



Institute for Molecular Medicine Finland
Nordic EMBL Partnership for Molecular Medicine

HiLIFE UNIT



UNIVERSITY OF HELSINKI



Changes in the fine-scale genetic structure of Finland through the 20th century

Sini Kerminen, Helsinki-Boston 21 Oct 2020

Institute for Molecular Medicine Finland

University of Helsinki

Finland

FROM FINE-SCALE POPULATIONS TO ANCESTRY

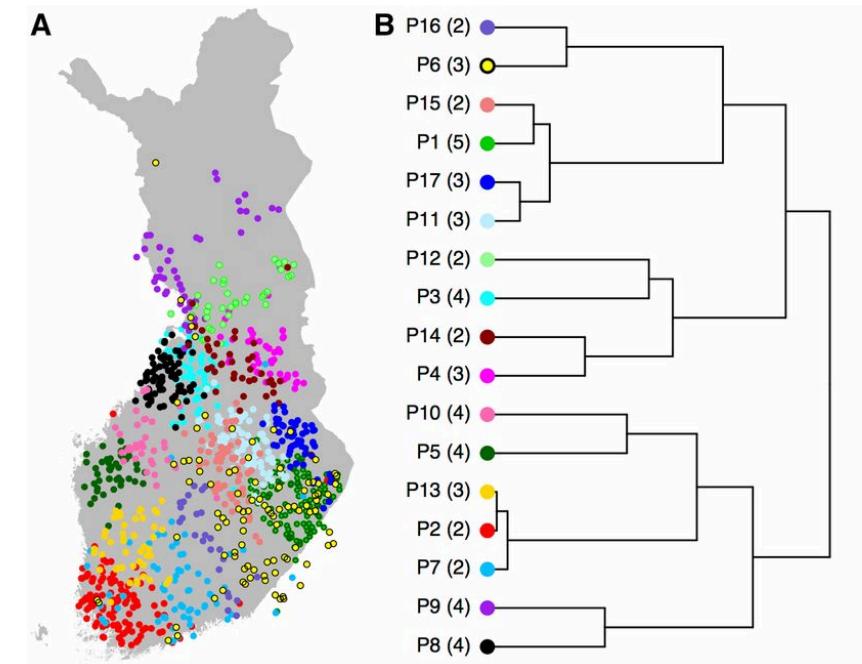
Previous results:

- 1,042 samples, parents born within 80 km

New project:

- >18,000 samples, parents born all over Finland
- How well can we estimate individual genetic ancestry?
- Can we detect changes in the genetic structure within Finland?
 - Focus on the 1900 century

Kerminen et al. 2017, G3:



FROM FINE-SCALE POPULATIONS TO ANCESTRY

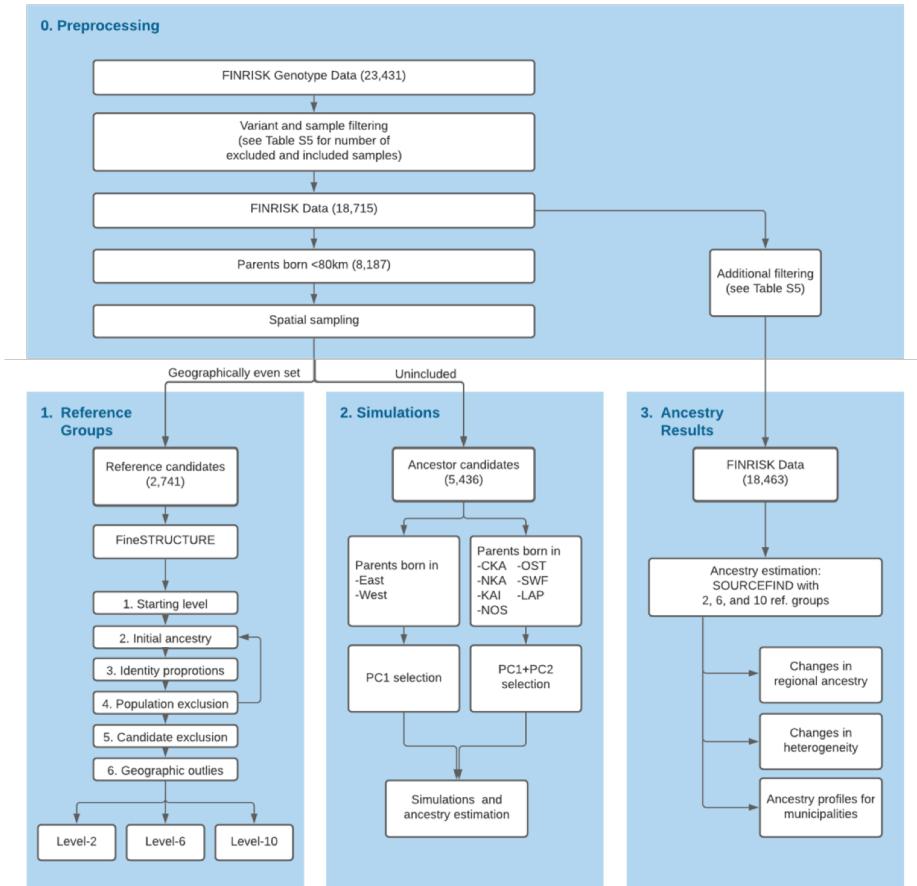
Previous results:

- 1,042 samples, parents born within 80 km

New project:

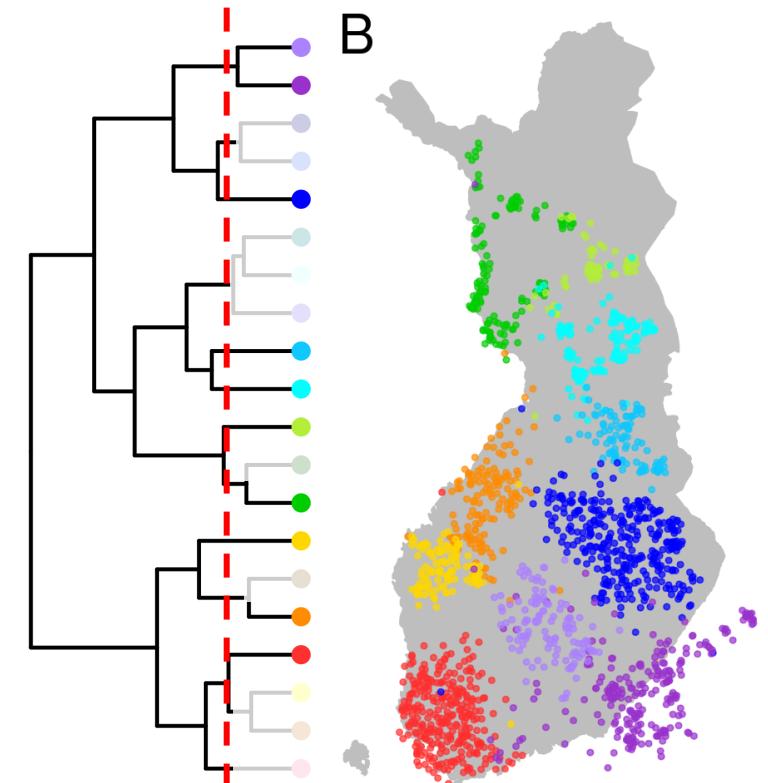
- >18,000 samples, parents born all over Finland
- How well can we estimate individual genetic ancestry?
- Can we detect changes in the genetic structure within Finland?
 - Focus on the 1900 century

Workflow:



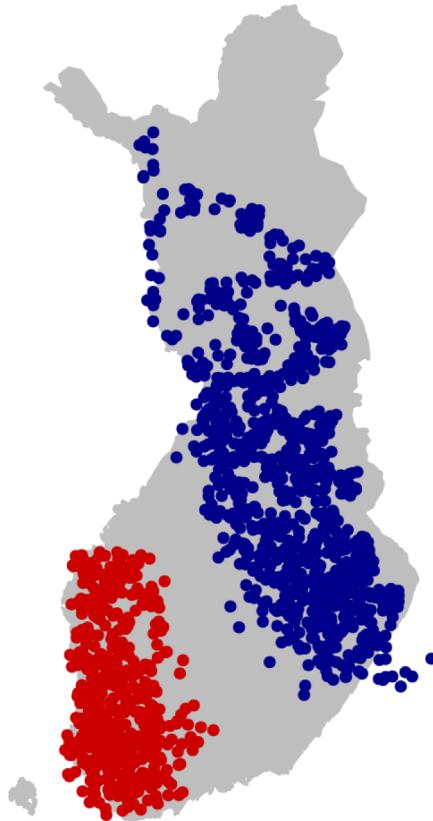
GENETIC ANCESTRY ESTIMATION

- Utilized SOURCEFIND software (Chacon-Duque et al. 2020, Nat Commun.)
- Built reference groups within Finland:
 - Identify fine-scale populations
 - Choose the starting number of populations, K
 - Estimate the genetic ancestry of reference candidates with respect to K
 - For each of the K populations, calculate the population's *identity proportion*
(= the average proportion of ancestry in that particular population across the individuals assigned to that population)
 - Exclude the populations with low identity proportion (< 50%)
 - Exclude reference candidate samples who show low levels of ancestry (thresholds used either <70% or <95%) from the population they were assigned to
 - Exclude manually possible geographic outliers.

