Estonian Biobank

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

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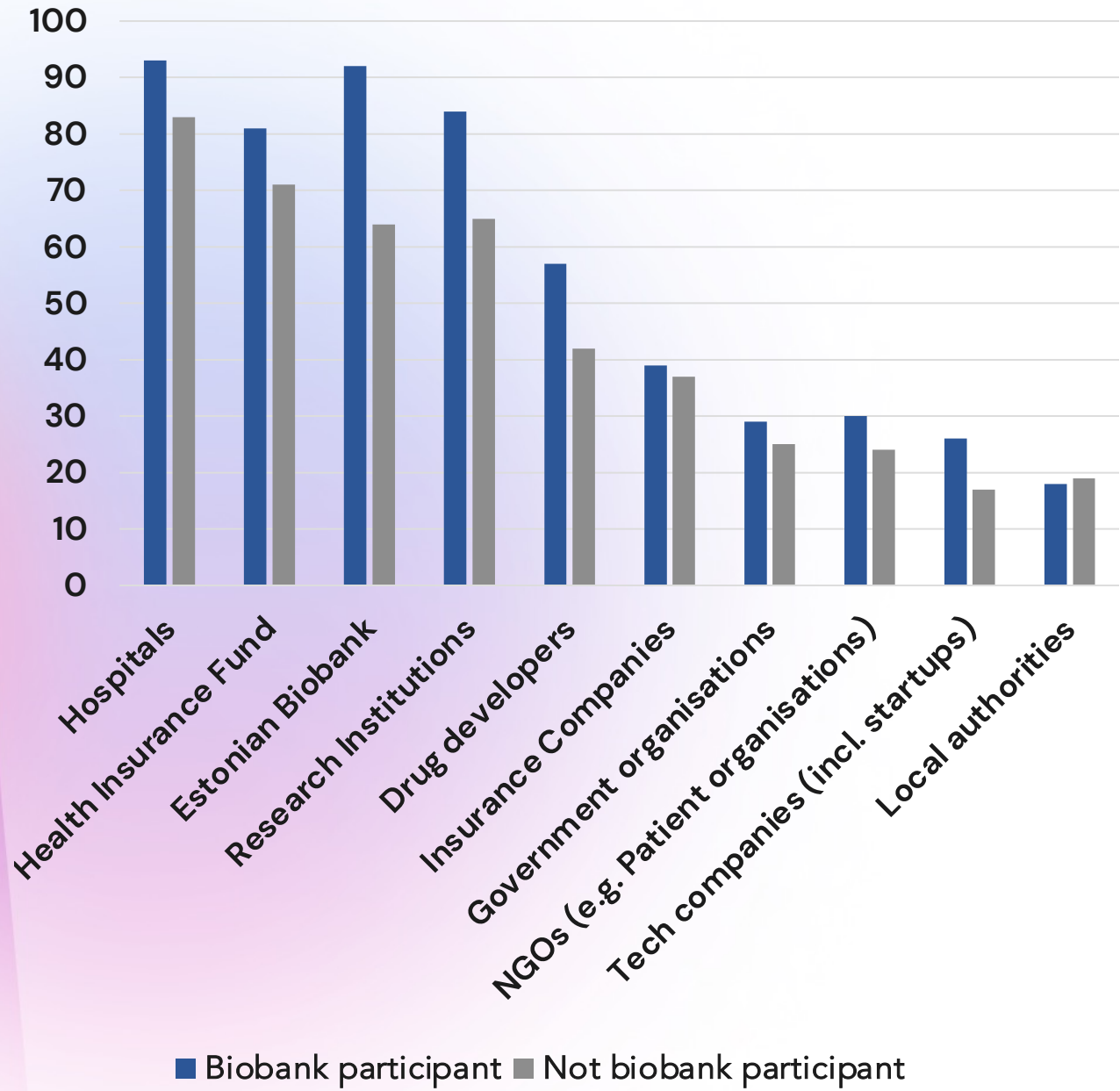
# Which organisations would you be willing to share your health and genetic data with?

Piret Hirv et al. 2025

Survey of 1466 individuals (31% Biobank participants)

Funded by Enterprise Estonia

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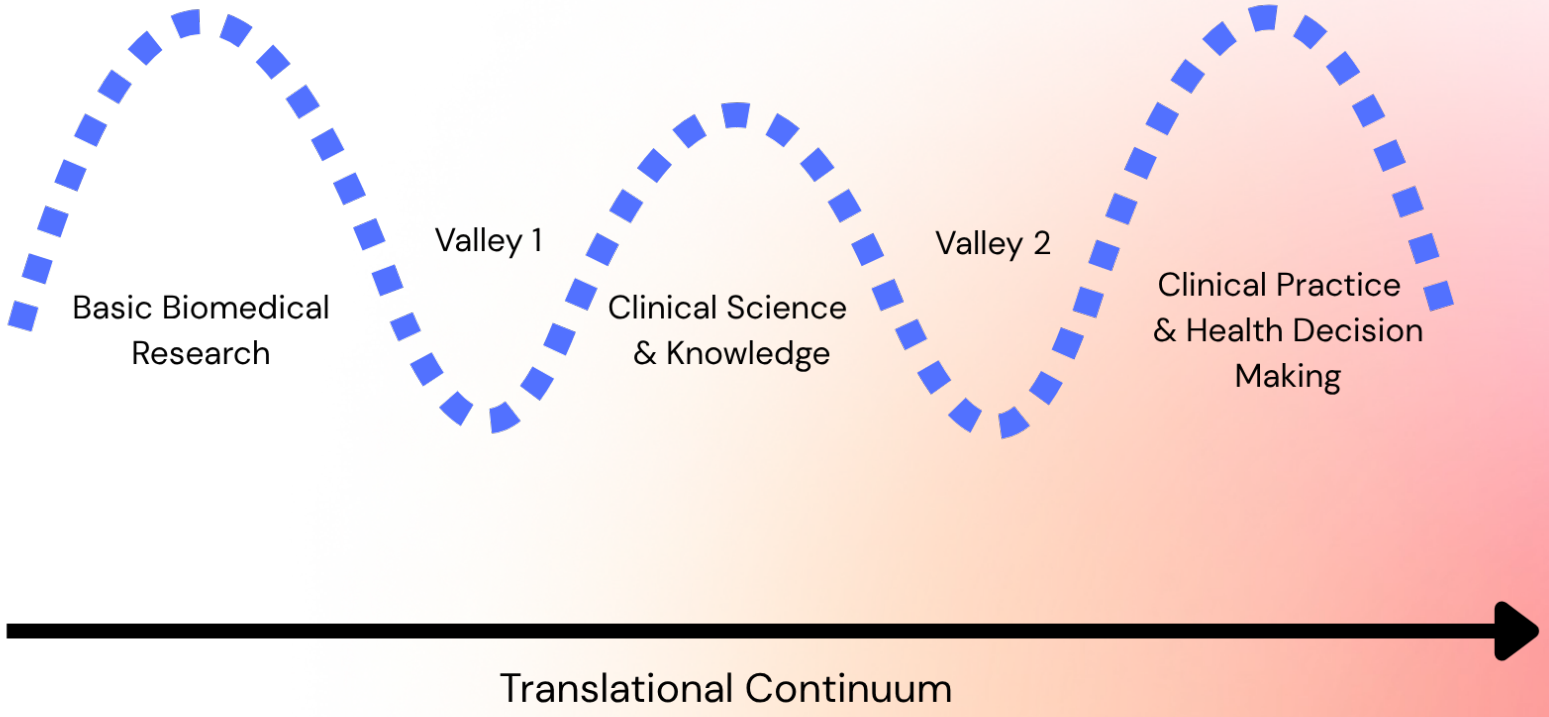
# Clinical and Translational Research: Valleys of Death

