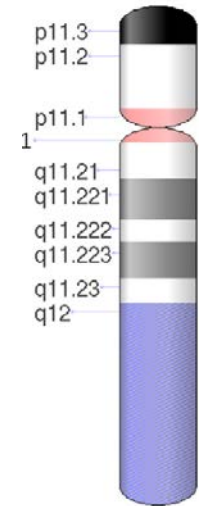


Simplicity can be confusing (understanding the manly chromosome partly through APL)



Y chromosome

Charles Brenner, Ph.D.

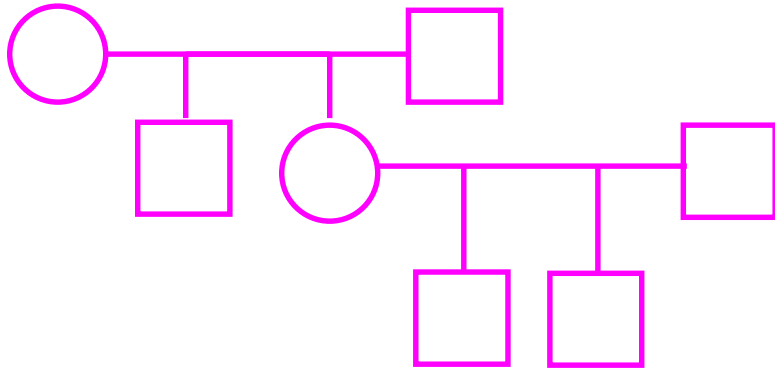
Purveyor of forensic mathematics,

DNA·VIEW®

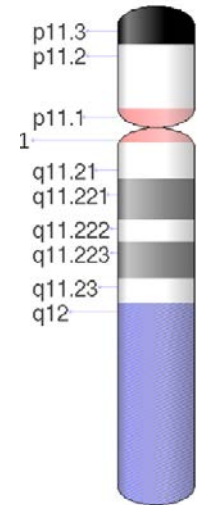
charles@dna-view.com

<http://dna-view.com>

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



Autosomal pedigree



Y chromosome

Charles Brenner, Ph.D.

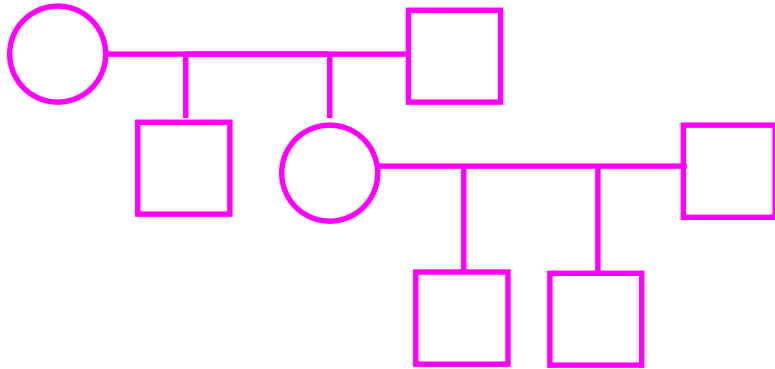
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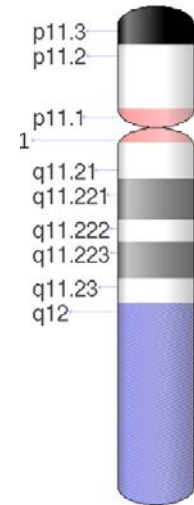
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Autosomal pedigree



Y pedigree



Y chromosome

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<http://dna-view.com>

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

Fighting for truth and/or justice

- I published some papers on Y DNA evidence

Fighting for truth and/or justice

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- FBI method for Y DNA is malpractice in tribal context

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Fighting for truth and/or justice

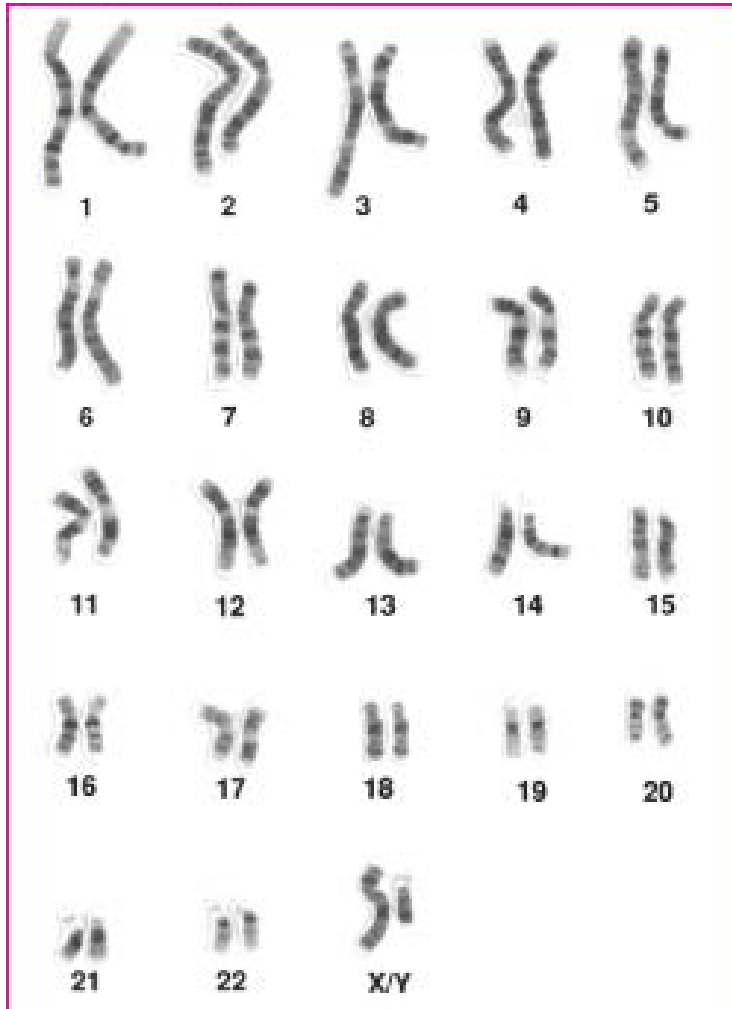
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Navajo 🙄 in New Mexico

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Hopi ★ & also Tohono O'odham ★ tribesmen / reservation in Arizona
Navajo 🙄 in New Mexico
Ashkenazi (!) in Boston

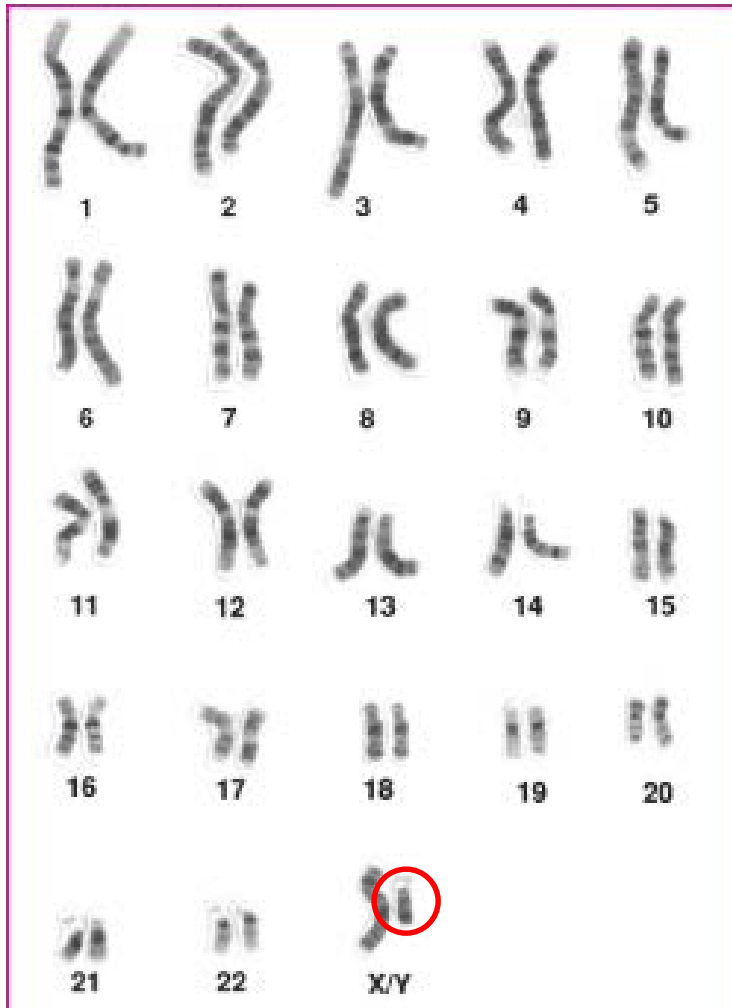
What's Y?

Karyogram of a human male



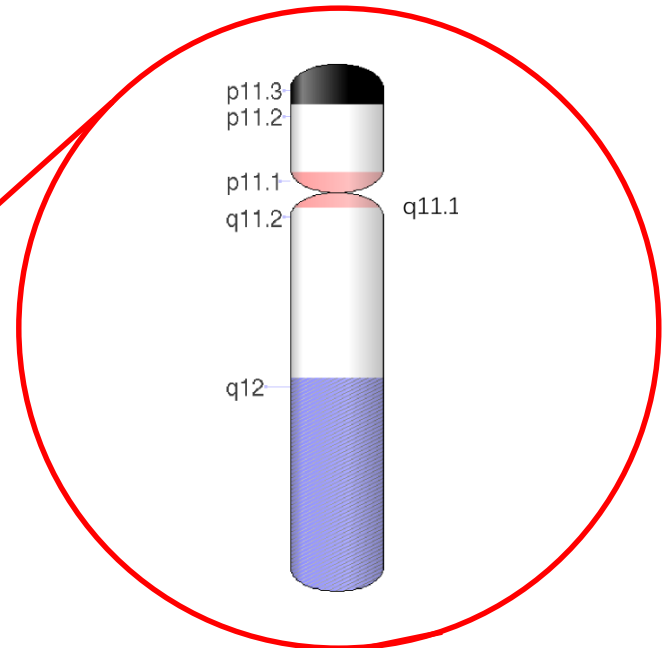
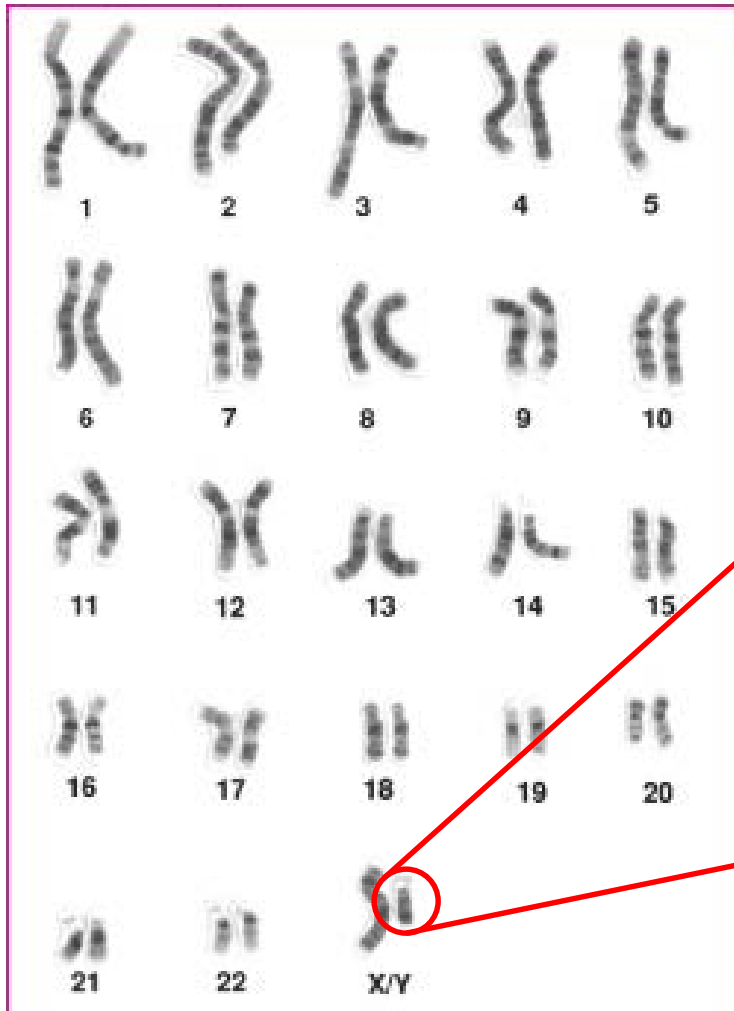
What's Y?

Karyogram of a human male



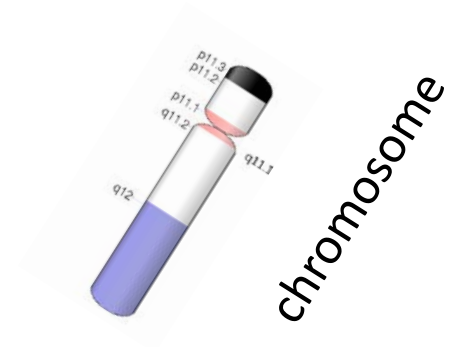
What's Y?

Karyogram of a human male

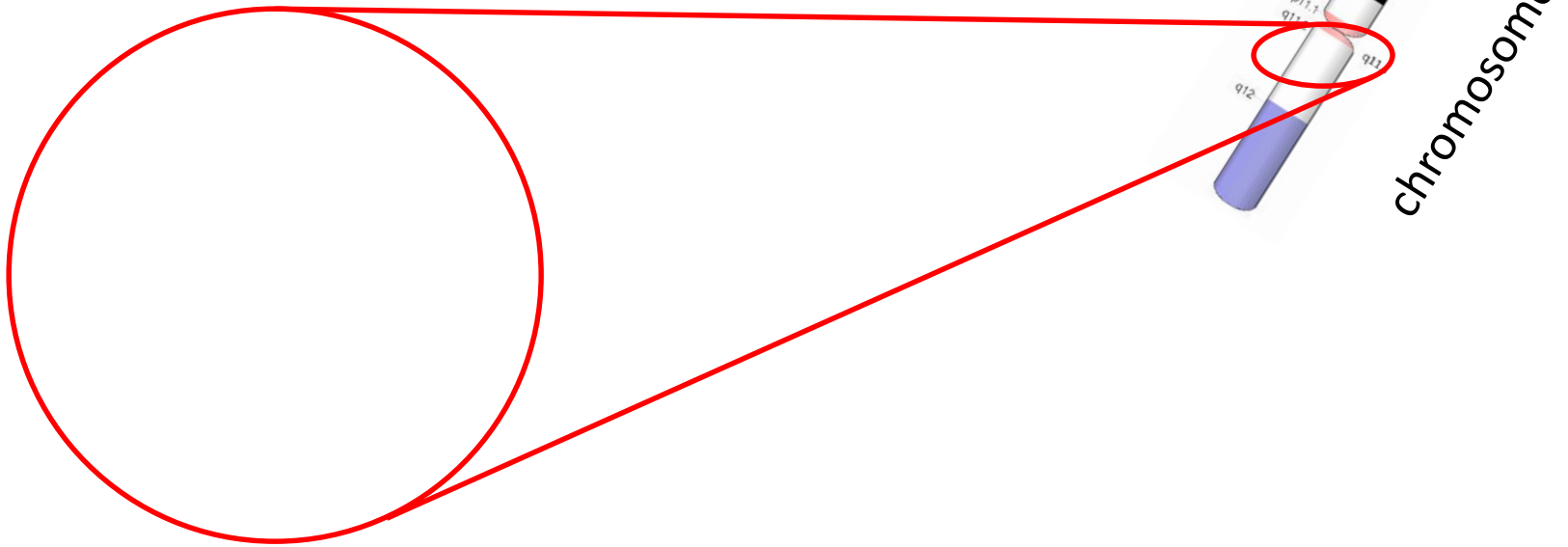


Y chromosome

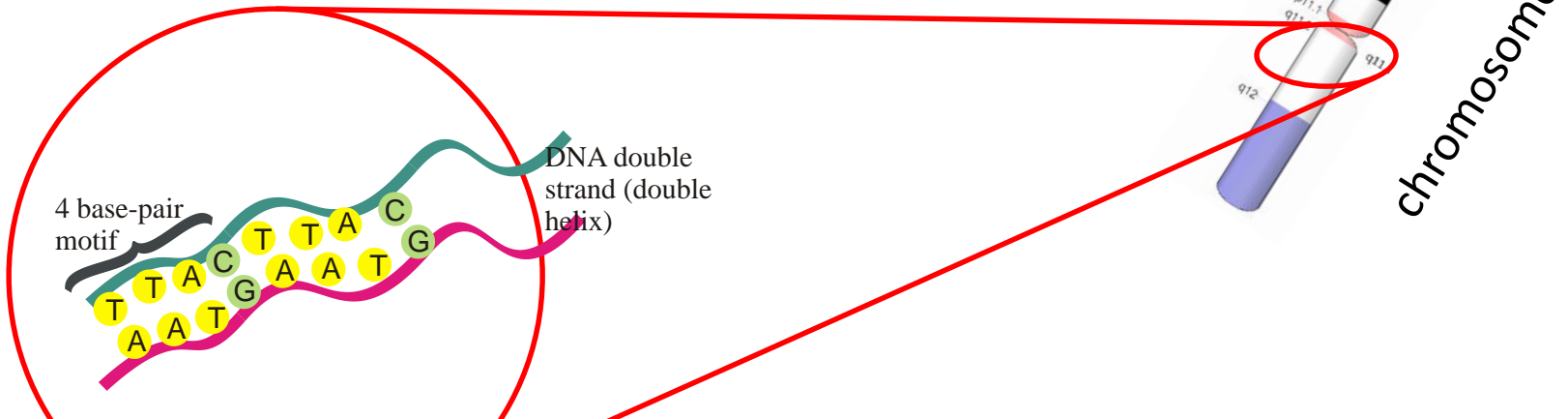
Typical identification locus within a chromosome



Typical identification locus within a chromosome



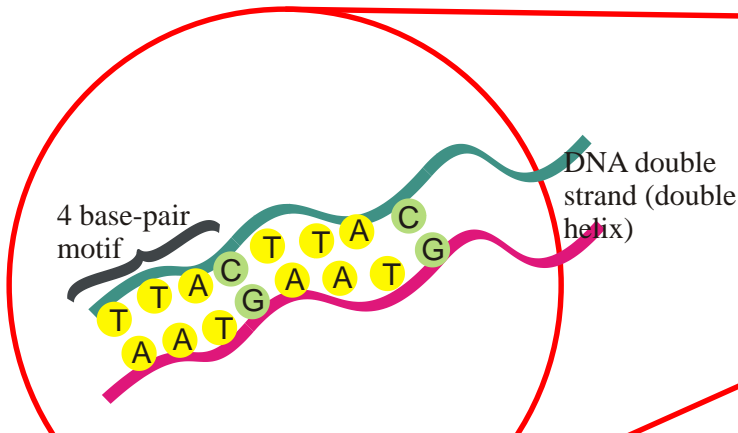
Typical identification locus within a chromosome



Electron microphotograph
showing 2 repeats of the motif.

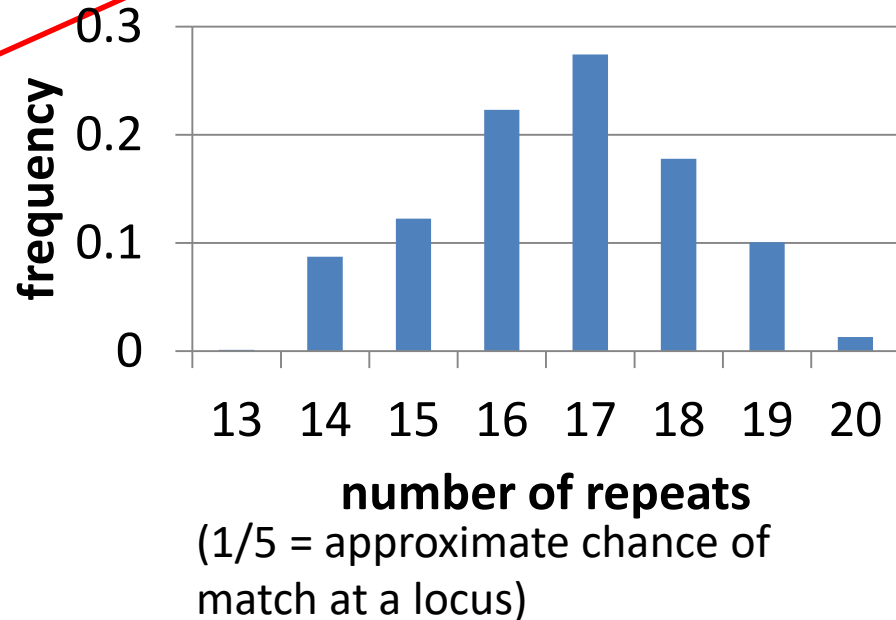
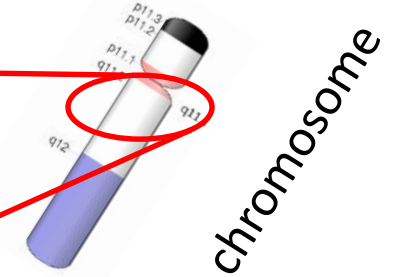
Number of repeats **varies** between people thanks to occasional replication slippage mutations over the eons.

Typical identification locus within a chromosome

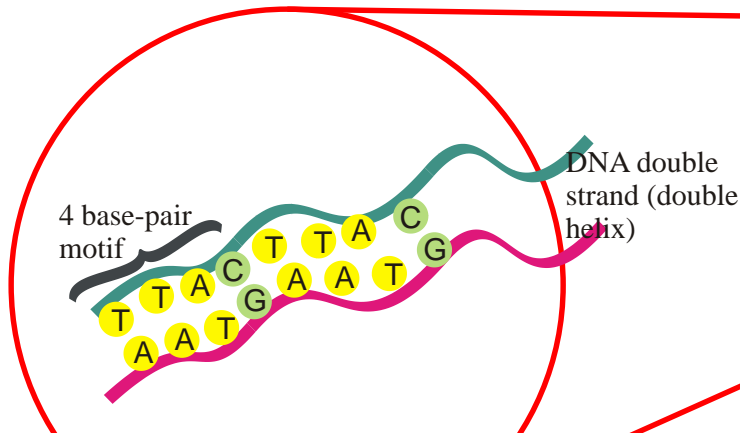


Electron microphotograph showing 2 repeats of the motif.

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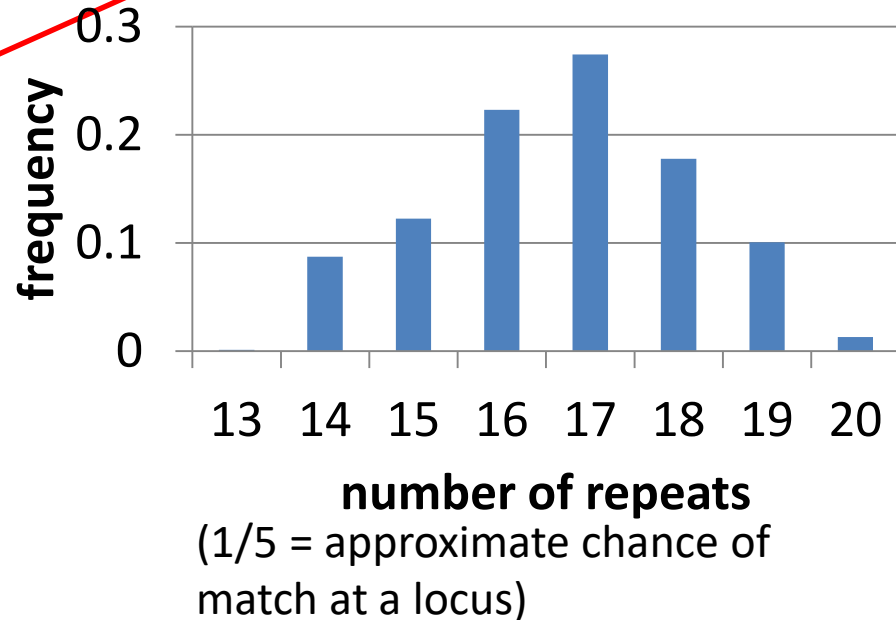
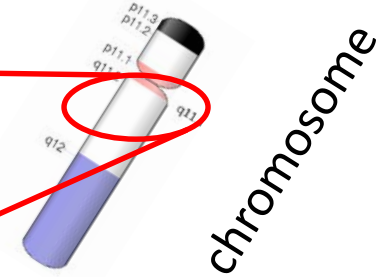


Typical identification locus within a chromosome



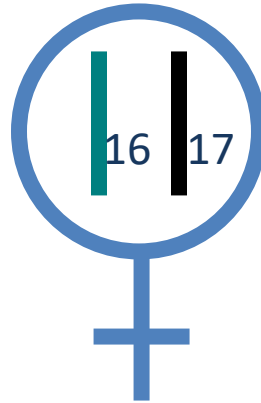
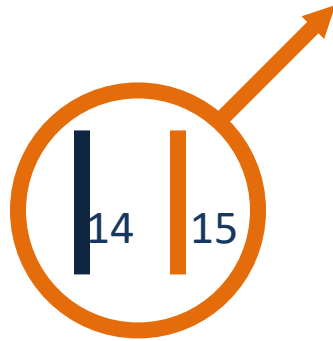
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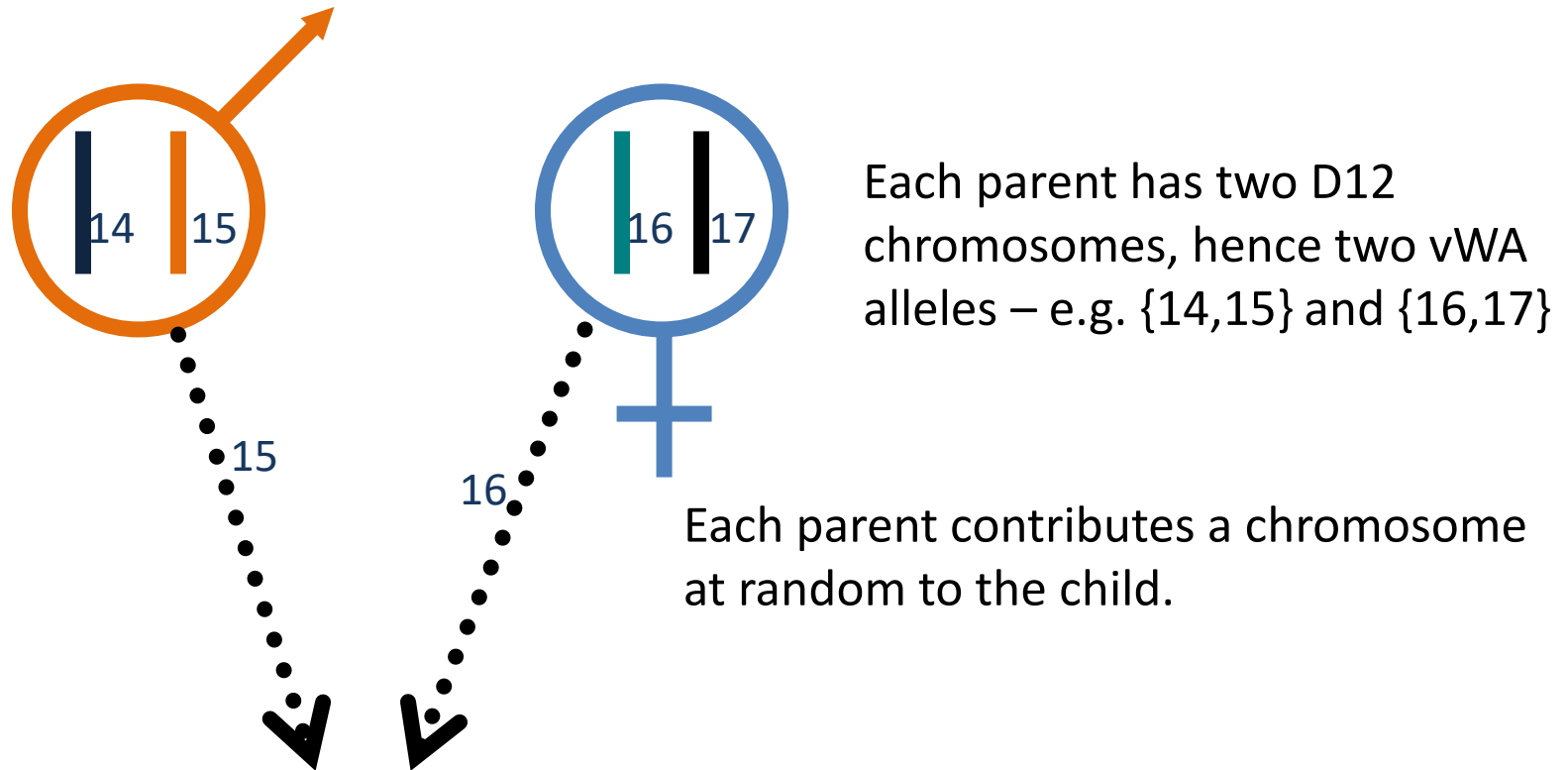
Forensic evidence: Suspect **allele** at locus matches crime scene allele
Evidential value: 5x more likely if suspect is the donor, than if not.
(NB: The cumulative evidence from 10 to 30 alleles can be very strong.)

Genetic inheritance (sexual), one locus

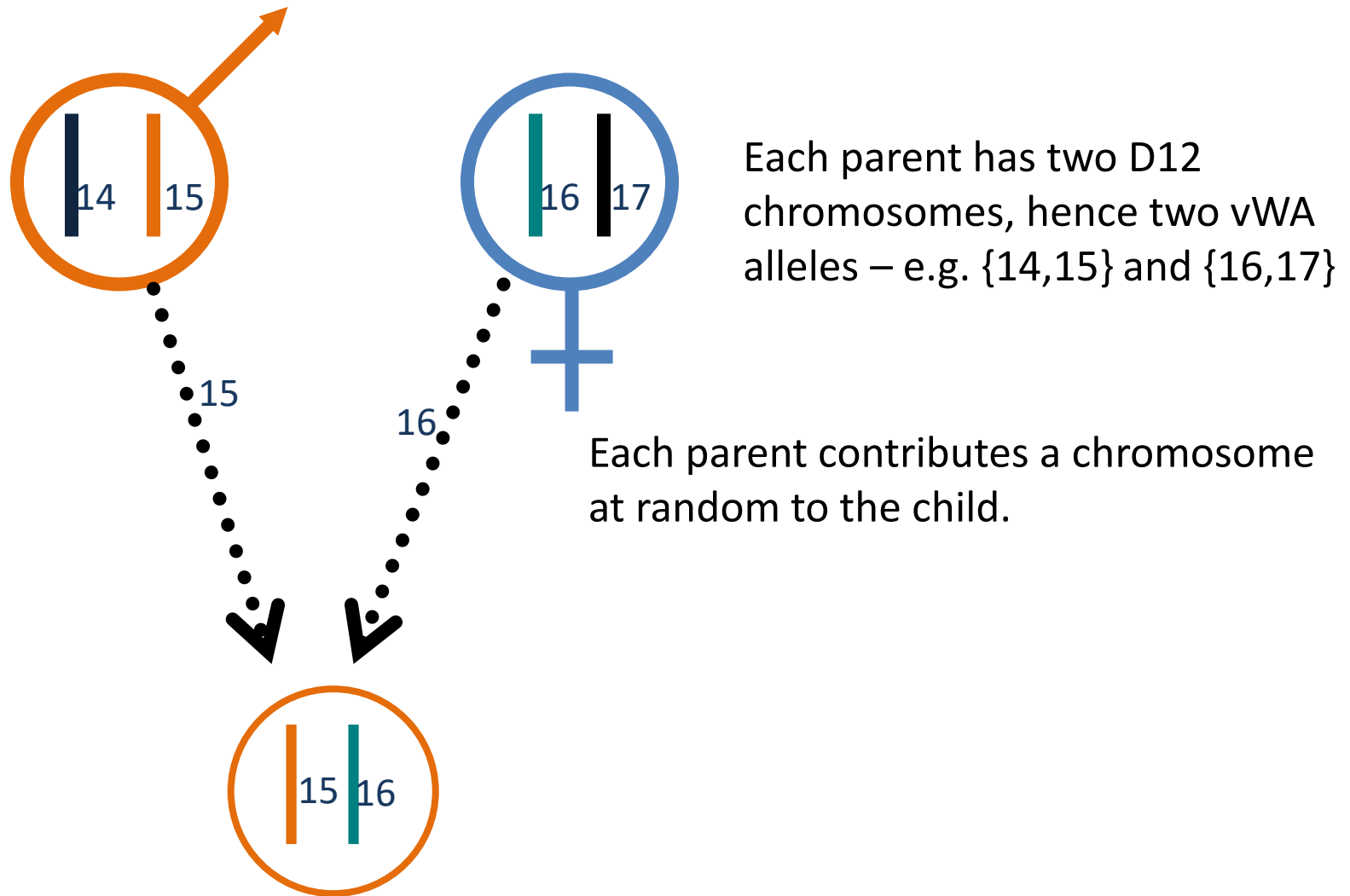


Each parent has two D12 chromosomes, hence two vWA alleles – e.g. {14,15} and {16,17}

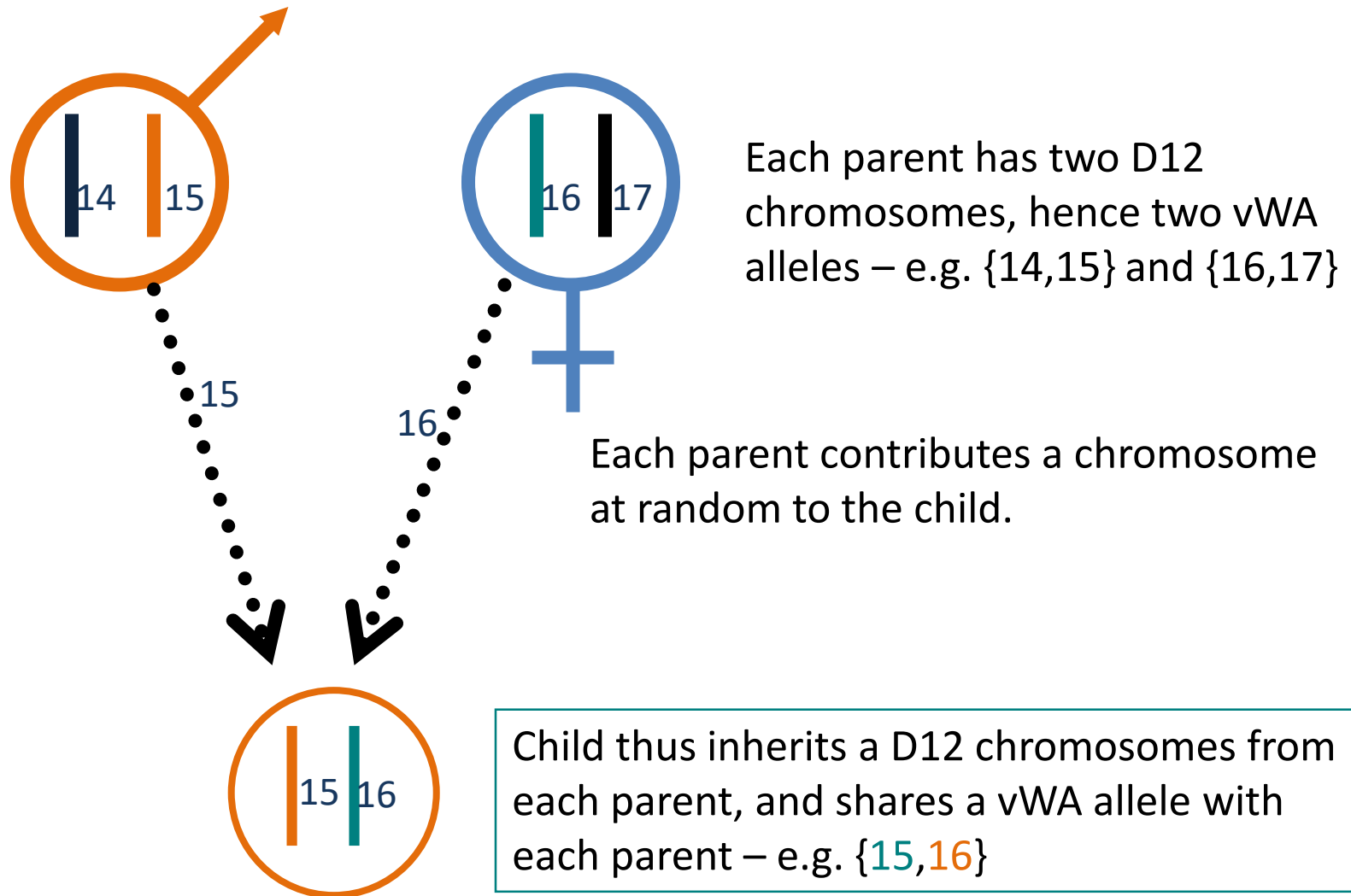
Genetic inheritance (sexual), one locus



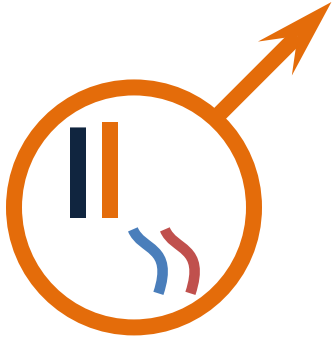
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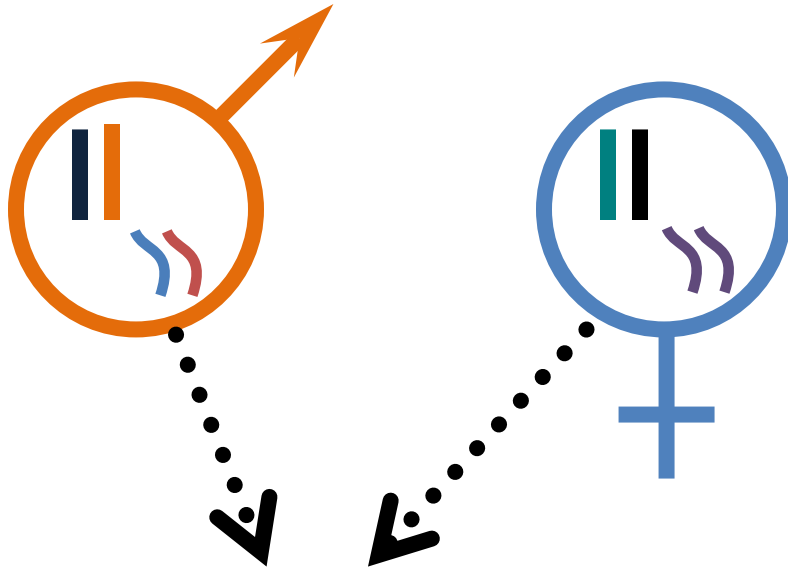


Genetic inheritance (sexual), multiple loci



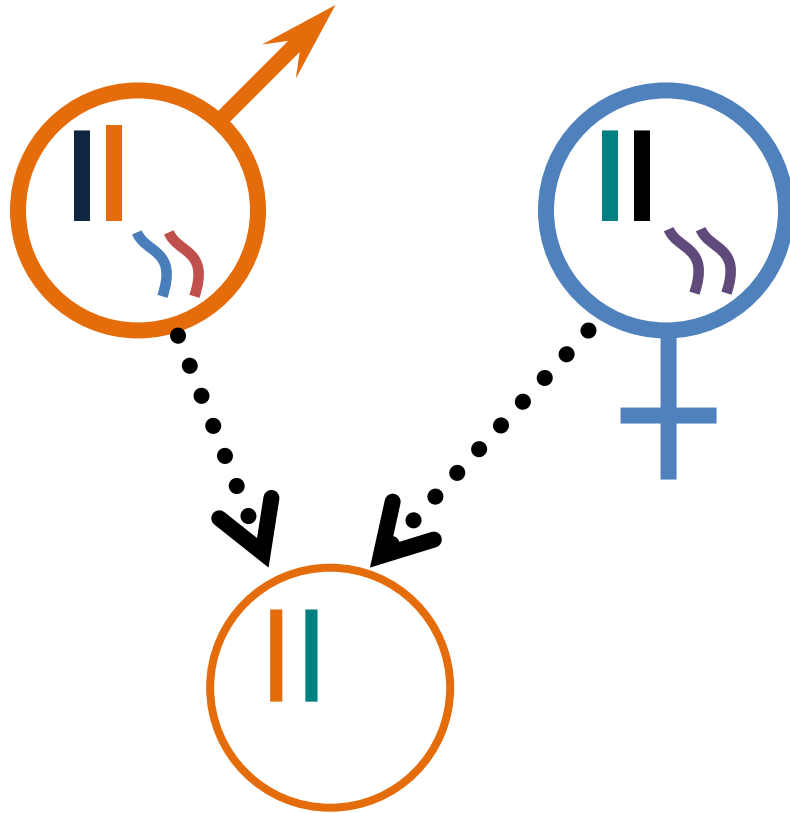
Each parent contributes one chromosome at random from each pair **independently**.

