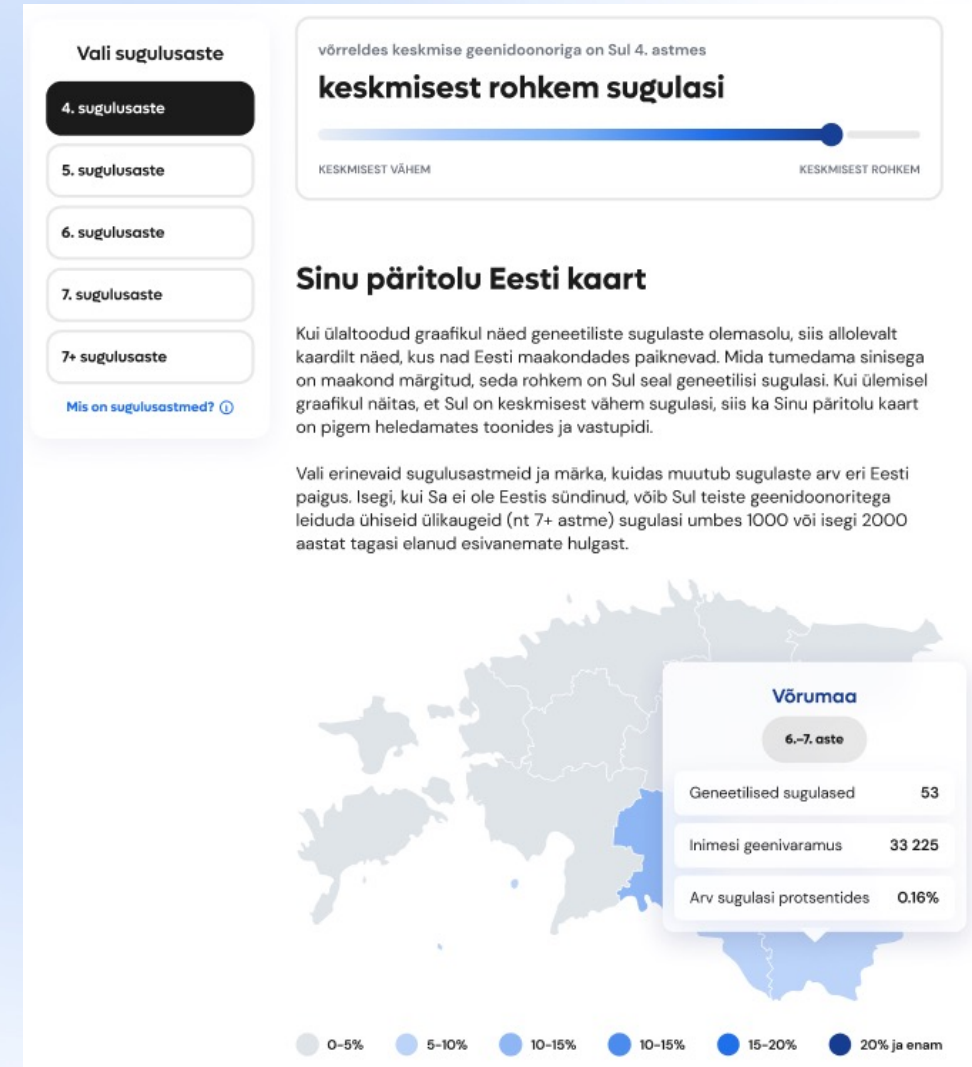
Gene CYP2C19 - Intermediate Metabolizer

Bousman CA, Stevenson JM, Ramsey LB, Sangkuhl K, Hicks JK, Strawn JR, Singh AB, Ruaño G, Mueller DJ, Tsermpini EE, Brown JT, Bell GC, Leeder JS, Gaedigk A, Scott SA, Klein TE, Caudle KE, Bishop JR. Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLCO6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clin Pharmacol Ther. 2023 Jul; 114(1):51-68. doi: 10.1002/cpt.2903. Epub 2023 May 30. PMID: 37032427; PMCID: PMC10564324.



