# Return of results to biobank research participants

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# Disclosures:

Scientific Advisor at Negen oy

Leave of Absence from GSK (scientific advisor)

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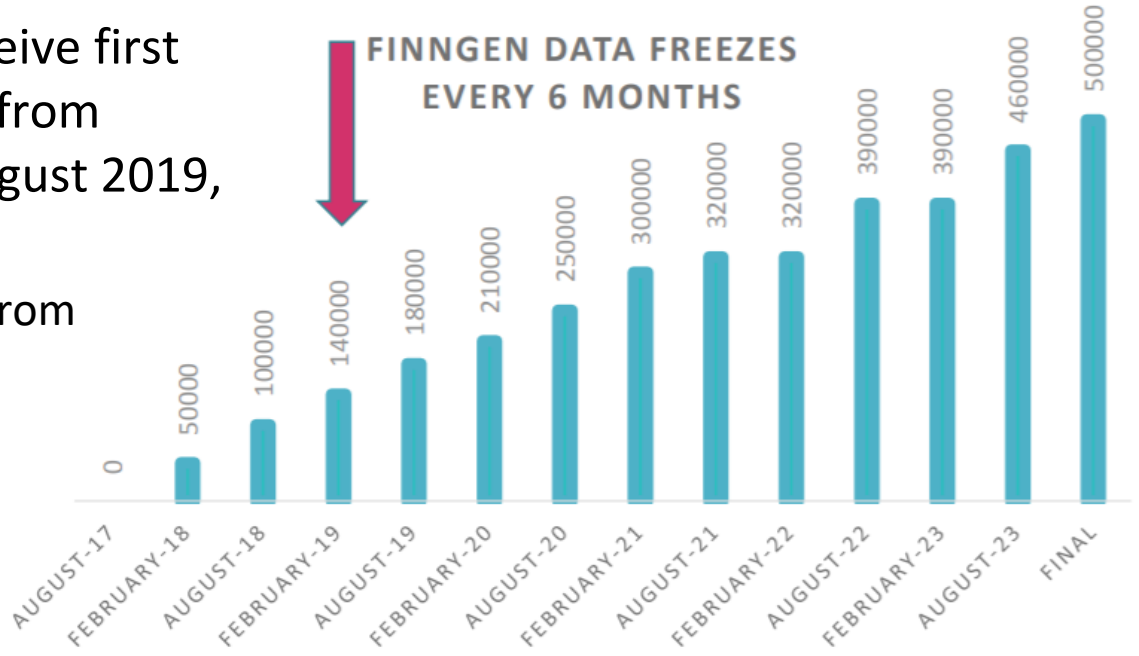
# Biobank samples

- ~300 000 new samples are collected for FinnGen from hospital biobanks by 2023
  - 106 500 Helsinki Biobank samples
- Data (genotypes) is returned to biobanks 1 year after data freeze
  - Currently 140 000 samples analyzed (data freeze #3, Feb 2019)
- Hospital biobanks will receive first FinnGen data (genotypes from ~24 000 individuals) in August 2019, increases every 6 months
  - 8/2019: data returned from 11 394 samples to Helsinki Biobank

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# Right to receive information

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- **The Finnish Biobank act** (39§) and EU data regulation (**GDPR**) guarantee a right to receive information on usage of personal samples and data as well as on research findings relevant to health
  - Biobank act 39§ 2 mom. : ….right to receive, **upon request**, information **concerning his or her health** as determined based on a sample. When providing information determined based on the sample, the person must be provided with an opportunity to receive an account of the **significance of the information**. **A fee may be charged** for clarifying the significance of the information…

- Biobank consent: *”I also authorize the biobank to contact me to inform me of findings from my samples which are of significance to my health”*
  - ~99 % agree

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# Convention on Human Rights and Biomedicine; Additional protocol concerning biomedical research

## Article 27 – **Duty of care**

- If research gives rise to information of relevance to the current or future health or quality of life of research participants, **this information must be offered to them**
- That shall be done within a framework of **health care or counselling**
- In communication of such information, due care must be taken in order to protect confidentiality and to respect any wish of a participant not to receive such information

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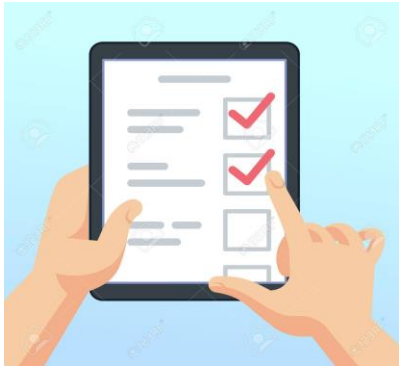
# Rationale for a Digital Participant portal for Finnish Biobanks