Genetic inheritance (sexual), multiple loci



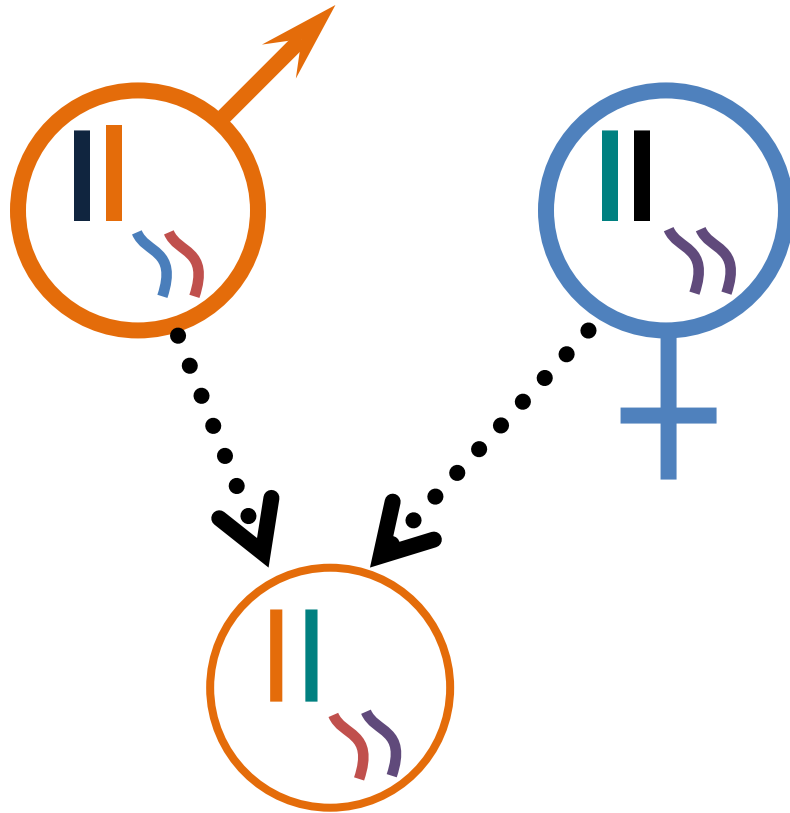
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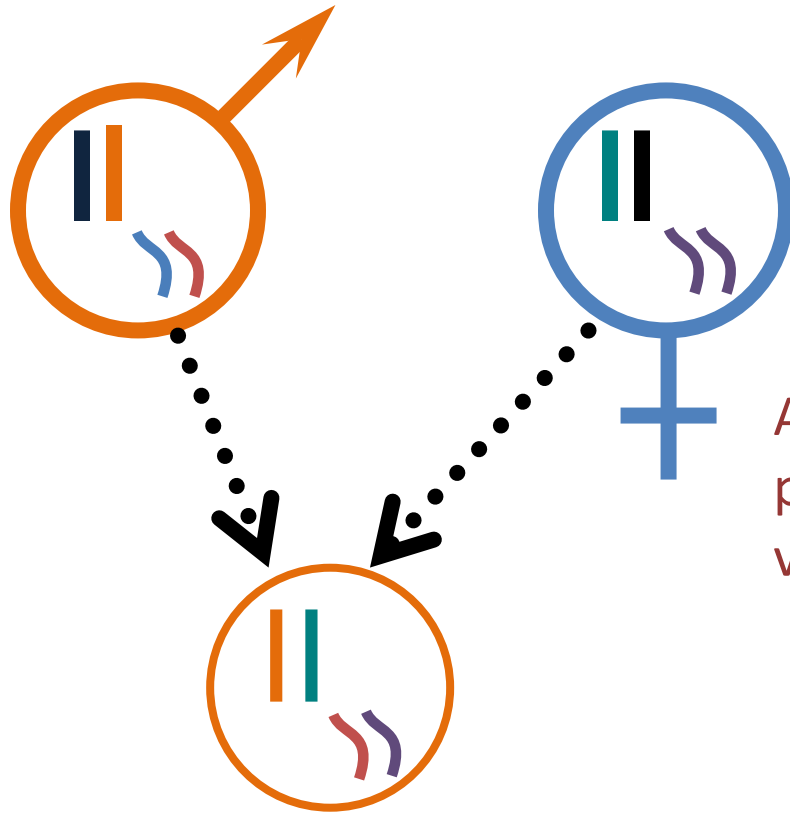
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Genetic inheritance (sexual), multiple loci



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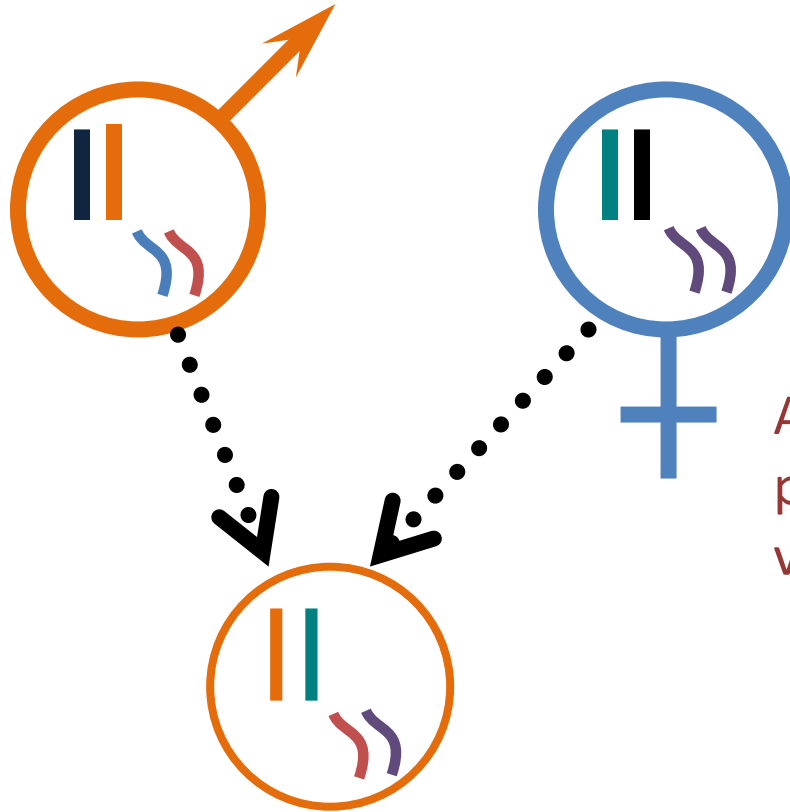
Genetic inheritance (sexual), multiple loci



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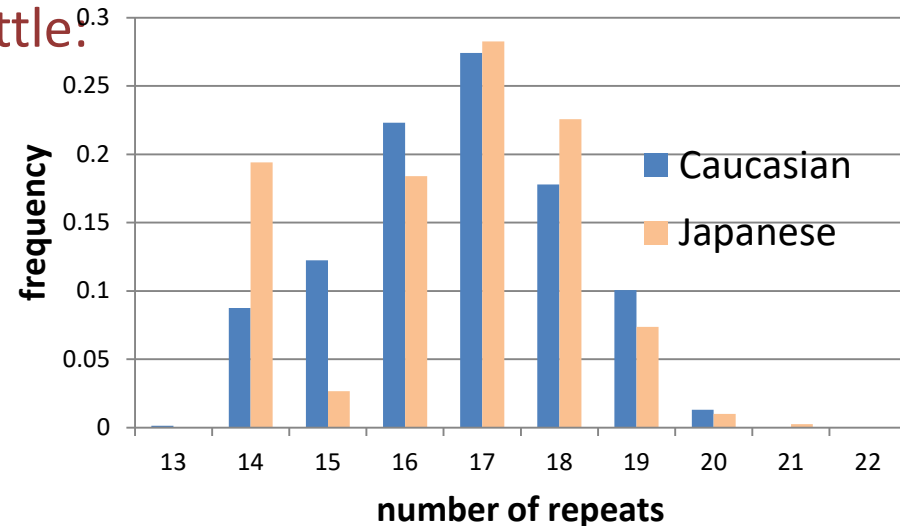
Alleles are common compared to population size, hence their frequencies vary little:

Genetic inheritance (sexual), multiple loci

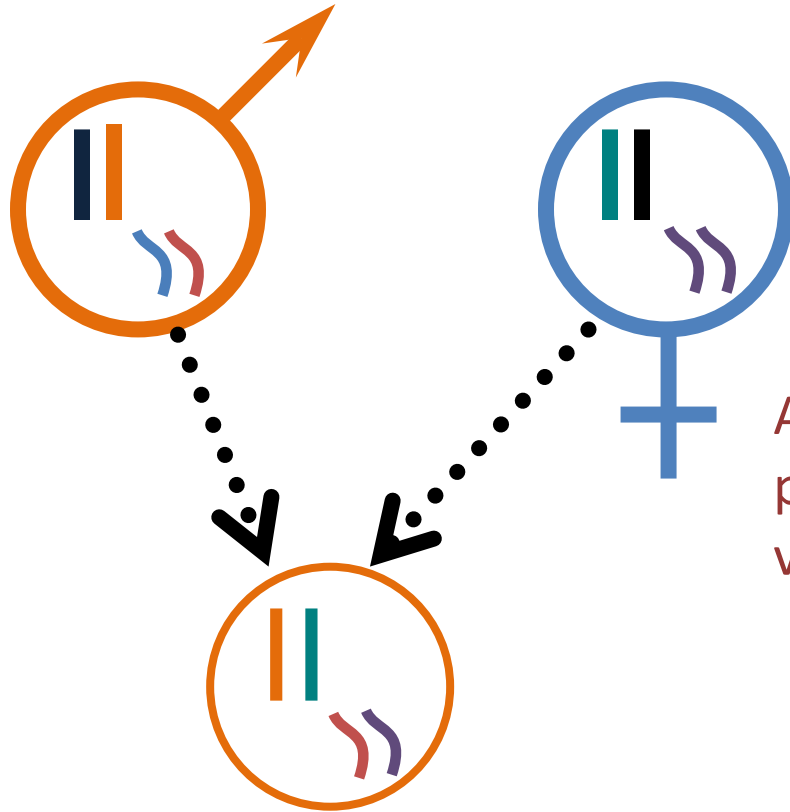


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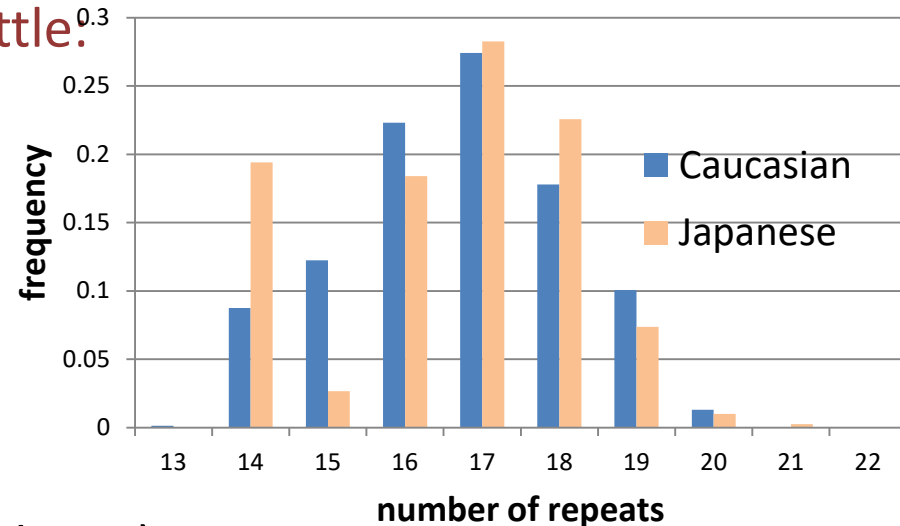


Genetic inheritance (sexual), multiple loci



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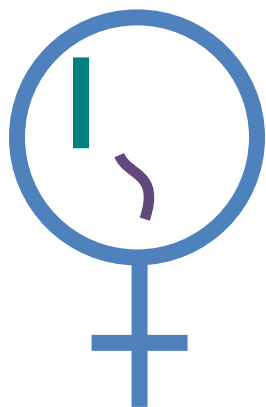
Alleles are common compared to population size, hence their frequencies vary little.



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*.

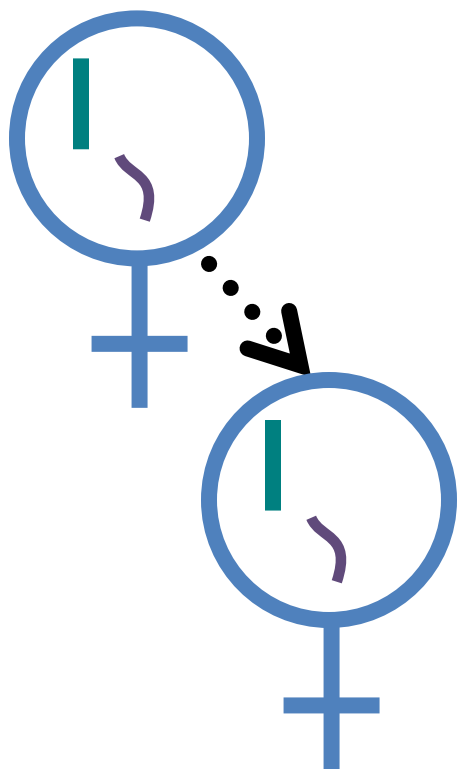
Evidential value: *cannot* multiply factor per locus.

Evidential value: ?

Treat haplotype as monster “allele”?

If 17 loci → 10000 haplotypes.

Genetic inheritance (Y), multiple loci



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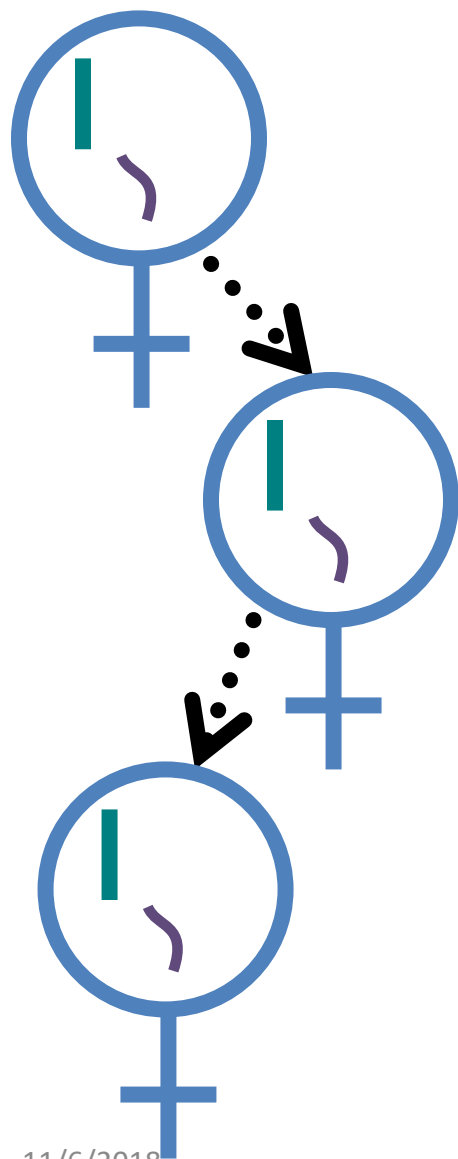
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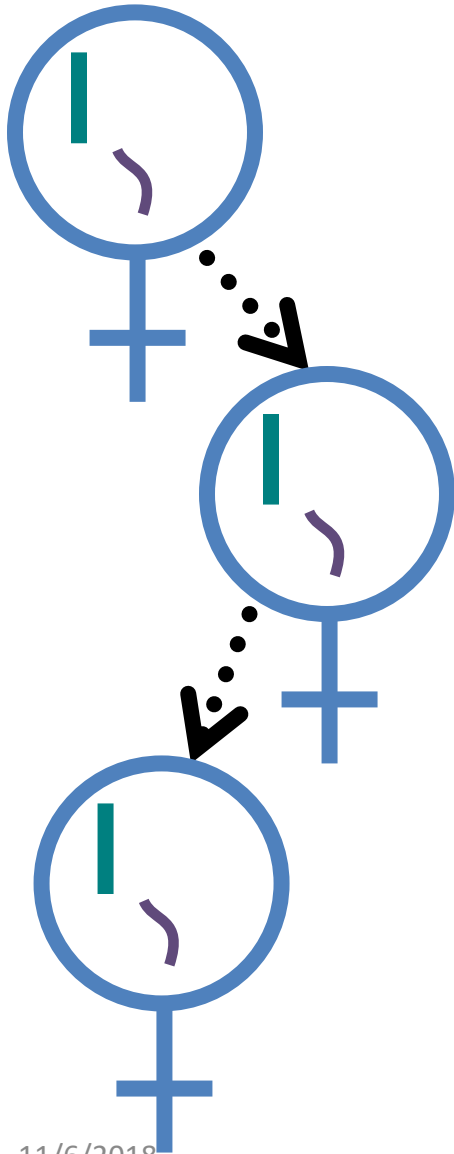
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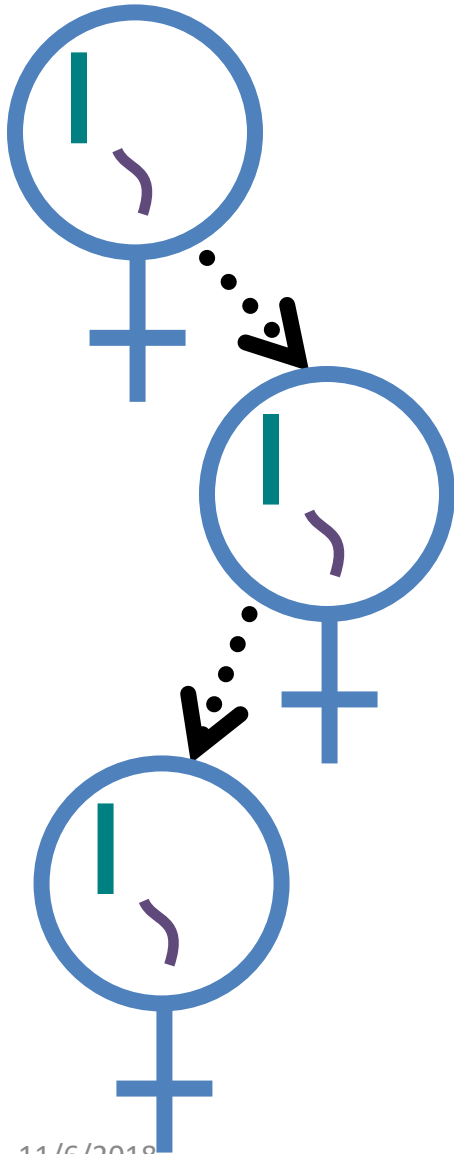
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Haplotypes are *rare* compared to population size; population frequencies vary enormously.

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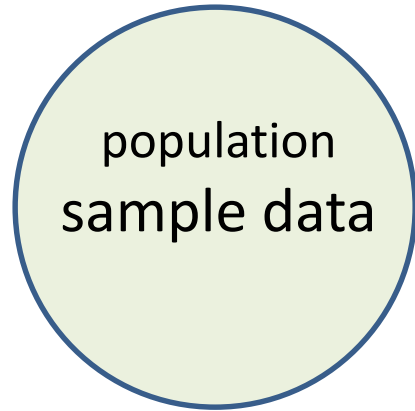
Haplotypes are **rare** compared to population size; population frequencies vary enormously.

Asexual; cloning

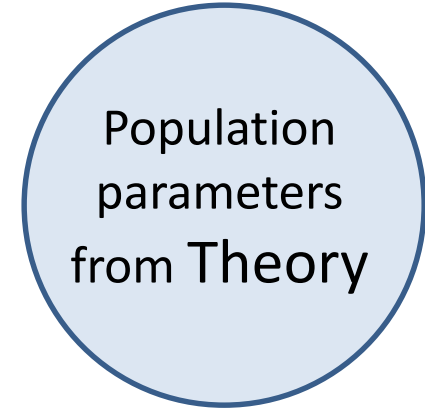
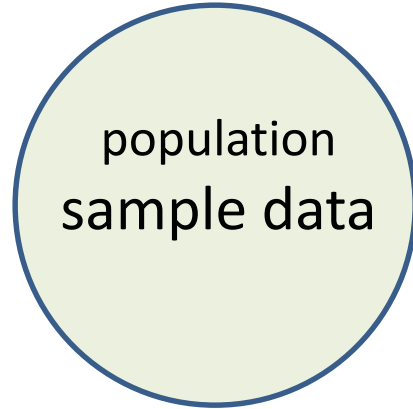


Y-evidence calculation approaches

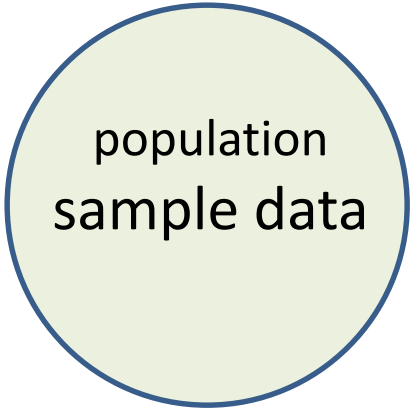
Y-evidence calculation approaches



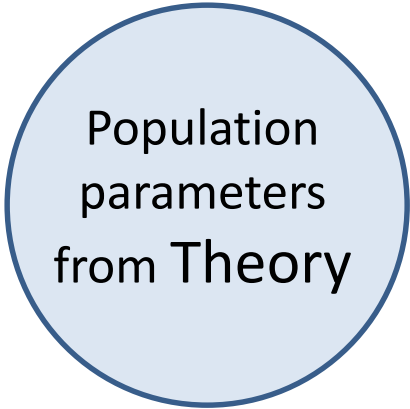
Y-evidence calculation approaches



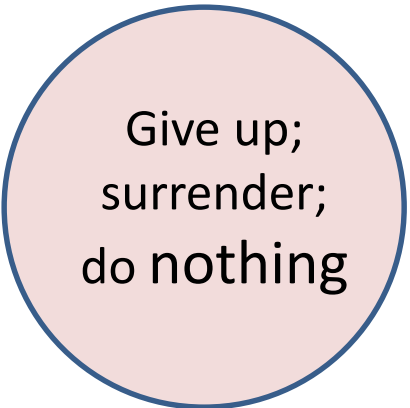
Y-evidence calculation approaches



population
sample data

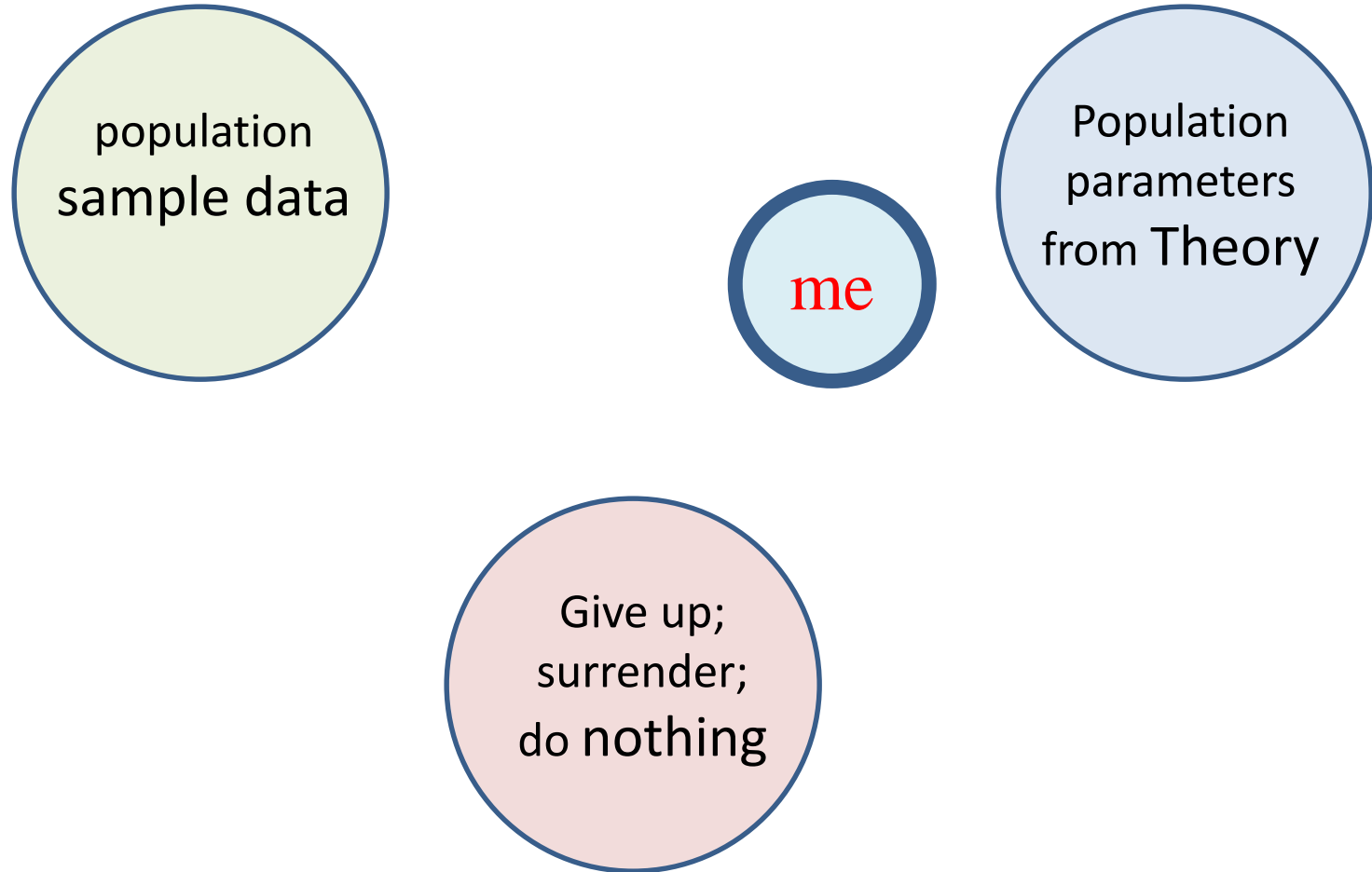


Population
parameters
from Theory



Give up;
surrender;
do nothing

Y-evidence calculation approaches



Evolution of the Yfiler lineages

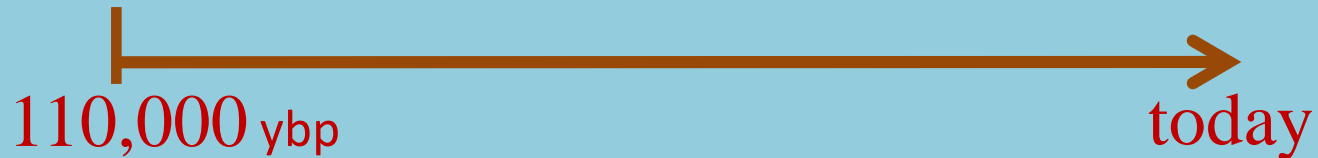


Y-chromosome
Adam”

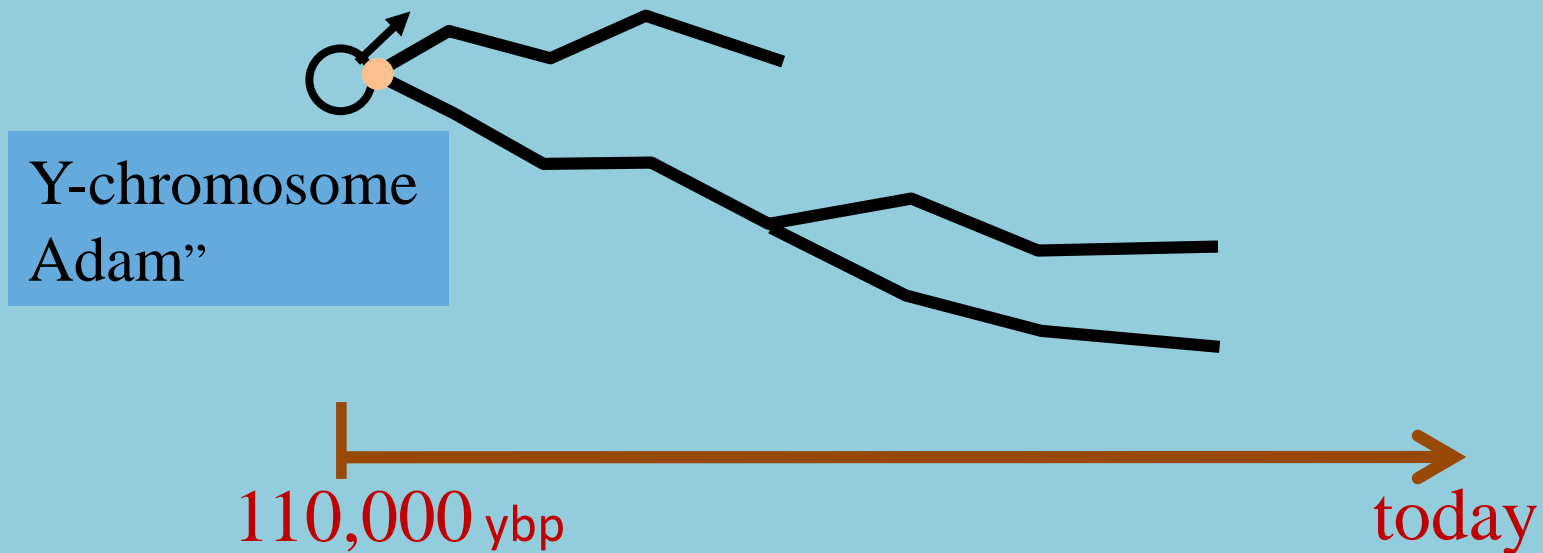
Evolution of the Yfiler lineages



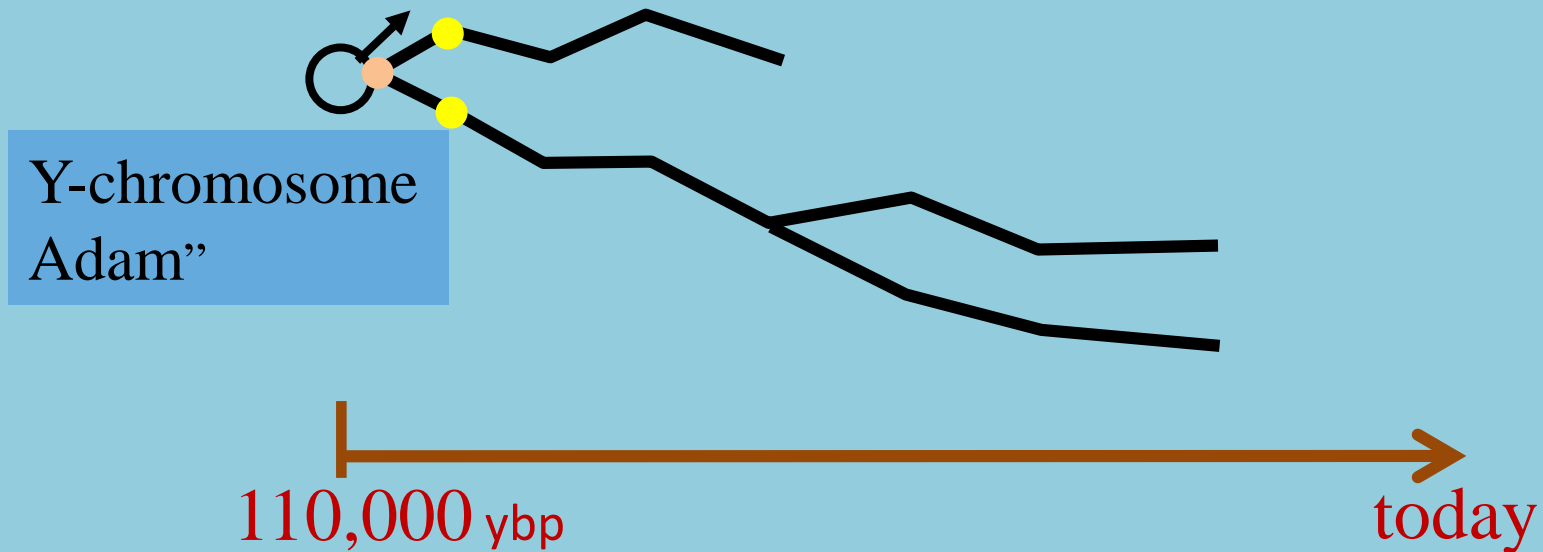
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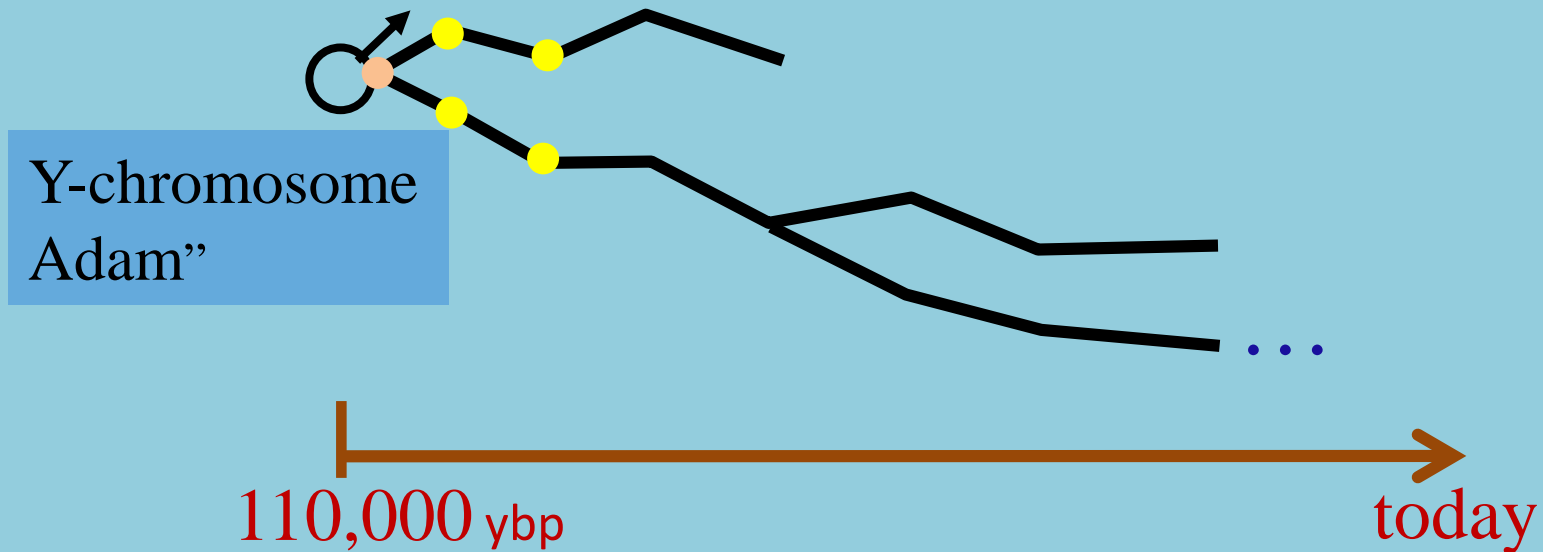
Evolution of the Yfiler lineages



