

GENETIC STRUCTURE OF HUMAN POPULATIONS IN GREAT BRITAIN

Introduction

As we've seen several times in this course, the amount of genetic data available on humans is vastly greater than what is available for any other organism. As a result, it's possible to use these data to gain unusually deep insight into the recent history of many human populations. Today's example comes from Great Britain, courtesy of a very large consortium [1]

Data

- 2039 individuals with four grandparents born within 80km of one another, effectively studying alleles sampled from grandparents (ca. 1885).
- 6209 samples from 10 countries in continental Europe.
- Autosomal SNPs genotyped in both samples (ca. 500K).

Results*

Very little evidence of population structure within British sample

- Average pairwise F_{ST} : 0.0007
- Maximum pairwise F_{ST} : 0.003

Individual assignment analysis of genotypes using `fineSTRUCTURE`. Same principle as `STRUCTURE`, but it models the correlations among SNPs resulting from gametic disequilibrium, rather than treating each locus as being independently inherited. The analysis is on *haplotypes* rather than on alleles. In addition, it clusters populations hierarchically (Figure ??)

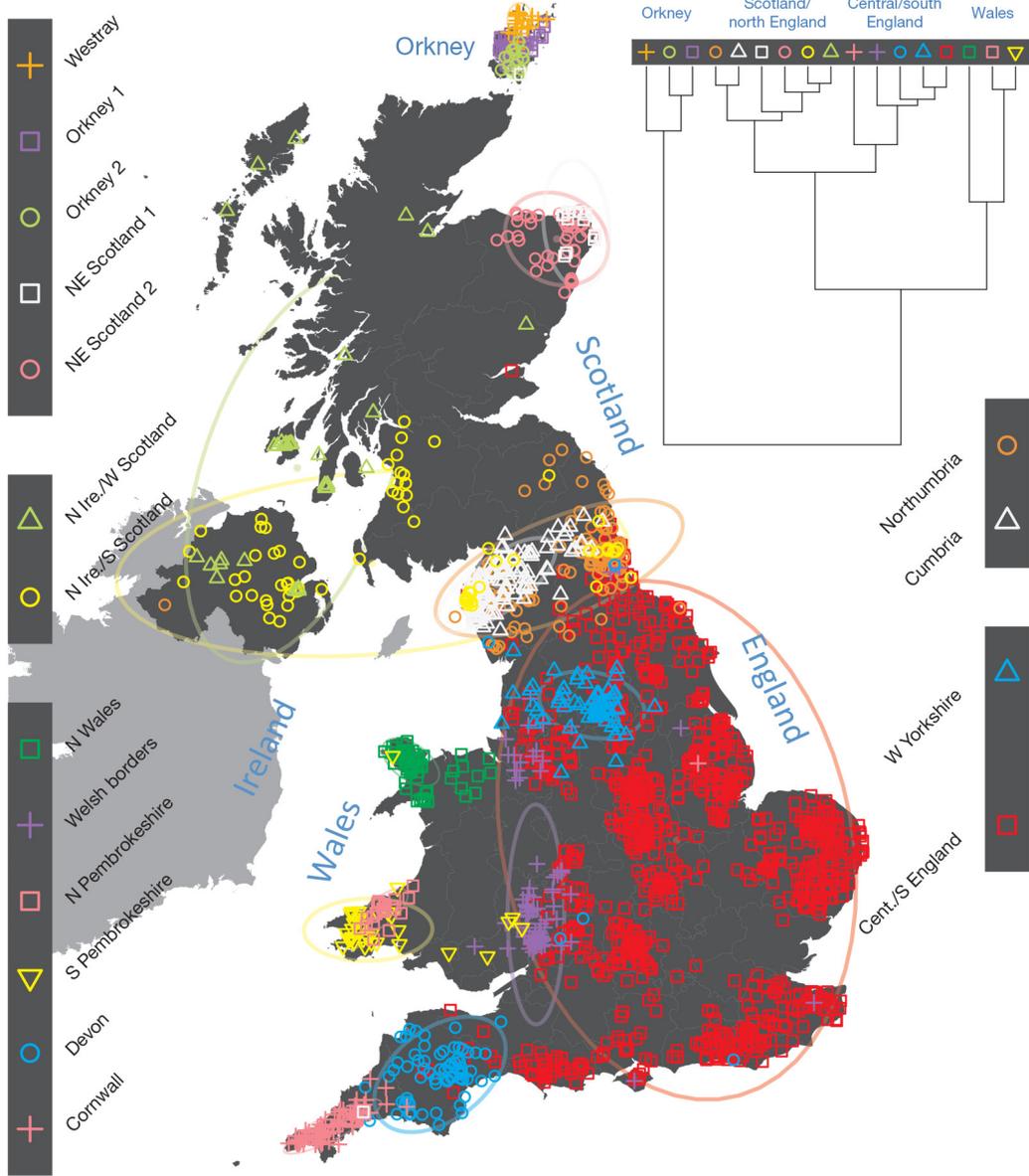


Figure 1: fineSTRUCTURE analysis of genotypes from Great Britain (from [1]).

