Practical: Changes of Allele and Genotype Frequencies

Main evolutionary forces

1. Mutation / Migration: new alleles in the population

2. Natural selection (fitness)

3. Genetic drift (Wright-Fisher's model of neutral evolution)

4. Demography: bottleneck / expansion

Main evolutionary forces affecting allele frequencies

1. Mutation / Migration: new alleles in the population

2. Natural selection (fitness)

3. Genetic drift (Wright-Fisher's model of neutral evolution)

4. Demography: bottleneck / expansion

Wright's Fixation Index (F_{ST})

Population structure = frequency differences between populations

Ancestral population

p_0

p_1

p_2

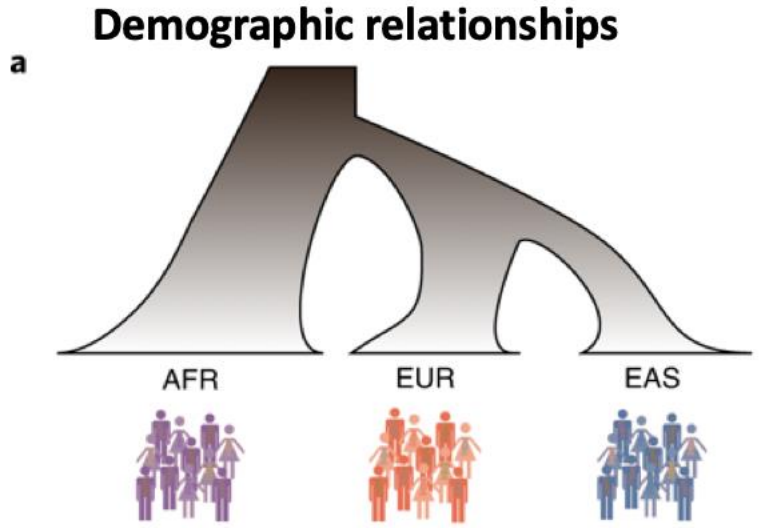
p_K

Derived population 1

Derived population 2

Derived population K

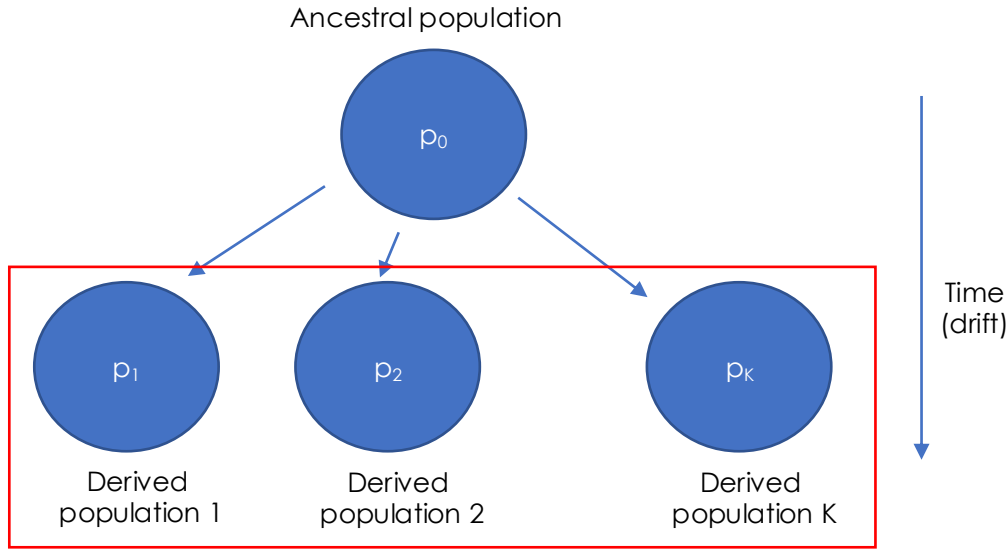
Time (drift)



Martin et. al., *Nature Genetics*, 2019

Many definitions and many estimators

- F_{ST} as a ratio of variance (Wright):
  $F_{ST}=var_B(X)/[var_B(X)+var_W(X)]$; X=0,1 (allelic state)
  F_{ST}=0: no frequency differences between populations
  F_{ST}=1: allele is fixed in one population (fixation index)