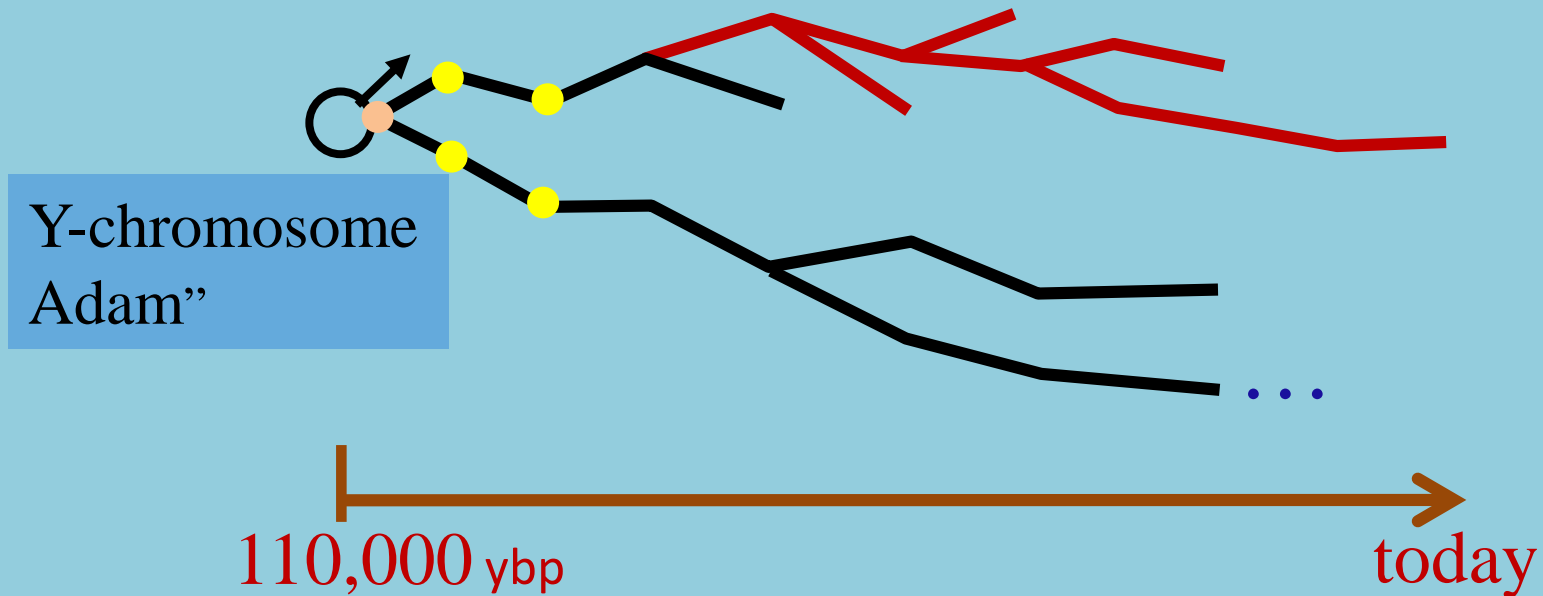
Evolution of the Yfiler lineages



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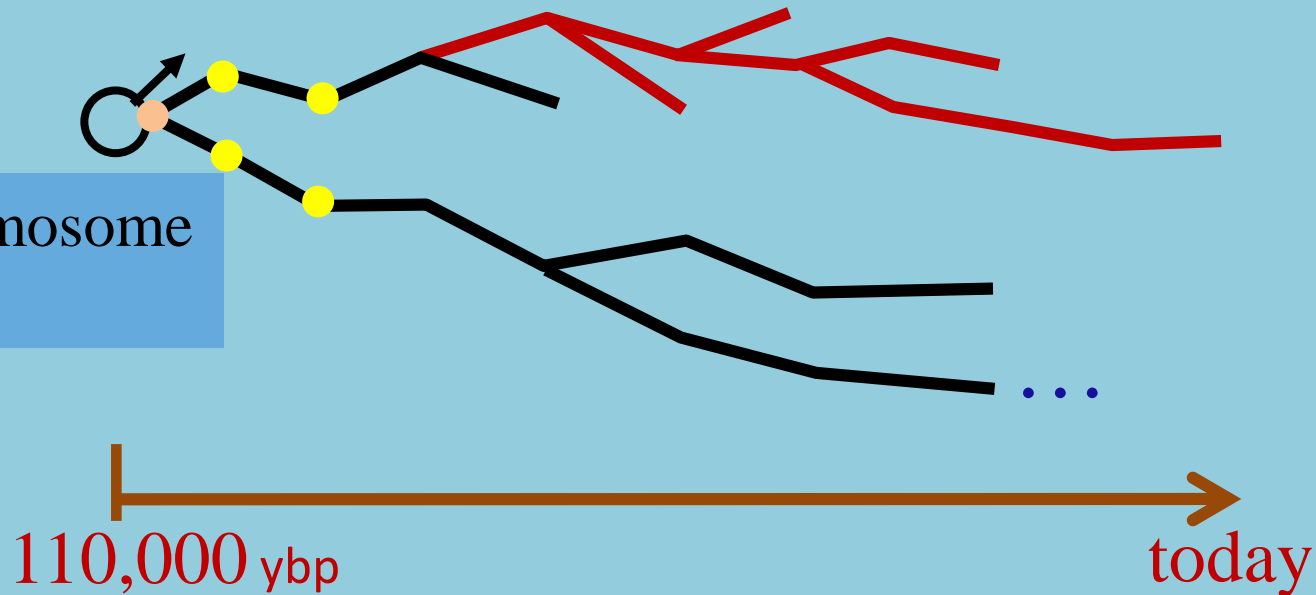
Evolution of the Yfiler lineages



Evolution of the Yfiler lineages

mutation

Y-chromosome
Adam''



Evolution of the Yfiler lineages

mutation

Y-chromosome
Adam''

110,000 ybp

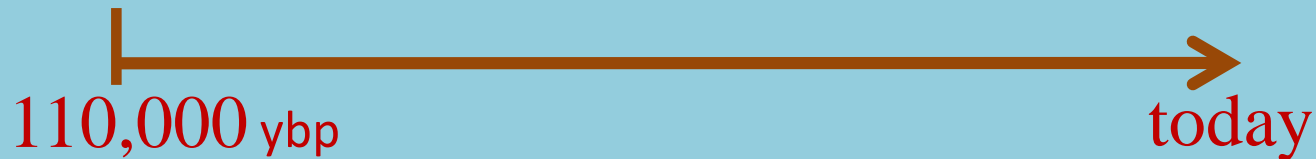
today

The diagram illustrates the evolutionary paths of the Y-chromosome. It starts at a single point labeled 'Y-chromosome Adam'' at 110,000 ybp. From this point, the lineage splits into several branches. One branch is highlighted in red, another in yellow, and others in black. A yellow arrow labeled 'mutation' points to a specific node on the yellow lineage, indicating a point of genetic change. The timeline at the bottom shows the progression from 110,000 ybp to 'today'.

Evolution of the Yfiler lineages

mutation

Y-chromosome
Adam''

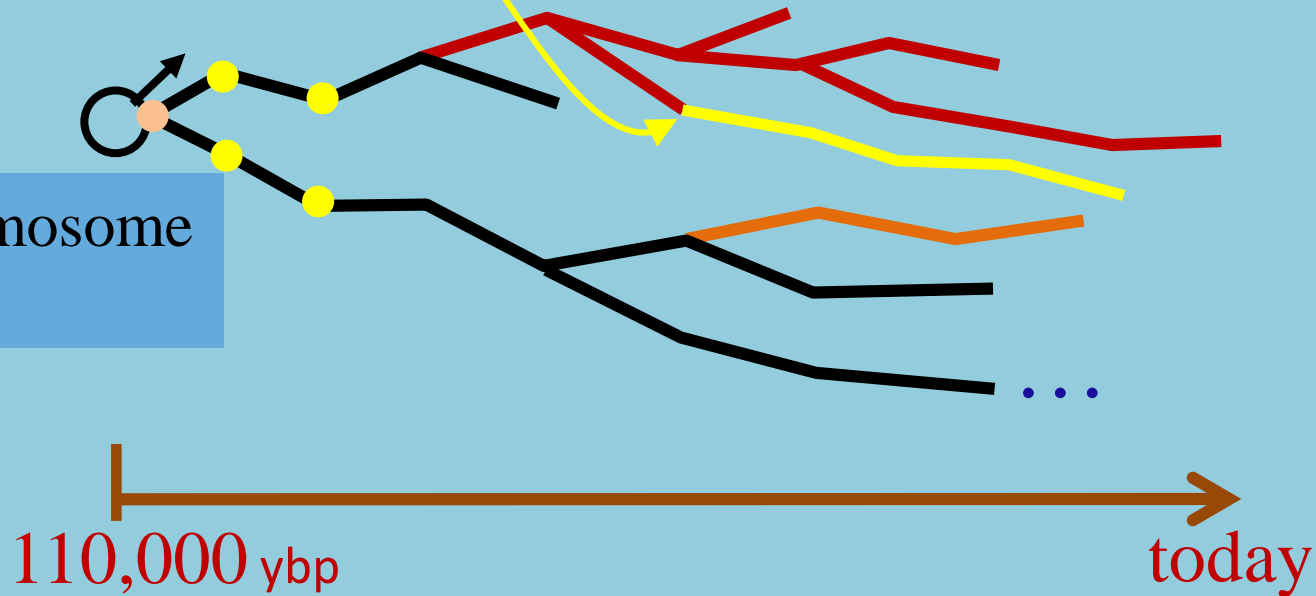


(Same color = same Y-haplotype)

Evolution of the Yfiler lineages

mutation

Y-chromosome
Adam''



(Same color = same Y-haplotype)

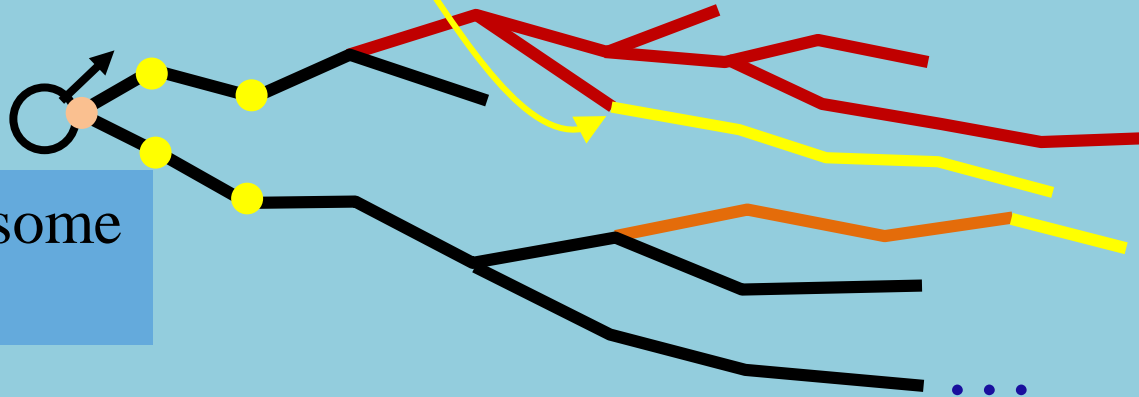
Evolution of the Yfiler lineages

mutation

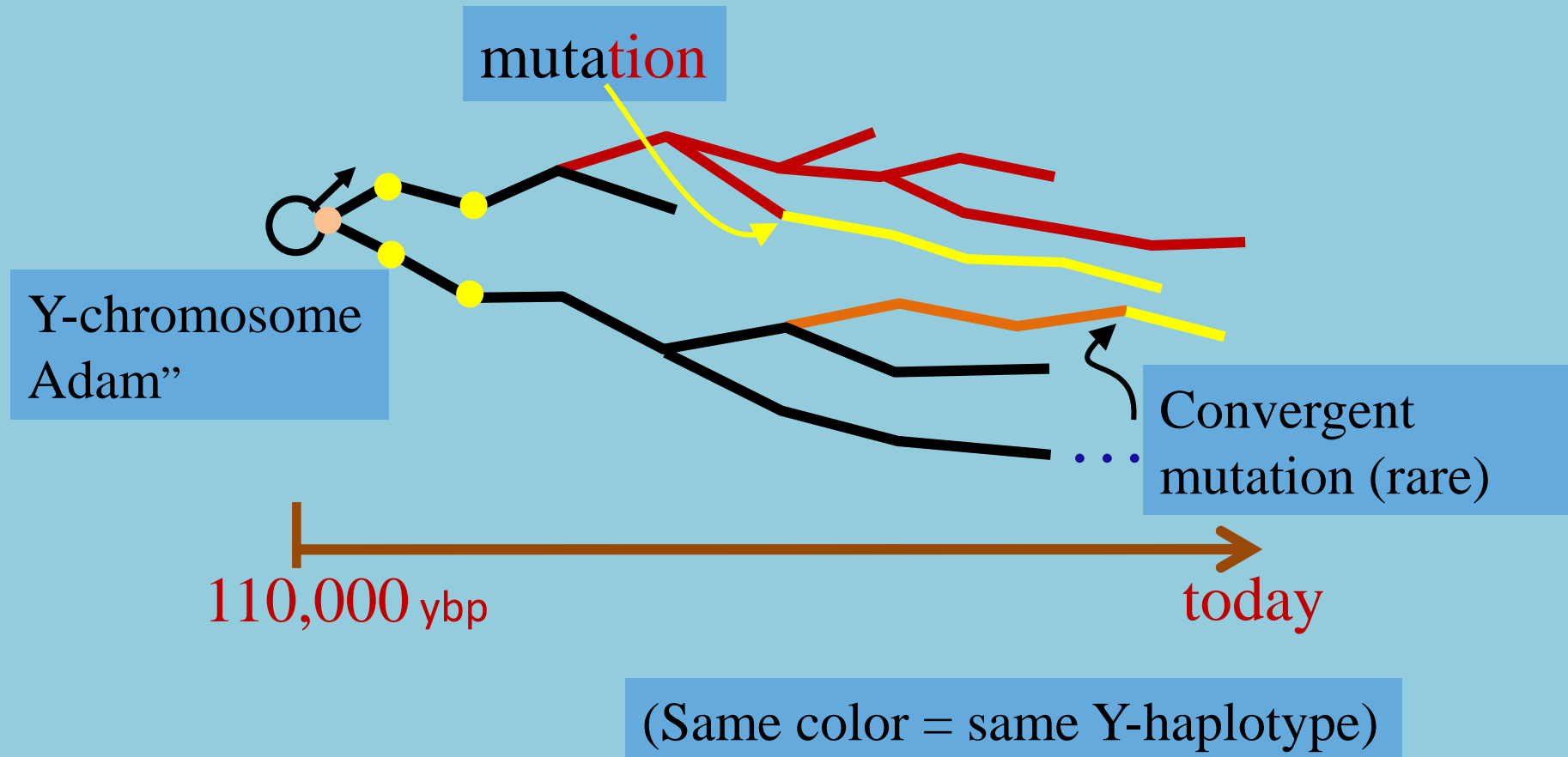
Y-chromosome
Adam''

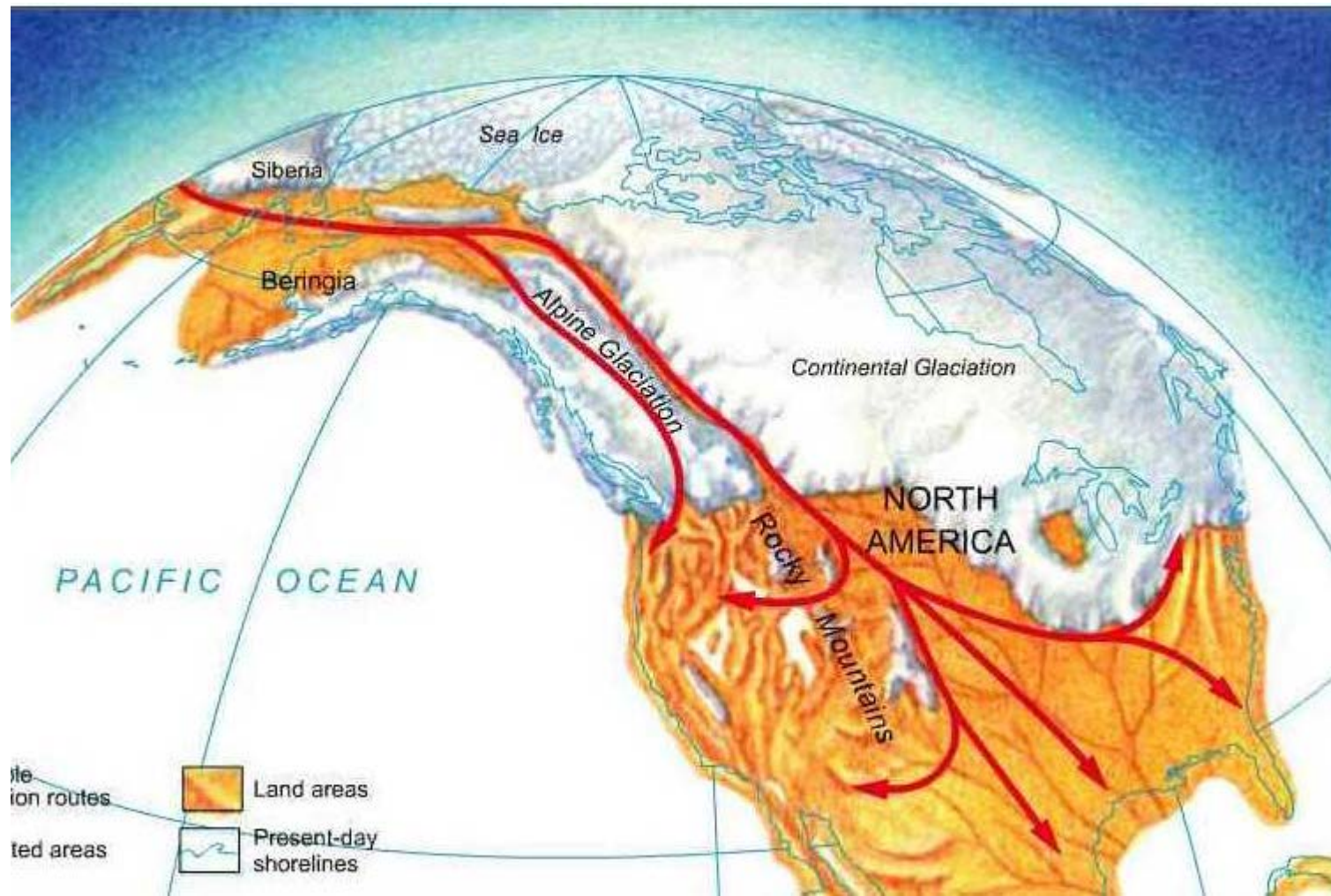
110,000 ybp → today

(Same color = same Y-haplotype)

