Biobanks and the Rise of Precision Medicine: Lessons from the Estonian Biobank

GenomeWebinar
August 22, 2019

Ben Butkus
GenomeWeb

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University of Tartu & Estonian Biobank Science Center

Outi Törnwall, PhD
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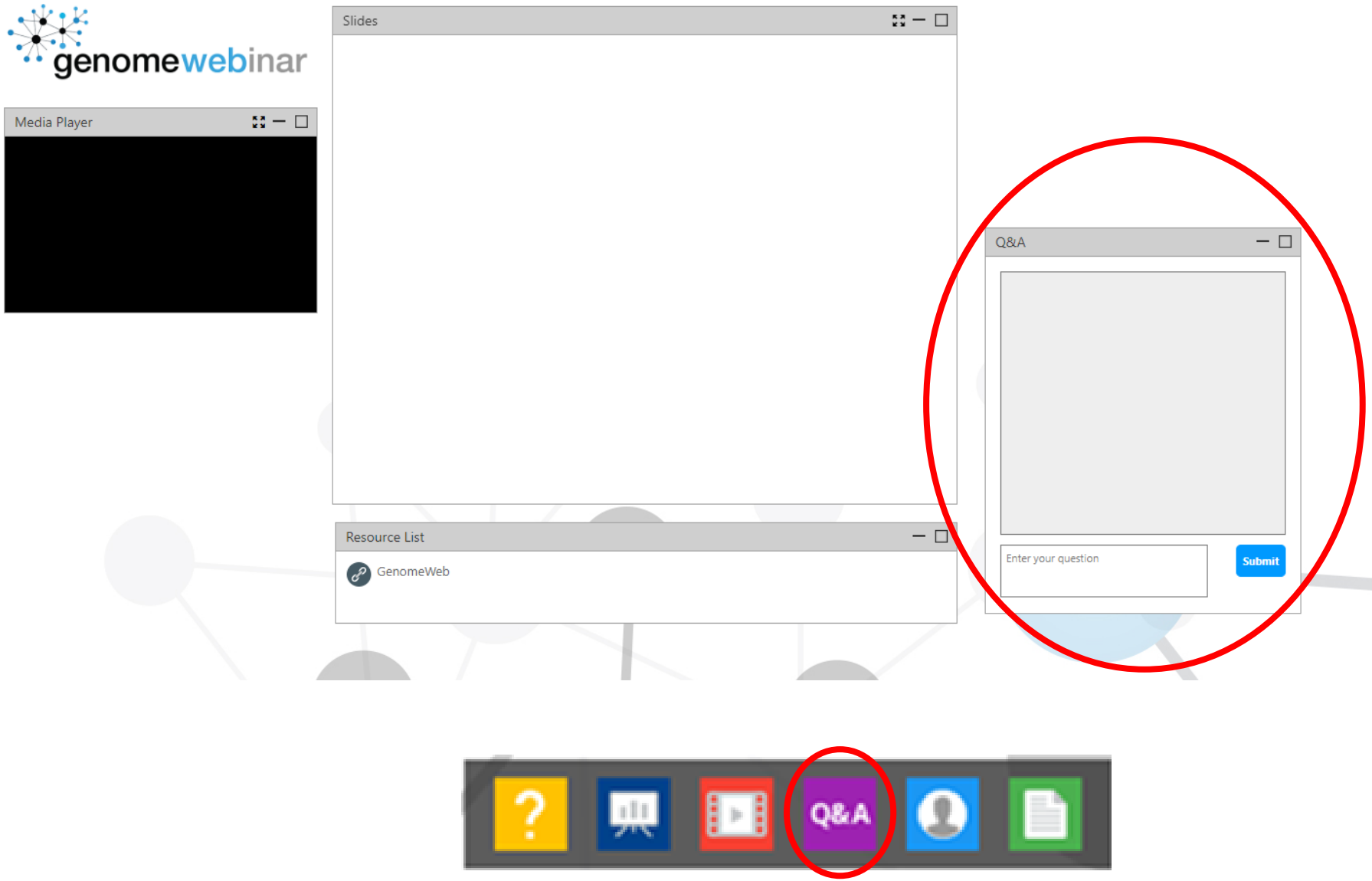
Today's Panelists

Ben Butkus
Editorial Director,
GenomeWeb
(Moderator)

