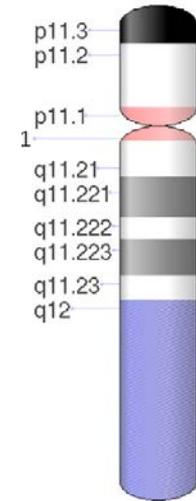


Simplicity can be confusing (understanding the manly chromosome partly through APL)



Y chromosome

Charles Brenner, Ph.D.

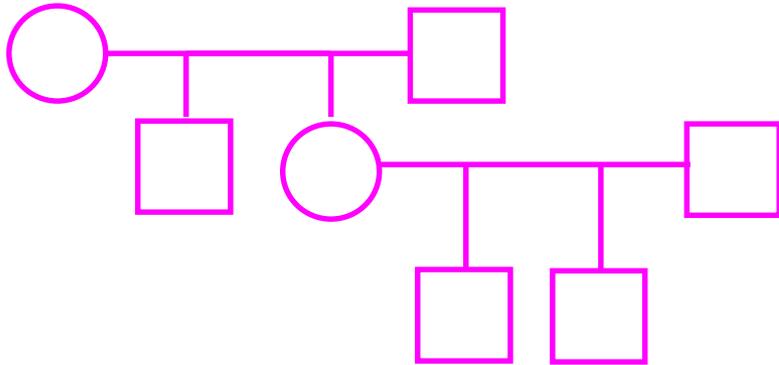
Purveyor of forensic mathematics,

DNA·VIEW®

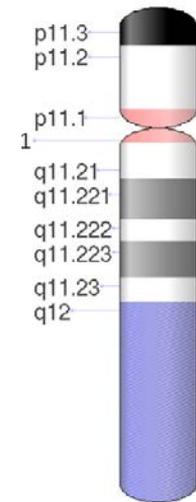
charles@dna-view.com

<http://dna-view.com>

Simplicity can be confusing (understanding the manly chromosome partly through APL)



Autosomal pedigree



Y chromosome

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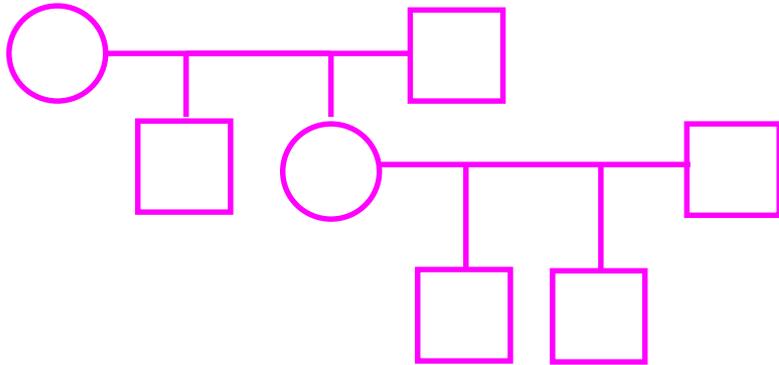
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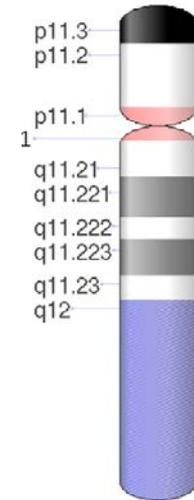
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Forensic DNA problem

- DNA matching Mr. Russell detected on victim.
- Probability of such a match by chance?
- Why Y?
 - Victim DNA may overwhelm assailant DNA.
 - Male-only Y DNA may solve that problem.

Fighting for truth and/or justice

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- I published some papers on Y DNA evidence

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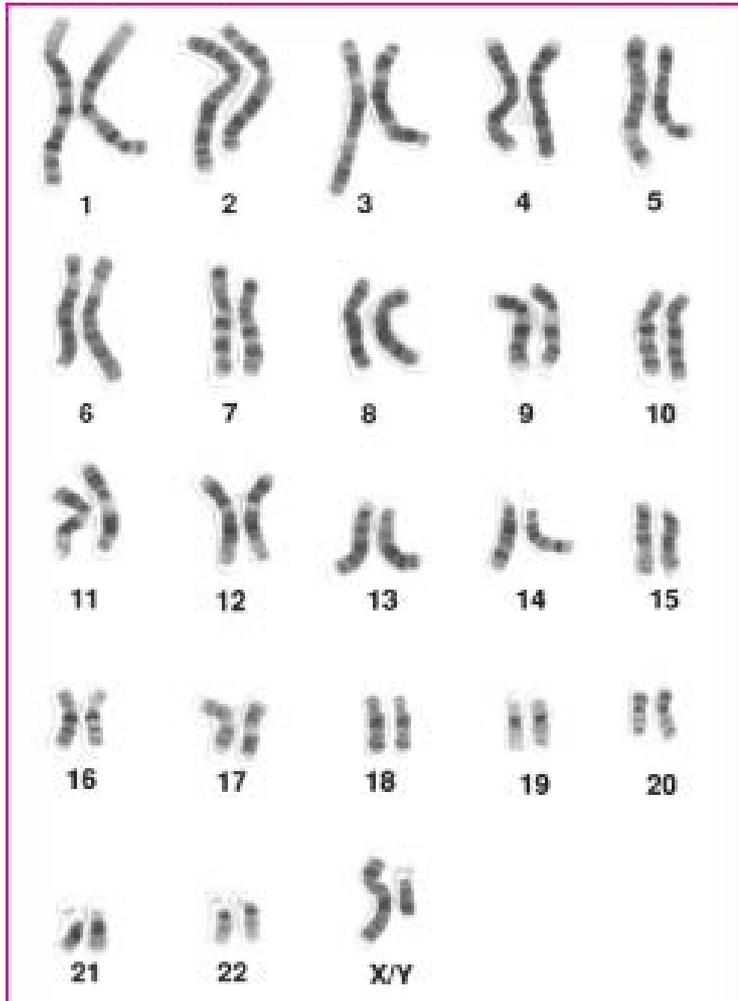
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 - Hopi  & also Tohono O’odham  tribesmen / reservation in Arizona
 - Navajo  in New Mexico
 - Ashkenazi (!) in Boston

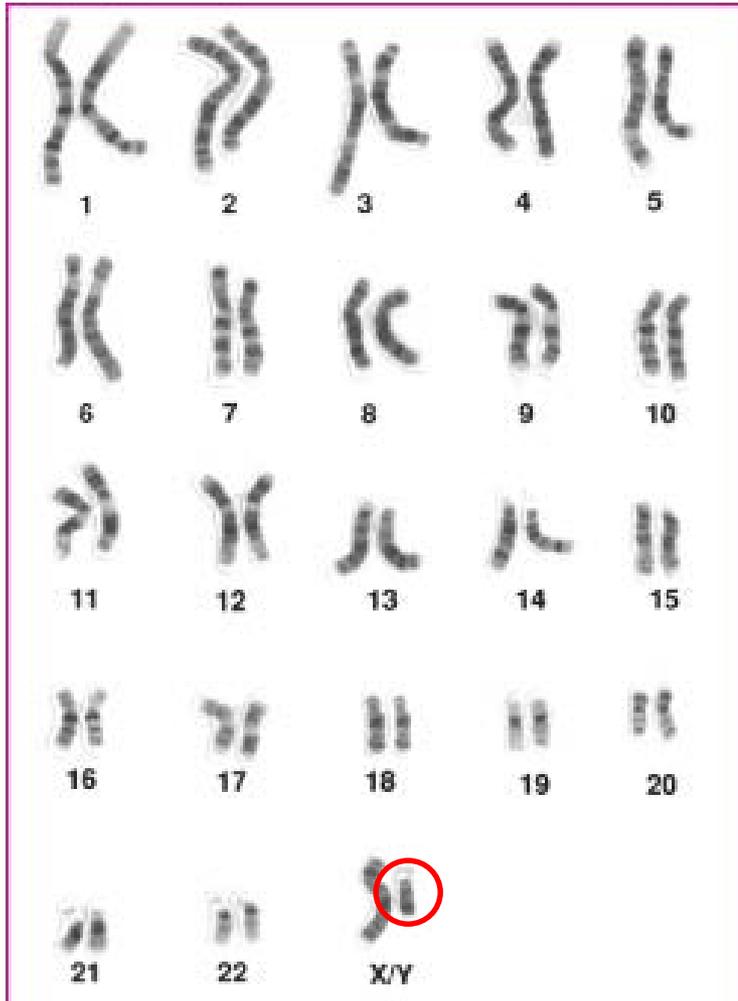
What's Y?

Karyogram of a human male



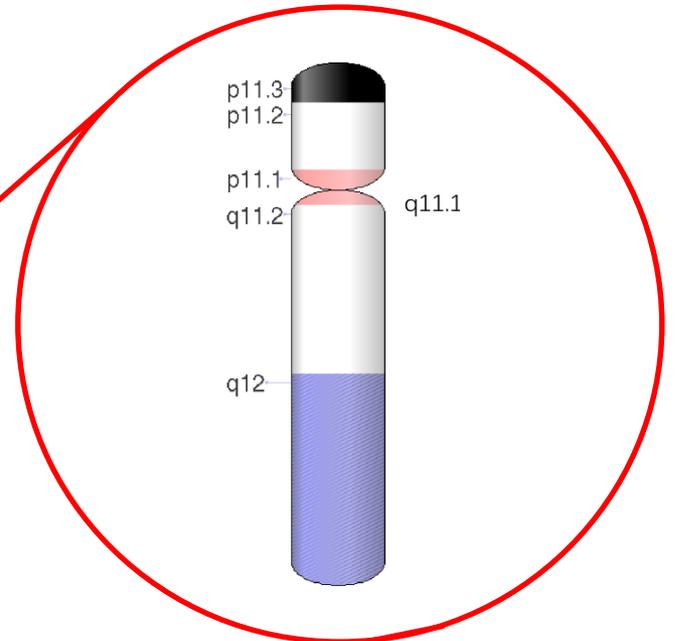
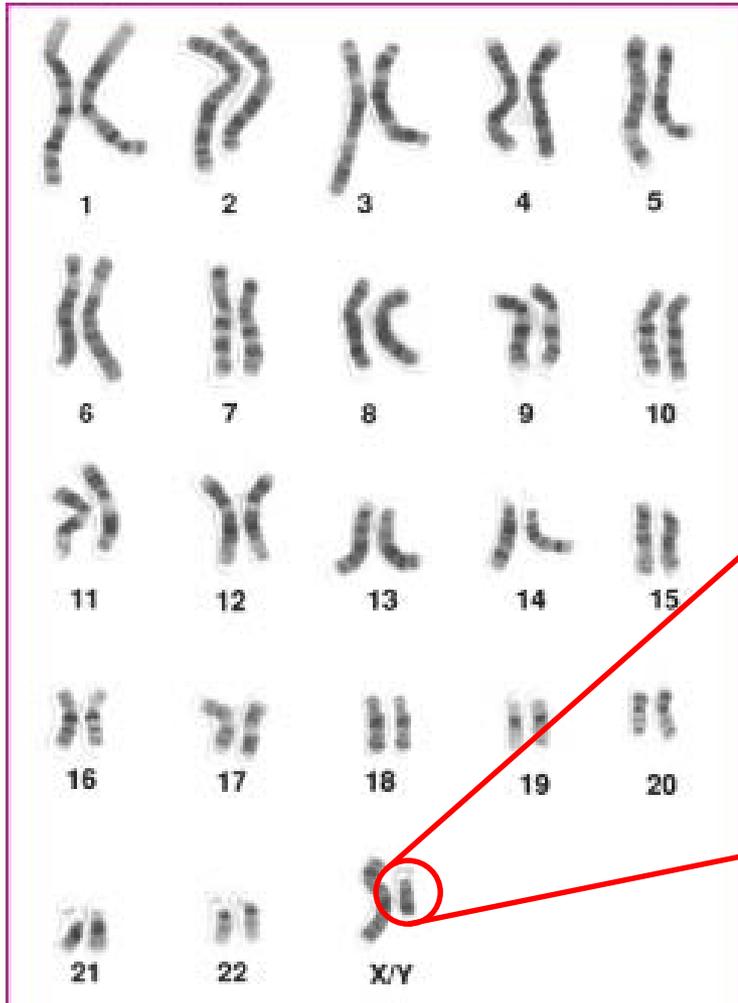
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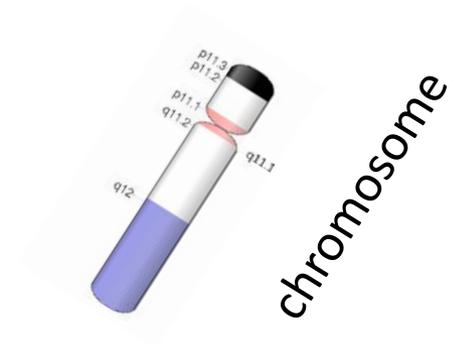
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Karyogram of a human male

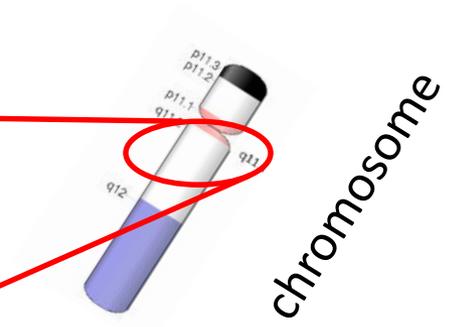
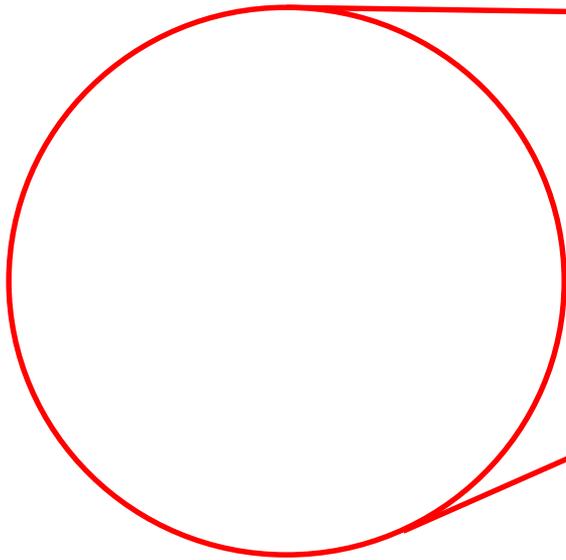


Y chromosome

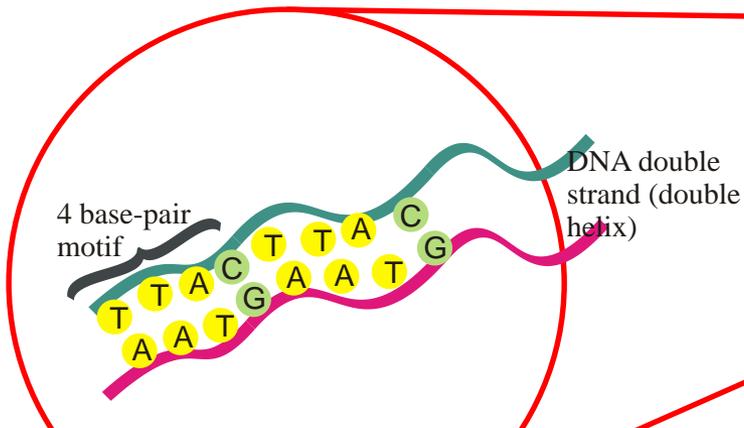
Typical identification locus within a chromosome



Typical identification locus within a chromosome



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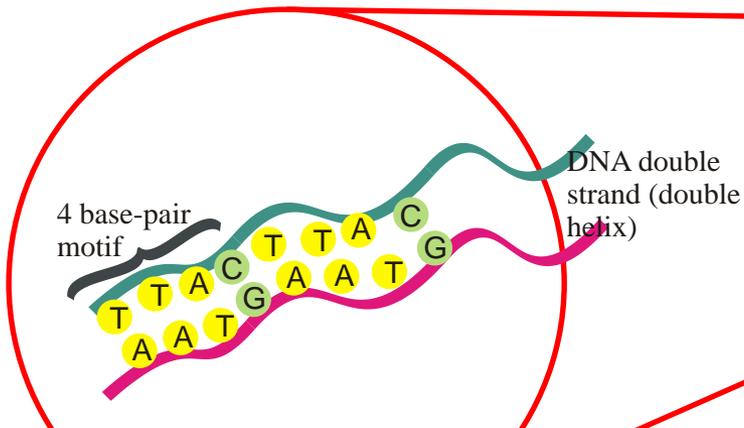
chromosome

Electron microphotograph

showing 2 repeats of the motif.

Number of repeats **varies** between people thanks to occasional replication slippage mutations over the eons.

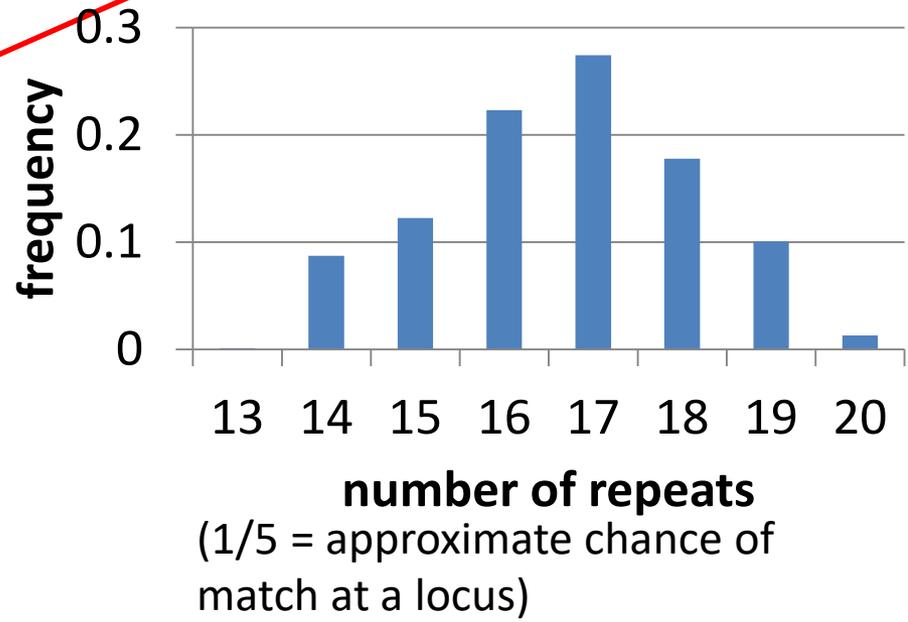
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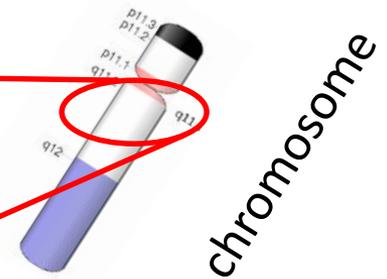
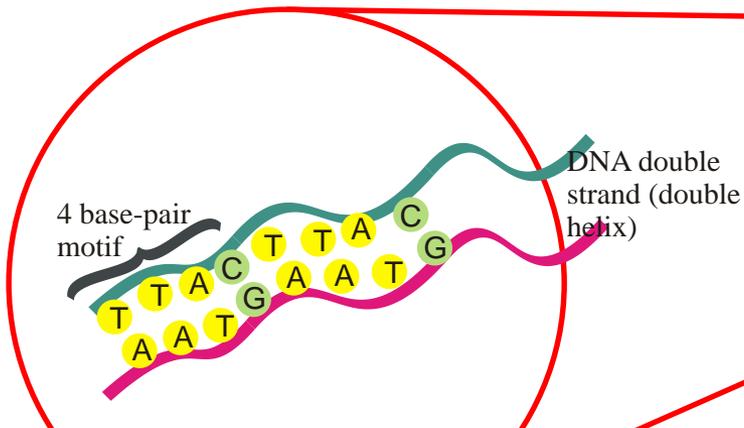
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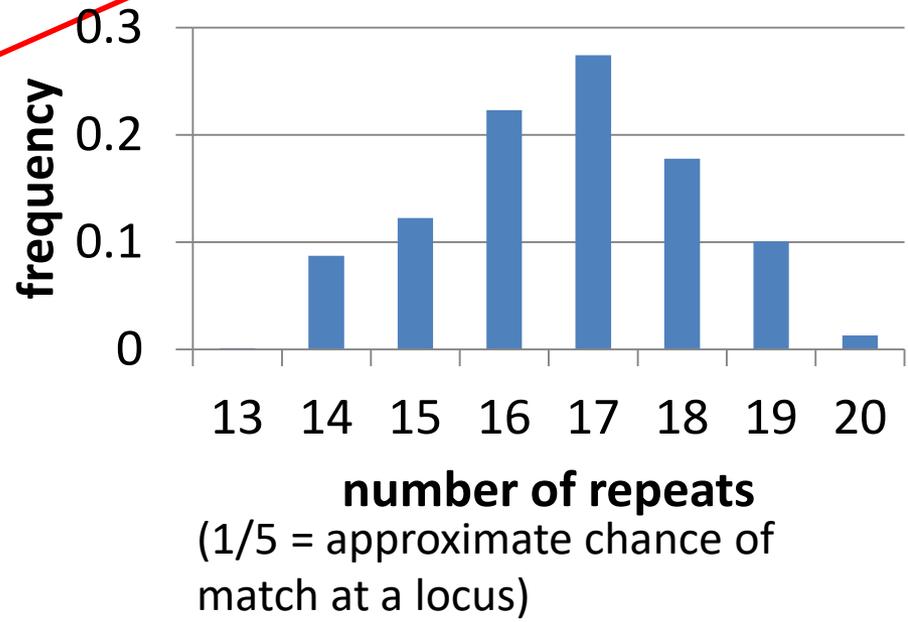


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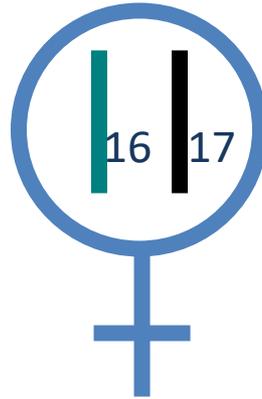
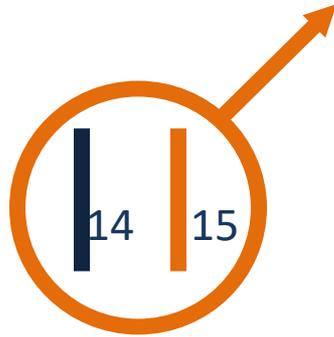
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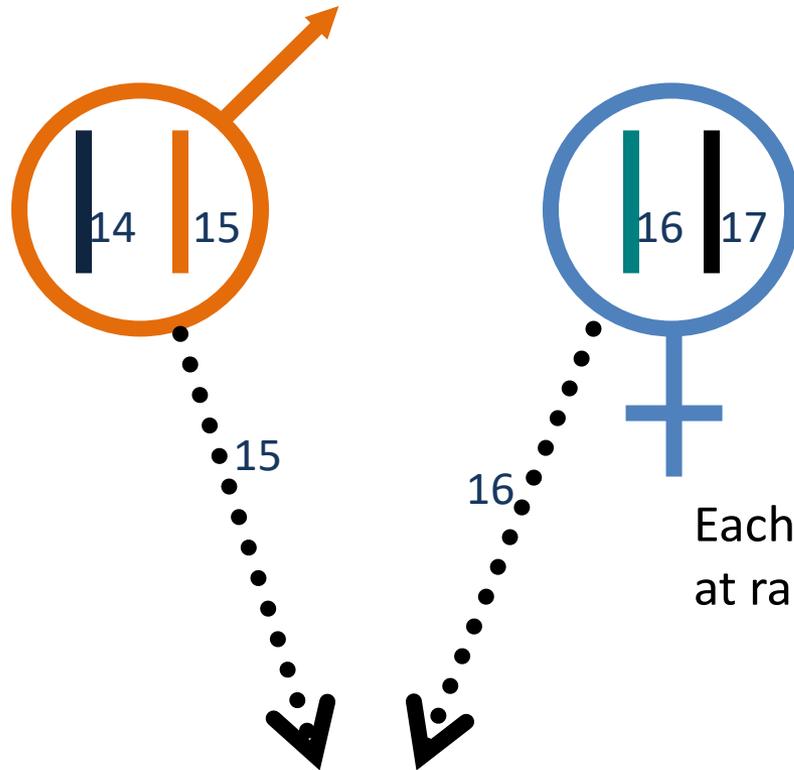
Forensic evidence: Suspect **allele** at locus matches crime scene allele
Evidential value: 5x more likely if suspect is the donor, than if not.
(NB: The cumulative evidence from 10 to 30 alleles can be very strong.)

Genetic inheritance (sexual), one locus



Each parent has two D12 chromosomes, hence two vWA alleles – e.g. {14,15} and {16,17}

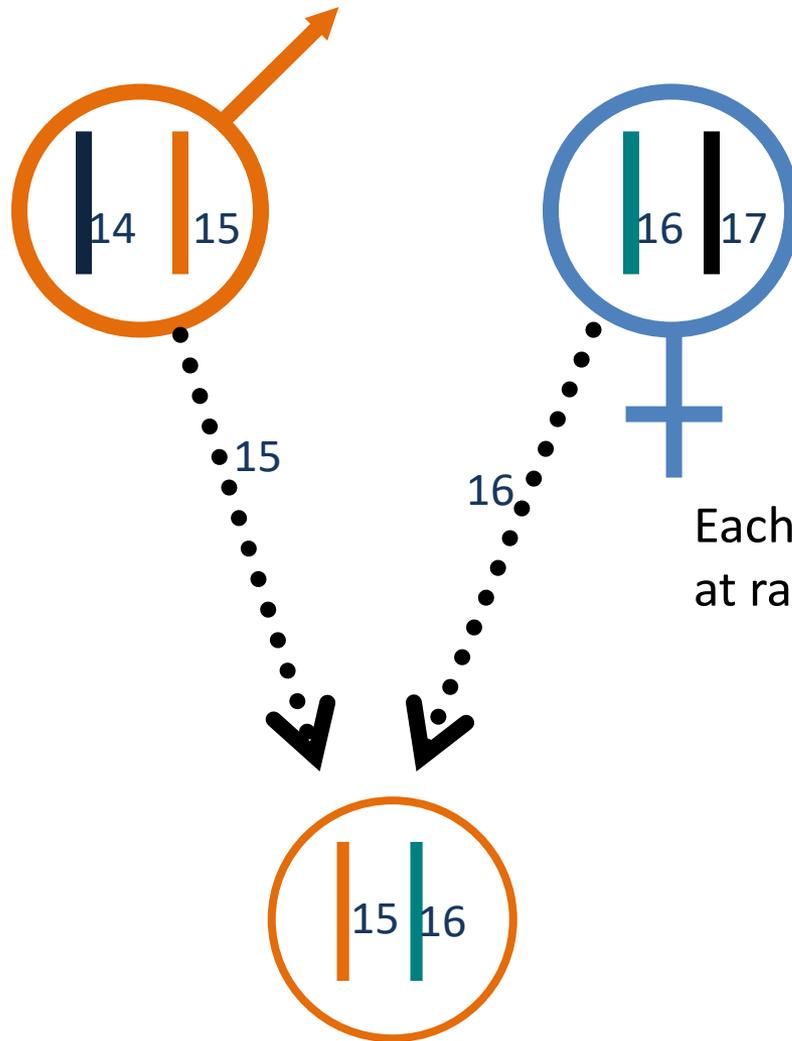
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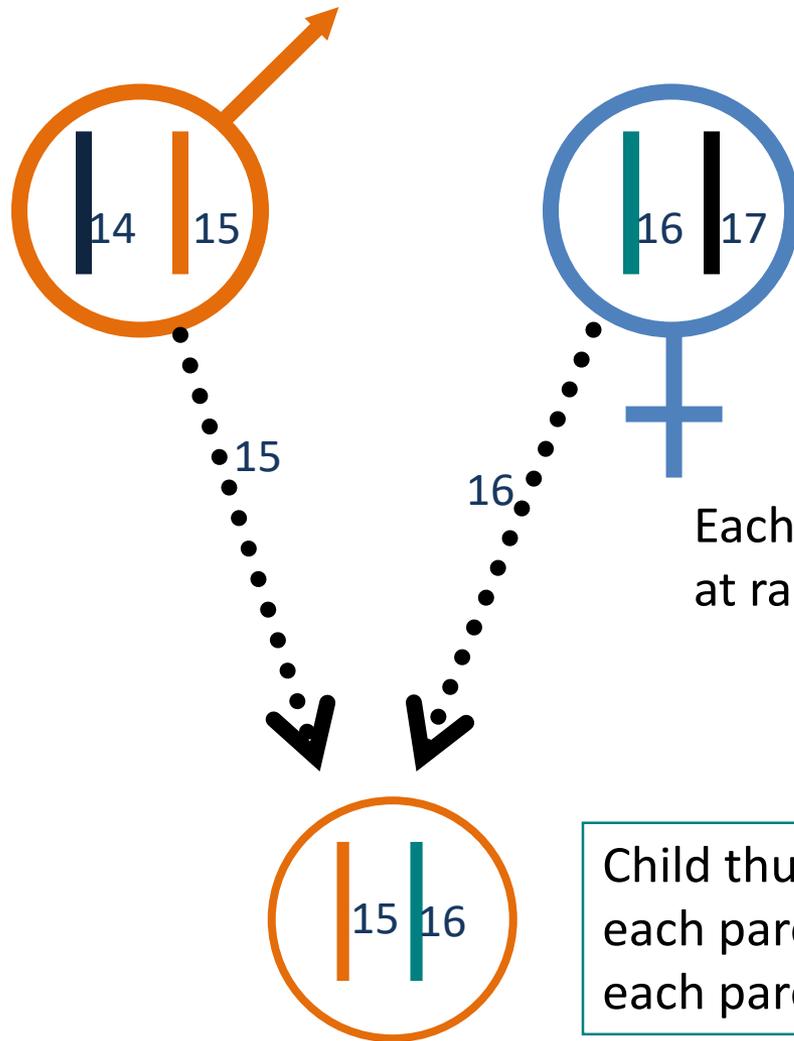
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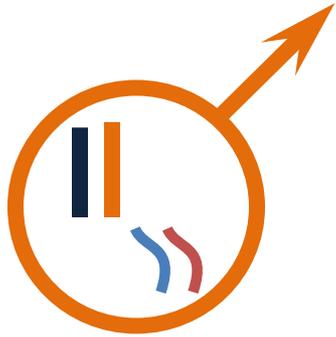


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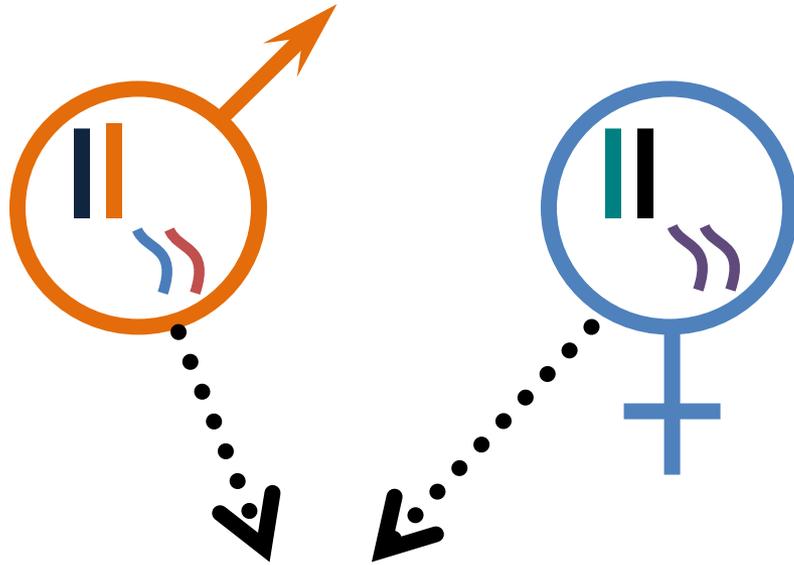
Child thus inherits a D12 chromosomes from each parent, and shares a vWA allele with each parent – e.g. {15,16}

Genetic inheritance (sexual), multiple loci



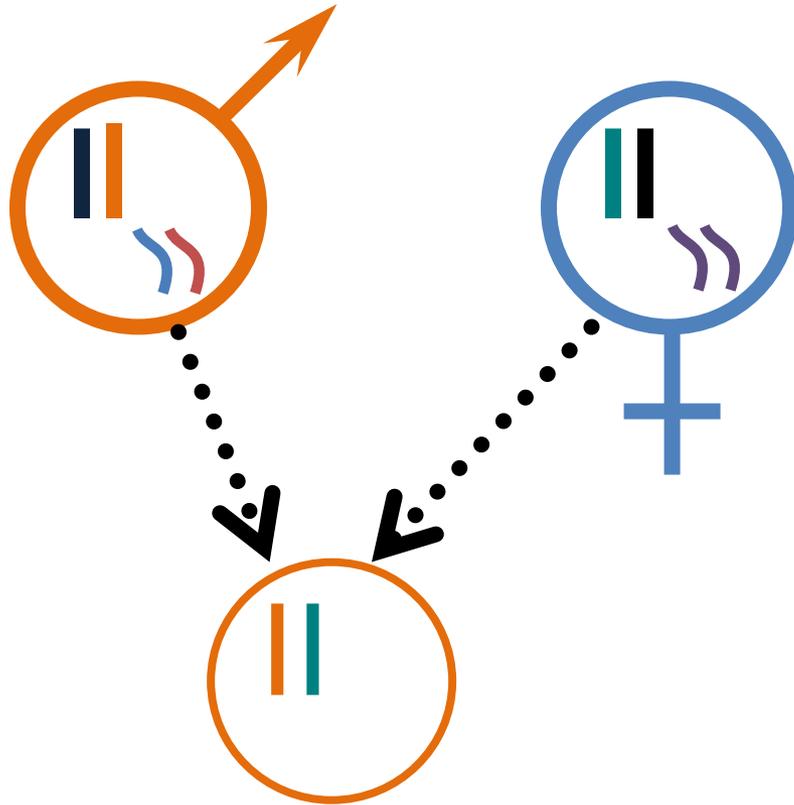
Each parent contributes one chromosome at random from each pair **independently**.

