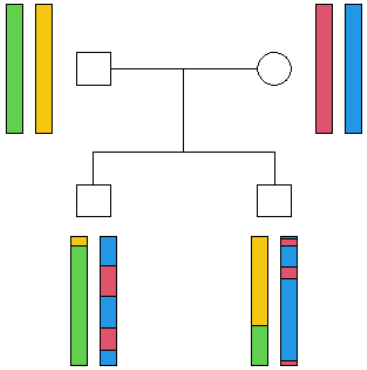


Lecture 5: IBD segments and realised relatedness

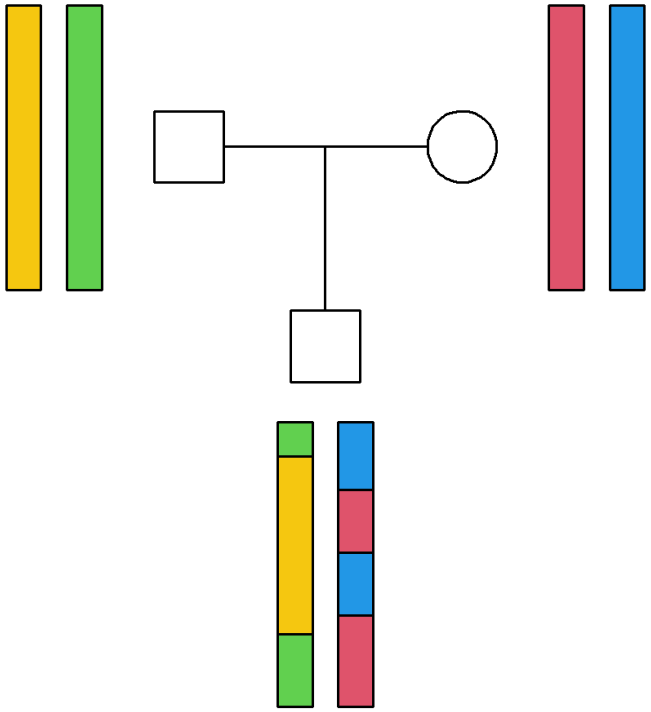
or

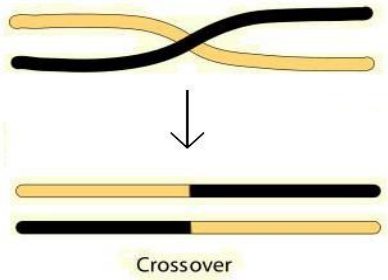
Why are some siblings more alike than others?