Tõnu Esko, PhD
Vice Director, Institute of Genomics,
University of Tartu;
Head of the Estonian Biobank
Science Center

Outi Törnwall, PhD
Data Partnerships Director,
BC Platforms

Please submit any questions in the Q&A panel



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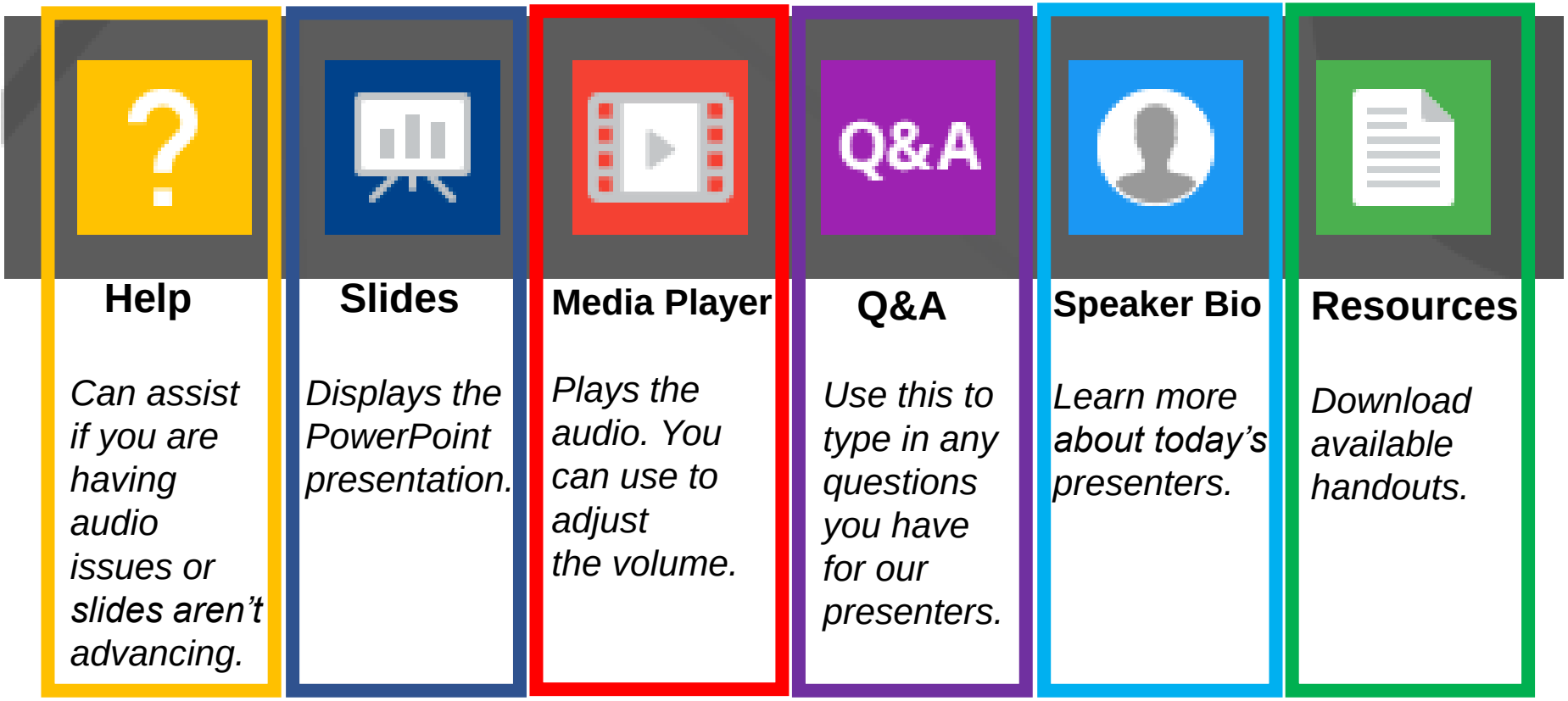

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Q&A

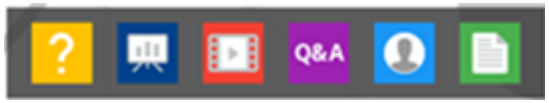
Use this to type in any questions you have for our presenters.

Speaker Bio

Learn more about today's presenters.

Resources

Download available handouts.