Genetic inheritance (sexual), multiple loci



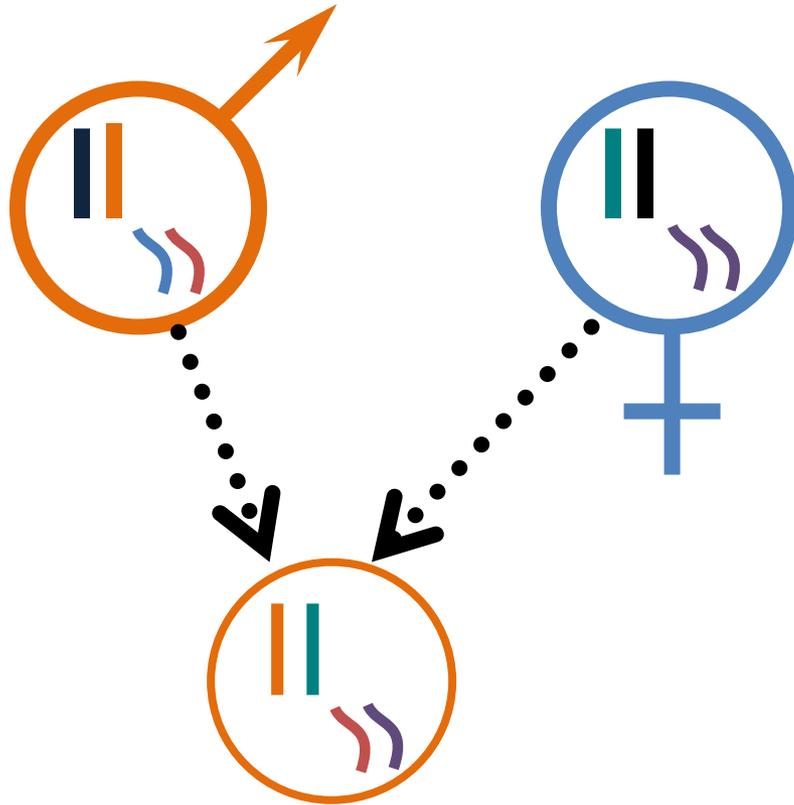
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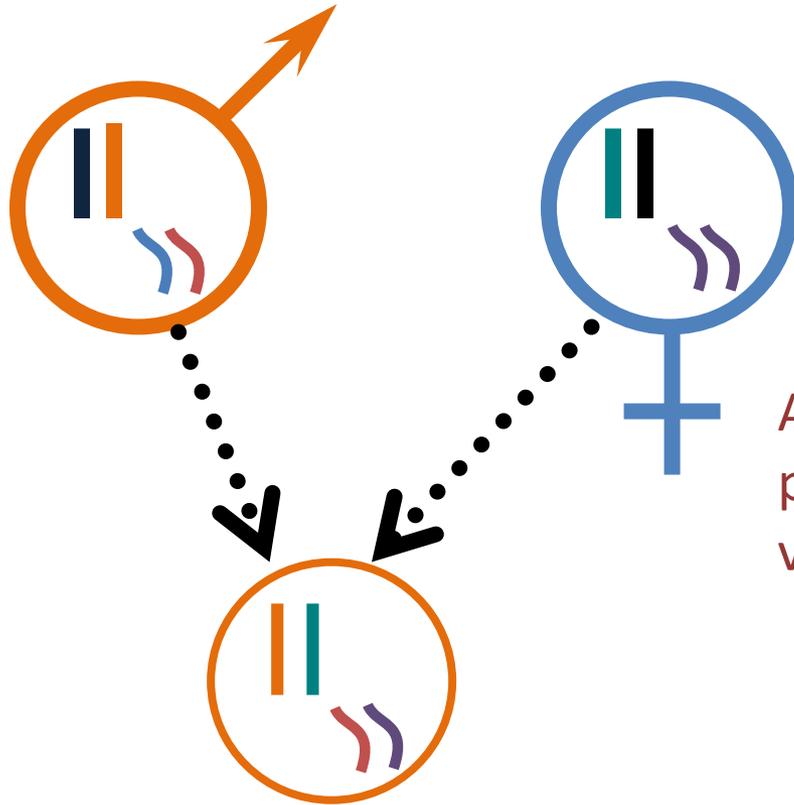
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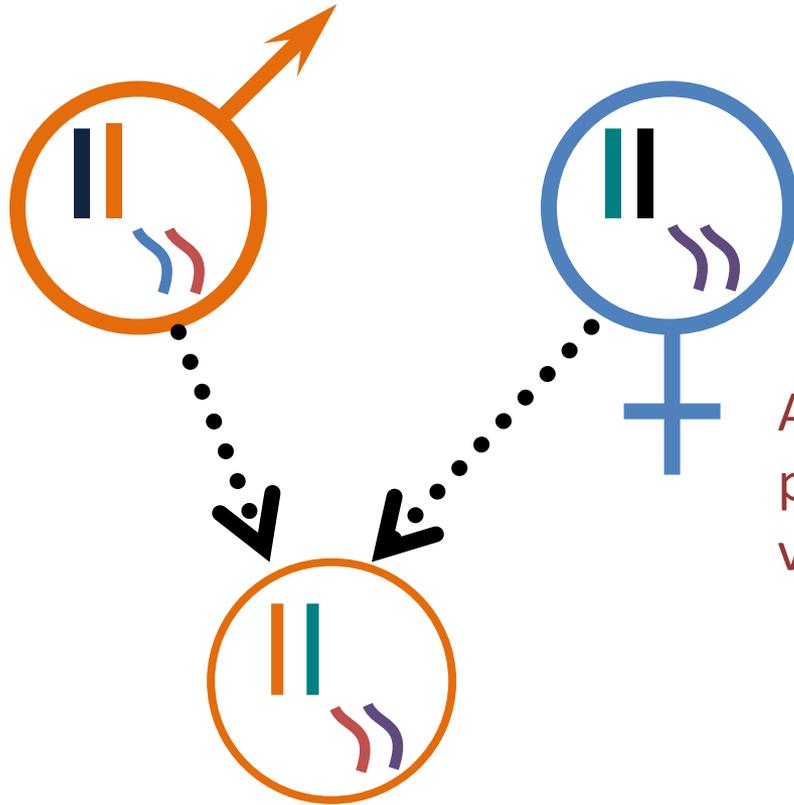
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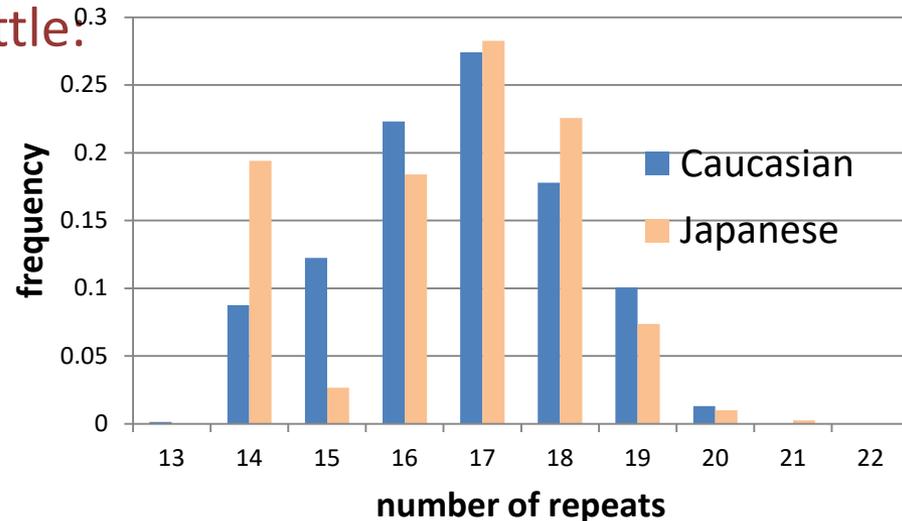
Alleles are common compared to population size, hence their frequencies vary little:

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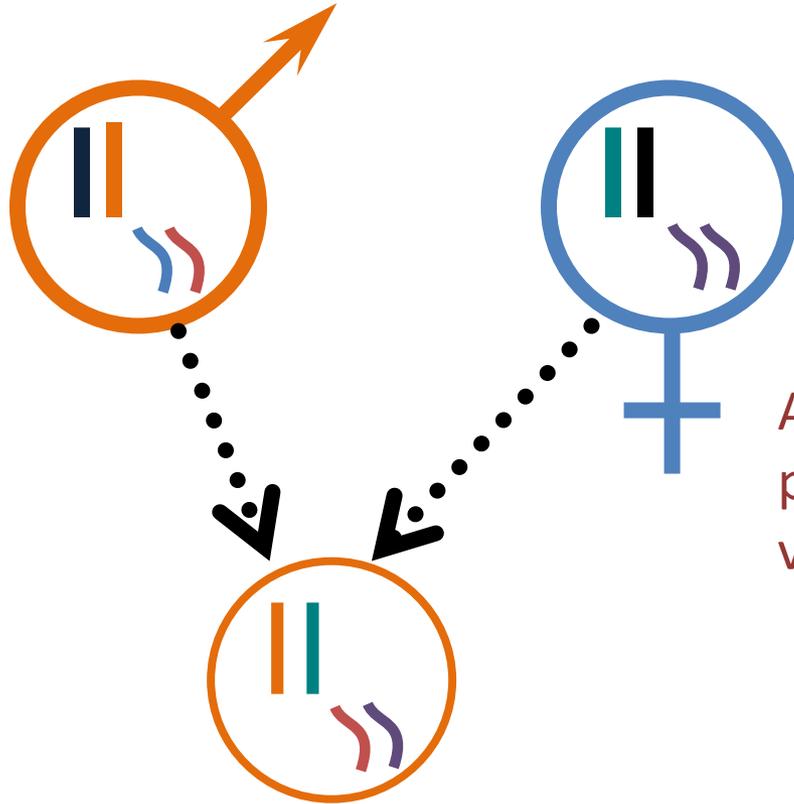


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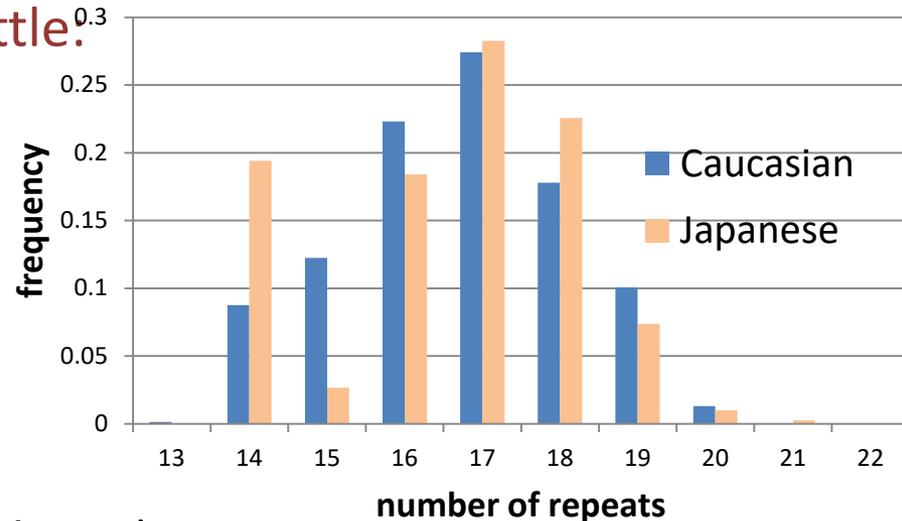


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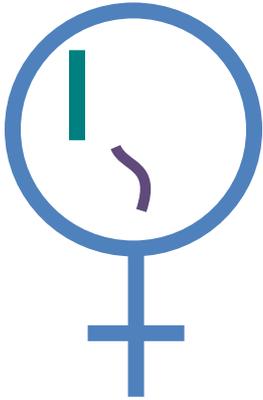
Alleles are common compared to population size, hence their frequencies vary little.



Evidential value: about $5 \times$ factor per allele†

† (but deduct ! 2 per locus, so $5^2 \div 2$ per locus)

Genetic inheritance (Y), multiple loci



Father→son transmission

Y chromosome (all loci) as a unit: “haplotype”.

No mixing in transmission, hence loci are *dependent*.

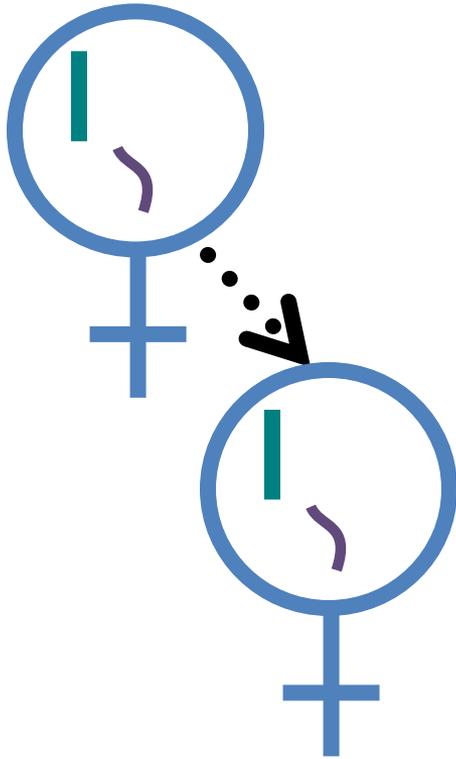
Evidential value: *cannot* multiply factor per locus.

Evidential value: ?

Treat haplotype as monster “allele”?

If 17 loci → 10000 haplotypes.

Genetic inheritance (Y), multiple loci



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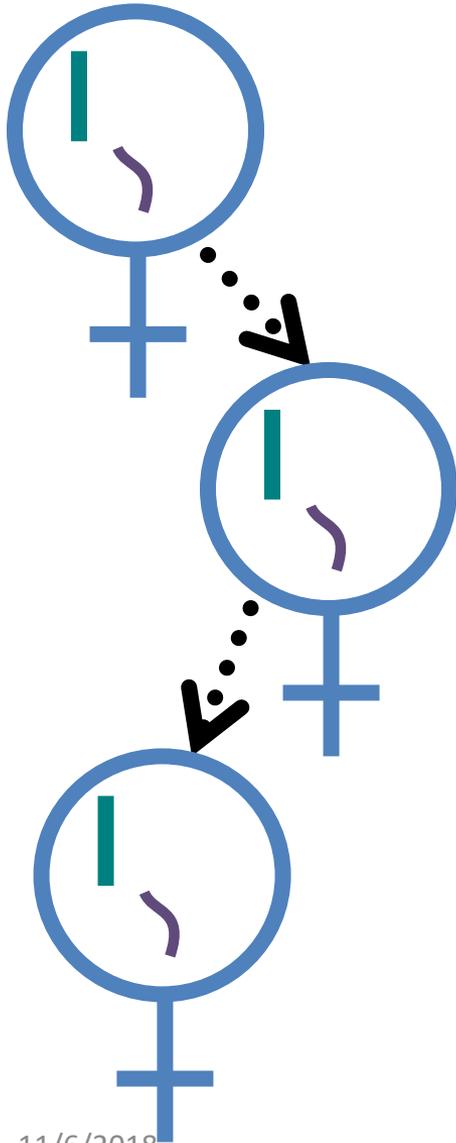
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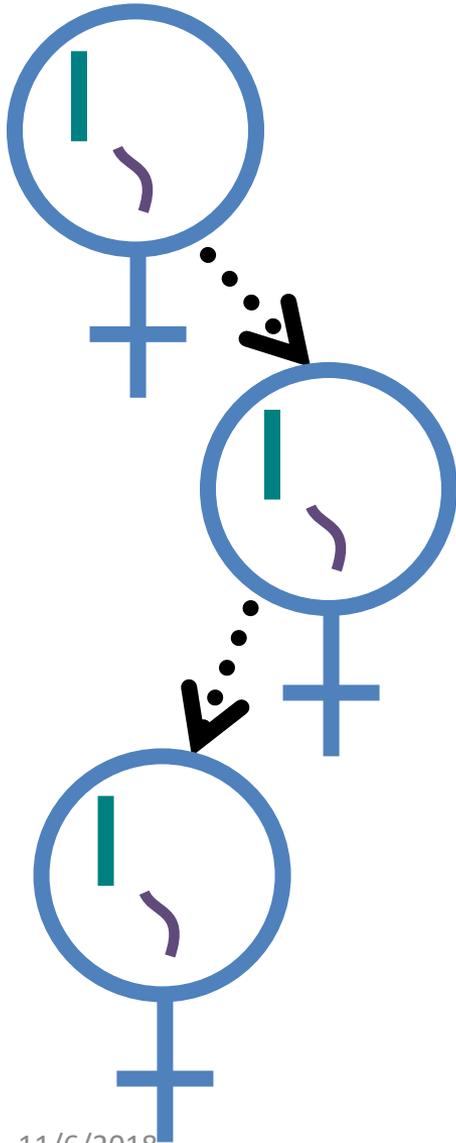
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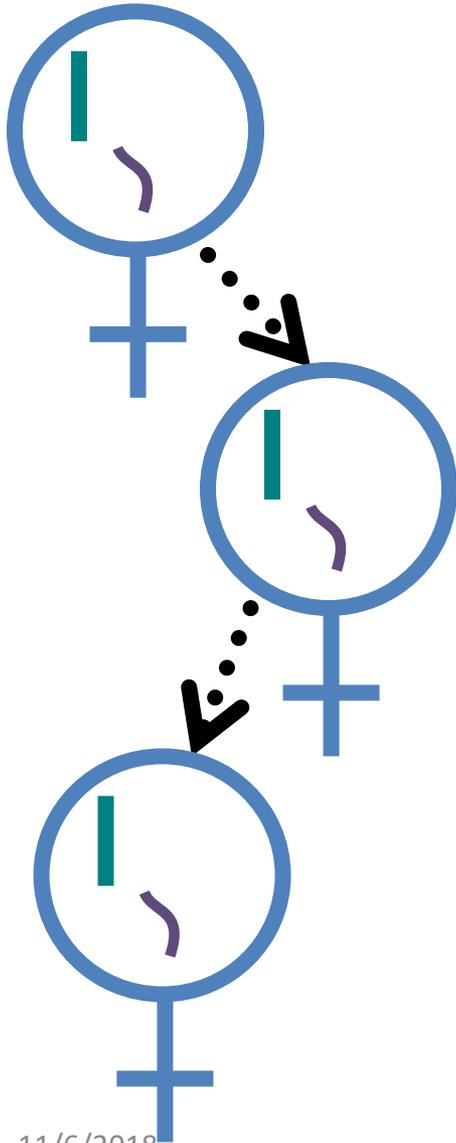
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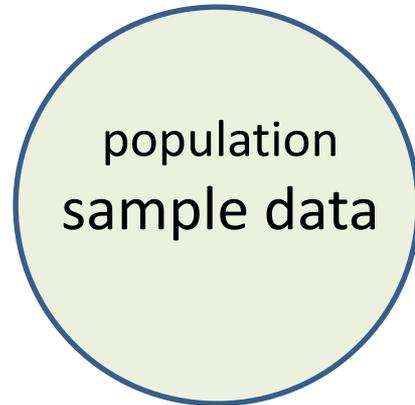
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Asexual; cloning

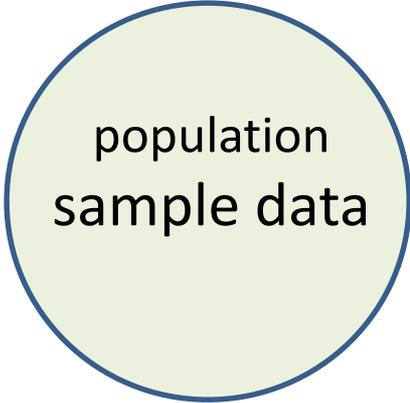


Y-evidence calculation approaches

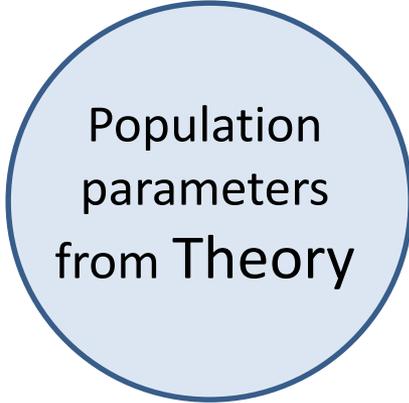
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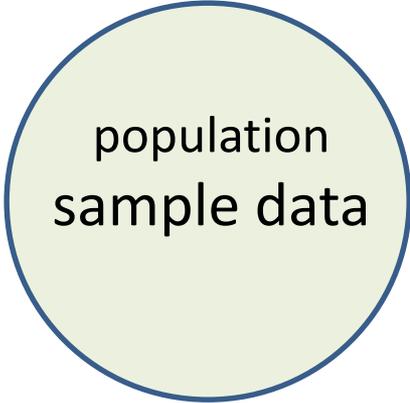


population
sample data

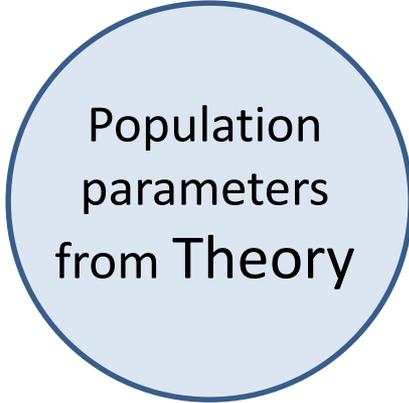


Population
parameters
from Theory

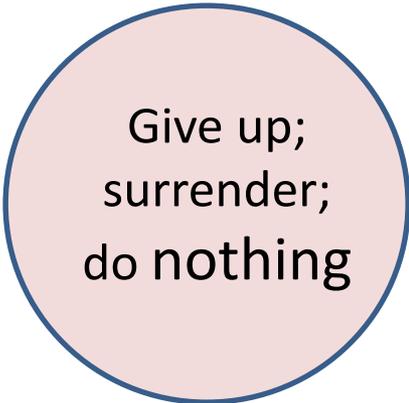
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population
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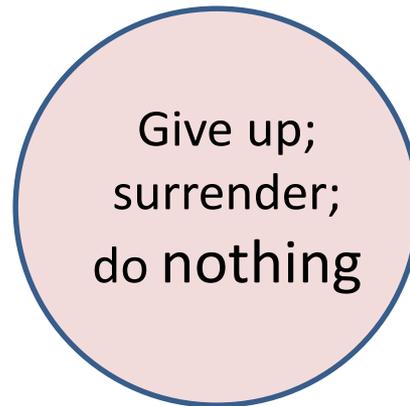
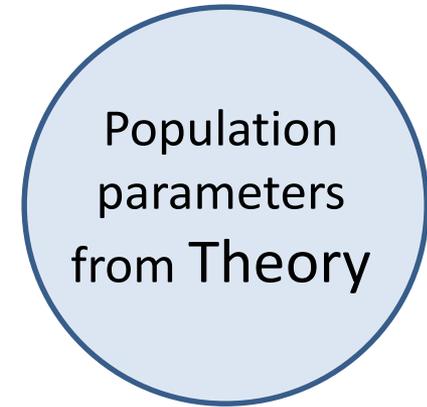
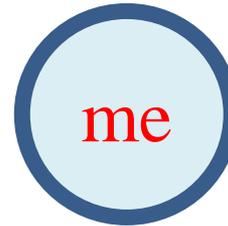
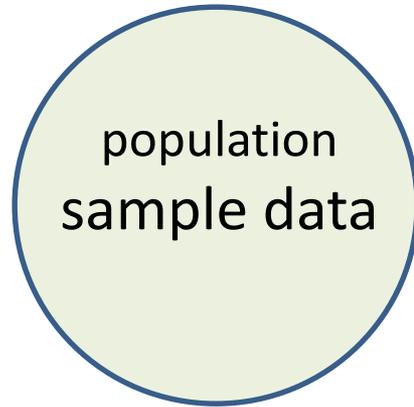


Population
parameters
from Theory



Give up;
surrender;
do nothing

Y-evidence calculation approaches



Evolution of the Yfiler lineages

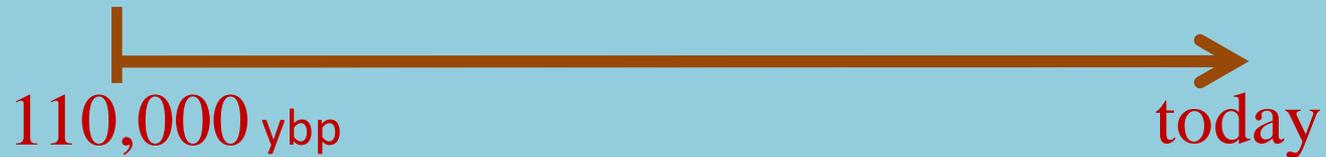


Y-chromosome
Adam”

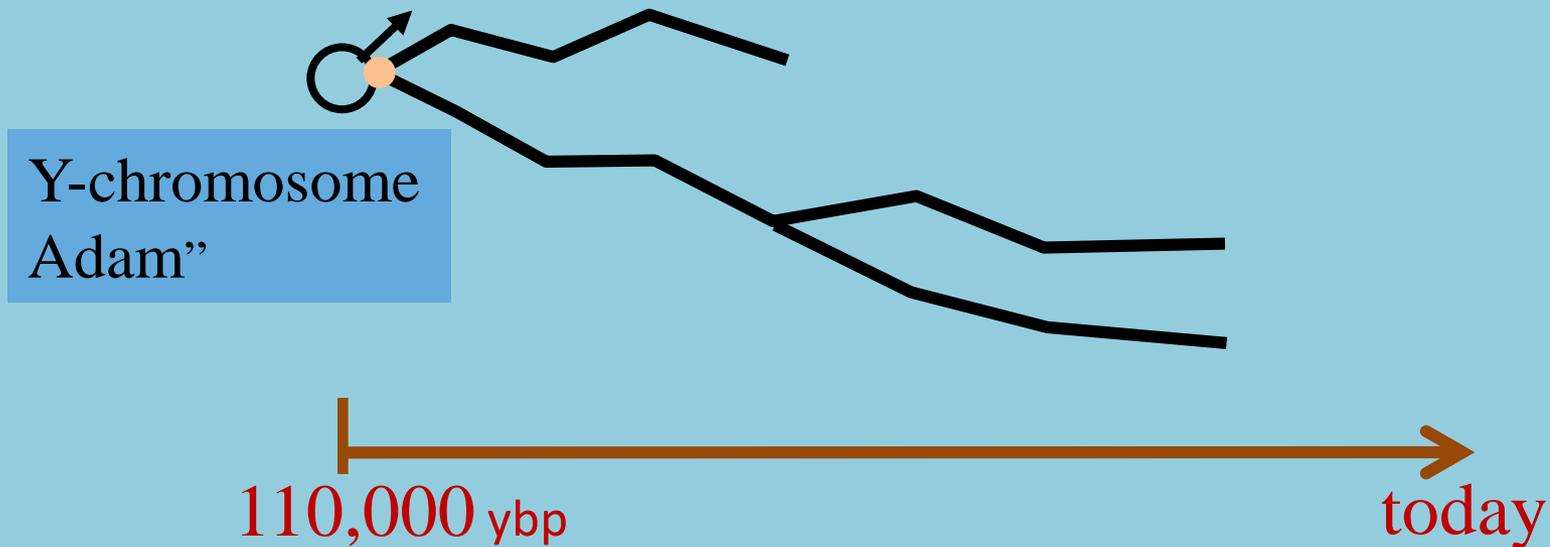
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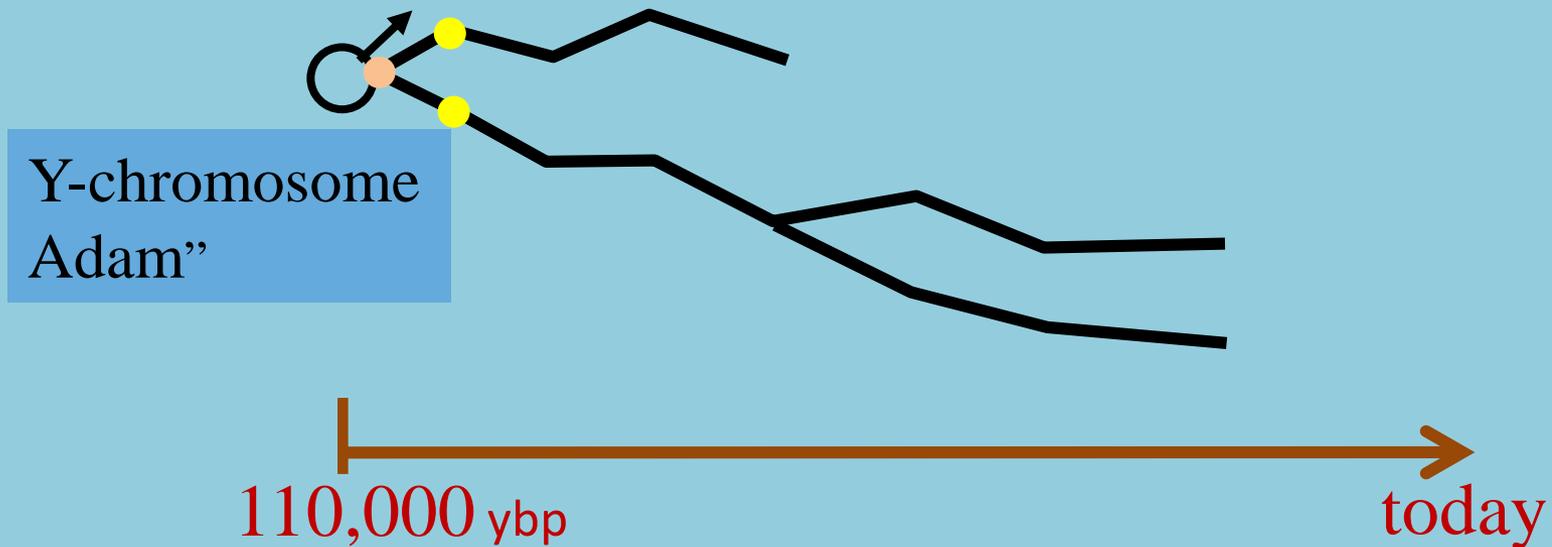
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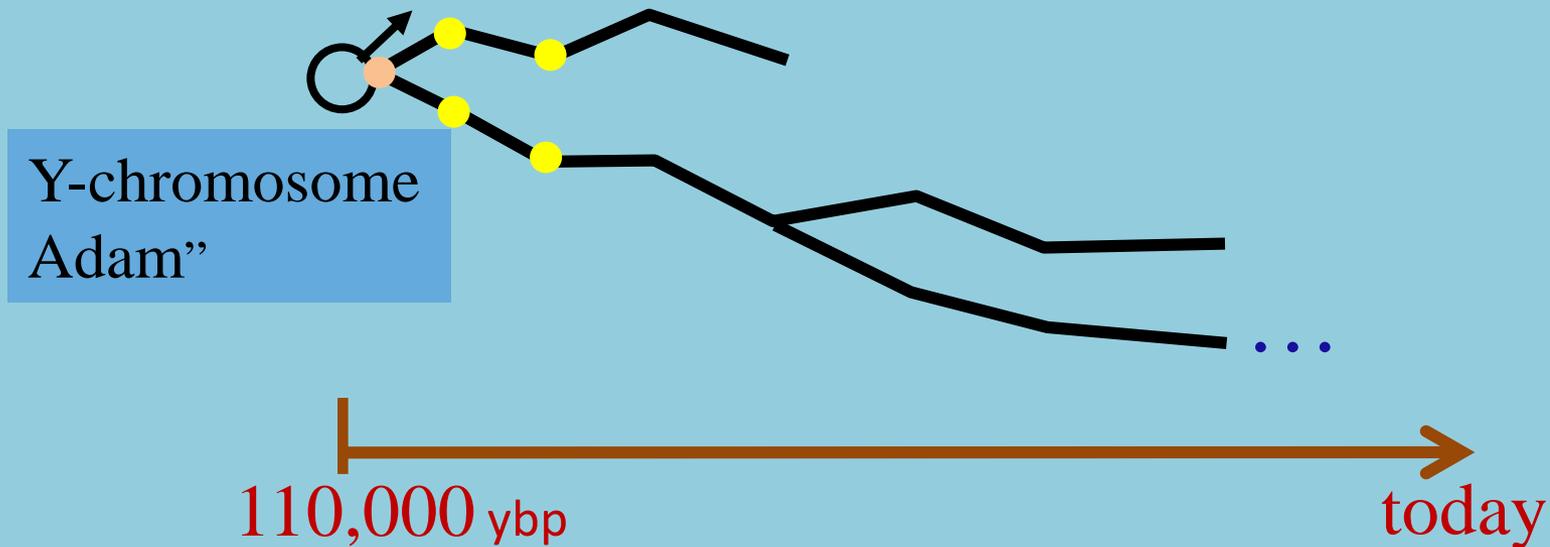
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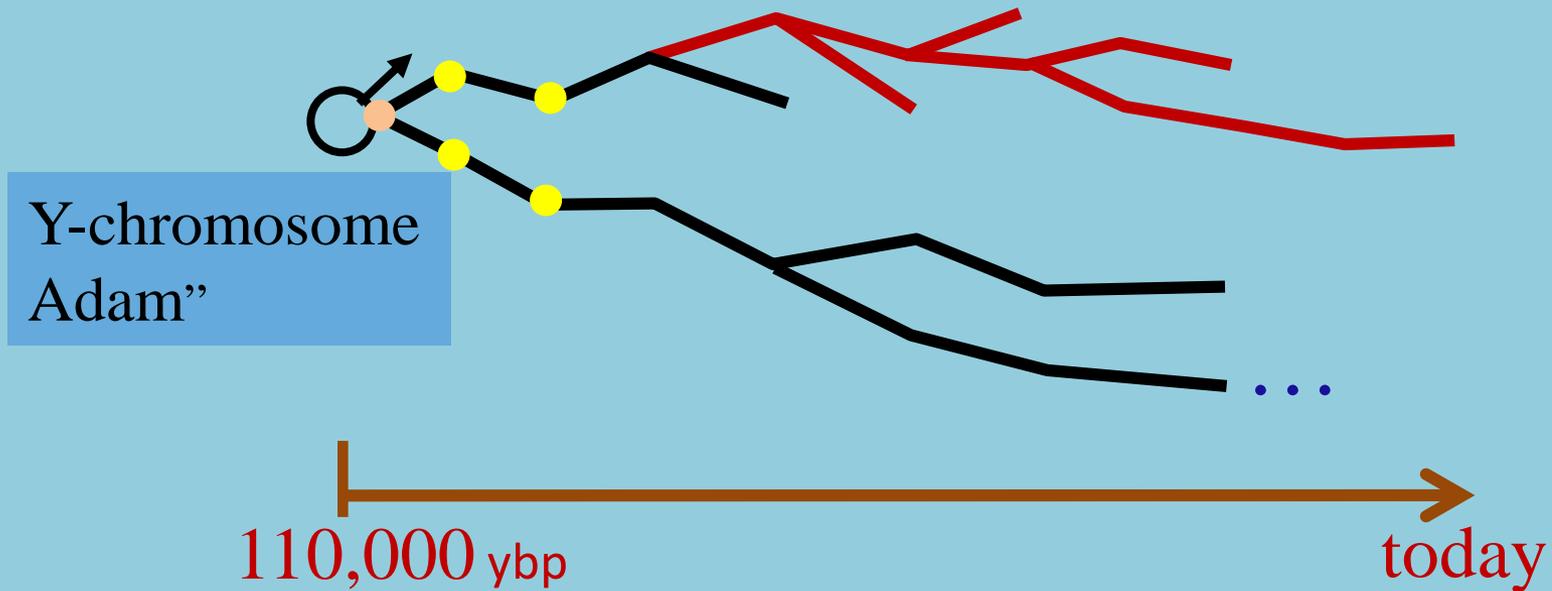
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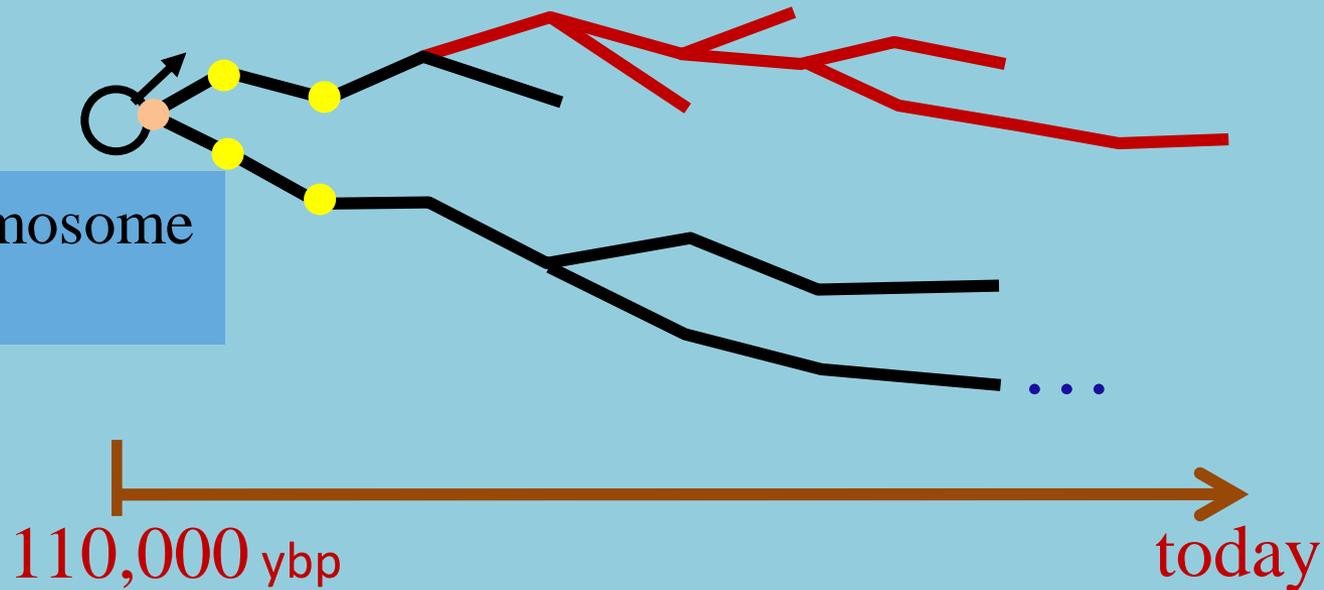
Evolution of the Yfiler lineages



