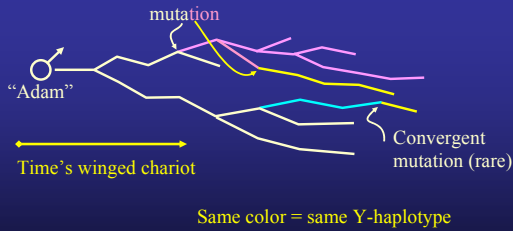


Y-chromosome lineage



Y haplotype evolutionary history

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Convergent Y mutation

- Y haplotype = 17 numbers = position in 17-space
- Mutation is random walk in 17 dimensions
 - Each step is +1 or -1 in some dimension.
- Random walks rarely return to start.
 - $\text{Pr}(\text{return at } 2^{\text{nd}} \mu \cdot n) = 1/(17 \cdot 2) = 3\%$
 - $\text{Pr}(\text{return at } 4^{\text{th}} \mu \cdot n) = 2/34^2 = 0.2\%$
 - $\text{Pr}(\text{return at } 6^{\text{th}} \mu \cdot n) = 10/34^4 = 0.001\%$
- Identical Y-filer haplotype => relationship to common ancestor without mutations

Y haplotype evolutionary history

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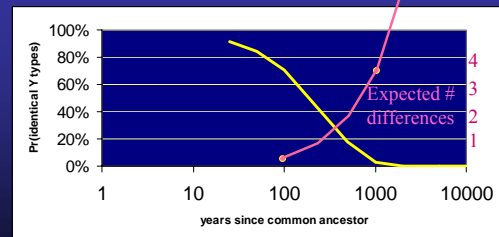
Convergence experiment

- Simulated Y-filer population (N=200000)
- Small proportion of pairwise matches
 - $\text{Pr}(\text{match}) = 1/23000$ (ok, parameters a little off)
- Given match (IBS), are all IBD?
 - $\text{Pr}(\text{IBD} | \text{IBS}) \approx 97\% = 33/34$.
 - Close to computed estimate of non-convergence (previous slide)
 - (But they are not the same experiment.)

Y haplotype evolutionary history

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Y-haplotype divergence



Y haplotype evolutionary history

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Comments on Y evolution

- Expect *no* common haplotypes between populations long separated
- ABI – 8 populations of data, 3561 men, 3347 distinct haplotypes, 3187 unique ones ($\kappa=89.5\%$)
 - 374 men had 160 non-unique haplotypes
 - 145 sets occurred in 2+ different populations
 - 95 were combinations of Cauc, Afr-Am, Hisp
 - 33 further include Native-Am plus the preceding
 - 7 were Asian + Filipino; 2 were Afr-Am + Afr
 - Remaining 8 were: 4 Cauc+Hisp+Vietnamese(!), 2 Afr-Am, 2 Filipino+Vietnamese

Y haplotype evolutionary history

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Comments on crime-suspect match

- If suspect *not* donor, then
 - 97% that suspect and donor are IBD
 - which is very unlikely if they are separated by more than a few centuries.
- A 17-locus haplotype is typically represented by a small number* of men descended without mutation from a common ancestor 200-300 years ago.
 - * 1/10000 of the population

Y haplotype evolutionary history

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