Tõnu Esko, PhD

Vice Director, Institute of Genomics,University of Tartu;
Head of the Estonian Biobank Science Center

Unlocking health with Big Data

Tõnu Esko

Vice Director, Institute of Genomics

Head, Estonian Genome Center

Research Scientist, Broad Institute, Boston, USA

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A unique place to fulfill the promises of Big Data:

World leader in public IT-services and ambitious start-up community

ESTONIA

Population: 1.3 Million
Size: 45 227 km²
Capital: Tallinn
Language: Estonian
Member of EU
Currency: Euro
GDP: 17B EUR

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Let's do it! World Cleanup 2015

Estonian Biobank

- Estonian Genome Center, University of Tartu
- A prospective, longitudinal, population-based database with health records and biological materials
- ~200,000 participants - 15% of the adult population
- Individuals are recruited by medical personnel
- Broad informed consent
- Legislation: Estonian Human Genes Research Act

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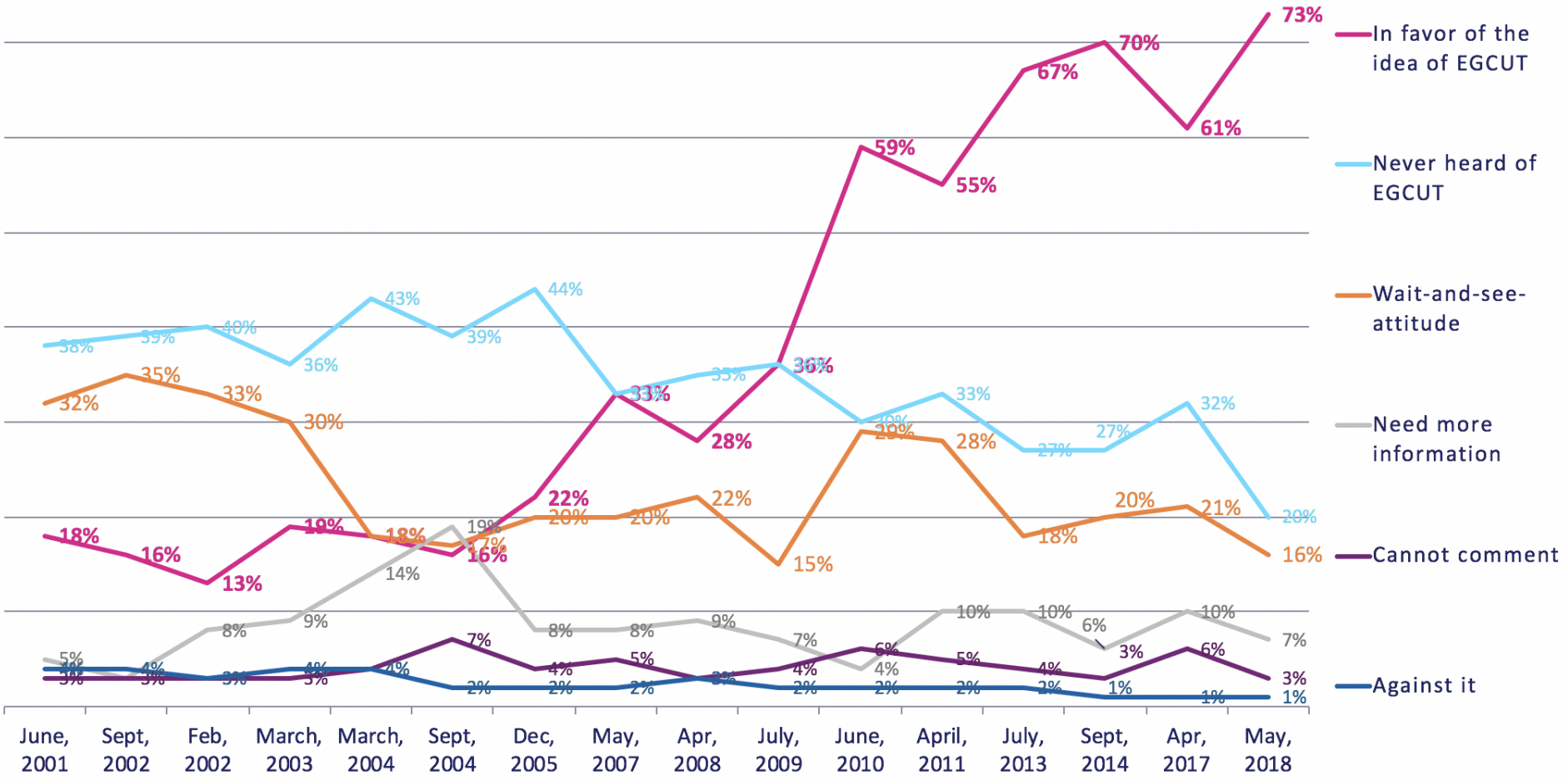
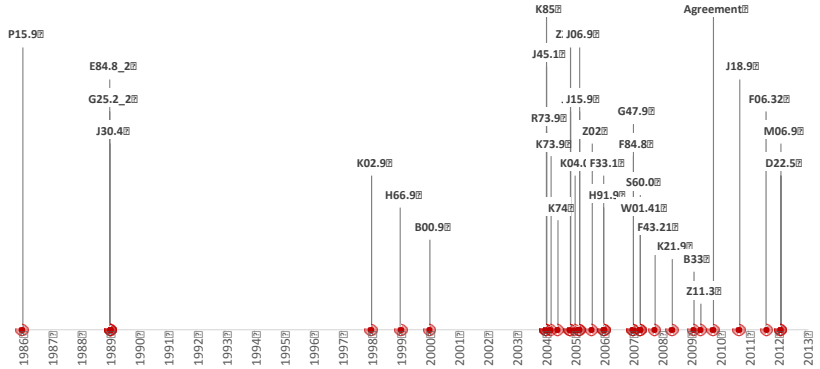
Public opinion and awareness