- Queries on usage of personal samples/data and their replies are performed manually in all Finnish biobanks
  - No digital means of exercising these rights exist
  - The right of access to information may be realized through an online viewing connection
- No centralized service: the participant needs to contact each biobank individually
- Currently the amount of returned results to biobanks is still modest → will change as returned results will greatly increase
- A Sitra funded investigation on feasibility of a common, FINBB-operated participant portal for Finnish Biobanks was made 8/2018-2/2019 by Helsinki Biobank

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# The Biobank Participant portal

- National, one-stop-shop to all biobank participants
  - Strong digital authentication method
  - FinnGen only one of the included research projects
- Portal, where participants can log in and receive information from all Finnish biobanks for:
  - What data and samples are stored and where
  - Where samples and data have been used
  - View research results
- Give/withdraw/restrict consent
- Recontact/recall possibility

- First implementations for the portal planned in 2019

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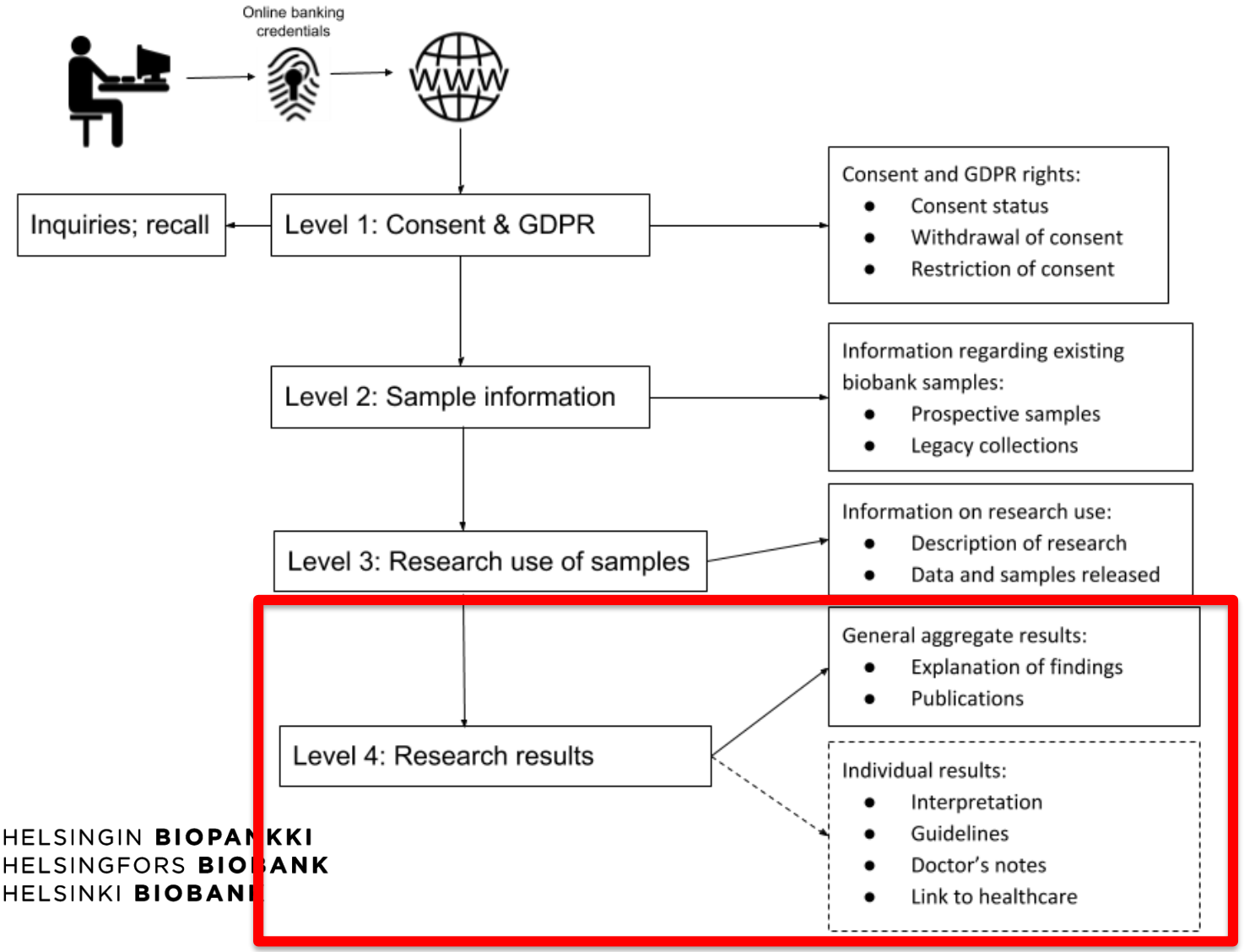
# Digital Participant portal for Finnish Biobanks