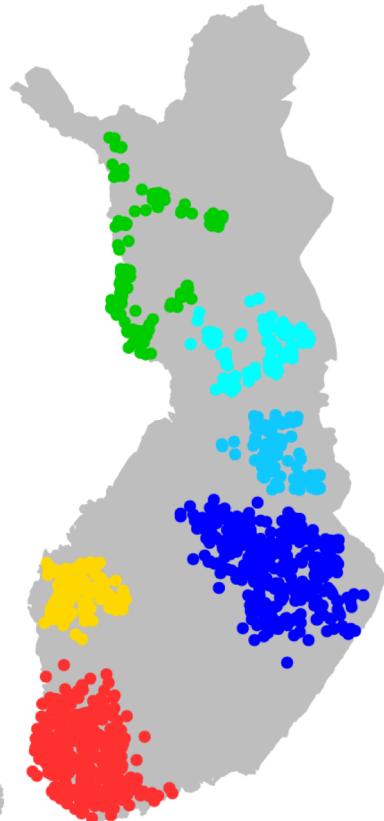


REFERENCE GROUPS

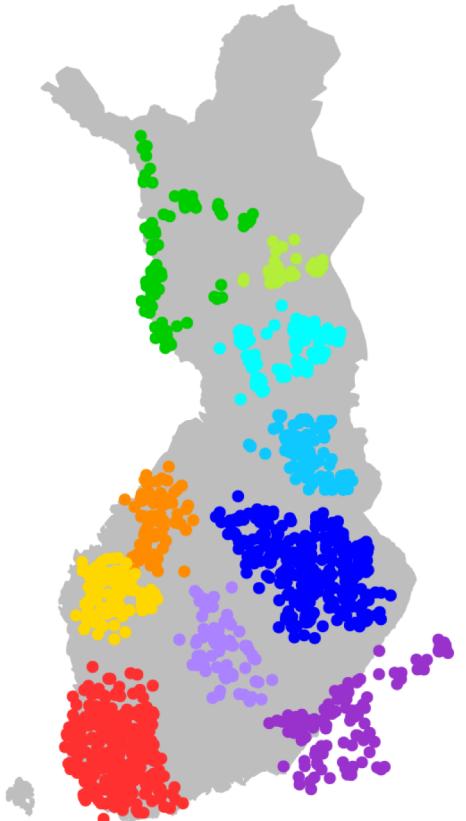
A) 2 groups



A) 6 groups



A) 10 groups



R2-West
R2-East

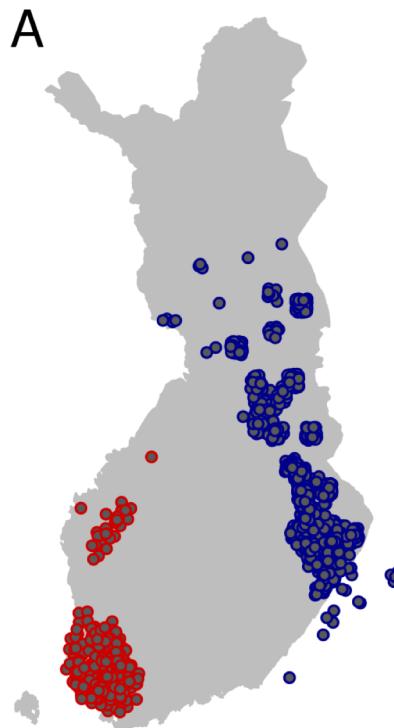
R6/10-Southwest
R6/10-West_Lapland
R6/10-Savo-Karelia
R6/10-Kainuu
R6/10-Bothnia
R6/10-Kuusamo
R10-Evacuated
R10-Kokkola
R10-Central_Finland
R10-East_Lapland



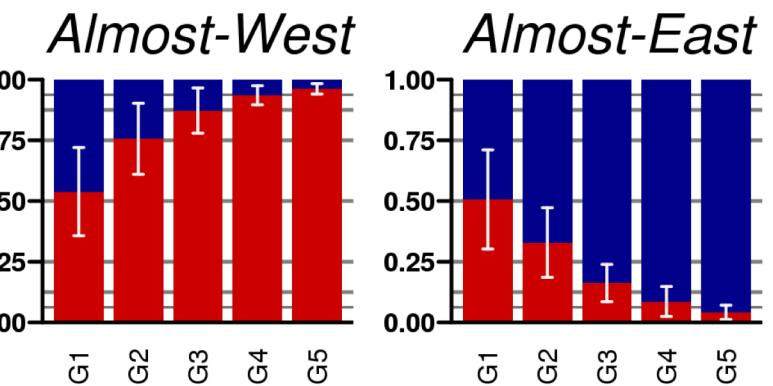
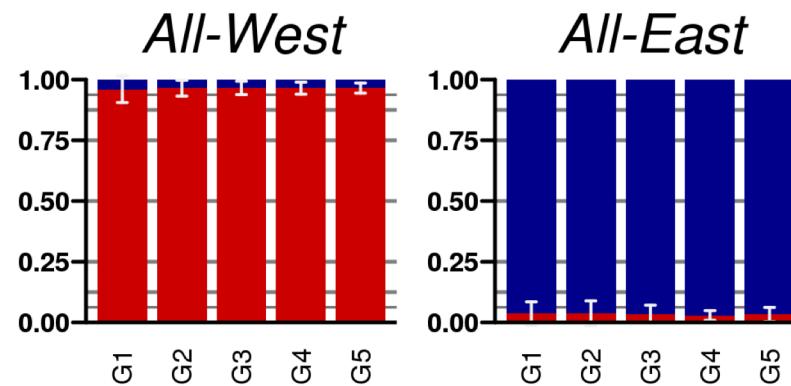
TESTING DETECTABILITY

- Simulated 20 offspring using real word ancestors originating from the different parts of Finland

Ancestors



Estimated ancestry

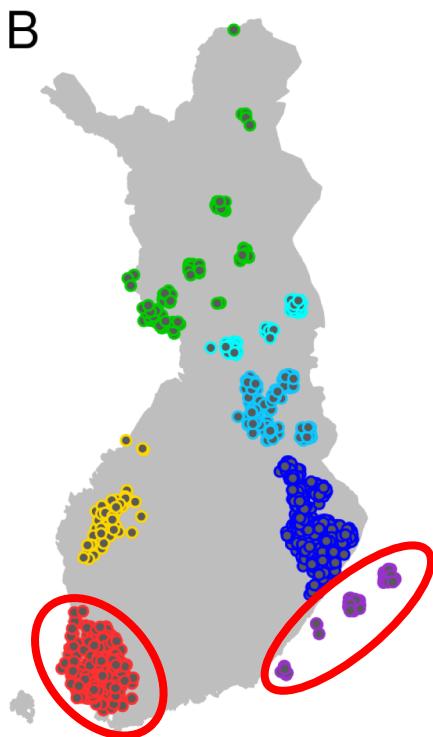


→ Eastern/Western ancestry can be detected 4 generations back in time (~6%)

