The GeneRISK Study

Information for researchers interested in using stored samples and data

Introduction

The GeneRISK-study is an ongoing prospective observational study including 7 342 randomly selected 45-65-year old individuals from Southern Finland. The goal of the GeneRISK-study is to test the longitudinal impact of communicating personal genome-based disease risk information directly to the study participants. The GeneRISK study has been funded by Business Finland as part of the SalWe's GET IT DONE program of personalized diagnostics and care.

GeneRISK is coordinated by the Institute for Molecular Medicine Finland (FIMM, University of Helsinki) and has been carried out in collaboration with CAREA - Kymenlaakso social and health care services in Southeastern Finland, Mehiläinen Oy and the Finnish Red Cross Blood Service.

Study participants have undergone a clinical health check-up and information regarding their disease history and lifestyle has been collected via electronic questionnaires. Blood glucose and serum lipids were determined from fasting samples, and the genetic profile was analyzed using the HumanCoreExome BeadChip (Illumina Inc, San Diego, CA, USA). All study participants consented both to receiving data on their personal ASCVD-risk, including genomic risk information, and to being re-contacted.

Ethical considerations

The GeneRISK study is being carried out according to the principles of the Helsinki declaration and the Council of Europe's (COE) Convention of Human Rights and Biomedicine. All study participants have given their informed consent that permits the use of collected samples and data for the original study and for biobank research through THL Biobank, and the study protocol has been approved by the Ethical Committee of the Helsinki and Uusimaa Hospital district on 9 December 2014.

Selection of study subjects

The recruitment has been carried out during 2015–2017 at three different sites: 1) 4 857 individuals were recruited from CAREA - Kymenlaakso social and health care services in Southeastern Finland, 2) 1 369 individuals were recruited from Mehiläinen Oy (Helsinki and Turku region offices), and 3) 1 116 individuals were recruited from the Finnish Red Cross Blood Service in Helsinki, ascertaining healthy blood donors.

Health-related exclusion criteria:

- pregnancy
- diagnosed myocardial infarction, stroke, cerebral hemorrhage or cerebral infarction
- diagnosed coronary heart disease, coronary artery bypass or coronary angioplasty

GeneRISK samples available for biobank research

The following samples are available from over 7 300 sample donors through THL Biobank

- DNA
- serum (blood collected into gel tube, processed within 60 min after sampling)
- plasma (EDTA, processed within 60 min after sampling)

Sample collection and processing details:

The study nurses were trained to perform sampling and sample processing in a similar manner at all centers of recruitment. Participants were instructed to fast overnight for 10 hours before the blood samples were collected. Blood samples were collected by venipuncture for DNA extraction and for serum (VF108SAS, gel) and plasma (VF-109SDK, EDTA) samples. Serum and plasma were let to settle for 30 (to maximum of 60) min and then centrifuged (2200 g for 10 min), aliquoted in 0,5 ml fractions (Fluid X tubes) and frozen immediately (-20^{0}C) on site. Serum and plasma samples were shipped in dry ice every week for long term storage in -185^{0}C. DNA extraction was performed using PerkinElmer Janus Chemagic 360i Pro Workstation with CMG-1074 kit. This workstation uses patented magnetic bead technology with fully automated liquid handling.

GeneRISK phenotype and omics data available for biobank research

Basic data

- Age
- Gender
- Sampling date, sample specifications and quality information

Data collected by questionnaires

- Sociodemographic characteristics
- Health status
- Use of health services
- Family history of disease
- Lifestyle
  - ✓ Physical activity
  - ✓ Drugs
  - ✓ Smoking
  - ✓ Alcohol use
  - ✓ Sleep
  - ✓ Nutrition
  - ✓ Lifestyle changes
- Functional ability
- Quality of life
- Women's questions (e.g. contraception)

Physical examination data

- Weight, height, BMI, waist circumference
- Blood pressure, pulse

Biological test results

- Blood lipid values (total cholesterol, HDL and LDL cholesterol, triglycerides, apolipoproteins A1, B)
- Glucose

Omics data

- Genome-wide SNP data
- Genotype data imputed to a population-specific reference panel

For availability of genome-wide genotypes and sequencing data, see more information in the 'THL Biobank Omics data availability table' at the THL Biobank sample collection page.

Registry data

Data from Finnish National Registers (such as National Social Welfare and Health Care registers e.g. Care Register for Health Care, Cancer Registry, Statistics Finland's registers, Kela's registers e.g. Drug Reimbursement Registers) can be linked to all sample donors by a separate application process.

Research group

Principal Investigator

Elisabeth Widén, University of Helsinki

Research coordinator

Johanna Aro

Key references:

CARDIoGRAMplusC4D Consortium, Deloukas P, Kanoni S, Willenborg C, Farrall M, Assimes TL, Thompson JR, Ingelsson E, Saleheen D et al. Large-scale association analysis identifies new risk loci for coronary artery disease. Nat Genet. 2013 Jan;45(1):25-33

European Society of Human Genetics. "Individual access to genomic disease risk factors has a beneficial impact on lifestyles." ScienceDaily, 15 June 2018

Ripatti S, Tikkanen E, Orho-Melander M, Havulinna AS, Silander K, Sharma A, Guiducci C, Perola M, Jula A, Sinisalo J, Lokki M-L, Nieminen MS, Melander O, Salomaa V, Peltonen L, Kathiresan S. A multilocus genetic risk score for coronary heart disease: case-control and prospective cohort analyses. Lancet 2010 Oct 23;376(9750):1393-400

Snell K, Helén I. 'Well, I knew this already' - explaining personal genetic risk information through narrative meaning-making. Sociol Health Illn. 2020 Mar;42(3):496-509

Tikkanen E, Havulinna AS, Palotie A, Salomaa V, Ripatti S. Genetic Risk Prediction and a 2-Stage Risk Screening Strategy for Coronary Heart Disease. Arterioscler Thromb Vasc Biol. 2013 Sep;33(9):2261-6