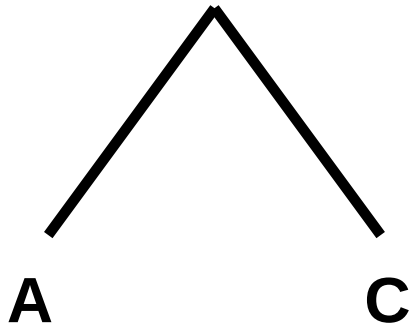
Quantifying relationships among populations: Simple tests

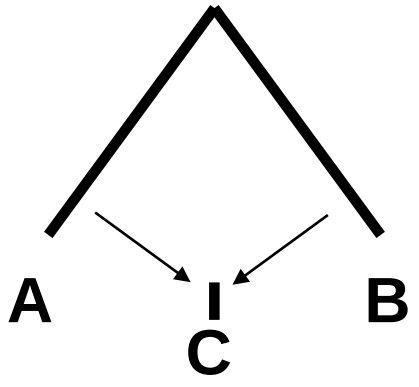
Andrea Manica

UNIVERSITY OF CAMBRIDGE

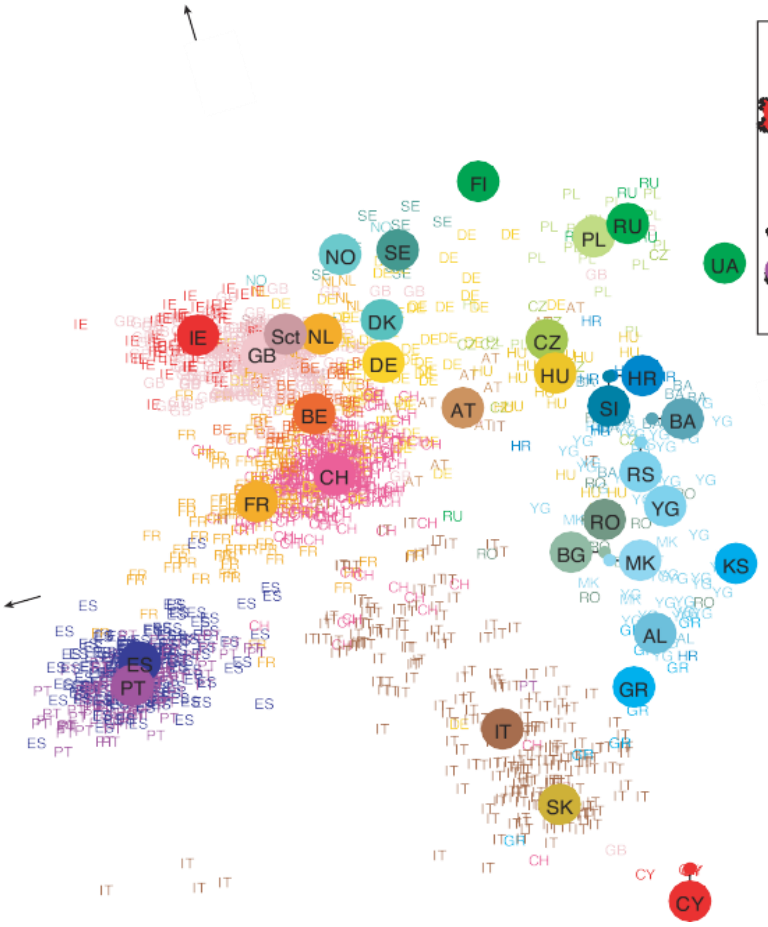
Outline

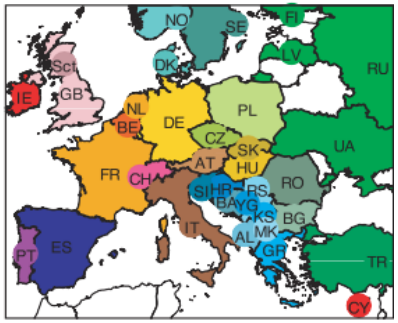
- Using F statistics (measure of drift) to compare populations
- Relationships among populations on a simple tree
- Admixture





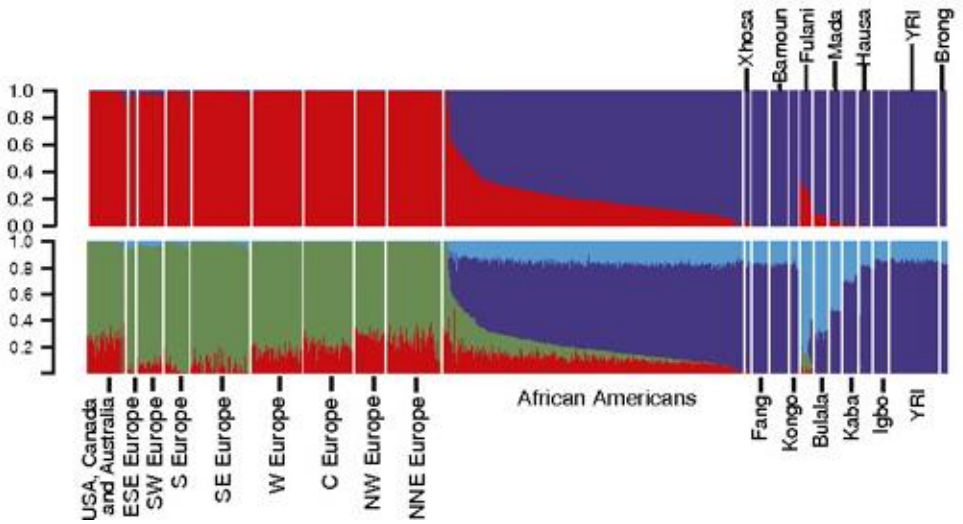
Relationships among populations





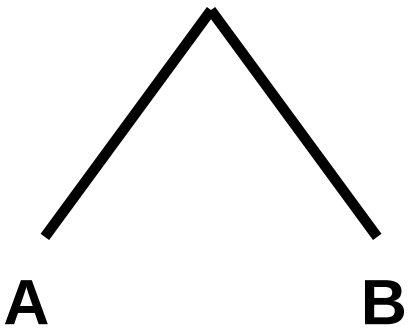
Proximity in PCA

Clustering algorithms (Garrett's lectures)