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# 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**

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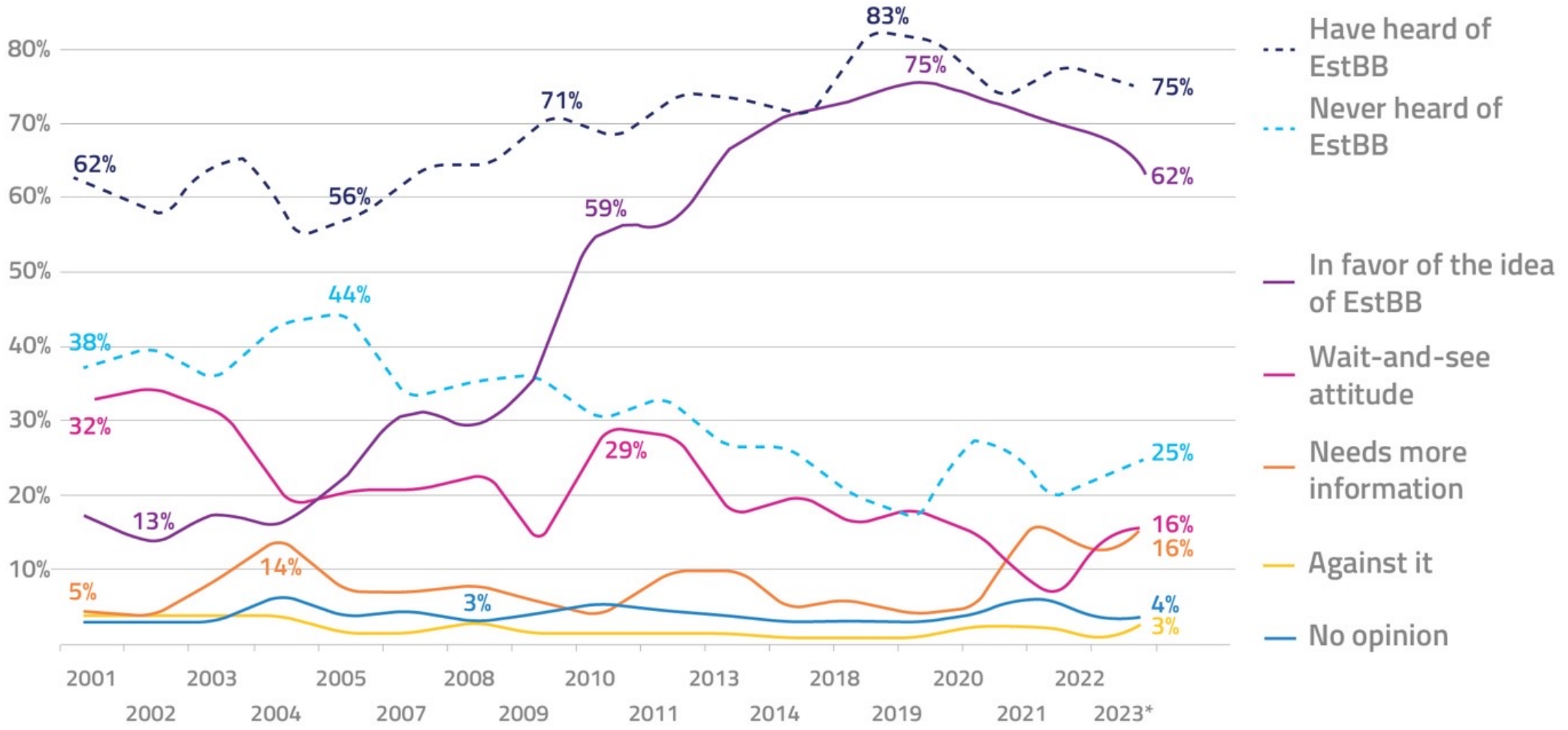
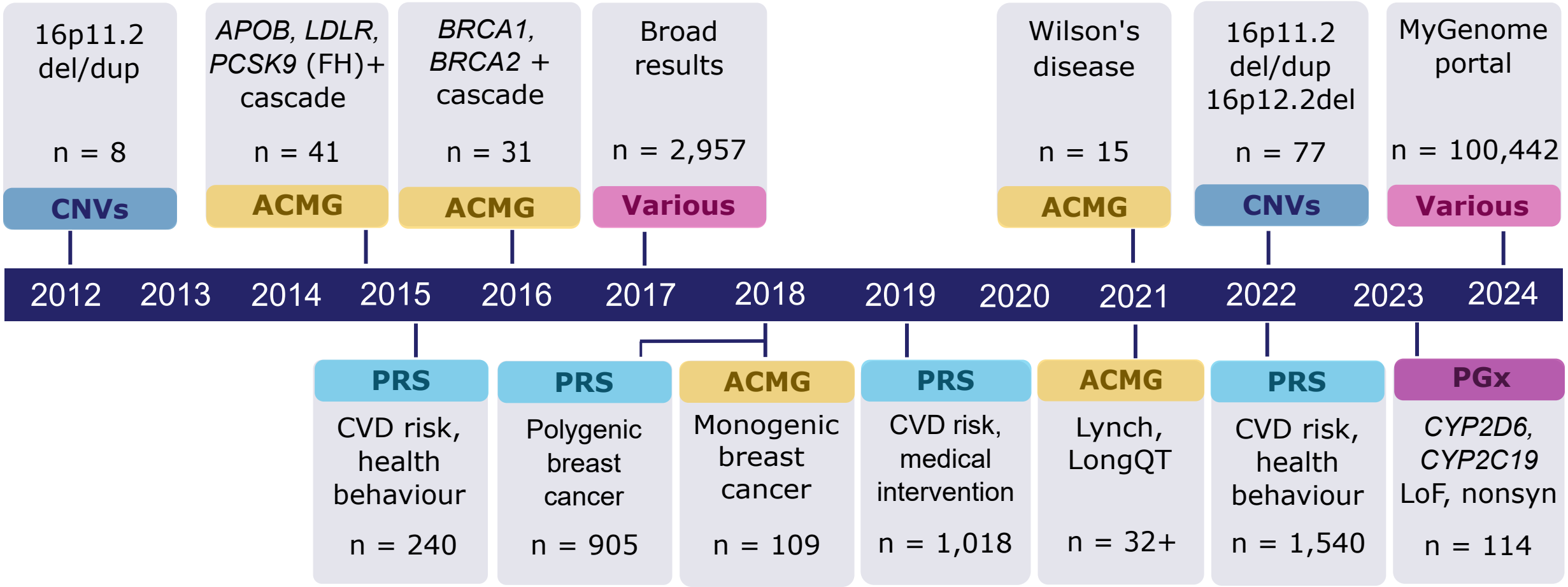
# Population survey (n=1,000): Public Awareness and Opinion



