



# Genetikanalyser i SCAPIS-kohorten – ett samarbetsprojekt

Tove Fall, professor i molekylär epidemiologi  
Uppsala Universitet

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through a collaborative effort between:

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# SCAPIS

en världsunik forskningsstudie som ska förhindra hjärt-lungsjukdom.

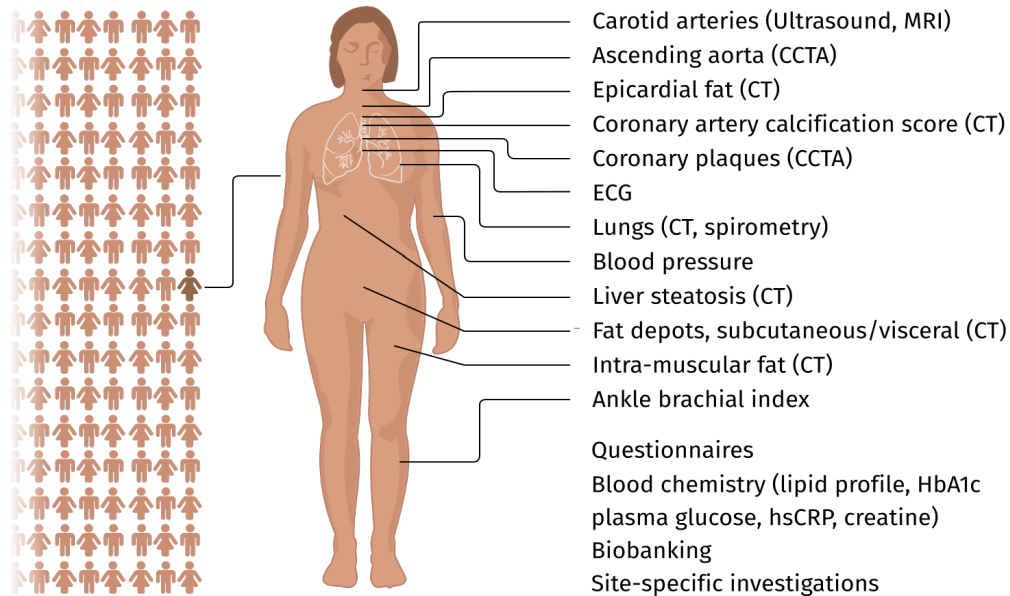
Den genomförs av forskare på sex universitetssjukhus tillsammans med Hjärt-Lungfonden.



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# SCAPIS insamling och analys 2012-2018



SCAPIS är en svensk befolkningsstudie med fokus på hjärt- och lungsjukdom  
30 000 slumpvis utvalda kvinnor och män i åldern 50-64 år  
Biologiska prover i sex olika biobanker  
Centralt Data Office

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# Omfattande extra lokala undersökningar

- Bukhöjd
- Kognitionstest
- Handstyrka
- Saturation
- Puls
- Kväveoxid i utandningen
- Accelerometri, lår
- 24 h EKG och BT
- Sömnregistrering
- Avföringsprov



Stor potential för massor  
av viktig forskning!



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# SCAPIS Datatjänst

- Göteborgs universitet
- Samtliga core-data från alla site
- Säker lagring och backup
- Administrativ support vid ansökningar
- Kontakt, maila [scapis@scapis.org](mailto:scapis@scapis.org)
- 1413 variabler i dagsläget