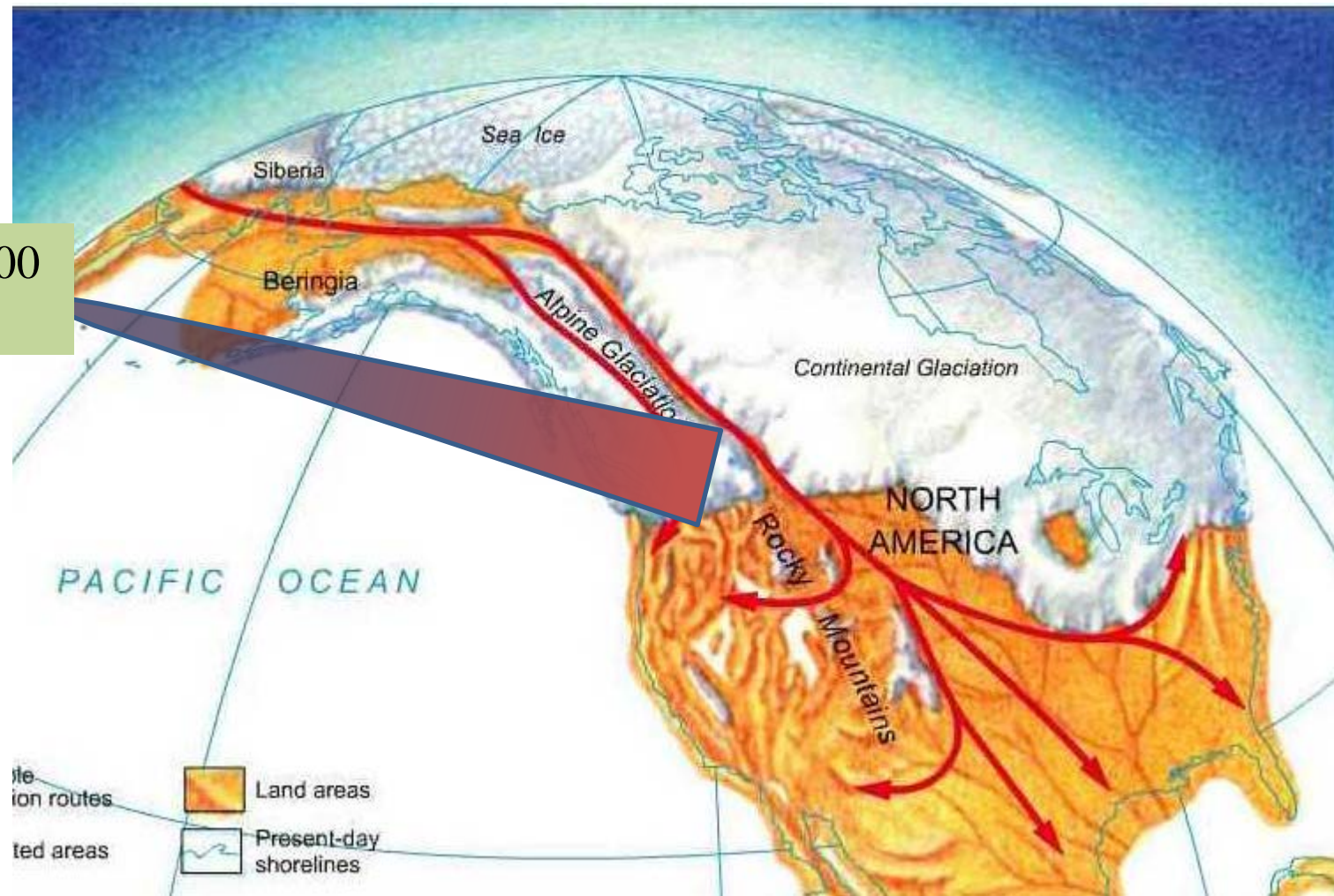


Evolution of the Yfiler lineages

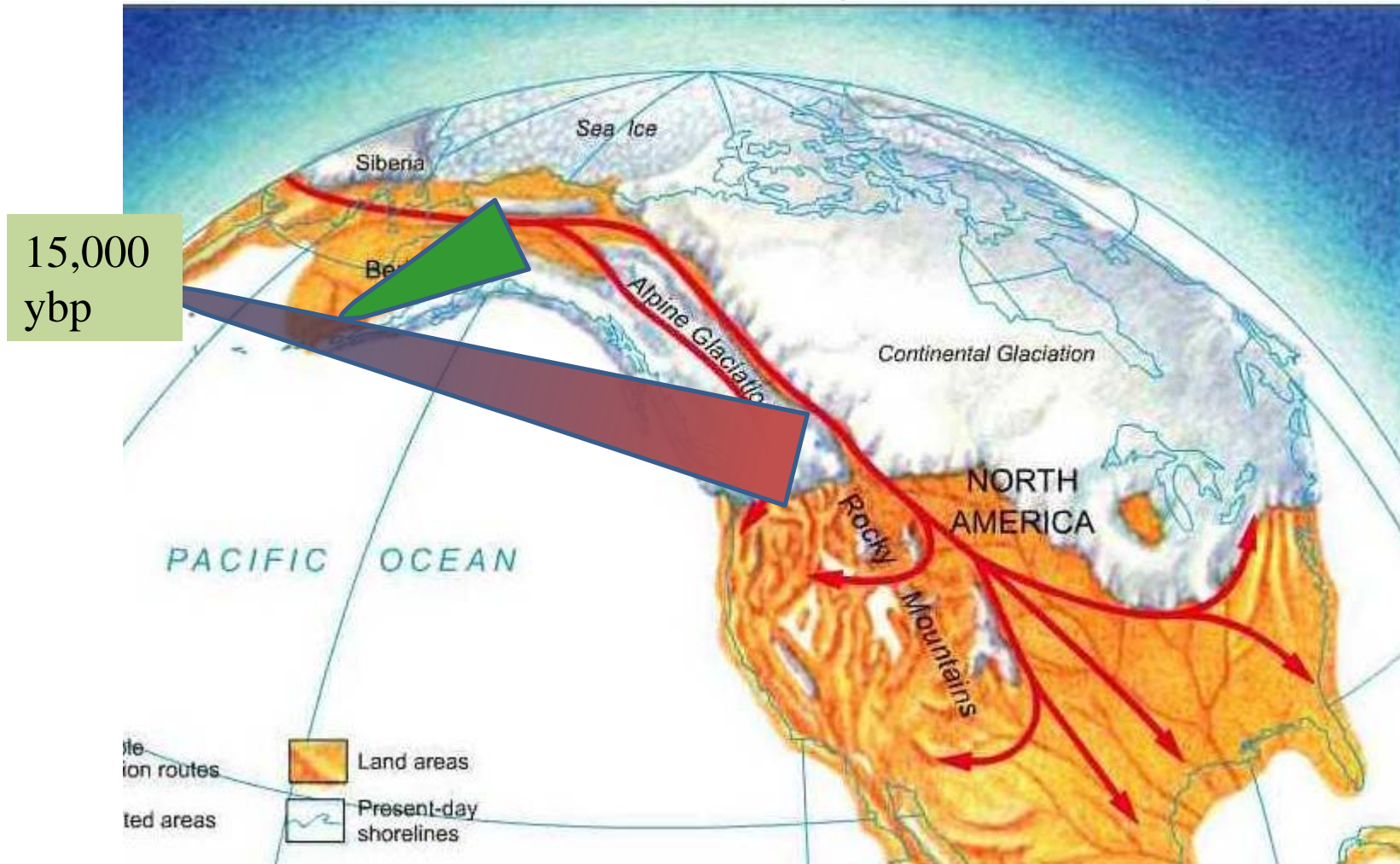




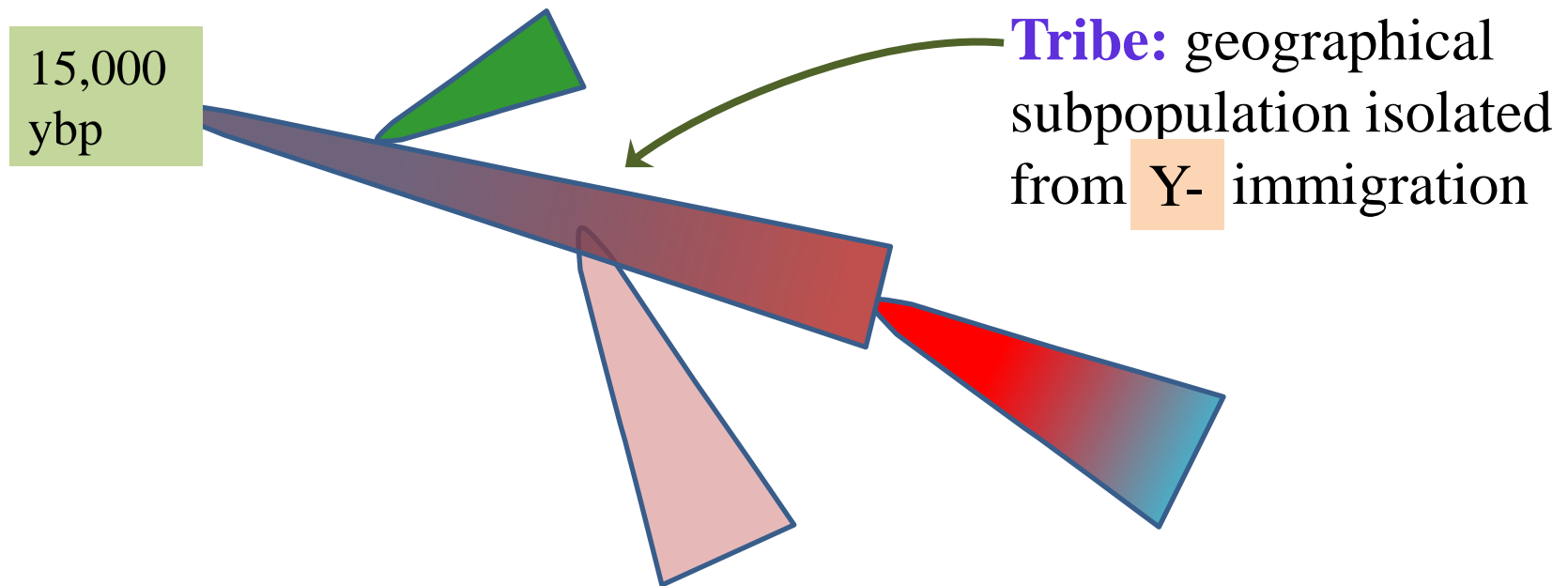
15,000
ybp



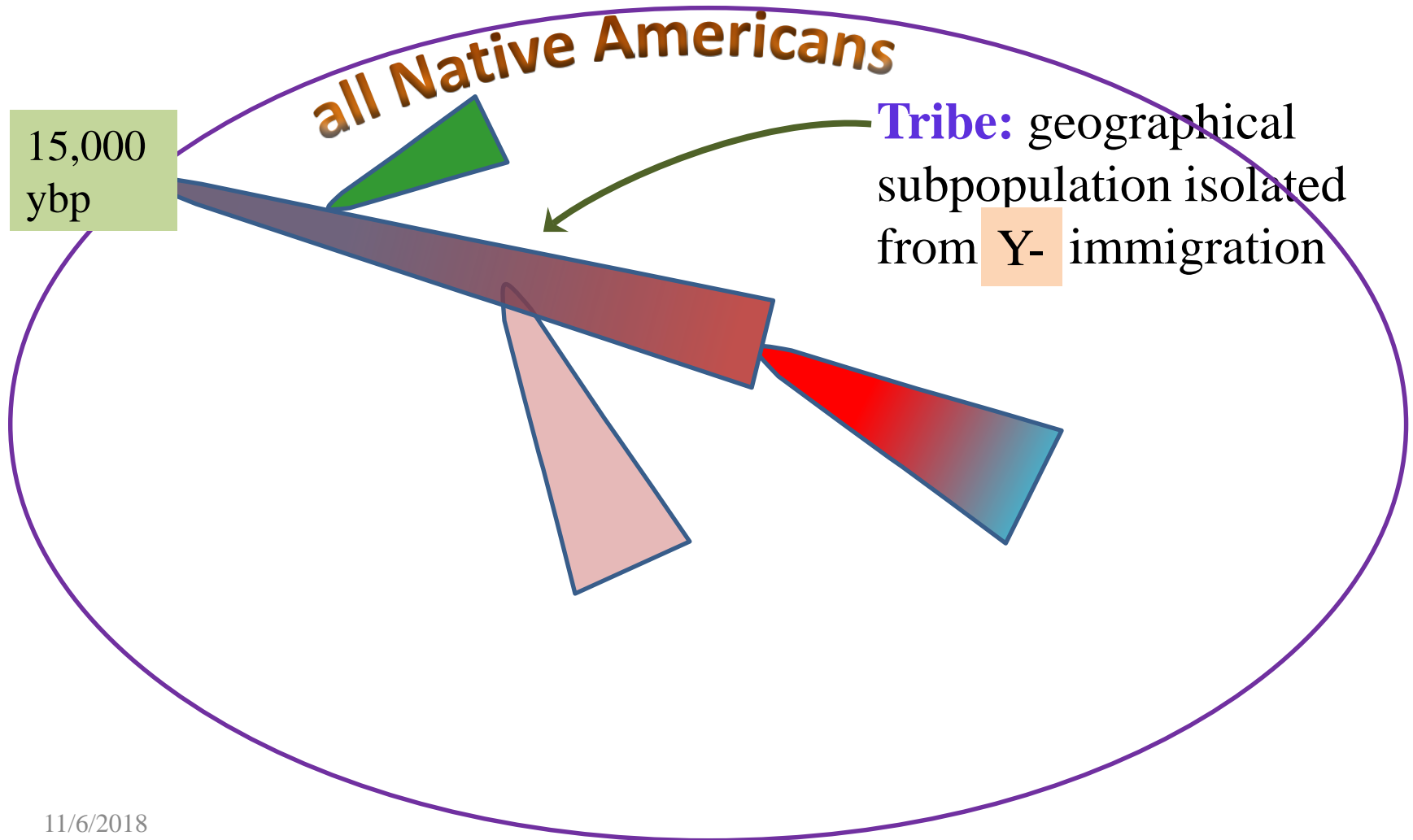
Manufacturing diversity



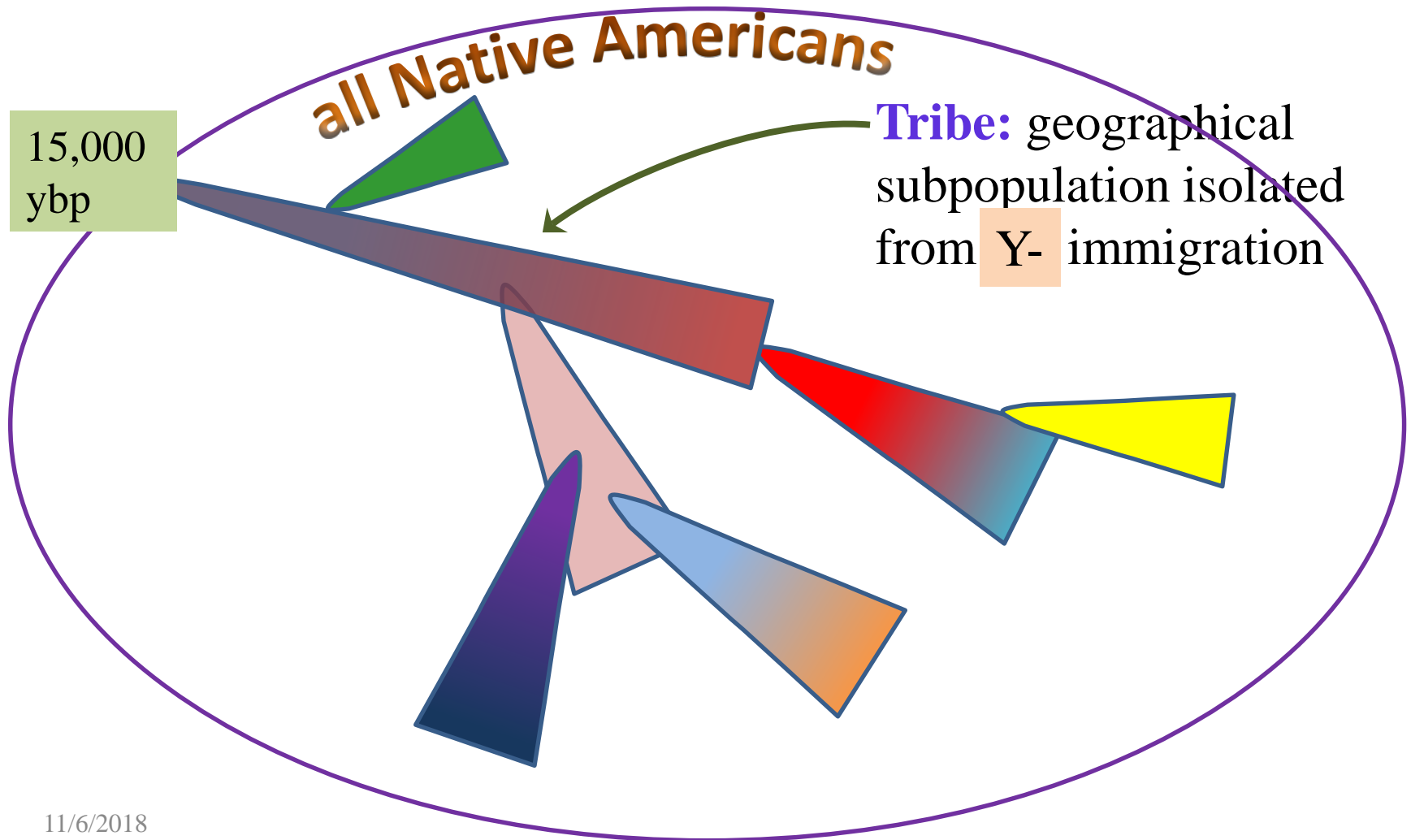
Manufacturing diversity



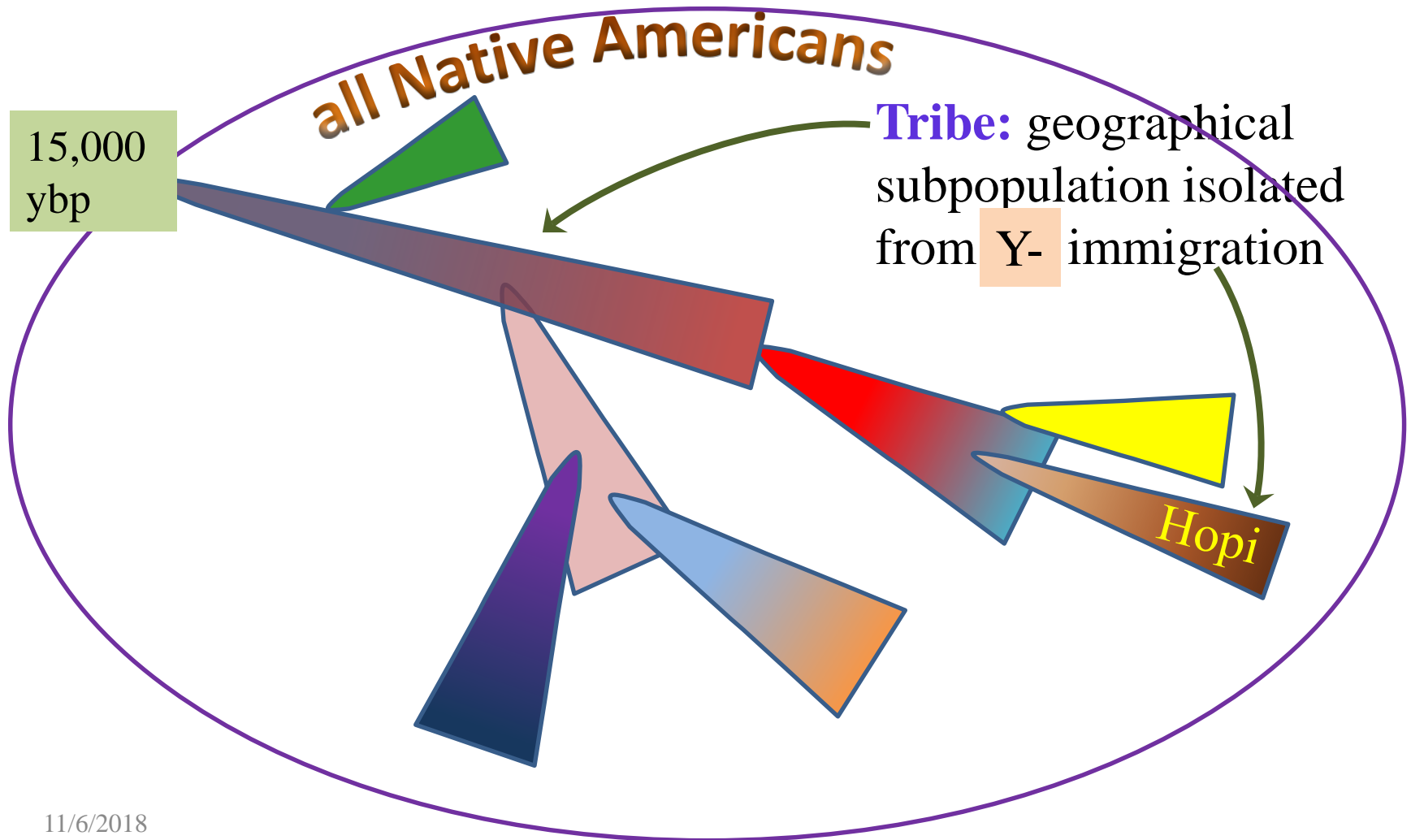
Manufacturing diversity



Manufacturing diversity



Manufacturing diversity



Manufacturing diversity

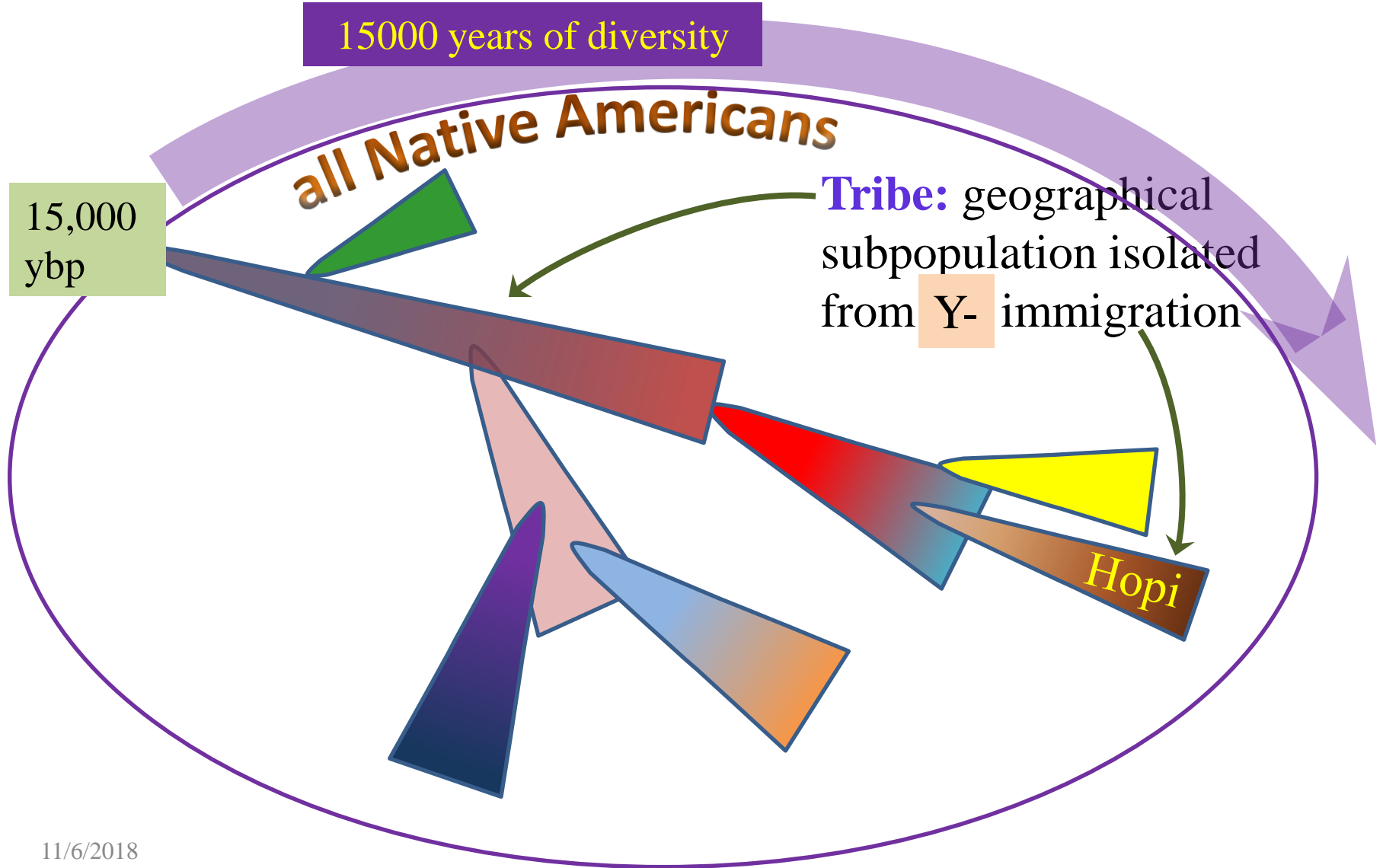
15000 years of diversity

all Native Americans

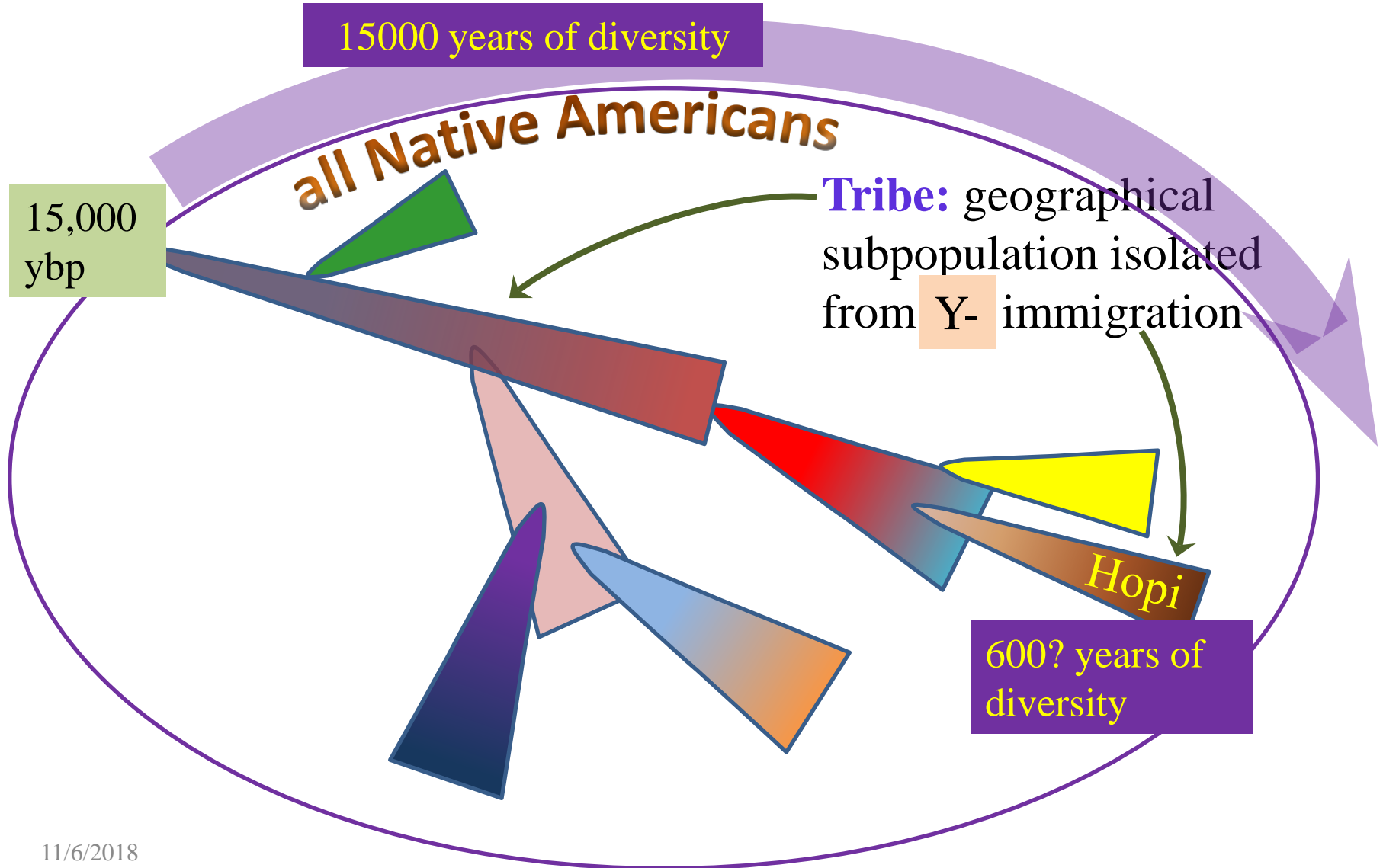
15,000
ybp

Tribe: geographical
subpopulation isolated
from Y- immigration

Hopi



Manufacturing diversity



Manufacturing diversity

15000 years of diversity

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

all Native Americans

15,000
ybp

Tribe: geographical
subpopulation isolated
from Y- immigration

Hopi

600? years of
diversity

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

Southwest Native Am. court cases

Southwest Native Am. court cases

- T. Kootswatewa (Feb 2016)

Southwest Native Am. court cases

- T. Kootswatewa (Feb 2016)
 - Child victim

Southwest Native Am. court cases

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 - Impressive judge agrees with me

Southwest Native Am. court cases

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Southwest Native Am. court cases

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Southwest Native Am. court cases

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Southwest Native Am. court cases

- T. Kootswatewa (Feb 2016)
 - Child victim
 - Impressive judge agrees with me
- A. Shirley (Aug 2016)
 - Lesser judge agrees with first judge
- M Russell (Oct 2017)
 - Can't quit on her

Y-haplotype mutation and matching

Y-haplotype mutation and matching

- Mutation model

Y-haplotype mutation and matching

- Mutation model

Adam ← 17p5 5 5 5 ...

A ancestral 17-locus Y haplotype

Y-haplotype mutation and matching

- Mutation model

Adam ← 17p5 5 5 5 ... A ancestral 17-locus Y haplotype

mu ← ÷350 8000 200 500 ... A per-locus mutation rates. Ave mu =
(÷350)

Y-haplotype mutation and matching

- Mutation model

Adam ← 17p5 5 5 5 ... A ancestral 17-locus Y haplotype

mu ← ÷350 8000 200 500 ... A per-locus mutation rates. Ave mu = (÷350)

SonOf ← {mu{α>?0:ω ⋄ ω+(.5>?0)⊃1 -1}·ω} A mutation at a locus is +1 or -1 step

Y-haplotype mutation and matching

- Mutation model

Adam ← 17p5 5 5 5 ... A ancestral 17-locus Y haplotype

mu ← ÷350 8000 200 500 ... A per-locus mutation rates. Ave mu = (÷350)

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Y-haplotype mutation and matching

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$$\Pr(\mathbf{Son} \equiv \mathbf{Father}) = (\times / 1 - \mu)$$

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Pr(**Son**≡**Father**) = (×/1-mu)

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

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NB: Time is reversible.

Y-haplotype population

Y-haplotype population

- Evolutionary model – Wright-Fisher growth + mutation

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Pop ←, cAdam ← 17/5 A generation 0, founder

Y-haplotype population

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

Y-haplotype population

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`Pop ← SonOf`Pop[?(NewSize ≠Pop)/≠Pop]` \mathcal{A} generation g

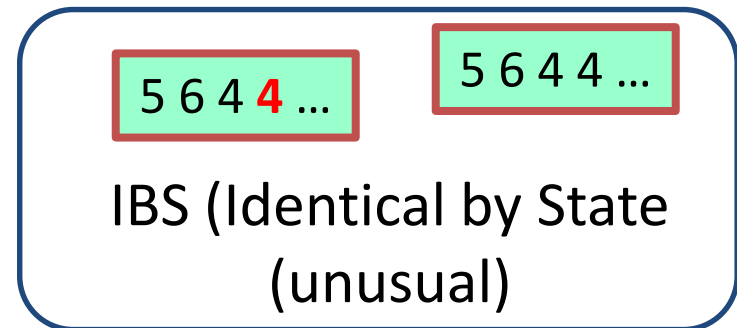
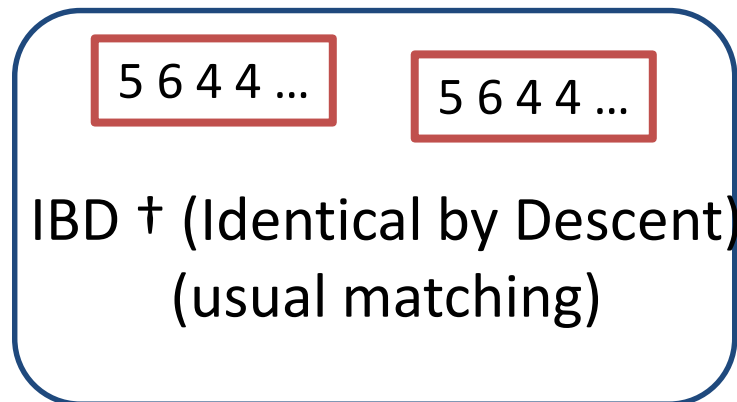
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Y-haplotype population

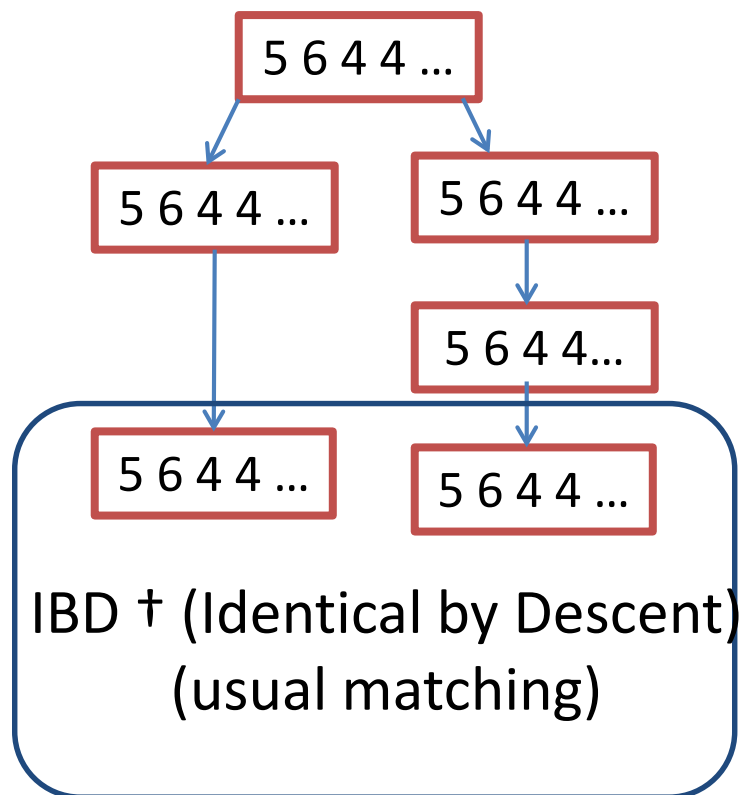
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- Iterating generations
 - “Diversity” \equiv accumulation of mutations
 - Time
 - Population size

All men are related

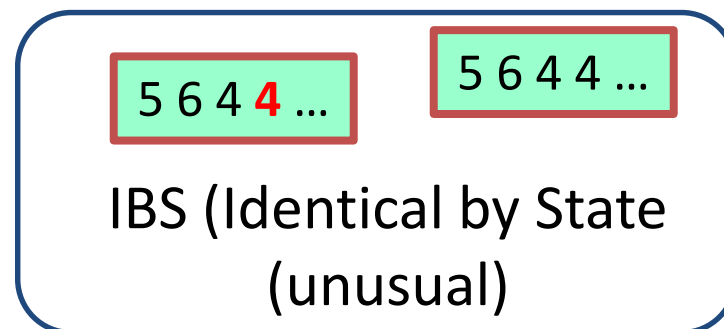


† Each box represents a 4-
locus Y haplotype

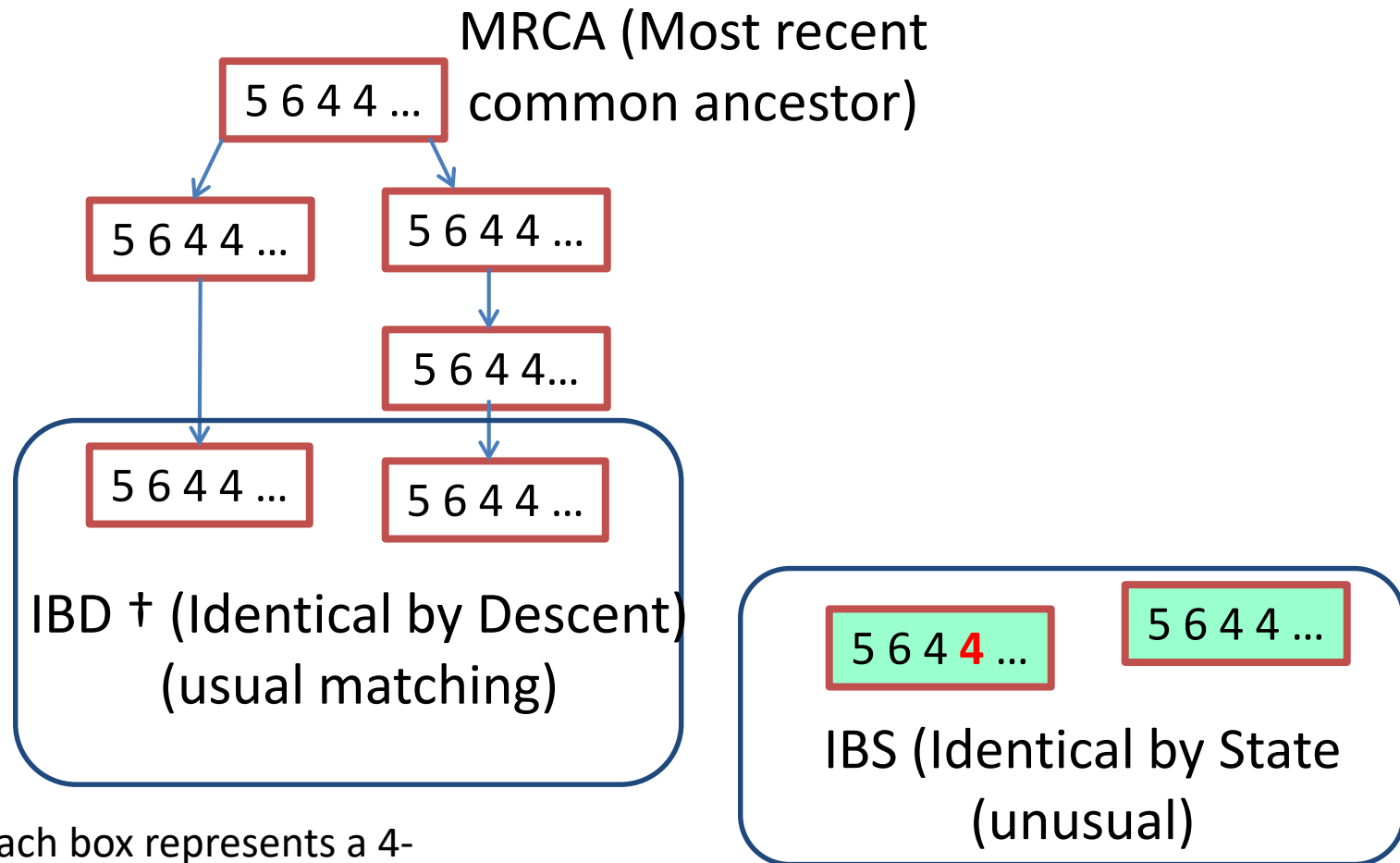
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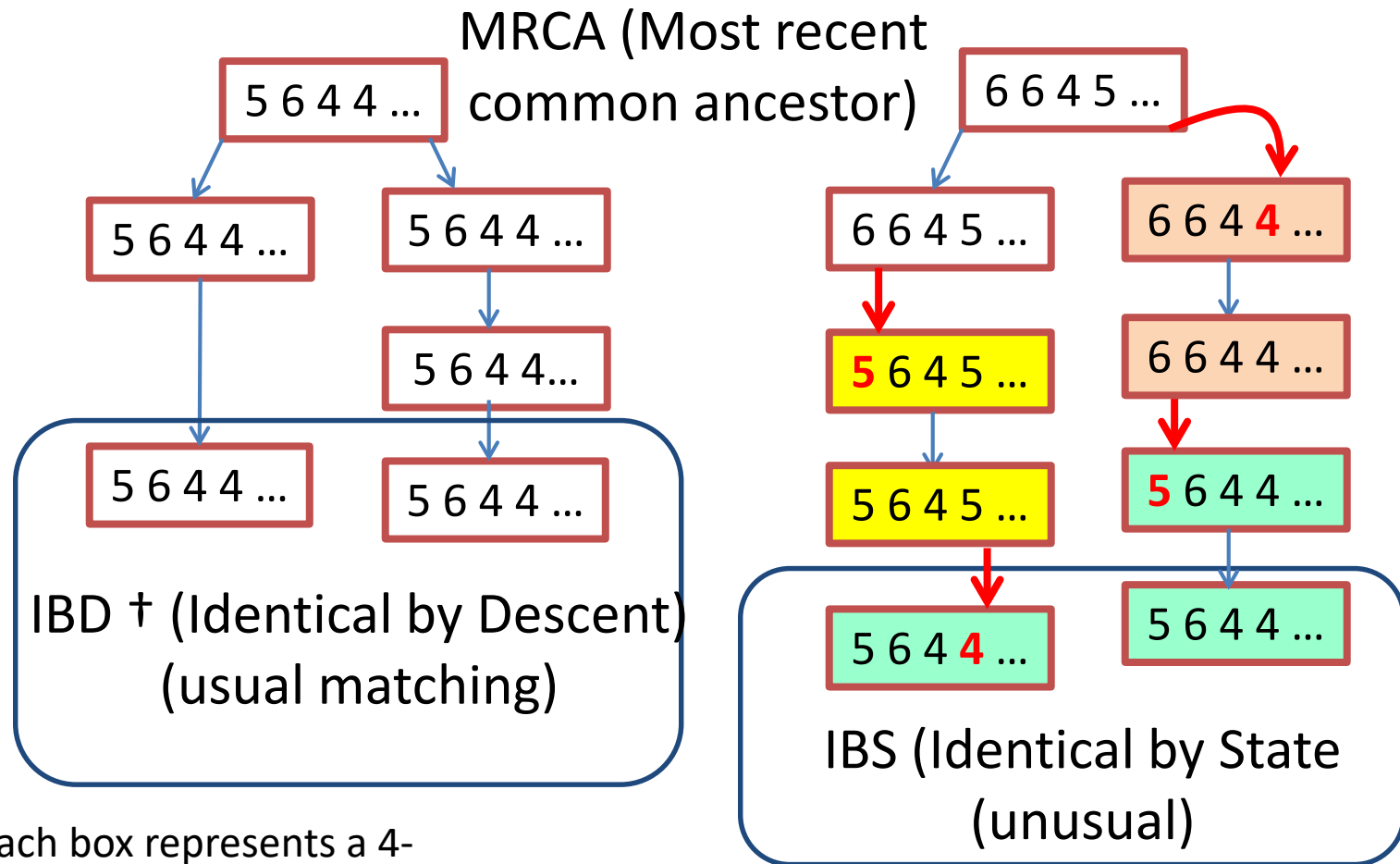


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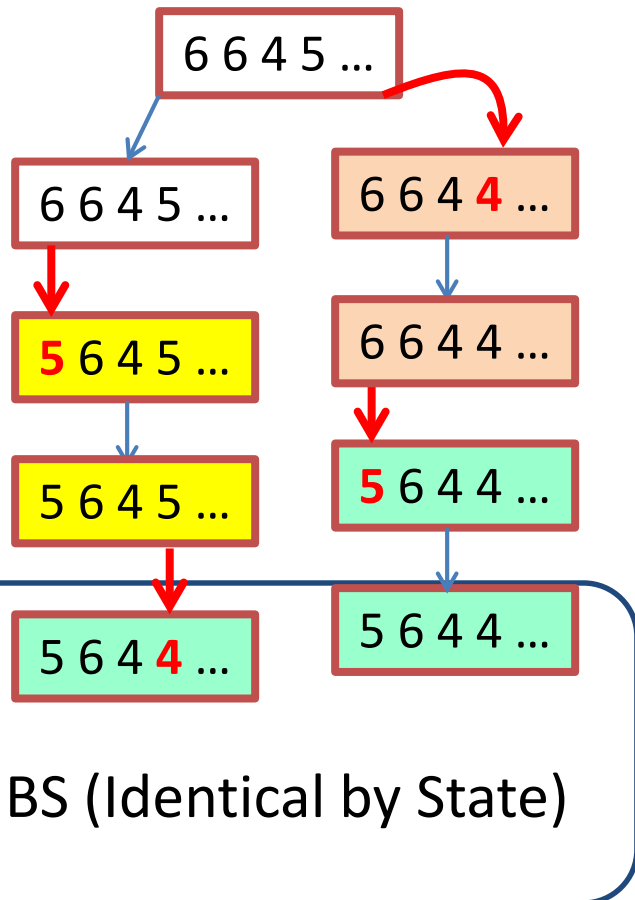
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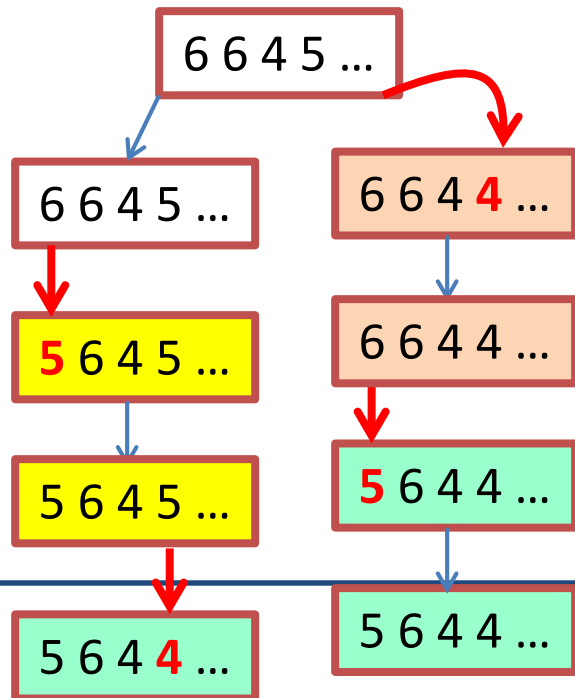
Identity by State – closer look

MRCA (Most recent
common ancestor)



Identity by State – closer look

MRCA (Most recent common ancestor)

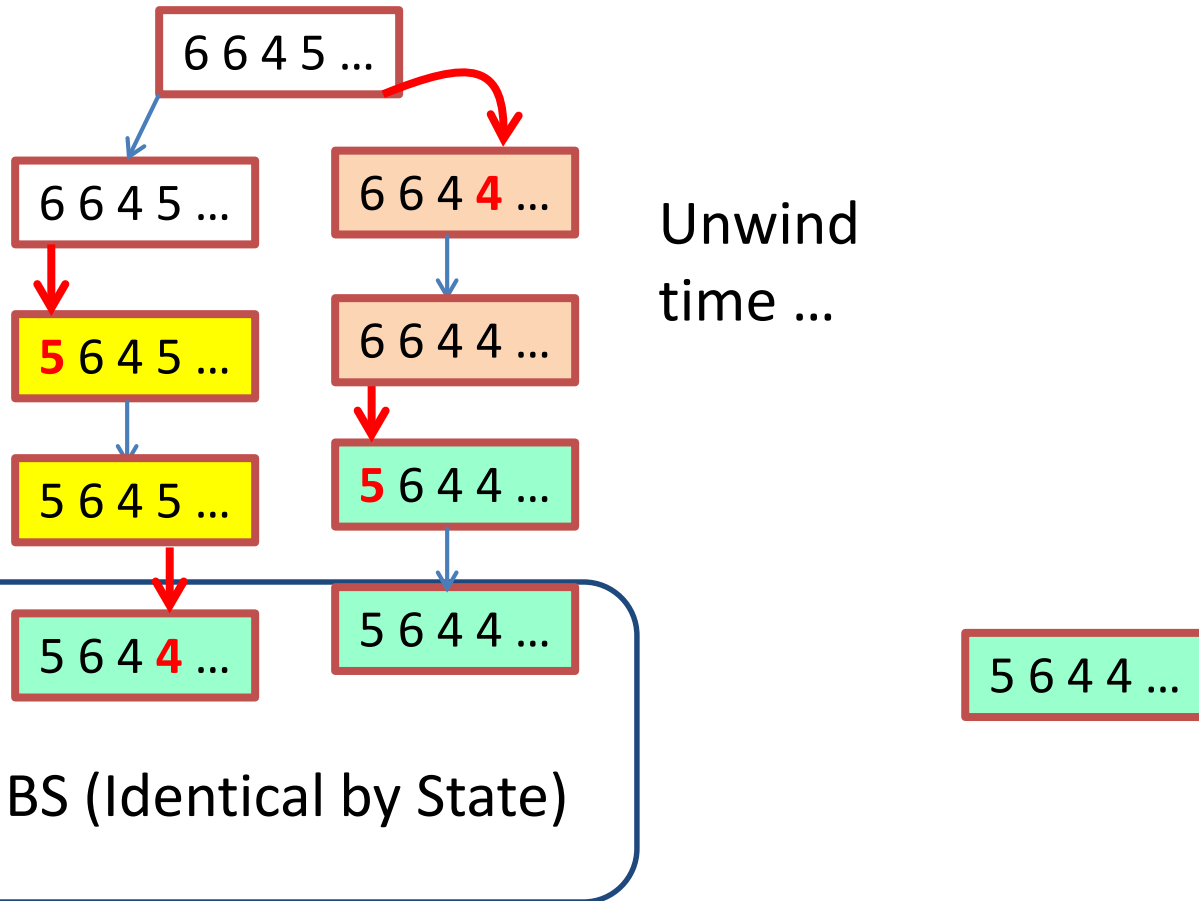


Unwind time ...

IBS (Identical by State)

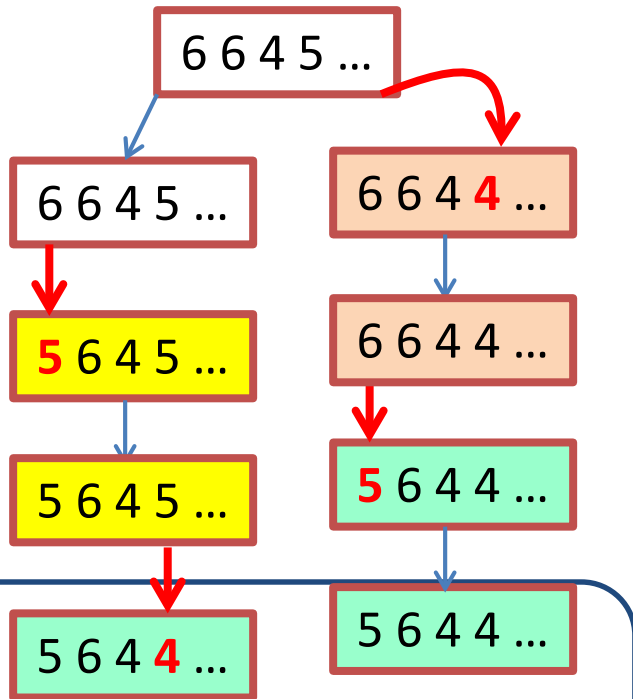
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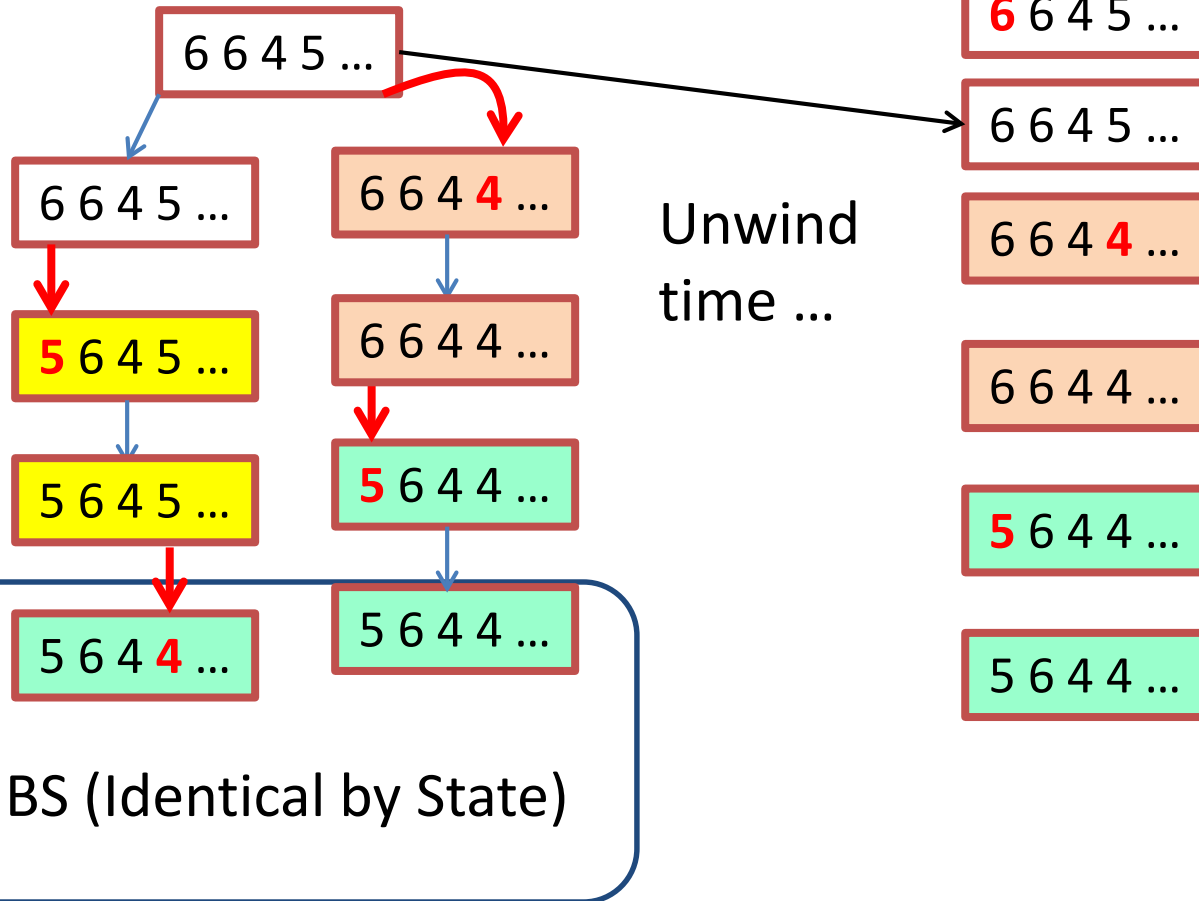
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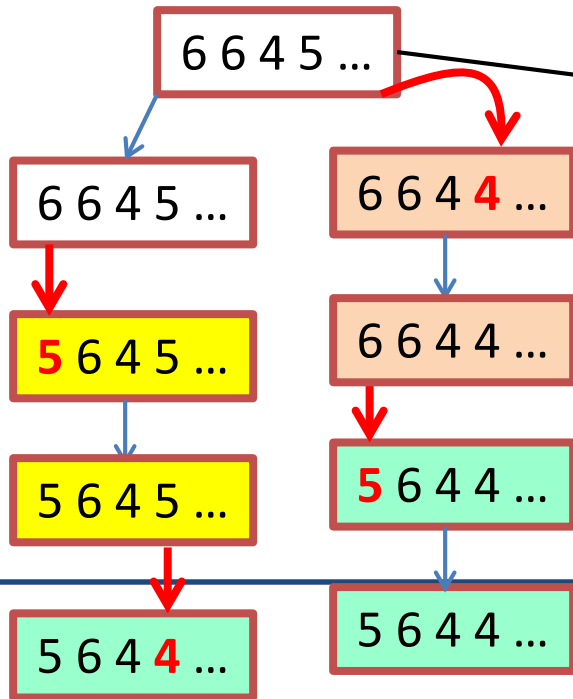
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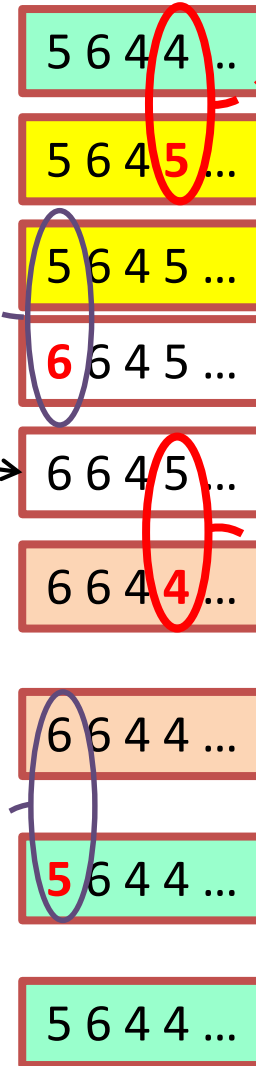


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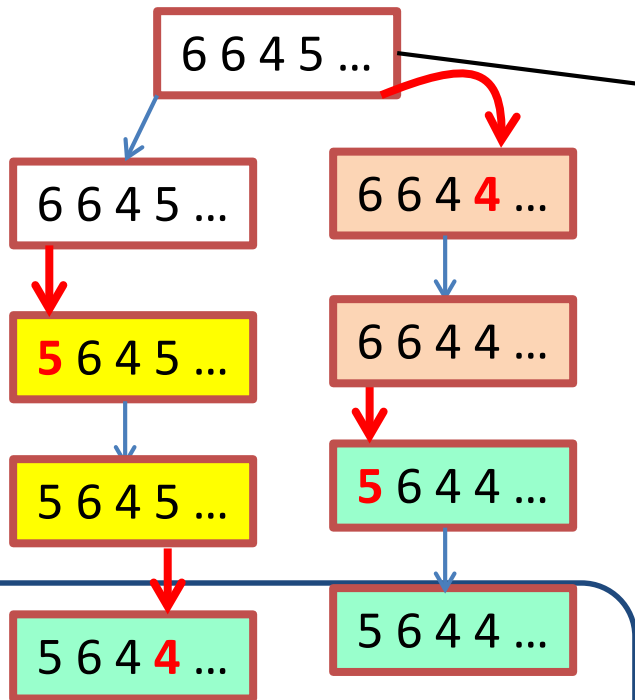
Unwind
time ...