# 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  *unpublished*

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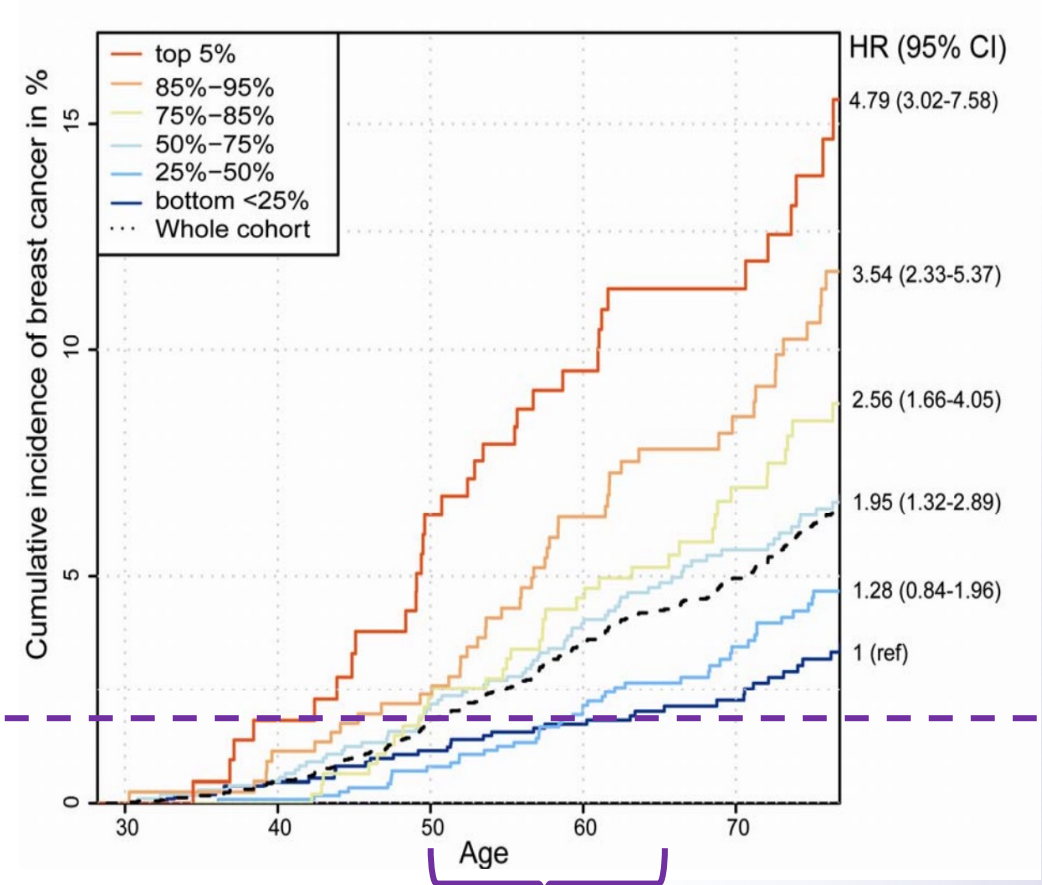
**Polygenic prediction of breast cancer: comparison of genetic predictors and implications for screening**

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




State provided mammography screening programs

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# 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.