Outgroup f_3 to define similarity

$$F_2(A,B) = E[(p_A - p_B)^2]$$



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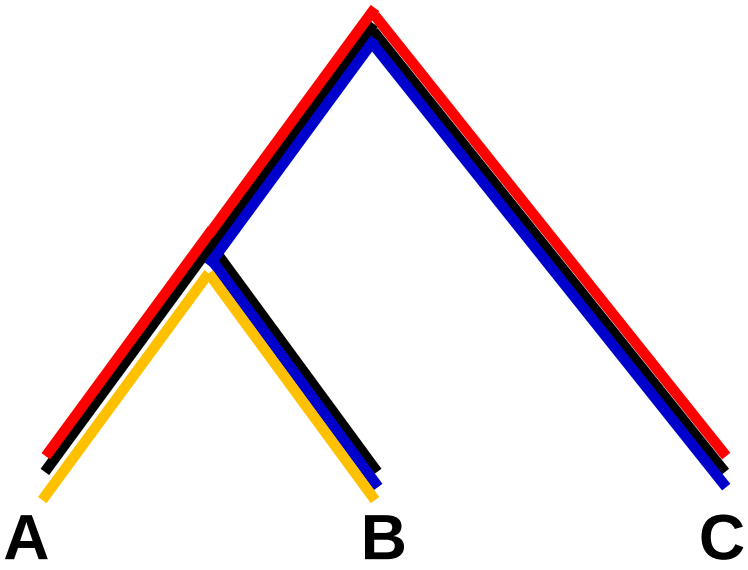
$$F_2(A,B) = E[(p_A - p_B)^2]$$

$$F_3(C; A,B) = E[(p_C - p_A)\,[(p_C - p_B)] =$$

$$= \tfrac{1}{2}\,[F_2(C,A) + F_2(C,B) - F_2(A,B)]$$



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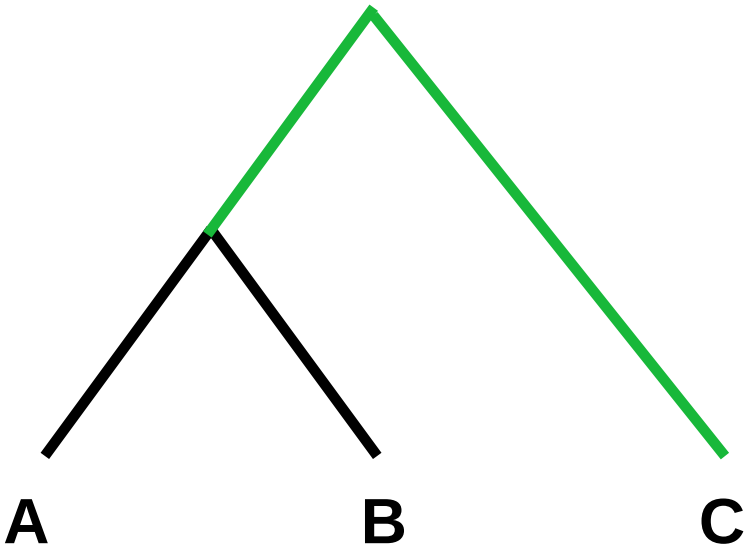
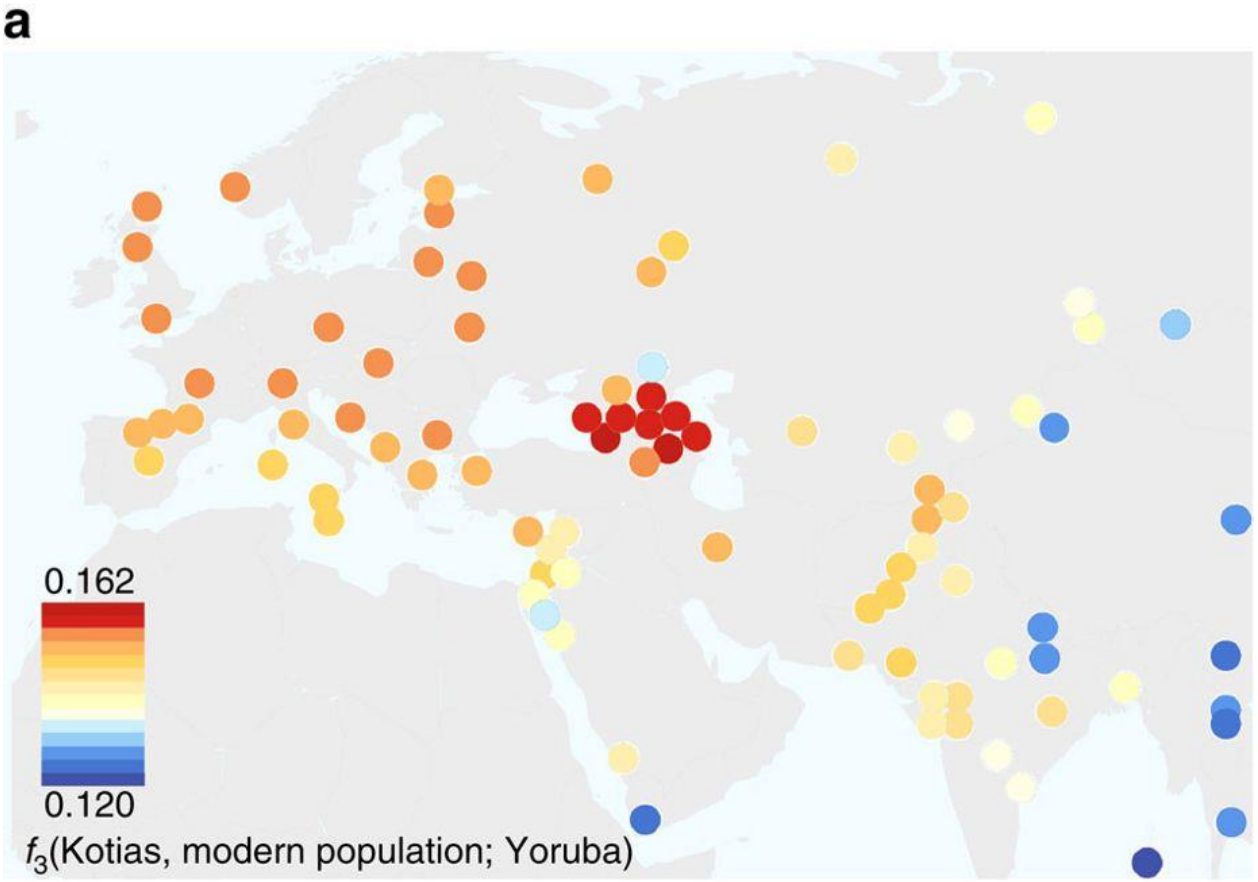