Research infrastructure

Sample collection (DNA, plasma, WBC)
200,000+ by 2020

Longitudinal Health data – linking with national health registers



Disease trajectories

Linked digital health repositories *(most since 2007)*

- Health insurance (since 2004) – *diagnosis, treatments, procedures (3.5M+ documents)*
- ePrescriptions – *prescribed drugs [3.5M+ documents]*
- Regional hospitals – *diagnosis, treatments, clinical labs [2.5M+ clinical labs]*
- Registries (MI, Cancer, Death etc) – *state registries*
  Nation-wide health records – *doctors notes and all procedures [1M+ documents]*
- Medical image database – *all digital images in central database [linking on project basis]*

CUTTING EDGE IN SEVERAL QUALITATIVE CHARACTERISTICS

	PATIENT RECONTACT	SIMPLE ECOSYSTEM*	CONTINUOUS MEDICAL DATA	CROSS-BORDER DATA TRANSFER	SAMPLE SIZE
EST Biobank	☑	☑	☑	☑	200 000
UK Biobank	☑	☒	☒	☑	500 000
Finngen	☑	☑	☑	☑	279 000
DEN Biobank	☑	☑	☒	☒	450 000
deCODE	☒	☑	☑	☑	120 000
Kadoorie	☒	☒	☒	☒	515 000
GoSHARE	☑	☒	☒	☑	55 000
GAPS	☑	☑		☑	225 000
HUNT	☒	☑		☑	250 000

STRENGTHS

- Cross-border transferrable data
- 20 years of continuous medical data
- Simple decision-making procedure
- Proven patient recontact (see Alver et al 2018)

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Do we have enough information?

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TARTU ÜLIKOOL 1632

eesti geenivaramu

Jaan Tamm

Your Data

Male ○ Female Age 54

Weight 89 Height 171 Waist 86

☐ Hypertension ☐ Myocardial Infarction

Genetic risk of type 2 diabetes

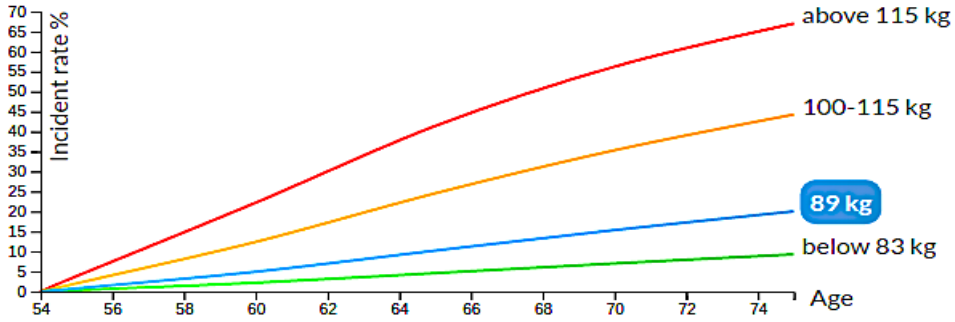
Seven persons out of 10 have lower genetic risk than you.