2 pairs of
cancelling
mutations
.

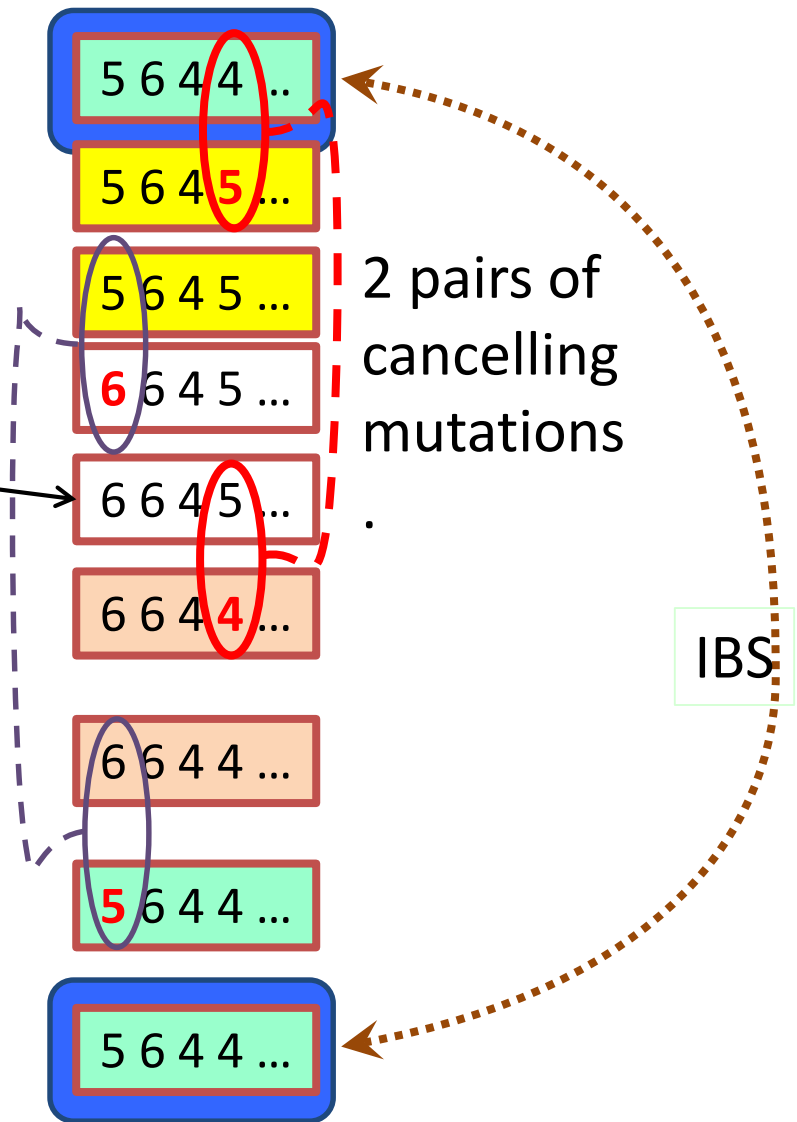
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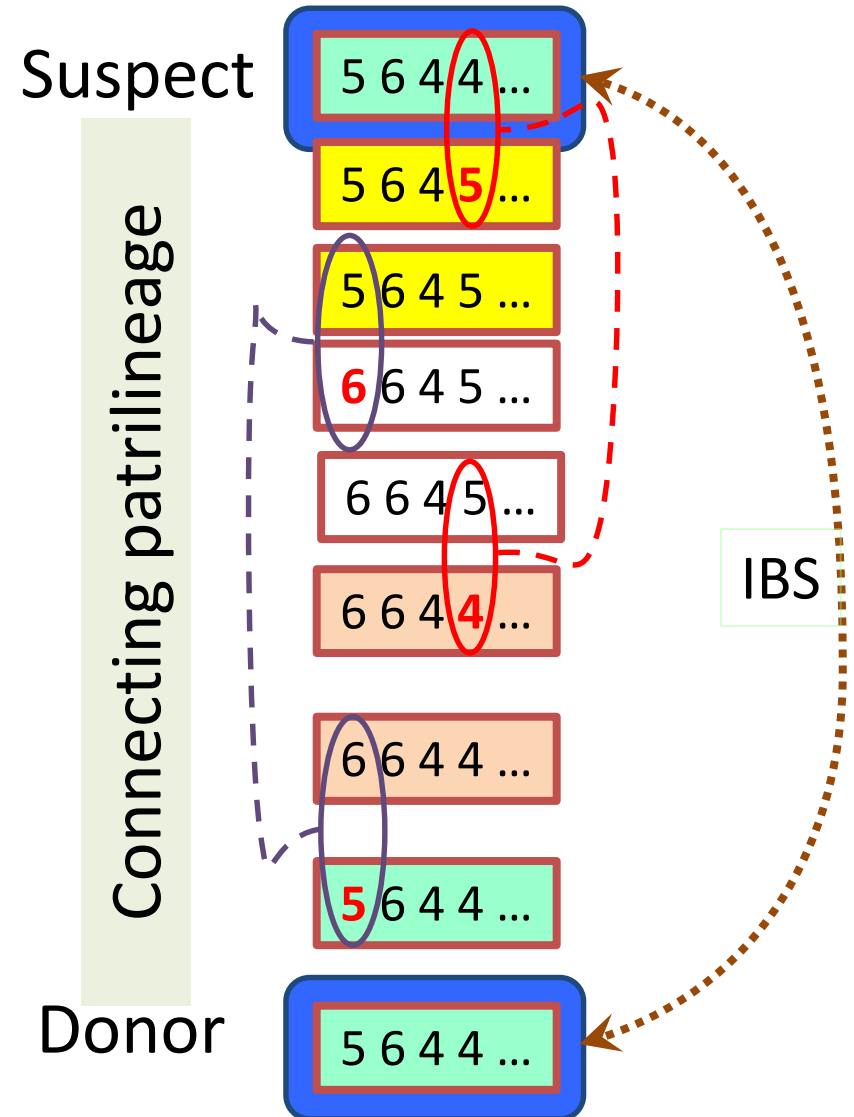


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Unwind
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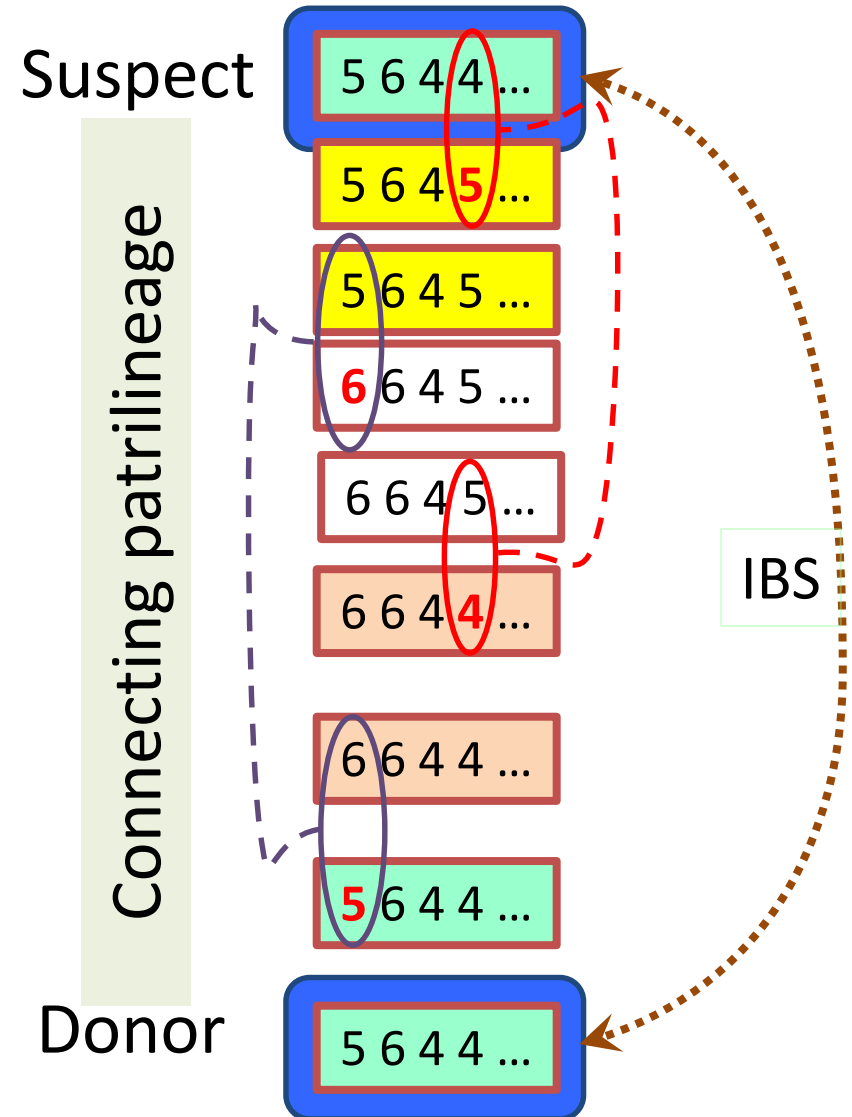


Y haplotype match condition



Y haplotype match condition

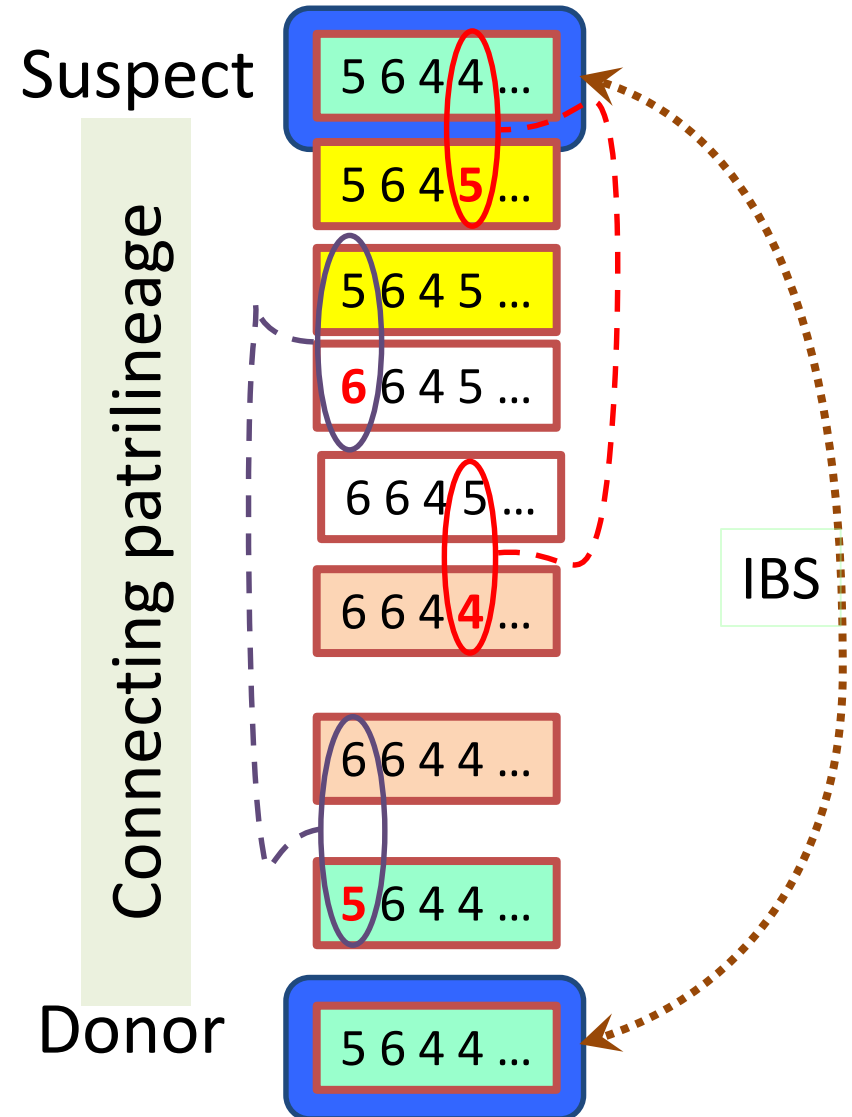
Y haplotypes match (IBS)
if and only if the connecting
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for some $n \in 0, 1, 2, \dots$.



Y haplotype match condition

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



Y match calculation #1 (primitive)

`matchpr ← n ConvMut (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

`ways ← n × .!gen, gen - n`

⌘ ways to position mutations

`pr ← (mu ÷ 2) (1 - mu) × . * (2 × n) (gen - 2 × n)`

⌘ probability of each way

`matchpr ← ways × pr`

⌘ total probability of all ways

Y match calculation #2

$\Pr(\text{match})$

given **g** generations of separation

IO ← 0

maxn ← 11 A 10 mutation pairs per locus is plenty

gens ← 1 to **ngen** + 1 A **gens** ← 0, 1, ..., **ngen** generations separation

ibS ← (1 to **maxn**) × **ConvMut gens** × **mu** A **p** ↔ **maxn ngen** (≠ **Yloci**)

A **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:

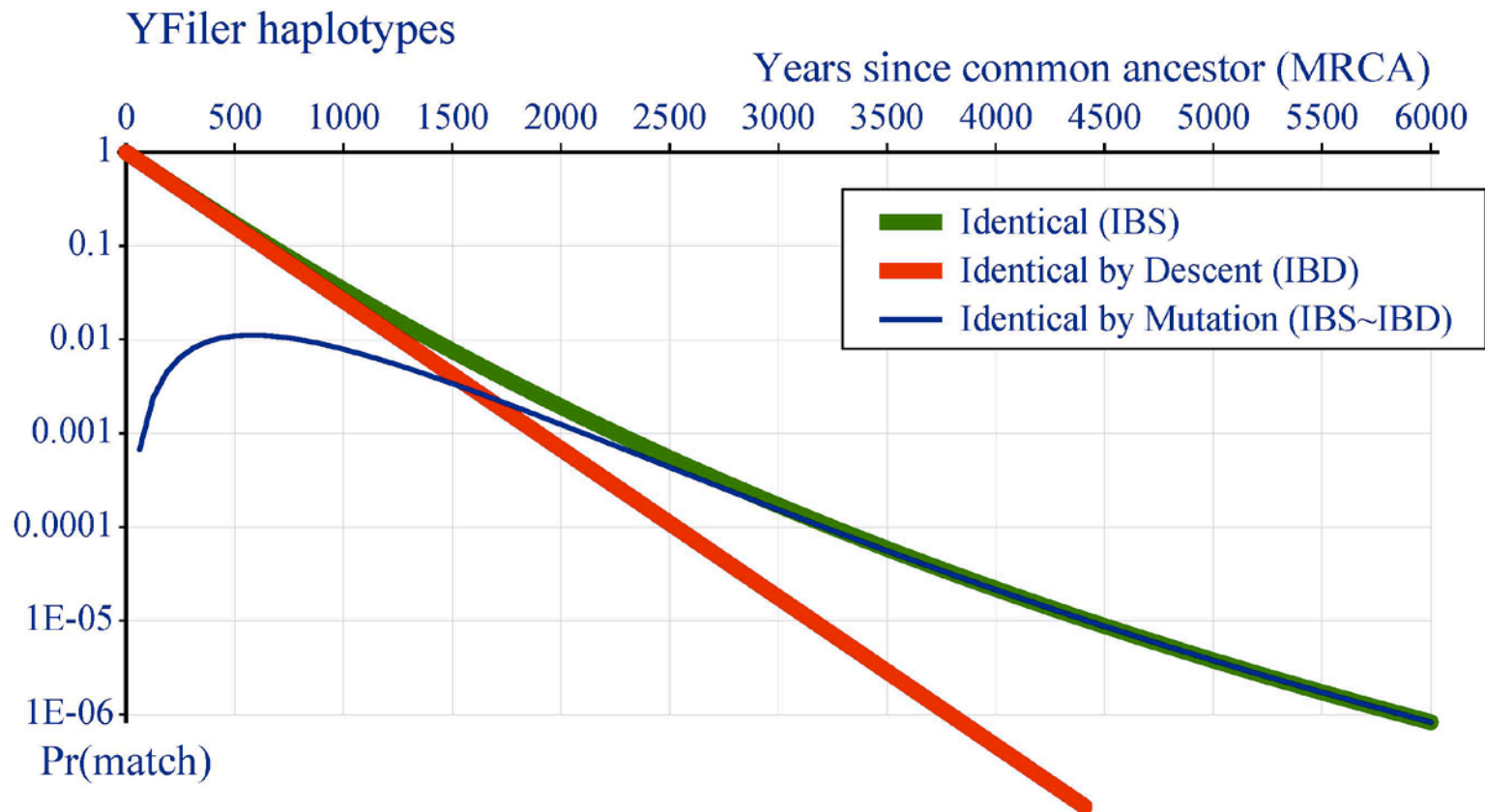
IBD ← **x** / **ibS**[0;;] A match all loci, no mutations

IBS ← **x** / **+/ibS** A match all loci, allow mutations

sIBS ← **IBS** - **IBD** A strictly IBS (some mutations)

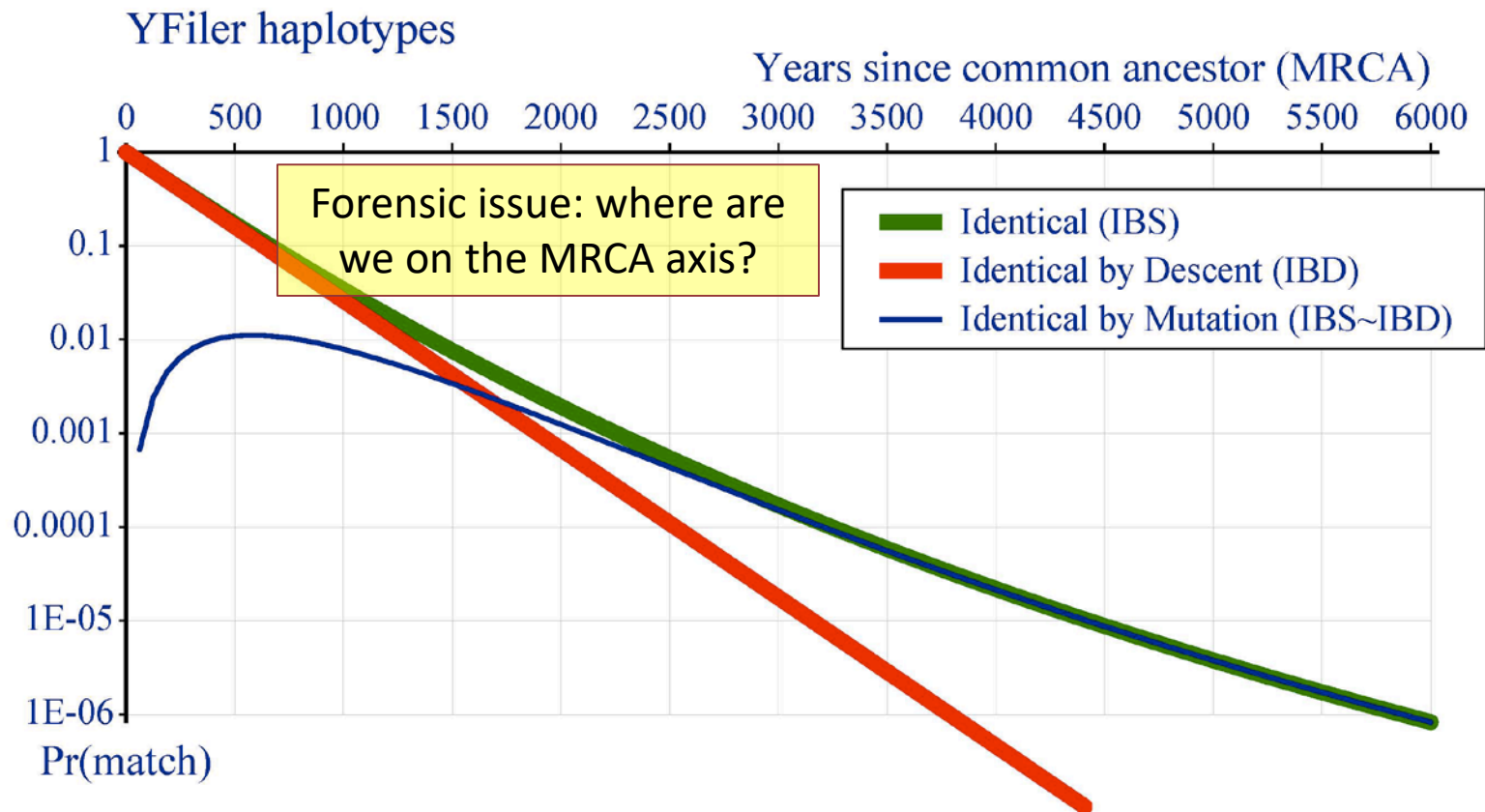
IBD dominates Yfiler matching

Identical by Descent vs by Mutation



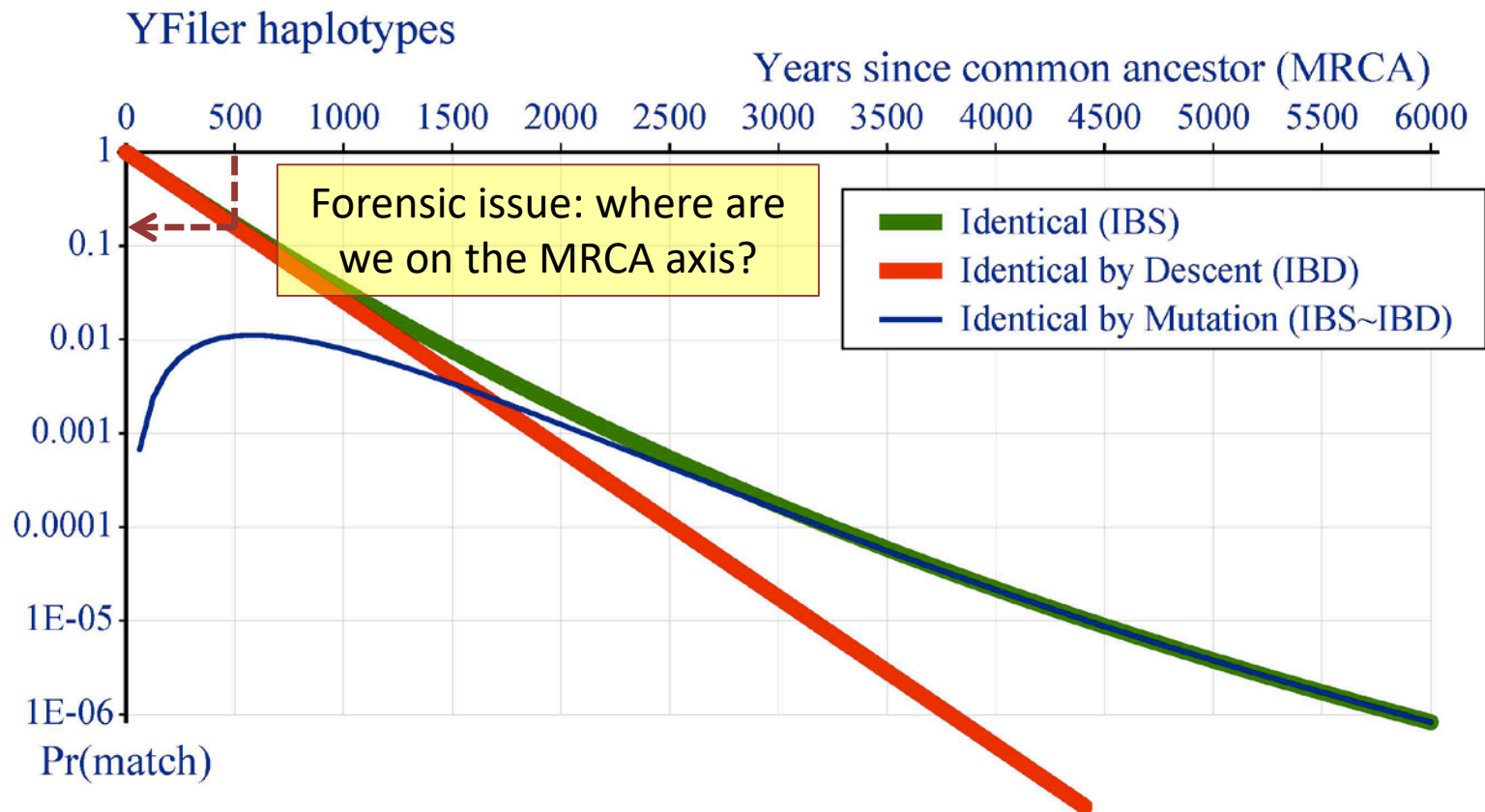
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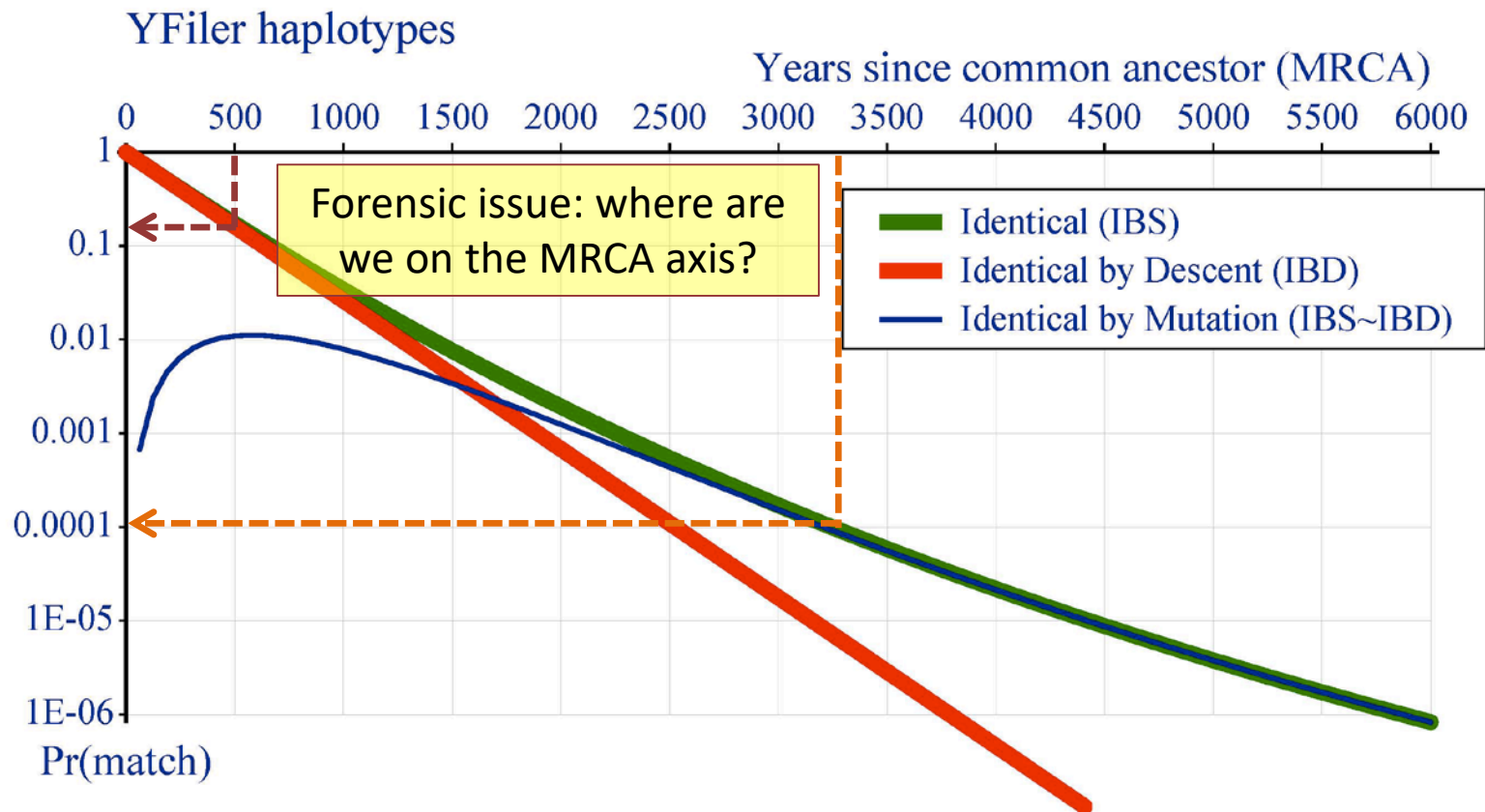
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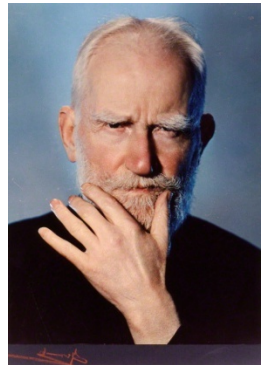
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Common boilerplate from forensic lab DNA matching report

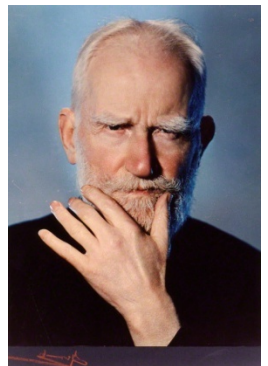
The longer I live the more I see that I am never wrong about anything, and that all the pains that I have so humbly taken to verify my notions have only wasted my time.