Evolution of the Yfiler lineages

mutation

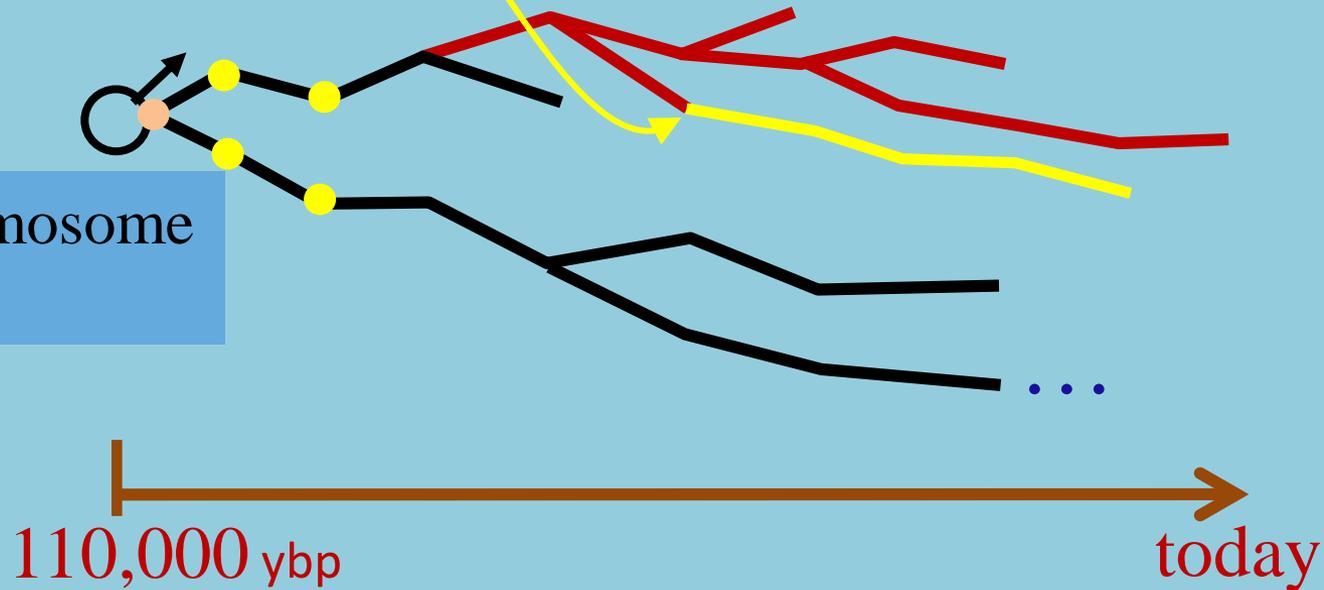
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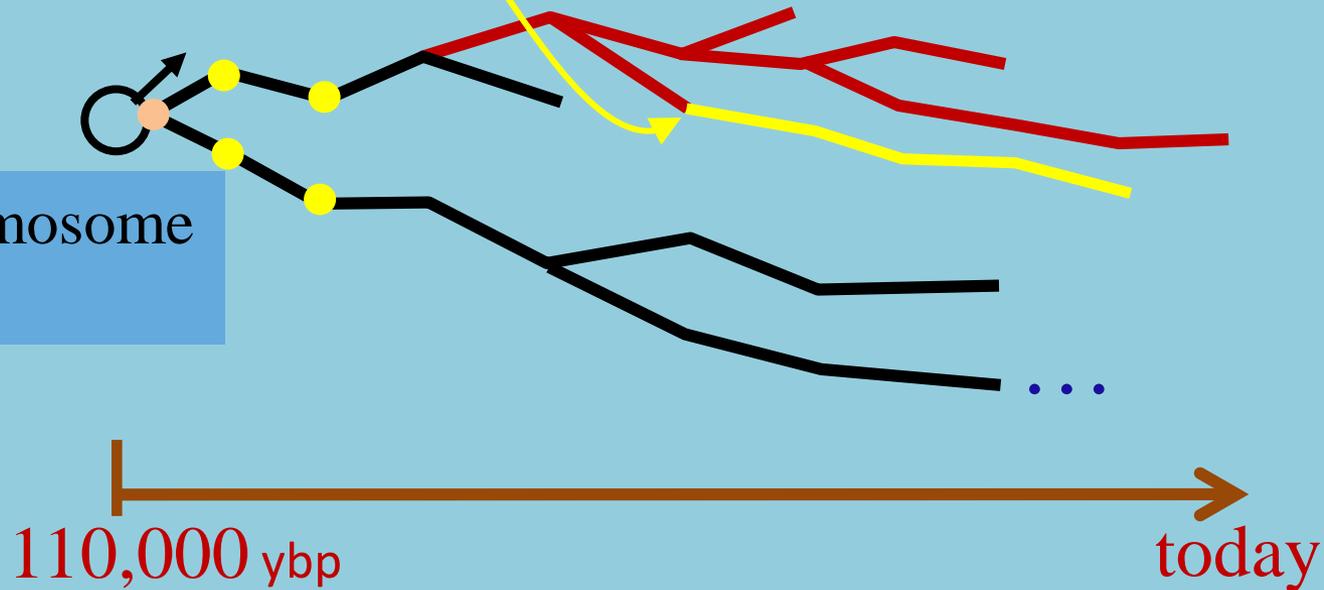
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Evolution of the Yfiler lineages

mutation

Y-chromosome
Adam"

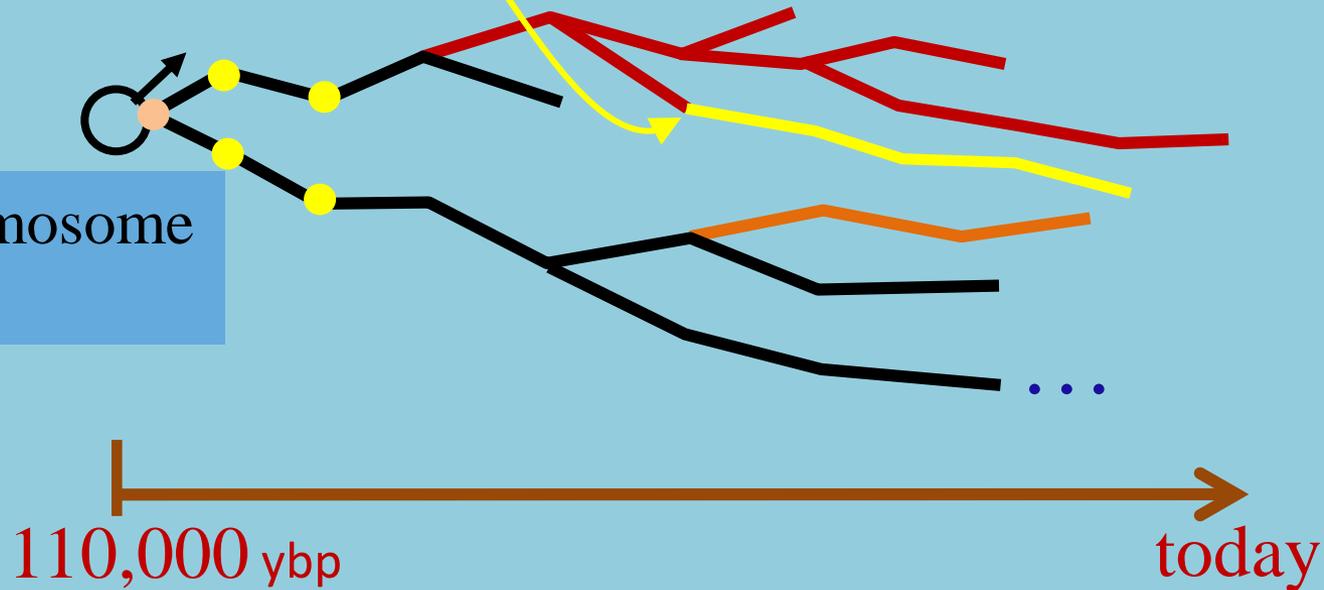


(Same color = same Y-haplotype)

Evolution of the Yfiler lineages

mutation

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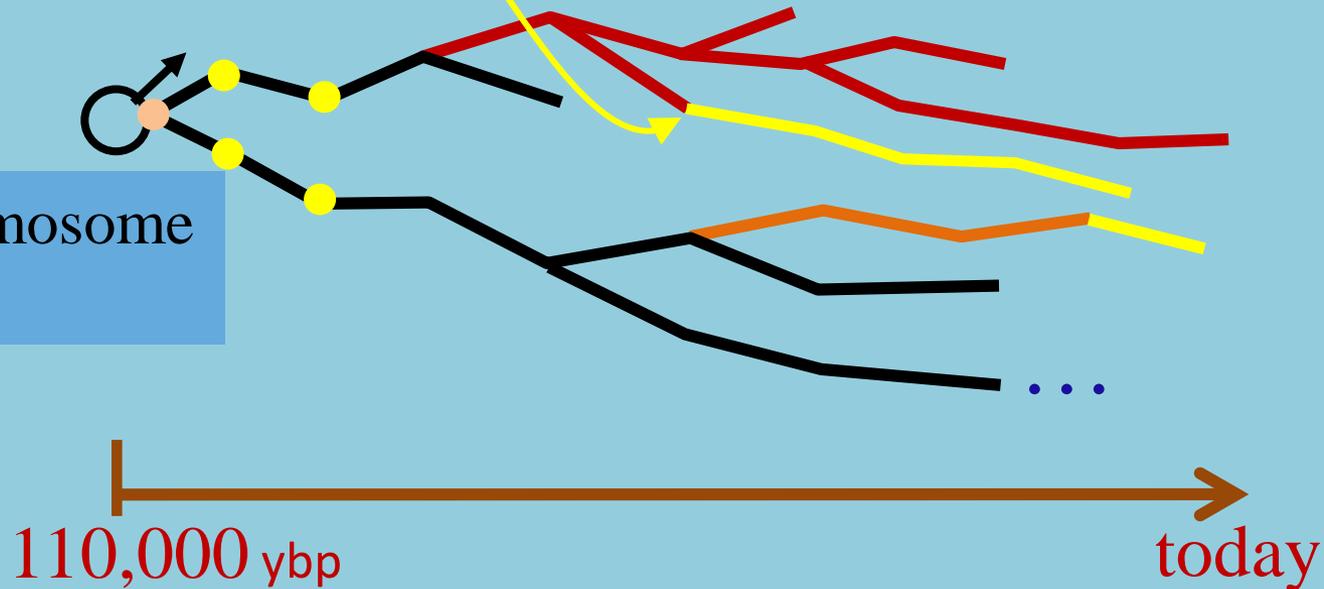


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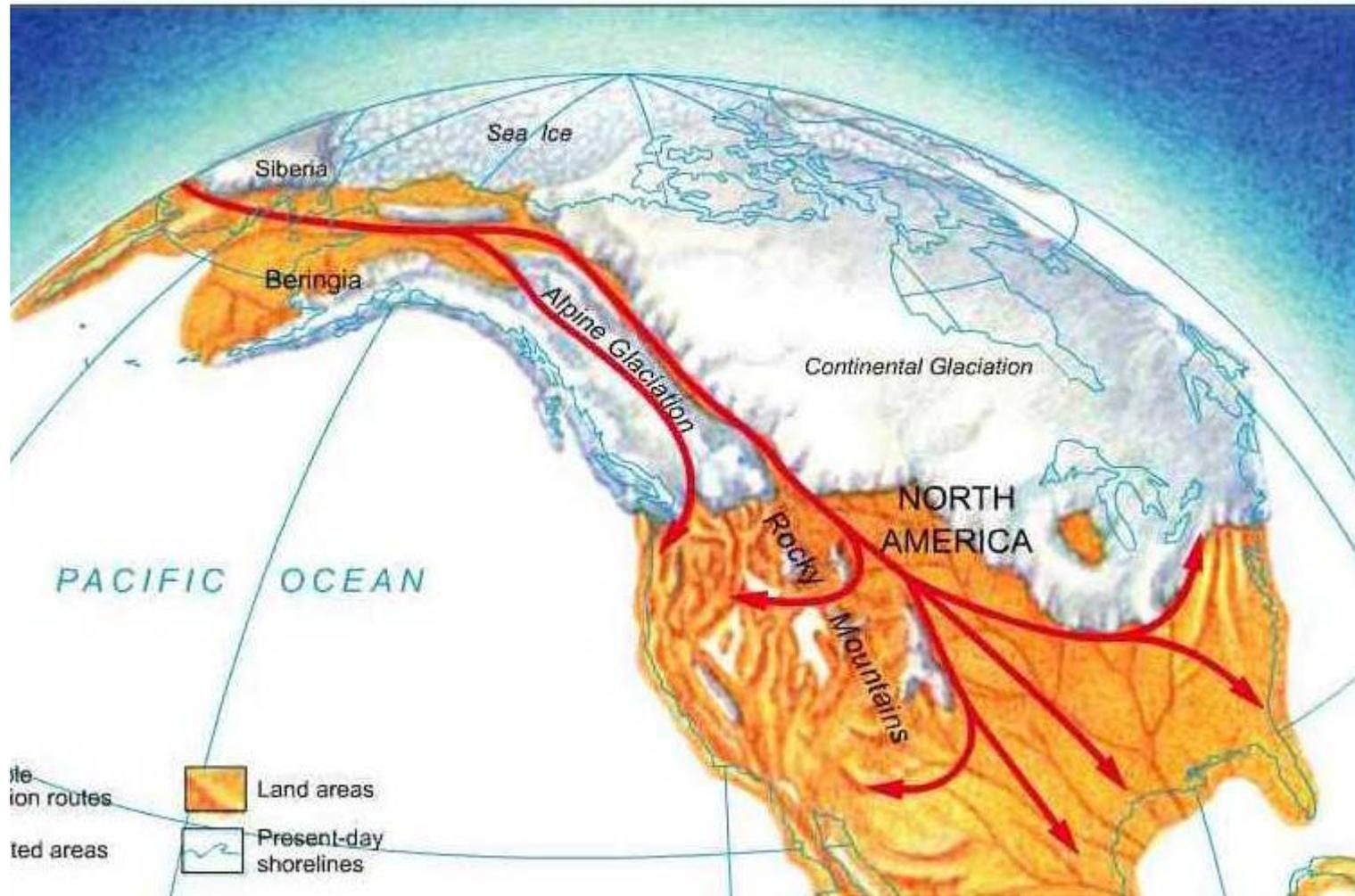
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mutation

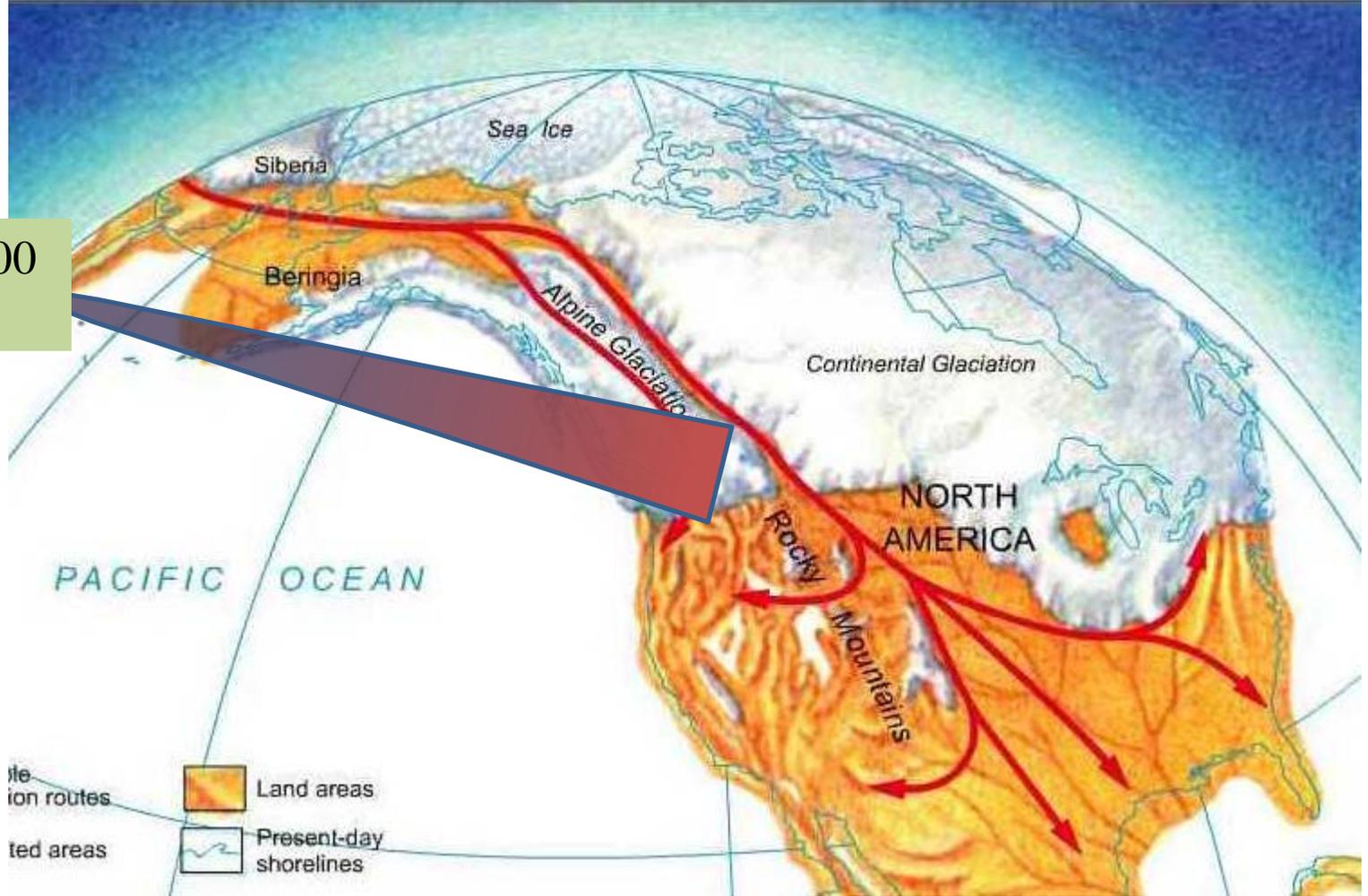
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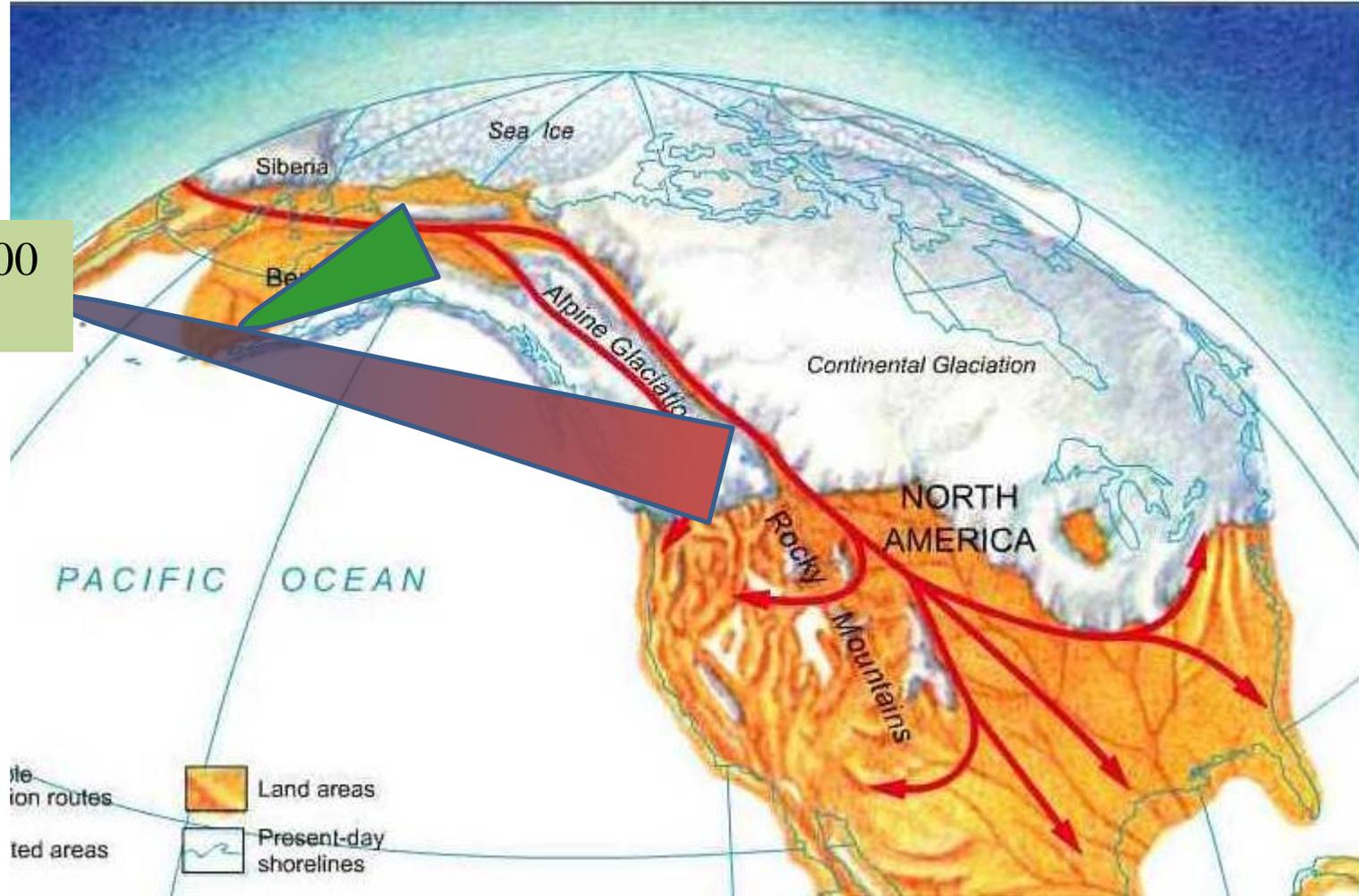


15,000
ybp

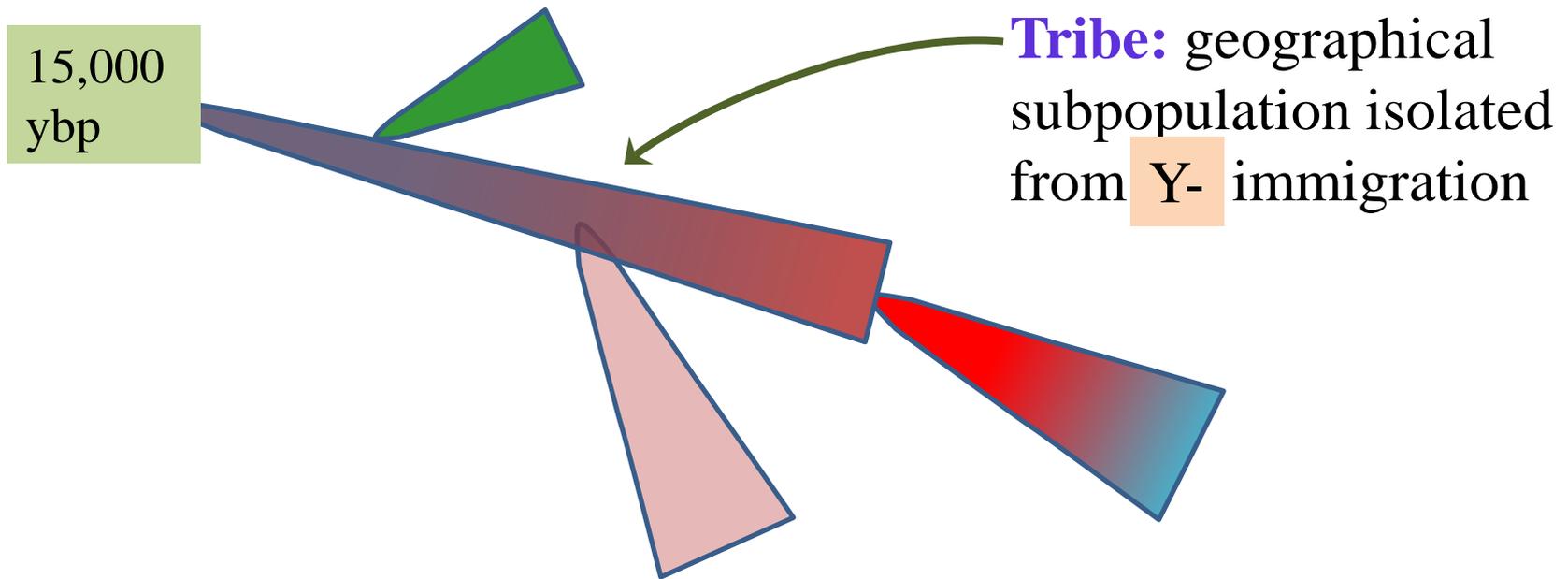


Manufacturing diversity

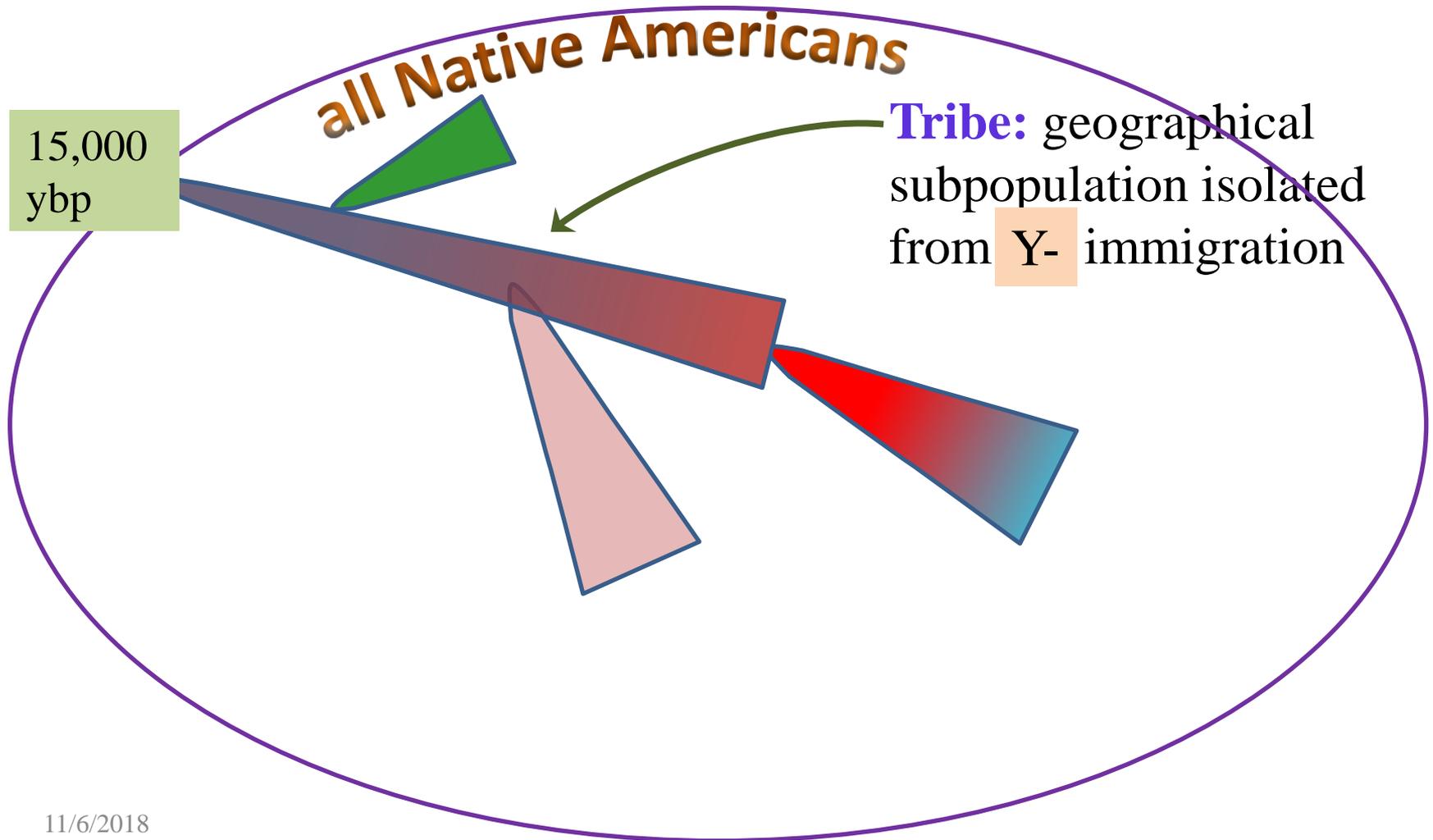
15,000
ybp



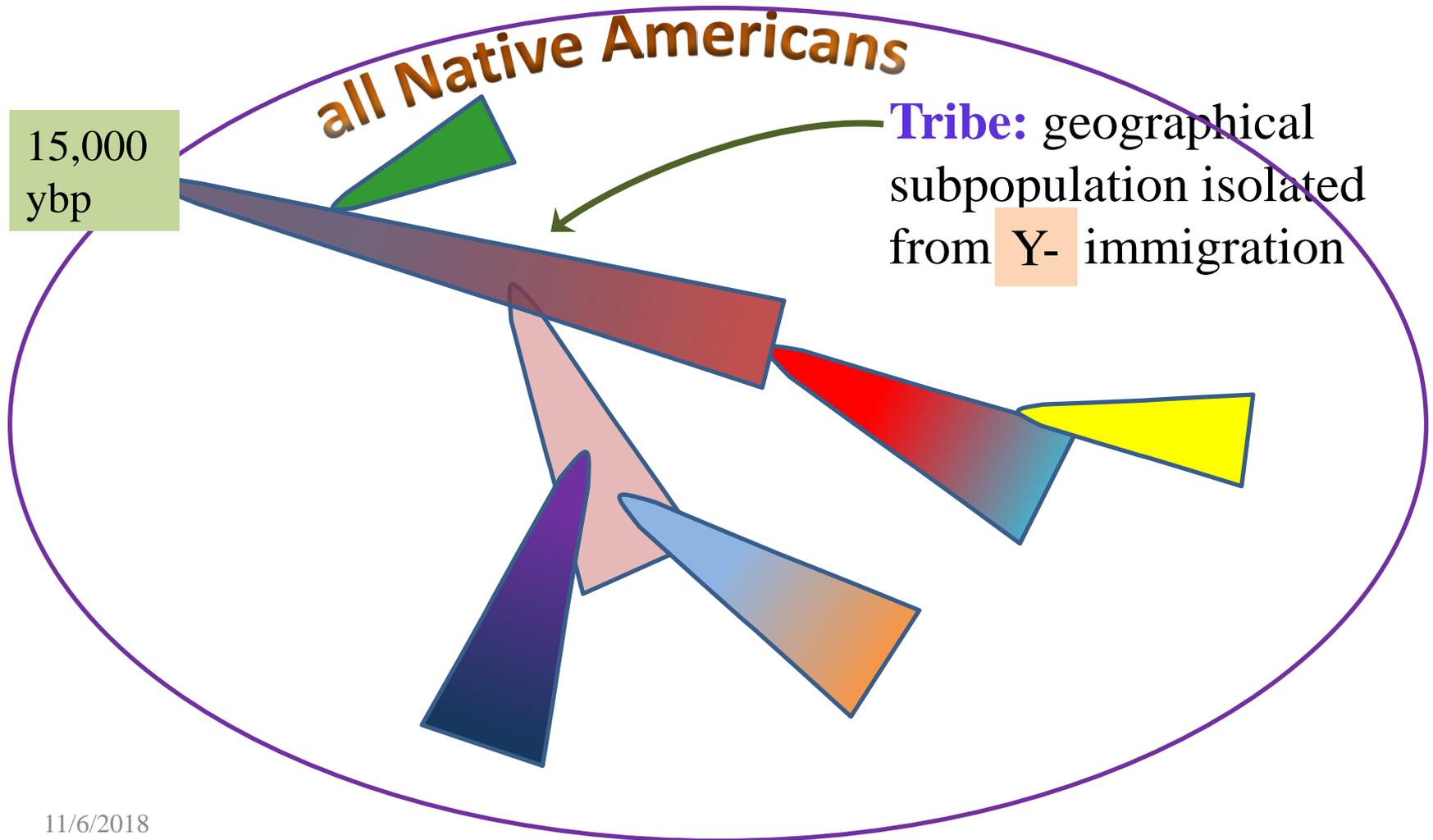
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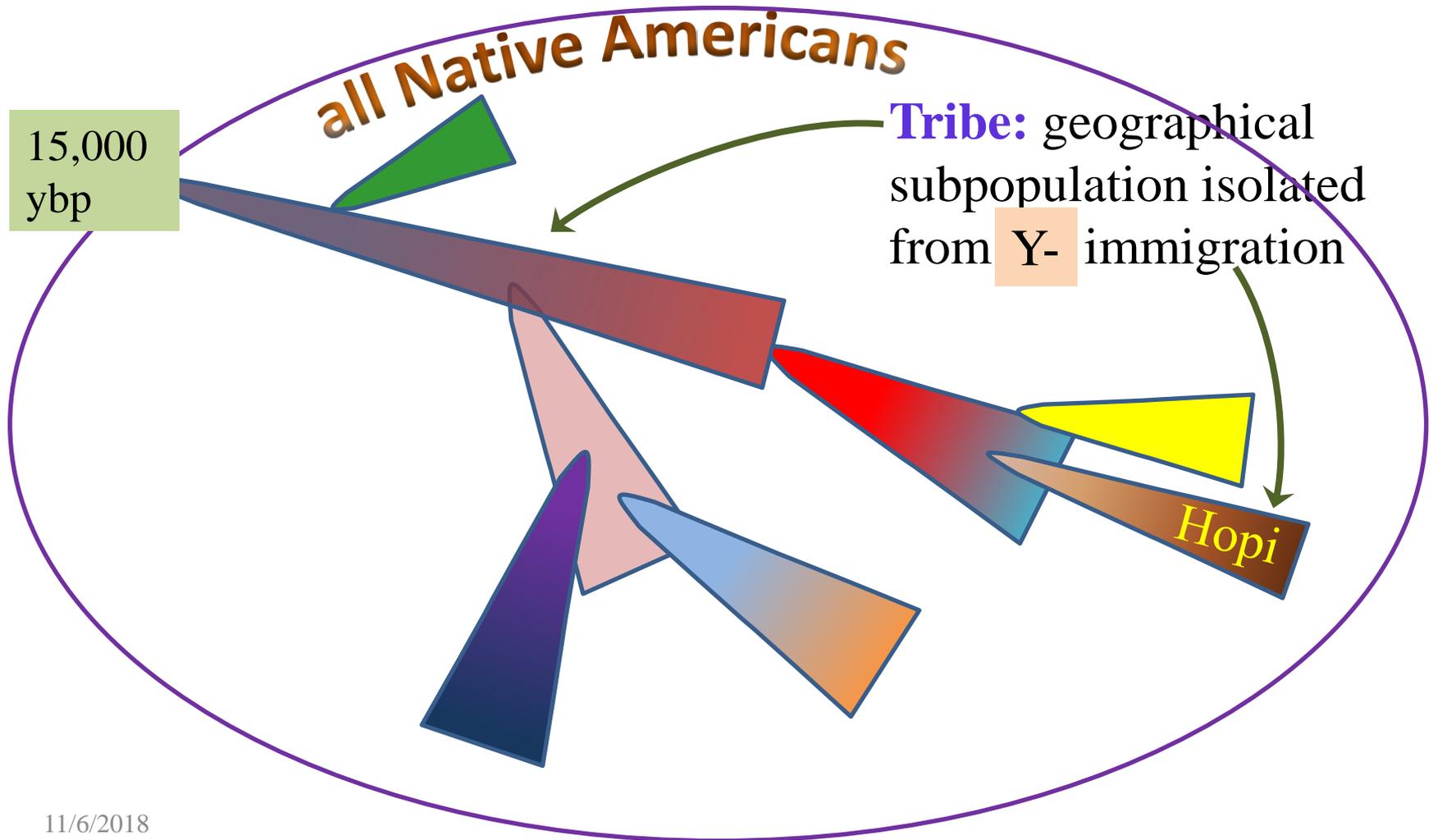
Manufacturing diversity