Genetic ancestry

1. Global diversity snapshot: global population proportions
2. Estonian kinship connection: from 4th degree relatives and further
3. Proportion of Neanderthal DNA





Questionnaires

Survey of the comprehensiveness and effect of each report



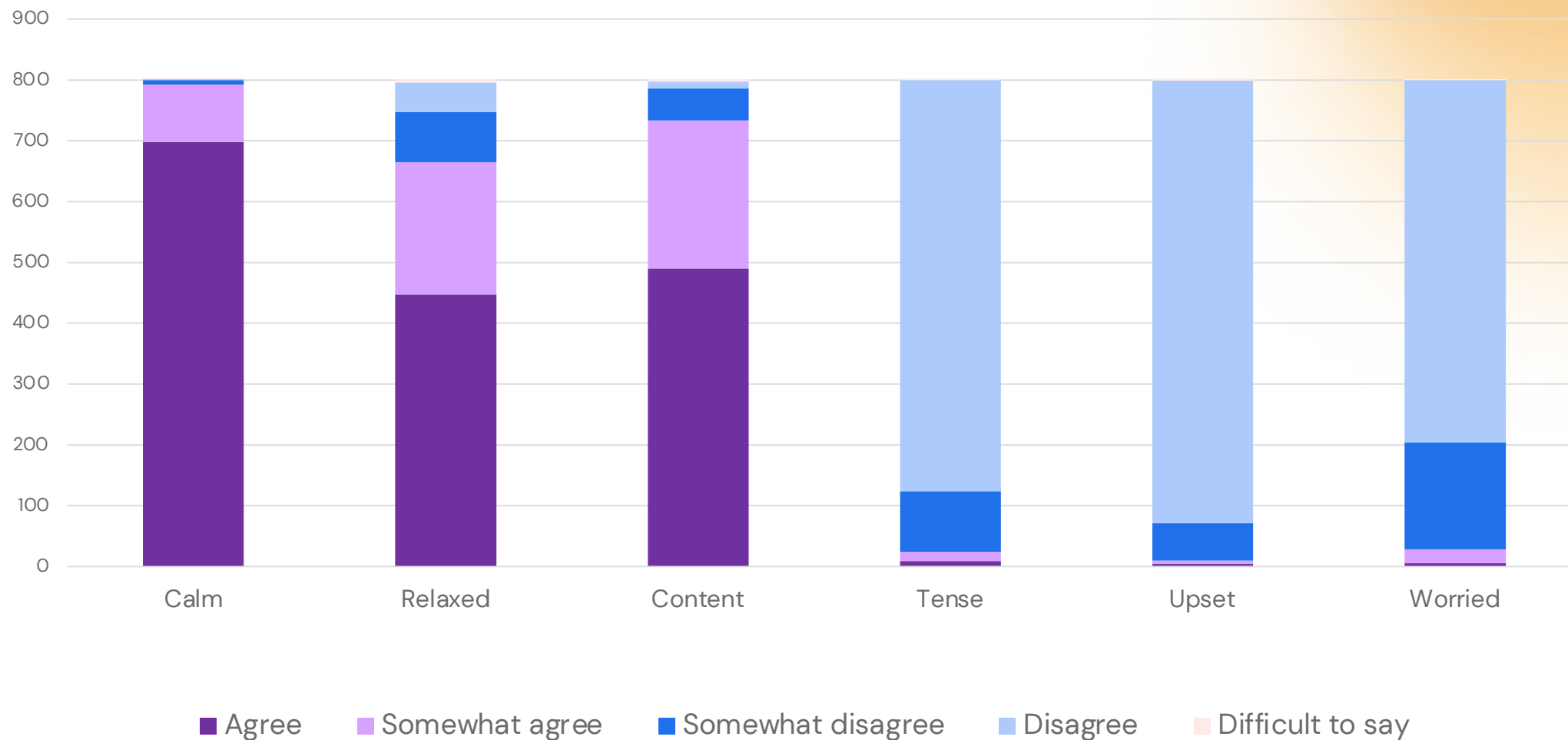
1. The information was **valuable** for me.