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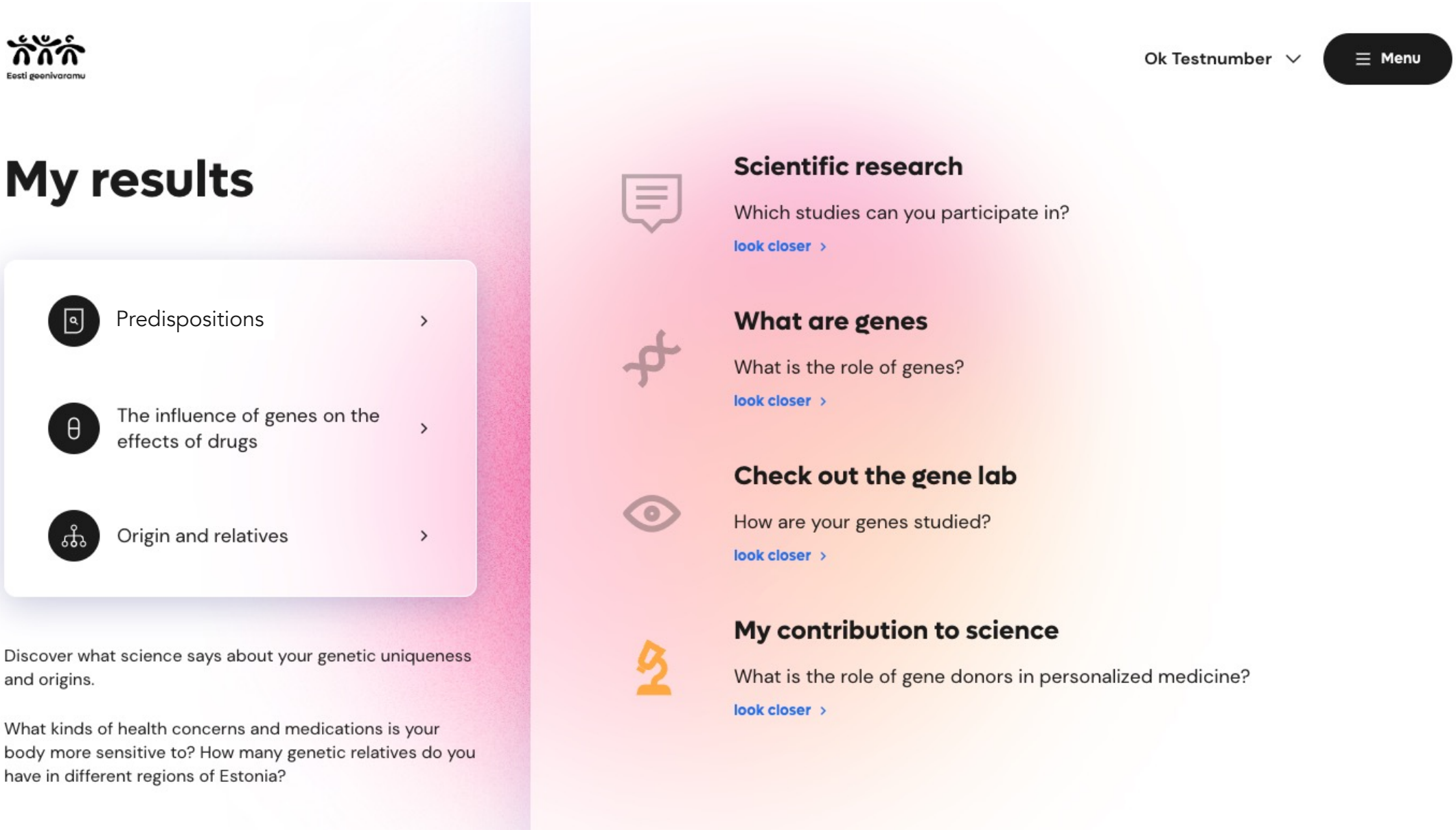
# Content available at release

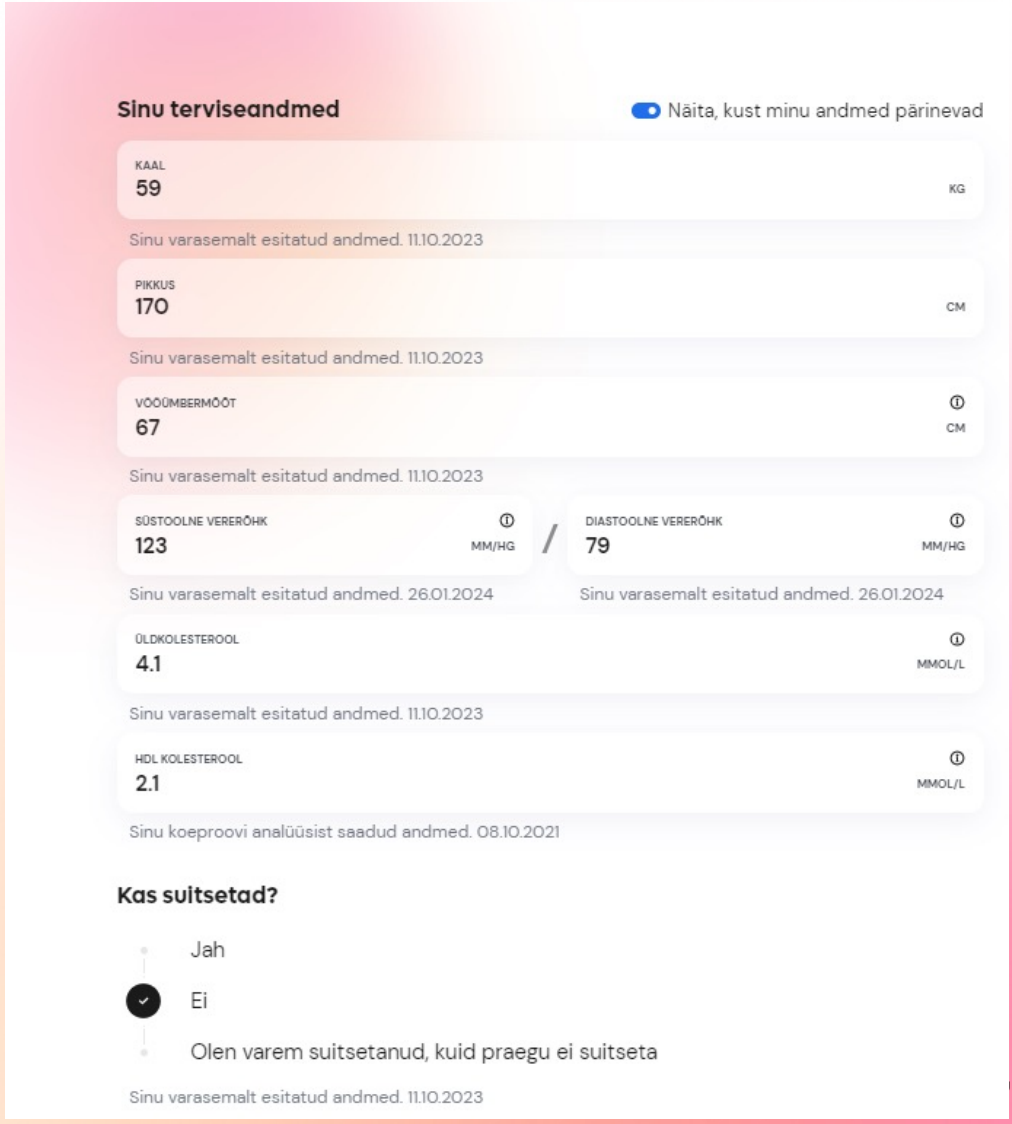
1. Personalized reports across five domains:
    - Cumulative risk for type II diabetes
    - Cumulative risk for coronary artery disease
    - Pharmacogenetics reports
    - Caffeine metabolism profile
    - Genetic ancestry reports
2. Research:
    - “Evaluating the clarity and impact of provided genetic results”, L. Leitsalu et al.
3. Educational resources on genetics