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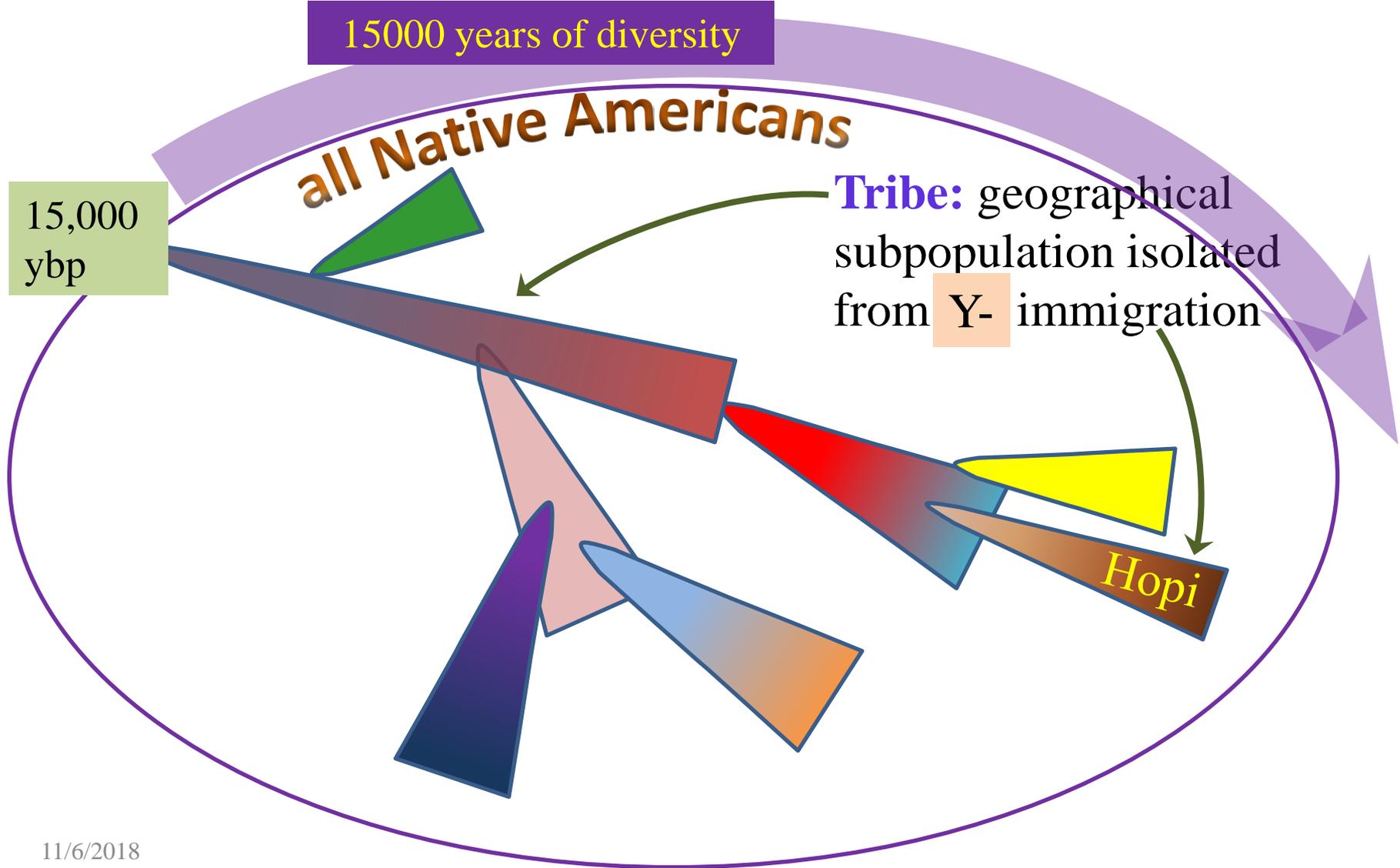
15000 years of diversity

all Native Americans

15,000
ybp

Tribe: geographical subpopulation isolated from Y- immigration

Hopi



Manufacturing diversity

15000 years of diversity

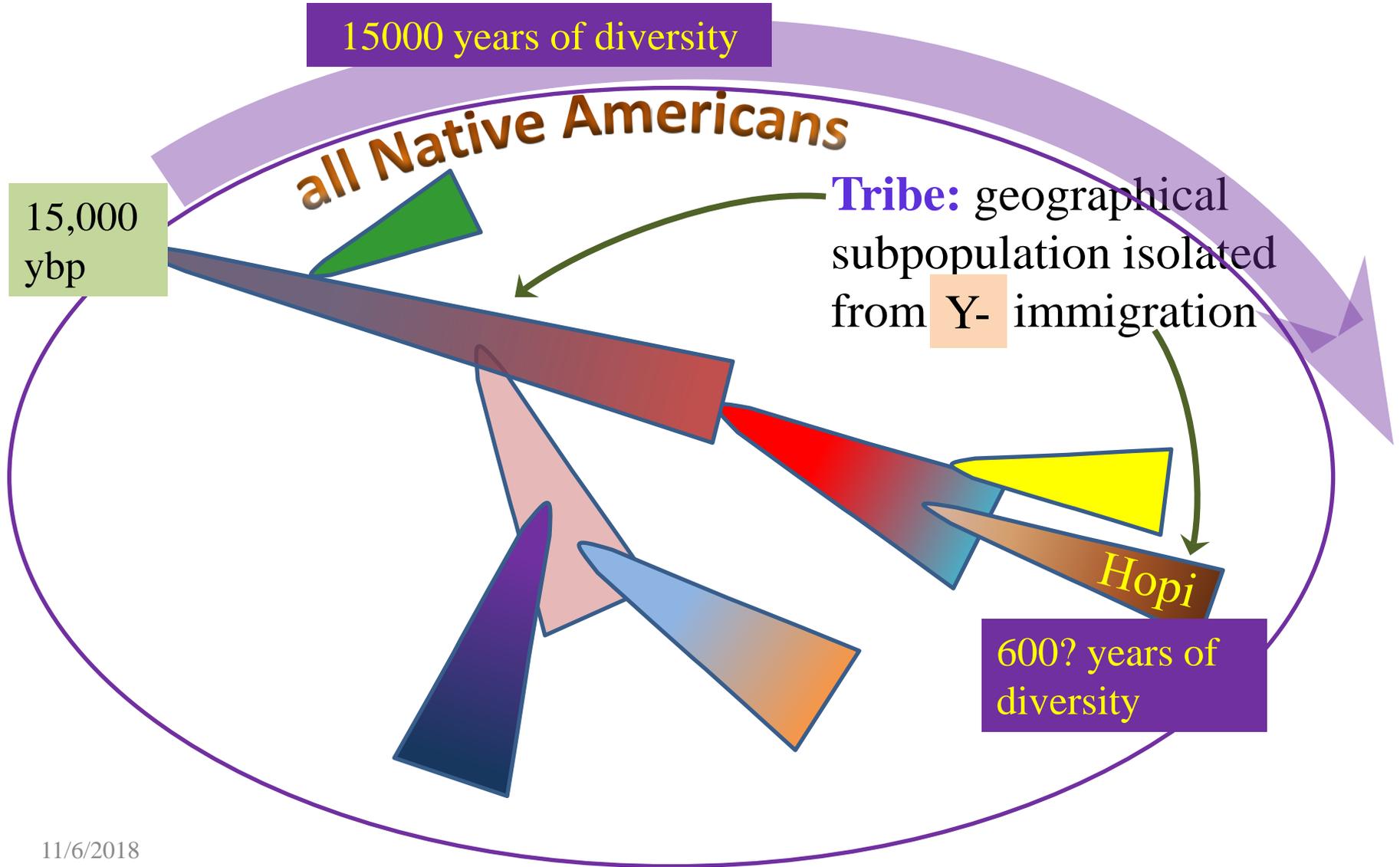
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600? years of diversity



Manufacturing diversity

15000 years of diversity

$\Pr(A|A) \approx 1/3000$

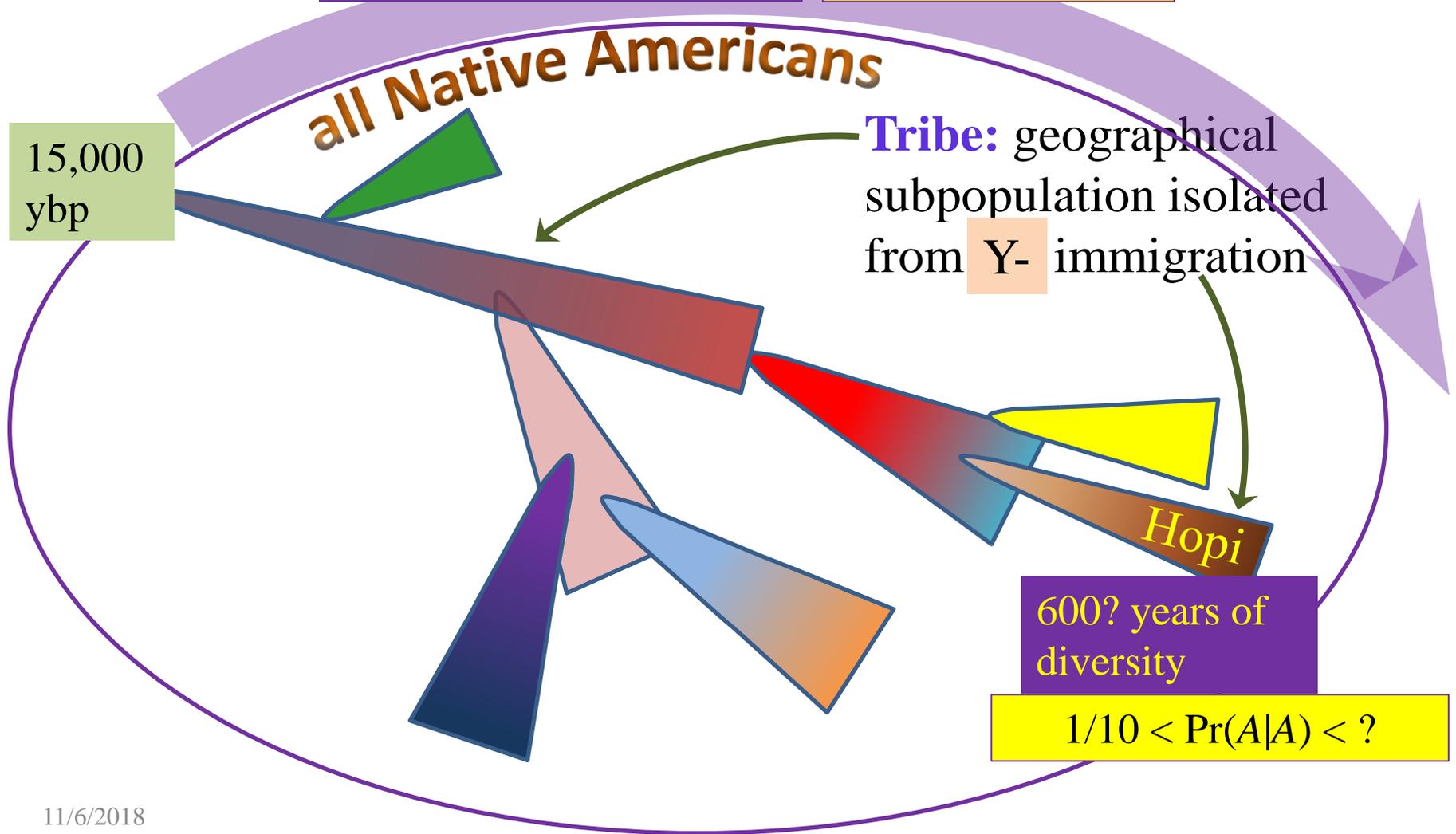
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Tribe: geographical subpopulation isolated from Y-immigration

600? years of diversity

$1/10 < \Pr(A|A) < ?$



Southwest Native Am. court cases

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Southwest Native Am. court cases

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Y-haplotype mutation and matching

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- Mutation model

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Adam ← 17p5 5 5 5 ...

A ancestral 17-locus Y haplotype

Y-haplotype mutation and matching

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Adam ← 17 ρ 5 5 5 5 ...

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μ ← ÷ 350 8000 200 500 ...
(÷ 350)

μ per-locus mutation rates. Ave μ =

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$$\Pr(\mathbf{Son} \neq \mathbf{Father}) = 17 \div 350 = 5\%. \text{ Patrilineage mutates every 500 years.}$$

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$$\Pr(\neq / \text{brothers}) = 10\%.$$

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- Implies

Pr(**Son** ≡ **Father**) = (x / 1 - mu)

Pr(**Son** ≠ **Father**) = 17 ÷ 350 = 5%. Patrilineage mutates every 500 years.

Pr(≠ / brothers) = 10%.

NB: **Time is reversible.**

Y-haplotype population

Y-haplotype population

- Evolutionary model – Wright-Fisher growth + mutation

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Pop \leftarrow , cAdam \leftarrow 17/5 ♀ generation 0, founder

Y-haplotype population

- Evolutionary model – Wright-Fisher growth + mutation
 $Pop \leftarrow c \cdot Adam$ $\leftarrow 17/5$ \mathcal{A} generation 0, founder
 $\nabla s \leftarrow NewSize$ $s \nabla$ \mathcal{A} some rule for population growth

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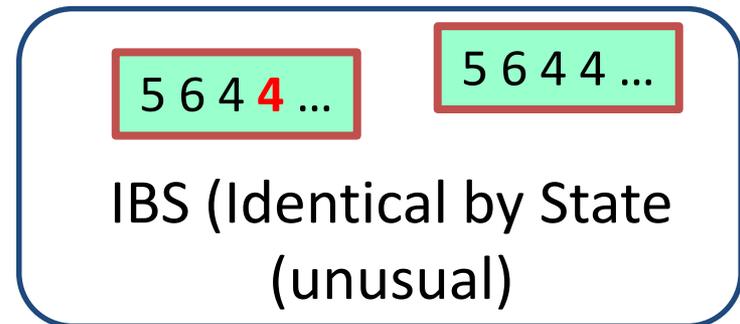
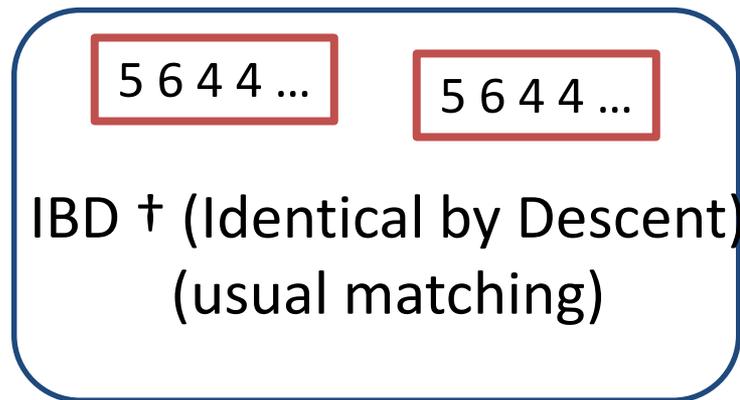
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- Iterating generations

Y-haplotype population

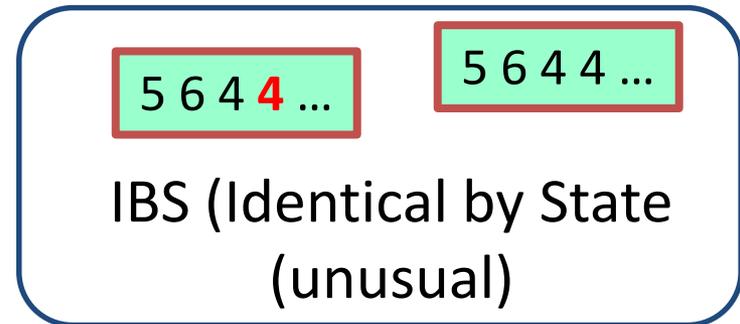
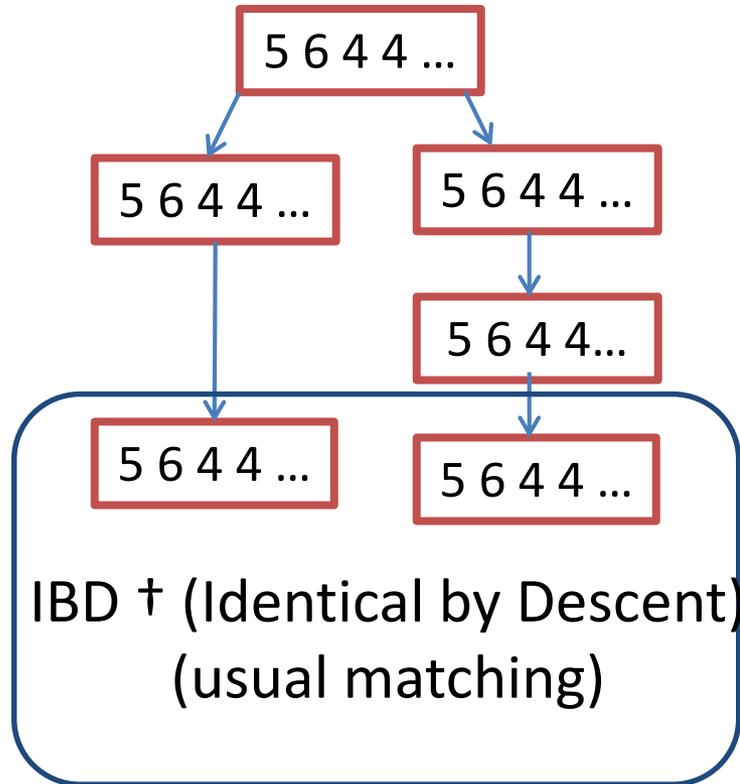
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- Iterating generations
 - “Diversity” \equiv accumulation of mutations
 - Time
 - Population size

All men are related



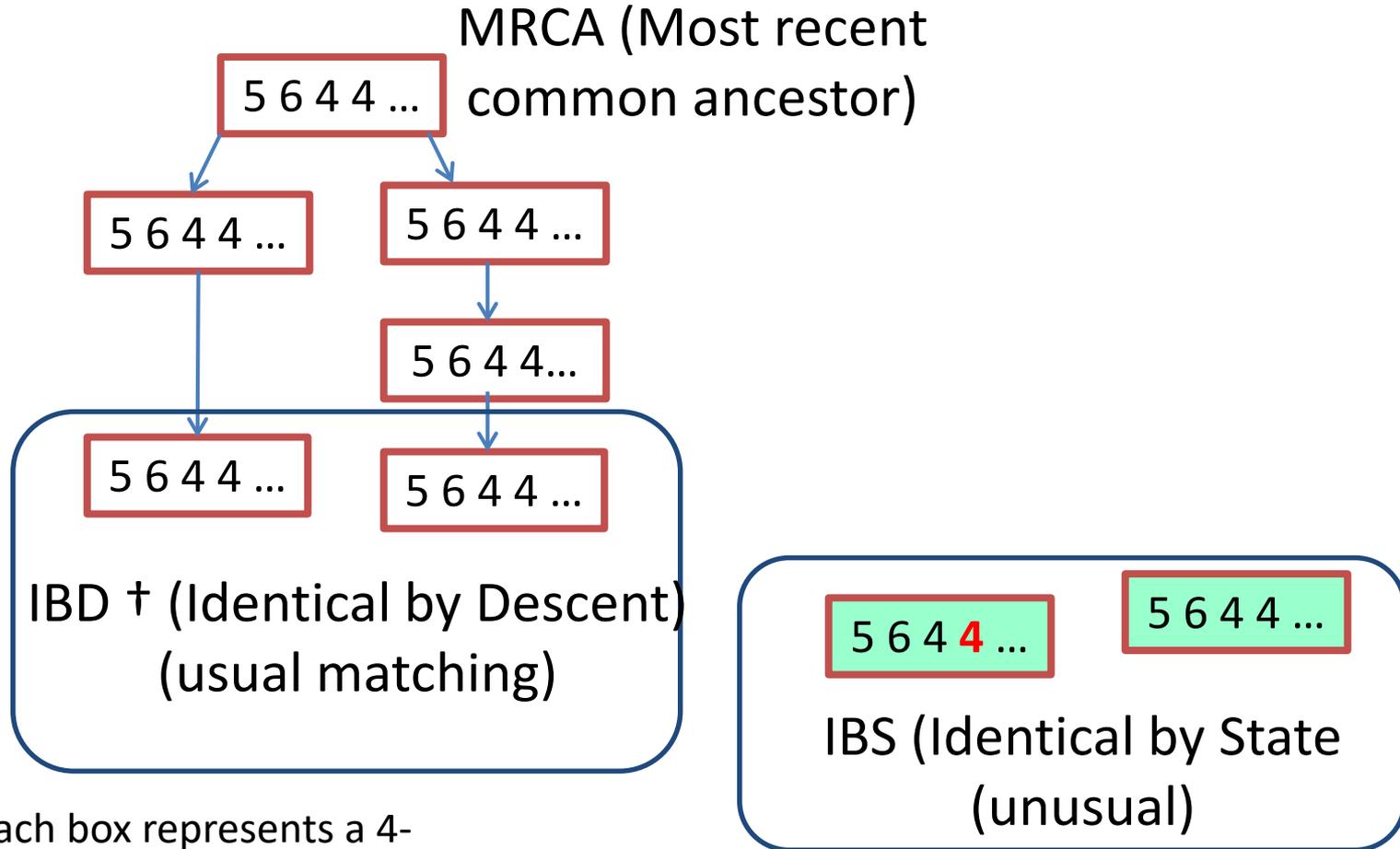
† Each box represents a 4-locus Y haplotype

All men are related



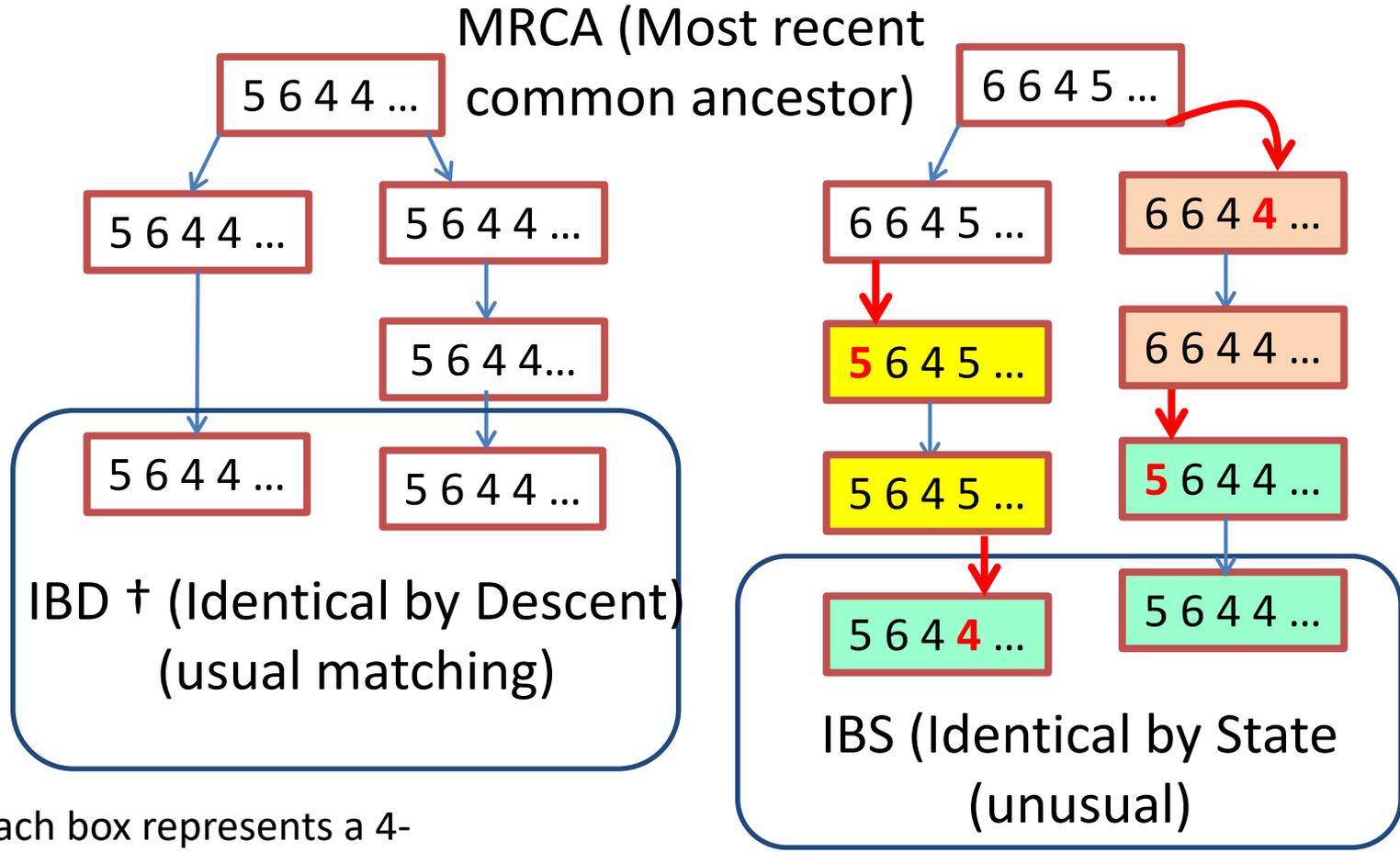
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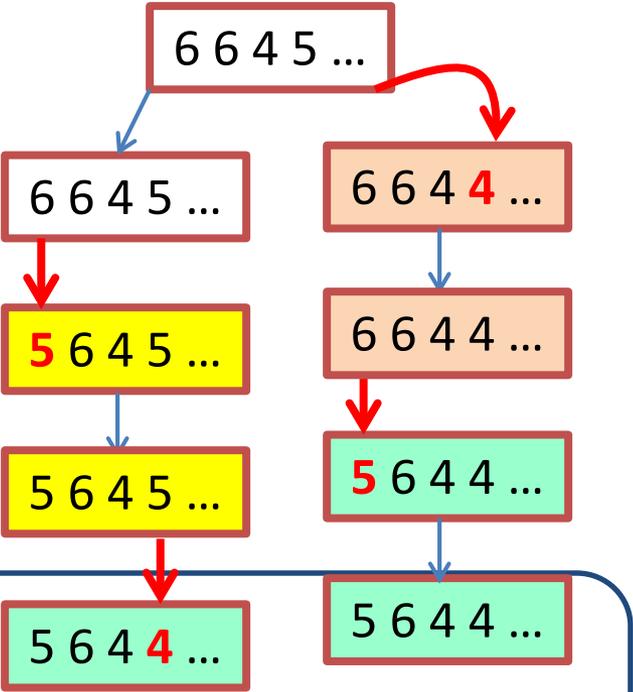
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Identity by State – closer look

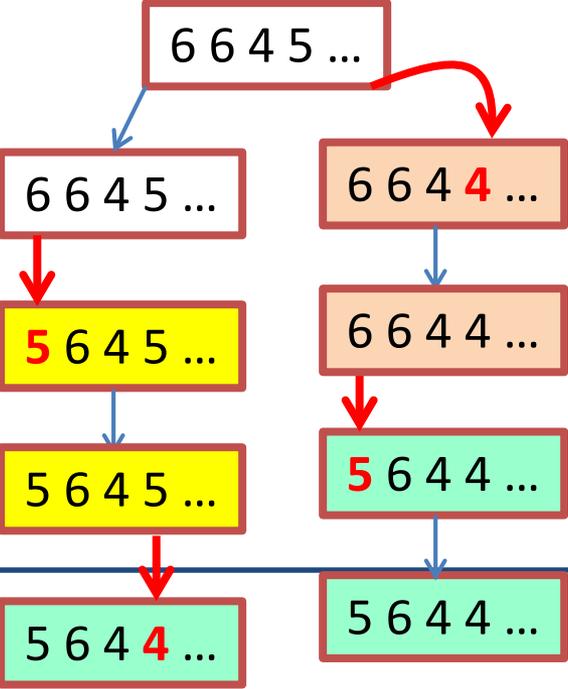
MRCA (Most recent
common ancestor)



IBS (Identical by State)

Identity by State – closer look

MRCA (Most recent
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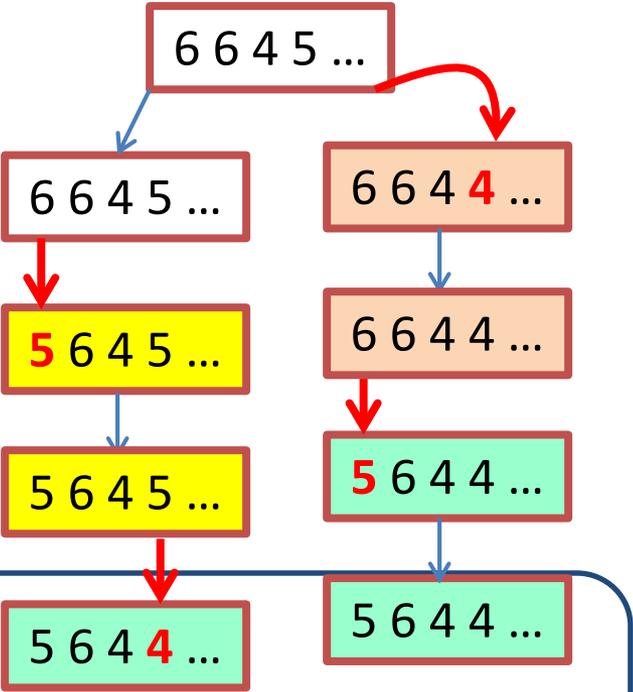


Unwind
time ...

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Identity by State – closer look

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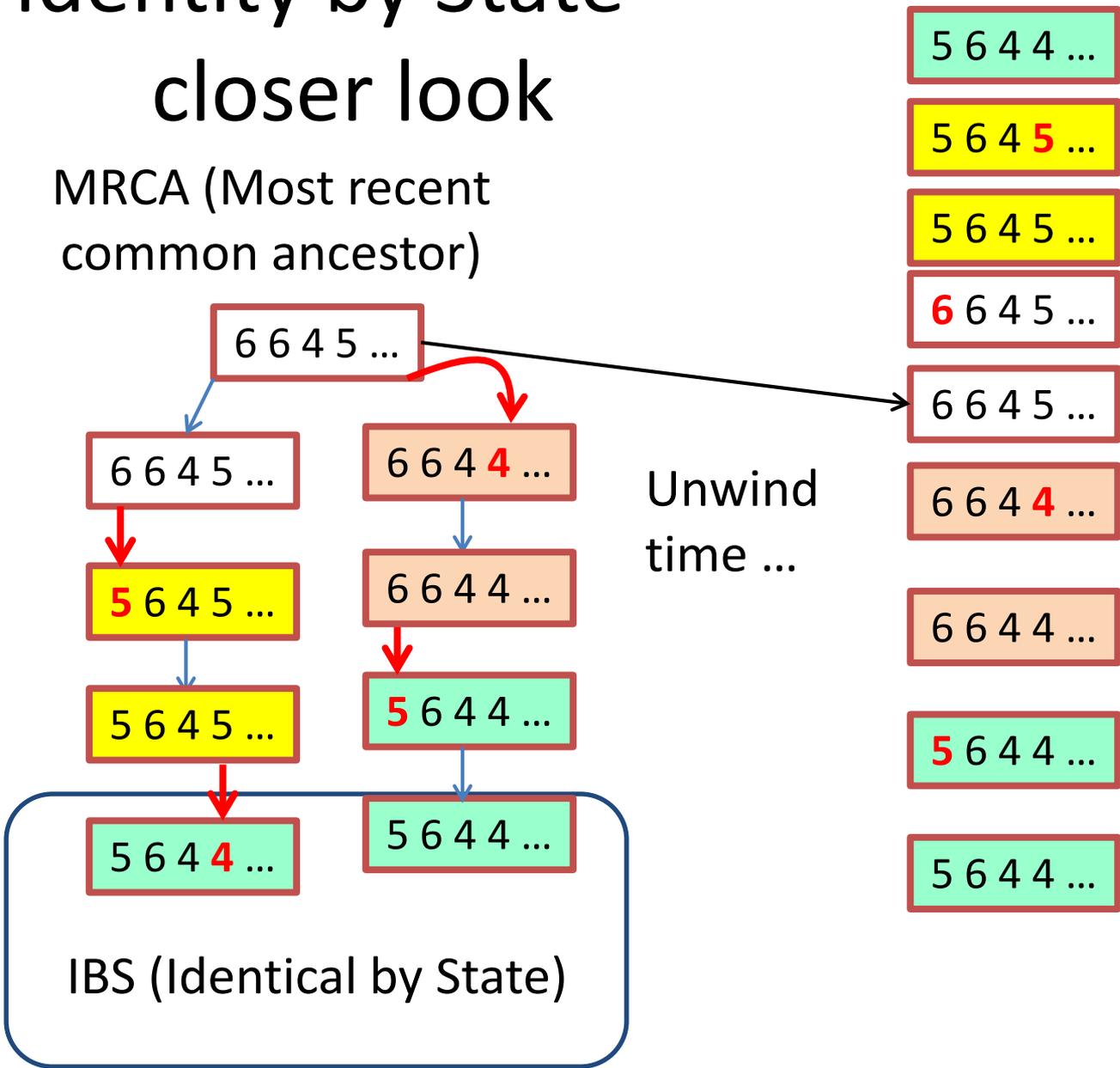
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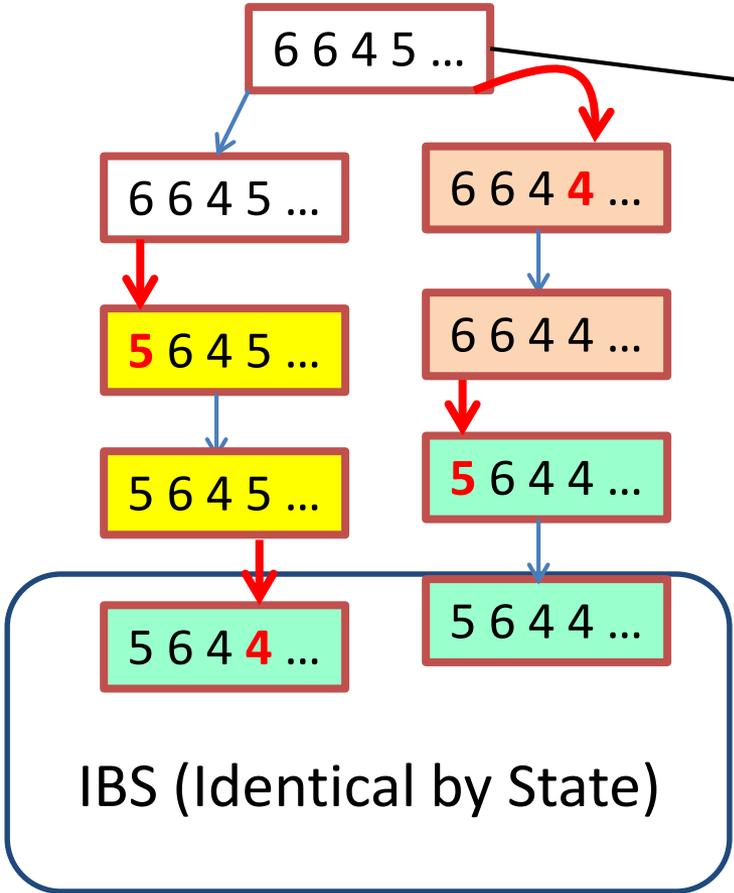
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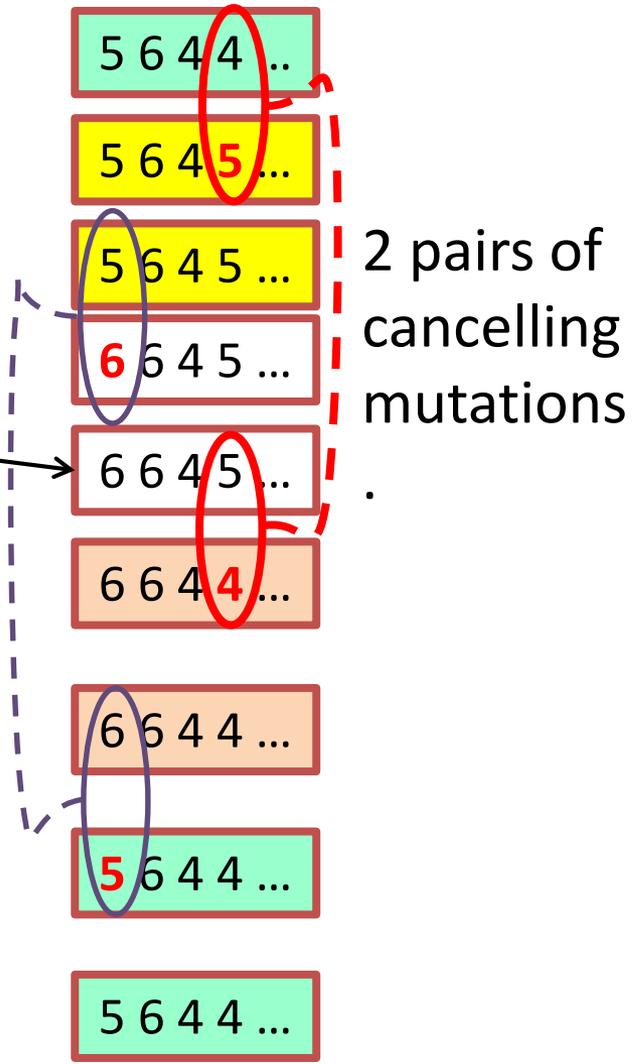


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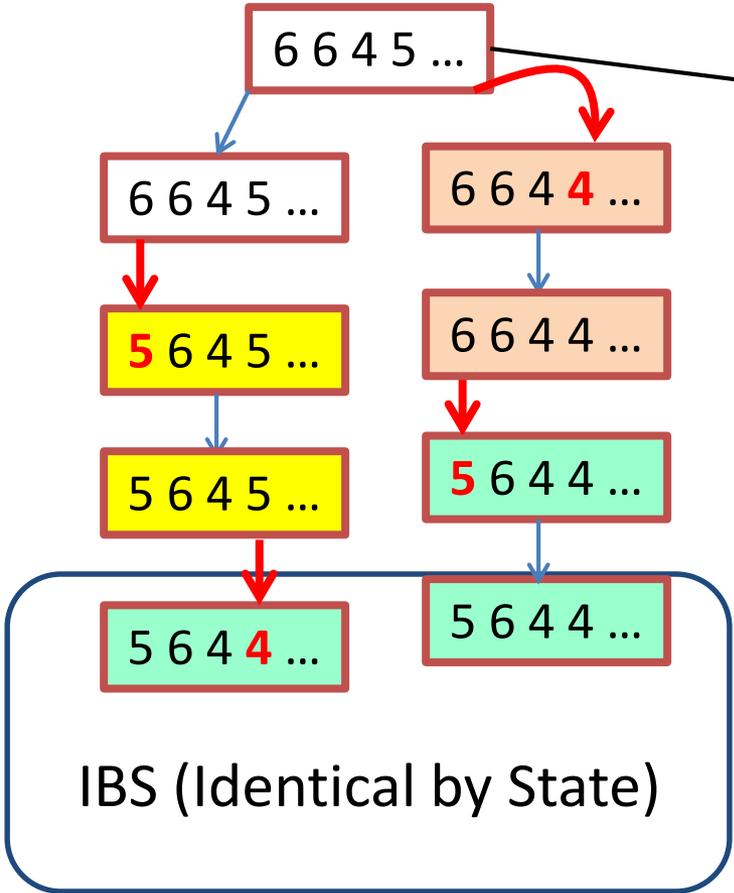


Unwind
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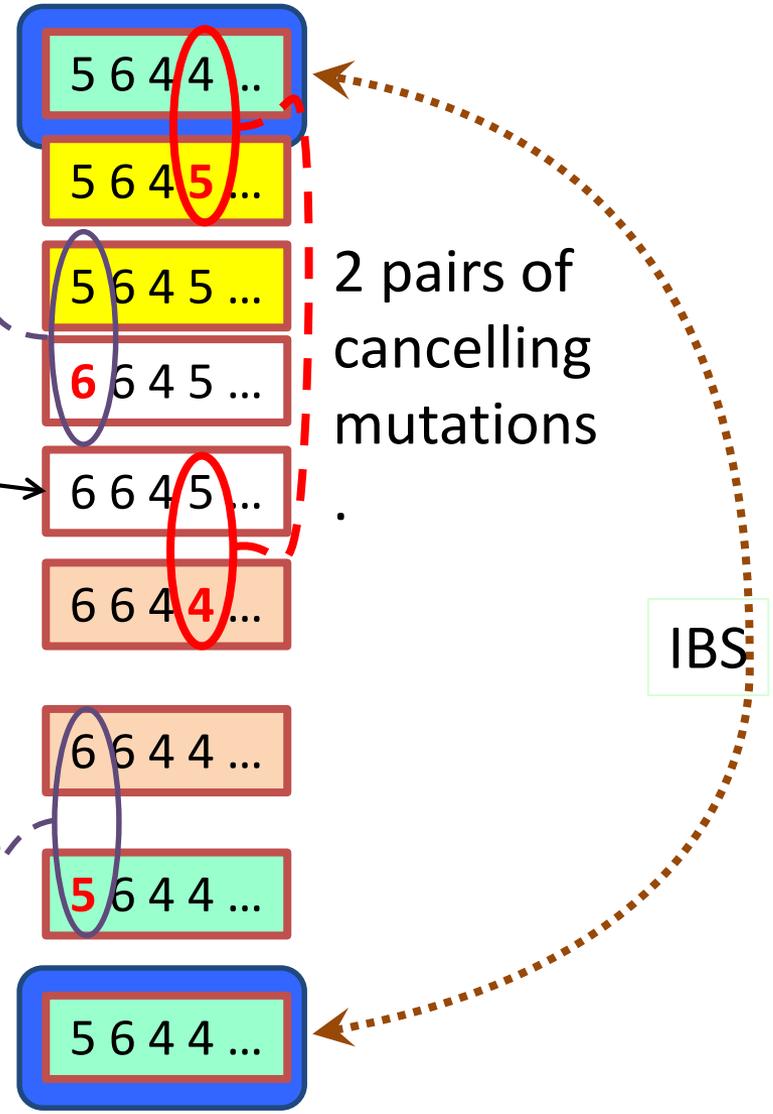


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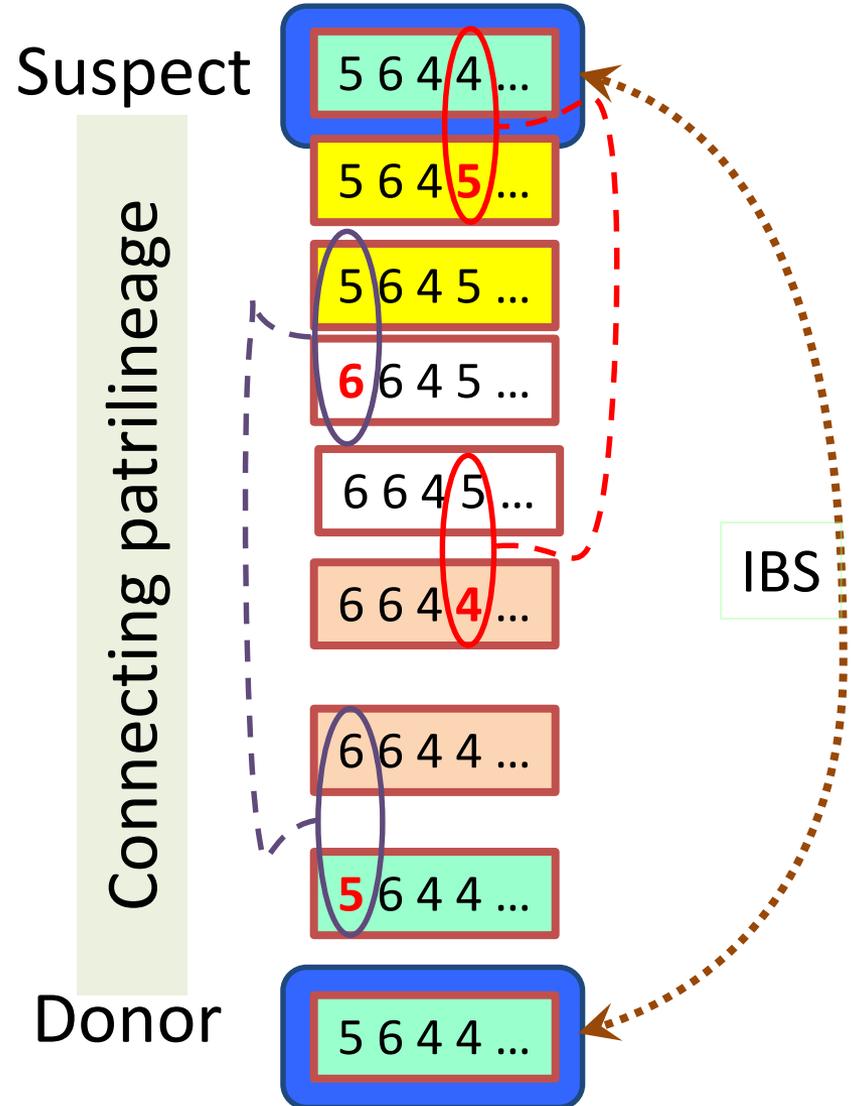
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Unwind time ...