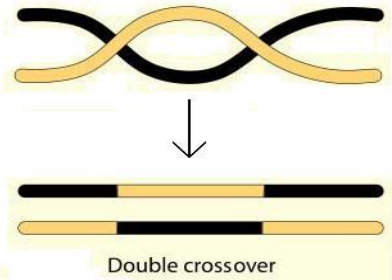
Magnus Dehli Vigeland

Workshop - Alcalá de Henares - 2026

Meiotic recombination

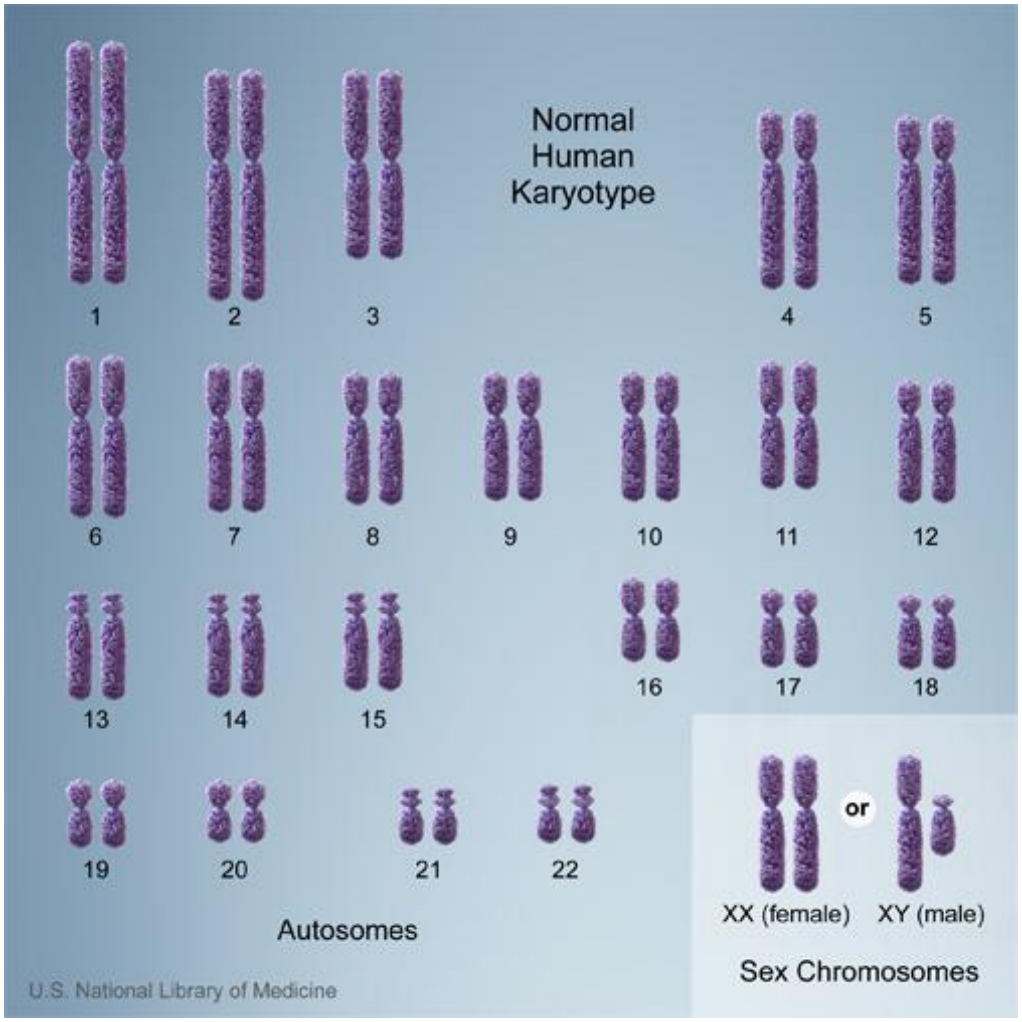


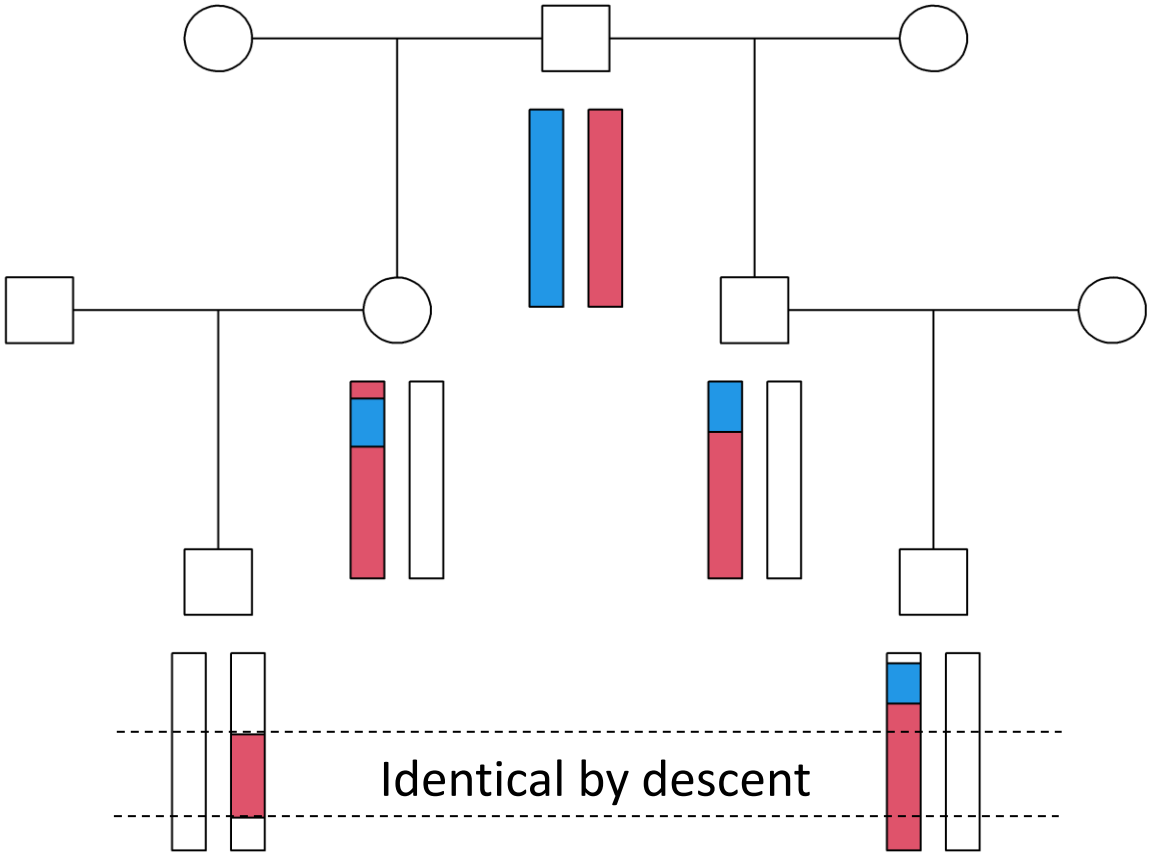


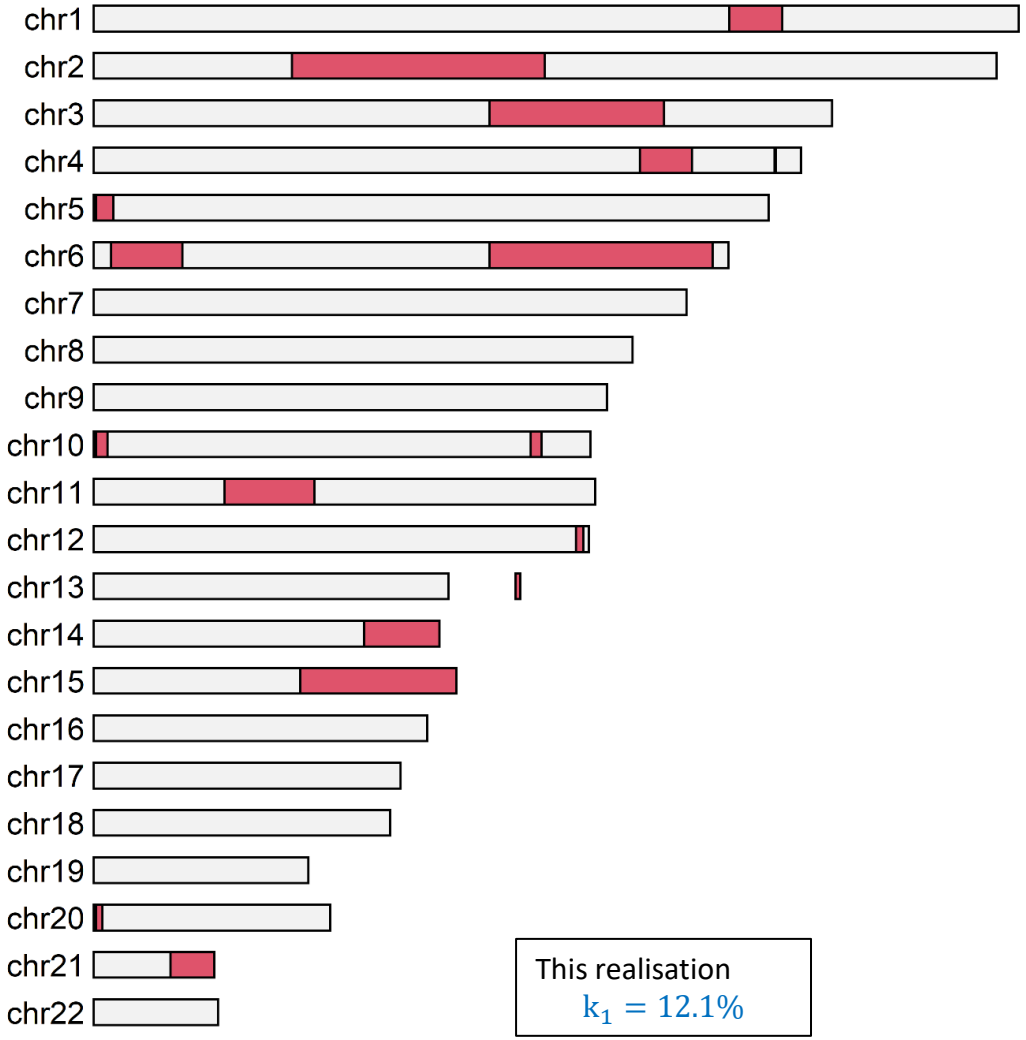


- Genetic distance between two loci:
  = *average # crossovers per meiosis*
- Units:
  - 1 Morgan (M) = 1 crossover per meiosis
  - 1 centiMorgan (cM) = 0.01 M
- The human genome: Ca 30 Morgan

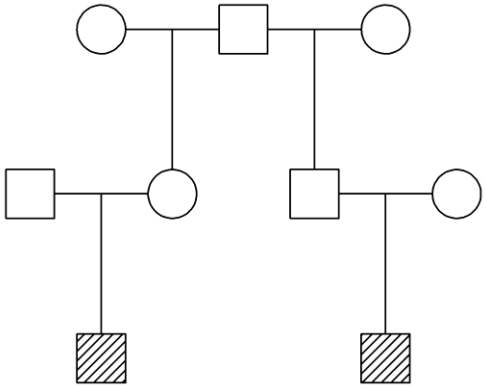
Rule of thumb: One crossover per chromosome arm




Identical by descent

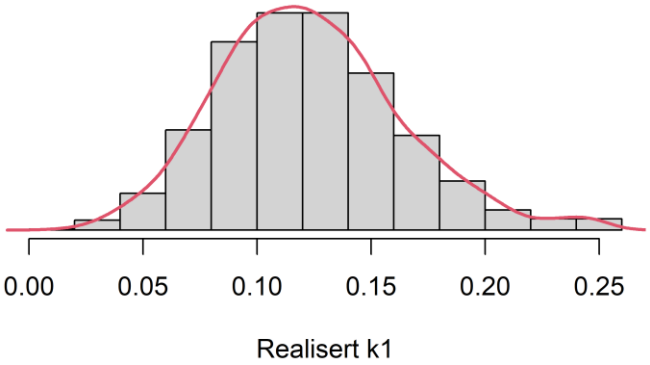


This realisation
$k_1 = 12.1\%$

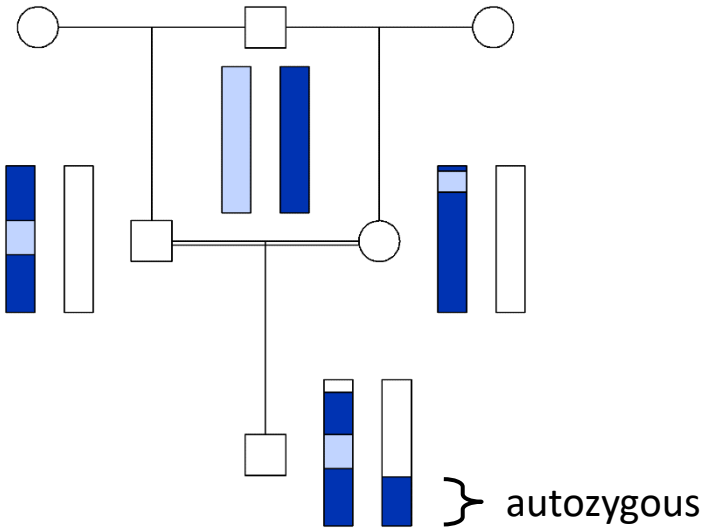


Half first cousins, expected sharing:

$$\kappa_1 = 2 \cdot \left(\frac{1}{2}\right)^4 = \frac{1}{8} = 12.5\%$$

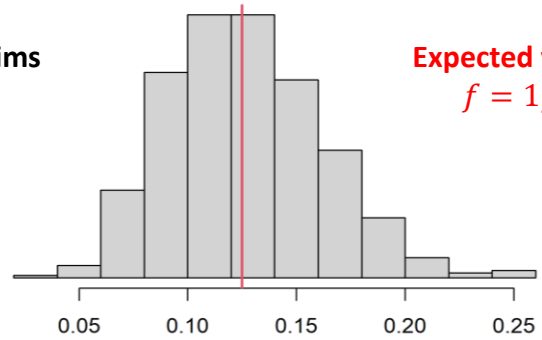


Realised inbreeding



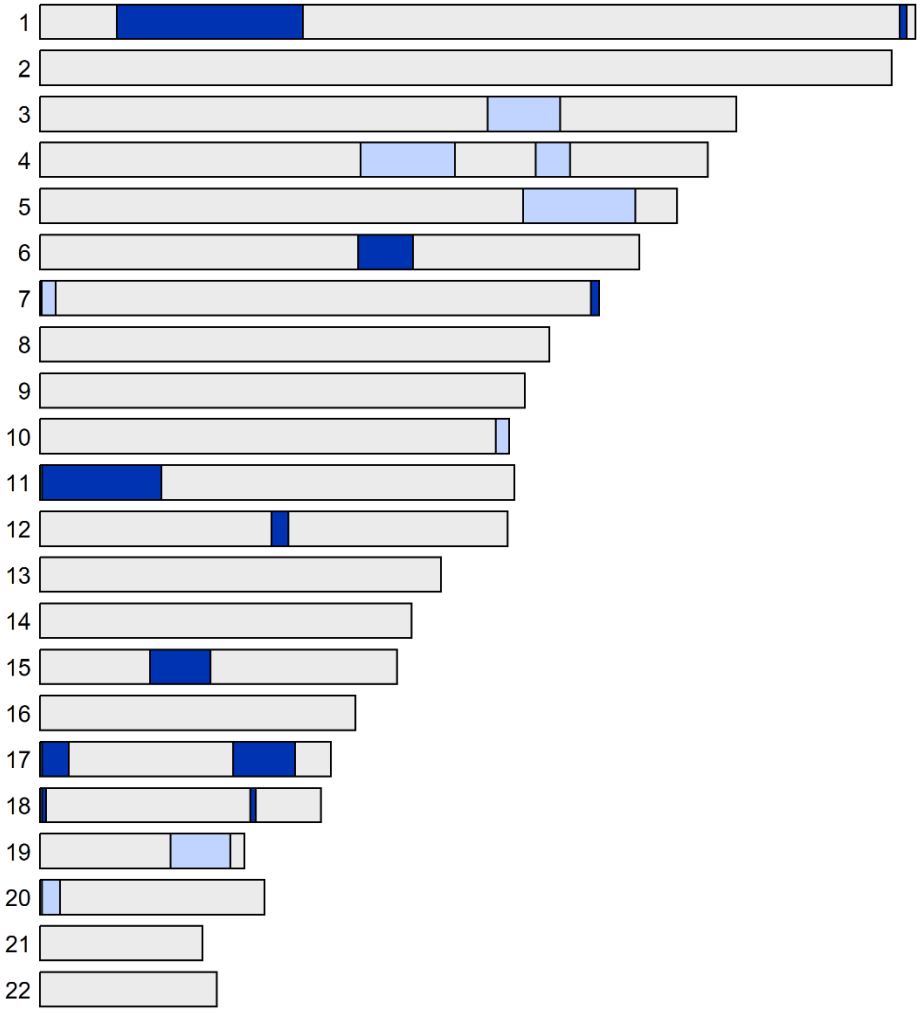
f_R = autozygous fraction of genome

500 sims

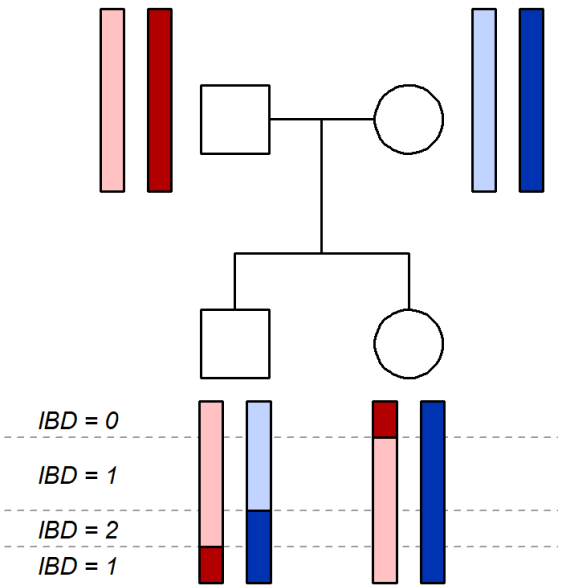


Expected value:
$f = 1/8$

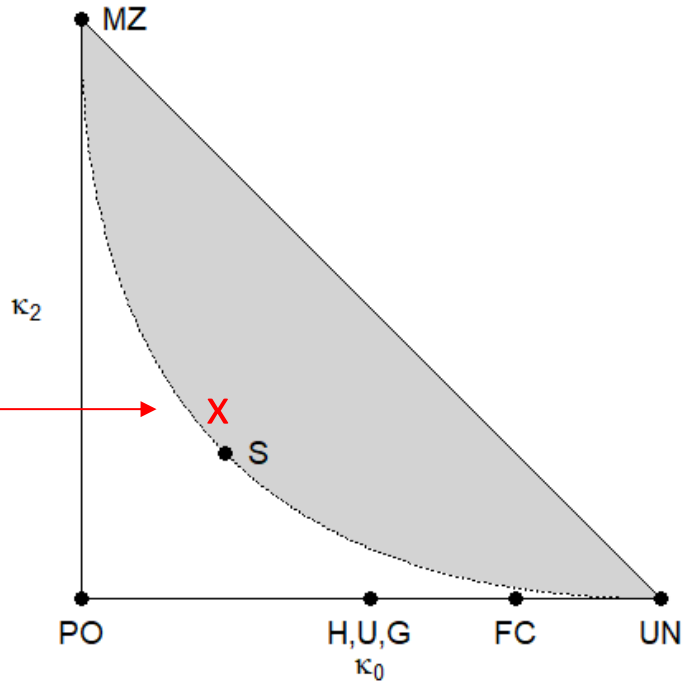
Autozygous segments



Realised IBD coefficients

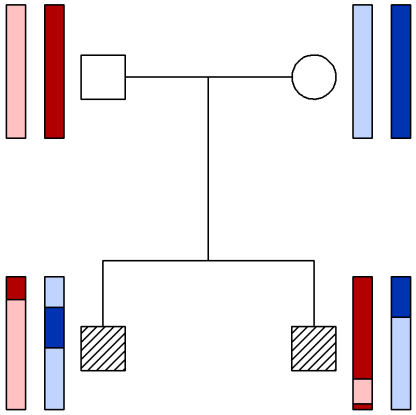


Realised IBD coefficients:
Proportions of genome with IBD = 0, 1, 2



Variation in realised IBD coefficients

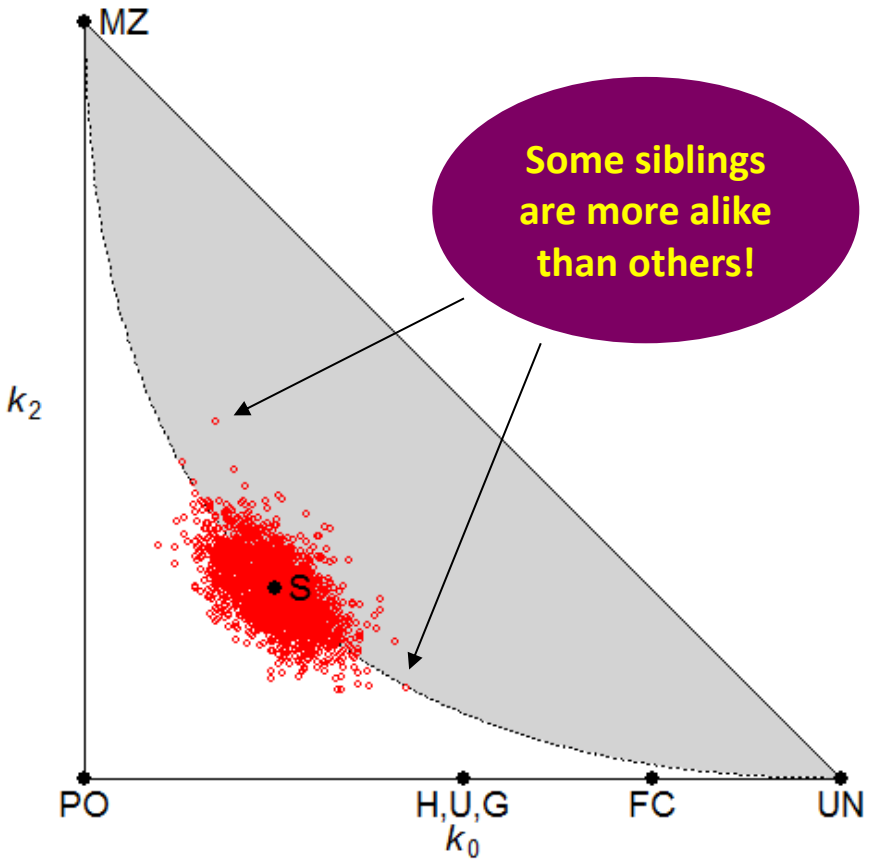
1000 simulations



```
> library(pedsuite)
> library(ibdsim2)

> x = nuclearPed(2)
> s = ibdsim(x, N = 1000)
> k = realisedKappa(s, ids = 3:4)

> showInTriangle(k)
```