<https://SCAPIS.org/portal/variables>

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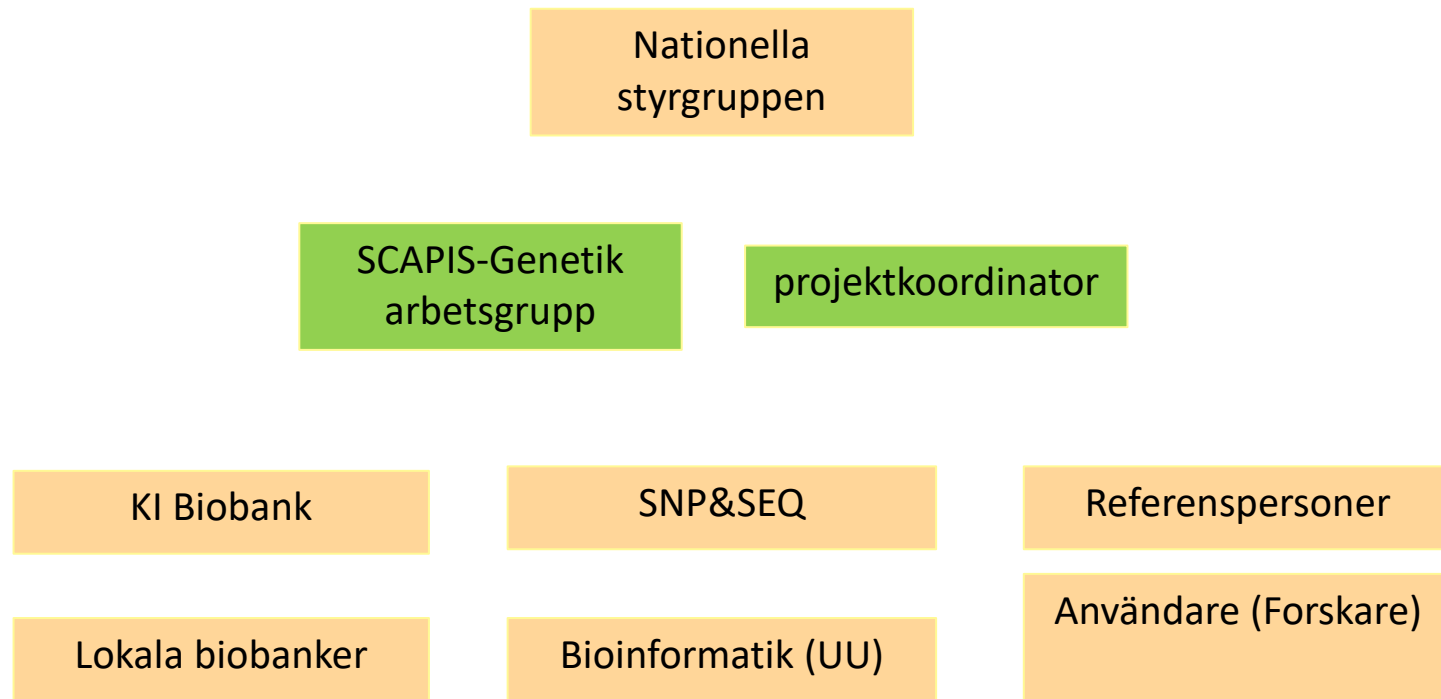
# SCAPIS-Genetik

- Med genetiska data kan vi studera genetiken bakom många fenotyper och berika nuvarande frågeställningar
- Projektet startade tidigt 2020 med målet att genotypa alla individer
- Genetikprojektet finansieras av Hjärtlungfonden, budget 20M
- Jag leder SCAPIS-Genetik arbetsgrupp (representanter från alla sites) på uppdrag av SCAPIS nationella styrgruppen (ordf Göran Bergström)
- Maria Storgärds, UCR projektkoordinator
- Proverna lagrade i sex olika biobanker > utlämnande till UU > MTA KI Biobank
- Projektet går relativt snabbt
- Nyckel: **parallella processer, lokal förankring, konsultationer med experter**