Two persons out of 10 have higher genetic risk than you.

Your genetic risk of type 2 diabetes is **average**. Your added lifestyle risk is **low**.

Your total risk of type 2 diabetes is **low**.

Risk of type 2 diabetes depends on body weight



Your 10-year risk of developing type 2 diabetes is **2%**. Your probability of developing type 2 diabetes before age 70 is **15%**.

An average person similar to you, but with lower body weight, has up to **50% lower** diabetes risk.

TARTU ÜLIKOOL

eesti geenivaramu

Farmakogeneetika

Üheks põhjuseks, miks inimesed reageerivad ravimitele erinevalt on variatsioonid geenides, mis on vajalikud ravimi toimimiseks organismis. Praeguseks on tuvastatud mitmeid ravimivastuses olulisi geneetilisi markereid. Käesolevas testis määrati Teil 6 geeni markereid, mis mõjutavad 16 erinevat ravimi toimeainet.

Testitud geenivariantide põhjal on võimalik anda alljärgnevad suunised:

Geen	Genotüüp	Hinnang	Soovitus	Mõjutatud ravimi toimeained
CYP2C19	*1/*1	Tavapärane ravimi lagundamine	✚	Estsitalopraam, Tsitalopraam, Klopidogrel, Sertraliin,Vorikonasool,Esomeprasool, Lansoprasool,Pantoprasool, Klomipramiin, Amitriptüliin
CYP2C9	*1/*1	Tavapärane ravimi lagundamine	✚	Fenütoiin
CYP3A5	*3/*3	Aeglane ravimi lagundamine, Tavapärane muster	✚	Takroliimus
DPYD	*5/*9A	Tavapärane ravimi lagundamine	✚	Kapetsitabiin, Fluorouratsiil
CYP2C9&VKORC1	*1/*1 & 1639G>A, genotüüp; GA	Tavapärane doosisoovitus	✚	Varfariin
SLCO1B1	TC	Tavapärasest kõrgem müopaatia risk	!	Simvastatiin

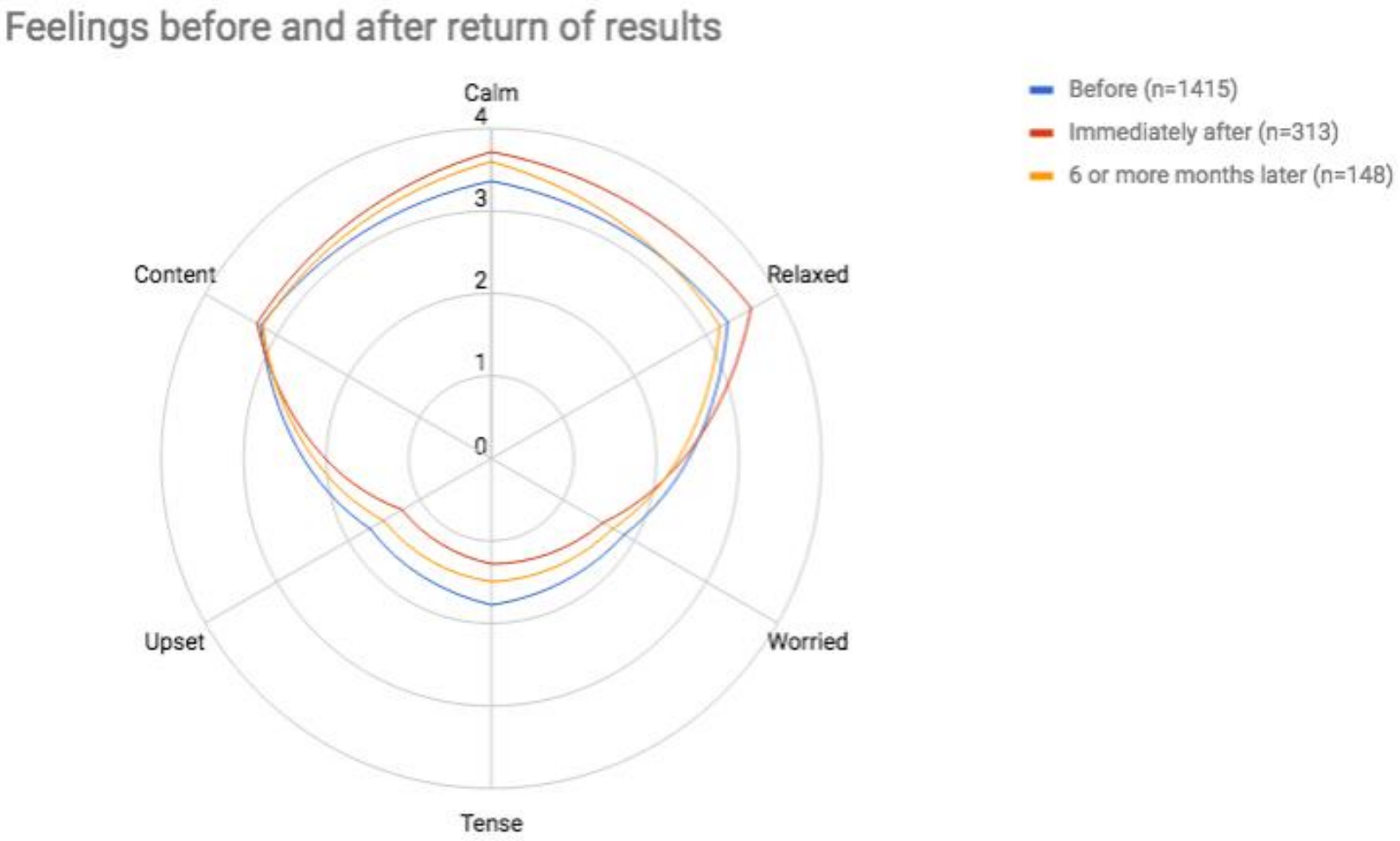
✚ - Tarvitada tavapäraselt. ! - Tarvitada ettevaatusega, võib vajada doosi muutmist ! - Tarvitada väga ettevaatlikult, oht kõrvaltoimeteks.