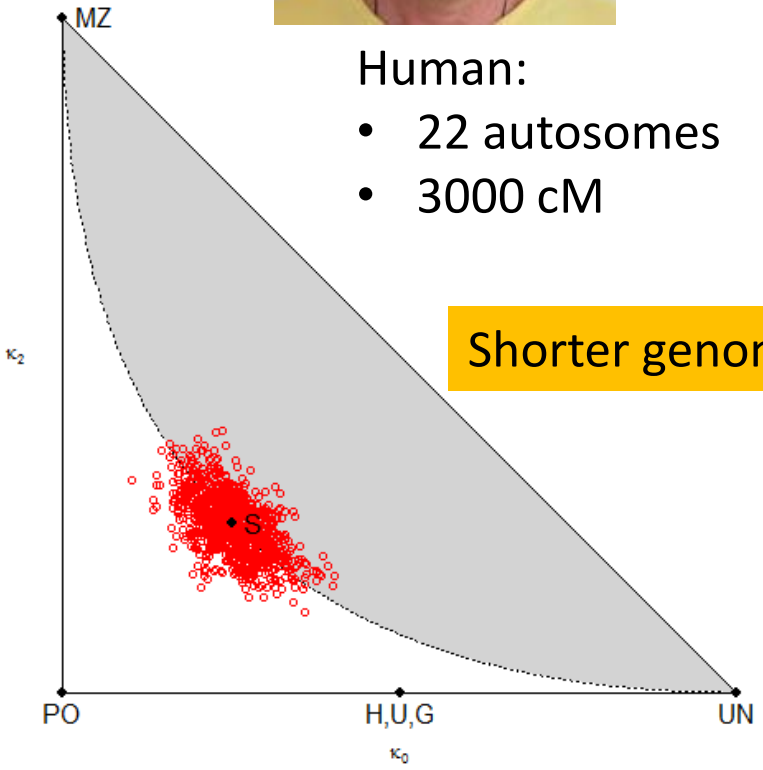
Variation depends on the genome

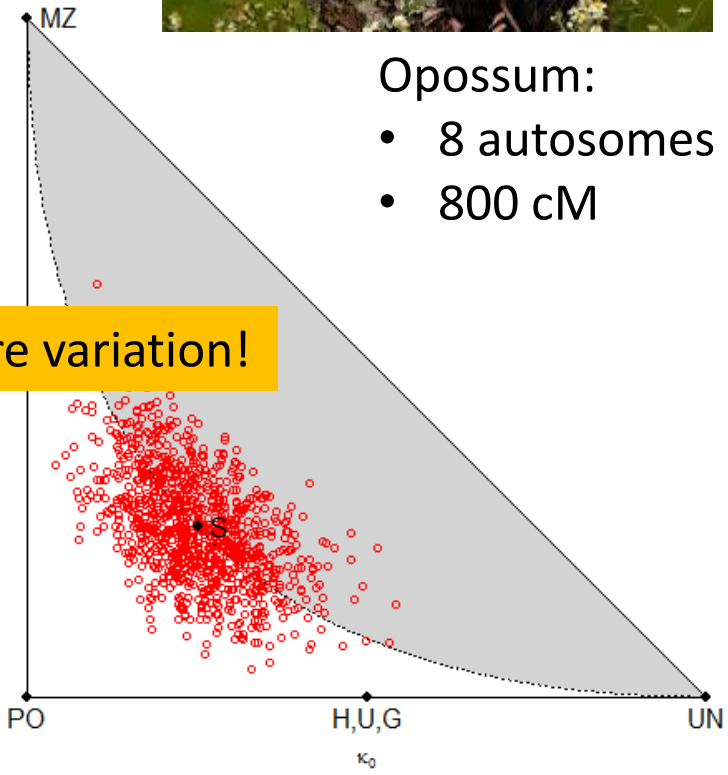
Human:

- 22 autosomes
- 3000 cM

Opossum:

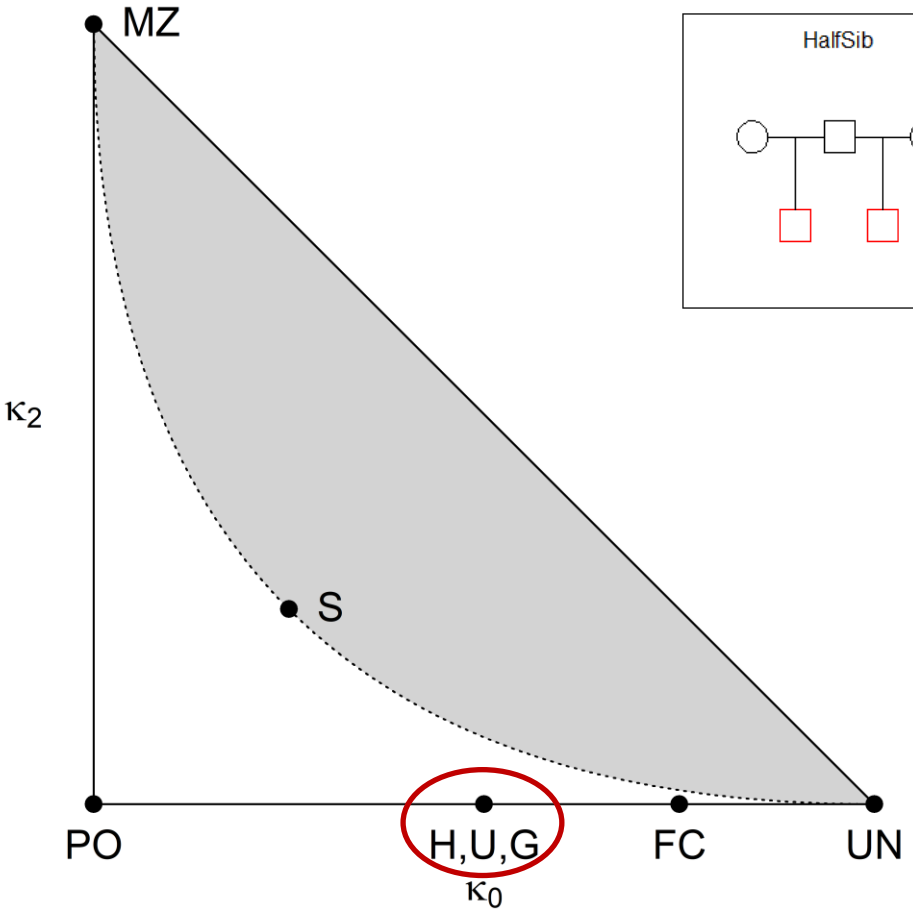
- 8 autosomes
- 800 cM

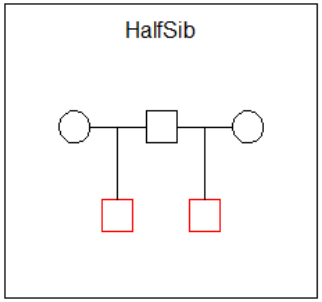


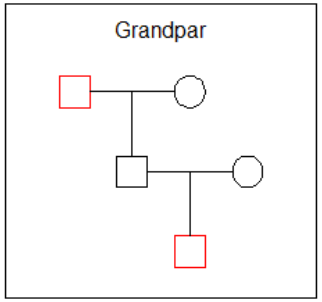


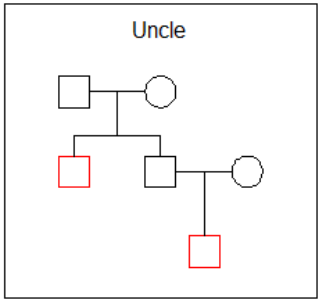
Shorter genome = more variation!

Indistinguishable relationships?



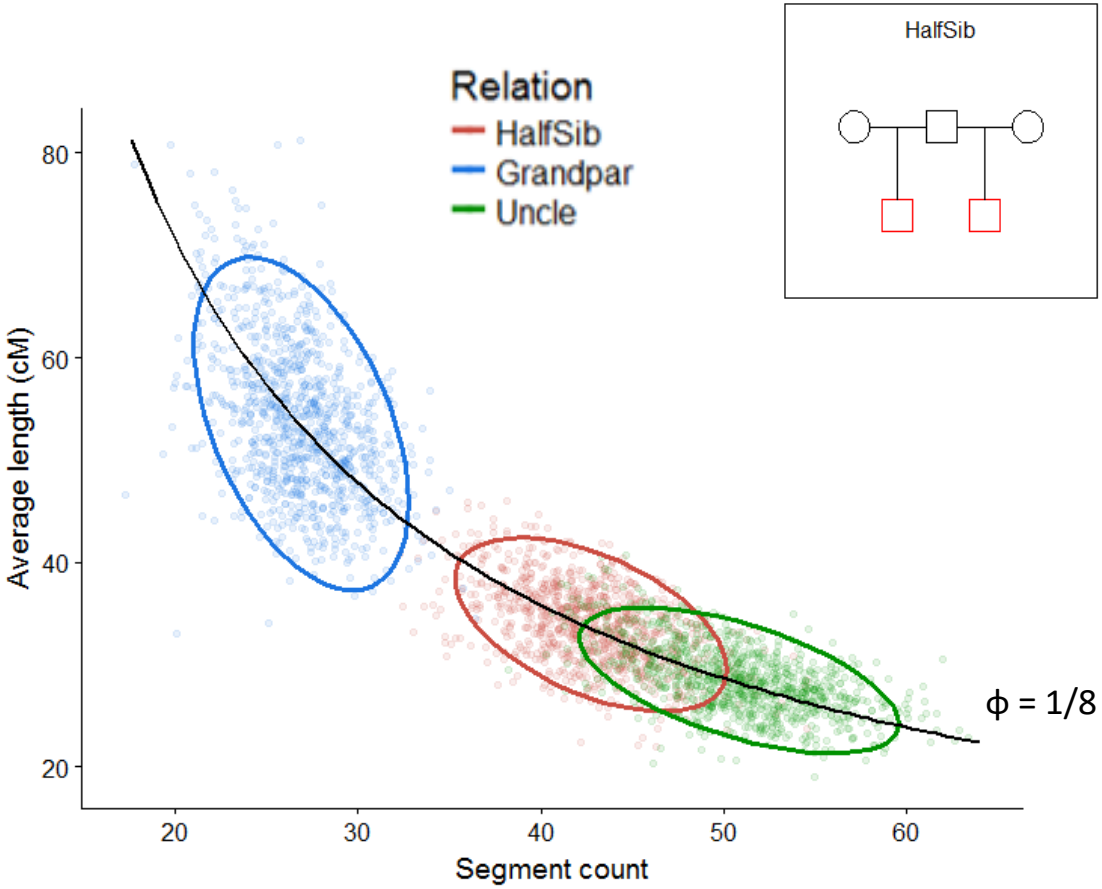


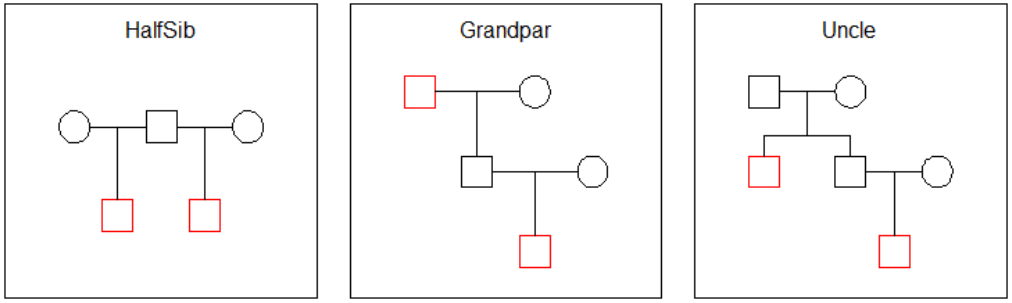




$$\kappa_0 = 0.5$$
$$\kappa_1 = 0.5$$
$$\kappa_2 = 0$$

Simulated IBD distributions





$\phi = 1/8$

In theory distinguishable with linked markers!