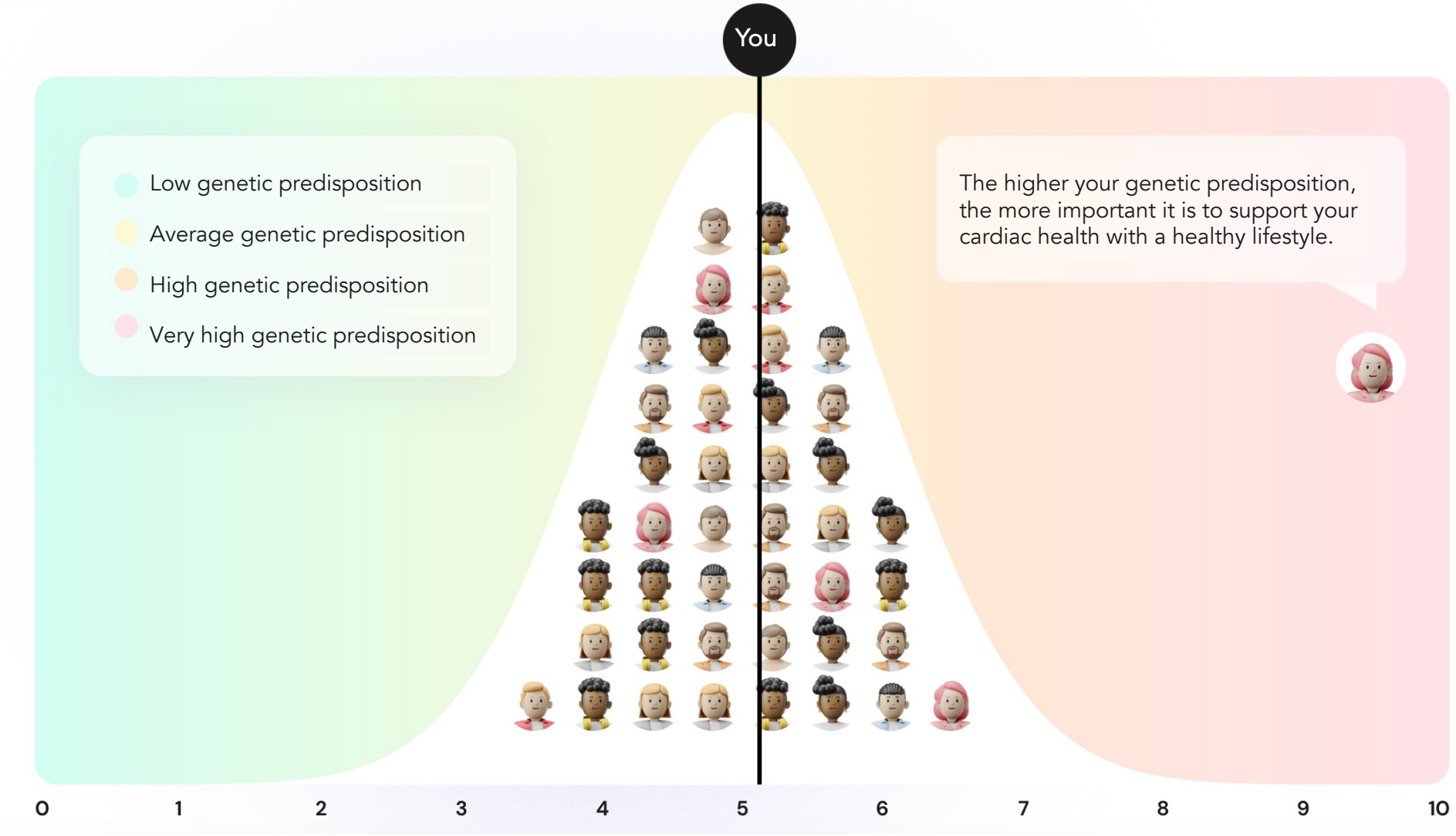

*In-house medical device, in accordance with Article 5(5) of Regulation (EU) 2017/746*