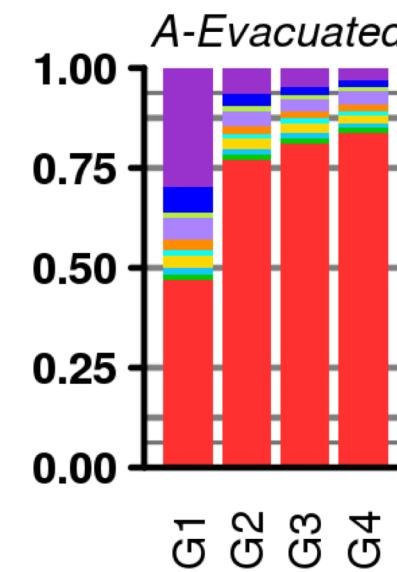
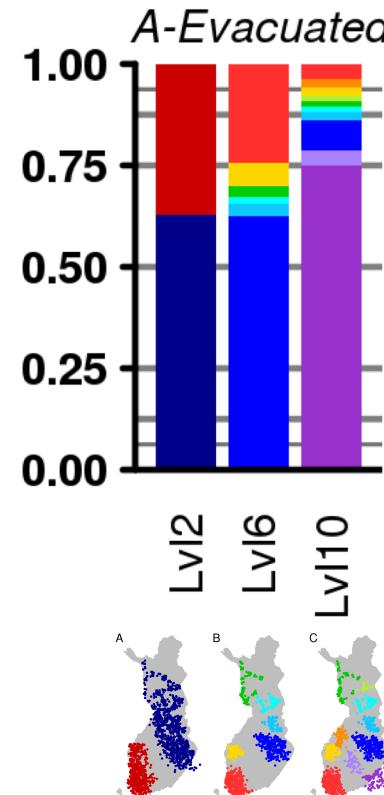
TESTING DETECTABILITY

- Simulated 20 offspring using real word ancestors originating from the different parts of Finland

Ancestors

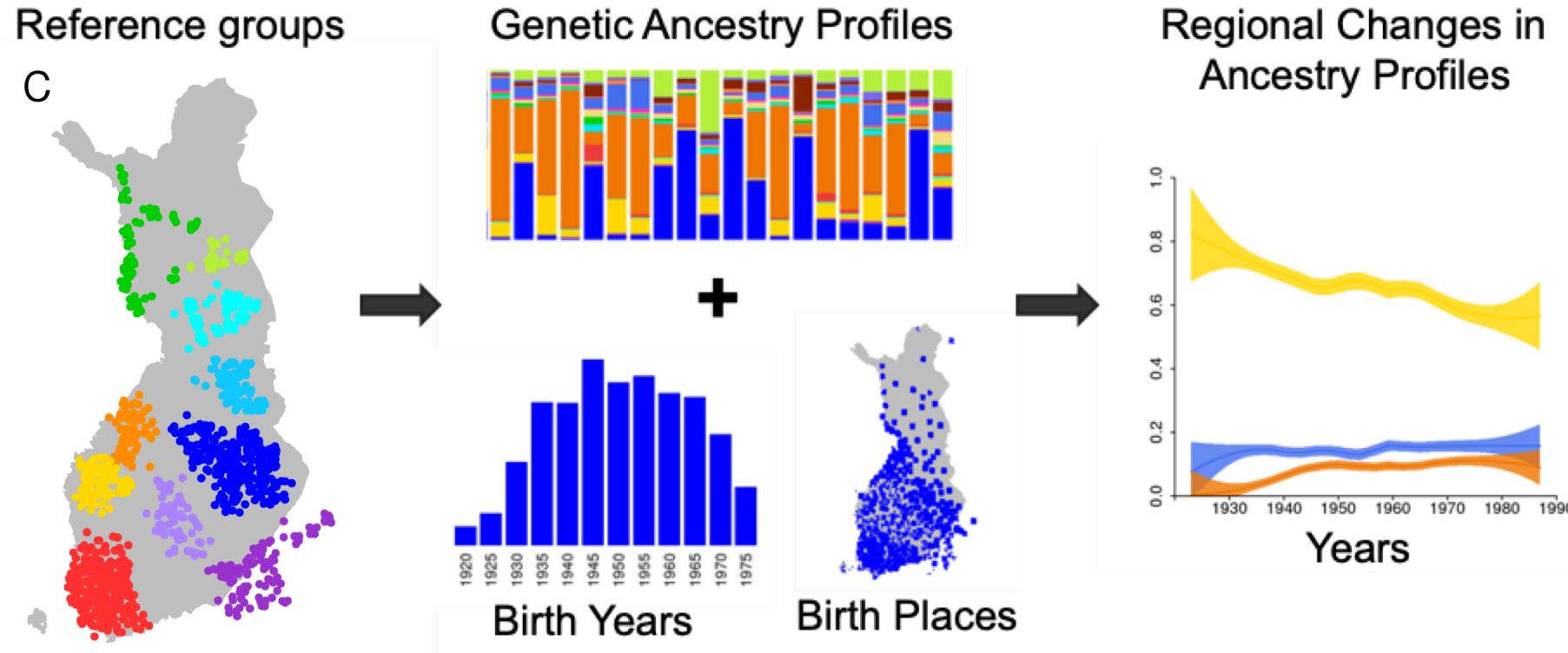
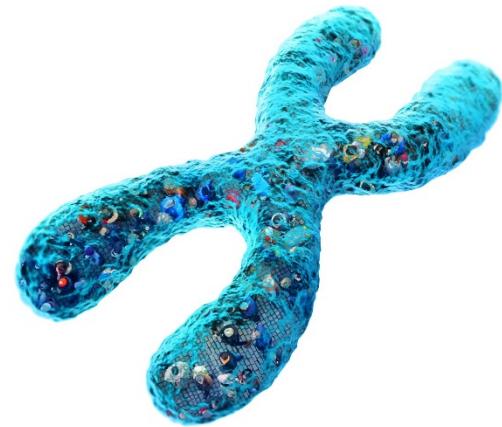