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# Return of biobank results:

## AGGREGATE RESULTS

- General results, not individual
  → equal to all participants
- Popular, easy to be comprehended
- In Finnish and English
- Researcher generates, biobank curates
- Ethical challenges minor
- Implemented relatively quickly

## INDIVIDUAL RESULTS

- Individual, personalized results
  → participant specific
- Popular, easily comprehended
- Raw data vs. interpreted information
- Ethically challenging
- Possibly relevant to participant's health
- Implementation challenging

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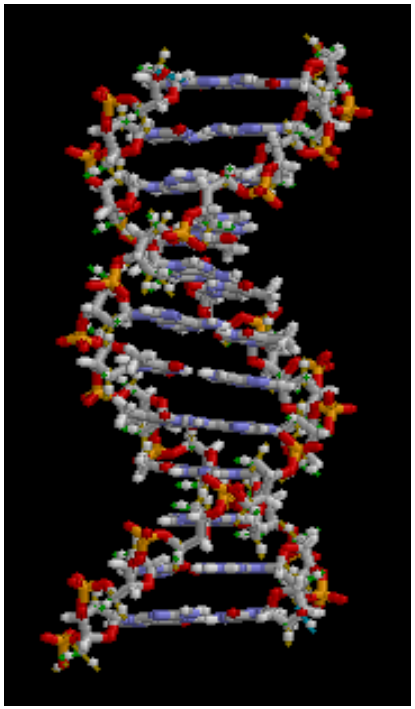
# Return of genetic research results

- There is no precedent on what research findings are considered relevant to biobank participant's health
  - Clinical relevance
- National criteria/guidelines for genetic data return is required
  - Genome center will instruct
- Return of results could be partly automatized, simultaneously clinically relevant results need to be validated and counselled
- "Right not to know"



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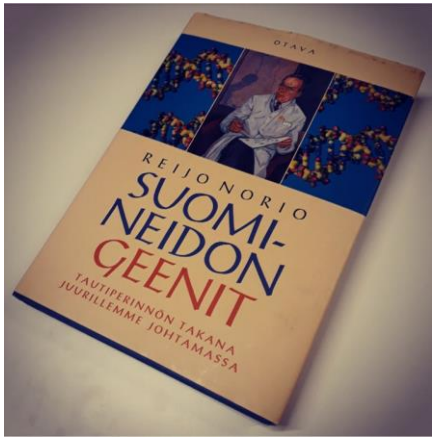
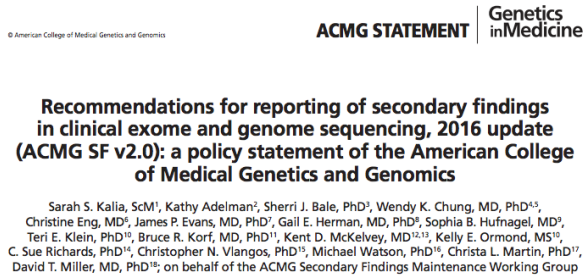
# What kind of results could be returned to FinnGen participants?




- ~700 000 variants genotyped with FinnGen array
- **Medically relevant information, actionable findings**

- Monogenic high risk disease variants
  - Ex. BRCA, long QT
- Predisposing low risk variants,
  - Ex. lactose intolerance variant, Factor V Leiden
- Pharmacogenetic results
  - Ex. Statins
- Polygenic risk scores

- ”Raw data” (genotypes)?



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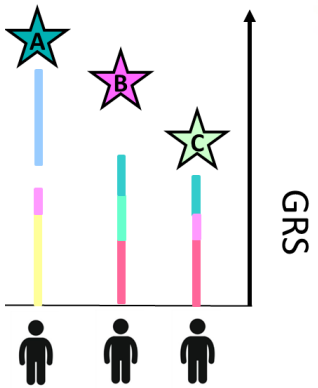
ACMG STATEMENT | Genetics inMedicine

**Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics**

Sarah S. Kalia, ScM[1], Kathy Adelman[2], Sherri J. Bale, PhD[3], Wendy K. Chung, MD, PhD[4,5], Christine Eng, MD[6], James P. Evans, MD, PhD[7], Gail E. Herman, MD, PhD[8], Sophia B. Hufnagel, MD[9], Teri E. Klein, PhD[10], Bruce R. Korf, MD, PhD[11], Kent D. McKelvey, MD[12,13], Kelly E. Ormond, MS[10], C. Sue Richards, PhD[14], Christopher N. Vlangos, PhD[15], Michael Watson, PhD[16], Christa L. Martin, PhD[17], David T. Miller, MD, PhD[18]; on behalf of the ACMG Secondary Findings Maintenance Working Group