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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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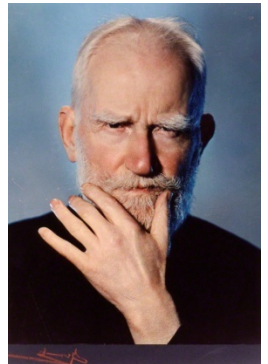


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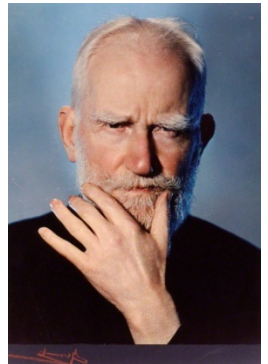


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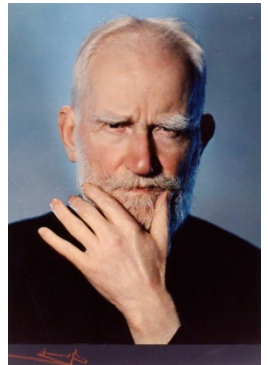


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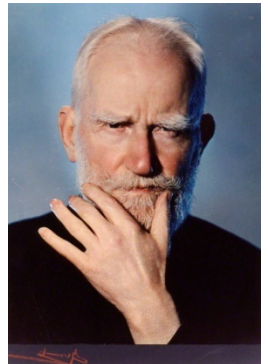


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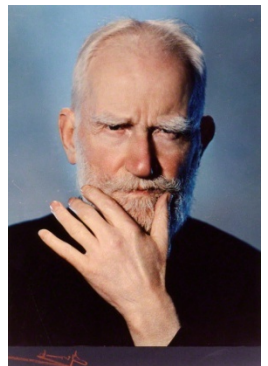


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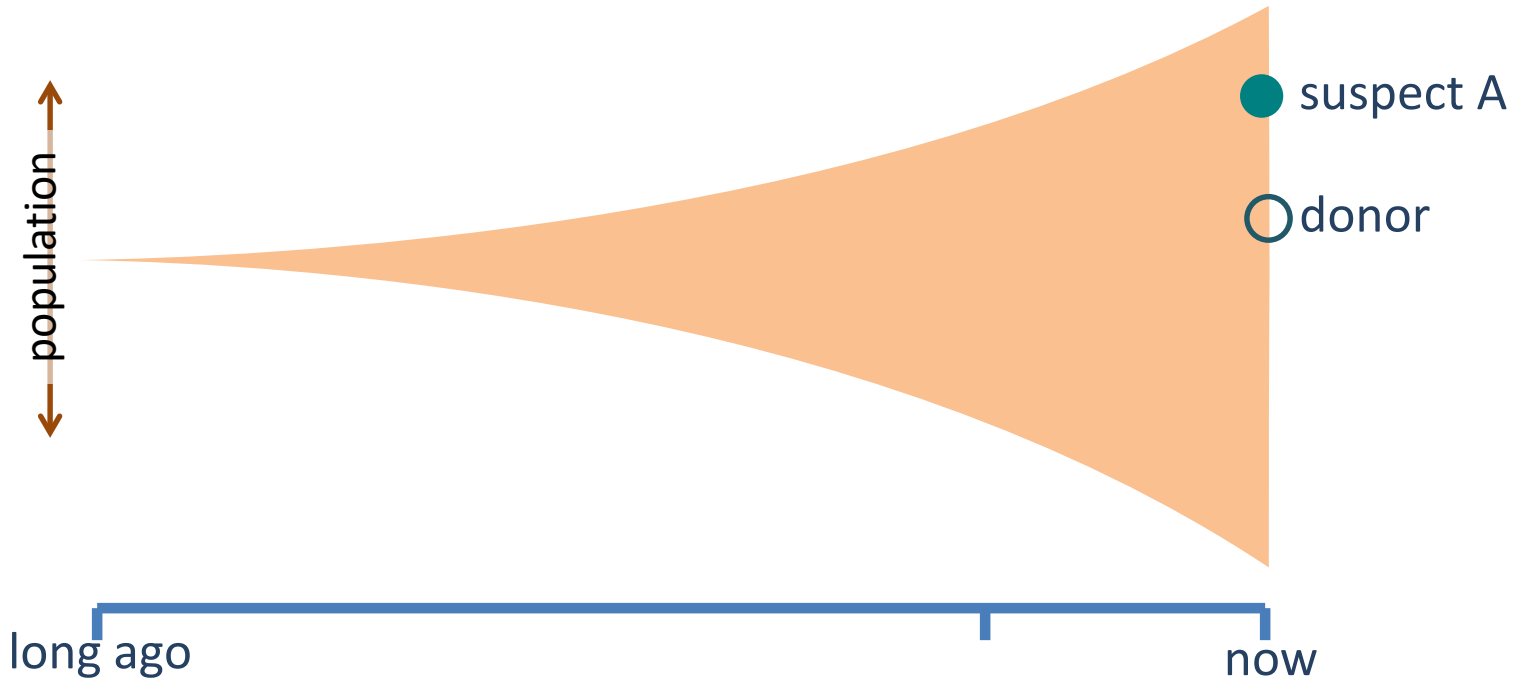
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 - really stupid

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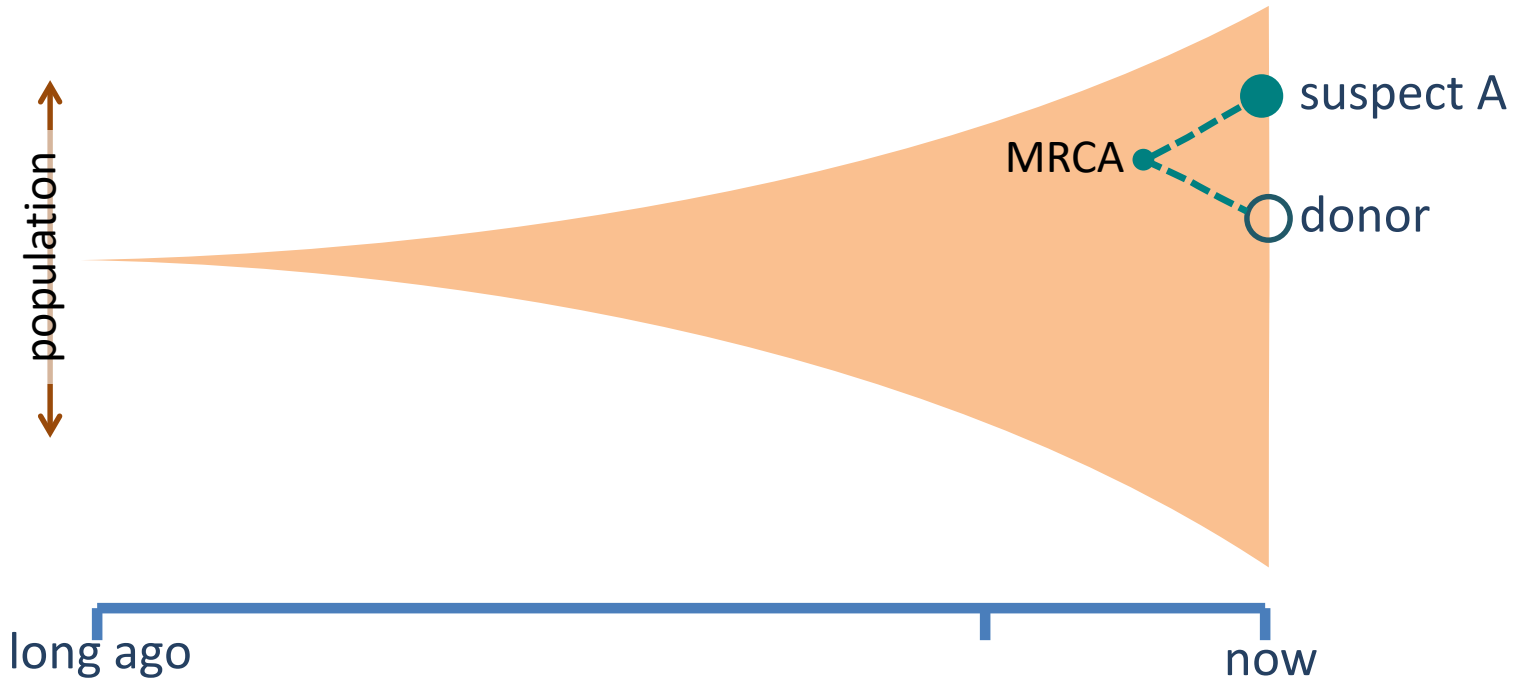
Modeling a population

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



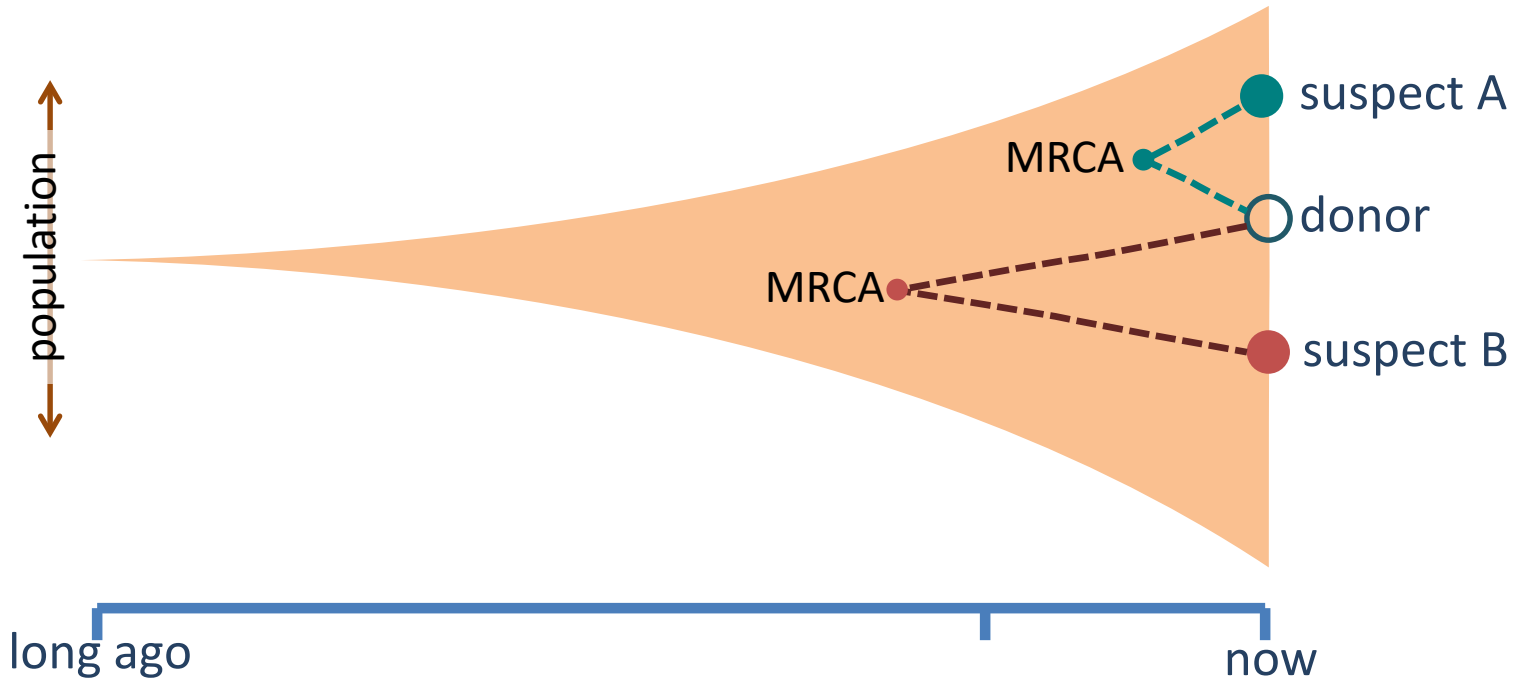
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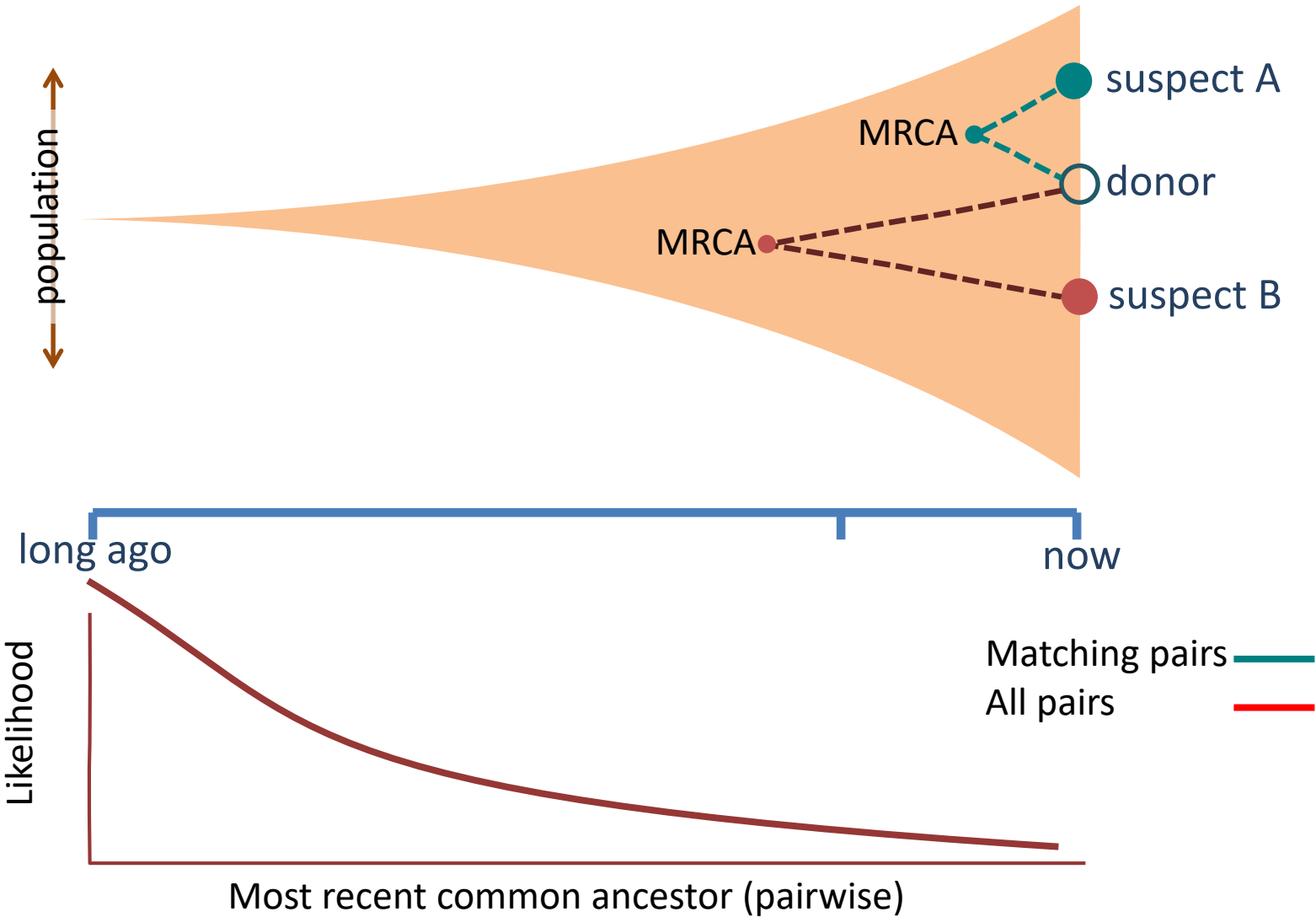
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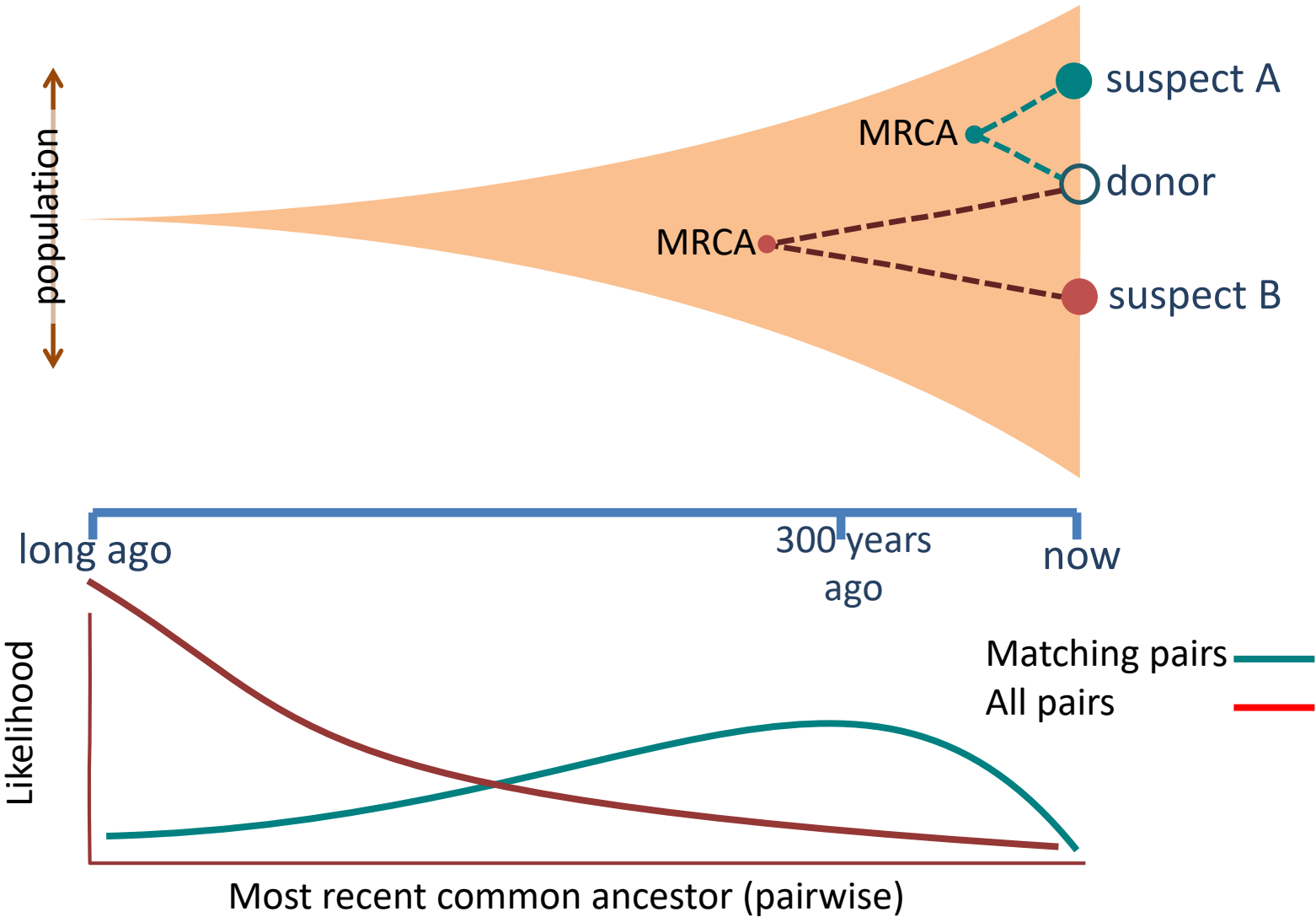
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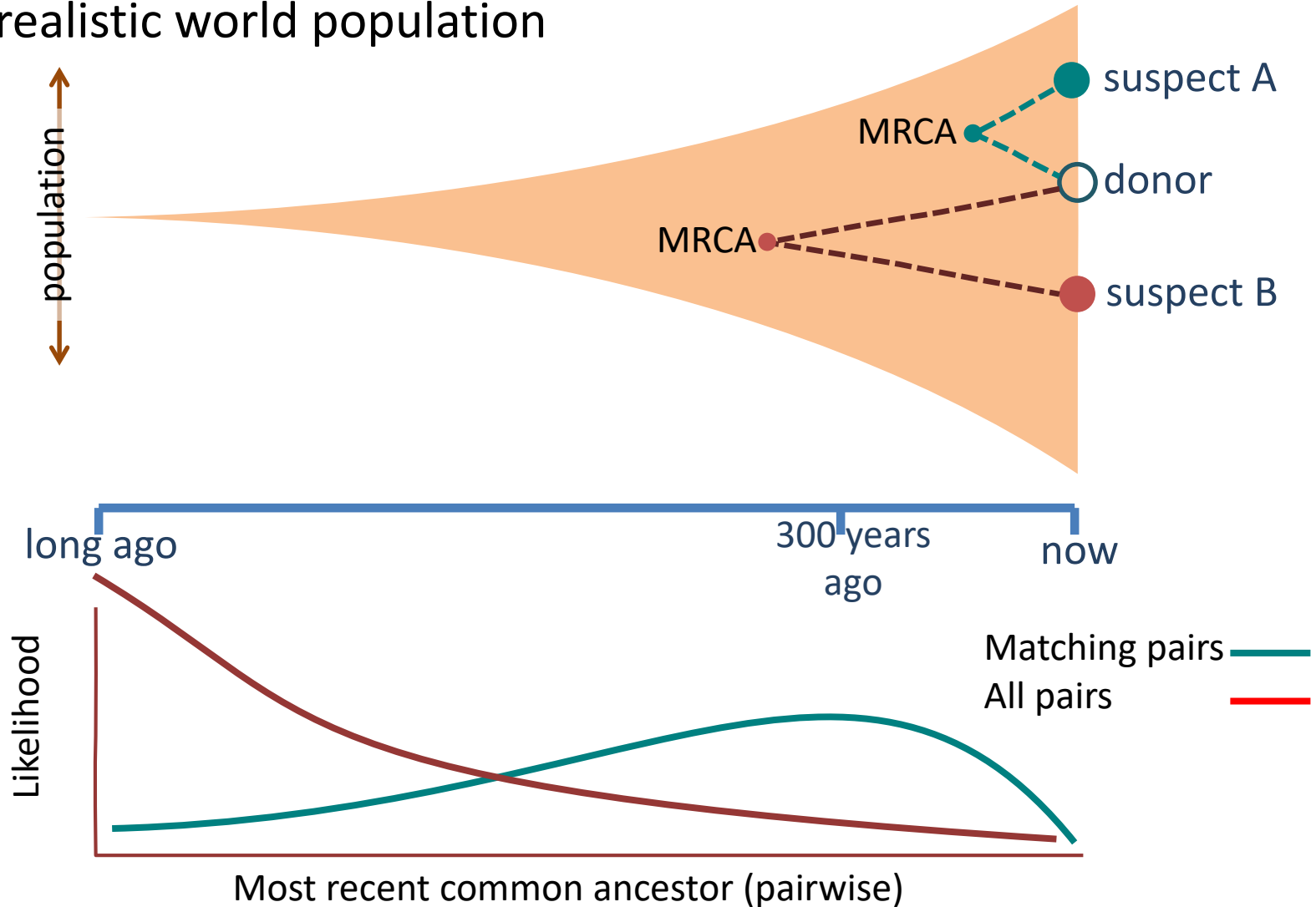
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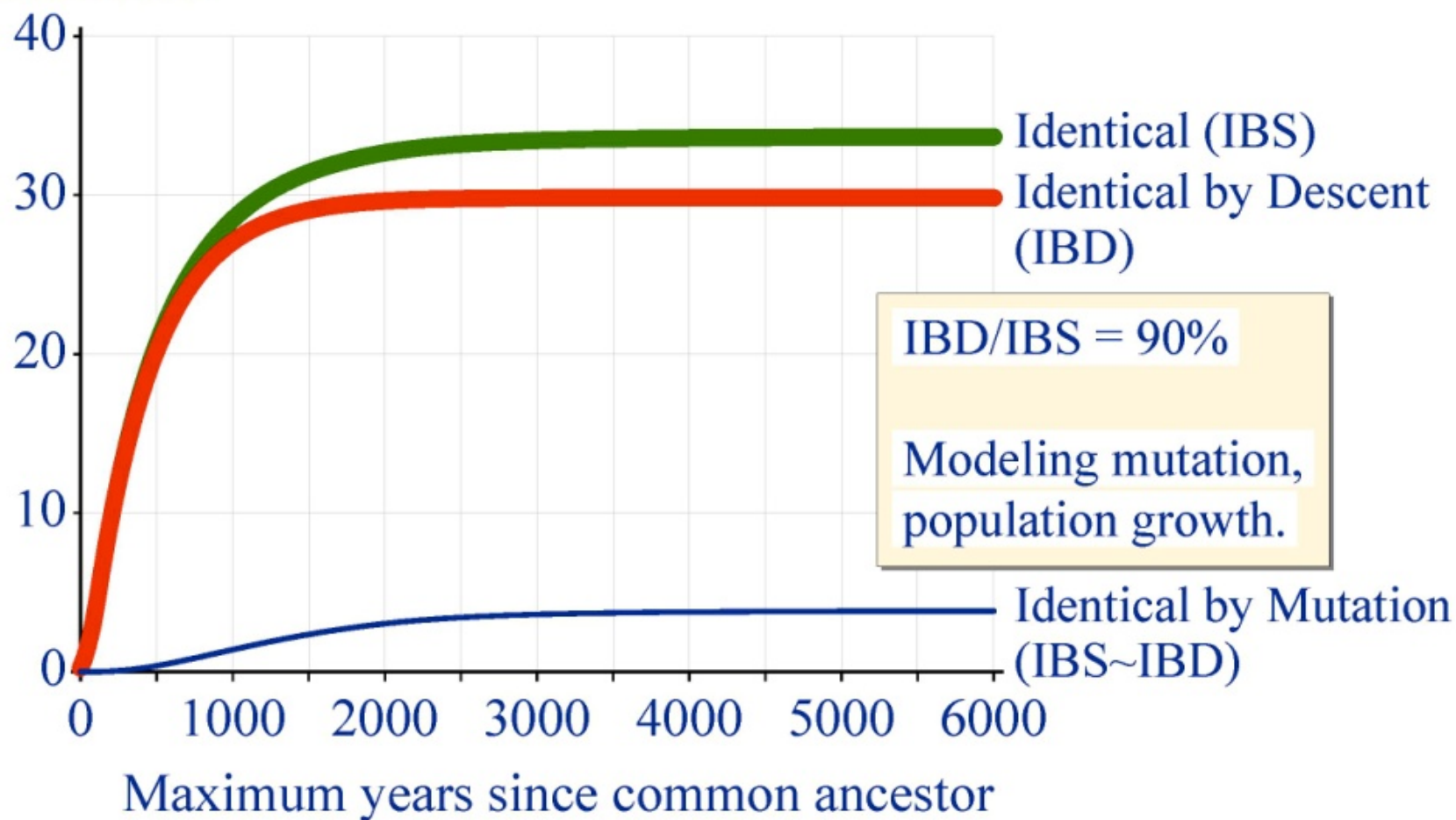
Modeling a population

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



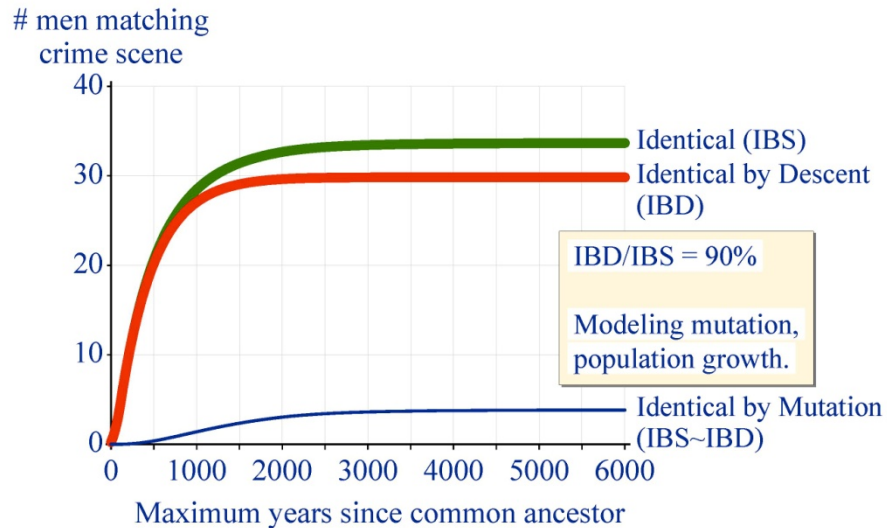
Identity by Descent vs by Mutation, YFiler

men matching
crime scene



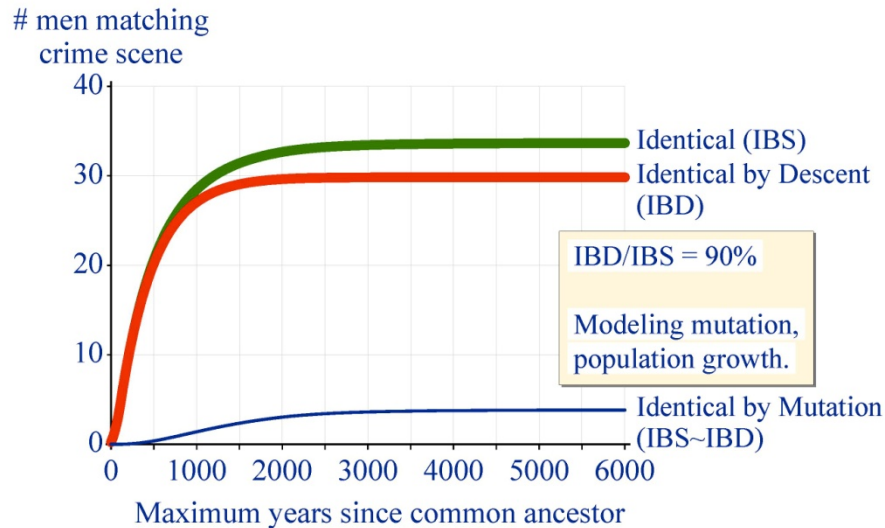
Haplotypes are NOT just super-polymorphic loci

Identity by Descent vs by Mutation, YFiler



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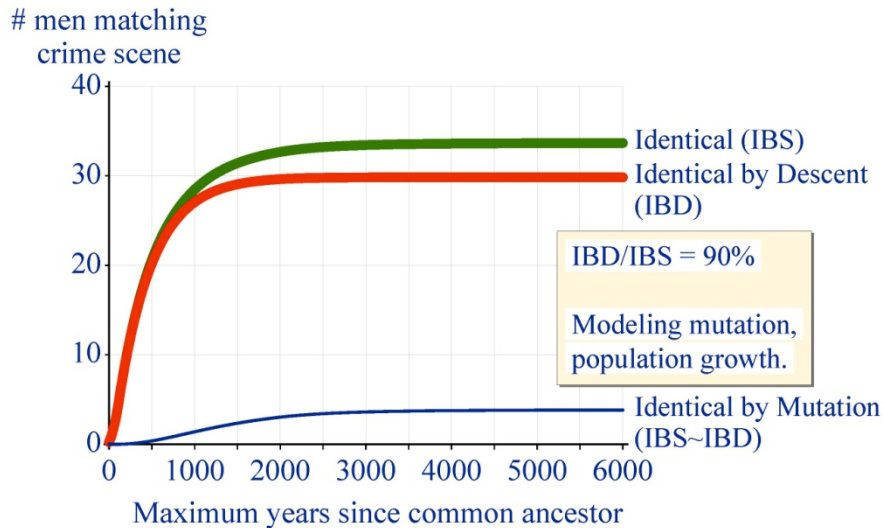
Haplotype

matching is dominated by

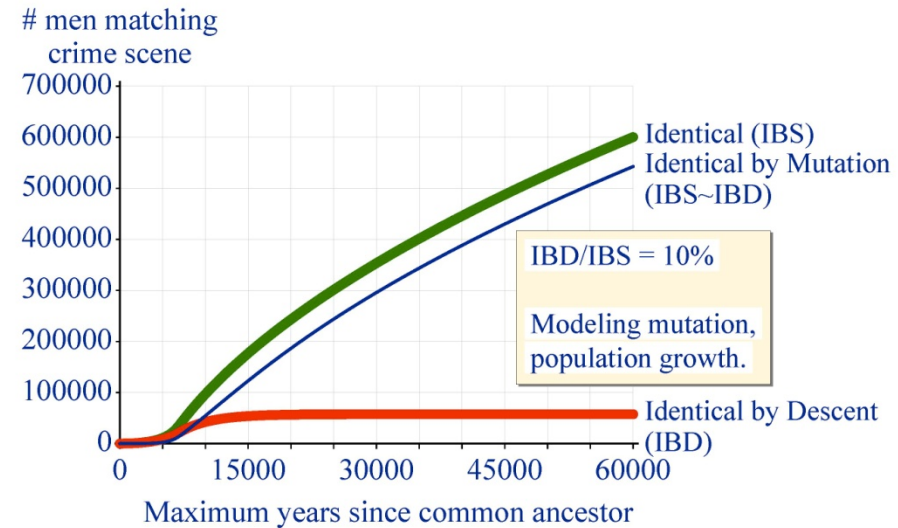
IBD – patrilineal link without intervening mutation.

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

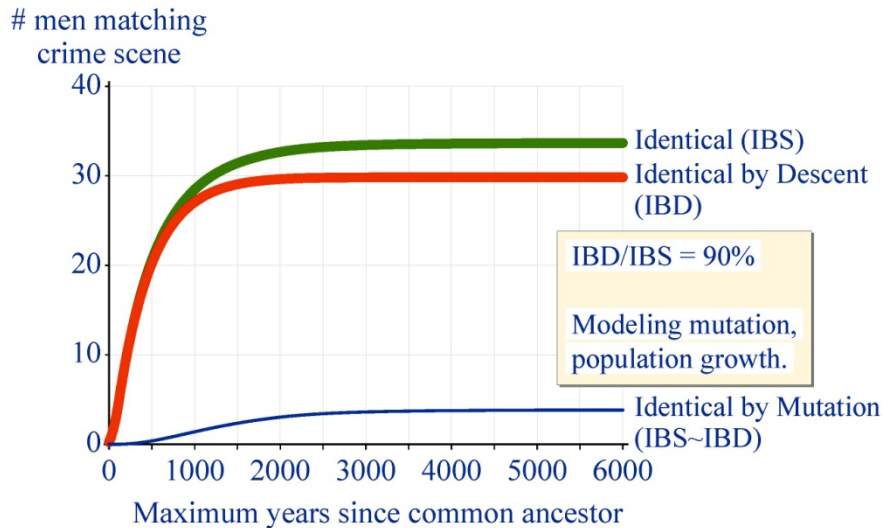
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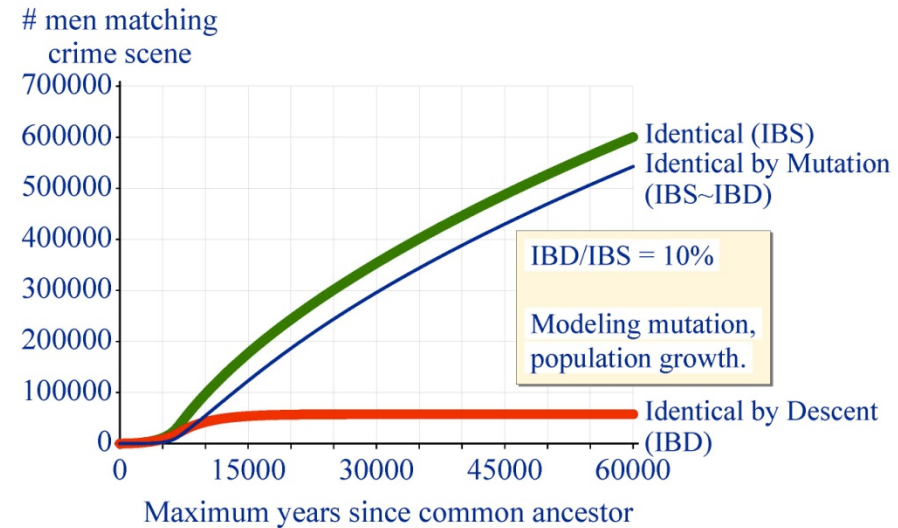
coincidence.

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

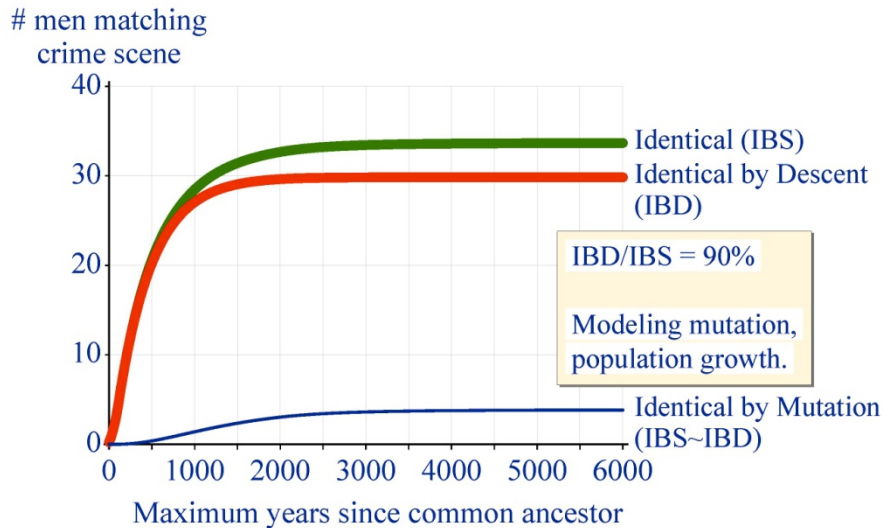
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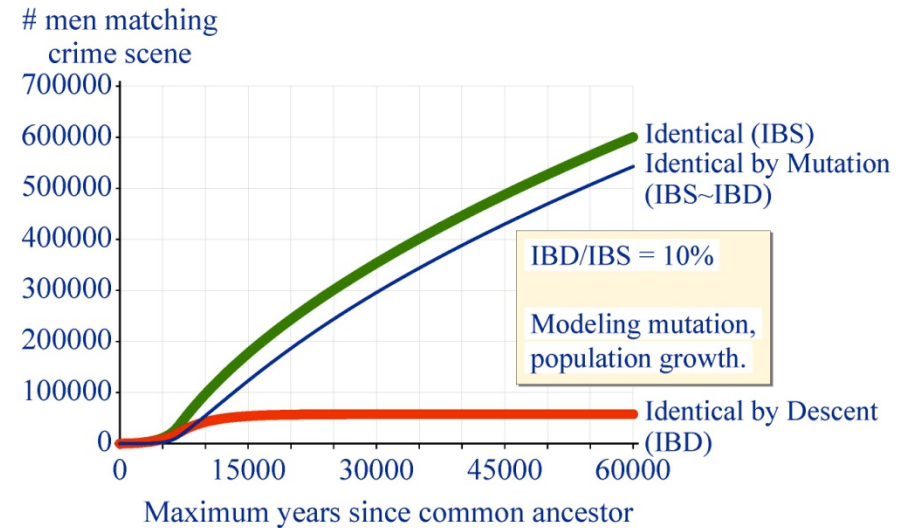
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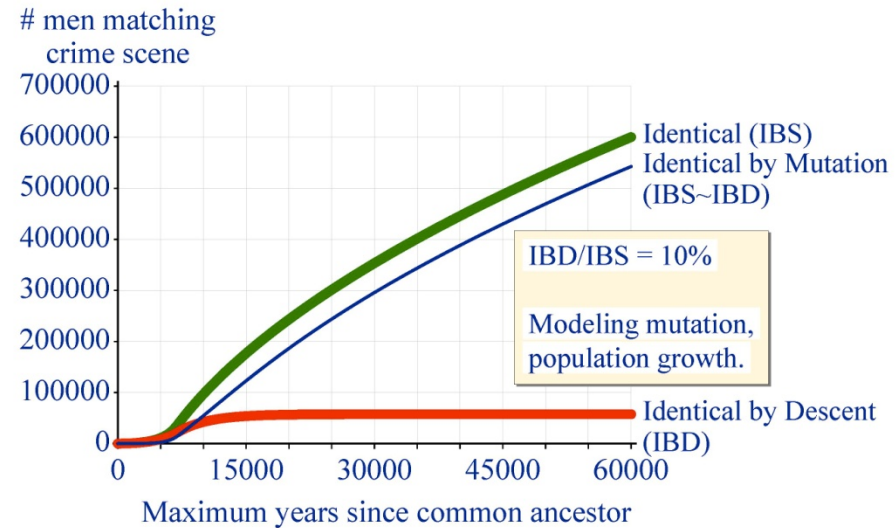
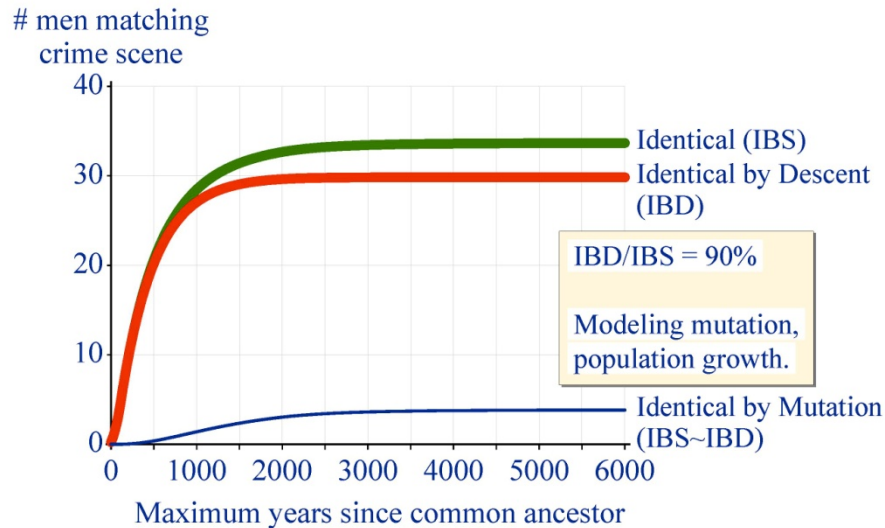
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Moral –