Estimated ancestry

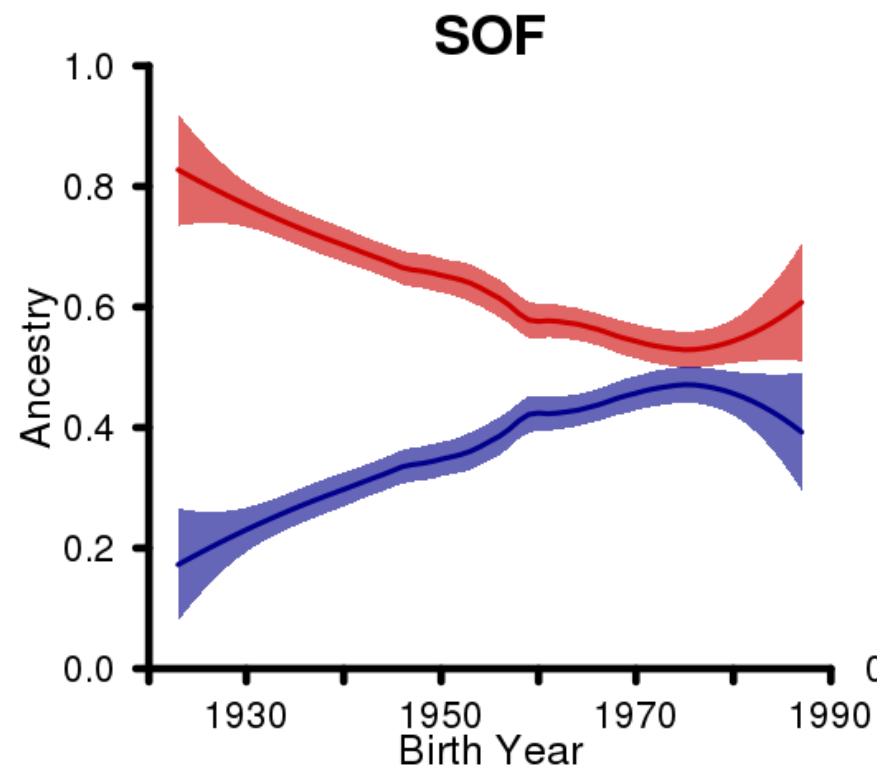
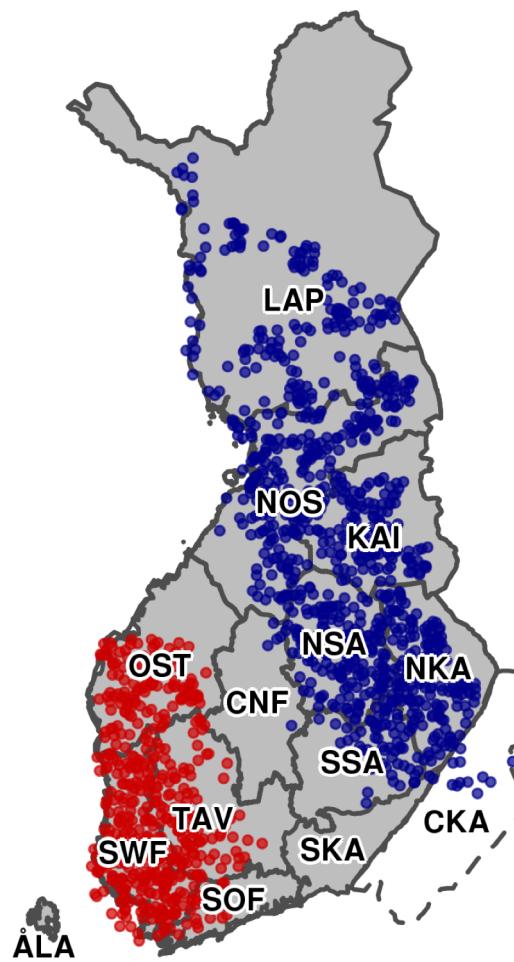