Shared drift for A and B from C

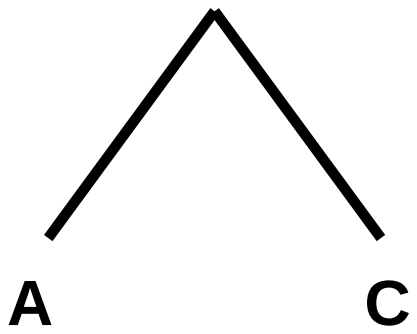
Overlap of paths C->A and C->B

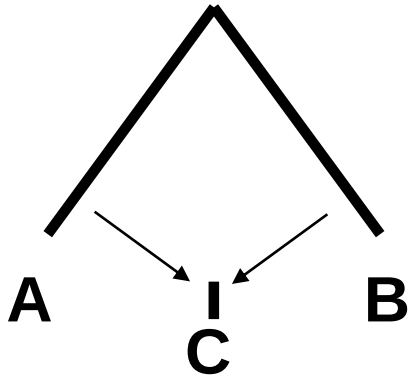
Outgroup f_3 to define similarity



Admixture f_3 if topology is not a simple tree

$$F_2(C,A) = E[(p_C - p_A)^2] = E[(p_C - p_A)\,(p_C - p_A)]$$





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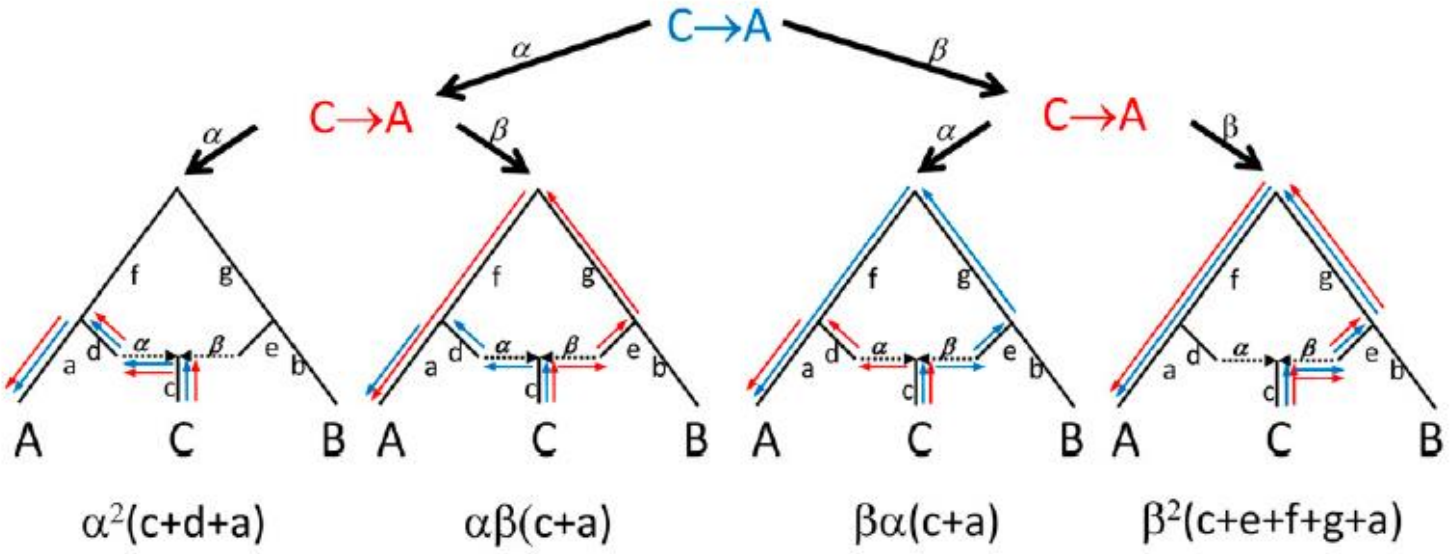
$$F_2(C,A) = E[(p_C-p_A)^2] = E[(p_C-p_A)\,(p_C-p_A)]$$