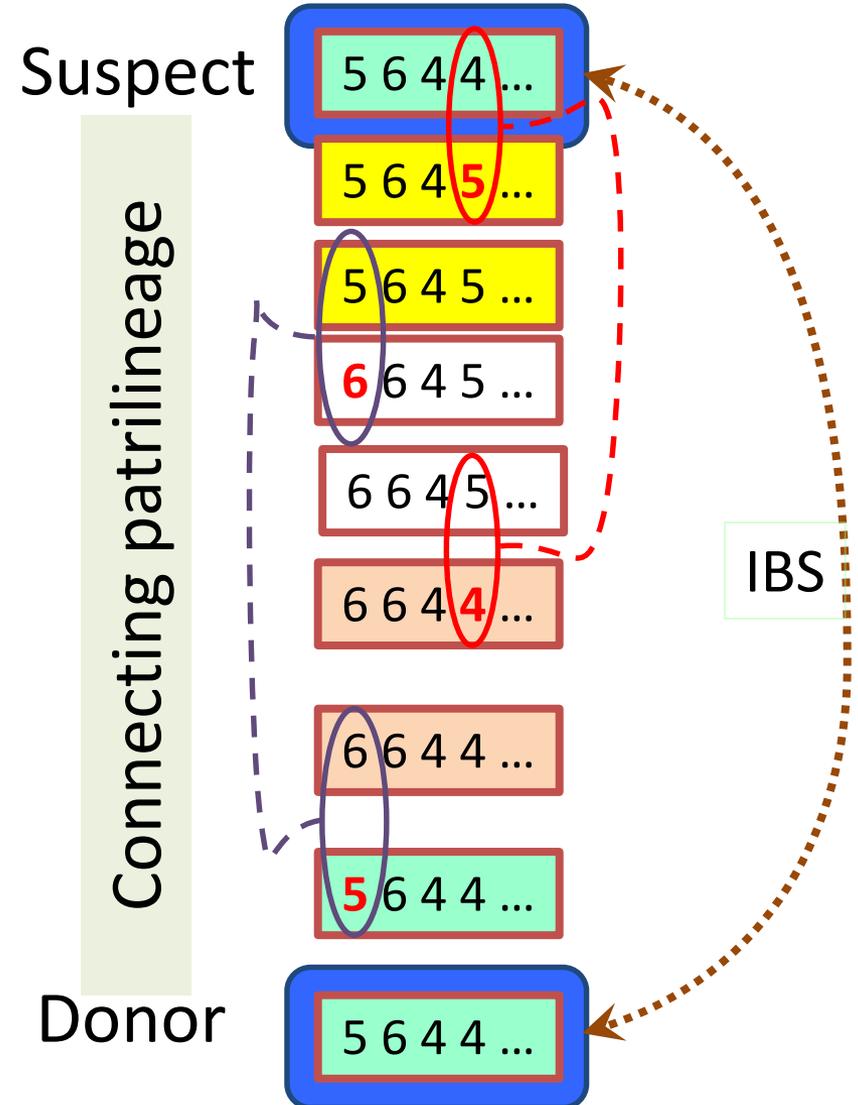


Y haplotype match condition



Y haplotype match condition

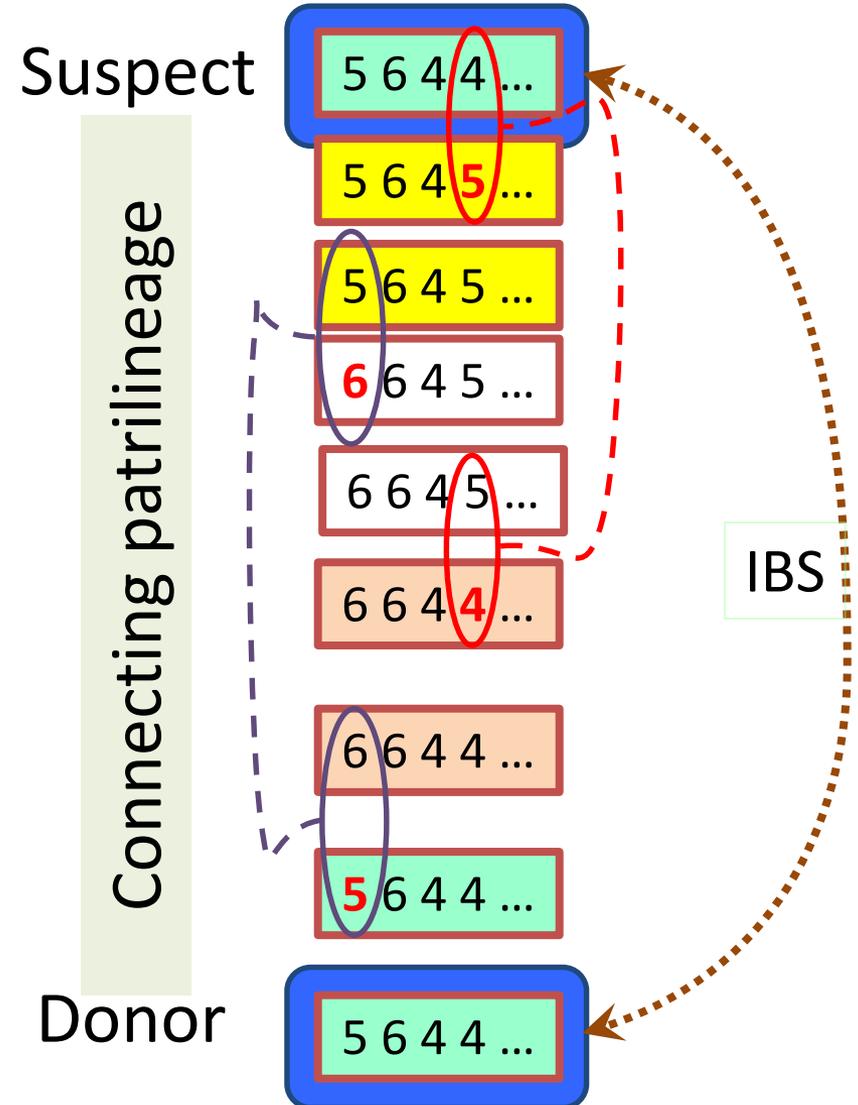
Y haplotypes match (IBS)
if and only if the connecting
patrilineage has n pairs of
cancelling mutations
for some $n \in 0, 1, 2, \dots$



Y haplotype match condition

Y haplotypes match (IBS)
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NB: Ok to apply rule per-locus.



Y match calculation #1 (primitive)

$\text{matchpr} \leftarrow n \text{ ConvMut } (\text{gen } \mu)$

- ⌘ Probability of Convergent Mutation at a locus
- ⌘ Pr(two haplotypes **gen** generations apart match)
- ⌘ **n** = # of (cancelling) mutation pairs
- ⌘ **mu** = Pr(mutation) at each generation

$\text{ways} \leftarrow n \times .! \text{gen}, \text{gen} - n$

⌘ ways to position mutations

$\text{pr} \leftarrow (\mu \div 2) (1 - \mu)^{\times} \cdot * (2 \times n) (\text{gen} - 2 \times n)$

⌘ probability of each way

$\text{matchpr} \leftarrow \text{ways} \times \text{pr}$

⌘ total probability of all ways

Y match calculation #2

$\Pr(\text{match})$

given g generations of separation

$\square IO \leftarrow 0$

$\text{maxn} \leftarrow 11$ A 10 mutation pairs per locus is plenty

$\text{gens} \leftarrow 1 \text{ngen} + 1$ A $\text{gens} \leftarrow 0, 1, \dots, \text{ngen}$ generations separation

$\text{ibS} \leftarrow (1 - \text{maxn})^{\text{ConvMut}} \text{gens}^{\text{mu}}$ A $\rho \leftrightarrow \text{maxn ngen}$ ($\neq Y \text{loci}$)

A $\text{ibS}[n; g; l] = \Pr(\text{match at locus } l \mid g \text{ generations including } n \text{ mut'n pairs})$

A Consider 3 matching probabilities:

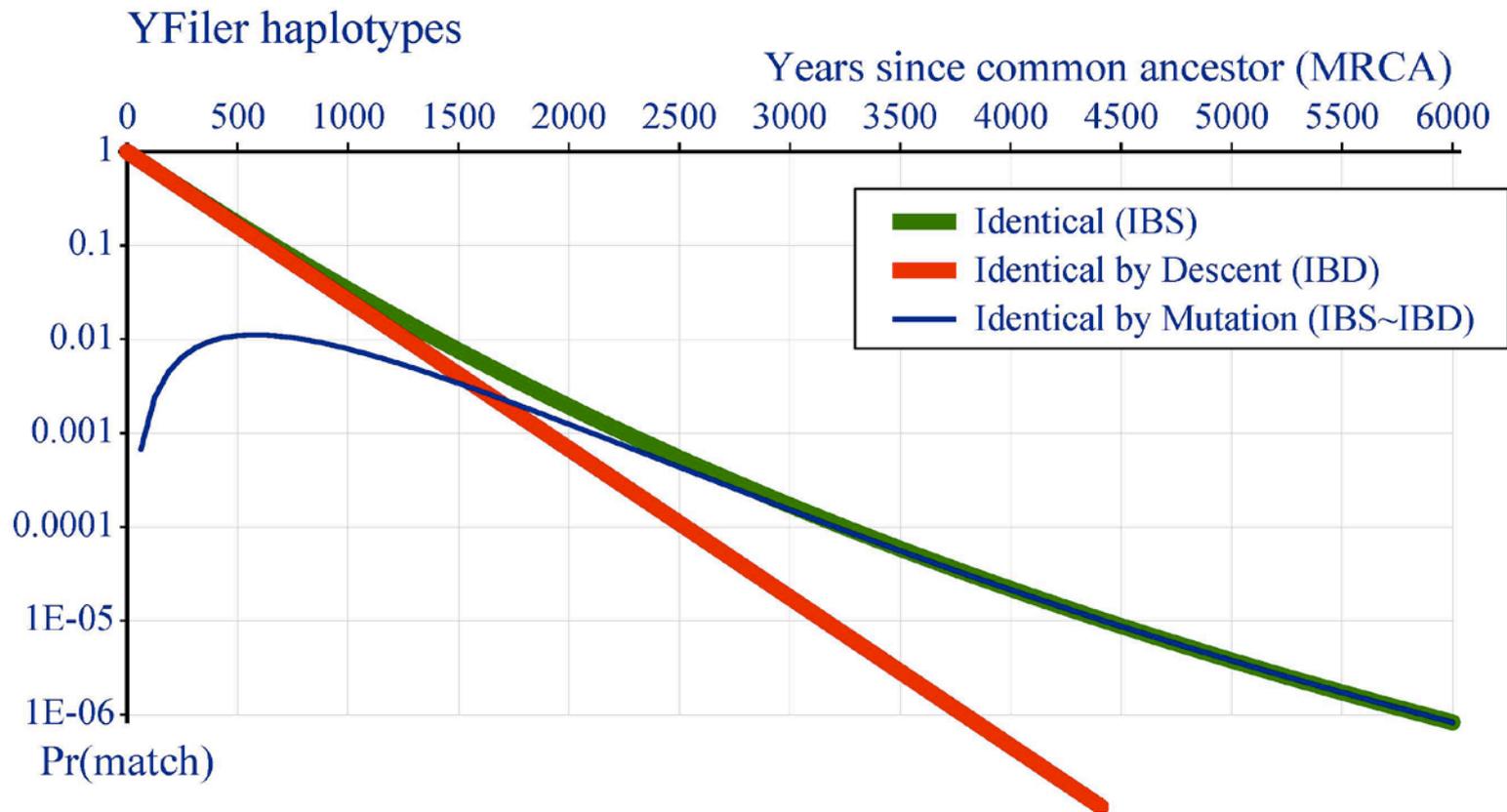
$\text{IBD} \leftarrow x / \text{ibS}[0; ;]$ A match all loci, no mutations

$\text{IBS} \leftarrow x / + \neq \text{ibS}$ A match all loci, allow mutations

$s\text{IBS} \leftarrow \text{IBS} - \text{IBD}$ A strictly IBS (some mutations)

IBD dominates Yfiler matching

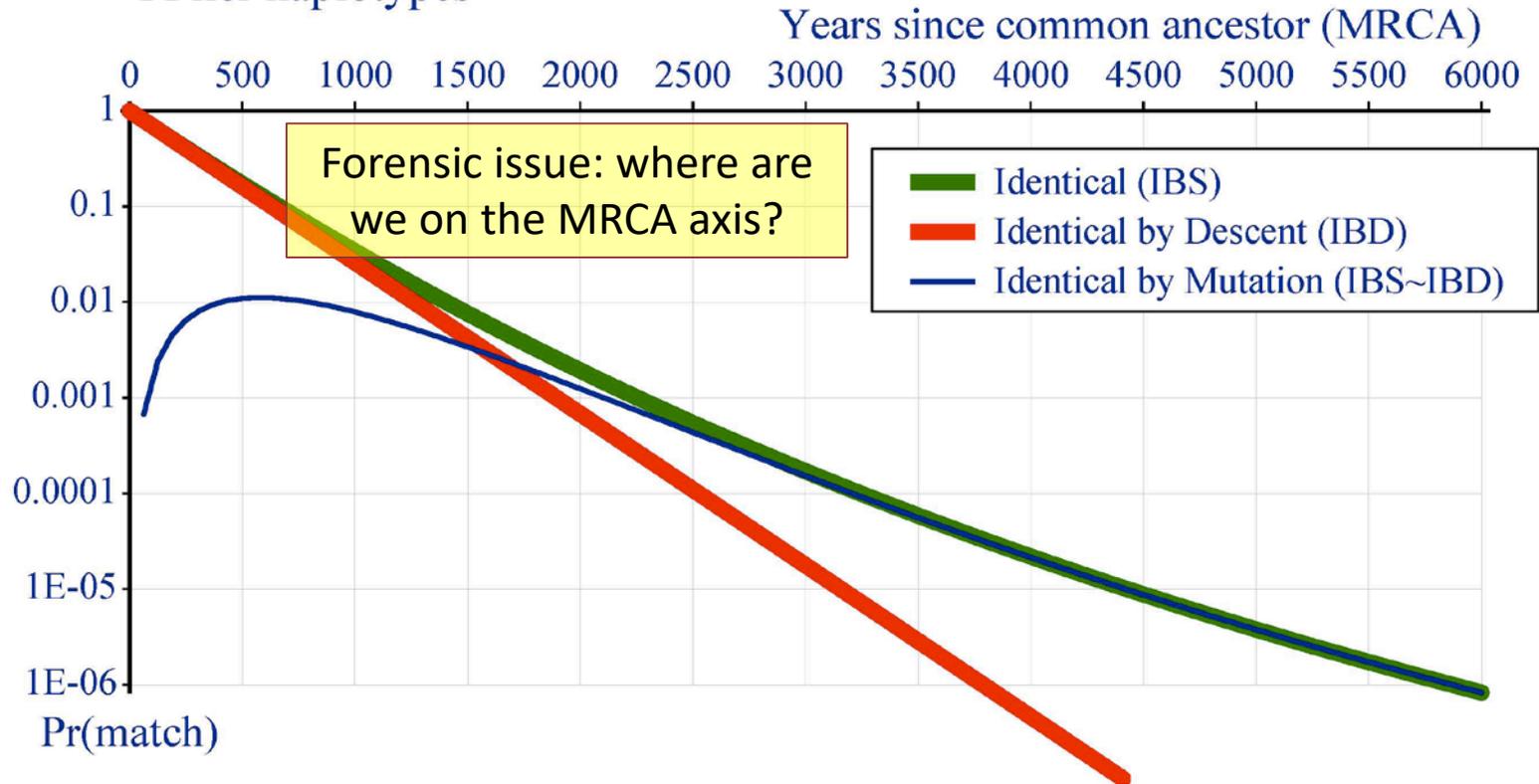
Identical by Descent vs by Mutation



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Identical by Descent vs by Mutation

Yfiler haplotypes

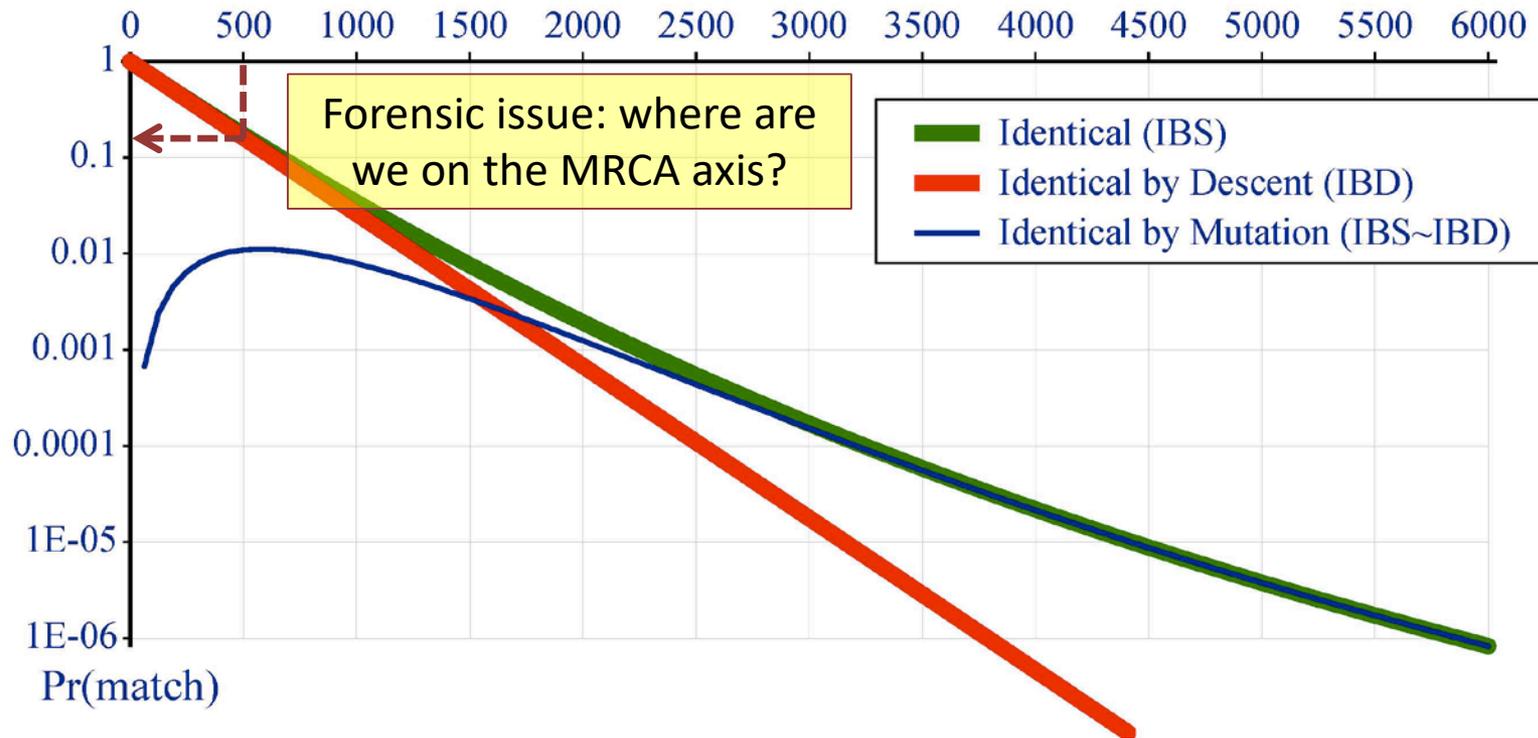


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Identical by Descent vs by Mutation

YFiler haplotypes

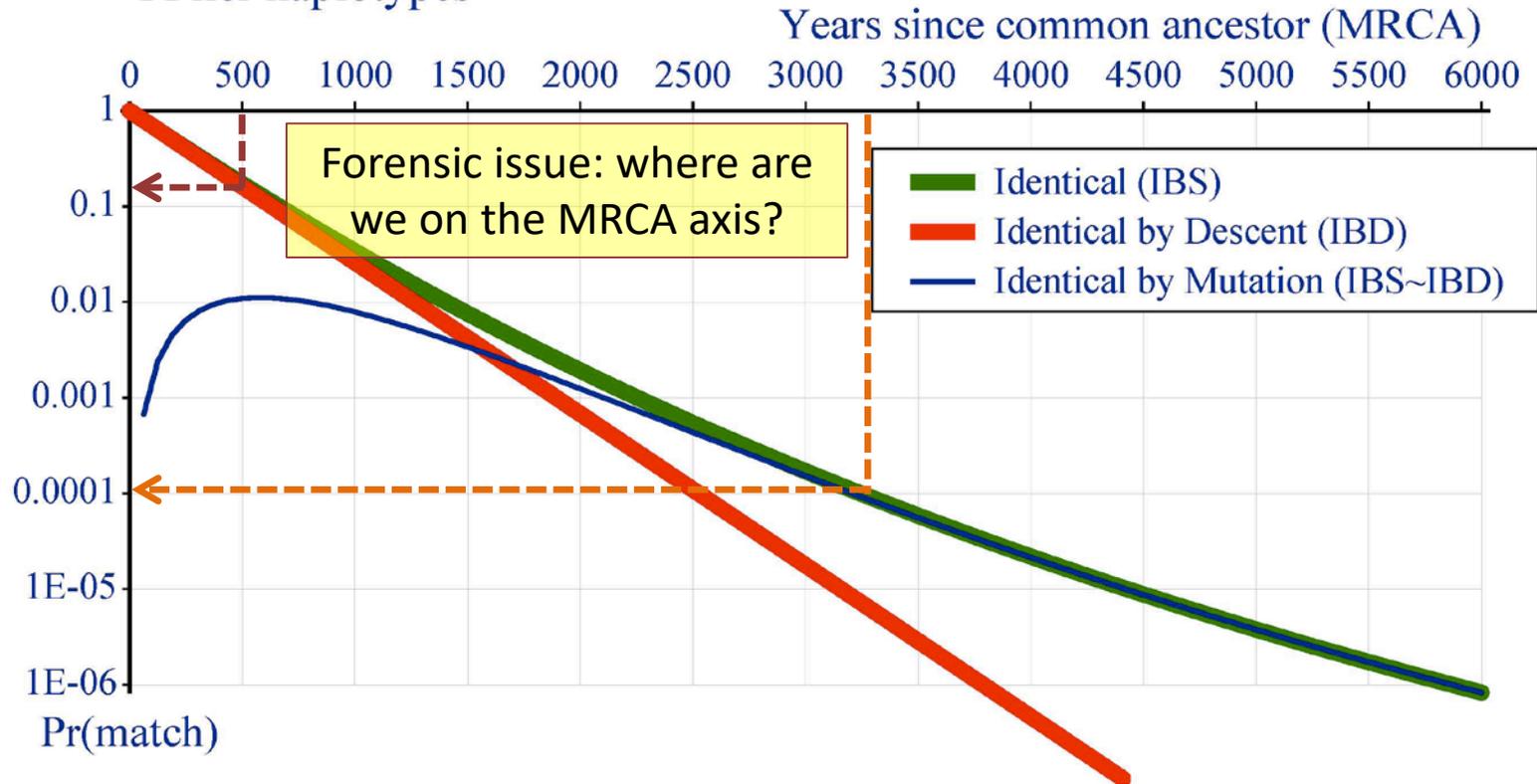
Years since common ancestor (MRCA)



IBD dominates Yfiler matching

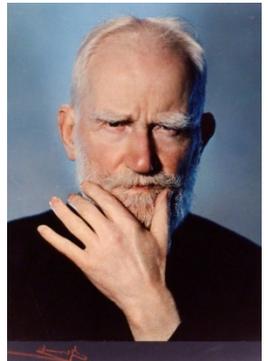
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Common boilerplate from forensic lab DNA matching report

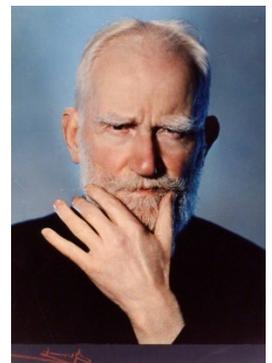
The longer I live the more I see that I am never wrong about anything, and that all the pains that I have so humbly taken to verify my notions have only wasted my time.



Common boilerplate from forensic lab DNA matching report

Due to the paternal inheritance of the Y-chromosome, all males from the same male lineage are expected to share the same Y-STR profile.

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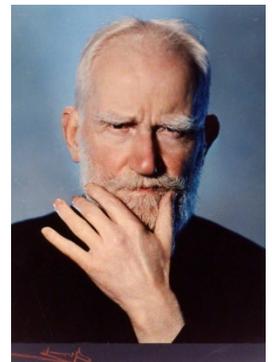


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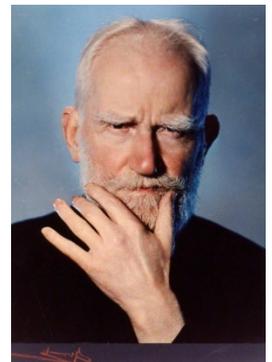


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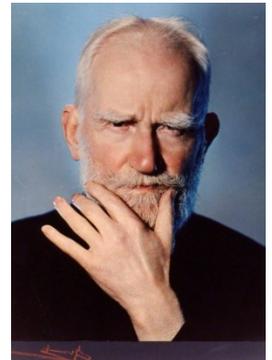


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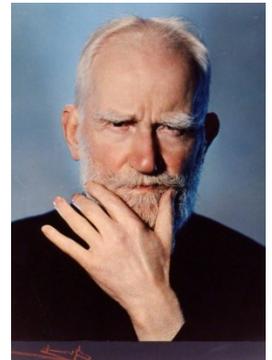


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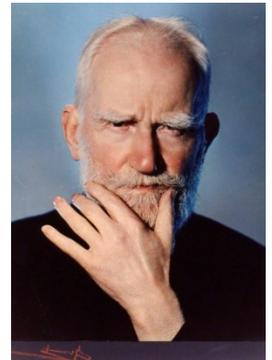


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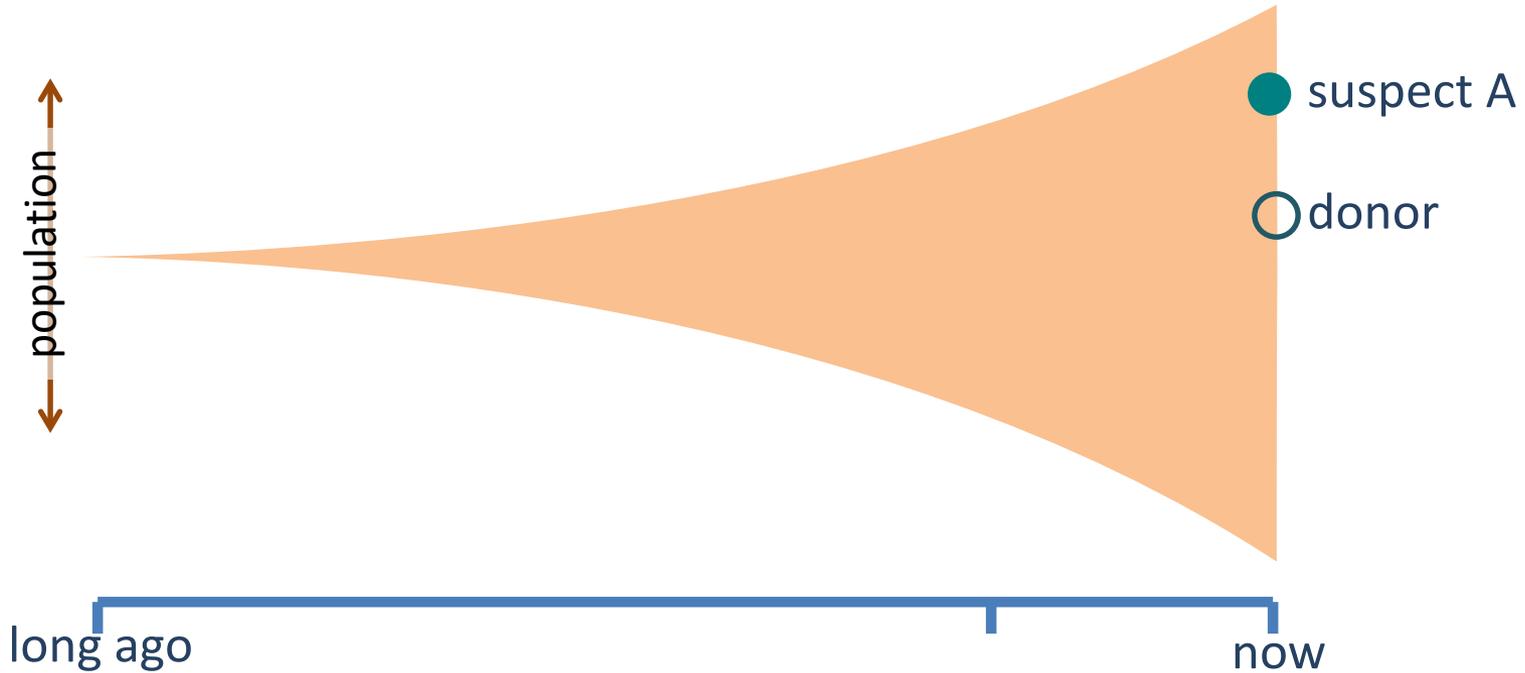
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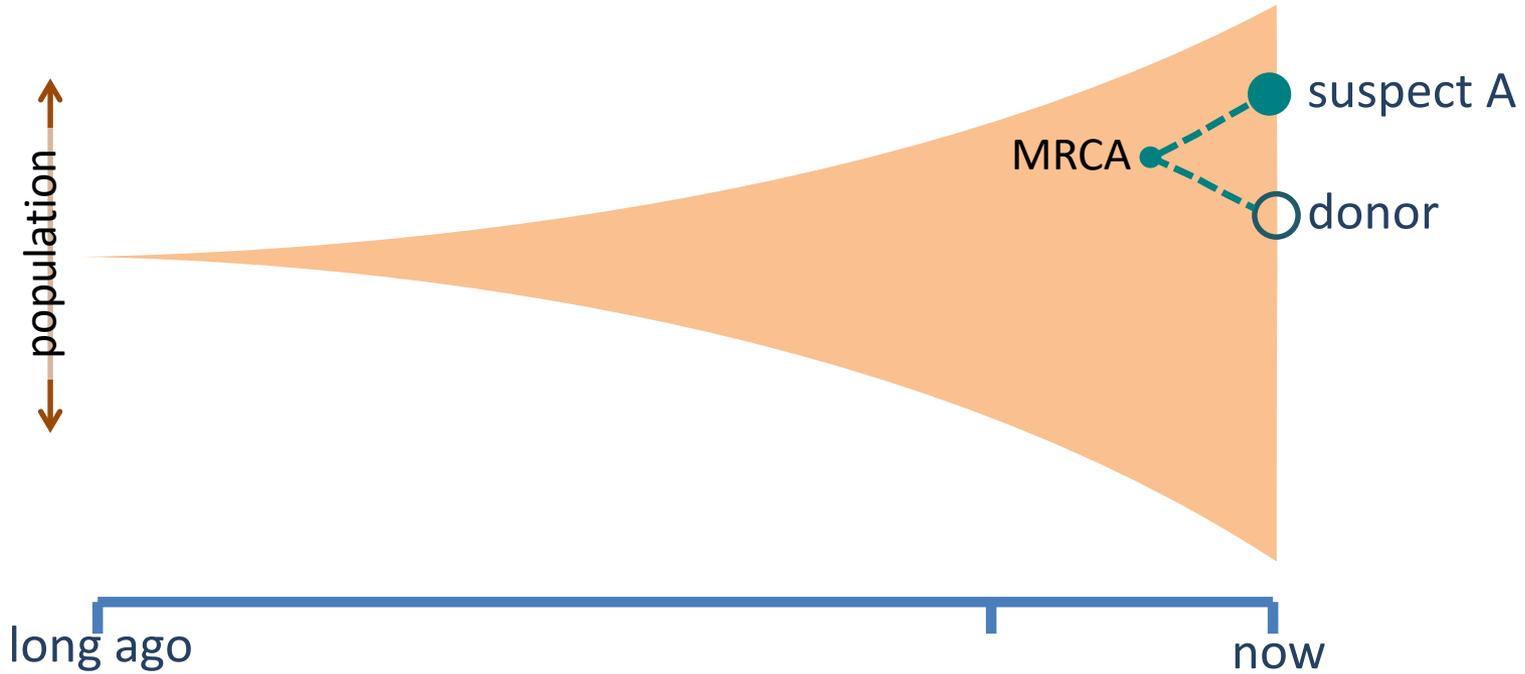
Modeling a population

1. constant population growth rate from 1 founding man to **N** men today.