→ Minor source of ancestry is detectable 2 generations back

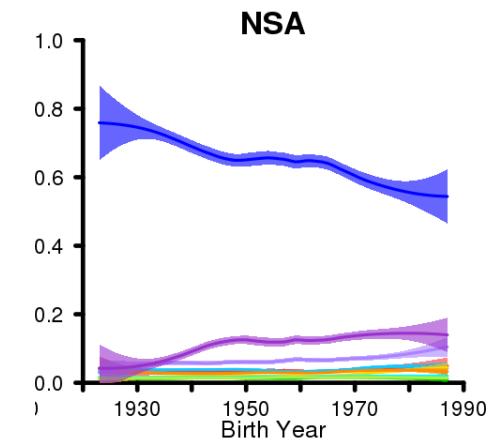
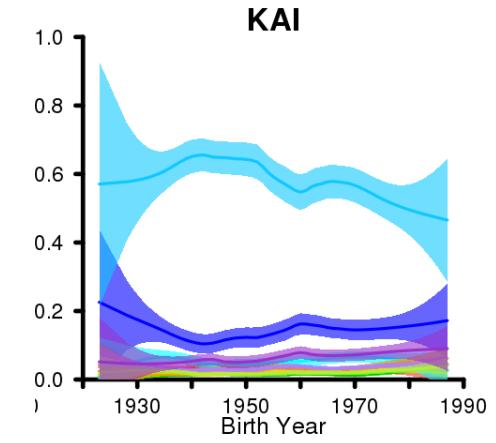
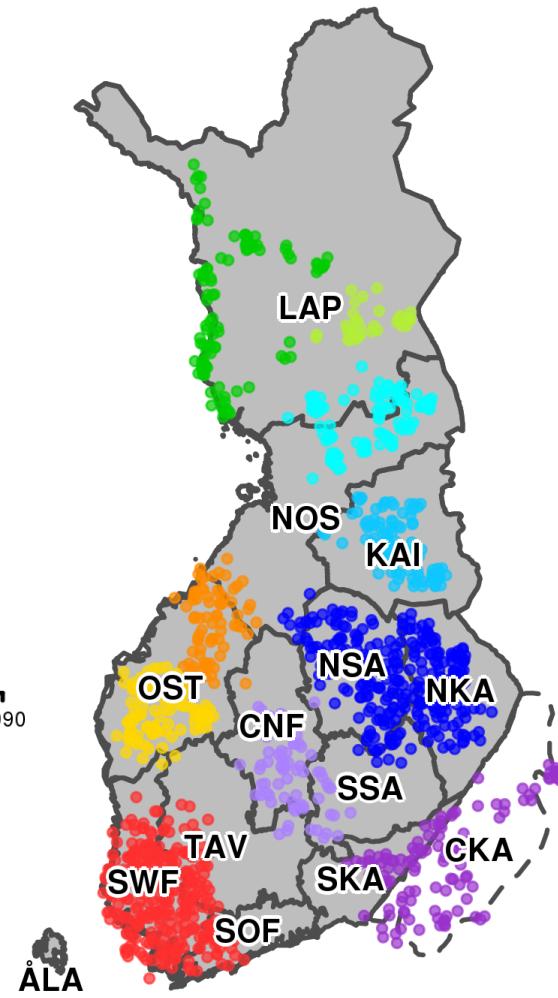
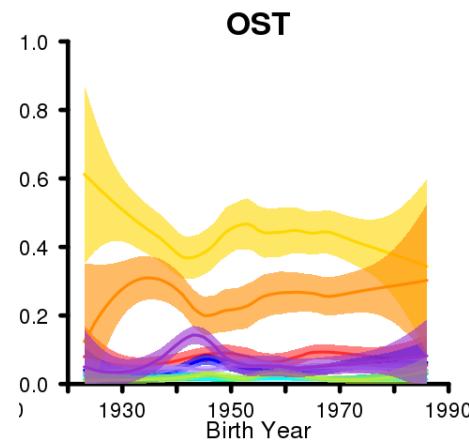
GENETIC ANCESTRY FOR 18,463 SAMPLES



CHANGES IN GENETIC STRUCTURE: 2 GROUPS

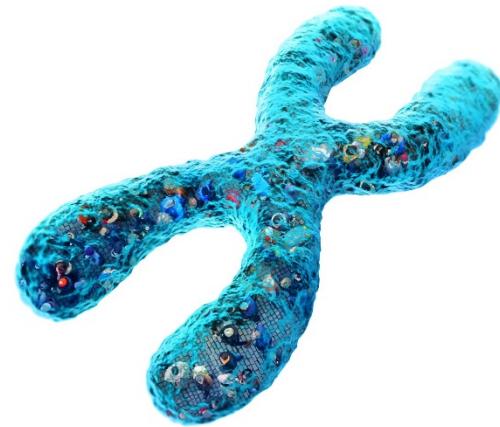
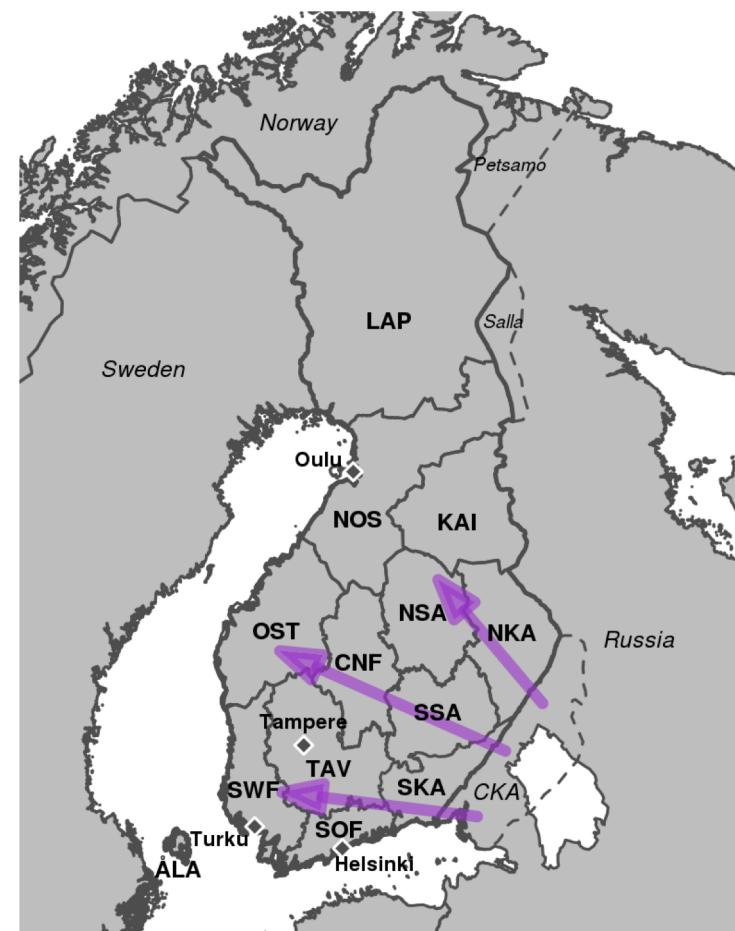