$$F_2(C,A) = a + c + \alpha^2 d + \beta^2(e+g+f)$$

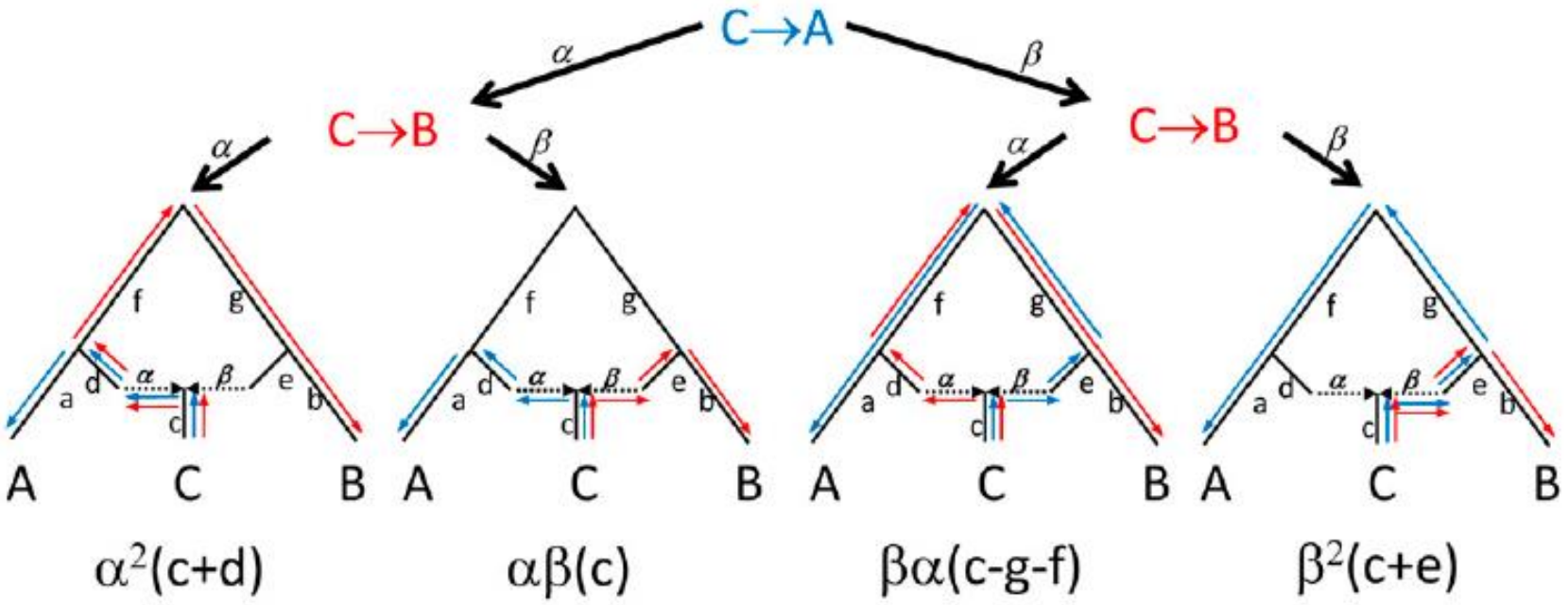


Tip: F statistics can be seen as overlap of paths from one sample to the other in the quadratic term