# 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.



# Disease predispositions



See how your lifestyle influences your total risk

Average genetic risk 5,7

Weight
90 (102) KG

Other health related data

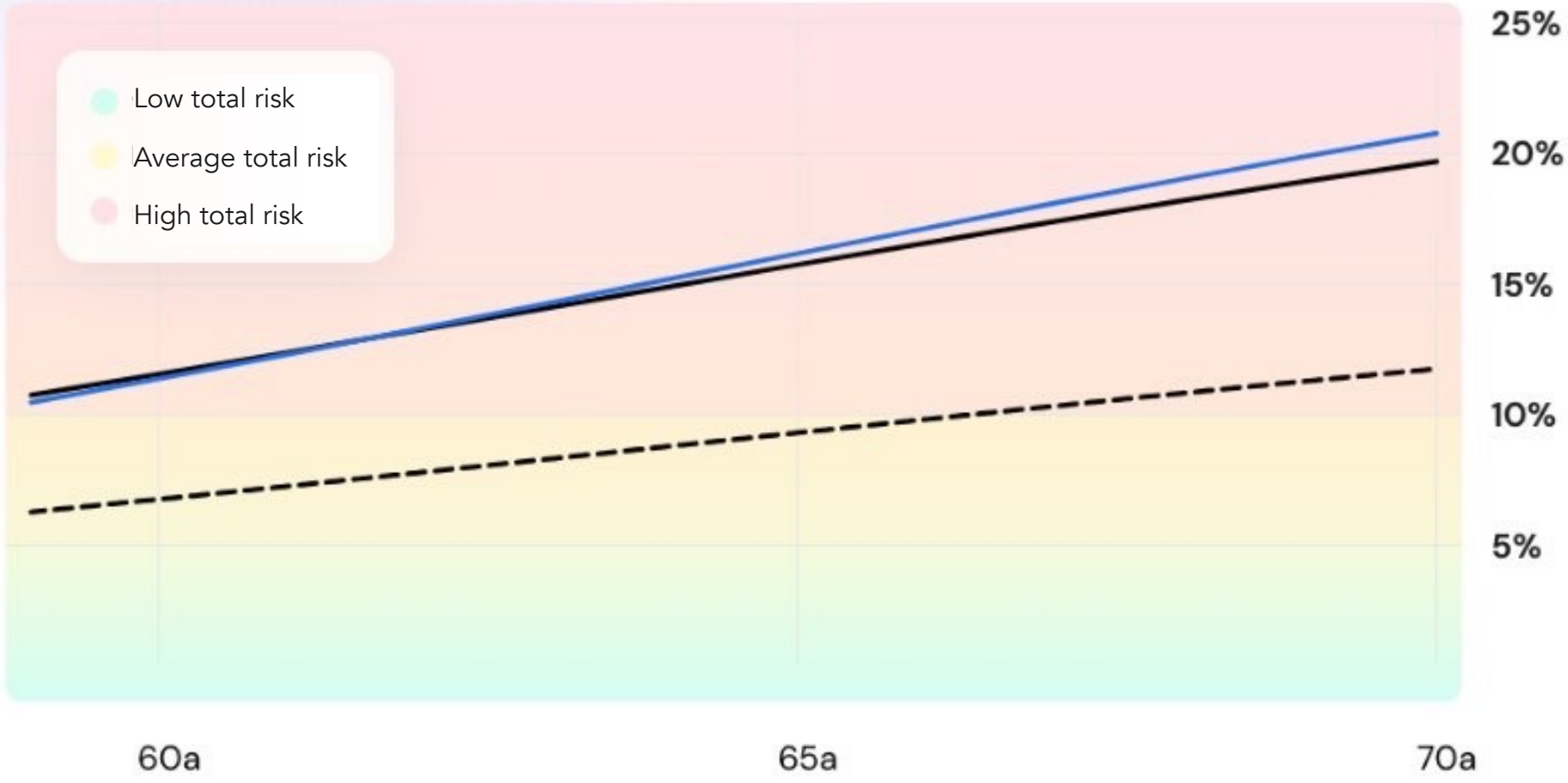
Systolic blood pressure
127 MM/HG

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.

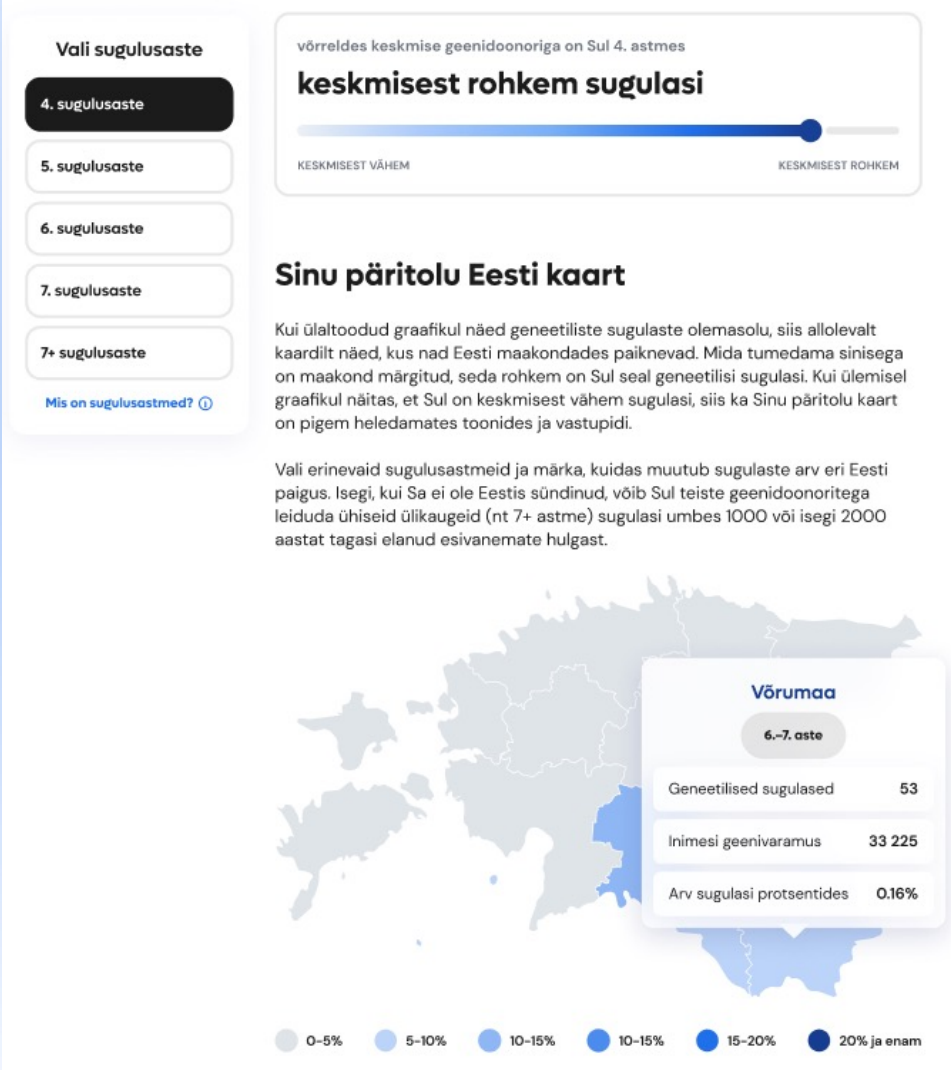
### Effect of gene on metabolism of the compound