CHANGES IN GENETIC STRUCTURE: 10 GROUPS

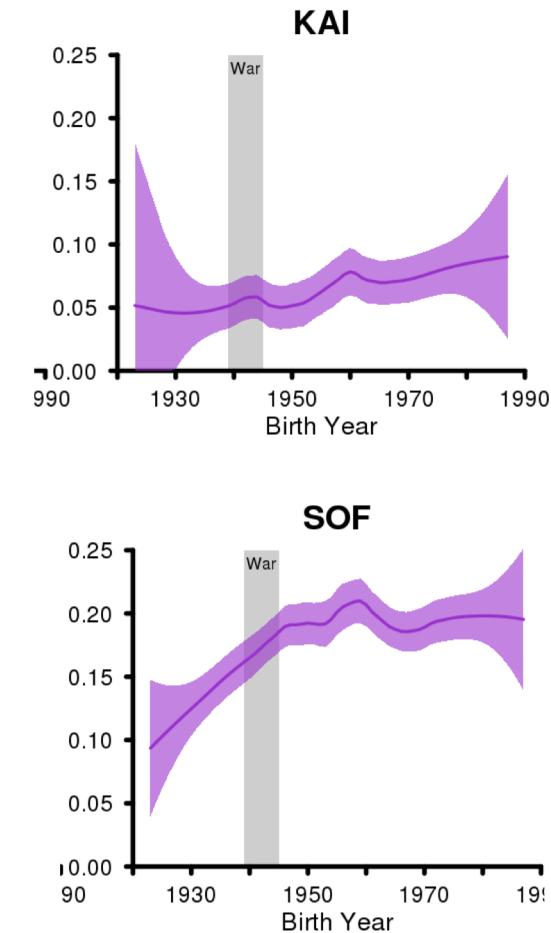
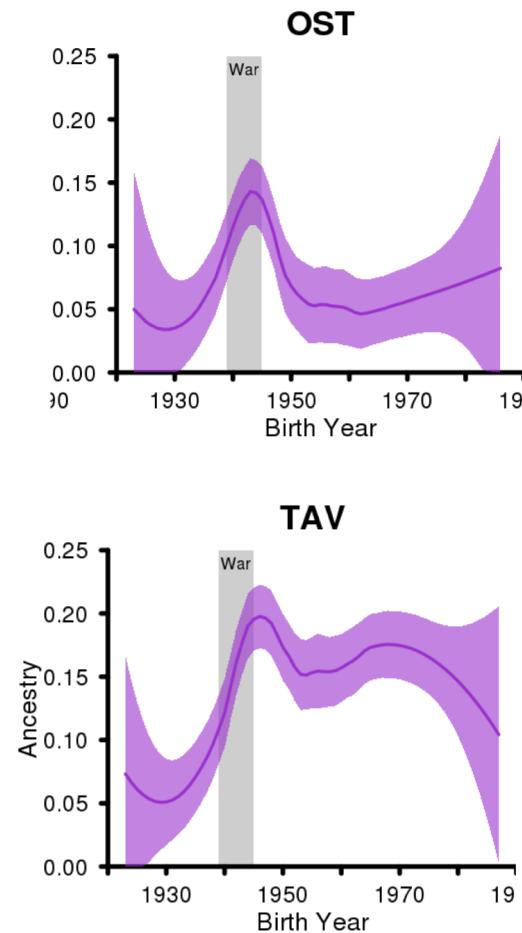