SLCO1B1

Statiinidest indutseeritud müopaatia ehk lihaskahjustus on harvaesinev seisund. Selle puhul esinevad ravimi võtmise foonil lihasnõrkus, -valulikkus, öised lihaskrambid, kõõluste valulikkus ja vereanalüüsides lihaskahjustusele viitavad muutused.

Kui Teile on antud ravimit kirjutatud ning on esinenud vastavaid tervisekaebusi, informeerige neist oma raviarsti.

Feelings towards genetic feedback



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Leitsalu et al unpublished

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Estonia to give genetic testing and advice to 100,000 residents

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Estonia will screen nearly one-tenth o its population's DNA in a controversia move to assess their disease risk

- Some 100,000 of the 1.3 million residents will provide blood samples
- Project aims to provide lifestyle advice and preventative measures to people
- Critics argue the programme could cause some participants 'great anxiety'
- Scientists maintain the information will be secure and made anonymous
- DNA will be analysed for more than 600,000 DNA variants linked to diseases

By ALEXANDRA THOMPSON HEALTH REPORTER FOR MAILONLINE

PUBLISHED: 15:45 BST, 2 April 2018 | **UPDATED:** 13:49 BST, 3 April 2018

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Estonia to DNA test 100,000 residents and give lifestyle advice based on results

Around one in 10 people to take part in programme to offer personalised health support

Chris Baynes | Tuesday 3 April 2018 23:32 BST | 5 comments

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Vision for future

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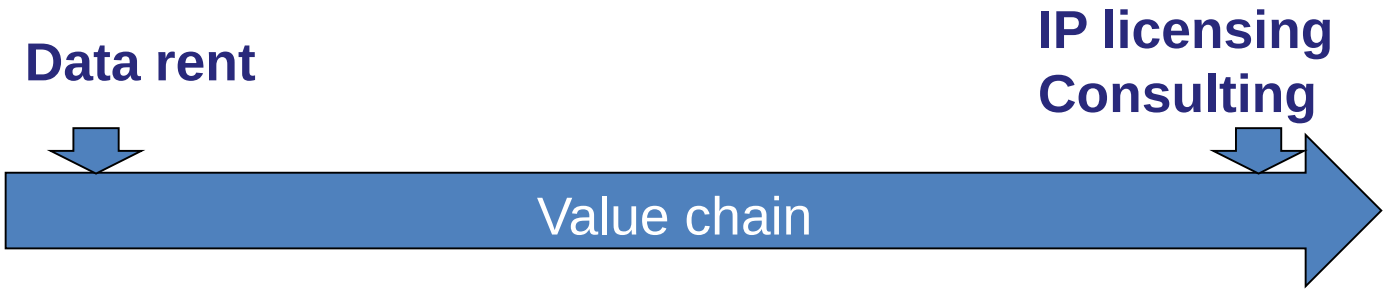
Digital Health Valley