METRO

HOME NEWS SPORT ENTERTAINMENT LIFES

UK WORLD WEIRD TECH

Let's not forget – Prince Harry and Meghan Markle are actually (very distant) cousins

Richard Hartley-Parkinson for Metro.co.uk Monday 27 Nov 2017 11:35 am

18.8k

FASHION

CULTURE

FASHION BEAUTY GIFT G

Ou, Awkward...Meghan Markle and Prince Harry are Apparently Related

By Meghan McKenna Date November 2, 2017

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ousins!

Brigitte

SPIELE NEWSLETTER VIDEO GEWINNSPIELE FORUM F-MAG ACADEMY SHOPPING ABO

Aktuell Mode Beauty Rezepte Gesund Liebe Familie Leben Horo

Brigitte → Aktuell → Stars und TV → Meghan Markle und Prinz Harry sind "verwandt"

Stammbaum erforscht: Prinz Harry und Meghan Markle sind Cousins!

STJERNEPAR: Meghan Markle og prins Harry viser endelig kjærligheten sin offentlig, etter å ha holdt forholdet svært privat i lang tid. Nå kommer det frem at paret, som er fra to forskjellige kontinenter, faktisk er i slekt. Foto: NTB scanpix

Prins Harry er i slekt med kjæresten

SE OG HØR

1.1K shares

View c

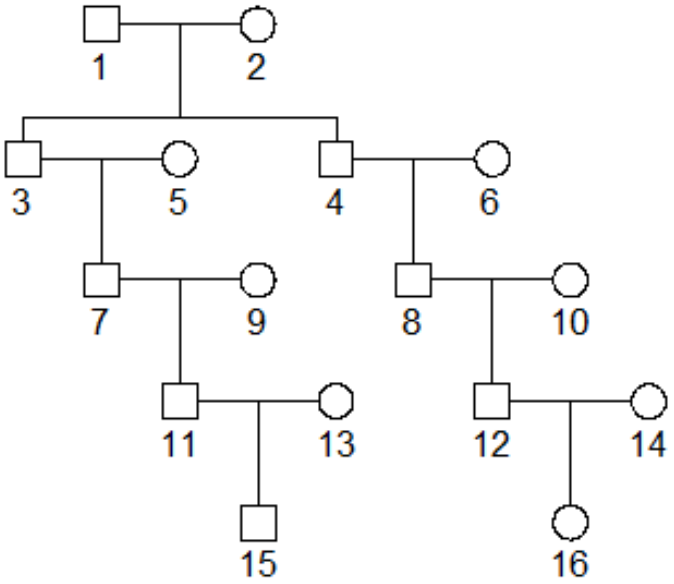
(Picture: Mail Online)

14 13 10 9 8 4 3 2 1

15 13 11 10 9 5 4 3 2 1

13th cousins once removed

The probability of zero IBD



N'th cousins	*P*(zero IBD)
first	0.0 %
second	0.0 %
third	1.5 %
fourth	28 %
fifth	67 %

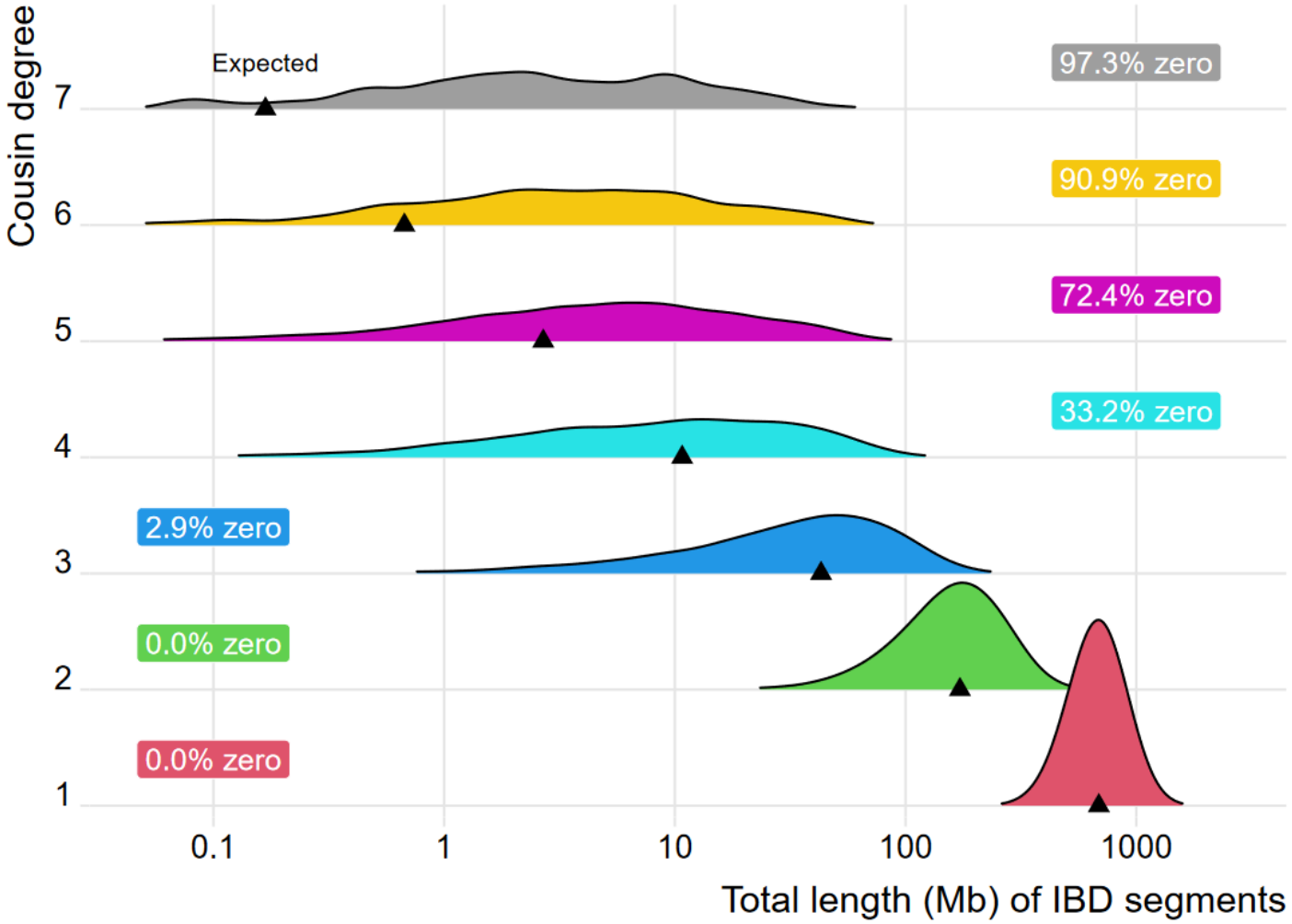
Third cousins

Expected fraction with IBD = 1:

$$k_1 = \frac{1}{64}$$

Two individuals can have a common ancestor without being genetically related

Distant cousins share either nothing or quite a bit



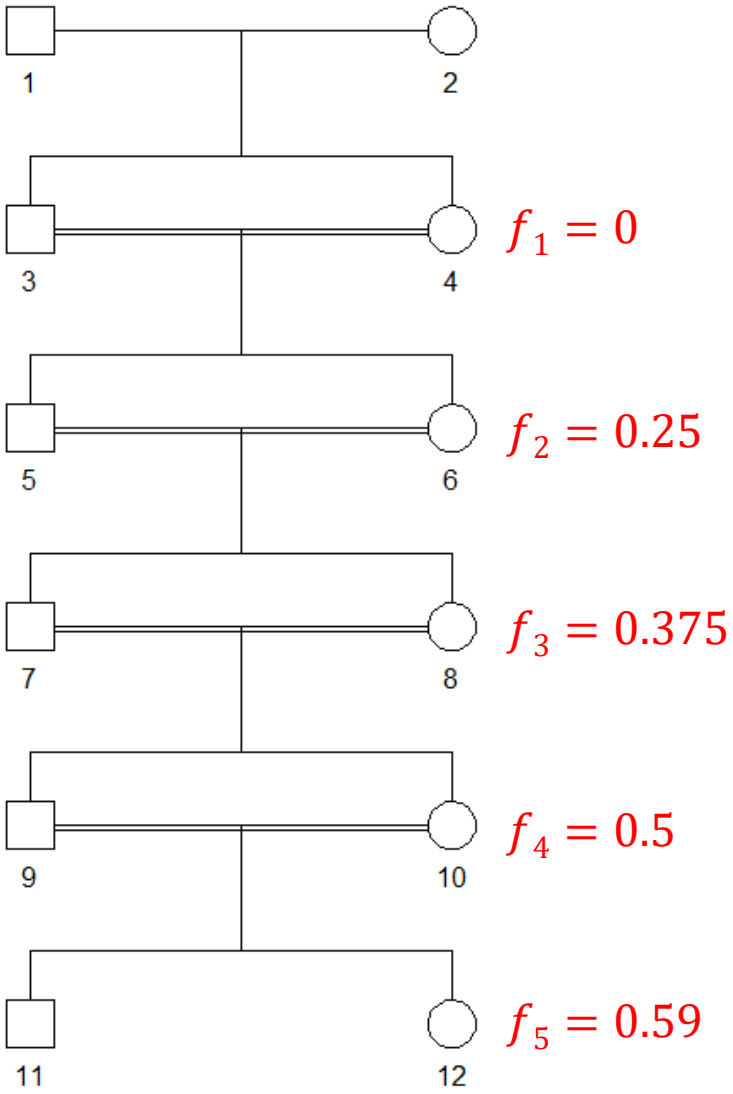
Reversely:
Is 100 % inbreeding possible?

- Continuous full-sib mating

- Easy to show:
  - inbreeding coefficient $f \rightarrow 1$

- In finite pedigree:
  - pedigree-based f will *never* reach 1

But: After ~30 generations, the **realised inbreeding** typically reaches 1

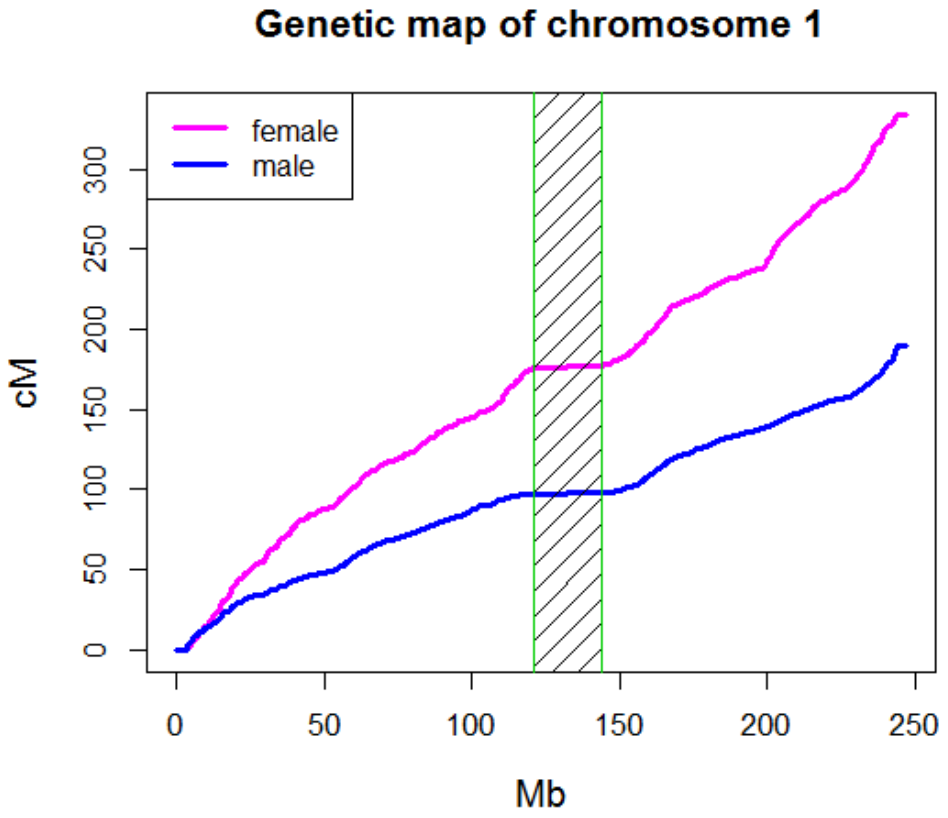


The importance of sex

- Rule of thumb:

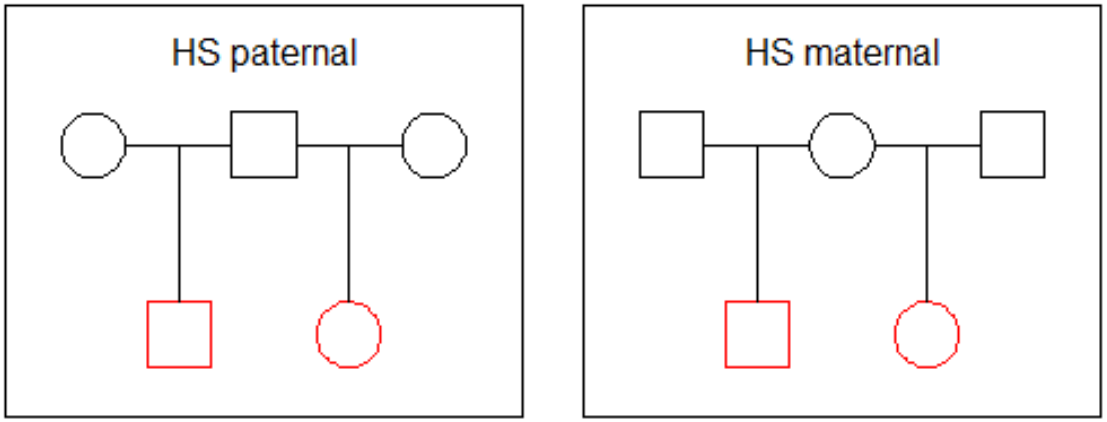
$$1\ \text{cM} \approx 1\ \text{Mb}$$

- But: crossover rates vary
  - across the genome
  - males vs. females

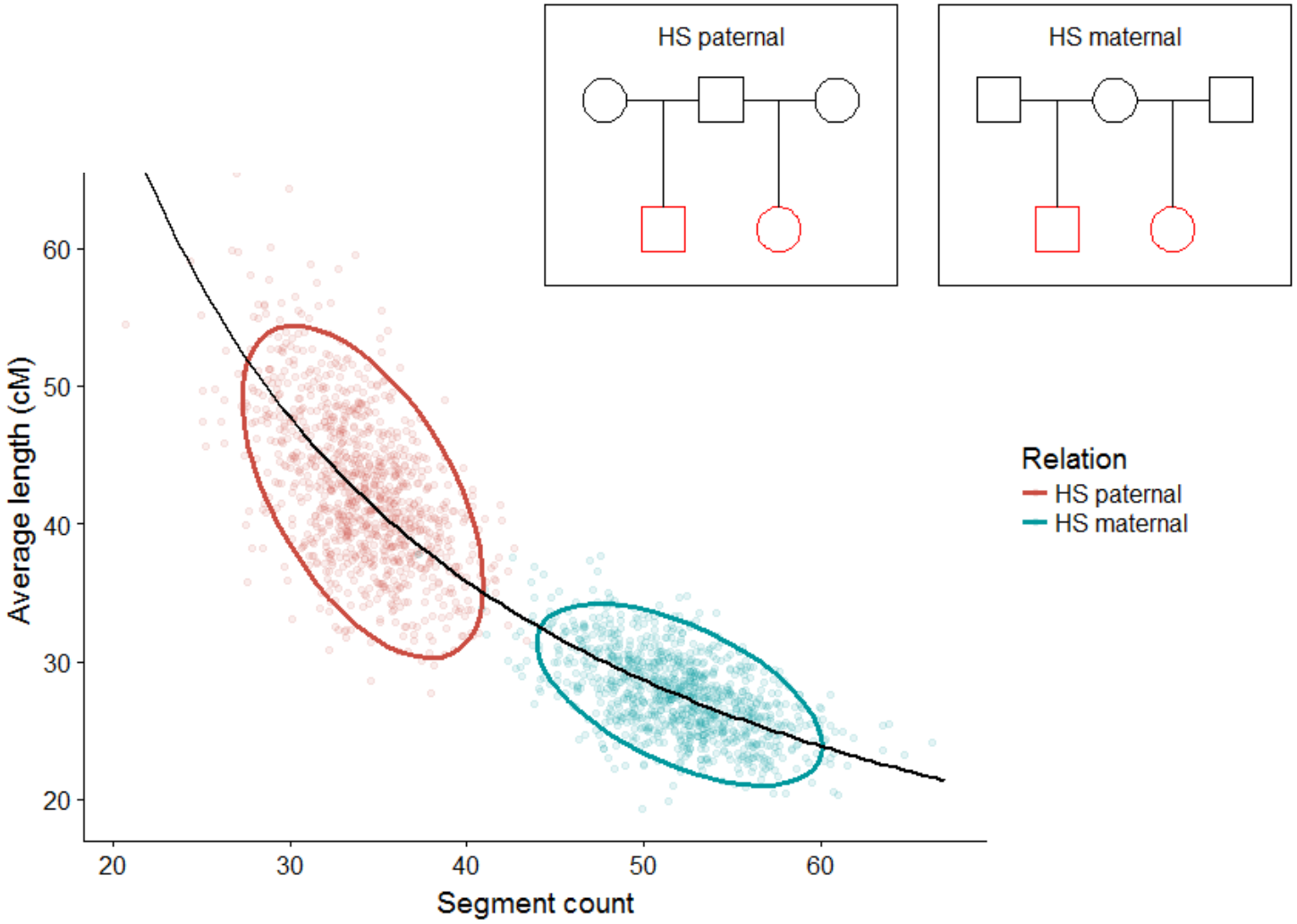


Females have a much longer genome!