Admixture f_3 if topology is not a simple tree

$$F_3(C; A,B) = E[(p_C - p_A)\,[(p_C - p_B)] =$$

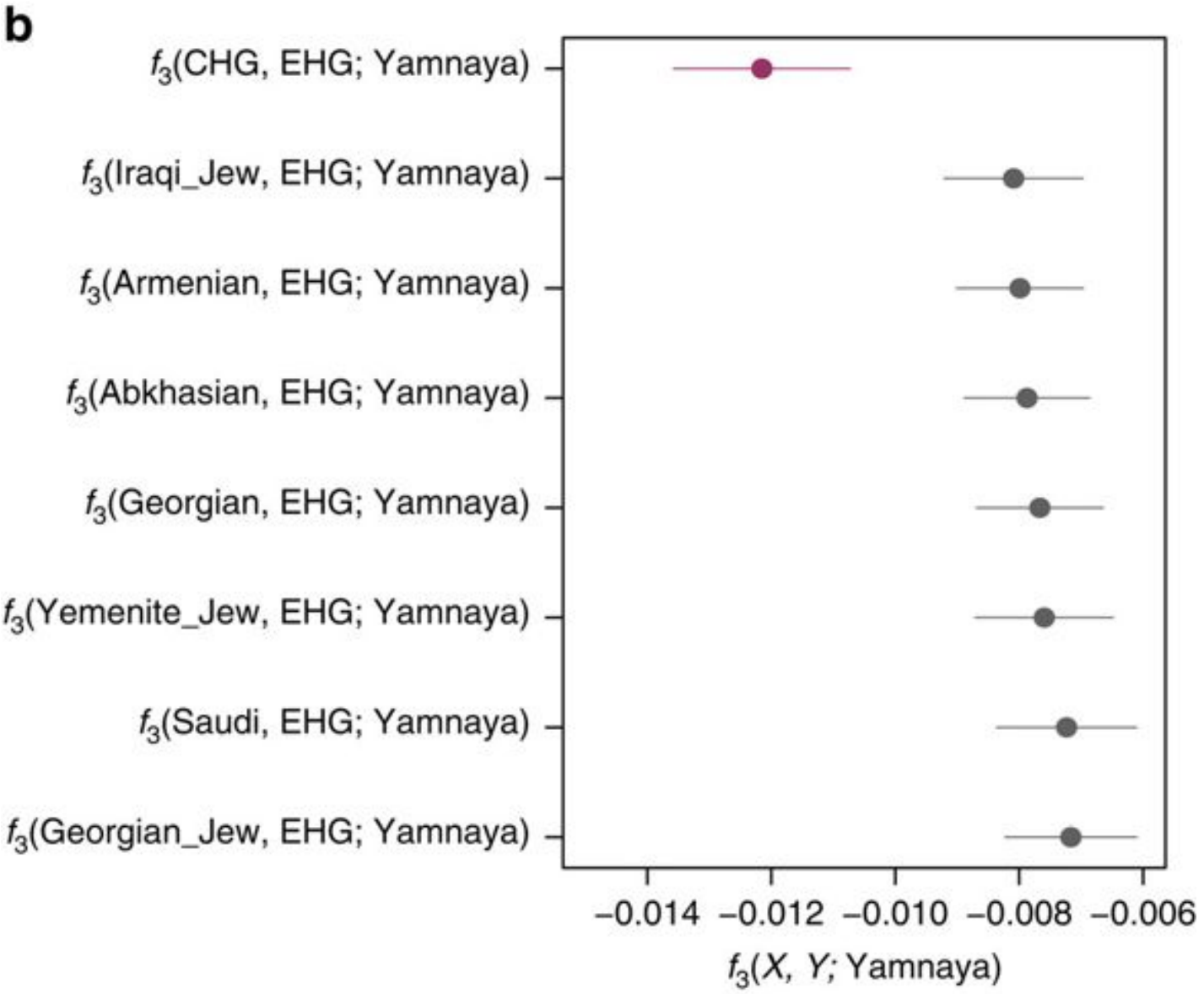
$$F_3(C;A,B) = c + \alpha^2 d + \beta^2 e - \alpha\beta(g+f)$$



Negative f_3 is a sign of admixture

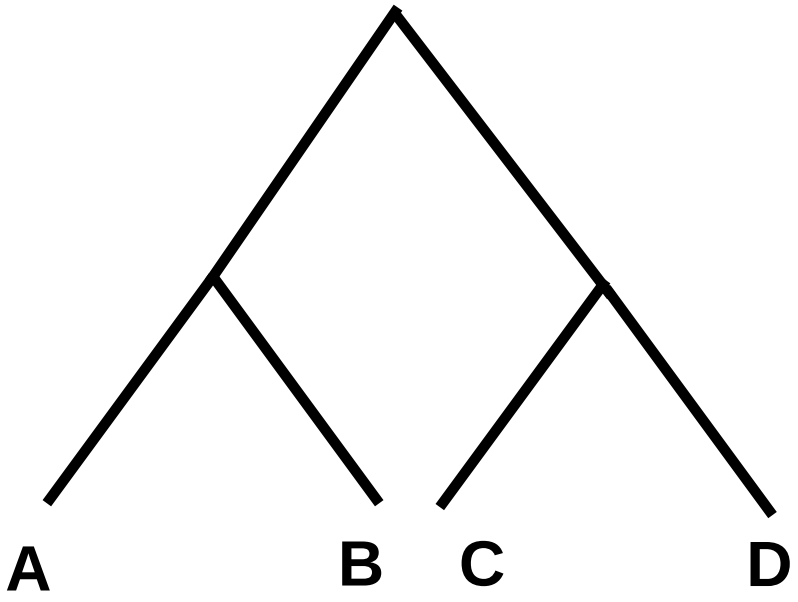
But note that a positive f_3 does not prove absence of admixture (e.g. large *c*, drift to C)