- **Vision** is to create a globally attractive digital health ecosystem with fully accessible interlinked health and molecular data to enable disruptive innovation in academic research, healthcare services and business creation
- **Motivation** (a) attract investments and talent; (b) attract R&D centers and high pay jobs; (c) create additional resources for public service innovation, academic research
- **Operations** across value chain, focus (where feasible) on export of knowhow, products and services



Potential business cases

(a) Subscription fees, equity ownership (University Holding), royalty fees

(b) “Sand box” for secure access and analysis on Biobank data

(c) Supporting drug development processes by combining health and “omic” profiles

(d) Developing algorithms for better prediction, prevention and care for diseases

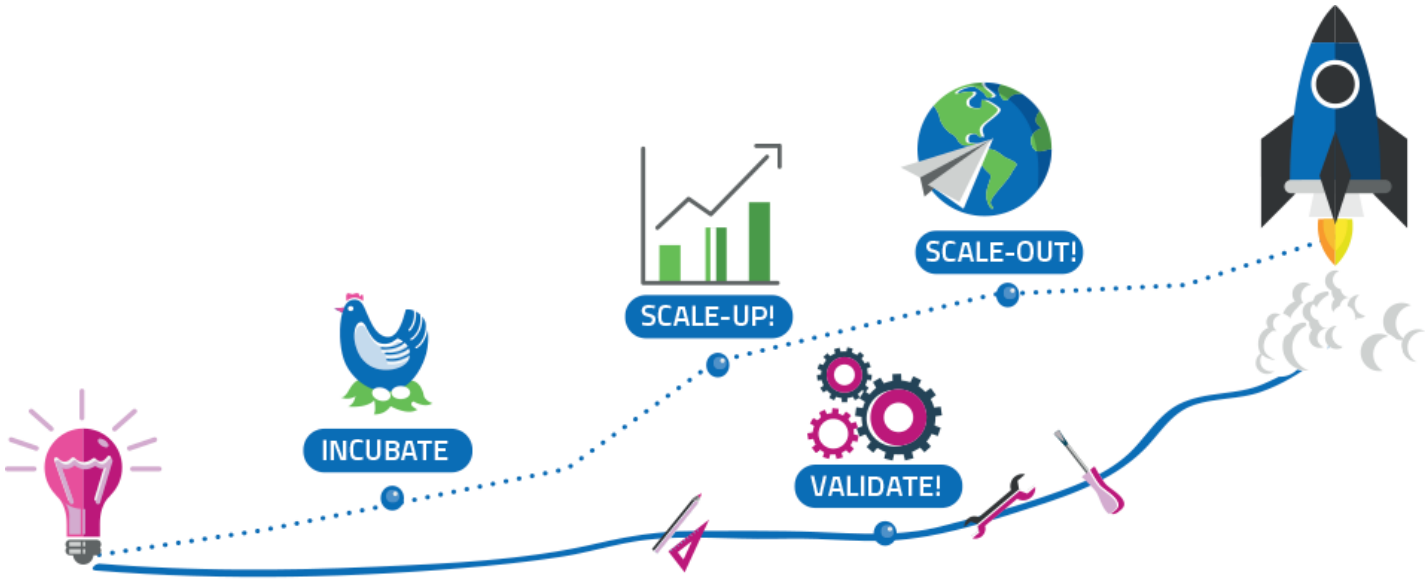
(e) Platform for trials for developing and testing technologies in real world setting

(f) Perform specific recall studies for deep phenotyping (Clinical Trials)

(g) Data accelerator for HealthIT and BioTech startups – combination of enhanced real world health data, access to test platform and circle of venture capital

(h) Hub for providing genetic profiling services (genotyping, sequencing)

Data accelerator for future products and services?