WORLD WAR II EVACUEES

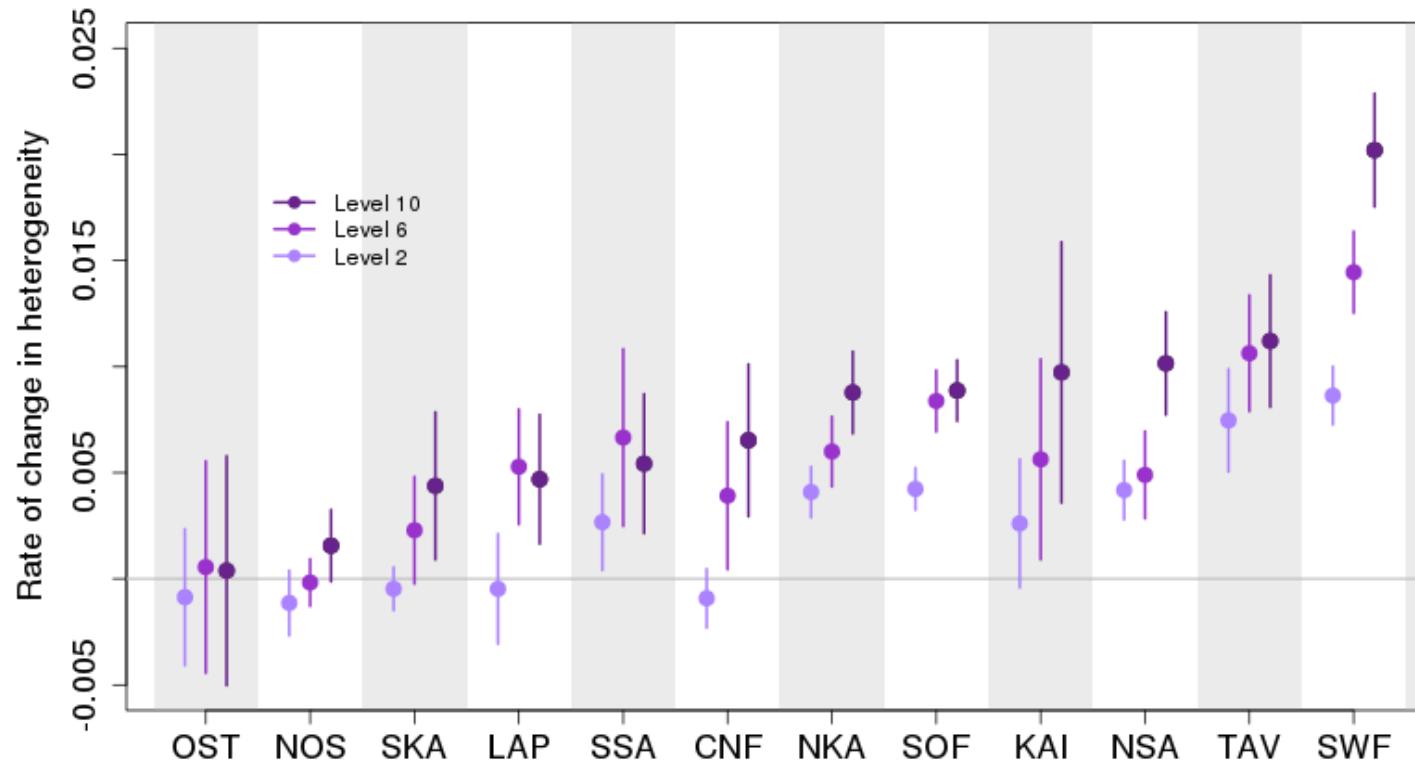
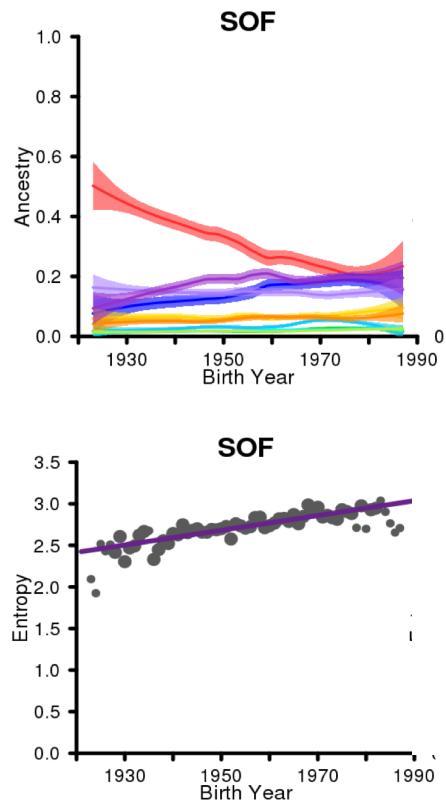
- In the World War II, Finland lost parts from East to the Soviet Union
- Over 400,000 Karelians were relocated from ceded areas
- Evacuations happened between 1939 and 1945



WORLD WAR II EVACUEES



INCREASING HETEROGENEITY – SIGN OF URBANIZATION?

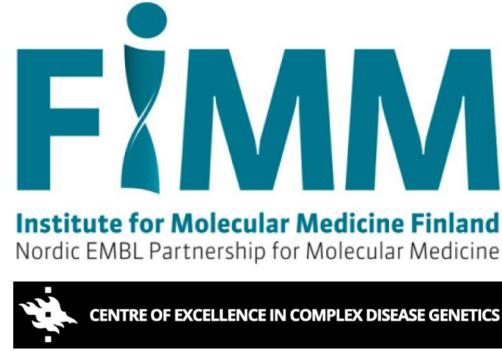


SUMMARY

- Workflow for individual-level genetic ancestry
 - Eastern/Western ancestry can be detected from 4 generation back (~6%), in more detail, ancestry can be identified 2 generations back (25%)
 - We detect clear regional changes in the ancestry profiles during the 20th century that match well with the known demographic events
-
- Great example of the gradually changing and mixing nature of real world populations
- Works as a basis for the future analyses of genetic ancestry within Finland



AKNOWLEDGEMENTS



Matti Pirinen
Samuli Ripatti
Aki Havulinna
Mark Daly



Veikko Salomaa
Markus Perola
Pekka Jousilahti



Rupesh Vyas
Darius Pacaukas
Nicola Cerioli