Analysis of the European data identifies 52 groups. The authors used **Chromopainter** to construct each of the haplotypes detected in their sample of 2039 individuals from the UK as a mosaic of haplotypes derived from those found in their sample of 6209 individuals from continental Europe. Since they know (a) the UK cluster to which each UK individual belongs and (b) the European group from which each individual contributing to the UK mosaic belongs they can estimate (c) the proportion of ancestry for each UK cluster derived from each European group. The results are shown in Figure 2

References

- [1] Stephen Leslie, Bruce Winney, Garrett Hellenthal, Dan Davison, Abdelhamid Boumertit, Tammy Day, Katarzyna Hutnik, Ellen C Royrvik, Barry Cunliffe, Consortium Wellcome Trust Case Control, Consortium International Multiple Sclerosis Genetics, Daniel J Lawson, Daniel Falush, Colin Freeman, Matti Pirinen, Simon Myers, Mark Robinson, Peter Donnelly, and Walter Bodmer. The fine-scale genetic structure of the British population. *Nature*, 519(7543):309–314, 2015.

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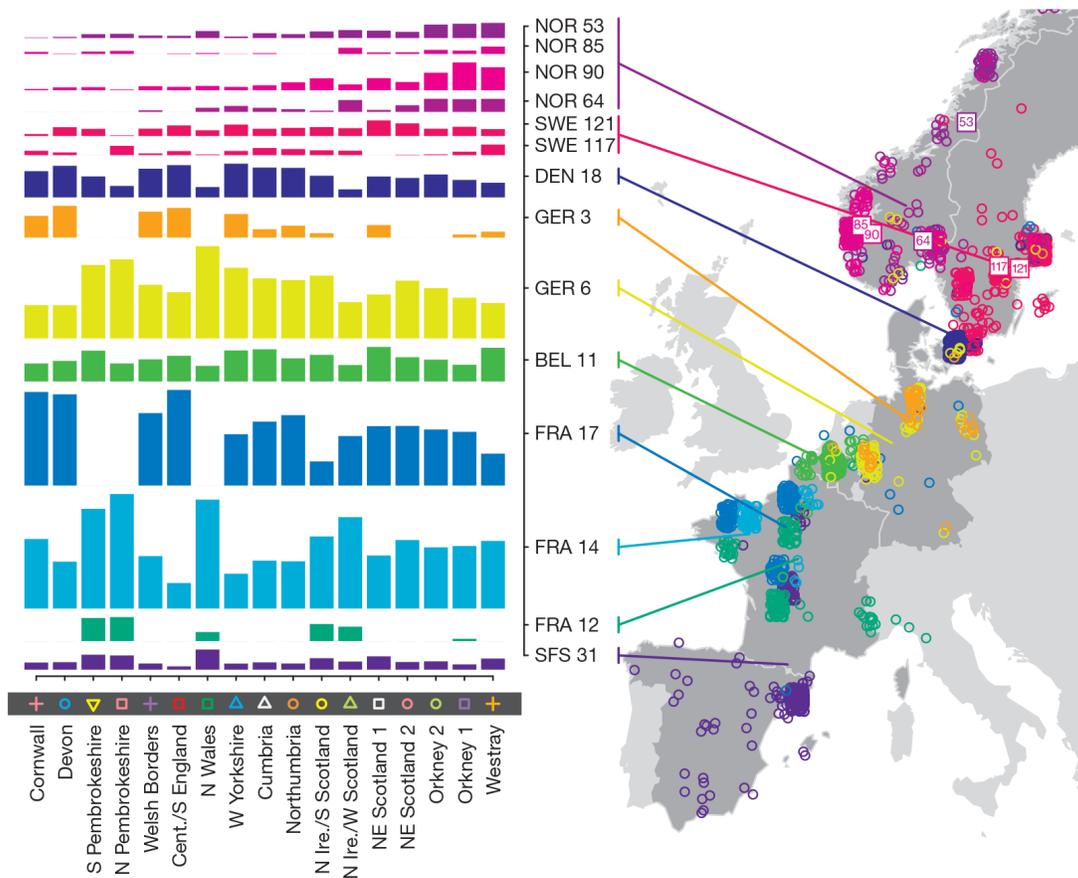


Figure 2: European ancestry of the 17 clusters identified in the UK (from [1]).