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

Identity by Descent vs by Mutation, YFiler



Haplotype

STR locus

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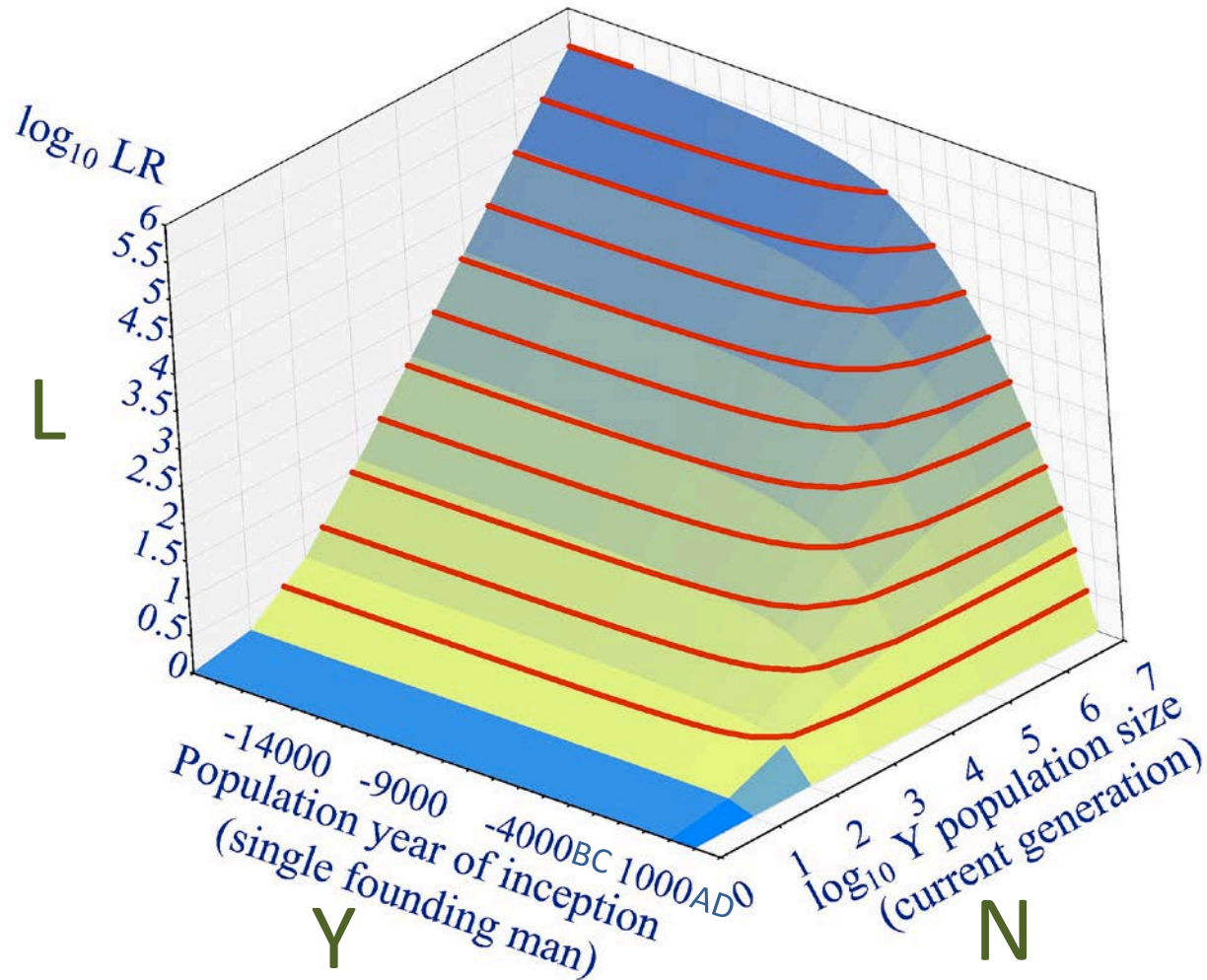
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Evidential strength of Y haplotype match

IBS matching probability

YFiler

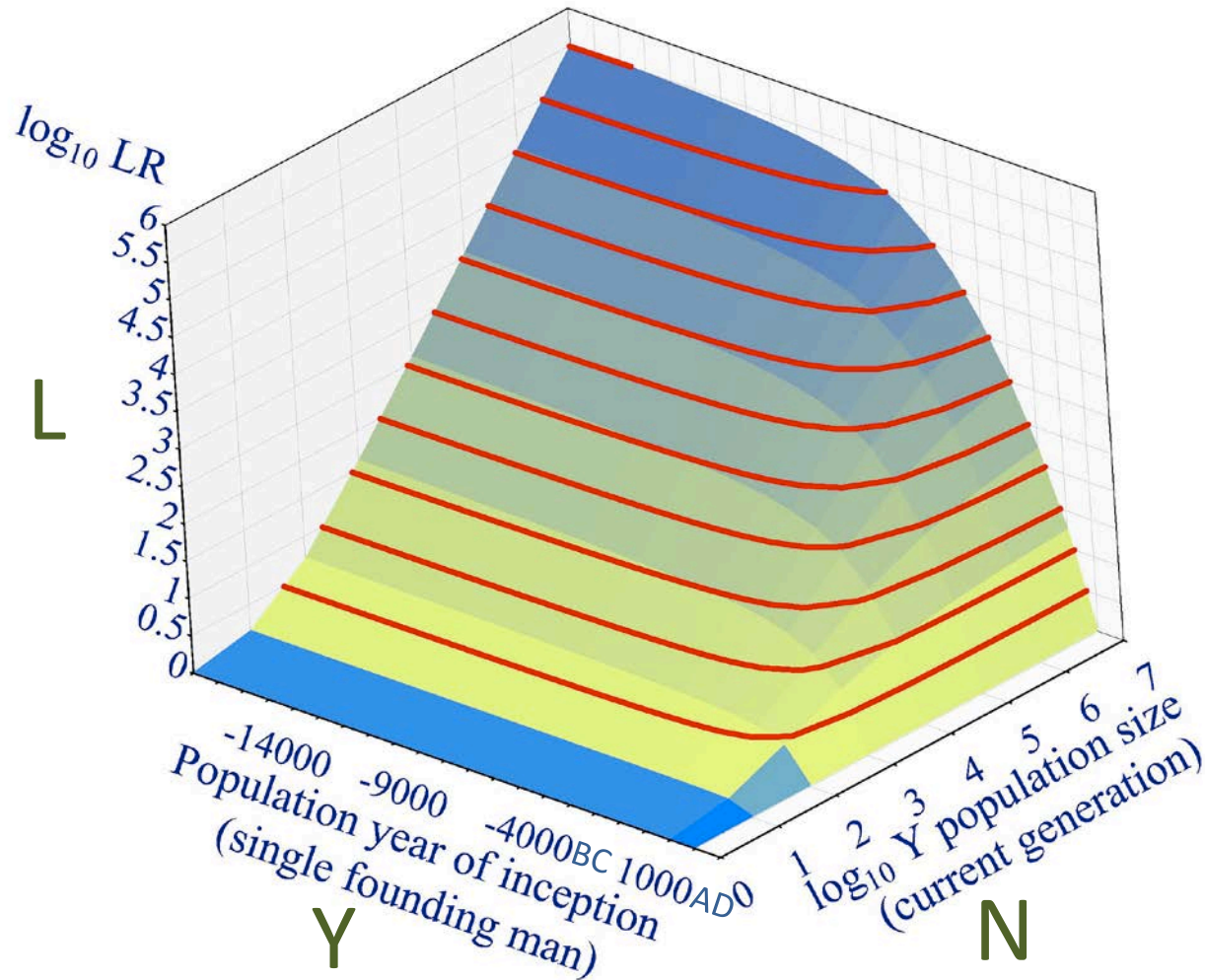


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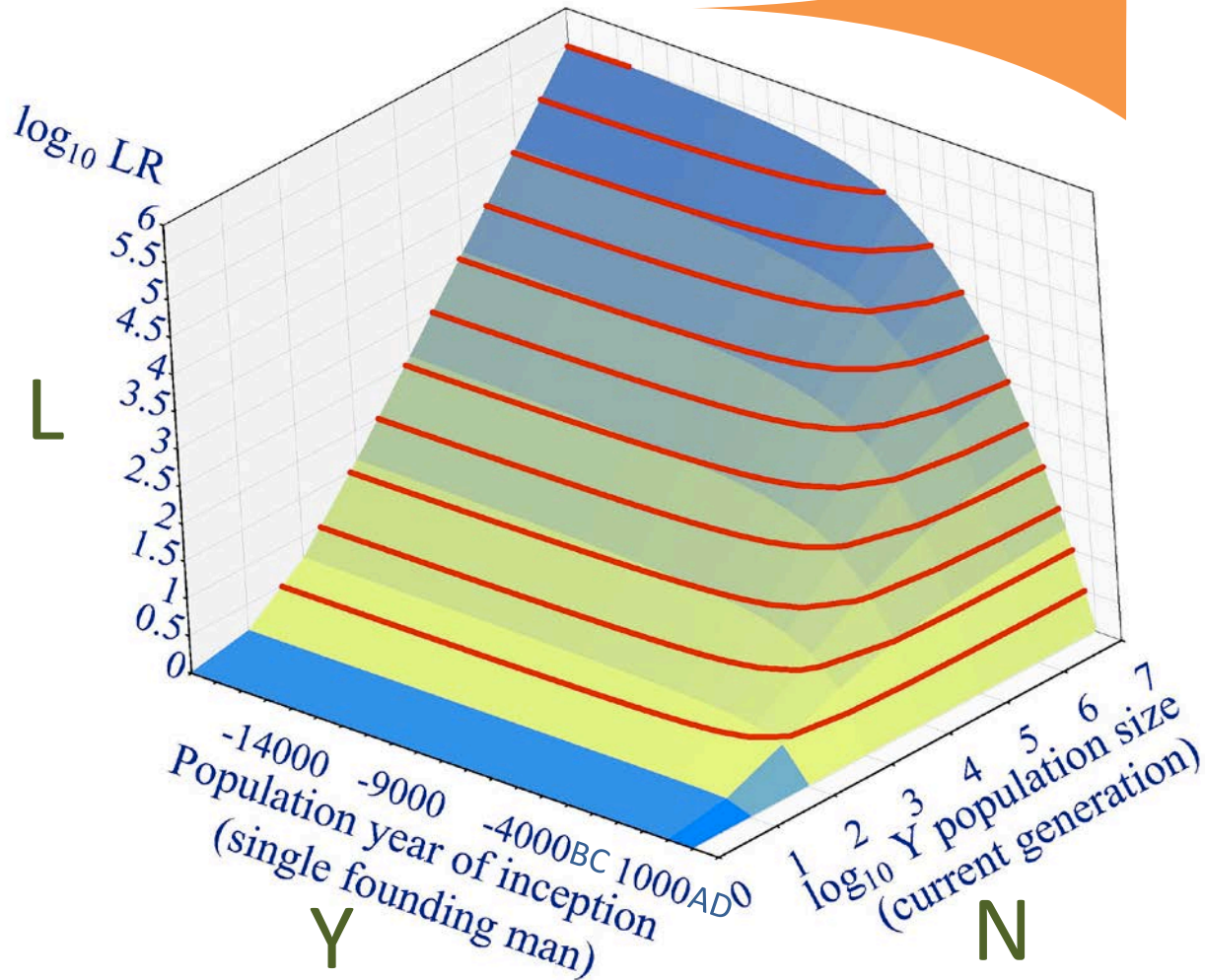
Model: constant
population growth rate



Evidential strength of Y haplotype match

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YFiler

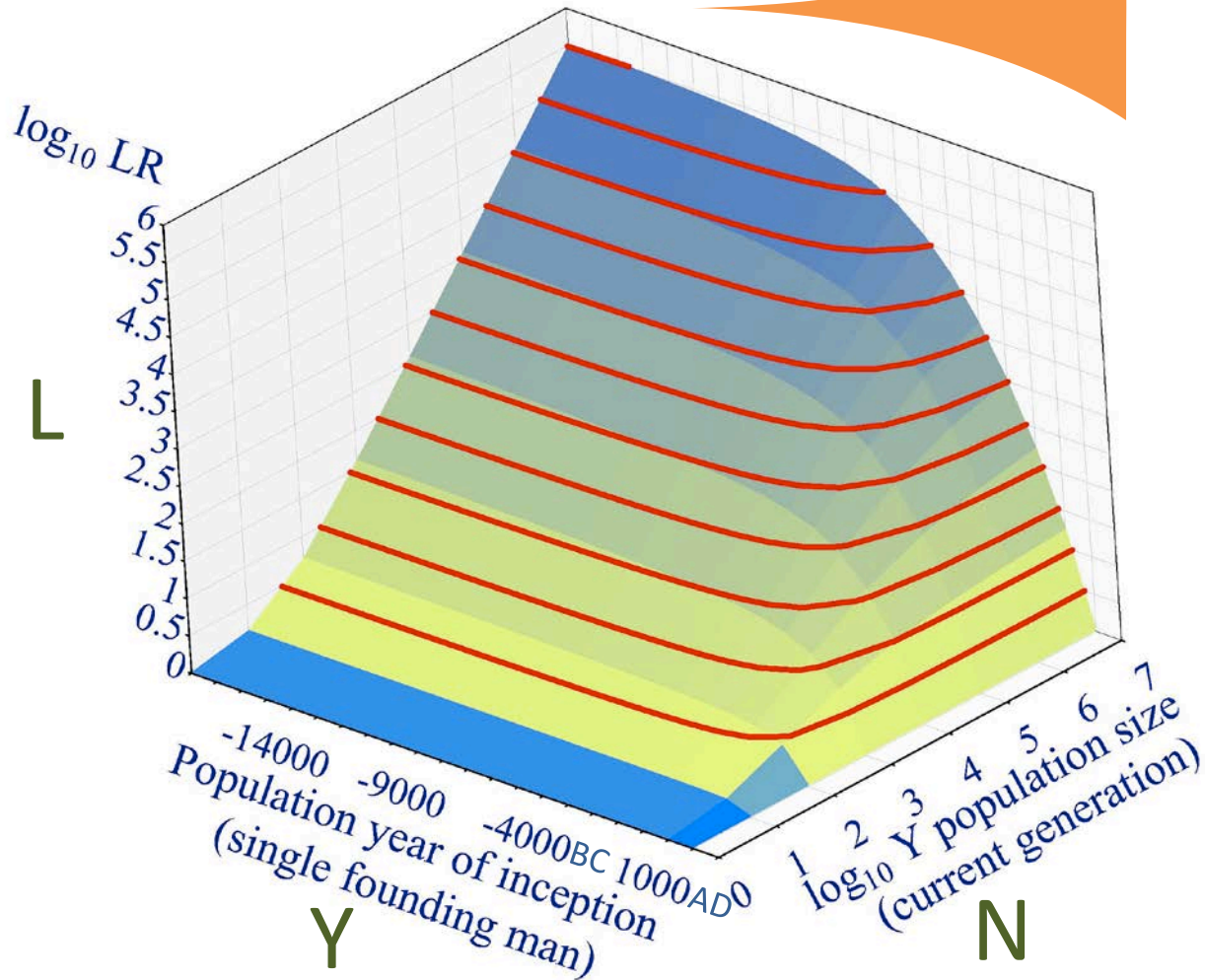


Model: constant population growth rate from 1 founding man to N men today..

Evidential strength of Y haplotype match

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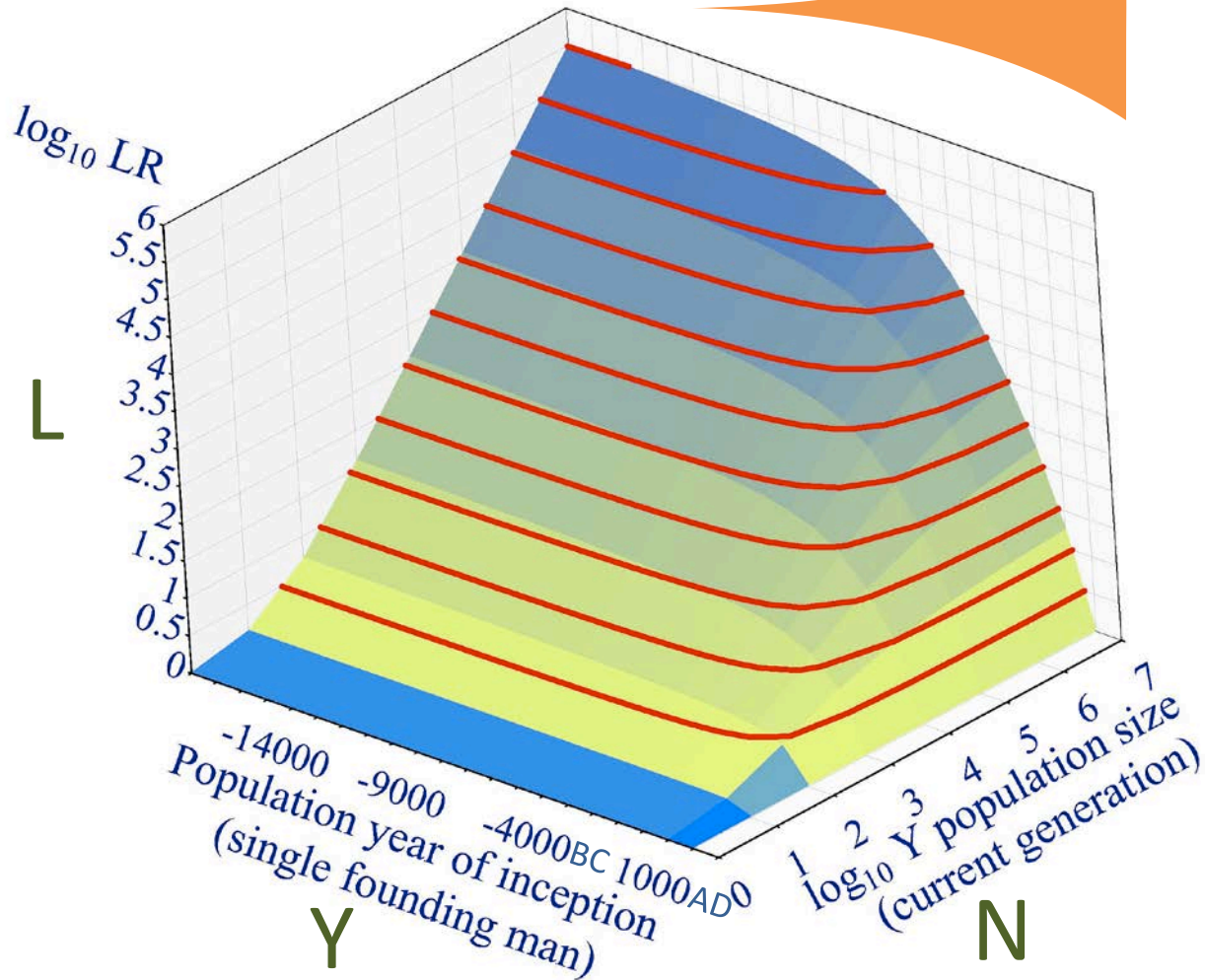
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Matching evidence

Evidential strength of Y haplotype match

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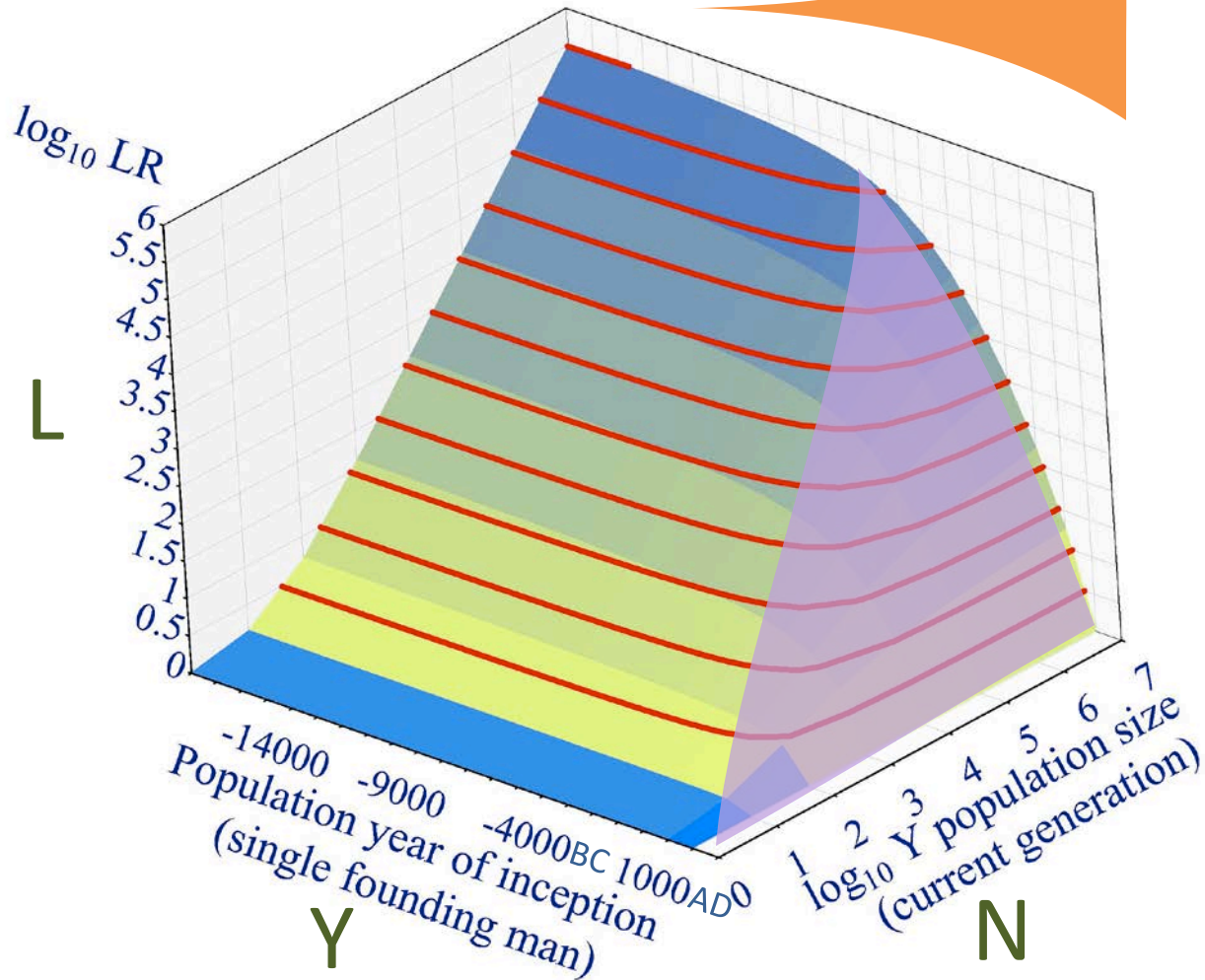
Matching evidence

- increases with population *size*.

Evidential strength of Y haplotype match

IBS matching probability

YFiler

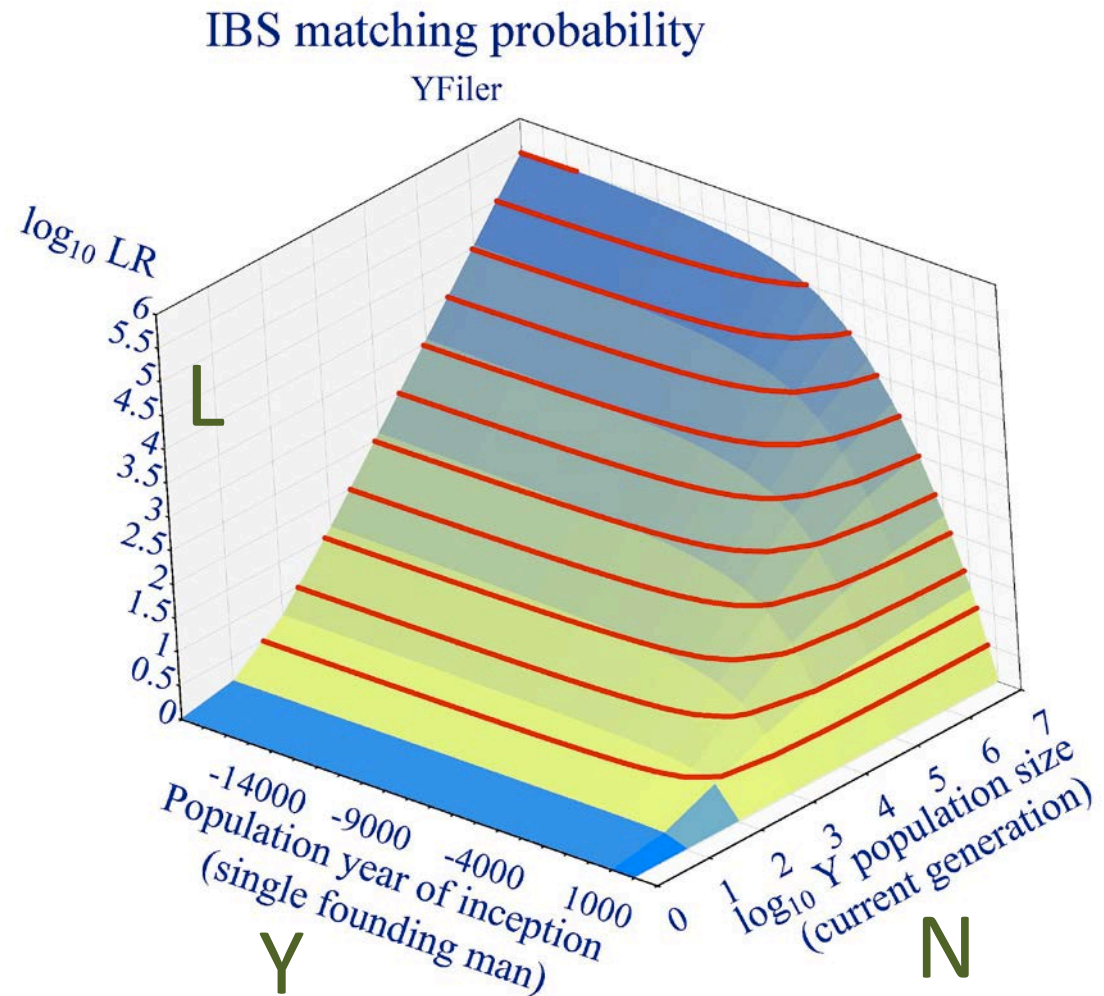


Model: constant population growth rate from 1 founding man to **N** men today..

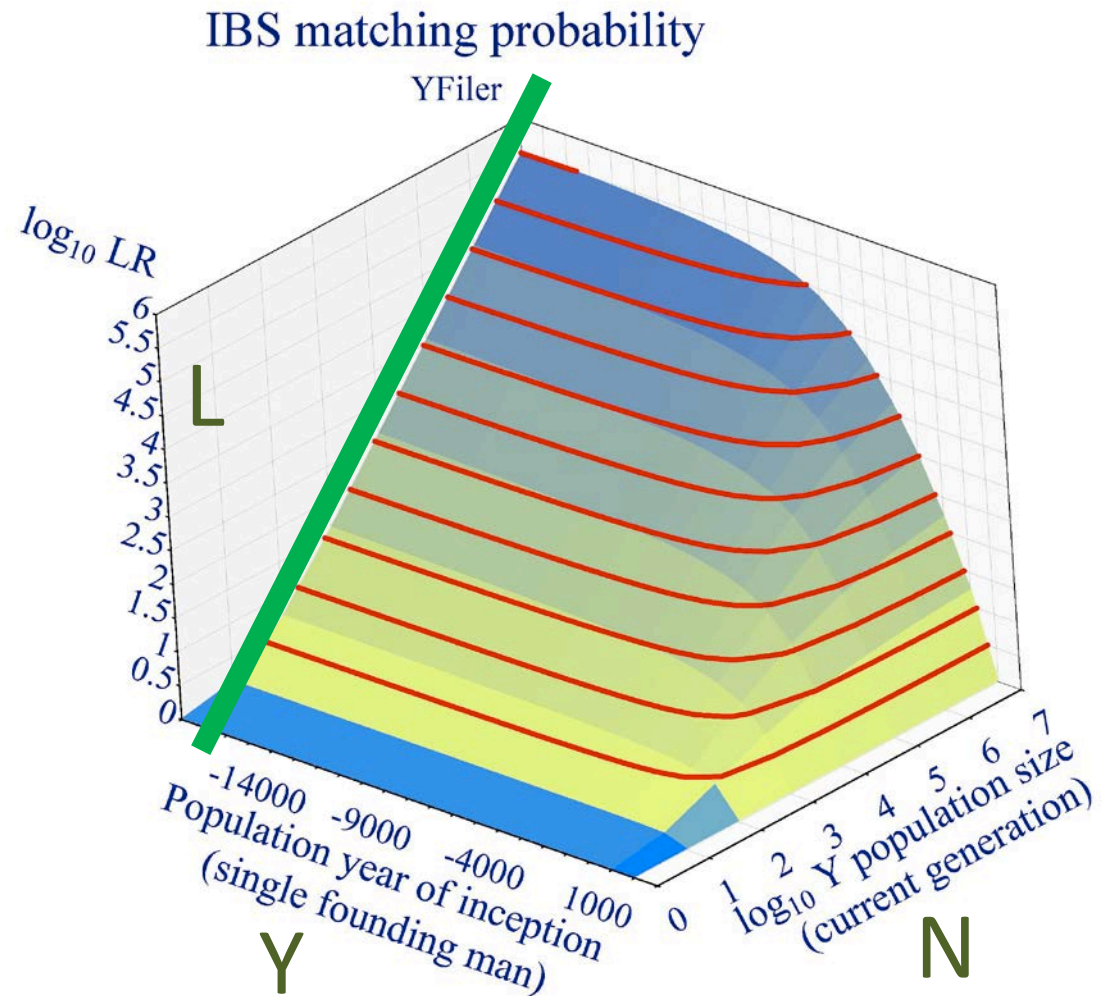
Matching evidence

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

Haplotype cohort size vs populations size

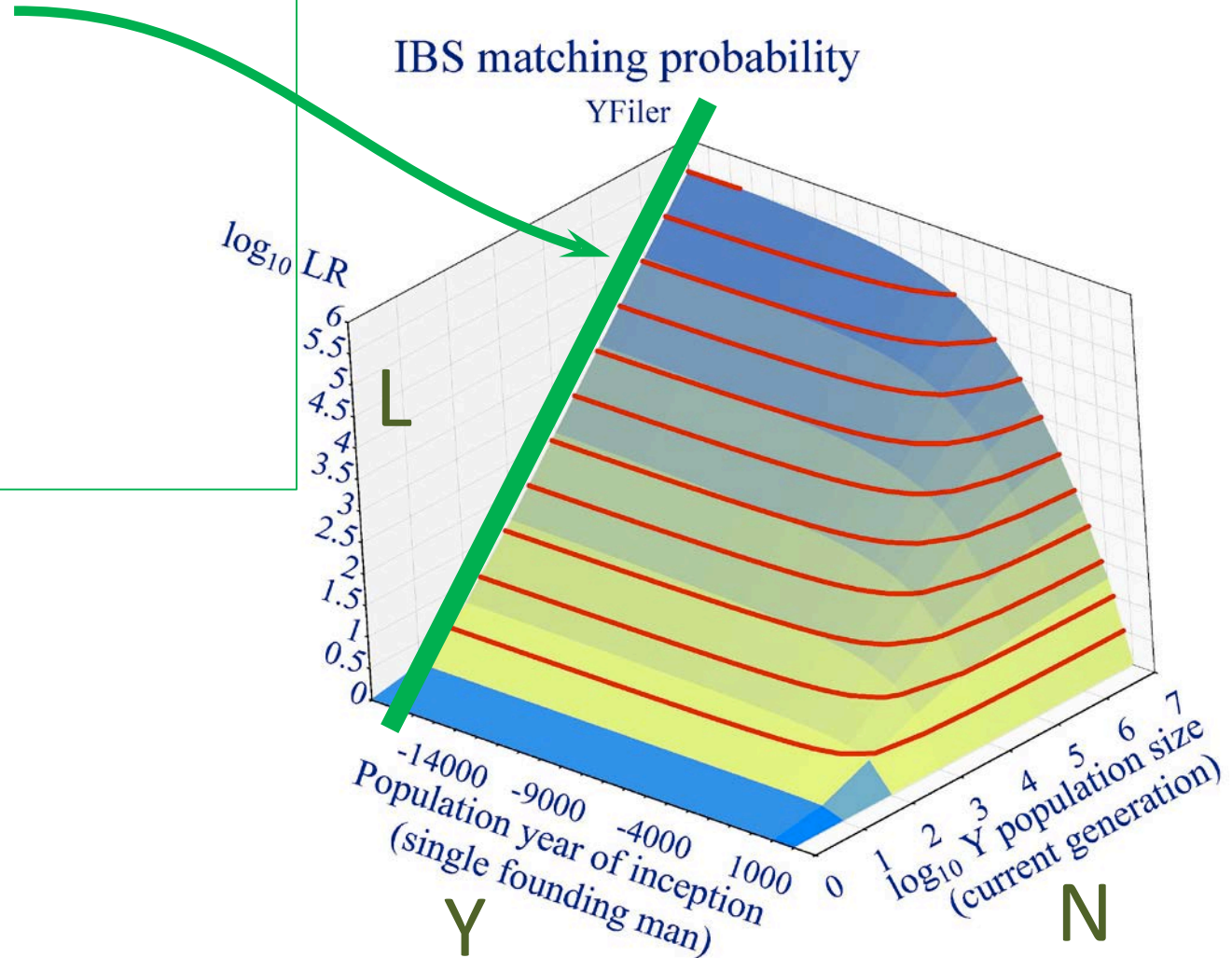


Haplotype cohort size vs populations size



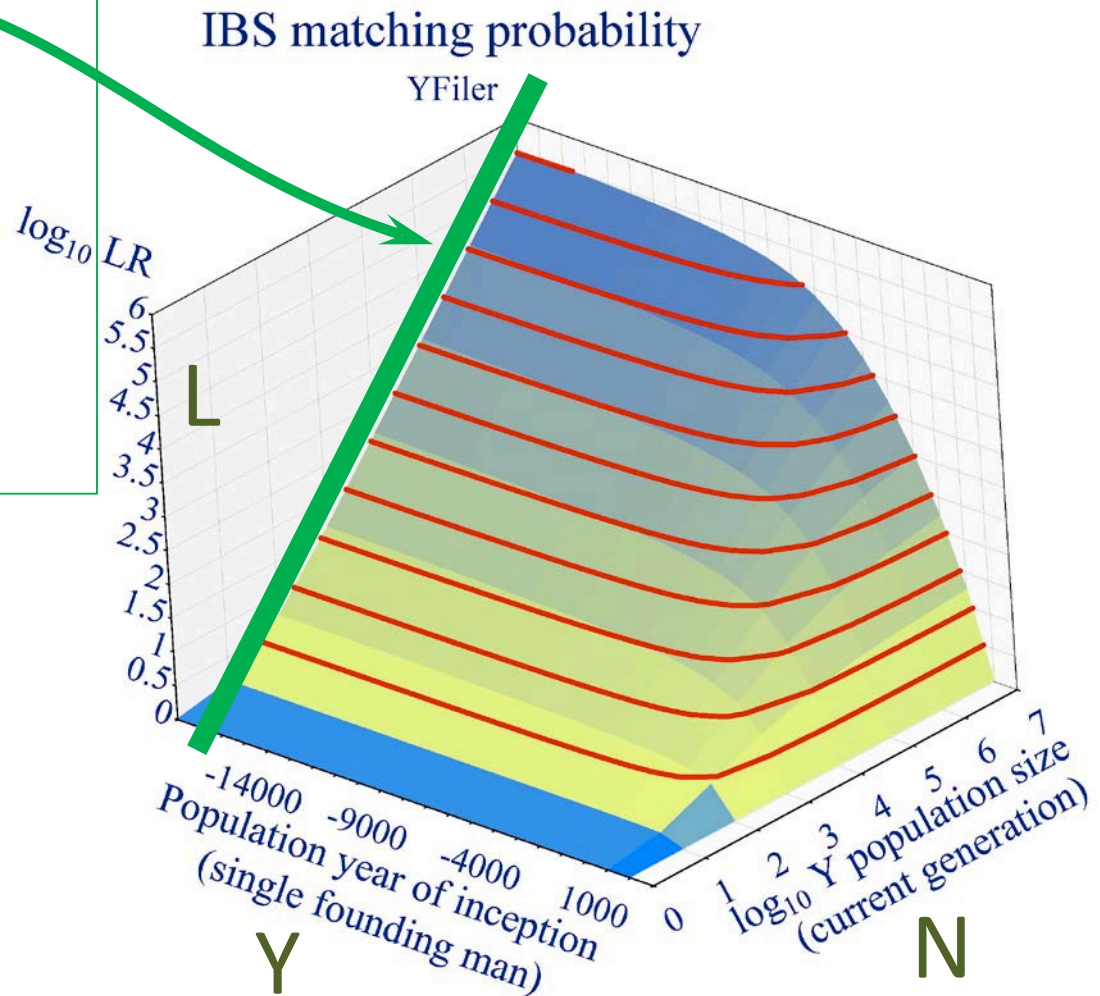
Haplotype cohort size vs populations size

- Straight? Yes



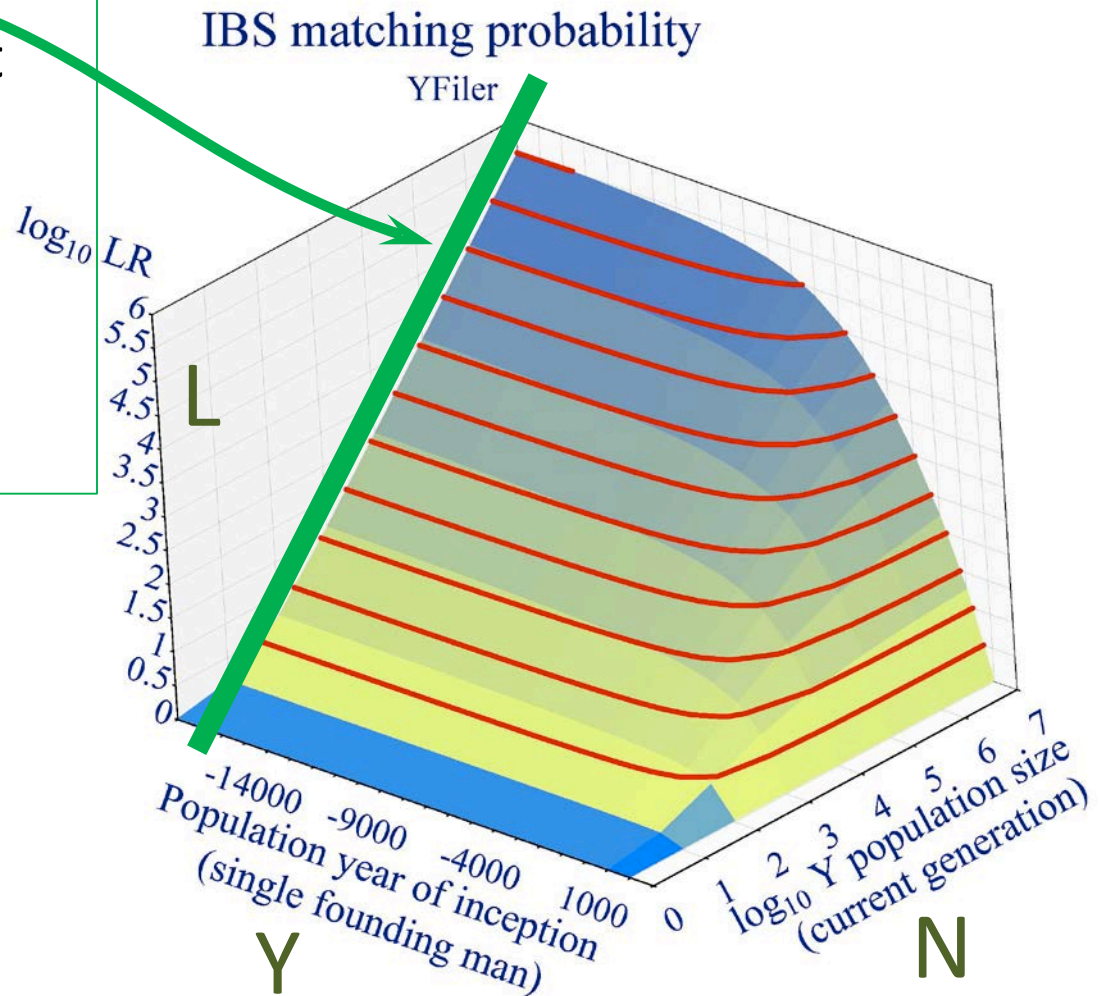
Haplotype cohort size vs populations size