Admixture f_3



f_4 for a simple tree

$$F_4(A,B;C,D) = E[(p_A - p_B)\,[(p_C - p_D)]] =$$



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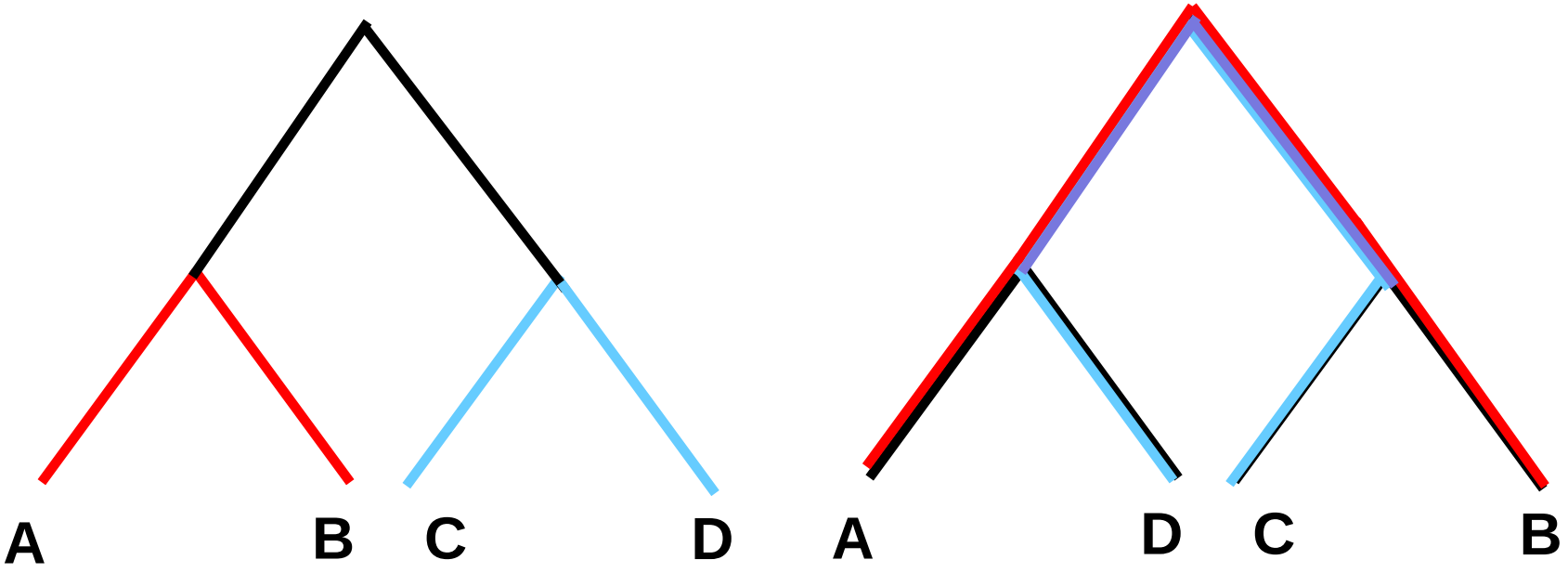
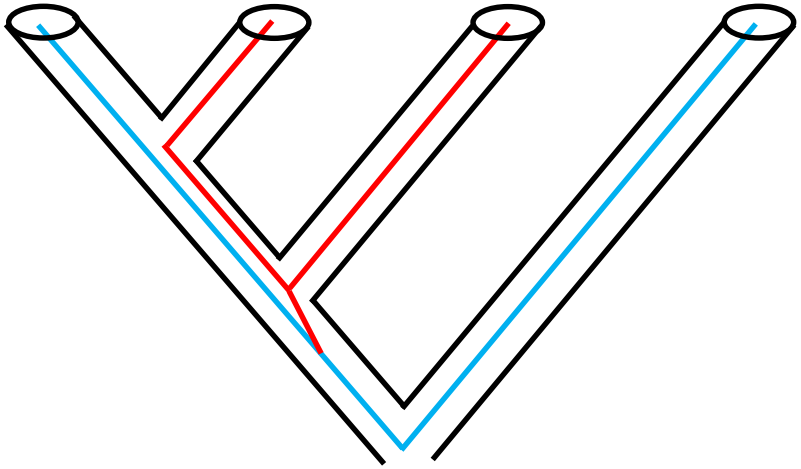


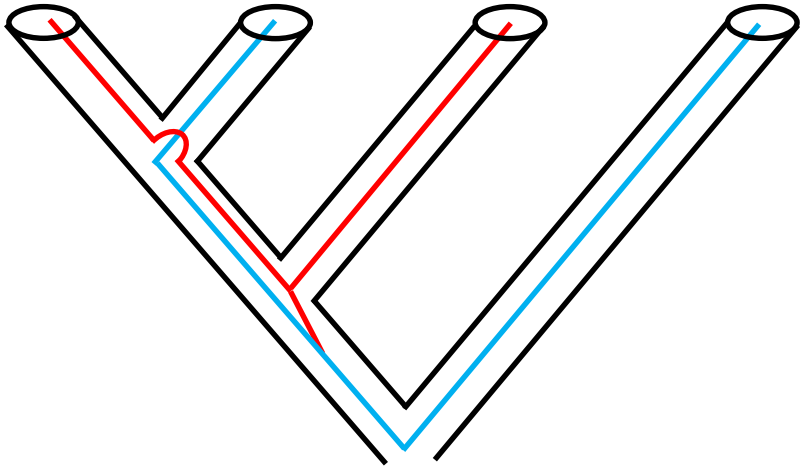
Expect f_4 to be zero if the two pairs form clades with respect to each other

D-statistics (ABBA-BABA)

$$f_4 = \frac{\#ABBA - \#BABA}{\#sites}$$

$$D = \frac{\#ABBA - \#BABA}{\#ABBA + \#BABA}$$





D(O,P_1; P_2,P_3)