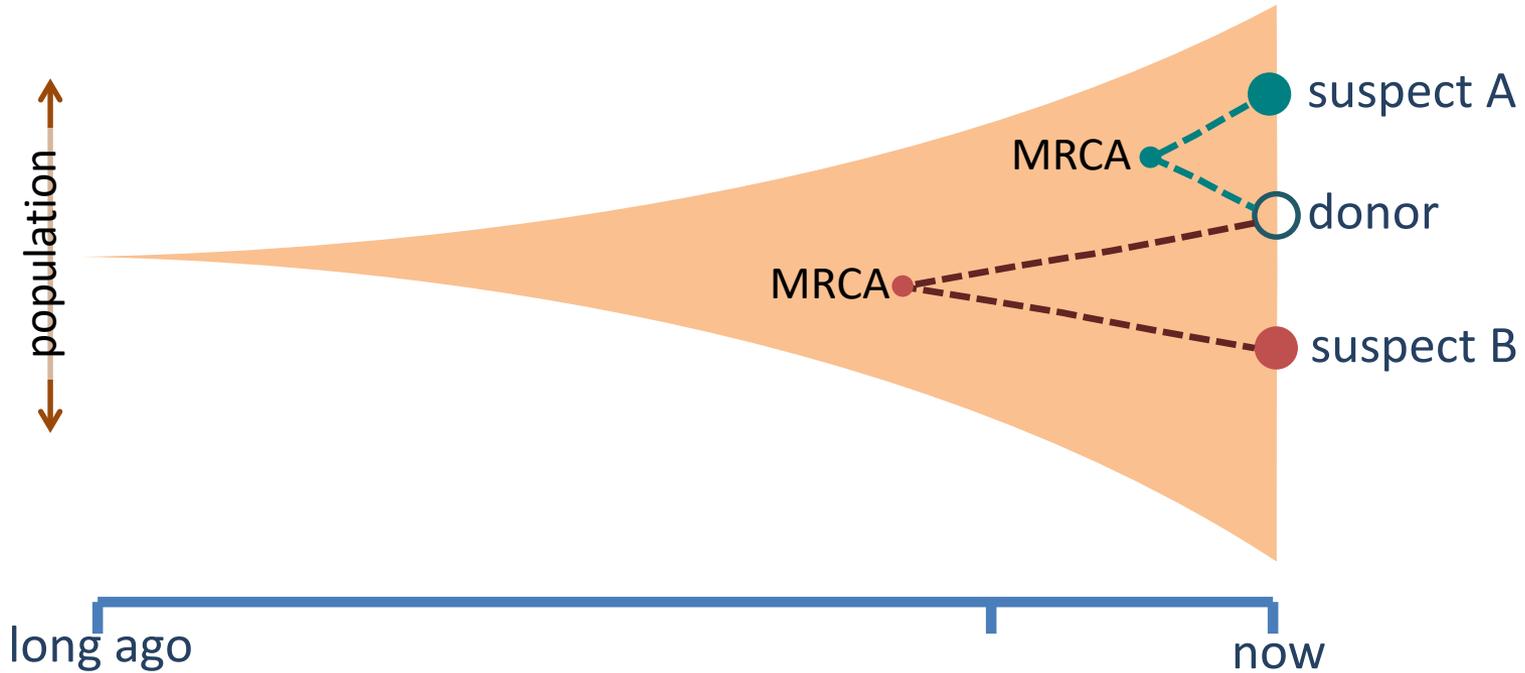
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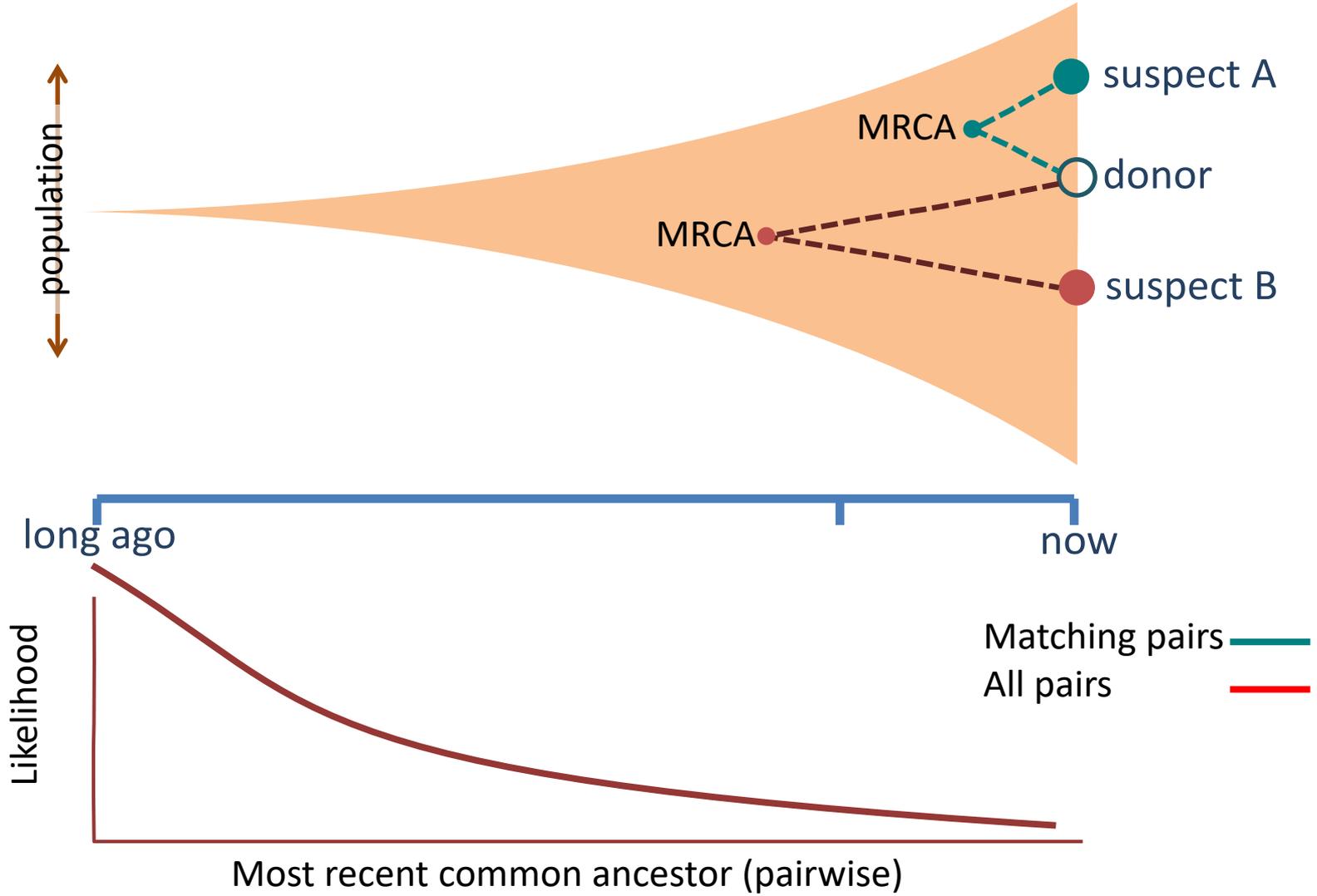
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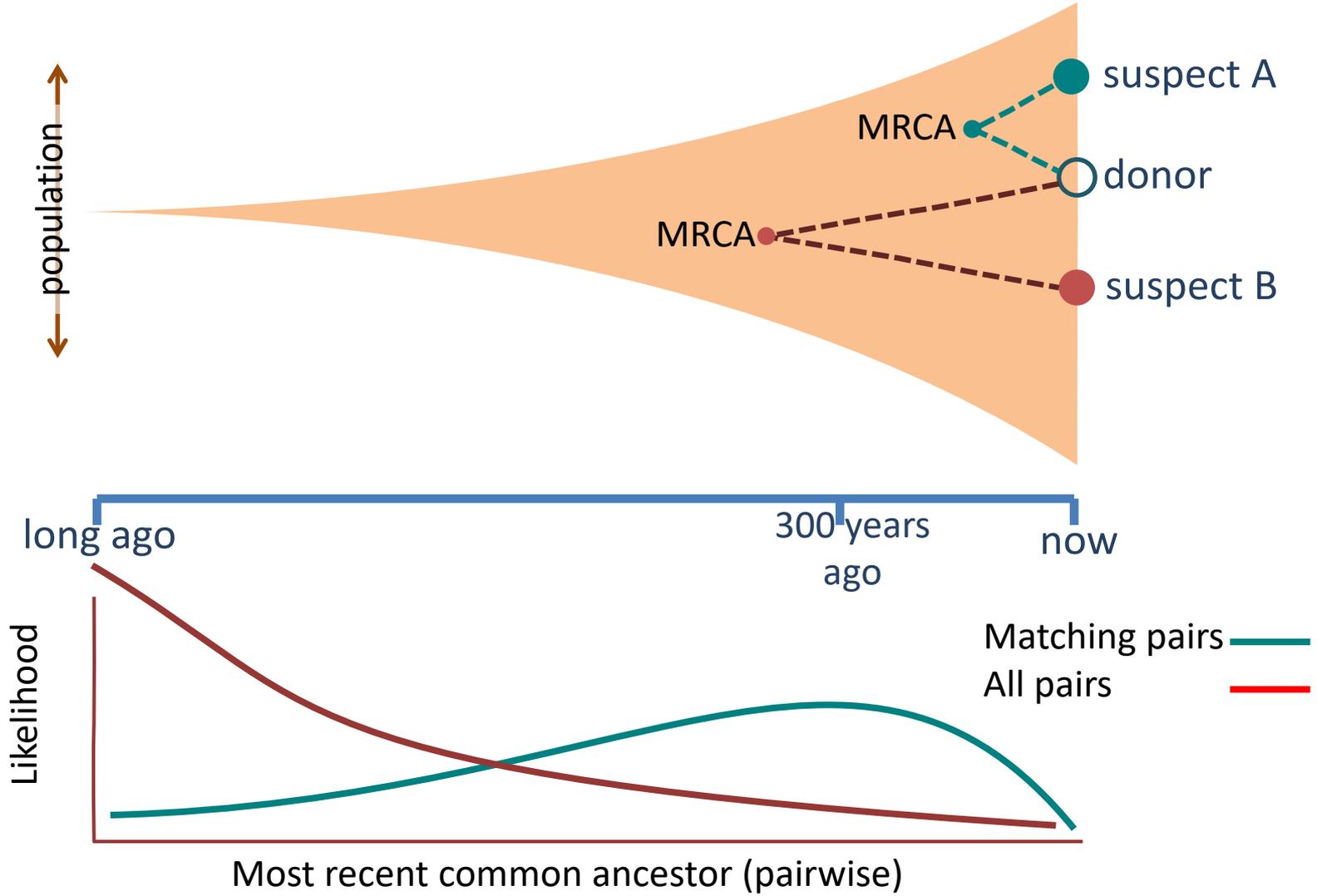
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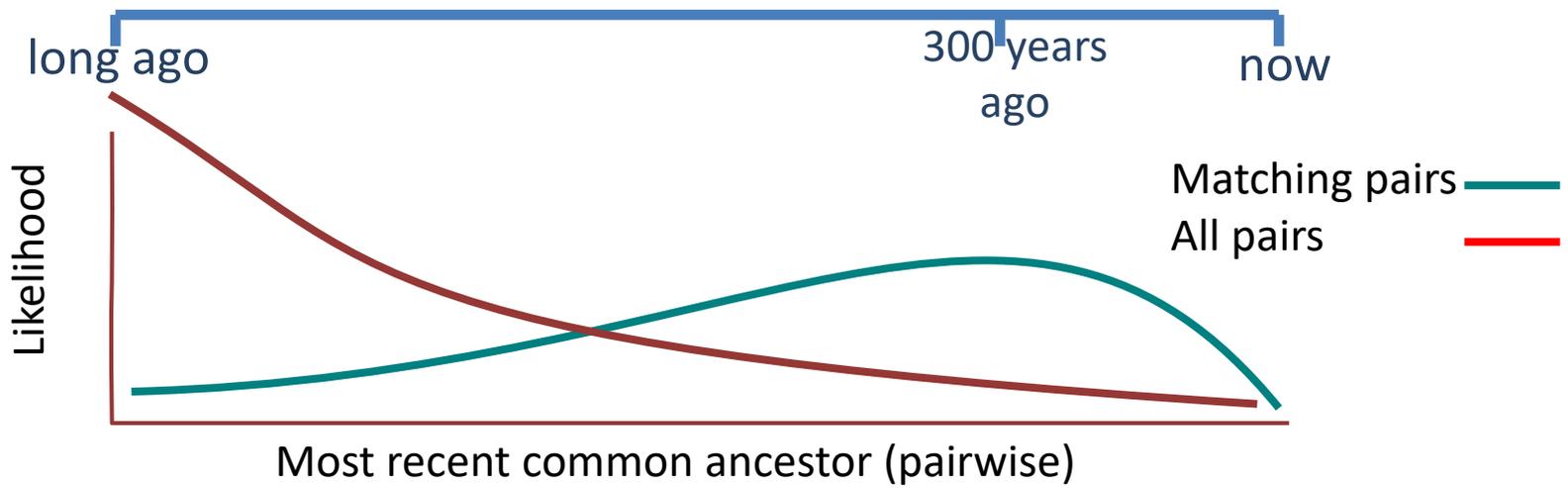
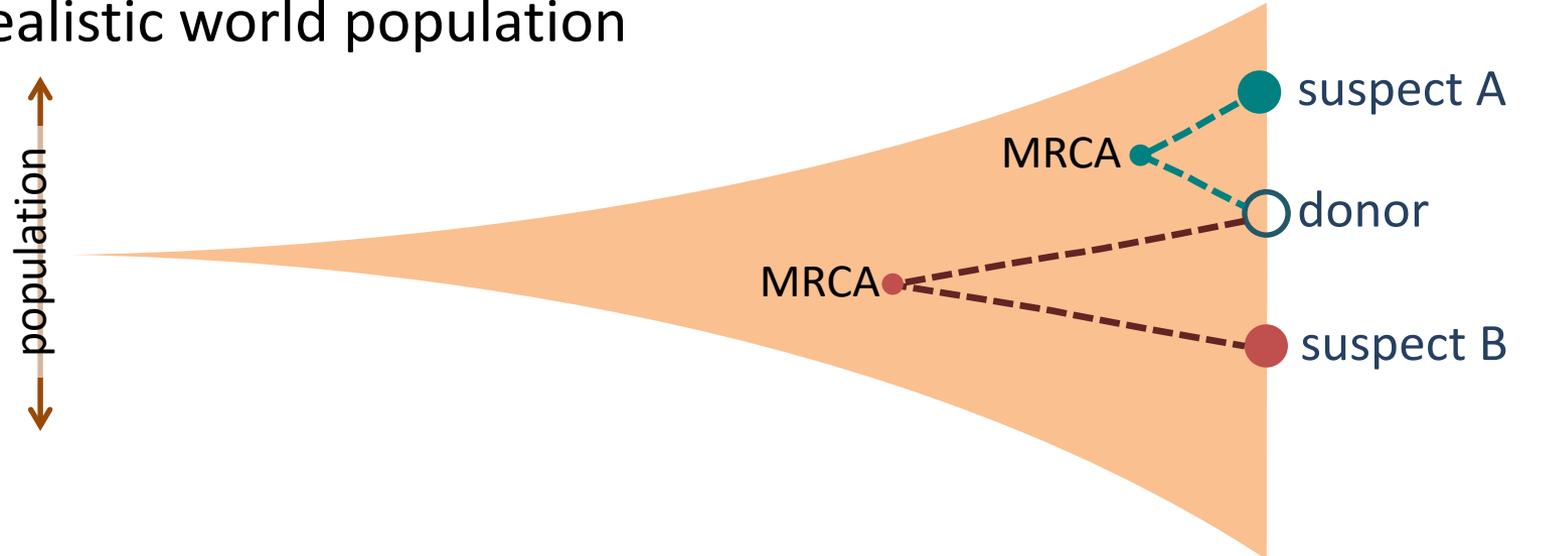
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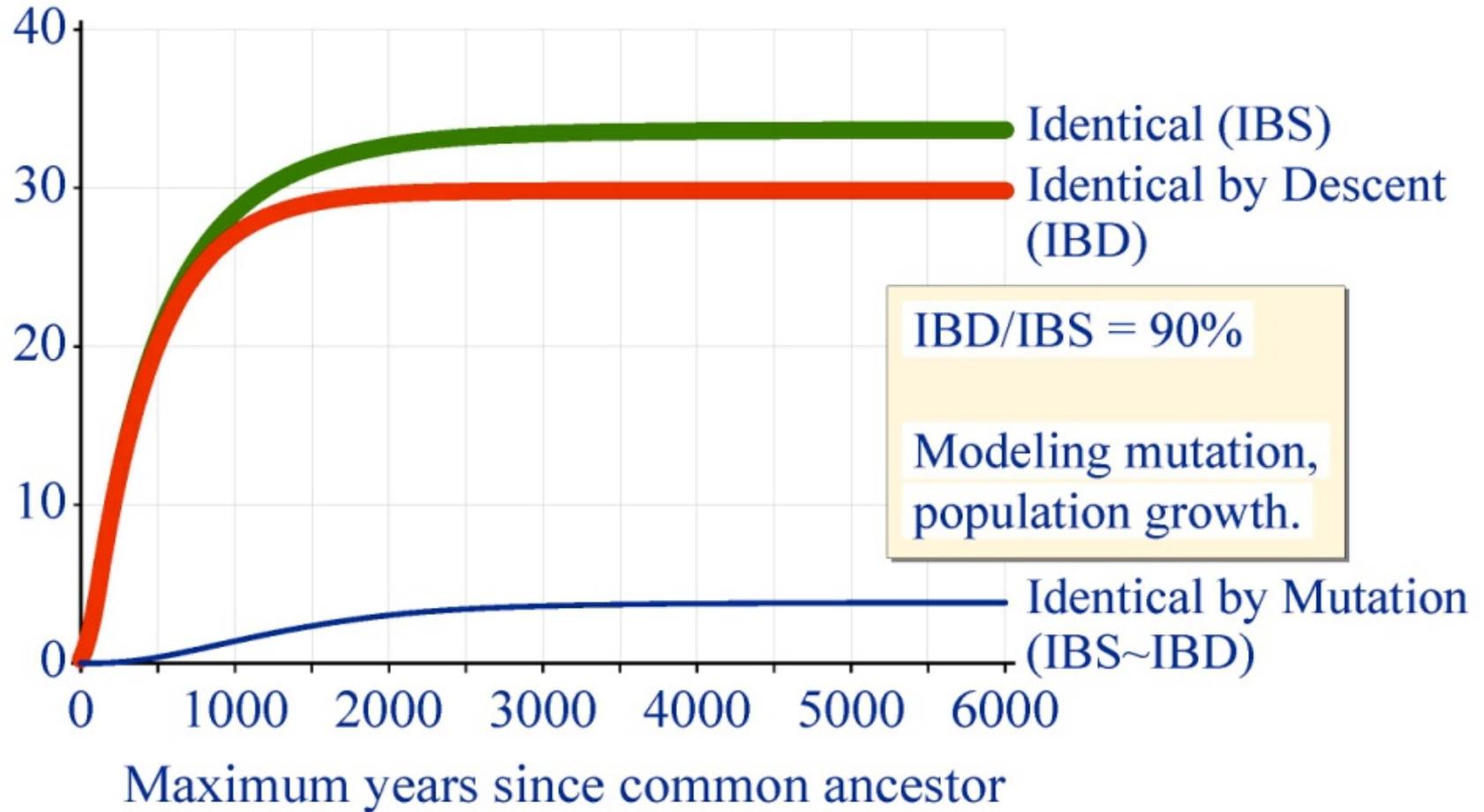
Modeling a population

- 1. constant population growth rate from 1 founding man to **N** men today.
- 2. realistic world population



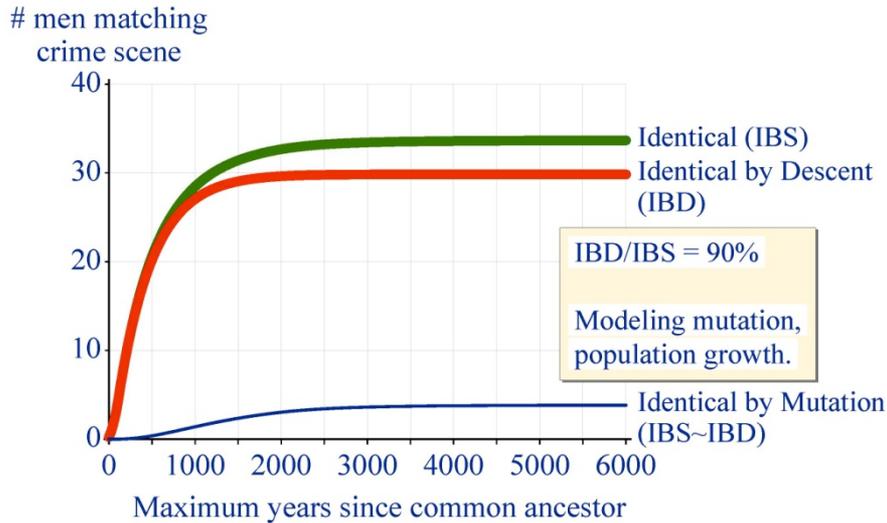
Identity by Descent vs by Mutation, YFiler

men matching
crime scene



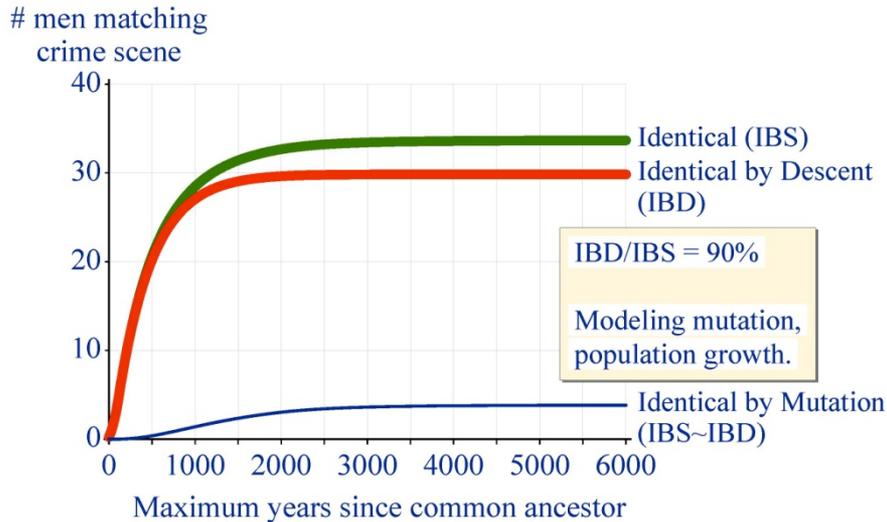
Haplotypes are NOT just super-polymorphic loci

Identity by Descent vs by Mutation, YFiler



Haplotypes are NOT just super-polymorphic loci

Identity by Descent vs by Mutation, YFiler



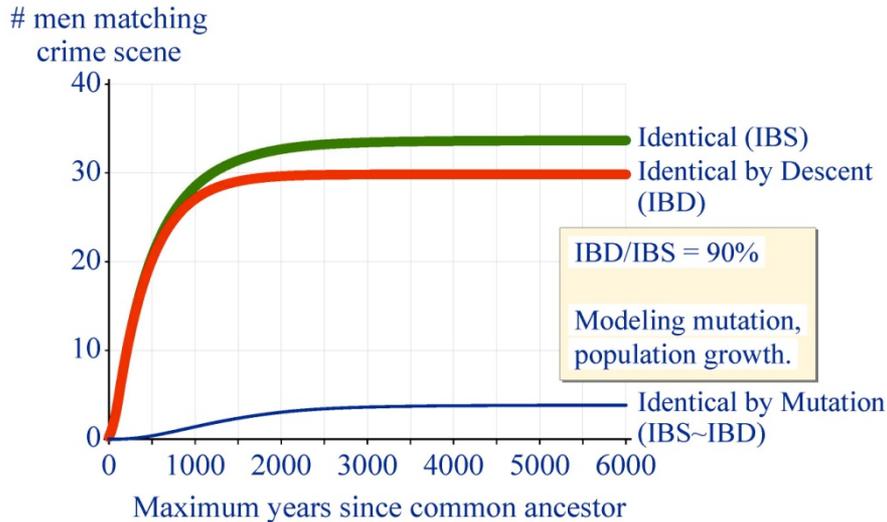
Haplotype

matching is dominated by

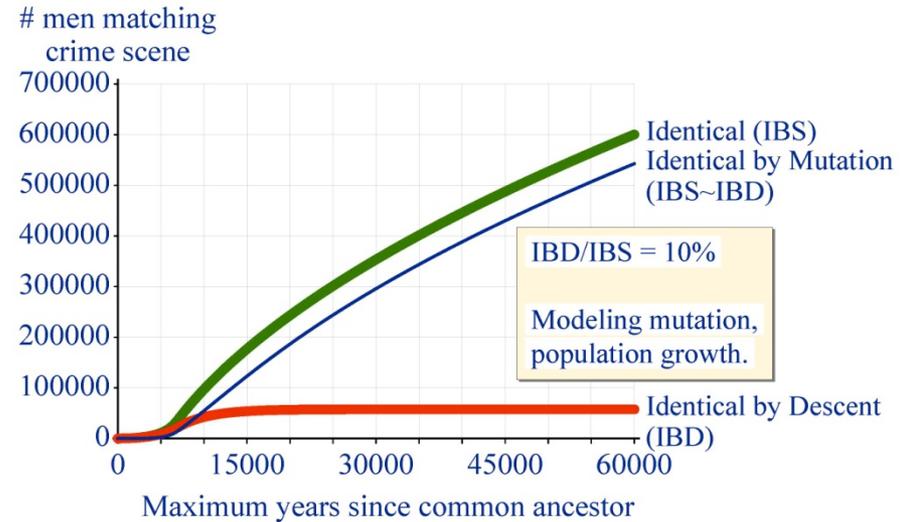
IBD – patrilineal link without intervening mutation.

Haplotypes are NOT just super-polymorphic loci

Identity by Descent vs by Mutation, YFiler



Identity by Descent vs by Mutation, 1 locus



Haplotype

STR locus

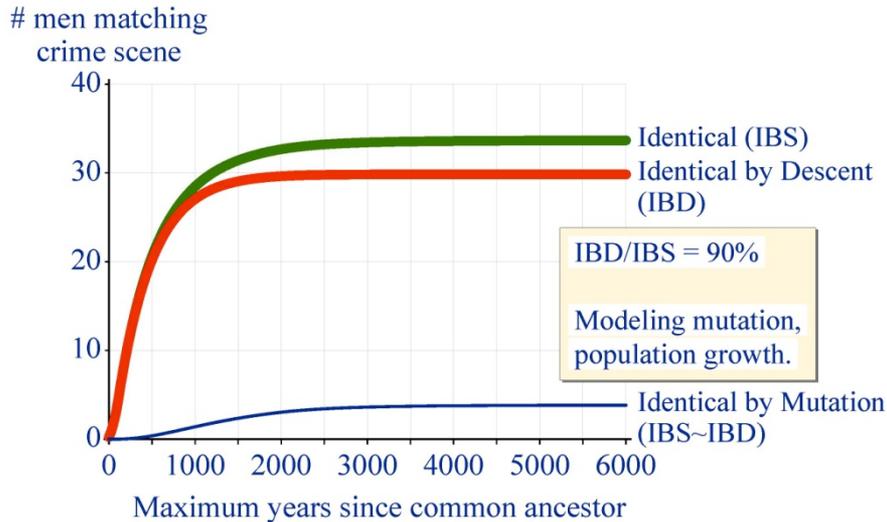
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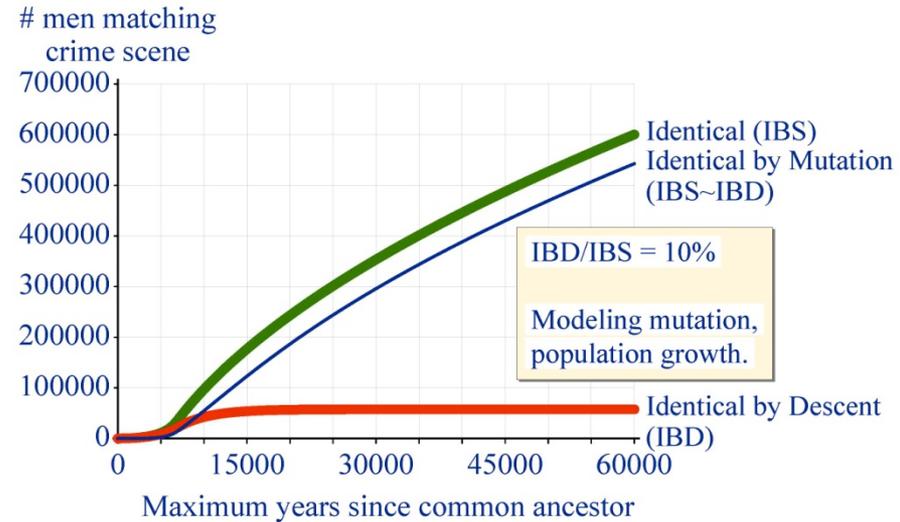
coincidence.

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Haplotype

STR locus

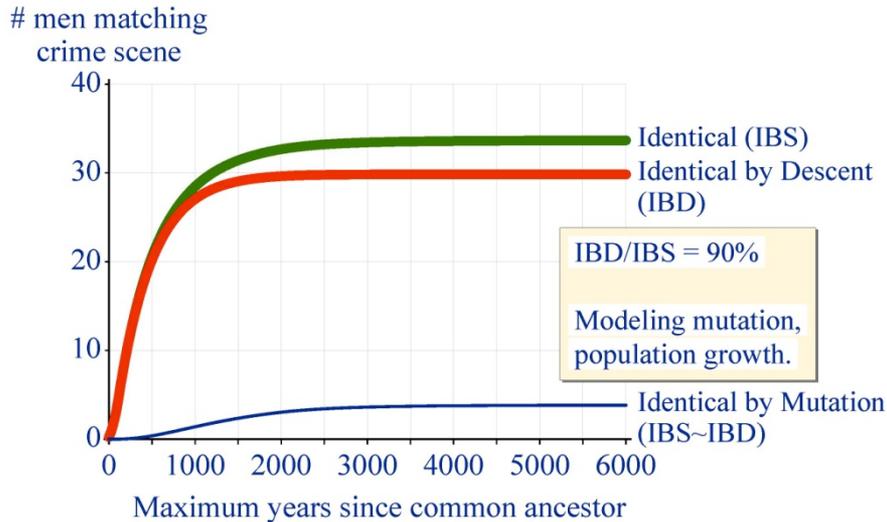
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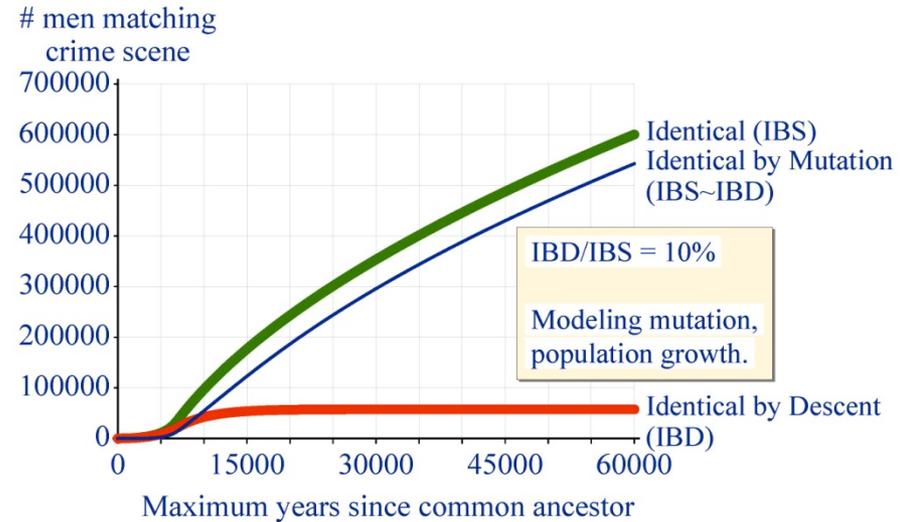
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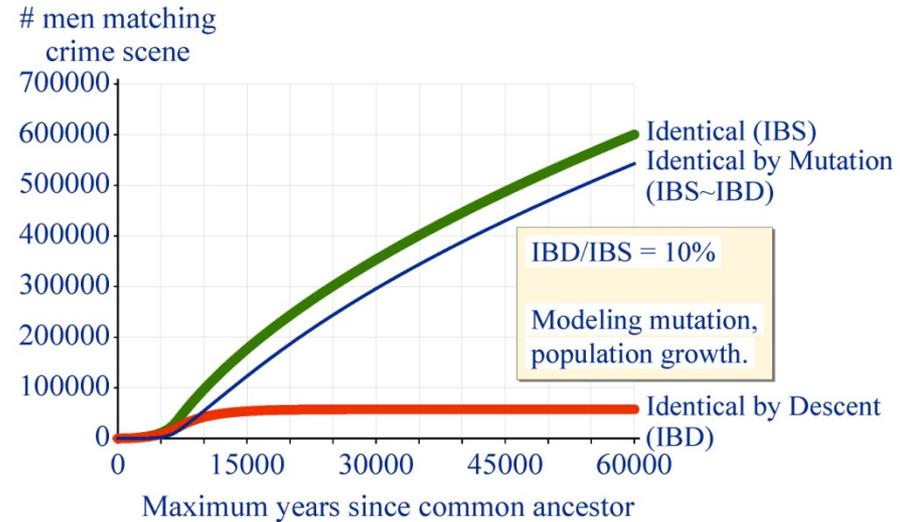
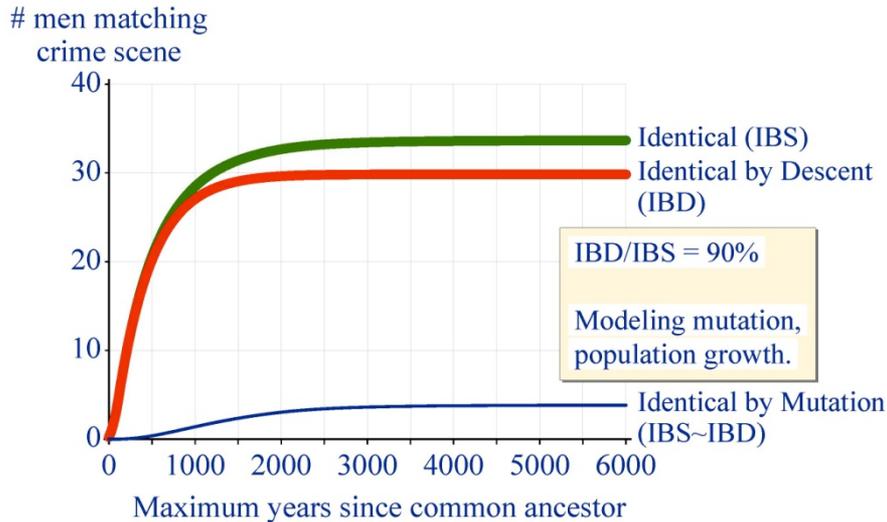
coincidence.

Haplotypes are NOT just super-polymorphic loci

Moral –

Y-haplotype modeling rule #1:
All men are related.