Can we separate these??



Yes!


HS paternal
HS maternal
Average length (cM)
60
50
40
30
20
20
30
40
50
60
Segment count
Relation
HS paternal
HS maternal

IBD sharing by family members

Purpose: Estimate and visualise distributions of genomic segments shared identical-by-descent (IBD) between related individuals, or within inbred individuals (autozygosity). Recombination is simulated down through the pedigree, using detailed, sex-specific crossover rates for the human genome (Halldorsson et al., 2019).

More information: This program is a frontend for the R package ibdsim2, which is part of the pedsuite ecosystem for pedigree analysis. Details about the simulations can be found in the package documentation, and in the book Pedigree analysis in R. Please cite this book if you use the app in your work.

Pedigree 1

Built-in pedigree

Half-sibs, maternal

Load ped file

Individuals

4, 5

Label

Ped 1

PARAMETERS

Chromosome

1 - 22 X

Crossover model

χ^2 Haldane

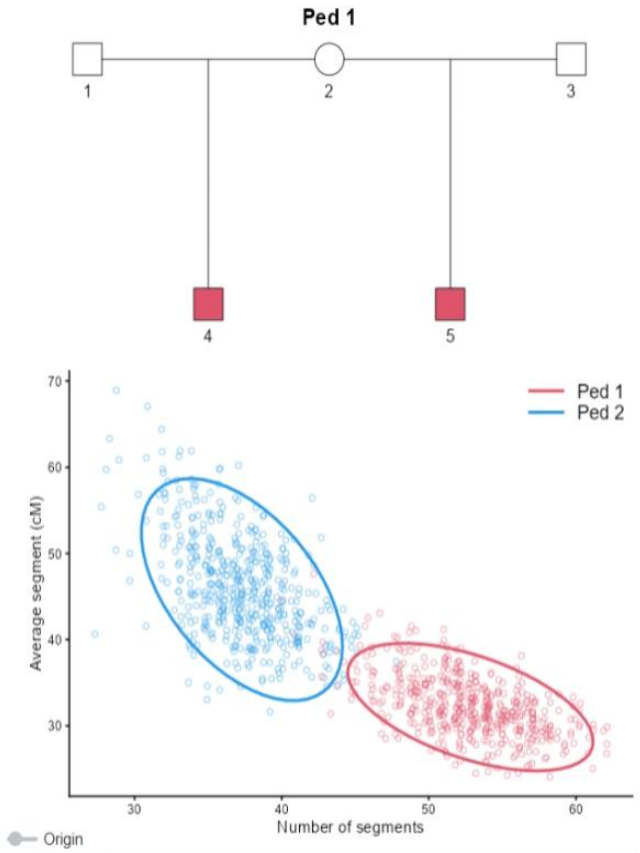
Sex-specific map

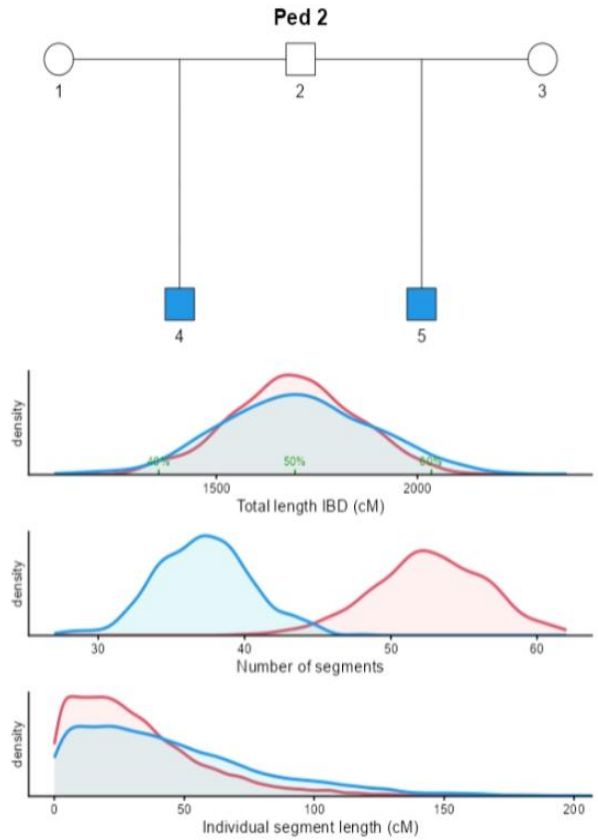
On Off

Cutoff **Seed**

0 123

Simulate!





Pedigree 2

Built-in pedigree

Half-sibs, paternal

Load ped file

Individuals

4, 5

Label

Ped 2

PARAMETERS

Chromosome

1 - 22 X

Crossover model

χ^2 Haldane

Sex-specific map

On Off

Cutoff **Seed**

0 123

Simulate!

Settings

Number of sims

500

Length unit

cM Mb

Analysis

Sharing Autozygosity

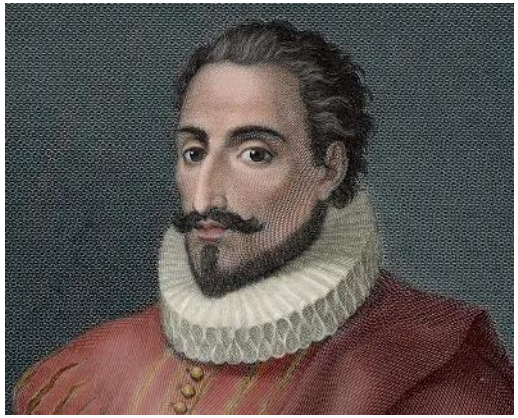
Download data

Observed data

Total length **Count**

Segments

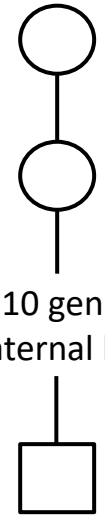
This is version 2.3.2.9000 of ibdsim2 (changelog). For bug reports, feature requests, or other comments, please file an issue at https://github.com/magnusdv/ibdsim2/issues.



Miguel de Cervantes

P(some IBD) = 38%

Isabella I of Castile



P(some IBD) = 51%

Summary of genetic relatedness

- Pedigree-based measures:
  - the kinship/inbreeding coefficient φ
  - the kappa coefficients $\kappa = (\kappa_0, \kappa_1, \kappa_2)$

- Each coefficient is
  - the probability of observing a certain IBD pattern in a random locus
  - the expected proportion of the genome in this state

- Realised relatedness:
  - IBD segments determined by meiotic recombination
  - females recombine more than males
  - may separate relationships with equal kappa's
  - 0% and 100% realised identity is possible!

So...what does it mean to be related?

- Pedigree based definition: $\varphi > 0$

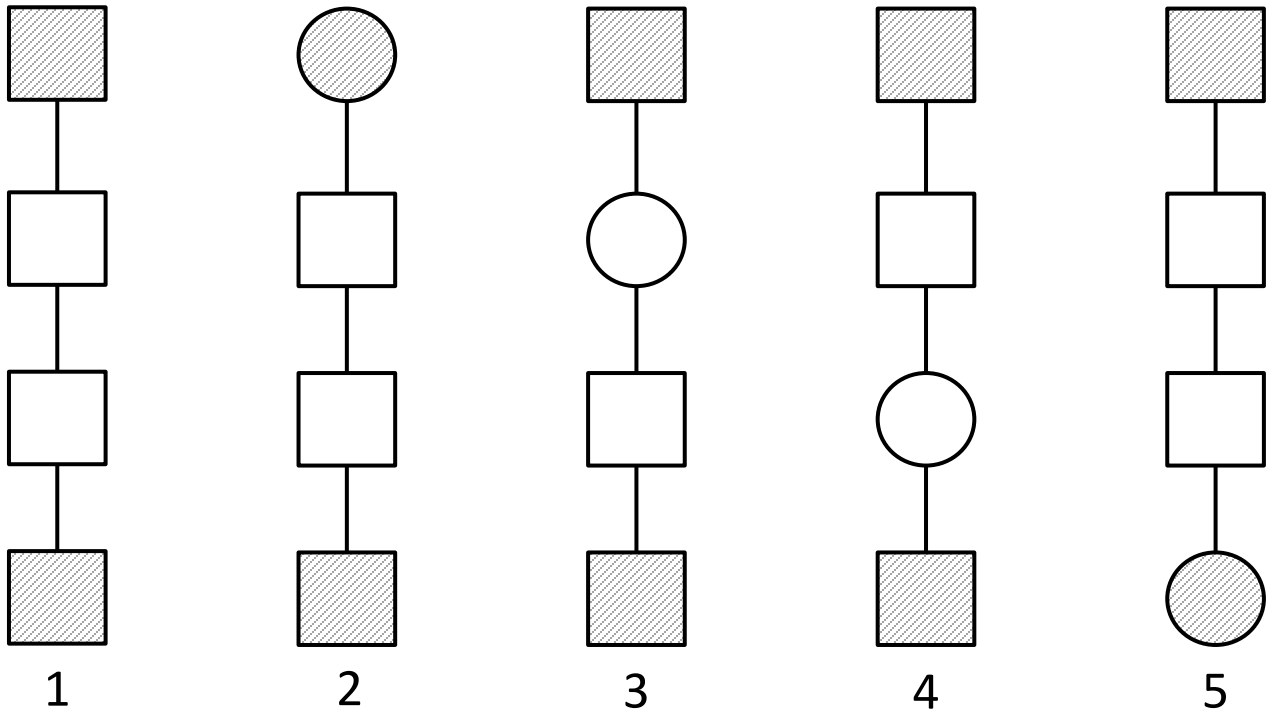
  potentially having alleles IBD

- Genomic definition (**realised** relatedness):