- Straight? Yes
- & 45°? Close



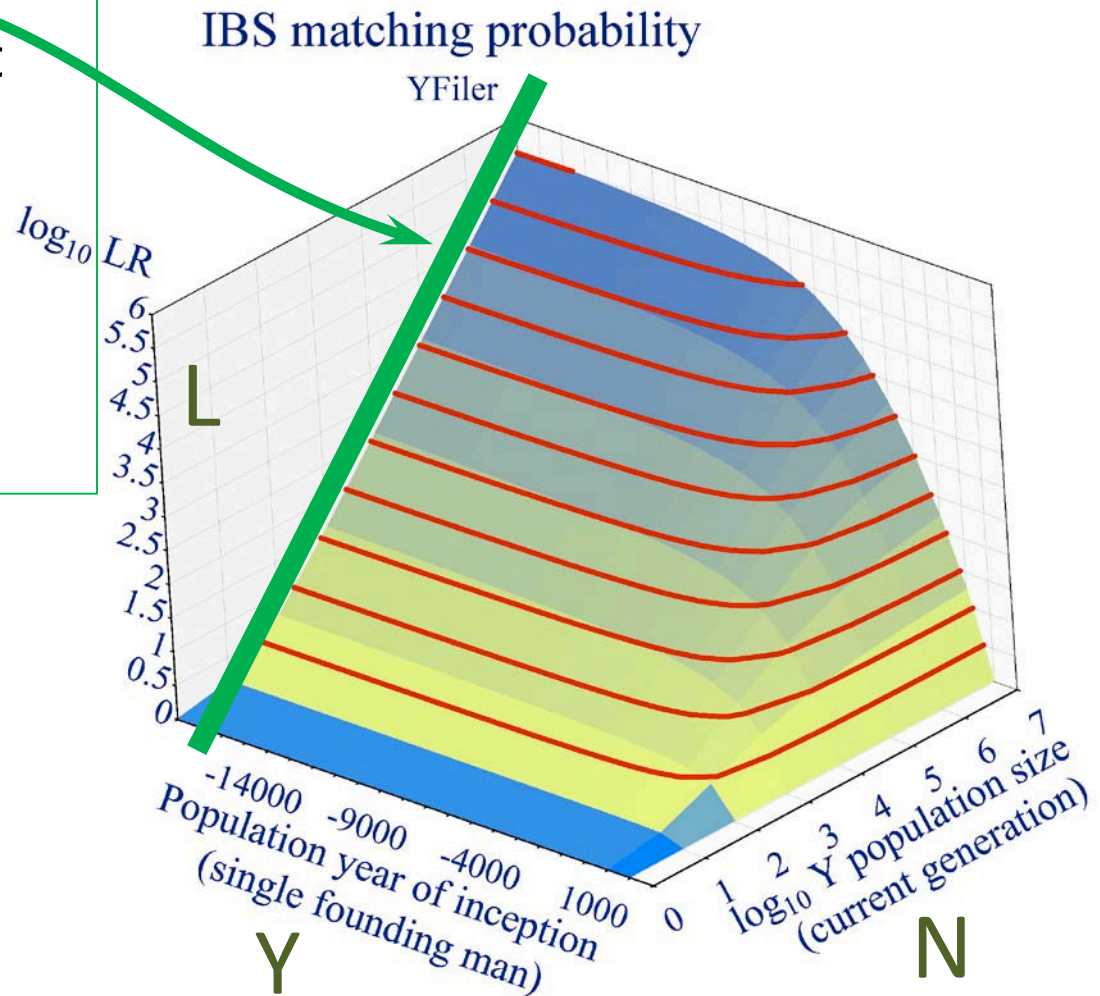
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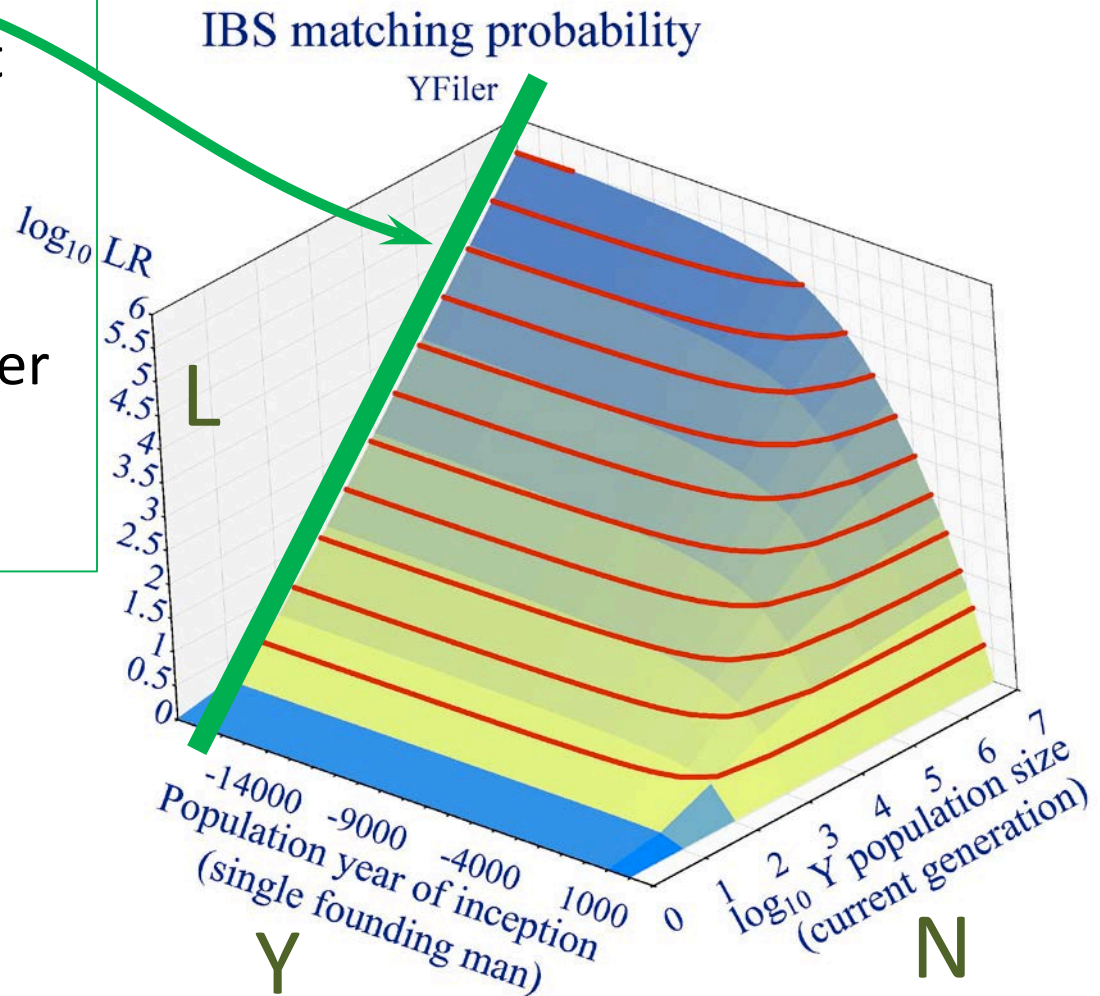
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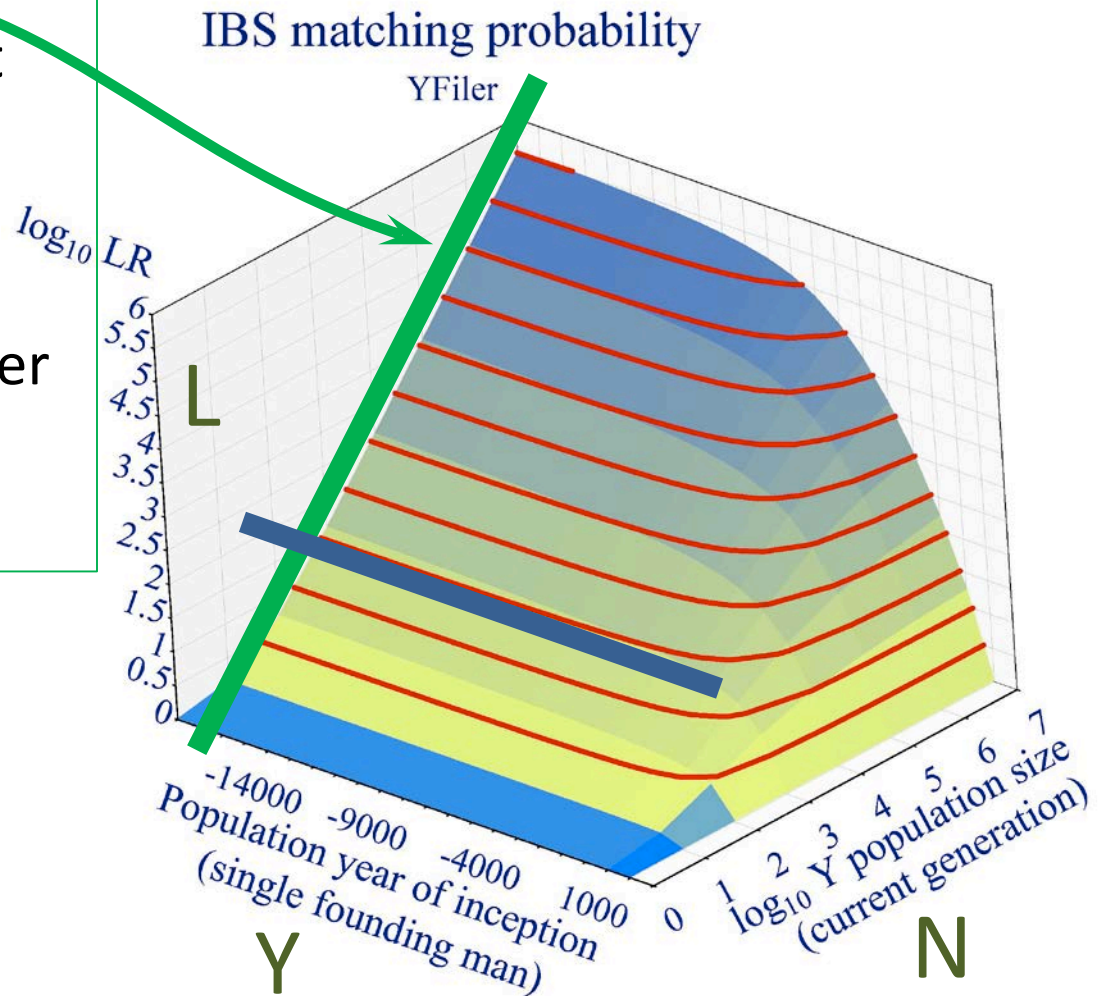
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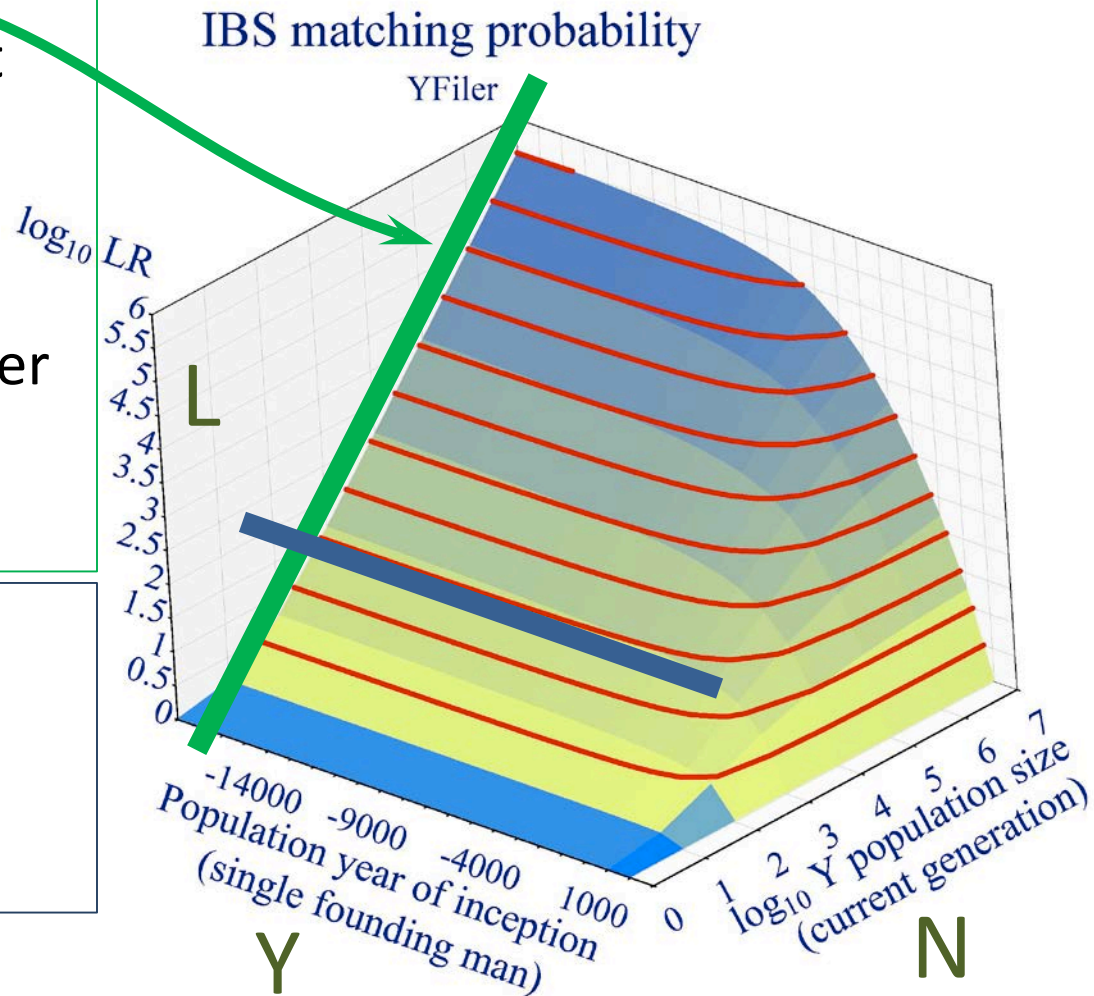
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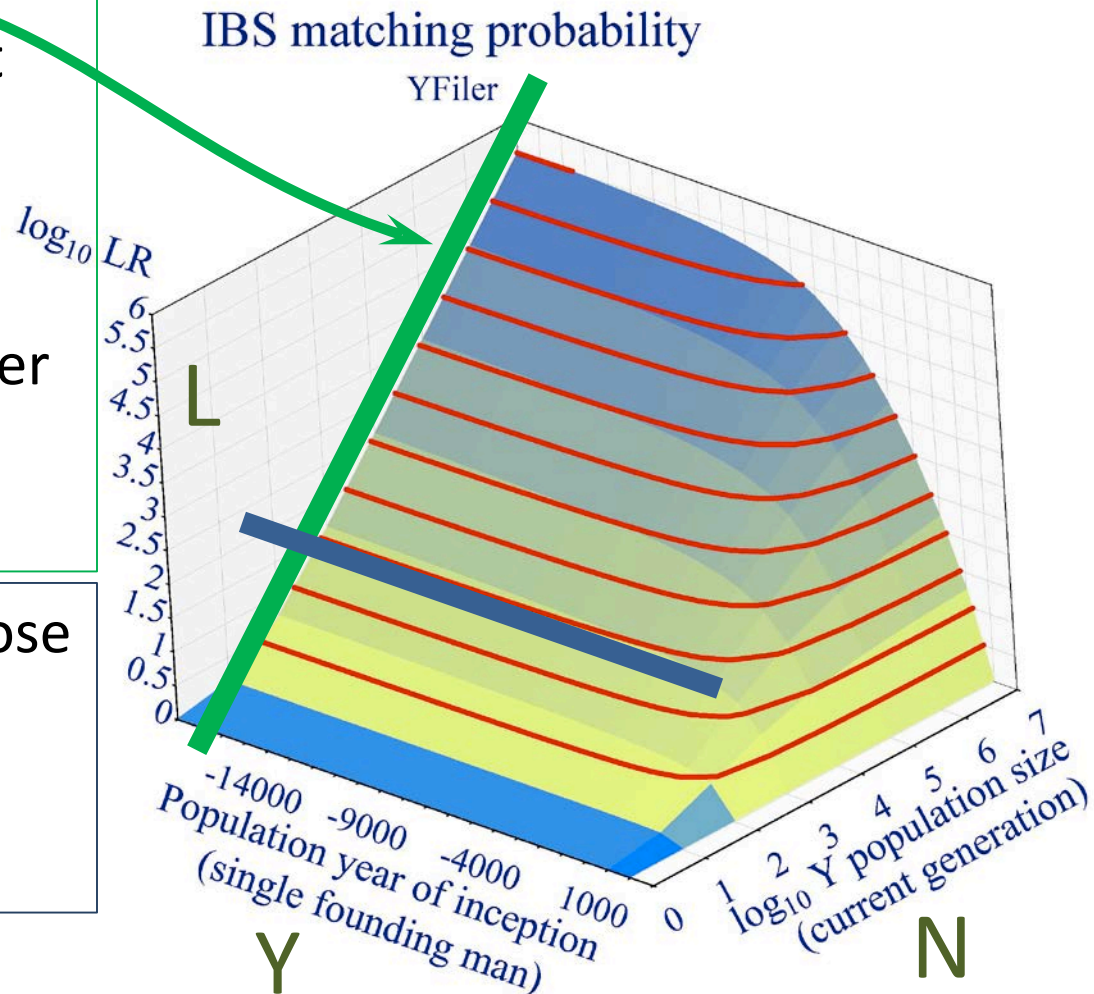
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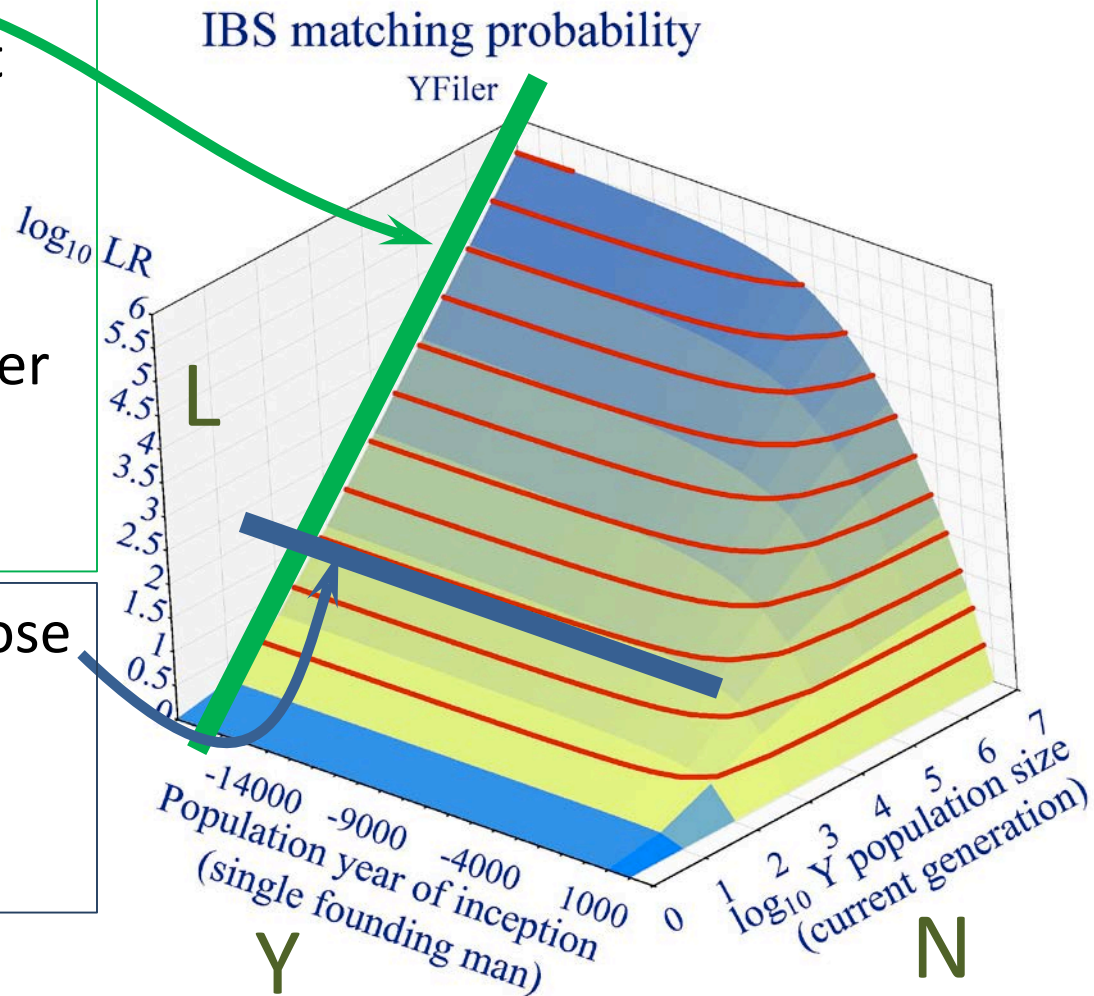
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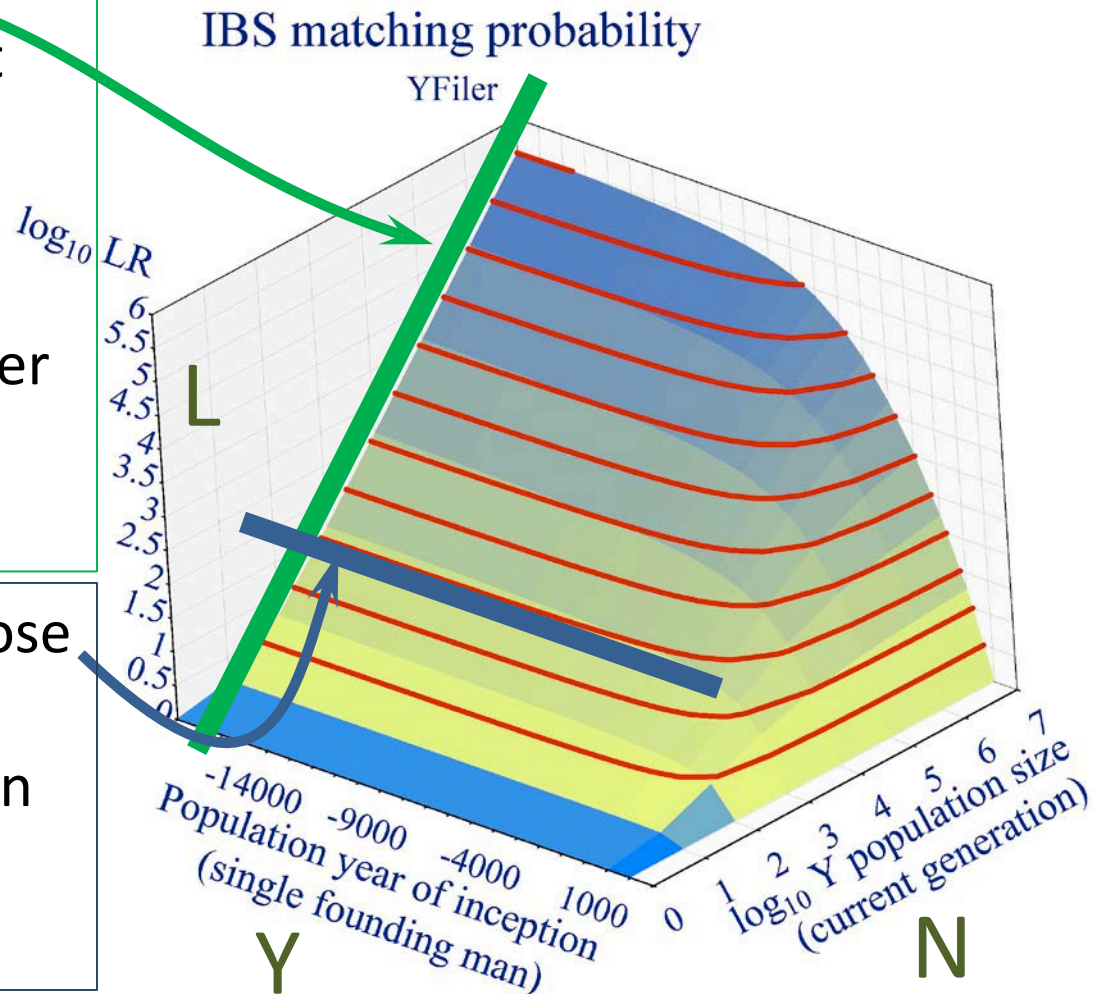
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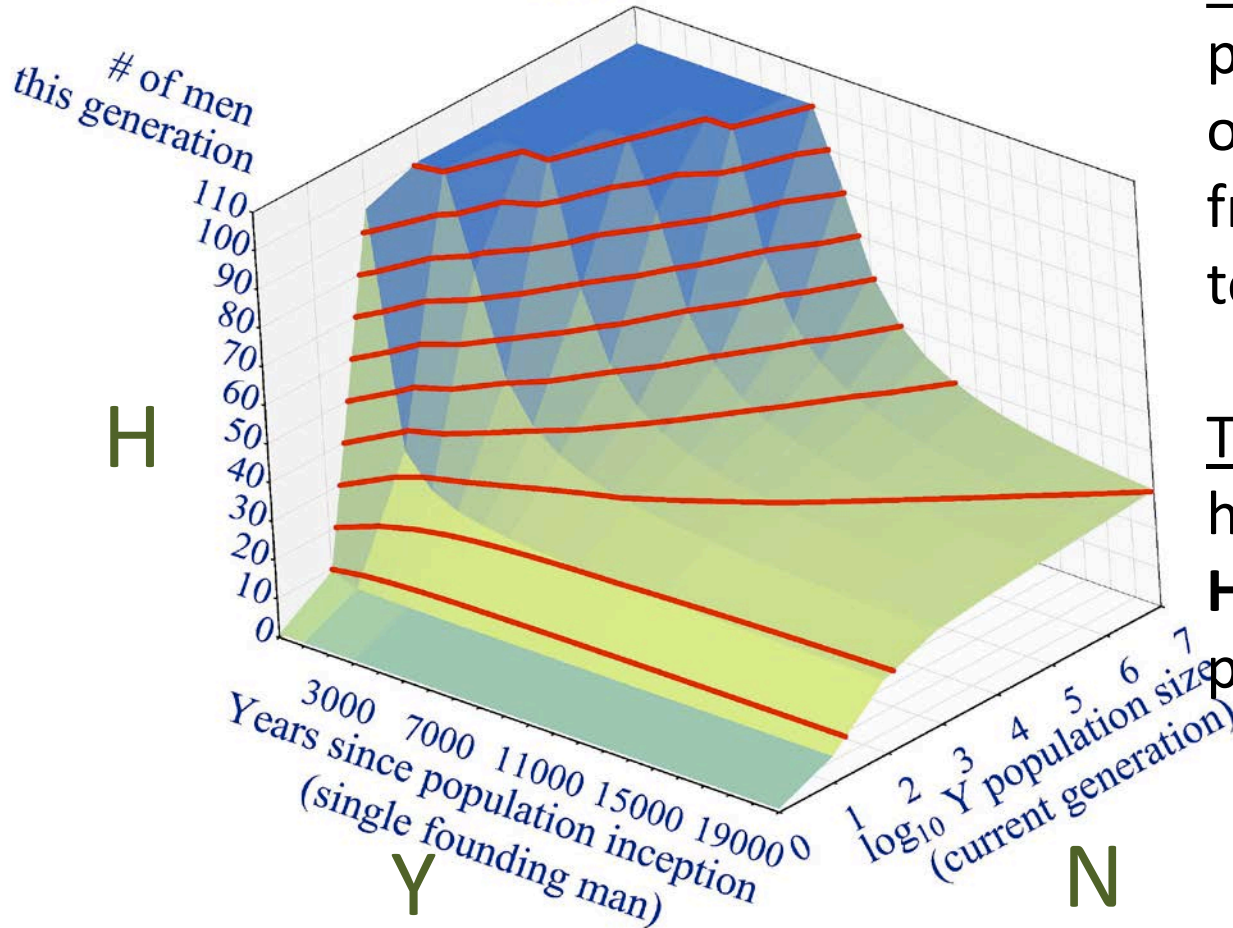
- Straight & horizontal? Close
 - Implies cohort size nearly independent of population growth rate



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

Summary

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 - Quick: 5 minutes to model back to Y-haplotype Adam
- I've (deliberately) chosen simplest model.
 - 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

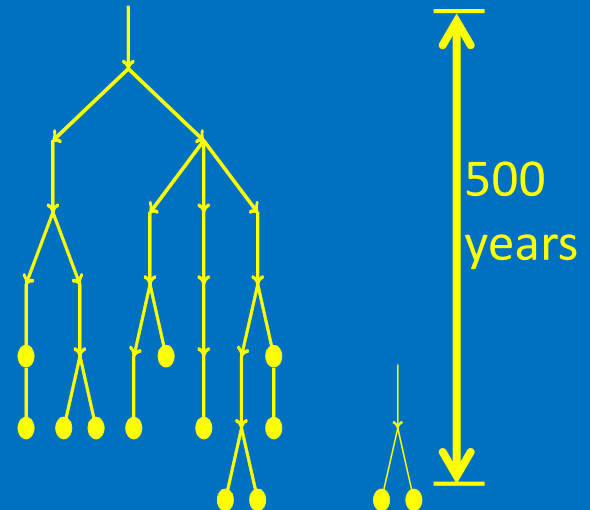
Y haplotype T

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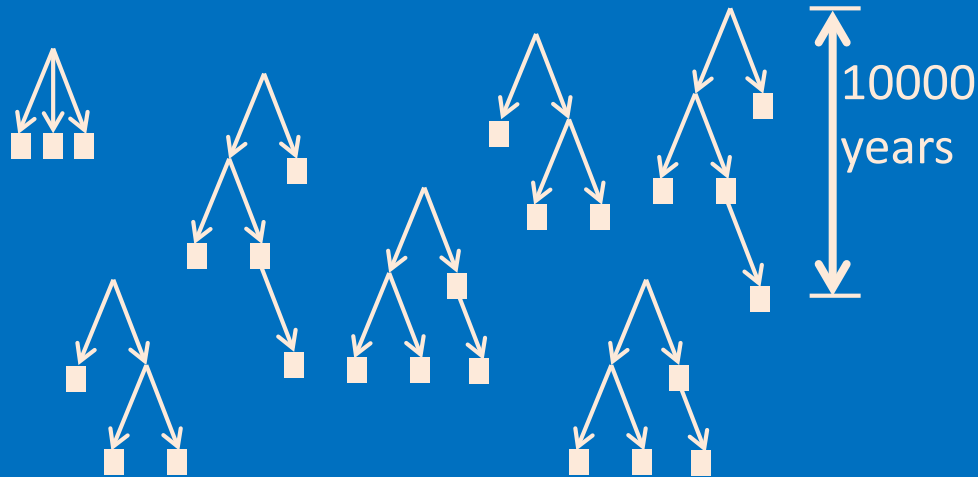
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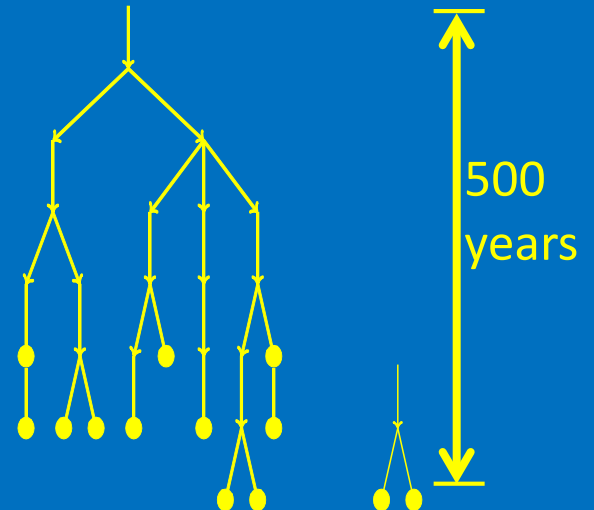
Autosomal STR allele A

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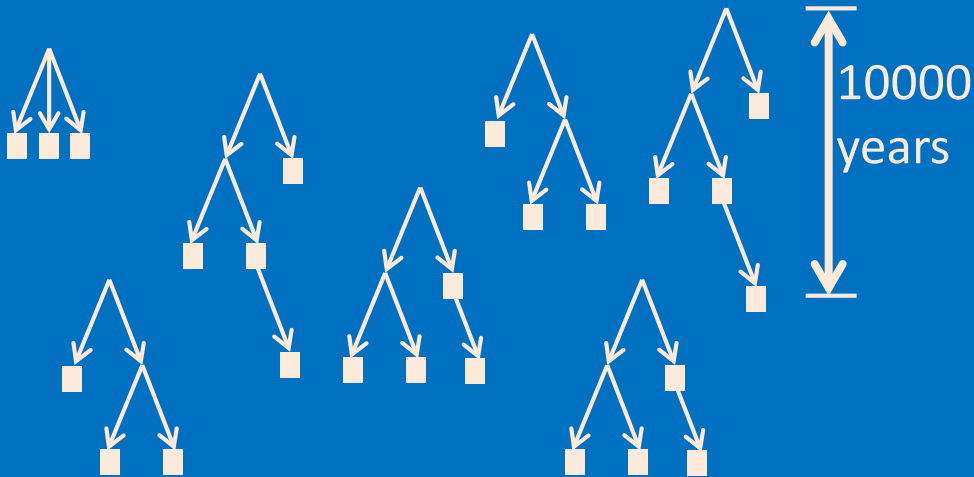
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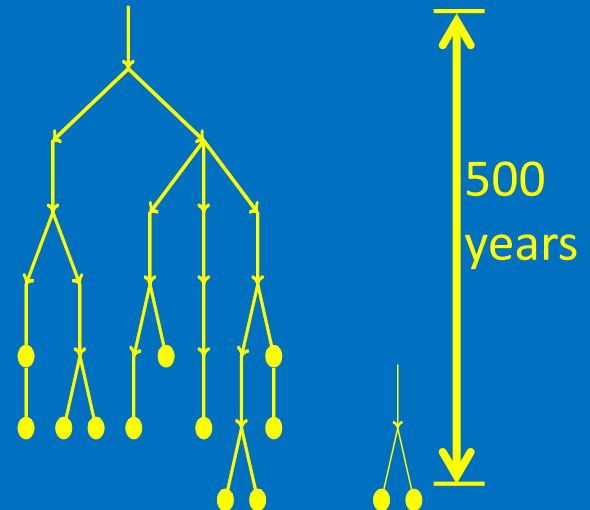
Autosomal STR allele A

- Many **A** families
- 5% of matching is family



Y haplotype T

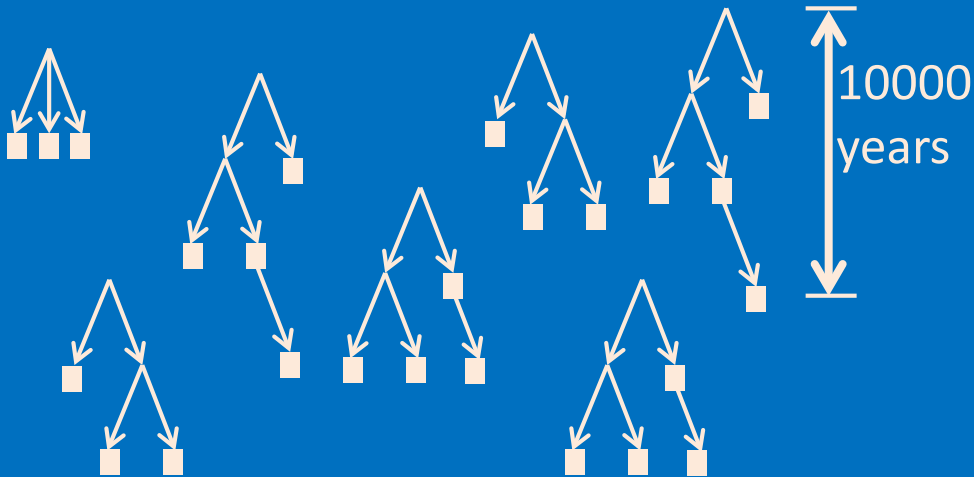
- One dominant **T** family
- 90% of matching is family



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