Identity by Descent vs by Mutation, YFiler



Haplotype

STR locus

matching is dominated by

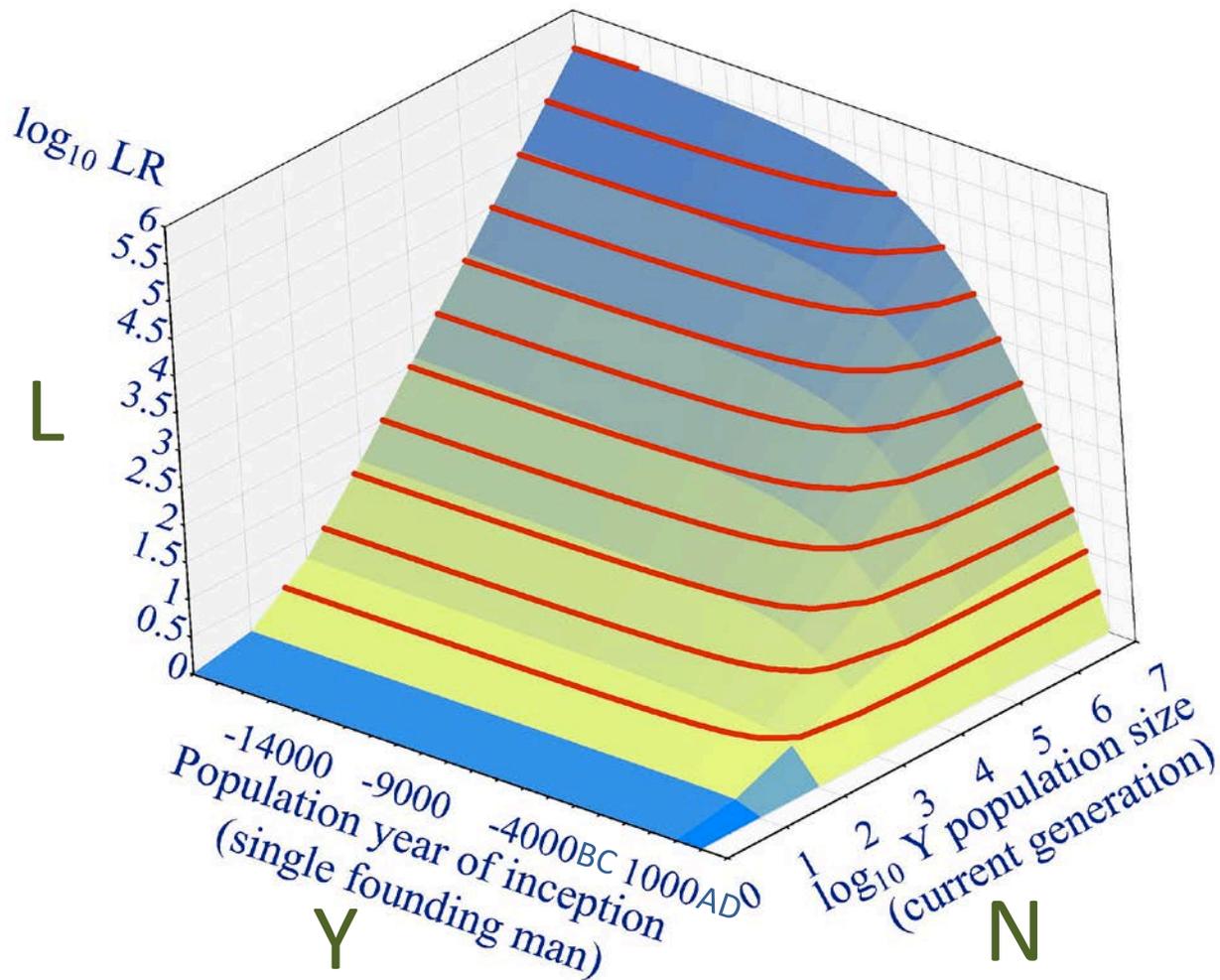
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Evidential strength of Y haplotype match

IBS matching probability

YFiler

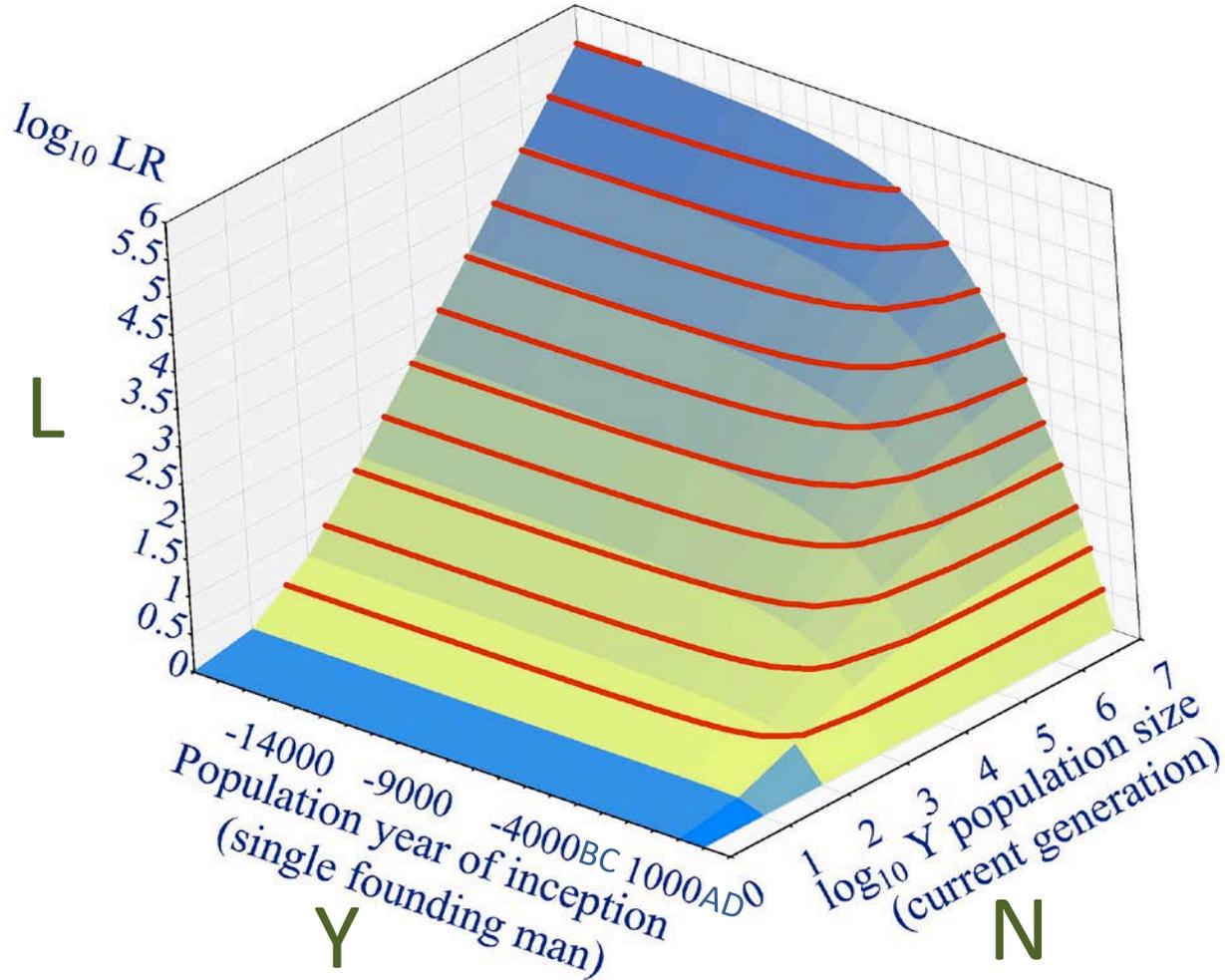


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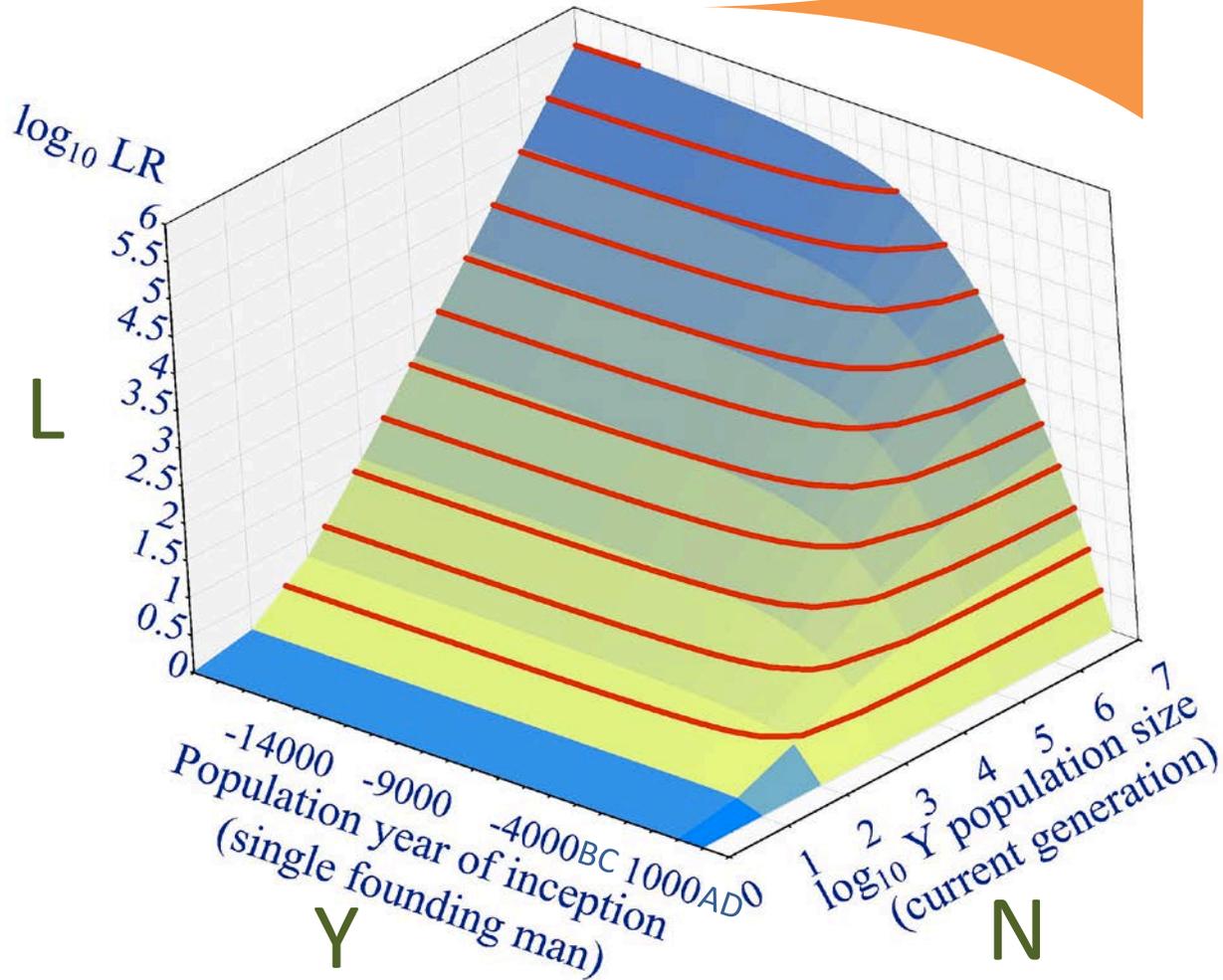
Model: constant population growth rate



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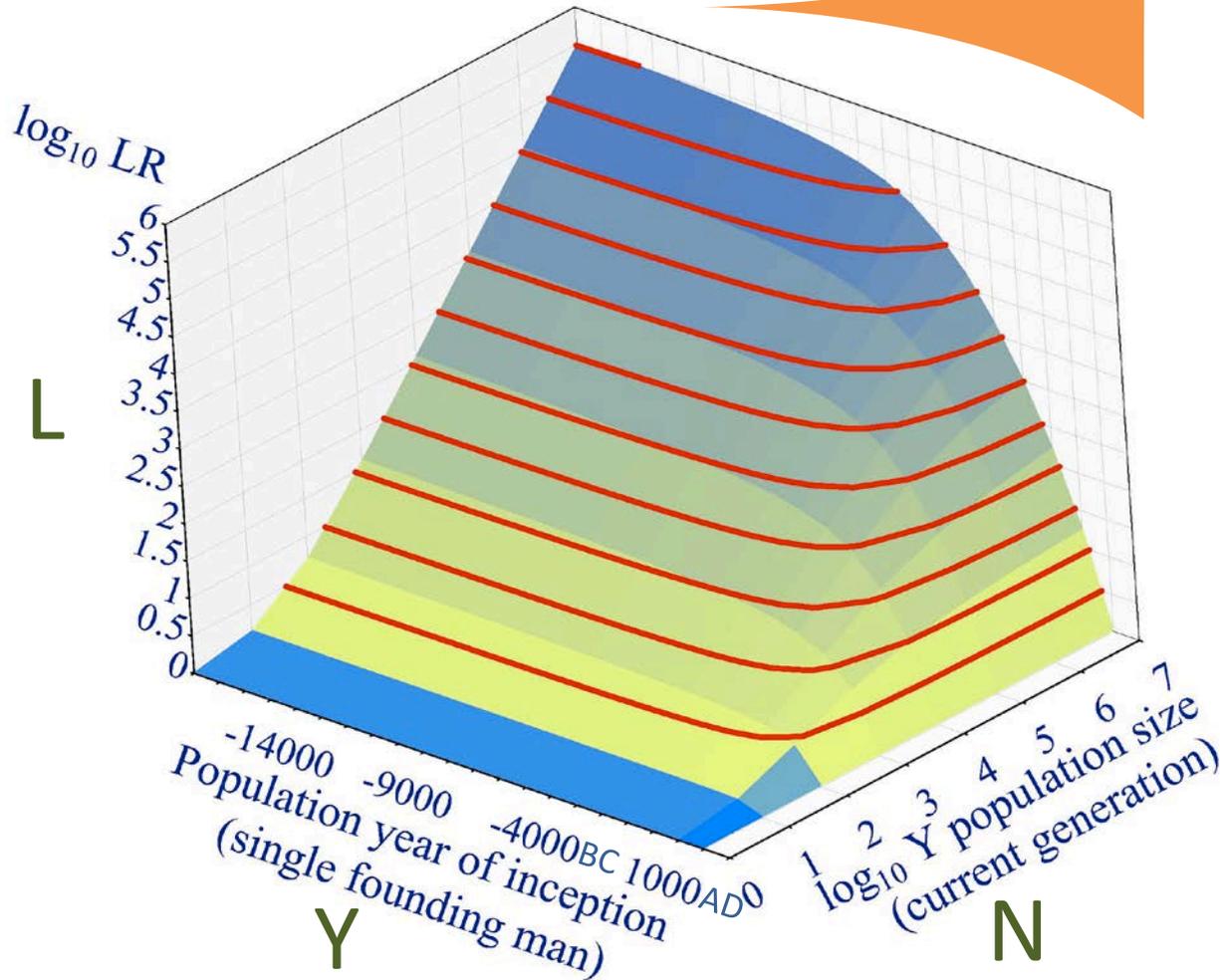


Model: constant population growth rate from 1 founding man to **N** men today..

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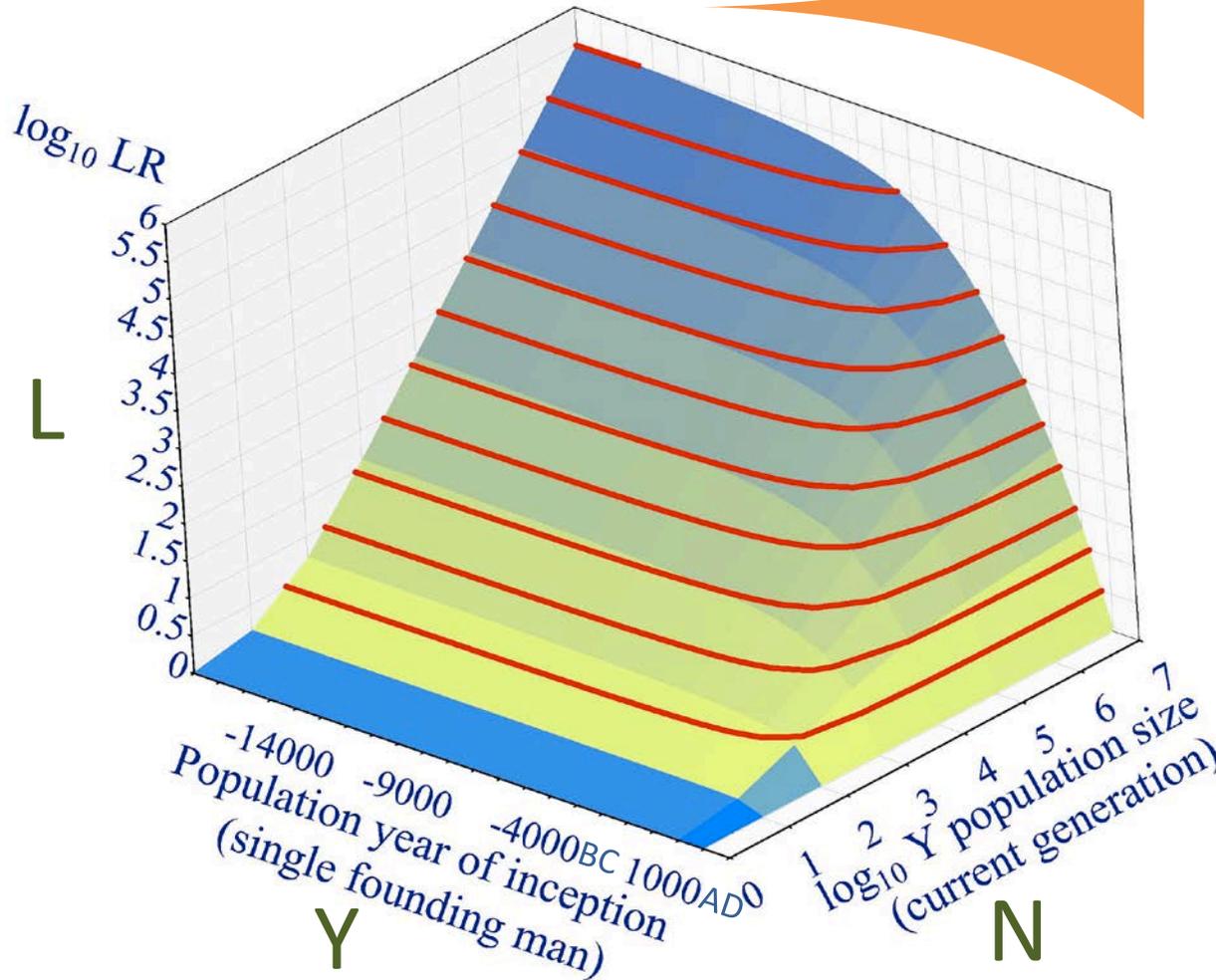
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Matching evidence

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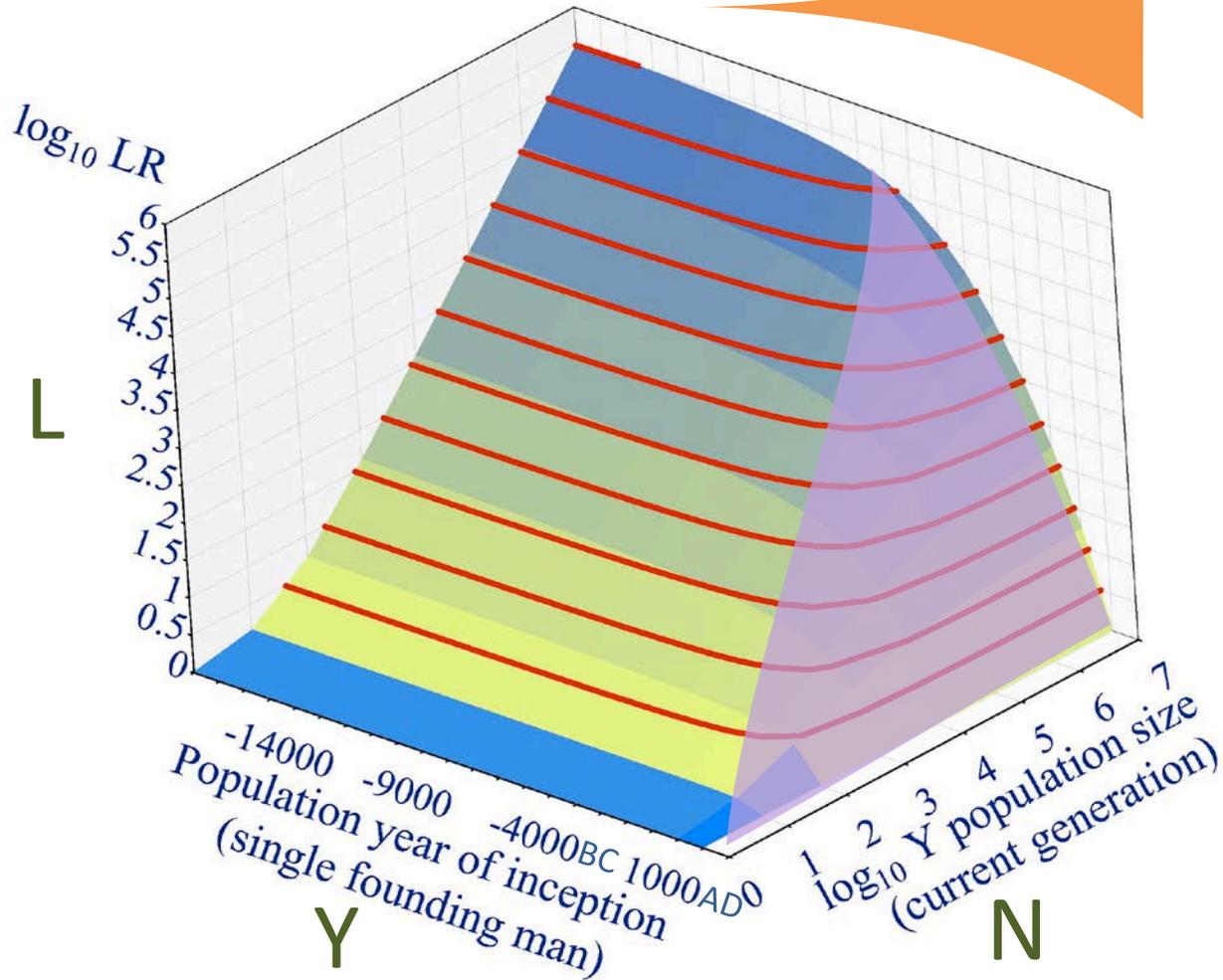
Matching evidence

- increases with population *size*.

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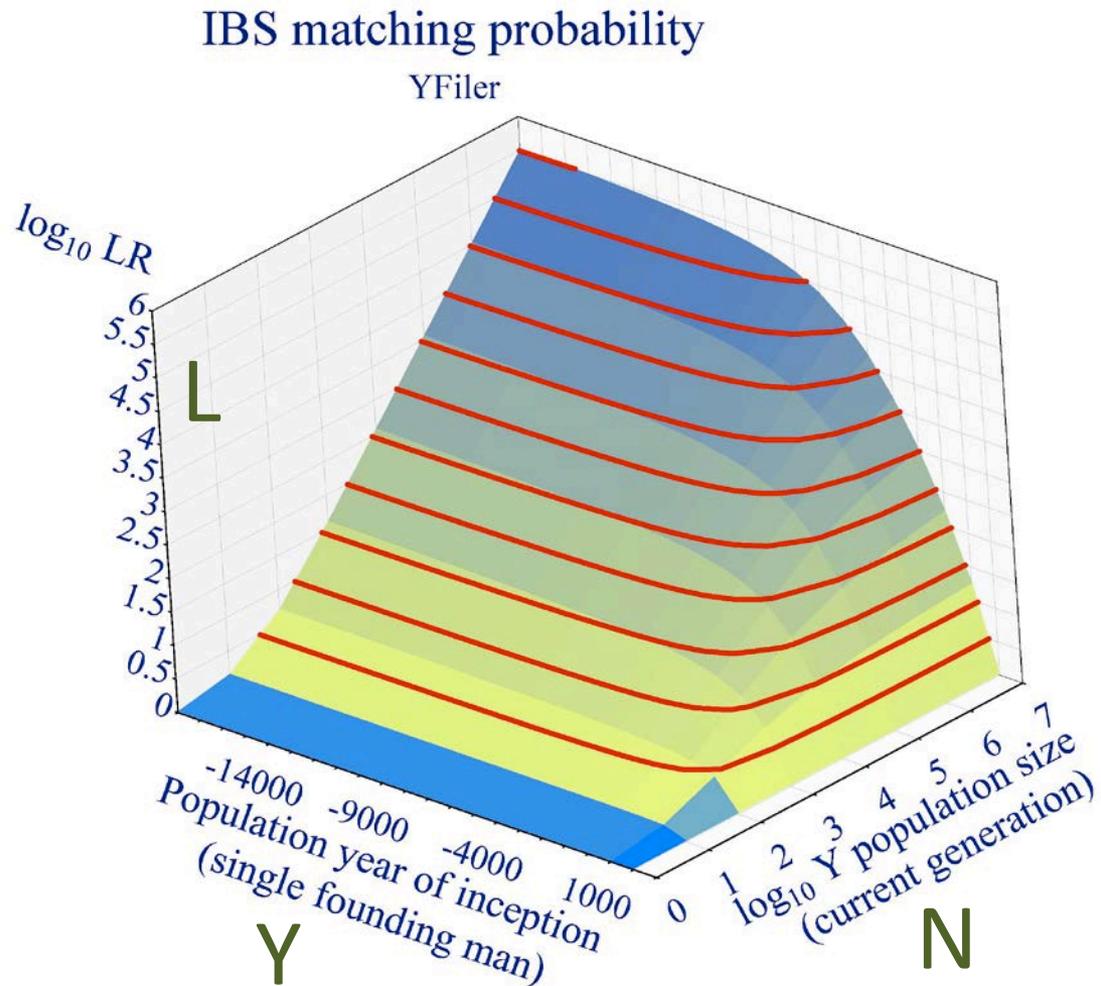


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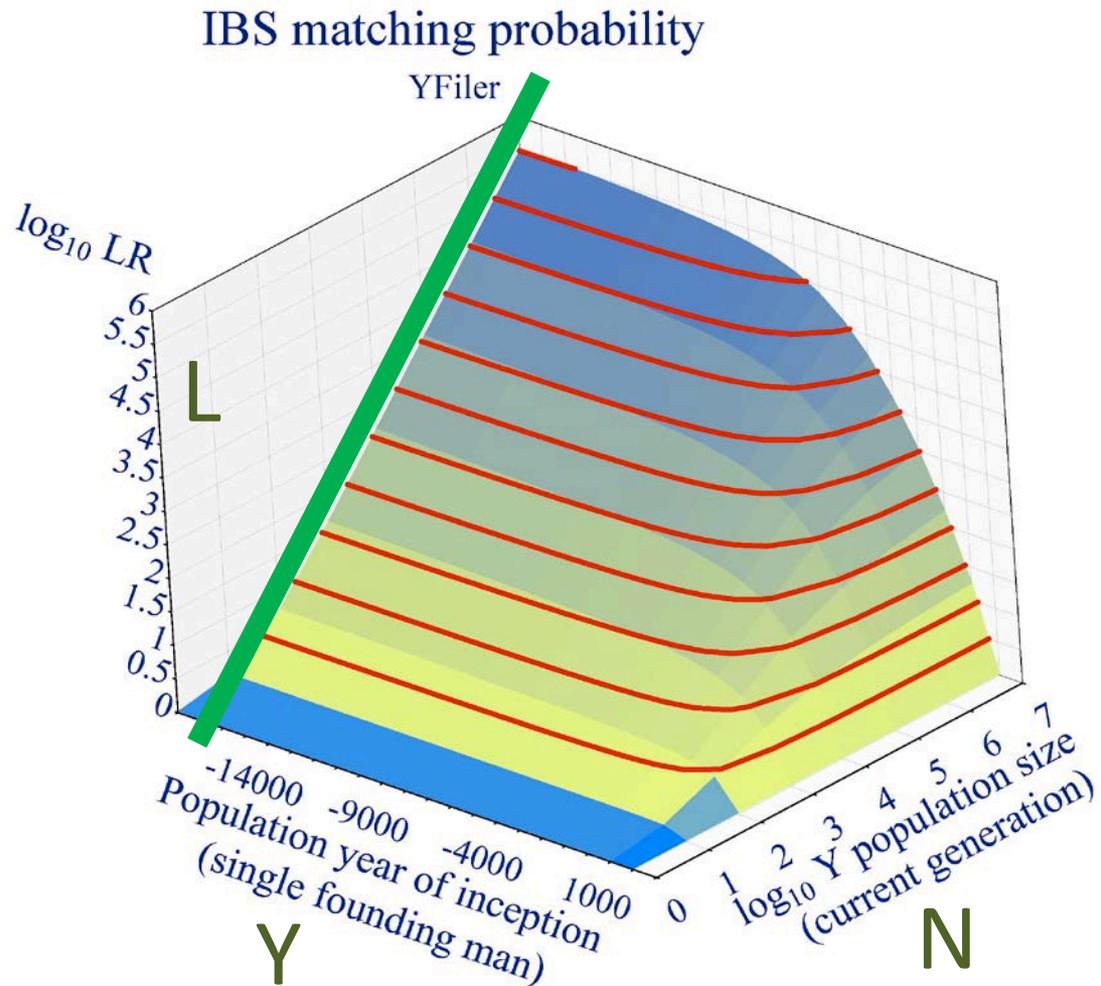
Matching evidence

- increases with population *size*.
- Population *age* is unimportant.

Haplotype cohort size vs populations size

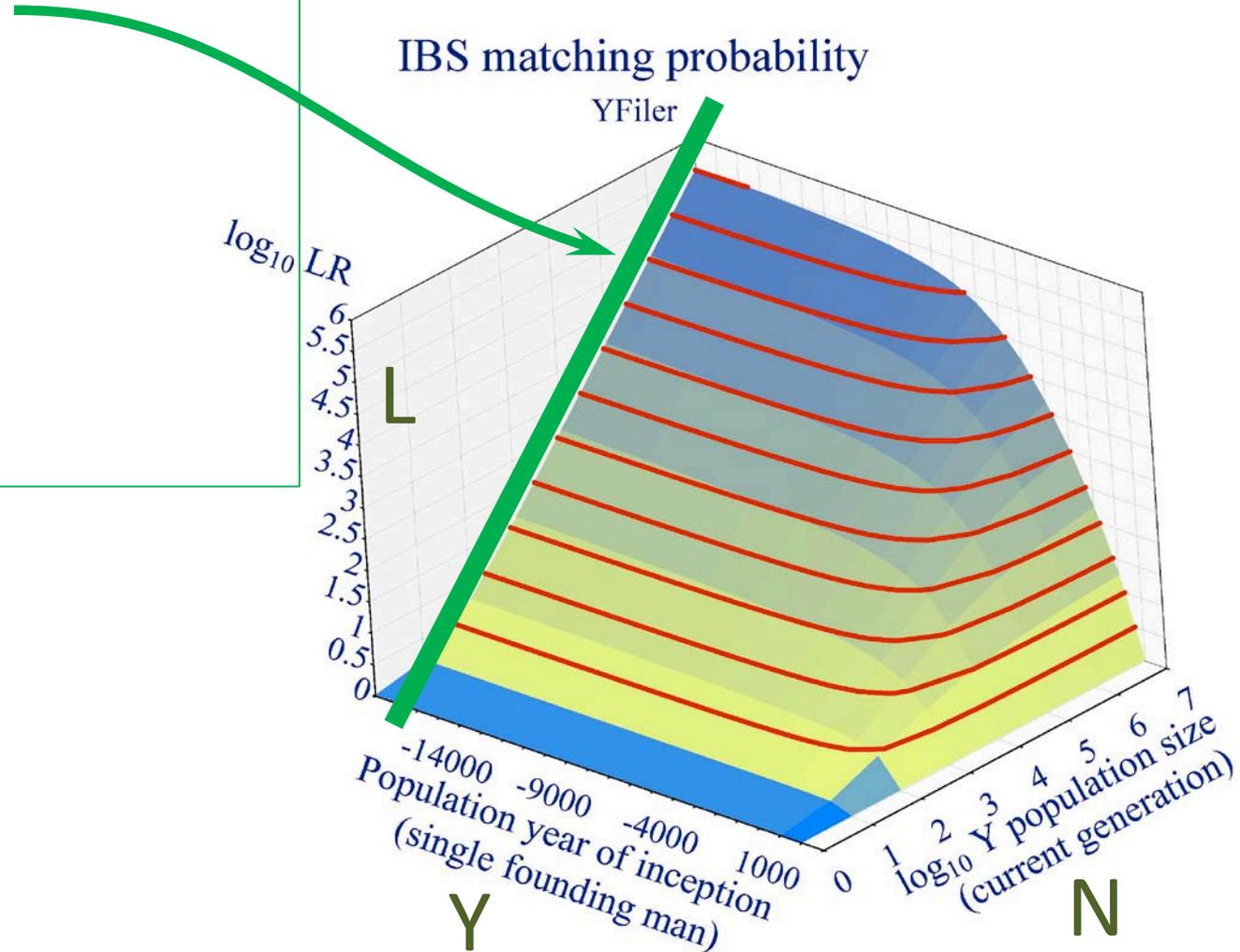


Haplotype cohort size vs populations size



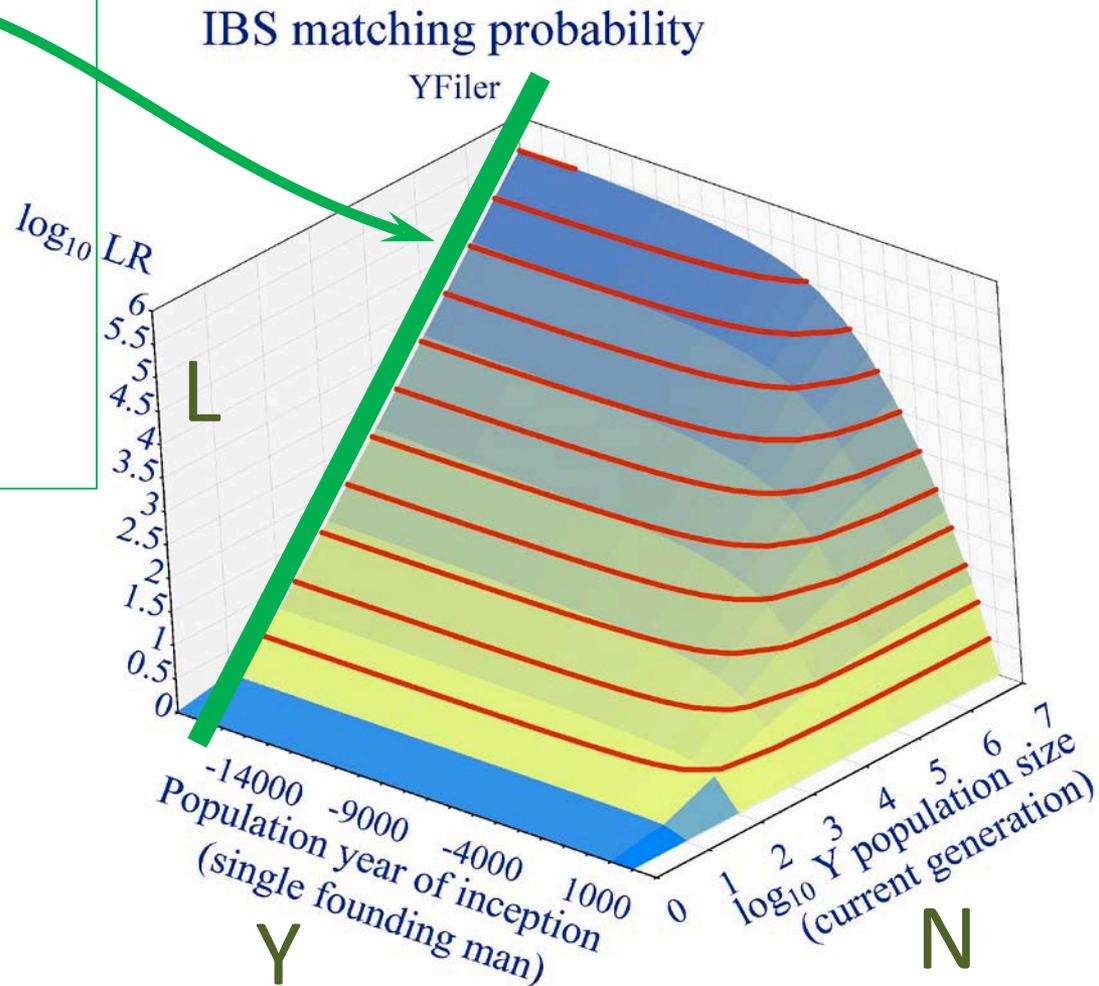
Haplotype cohort size vs populations size