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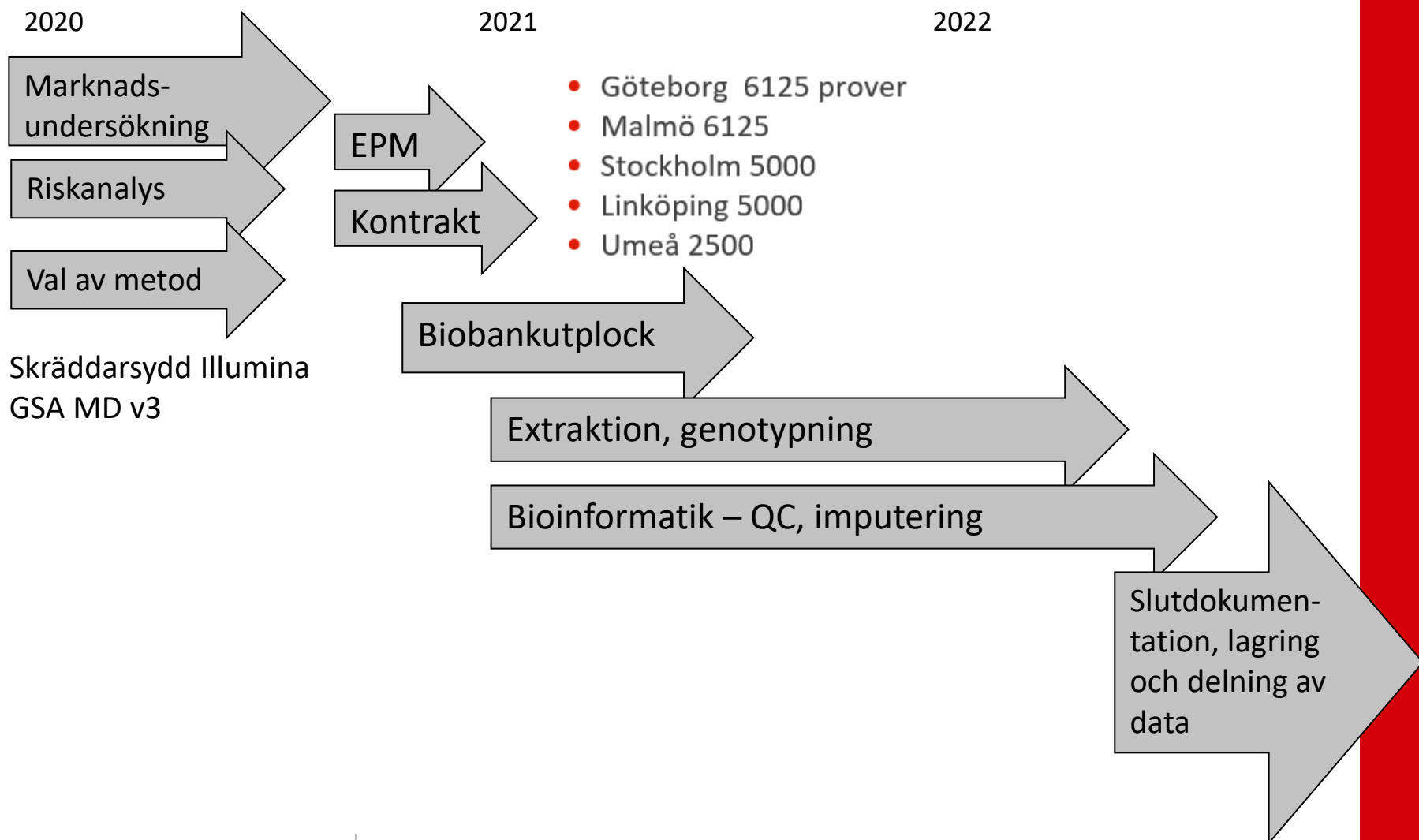
# SCAPIS-Genetik organisation



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# Projekttidsplan



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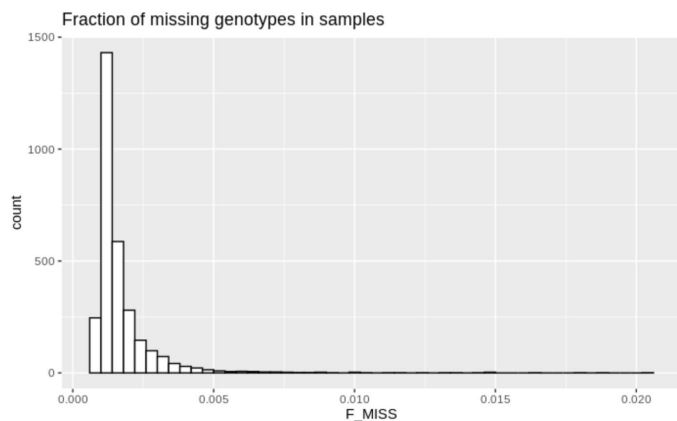
# Kvalitetssäkring

- På KIBB genomfördes ankomstkontroll och kontroll av nummerserier
- Extraktion sker randomiserad;
  - mellan siten
  - procentuell representation från site för varje extraktions-batch
- Koncentrationsmätning (NanoDrop) KIBB
- På varje platta lämnas en slumpvis position tom, för att kunna upptäcka en eventuell felaktig rotation av plattan
- DNA koncentrationsmätning upprepas på SciLifeLab (PicoGreen)
- Kvalitetskontroll genotypning SciLifeLab
- Kvalitetskontroll data UU