- Data already collected and access regulated
- Prompt testing of innovative ideas – fail fast model!
- Attractive model at global scale!
- Smart Clinical Trials – precision recruitment!

Proof of Concept StartUp acceleration – BioAgeLabs Inc

Understanding molecular signatures for human aging and all-cause mortality.

Duration: 2 years, March 2016 – Feb 2018

A start-up founded in 2015 (seed money 3M$) who aimed to go into the late-onset diseases/mortality space. Estonian Biobank provided the biological samples, study design and carried out the molecular profiling of 576 elderly Estonians.

The company carried out analyses, developed and protected IP for more than 100 molecules predictive for all-cause mortality.

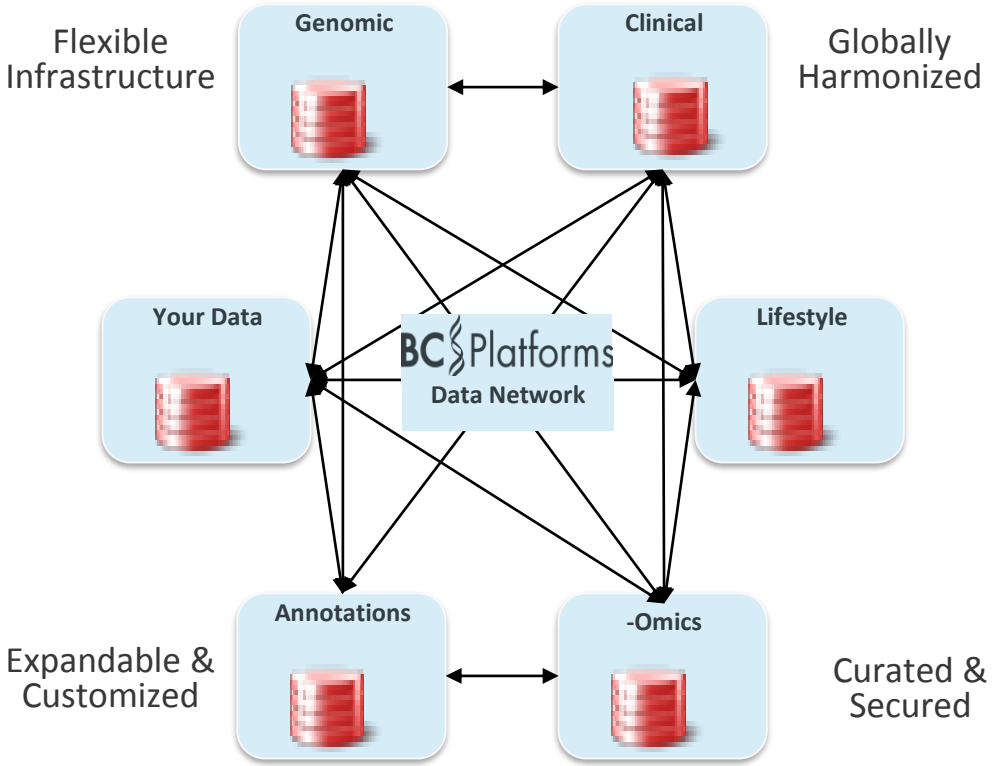
Based on the project results the company developed its IP and raised:
Round A – 10.9M$ (2017)
Round B – 23 M$ (2019)

Outi Törnwall, PhD

Data Partnerships Director,
BC Platforms

BC Platforms' Data Network Provides Unique Access to Diverse Data

Global Operations With Over *300K Patients Lives*
That Contain *Both Clinical & Genomic Data*



4.3 M Patients Contracted Today
(including 1.55 patients without full clinical & genomic data profile)

Flexibility for Your Researchers

- Integrate your proprietary data for analysis on the same platform
- Immediate availability to summary statistics for feasibility

We ensure Data Partners directly benefit from the value of their data

- **Provide access to source data while retaining ownership**
- **Receive the full value of data, no markups by BC Platforms**
- **Connect & collaborate with broad range of organizations**

- **Feasibility and data availability queries**
- **Aggregated analytics (PheWAS / GWAS)**
- **Proprietary data integration**
- **Individual patient level data studies**

Questions?

Ben Butkus
GenomeWeb

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University of Tartu & Estonian Biobank Science Center

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Please enter your questions in the Q&A panel on your screen.

Thank you for your participation!

Please be sure to fill out our post-webinar survey
to let us know how we did!