- Straight? Yes



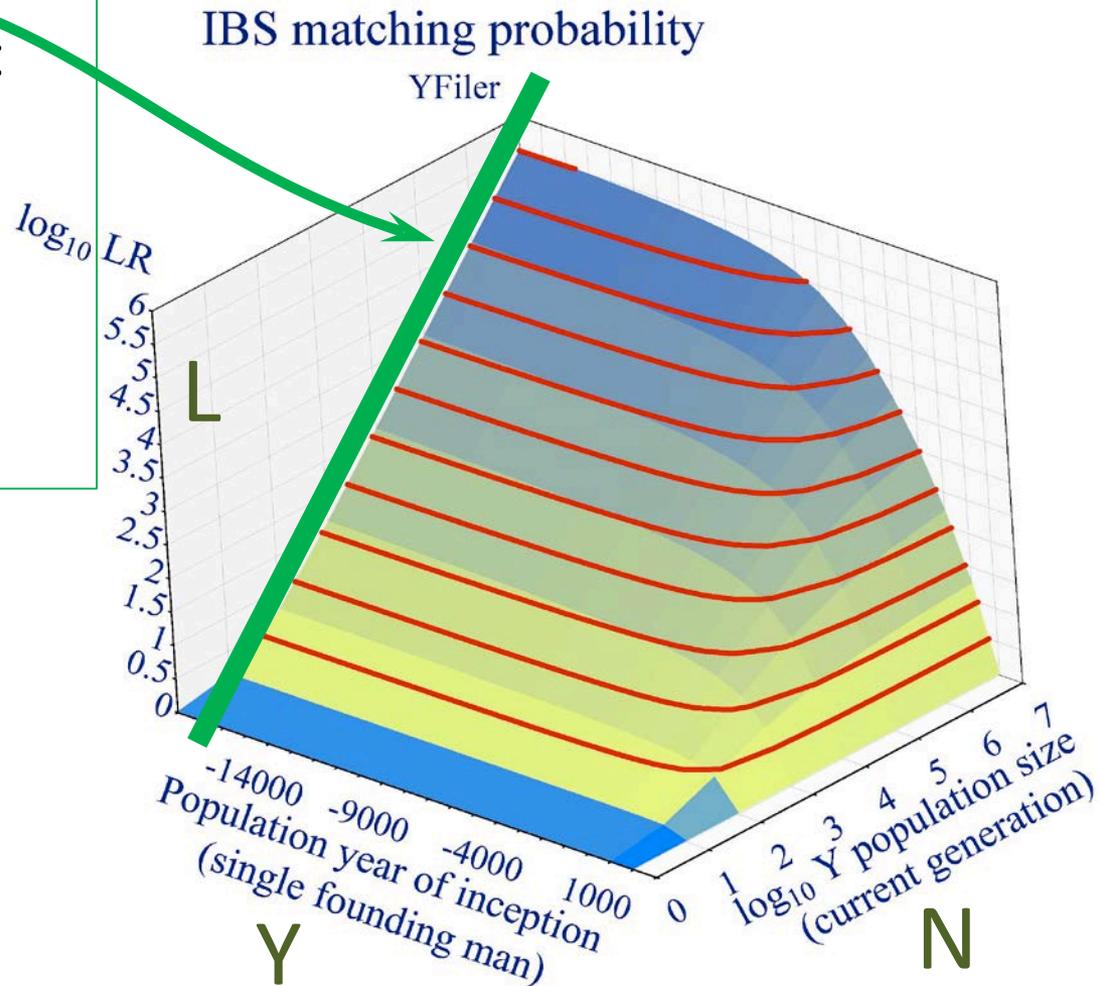
Haplotype cohort size vs populations size

- Straight? Yes
- & 45°? Close



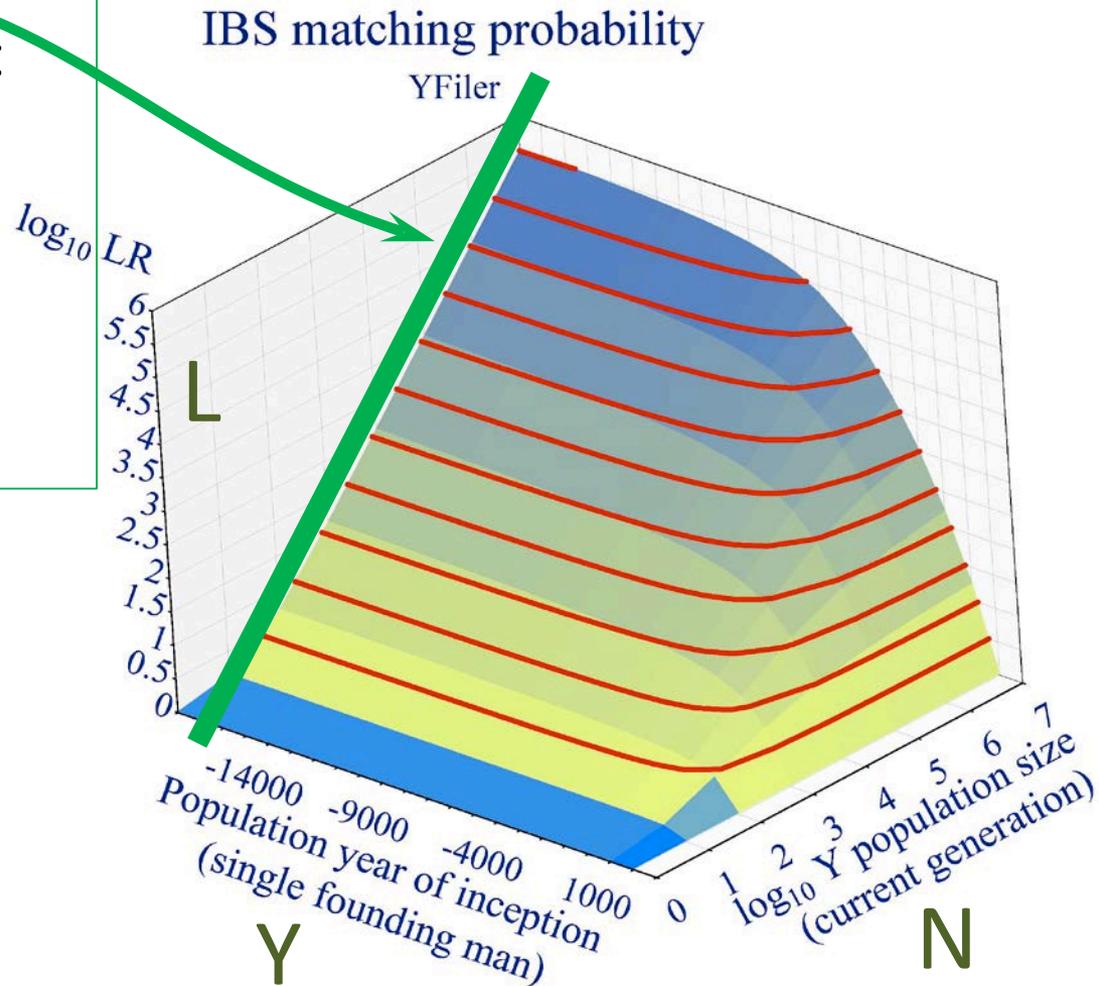
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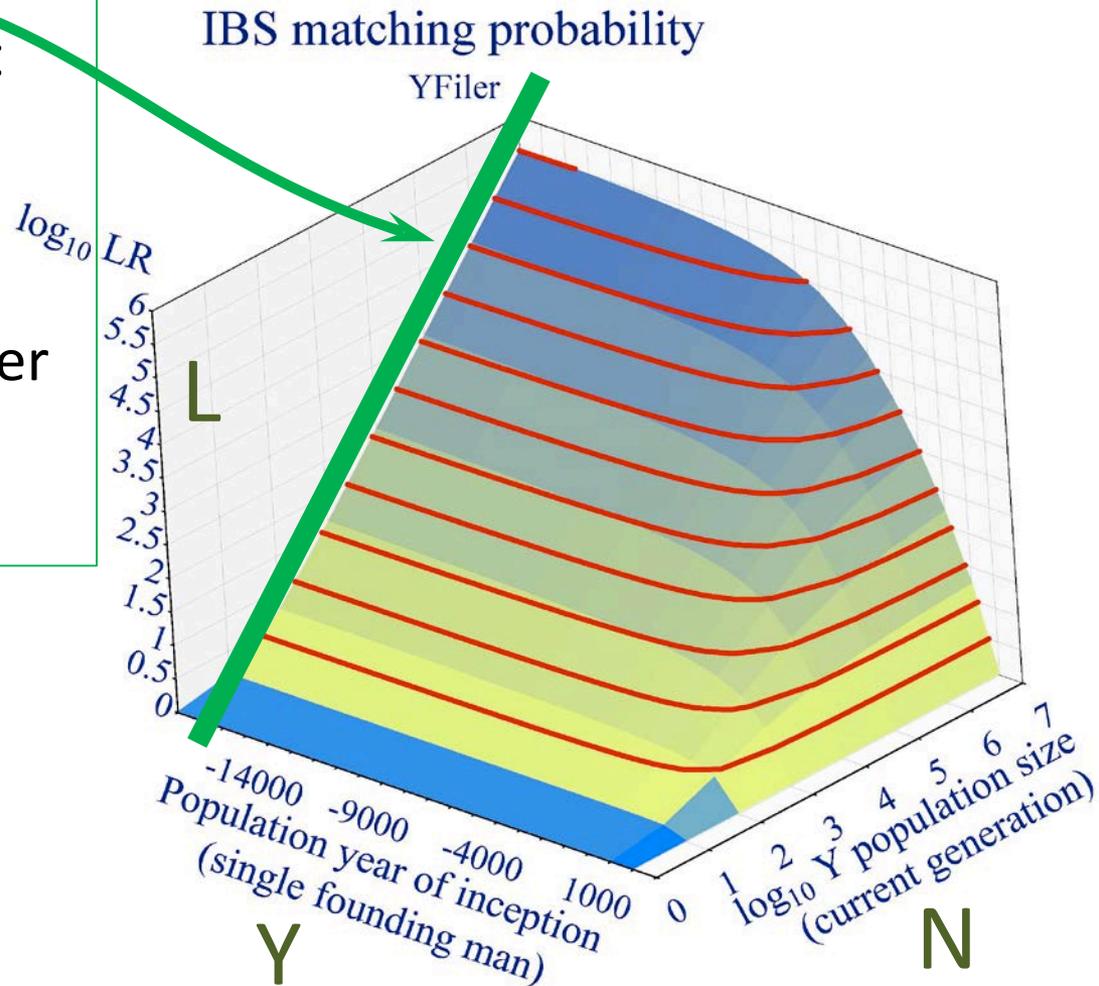
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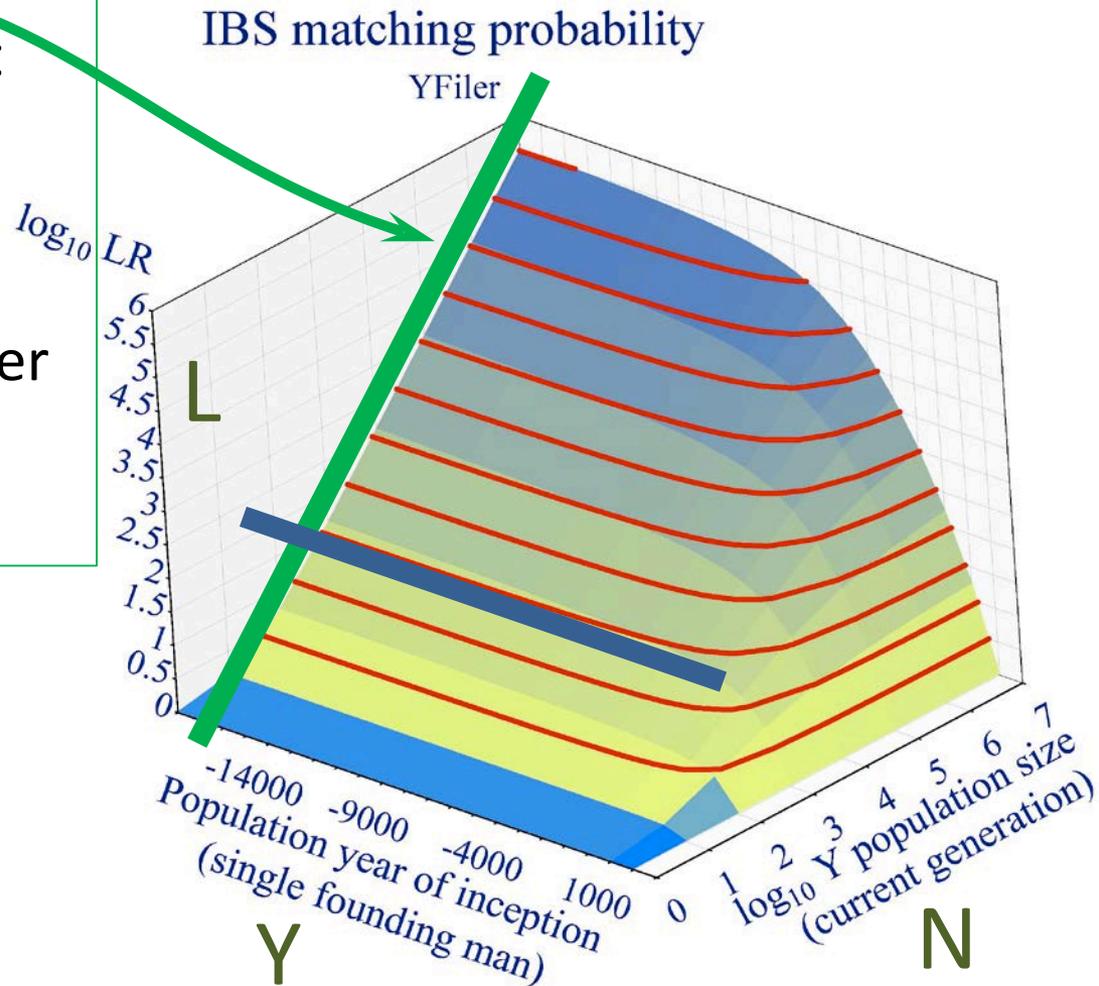
Haplotype cohort size vs populations size

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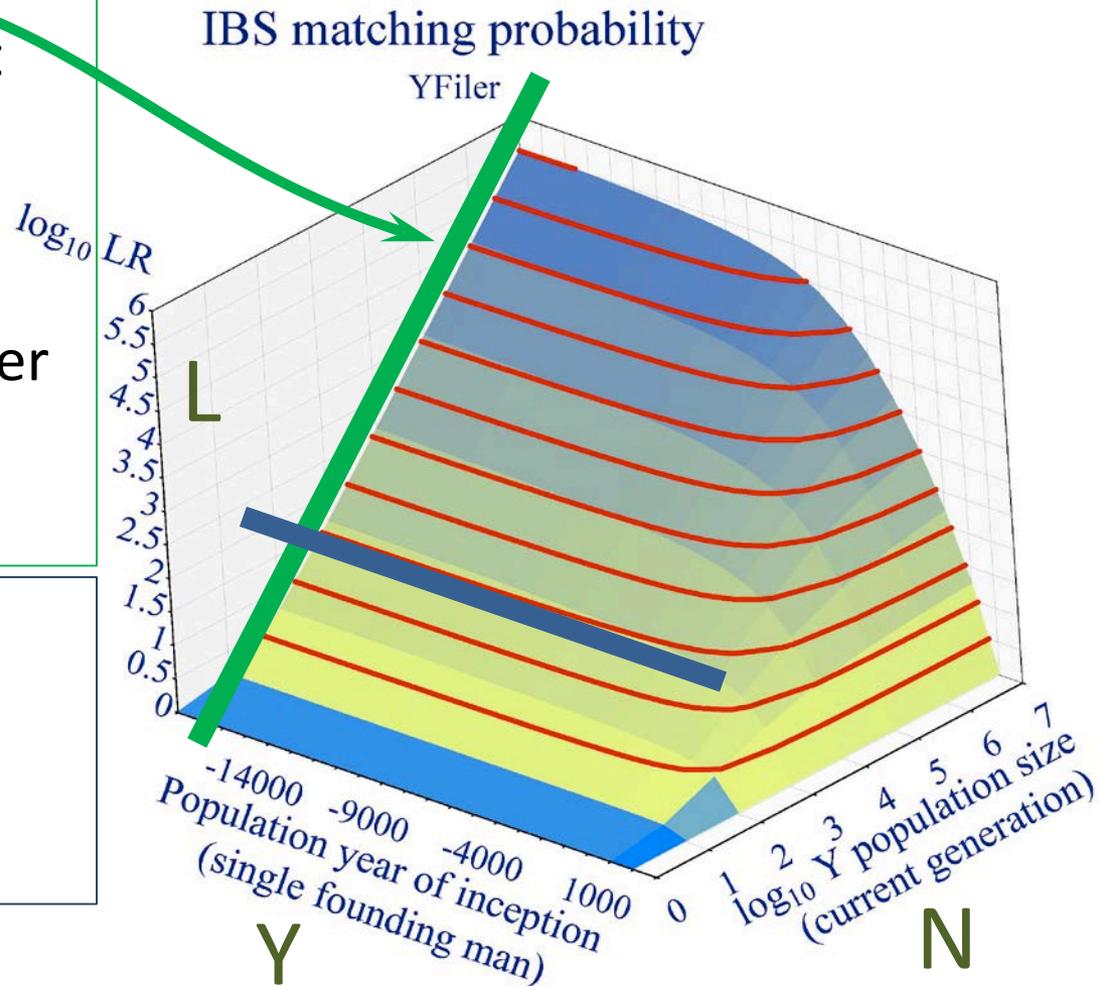
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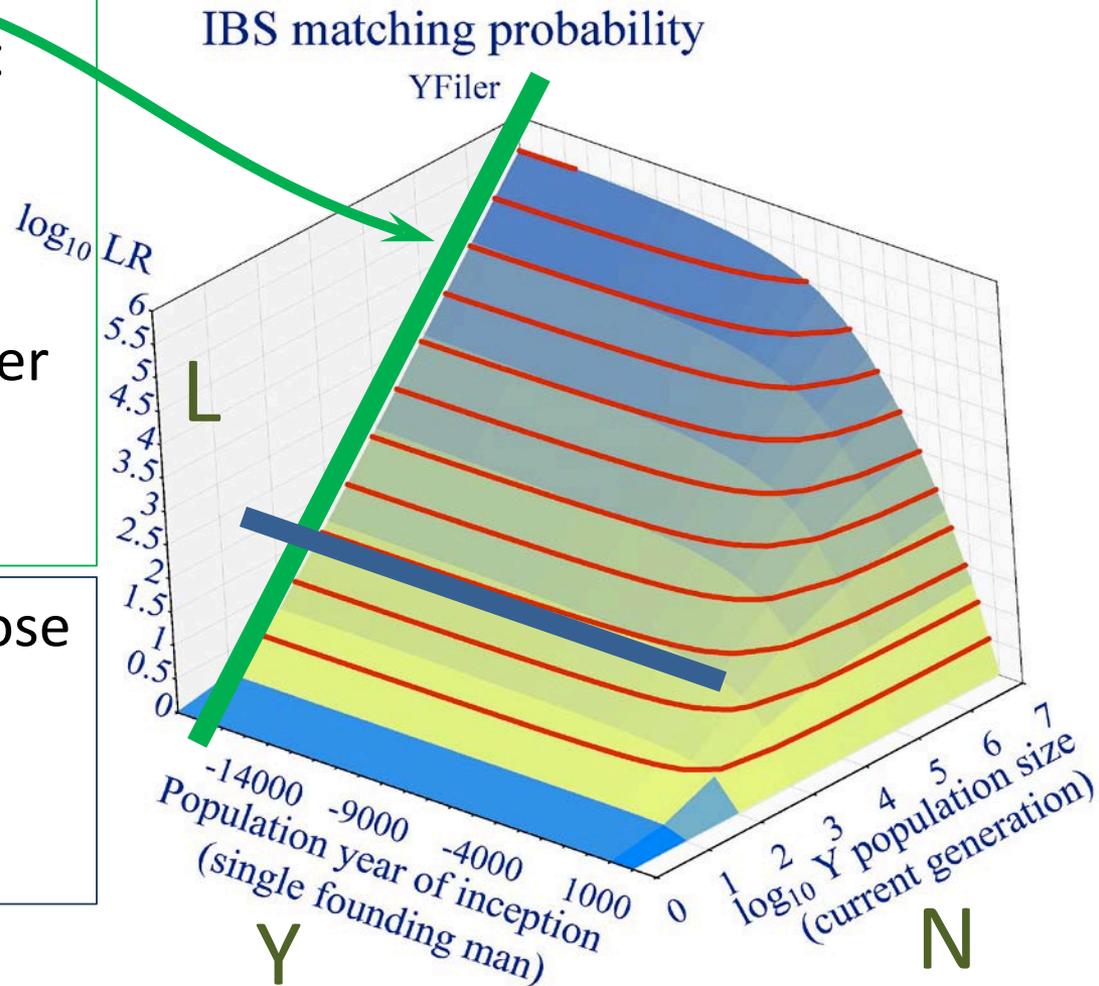
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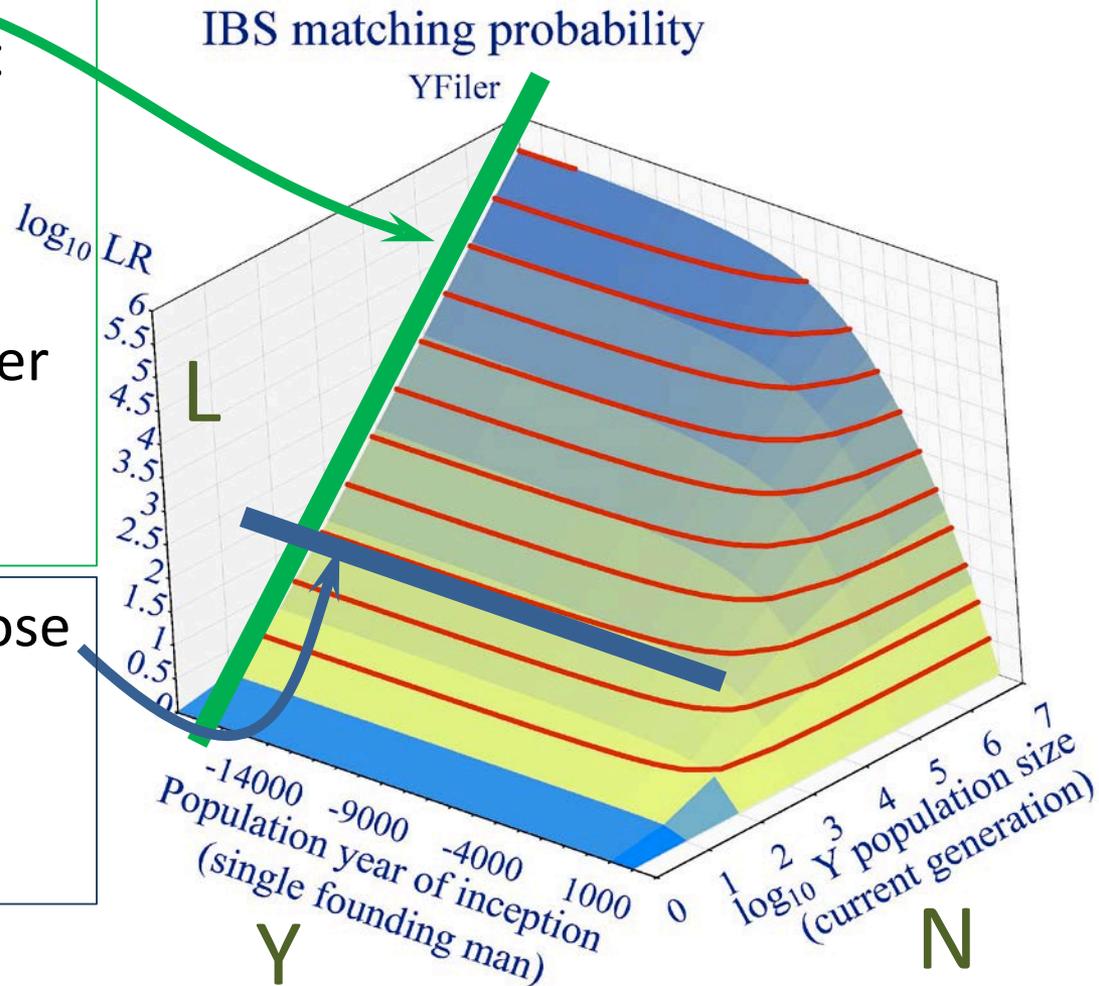
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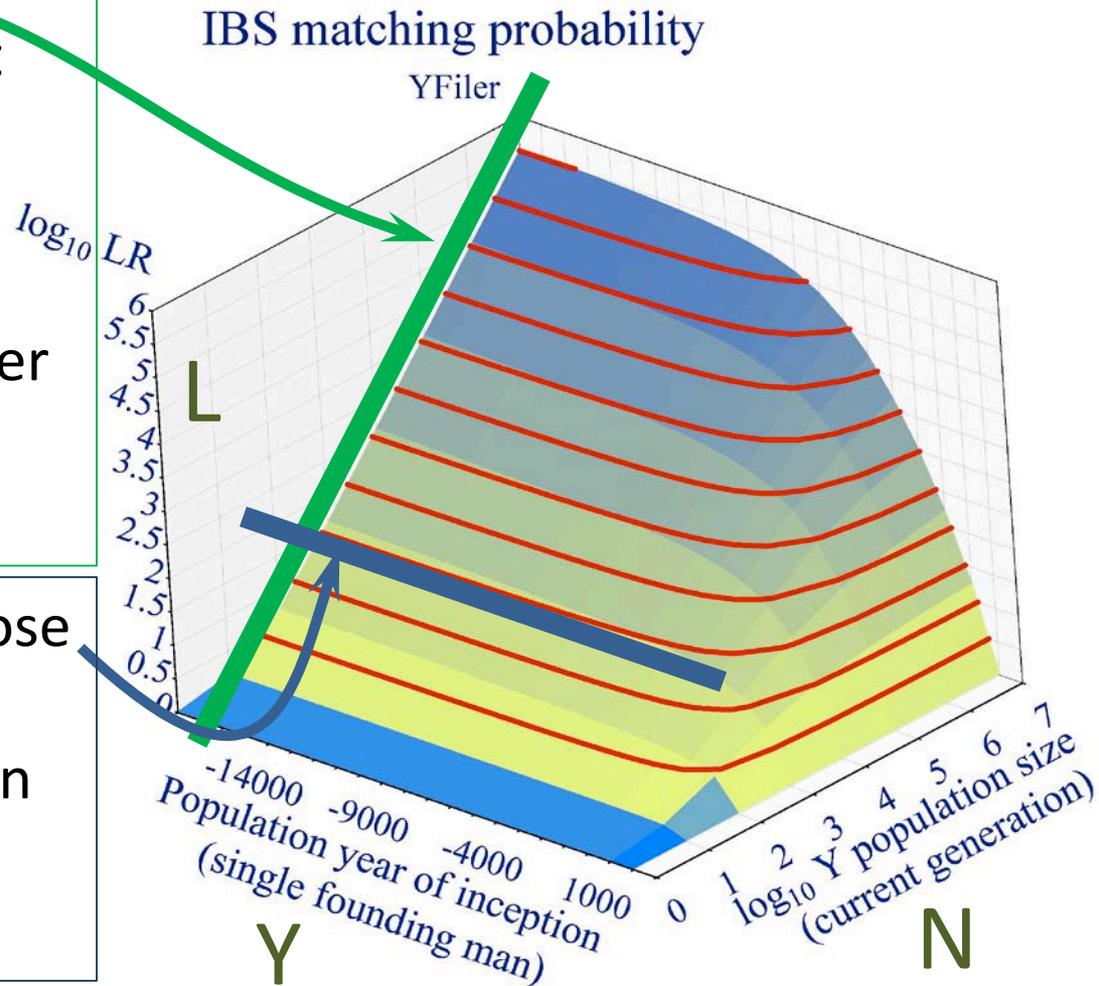
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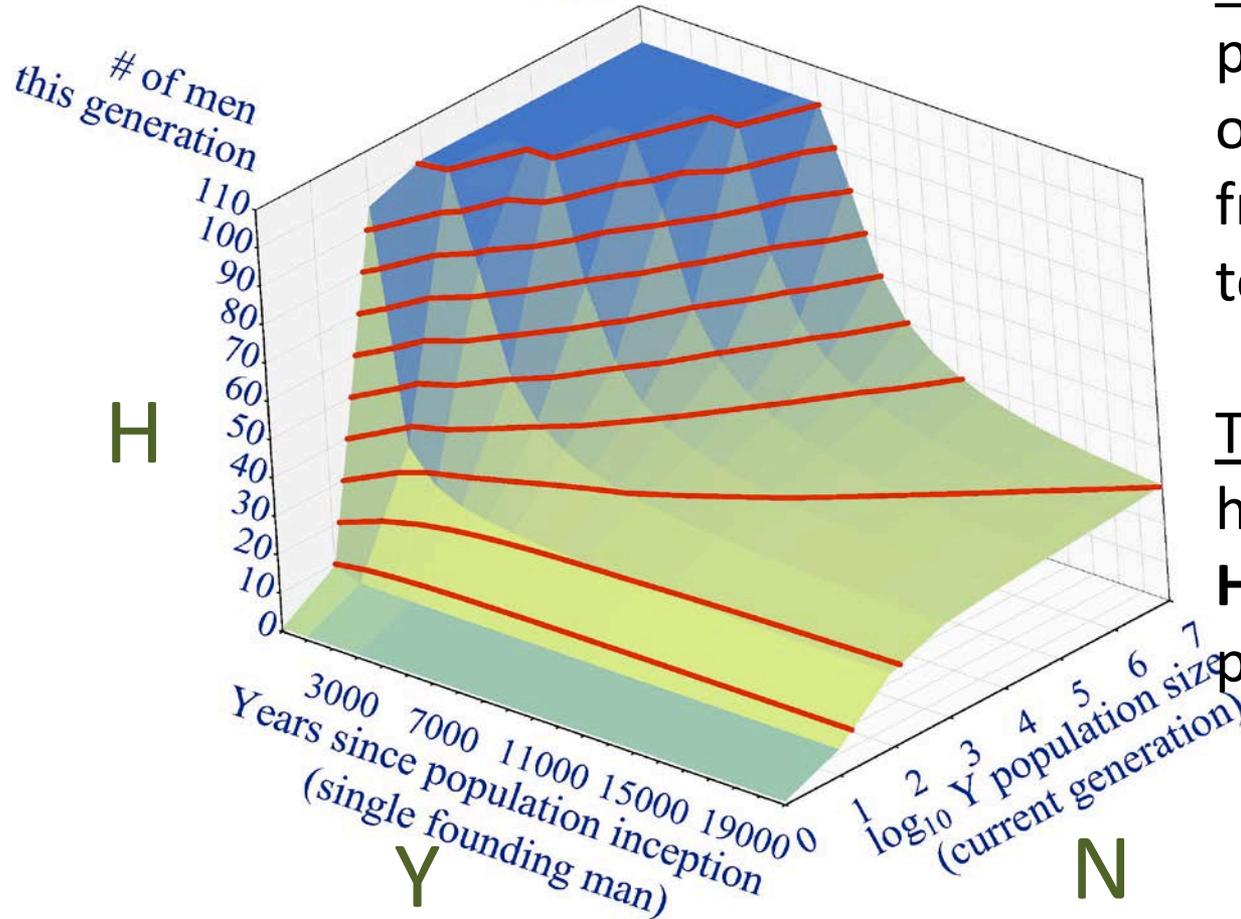
- Straight & horizontal? Close
 - Implies cohort size nearly independent of population growth rate



Number of men* with same haplotype

Average IBS cohort size

Yfiler



Assume constant population growth over Y years from 1 founding man to N men today.

Then typical Yfiler haplotype is shared by $H < 100$ men in the present generation.

* Nod to Andersen & Balding

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- *Number* of men per cohort is mostly *independent* of (unknowable, undefinable) population size.
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 - More work is possible.
- Current forensic practice is thoughtless



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The end

This work received no support from the NIJ, IMF, World Bank, Bill and Melinda Gates, or the Ford Foundation. Even Queen Isabella of Spain (usually a soft touch) wouldn't pitch in.

Ancestry dominates Y matching

Autosomal STR allele A

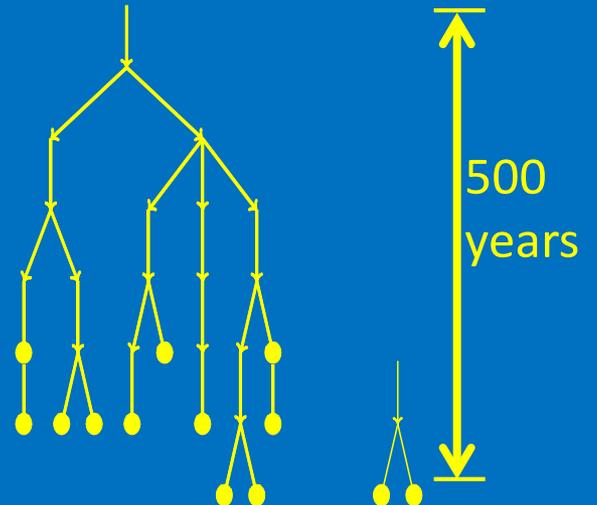
Y haplotype T

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Autosomal STR allele A

Y haplotype T

- One dominant T family



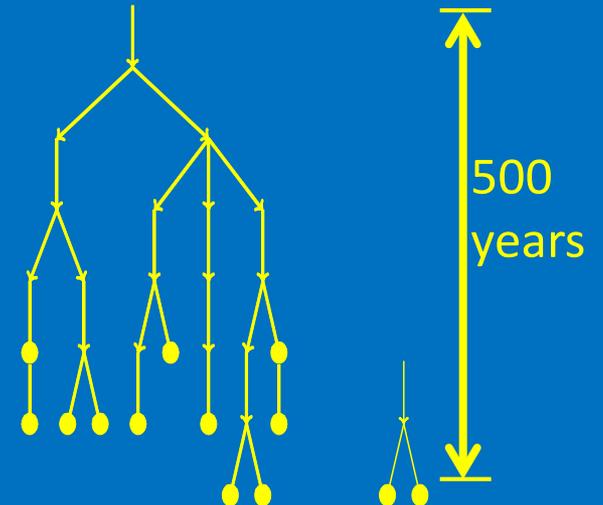
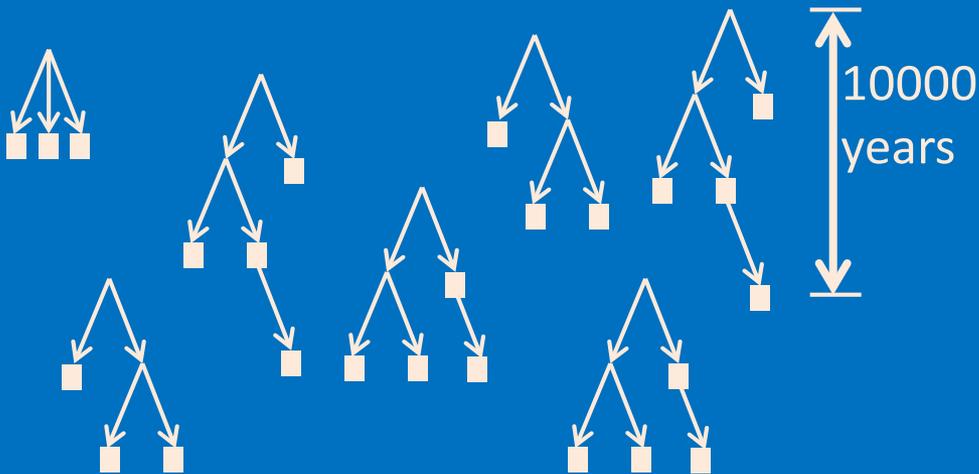
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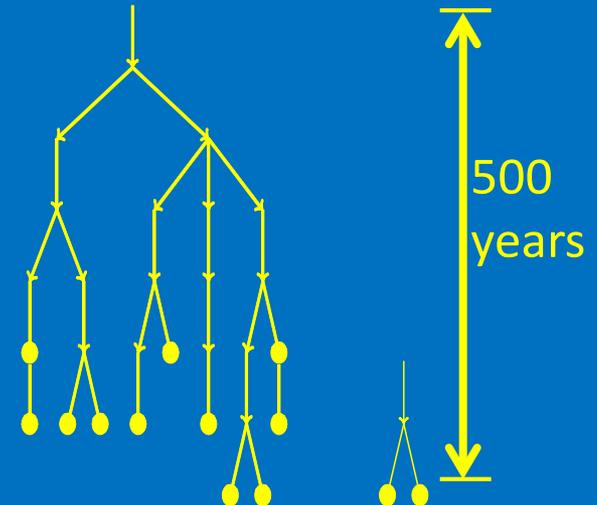
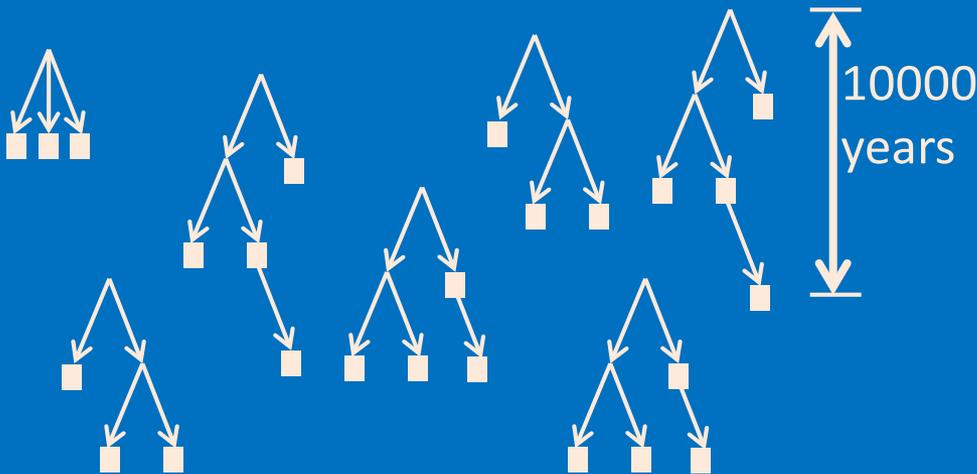
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Autosomal STR allele A

- Many A families
- 5% of matching is family

Y haplotype T

- One dominant T family
- 90% of matching is family



Ancestry dominates Y matching

Autosomal STR allele A

- Many A families
- 5% of matching is family
- Convergent mutation common

Y haplotype T

- One dominant T family
- 90% of matching is family
- Convergent mutation insignificant