- ☒ I agree
- ☐ I rather agree
- ☐ I rather do not agree
- ☐ I do not agree
- ☐ I do not know
- ☐ I do not wish to say

2. The information was **difficult to understand**.

- ☐ I agree
- ☐ I rather agree
- ☐ I rather do not agree
- ☒ I do not agree
- ☐ I do not know
- ☐ I do not wish to say

Reported feelings after visiting portal



[nature](#) > [news](#) > articleNEWS | 26 June 2024 | Clarification [27 June 2024](#)

Estonians gave their DNA to science – now they're learning their genetic secrets

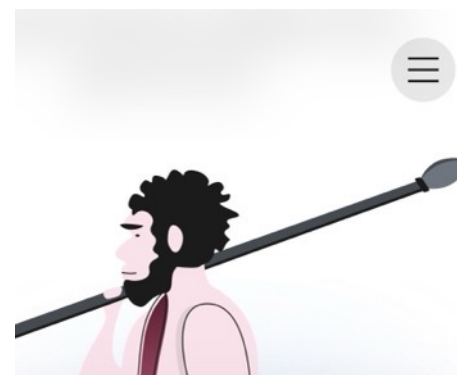
Project covering one-fifth of the country's population is one of the largest-ever efforts to share results on genetic health risks with research participants.

By [Ewen Callaway](#)**Annika Gagarinova** @annikakuld... · 12h

Ma teadsin, et ma olen seto juurtega Ida-Euroopa tšurka ❤️ Tänu, **Geenivaramu**.



11 1 60 7.7K



Neandertallaste osakaal Sinu genoomis:

1,71 %

Keskmiselt on geenidoonoritel neandertallaste osakaal genoomis 2%, kusjuures kõigi geenidoonorite seas varieerub see 0–3,2% vahel.

[Loen edasi ▾](#)**annemariorgla** 13h

Nagu moodne inimene ikka (tuleb täna threadsist välja) vaatasin enda geenivaramu andmeid. Tuleb välja, et minu rohke kofeiinitarbimine on selgelt põhjendatav selle kiire töötlemisega 🤪

Sinu geneetiline eelsoodumus kofeiini lagundamiseks on kiire



Sinu **CYP1A2 geen** toodab kofeiini keskmise kiirusega lagundavat ensüümi



Sinu **AHR geen** soodustab selle ensüümi keskmisest suuremas koguses tootmist

♡ 2 ↻ 🔍

**kaupo.kalda** 12h

Mul ka "kiire". Joon 3–4 tassi päevas, viimase nii kella 14 paiku. Siis rohkem ei taha.

♡ ↻ 🔍

Teams involved

Project Coordination

Legal & Ethical Oversight

Legal Counseling
Ethics

Technical Development

- Software Development
- Database Management
- Data Analysis
- Cybersecurity

Scientific & Clinical Expertise

Genetic Counseling
Laboratory Science
Bioinformatics
Biostatistics
Statistical Genetics
Medical Genomics
Pharmacogenomics
Evolutionary Genomics
Behavioral Science

User Experience & Communication

- Communication
- Graphic Design
- User Interface/User Experience (UI/UX) Design





Estonian Biobank

2nd most Googled question in
Estonia in 2024:

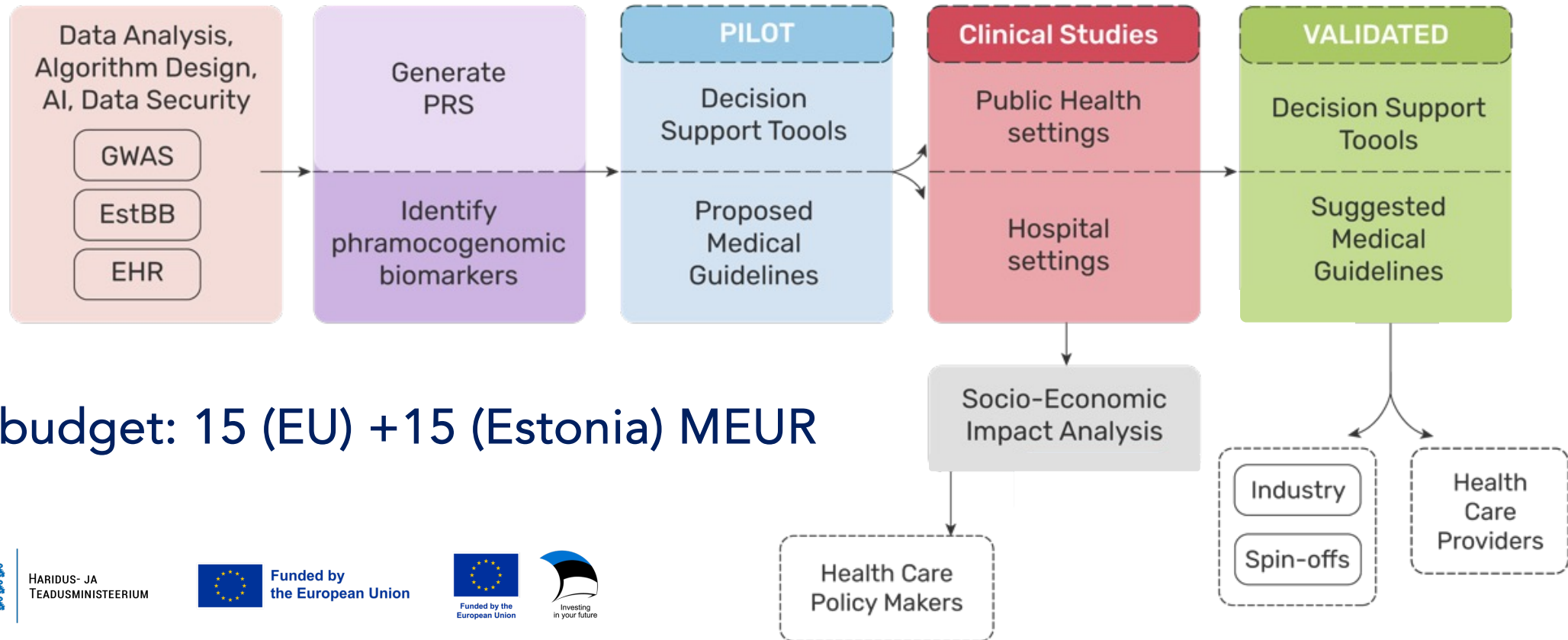
How do I join the biobank?

Only available for those who “trust”...

...how do we scale into the clinic?

TeamPerMed Centre of Personalised Medicine: Bringing partners and stakeholders together

University of Tartu, Tartu University Hospital, Erasmus Medical Center,
University of Helsinki, Erasmus University Rotterdam



Total budget: 15 (EU) +15 (Estonia) MEUR



HARIDUS- JA
TEADUSMINISTEERIUM



Funded by
the European Union



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European Union



Investing
in your future

Creativity & collaboration
>>> prosperity

1

Genetics first

2

Health equity

3

Precision health

Secure data storage and use
>>> trust

Portal team (2019-2025)

- **Project management:** Natalia Pervjakova, Silja-Riin Voolma, Evelin Tali
- **Development team:** Kristjan Metsalu, Priit Kleemann, Kaarel Kaasla, Lars Johannes Sissas, Marielle Lepson, Priit Pärkson
- **User experience and design concept:** Silja-Riin Voolma, Triin Jassov
- **Design:** Mari Potter & outl1ne
- **Writers of clear texts:** Liis Leitsalu, Karoliina Kruusmaa
- **Risk assessment models:** Krista Fischer, Karmel Teder, Tuuli Jürgenson
- **Ancestry reports:** Mait Metspalu, Vasili Pankratov, Georgi Hudjašov
- **Pharmacogenetics reports:** Kristi Krebs, Kadri Maal, Mari Nelis, Viktoria Kukuškina, Jana Lass, Georgi Hudjašov, Lili Milani
- **Caffeine reports:** Laura Birgit Luitva, Silva Kasela, Lili Milani
- **Ethics, legal and medical device expertise:** Kärt Pormeister, Helene Alavere, Tagne Ratassepp
- **Additional contributors:**
Reedik Mägi, Maarja Jõeloo, Liis Karo-Astover, Anu Reigo, Neeme Tõnisson, Kristi Läll, Sirje Lind, Diana Sokurova, Mairo Puusepp, Innar Hallik, Steven Smit, Triinu Temberg, Viljo Soo, Kelli Lehto, Tõnu Esko, Sander Kuusemets, Andres Metspalu, and many others!

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