# Resultat QC genetik – batch 1 (n=3033)

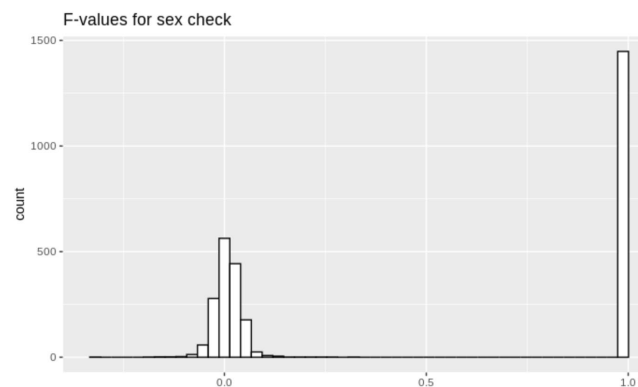
## Call rates of samples and markers

Total genotyping rate was 0.998. 14 samples had missing fractions > 0.01 with a maximum value of 0.02.

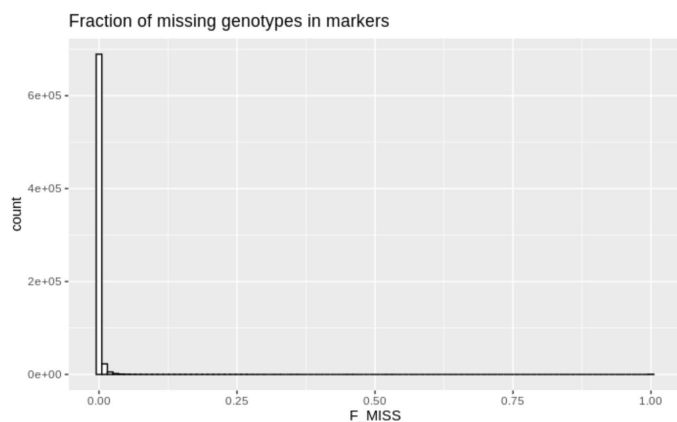


## Genetic vs phenotypic sex

Males/females were well separated in F values.

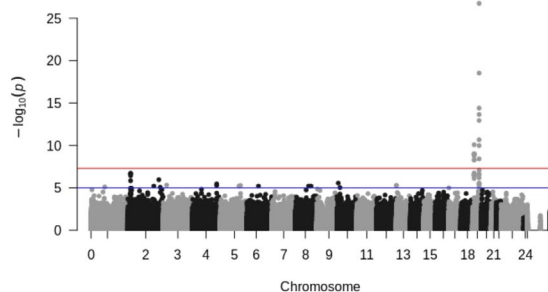


20726 markers had missing fractions > 0.01, 10917 markers > 0.02, 4989 markers > 0.1



## Association to LDL chol.

SNPs associated to LD cholesterol could be detected using the genotype data.



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# Framgångsfaktorer så här långt

- Riskanalys
- Konsultationer med experter genom hela processen
- I marknadsundersökningen lade vi stor vikt vid kvalitet
- Parallella processer
- Kvalitetskontroller i varje steg
- Lokal förankring - gott samarbete med lokala biobanker och forskare

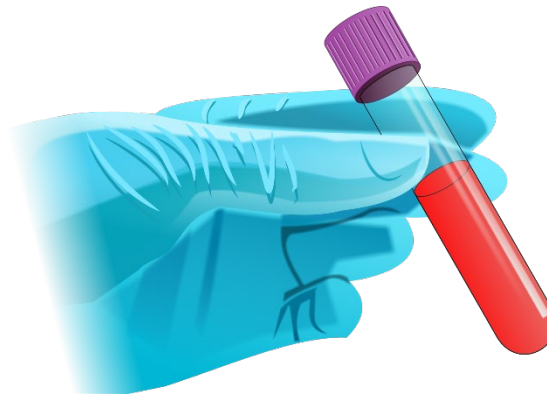
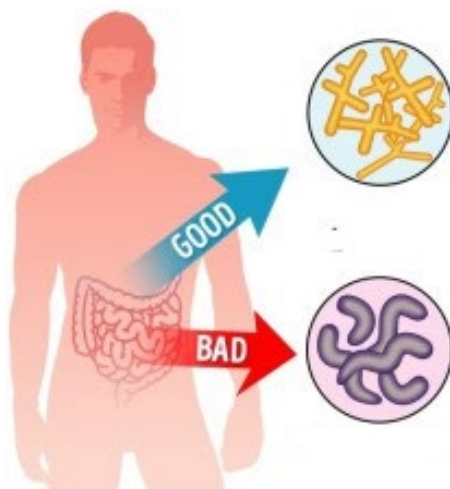
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# Projektet GUTSY bygger på SCAPIS