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, SLC6A4, 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**

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# 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

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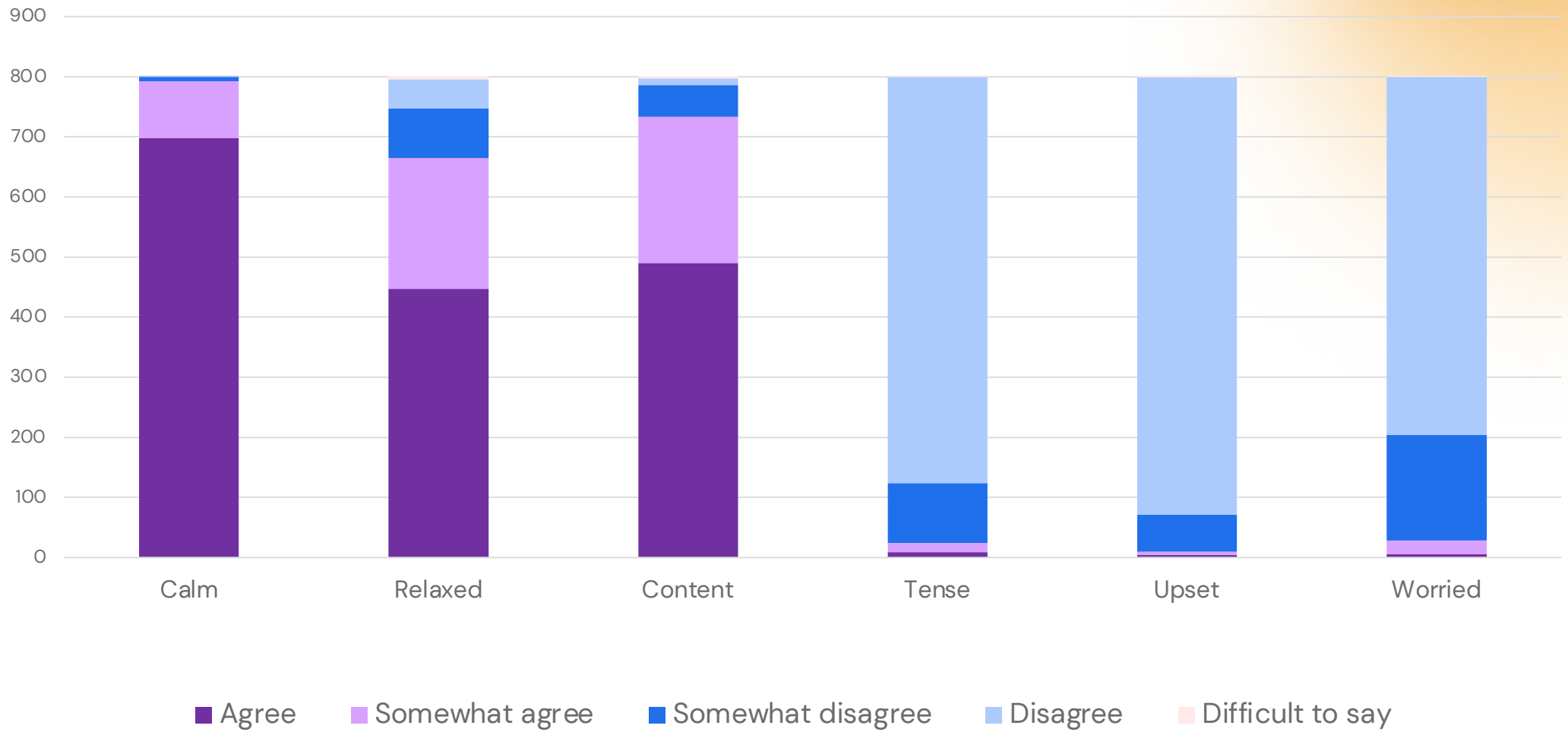
# Reported feelings after visiting portal

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NEWS | 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



# Teams involved

**Project Coordination**

**Technical Development**

- Software Development
- Database Management
- Data Analysis
- Cybersecurity

**User Experience & Communication**

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

**Legal & Ethical Oversight**

Legal Counseling
Ethics

**Scientific & Clinical Expertise**

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

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# 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?