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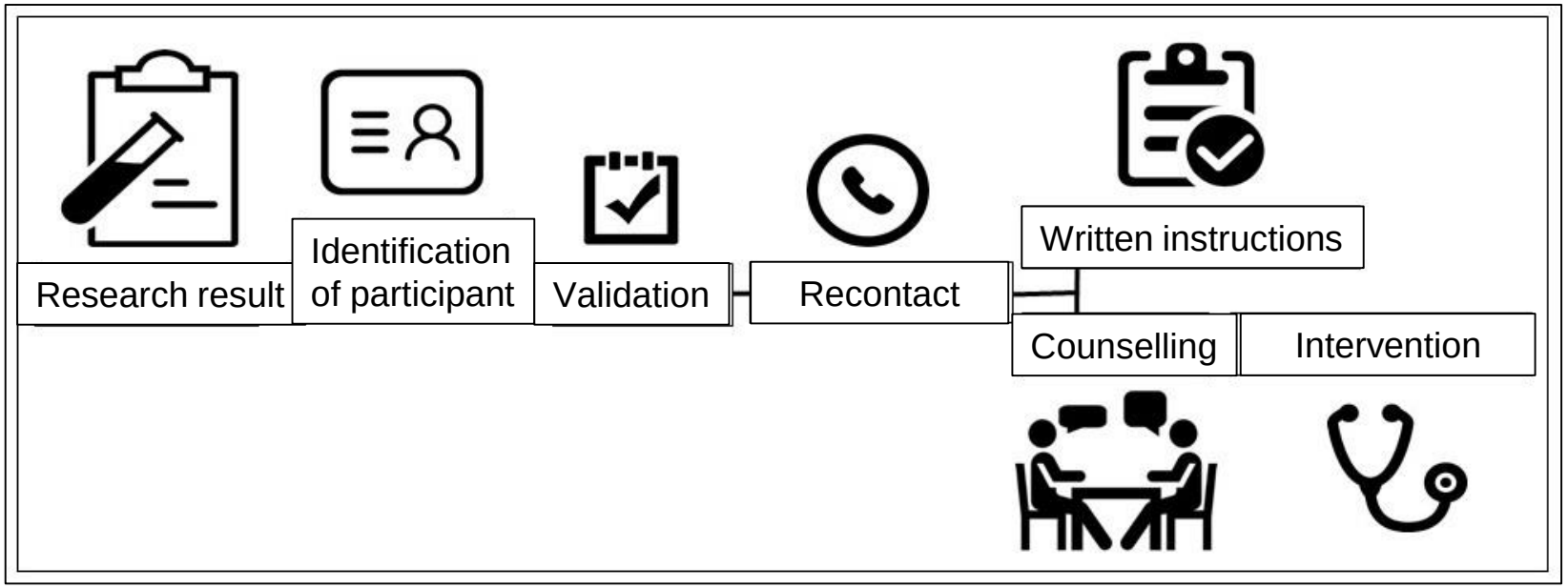
# Requirements for data return

- Consent: authorize the biobank to contact
- Criteria of returned results: definition of variants returned
  - National guidelines → equal procedures to all biobanks
- Validation of positive results (single clinical variants)
- Counselling process required
- Utilization of genomic information in clinical practice?: screening, treatment..



# Return of individual biobank results: possibilities and challenges

- Participatory: engages participants
- Empowering the public to use personal genetic information
- Desire to utilize genetic information in health care
- Recall: additional data collection possible

- Samples are not necessarily treated according to diagnostic procedures → validation required
- Collaboration between biobanks and genetic clinics is necessary
- Burden on healthcare vs. preventive healthcare

- Legal changes ongoing nationally: Genome act, Biobank act

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# Scenarios on returning biobank results to participants

1) No individual genetic results are returned
   - ”Research results” not medically relevant information (not meant for clinical use)?

2) Raw data returned upon request
   - Legal obligation to give significance on information
   - Ability to transfer the data to any service
   - Without interpretation → risk of wrong conclusions, validation?, counselling?, link to healthcare?

3) Specific results are returned <u>upon request</u>
   - Contact via participant portal
   - Participant actively choses to view results (disclaimers)
   - Link to healthcare?

4) High risk results are returned <u>to all participants</u>
   - Biobanks are not healthcare providers → link to healthcare
   - Requires resources but enables preventive healthcare

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# Thank you

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