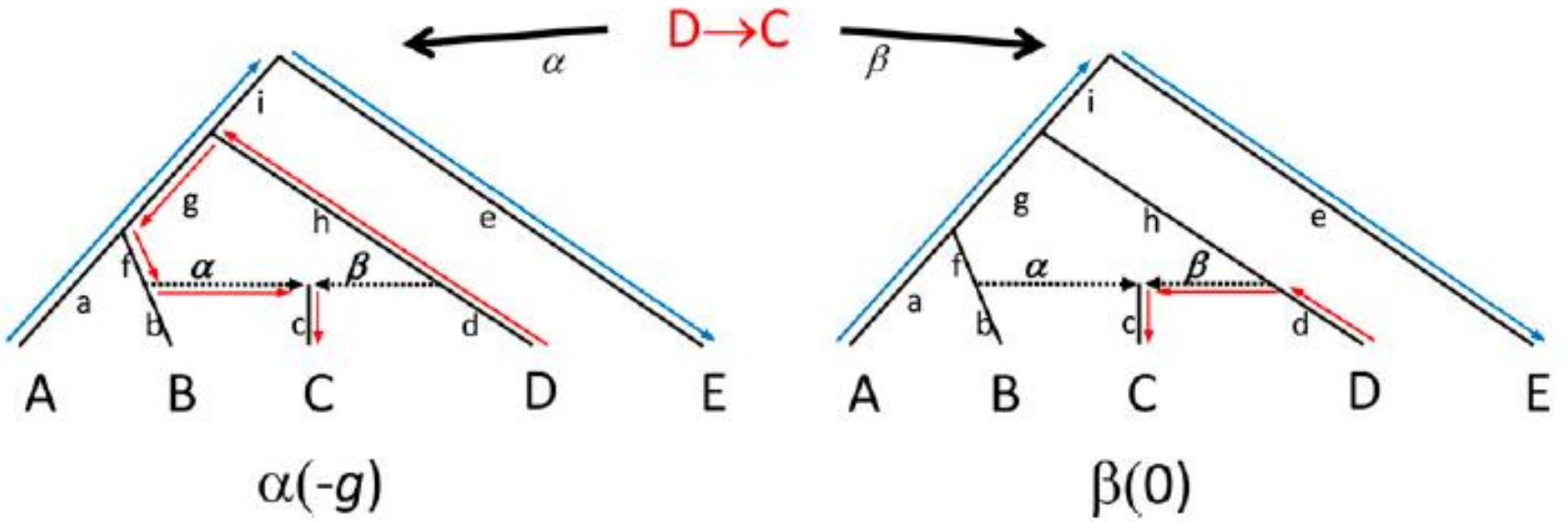
D = 0 true phylogeny

D < 0 gene flow P_1&P_2

D > 0 gene flow P_1&P_3

f_4 if topology is not a simple tree

$F_4(A,E;D,C) = -\alpha g$



A & E set the background, B & D are the sources

Testing significance of relationships

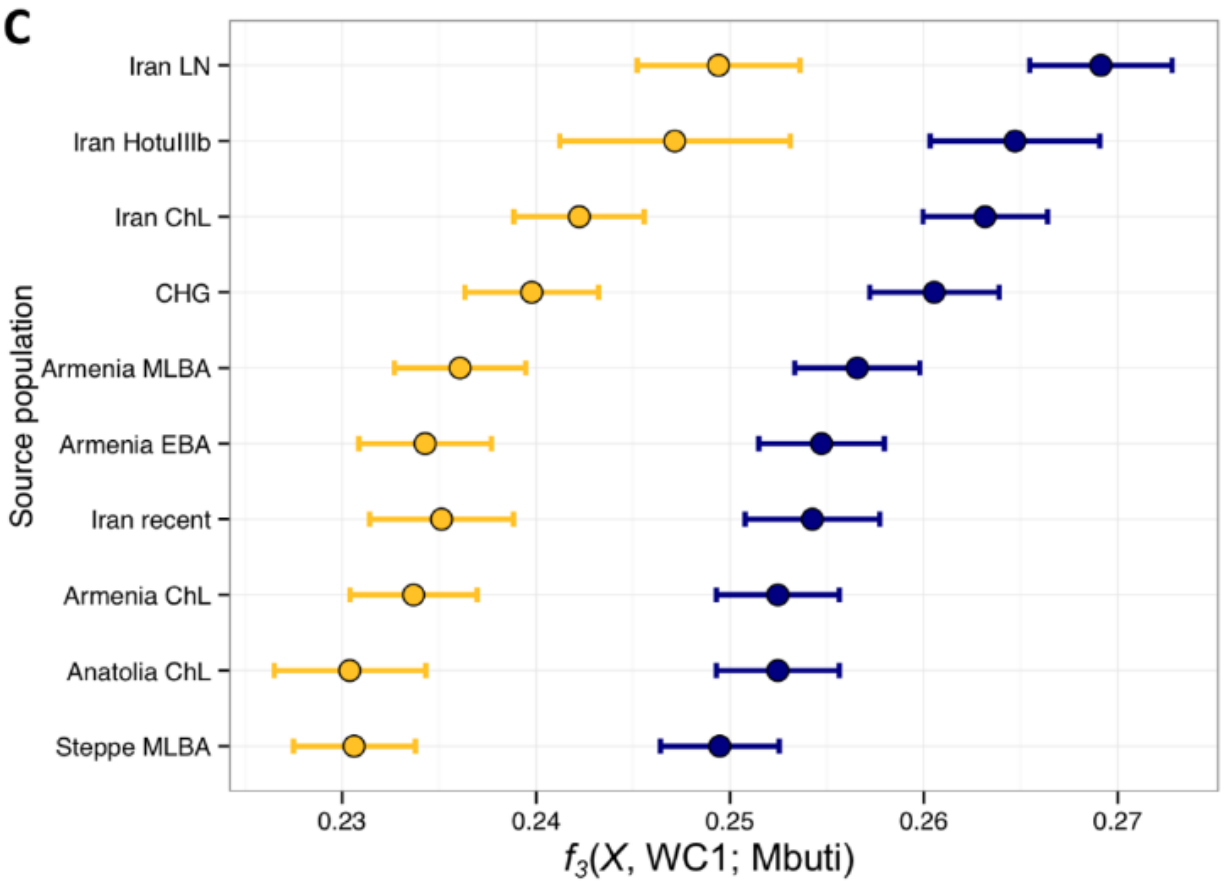
We can test significant by block jackknifing (usually using 5cM)