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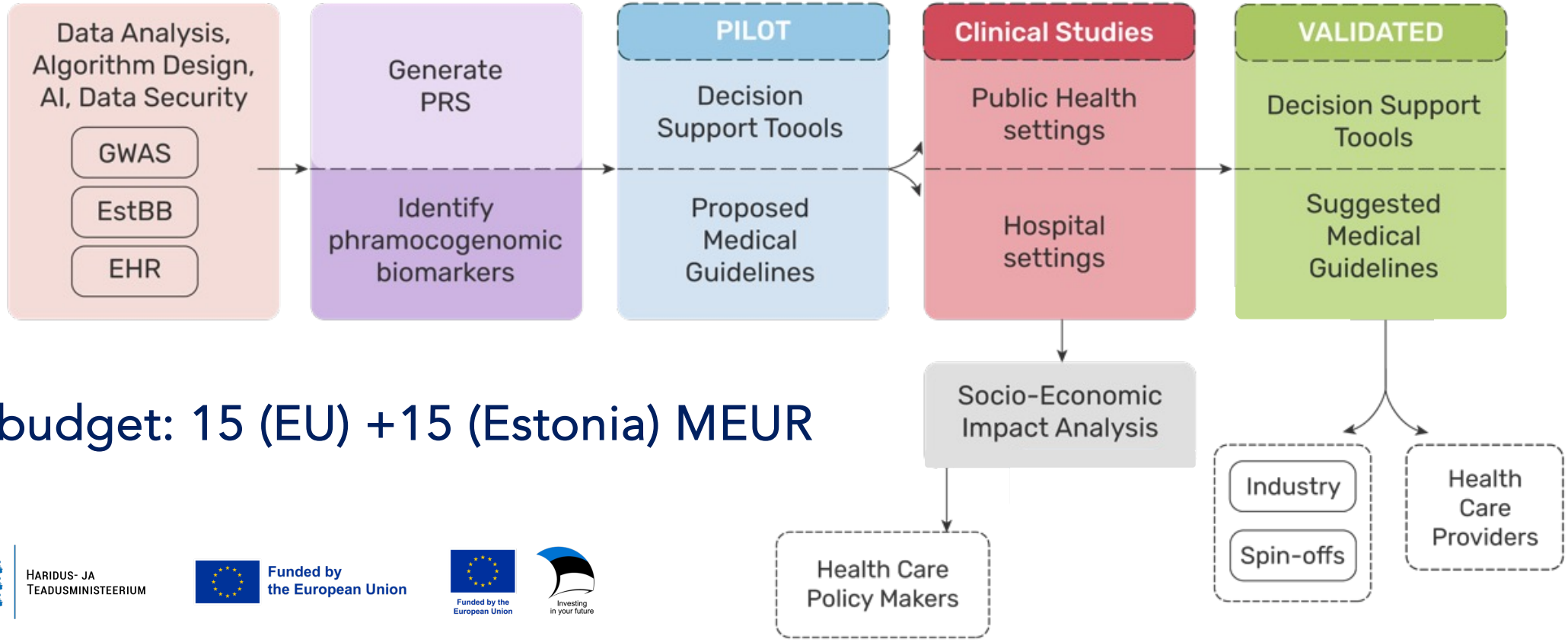
# TeamPerMed Centre of Personalised Medicine: Bringing partners and stakeholders together

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

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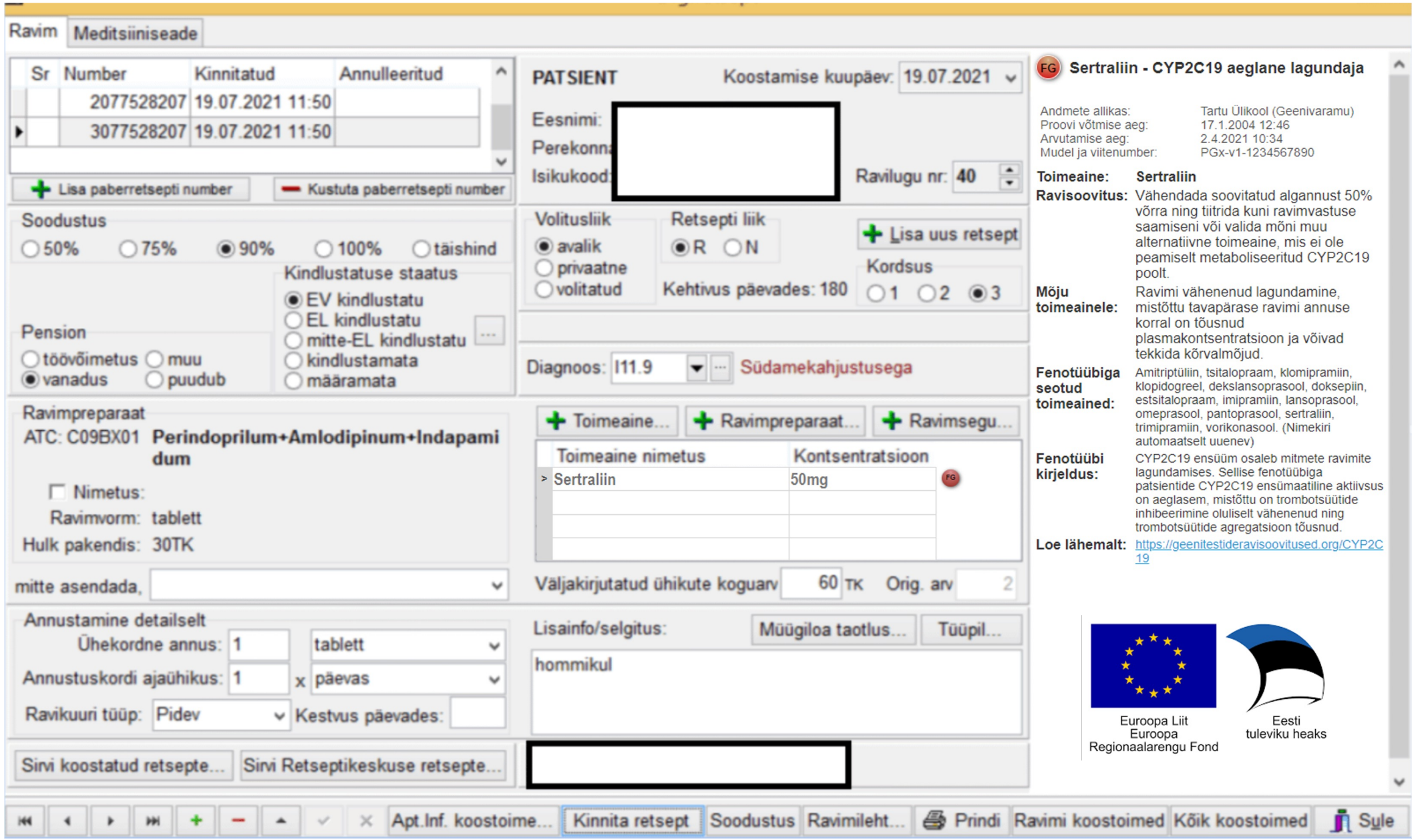
Investing in your future




*By Sulev Reisberg and Marko Arrak*

# Creativity & collaboration >>> prosperity

1 Genetics first

2 Health equity

3 Precision health

# Secure data storage and use >>> trust

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# 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!

*Seed funding from Enterprise Estonia (GreenICT), major funding from the Institute of Genomics.*

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