Estimators
Weir & Cochram (1984)
Nei (1973)
Hudson (1992)

Weir & Hill (2002) definition
$var(p_i | p_0) = F_{ST}\, p_0\, (1-p_0)$

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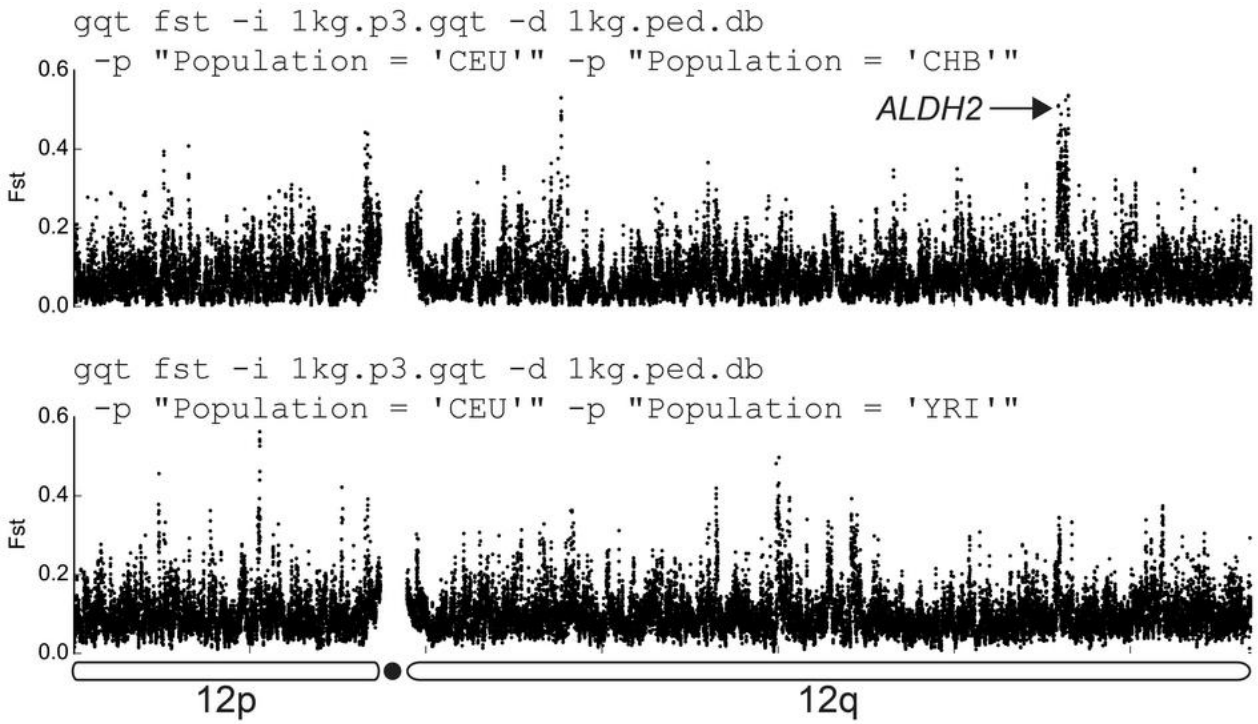
Genome Res. 2013 Sep; 23(9): 1514–1521.
doi: 10.1101/gr.154831.113
PMCID: PMC3759727
PMID: 23861382

Estimating and interpreting F_{ST}: The impact of rare variants

Gaurav Bhatia,[1,2,6,7] Nick Patterson,[2,6,7] Sriram Sankararaman,[2,3] and Alkes L. Price[2,4,5,7]

F_{ST} varies across the genome => *not just drift at play here!*



Layer R.M. et al. Efficient genotype compression and analysis of large genetic-variation data sets. *Nature Methods* (2016).

Typical F_{ST} ranges

F_{ST} is ~0.1- 0.2 between continental groups

F_{ST} is ~0.01- 0.05 within continental groups

F_{ST} is <0.01within countries