Mål:

1. Identifiera tarmflorebakterier kopplade till ateroskleros
2. Identifiera plasmabiomarkörer för skadliga tarmflora
3. Testa vilka länkar som motsvarar kausala samband



orsak

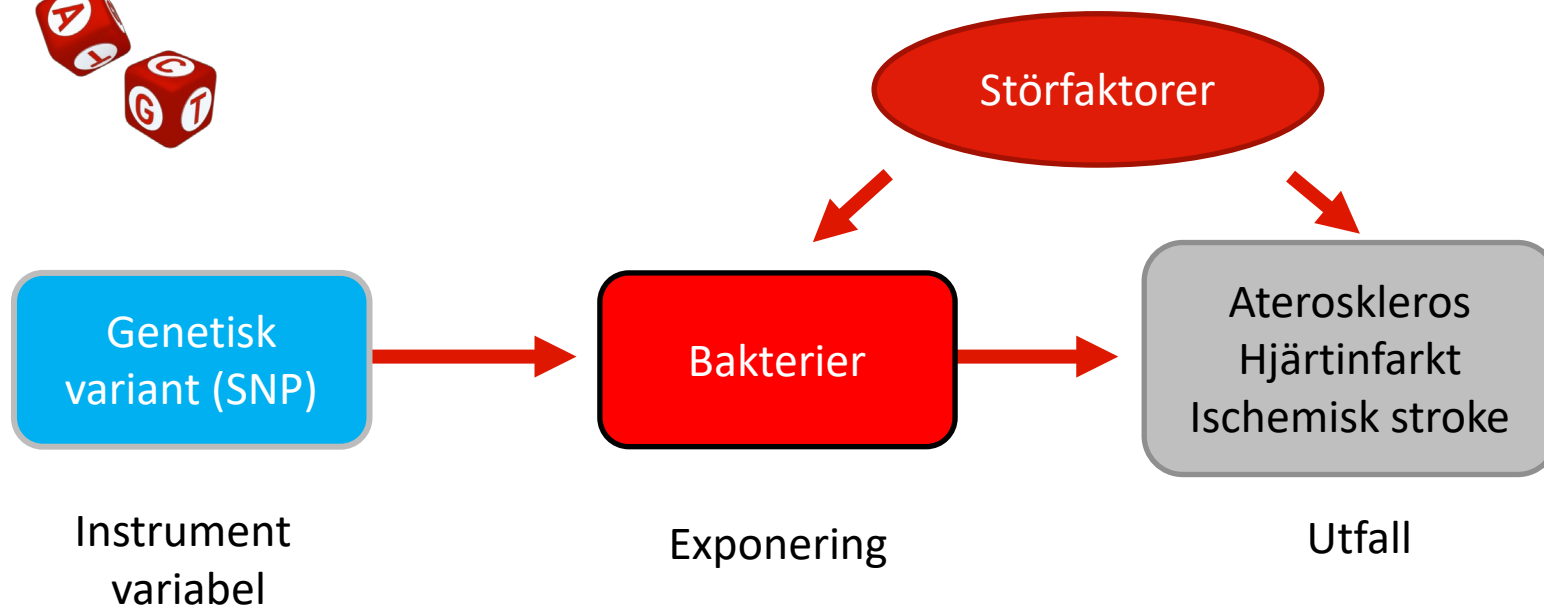


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# Mendelsk randomisering



Mendelsk randomisering testar **kausala samband**

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# Finansiering



# Hjärt- Lungfonden

*Knut och Alice  
Wallenbergs  
Stiftelse*

VINNOVA



Vetenskapsrådet

- Universitetssjukhusen och universiteten själva
- SNP&SEQ
- SNIC-SENS

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