Estimate F_x and its standard error, and then compute a standardized Z

$|Z| > 2$ considered significant (but note that the critical value has been changing… not always for the right reasons)

Be careful that you are not comparing apples and oranges (SNPs from the same panel)

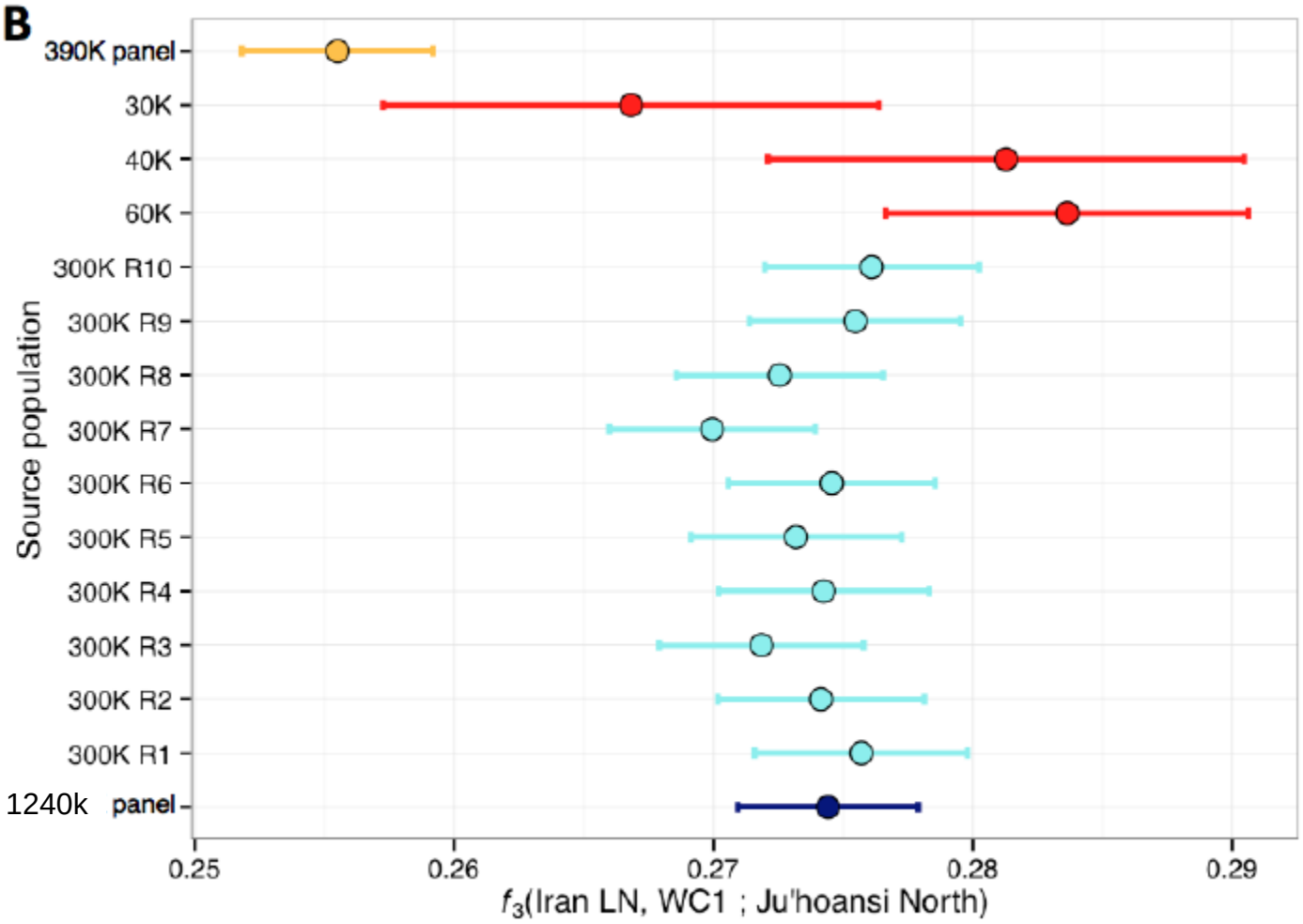
Ascertainment and F stats



Blue: 1240k SNP panel

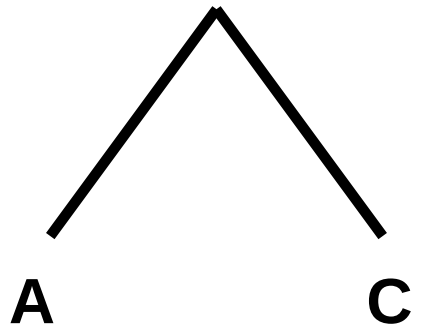
Yellow: 390k SNP panel (subset of 1240k)

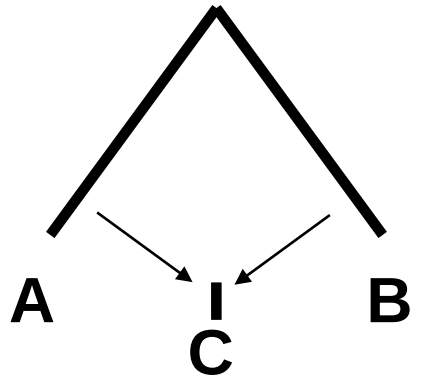
Ascertainment and F stats



Summary

- Using F statistics (measure of drift) to compare populations
- Relationships among populations on a simple tree
- Admixture





Practical

- Use Admixtools to compute different F stats and interpret them
- Human dataset with modern and ancient
- Commands and questions in partical1.sh