  actually having alleles IBD

Epilogue

- Which of these relationship are genetically different?

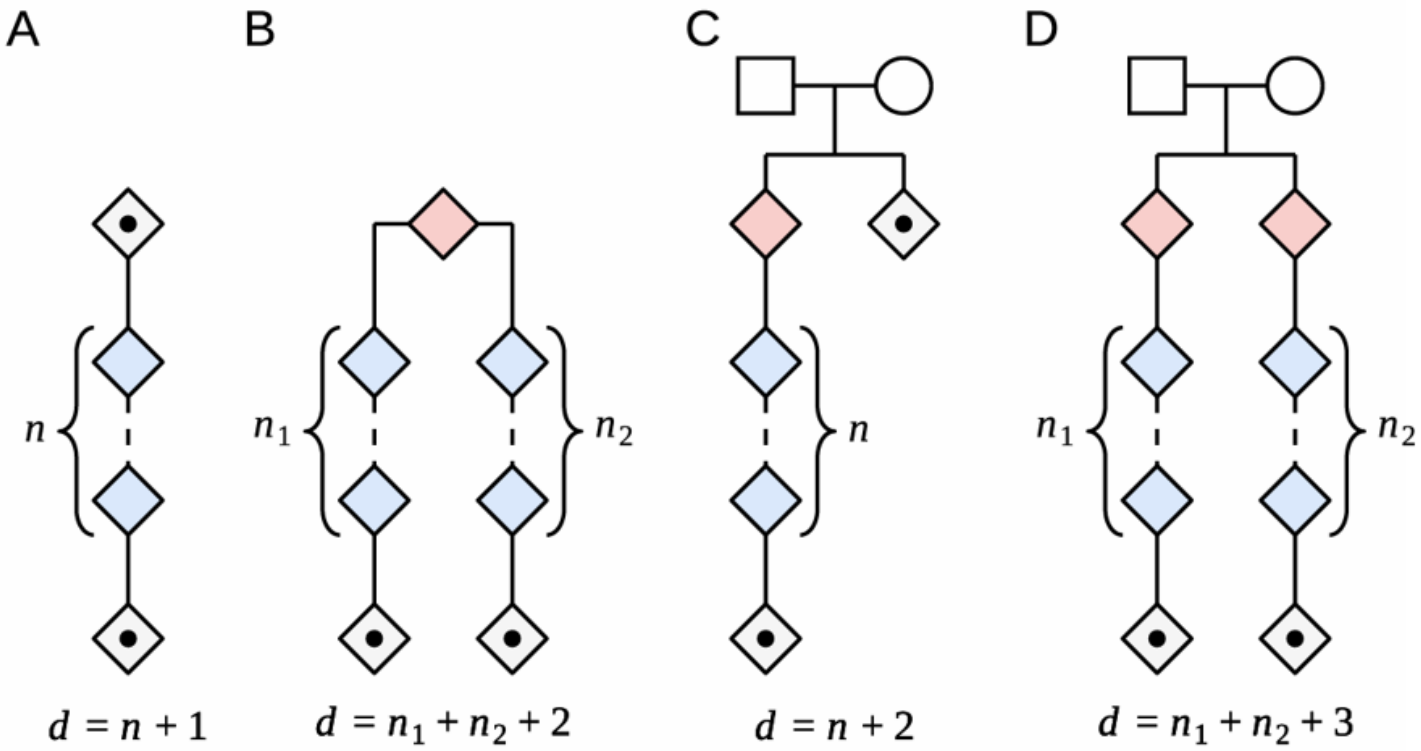


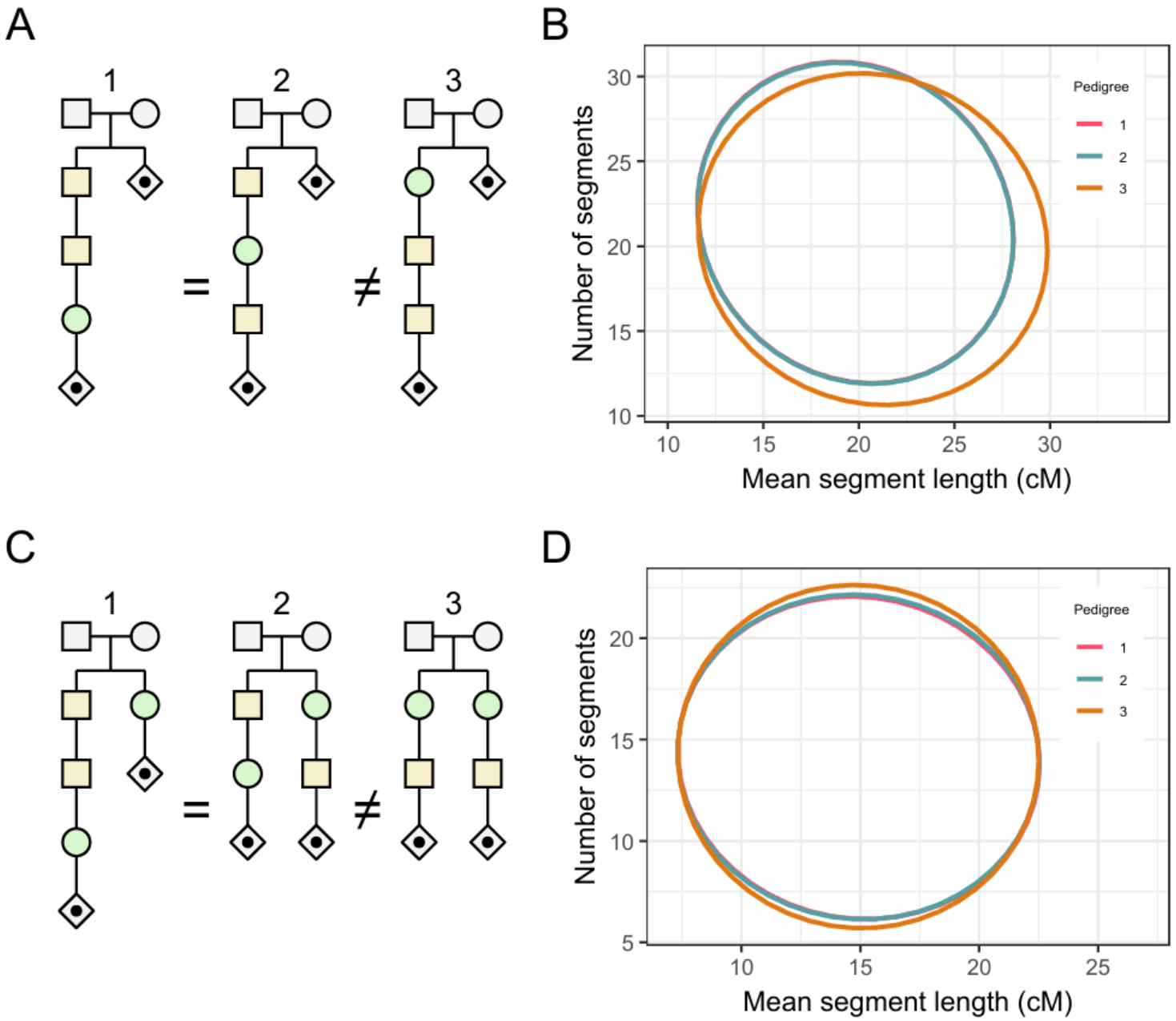
- Answer: 1 = 2 = 5 and 3 = 4

Henrik Nordtorp

Theorem
Two unilineal relationships have identical IBD distributions
if and only if they have the same ...

- ❖ Type (A-D)
- ❖ Degree
- ❖ Anchor females/males
- ❖ Free females/males




A
1
2
3
=
≠
B
Number of segments
Mean segment length (cM)
Pedigree
1
2
3
C
1
2
3
=
≠
D
Number of segments
Mean segment length (cM)
Pedigree
1
2
3