

Pedigrees analysis and relatedness inference



Teachers

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Home page:

https://magnusdv.github.io/pedsuite/articles/web_only/course-isfg2026.html

Schedule

Morning session – Pedigree analysis: Basic

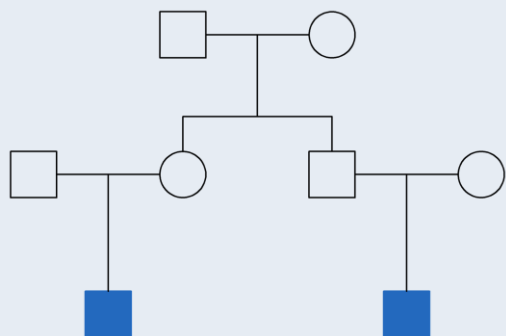
- 08:30–09:30 **Pedigrees and measures of relatedness** (MDV)
- 09:30–10:30 Exercises I
- 10:30–10:45 *Coffee break*
- 10:45–11:30 **Kinship testing** (TE)
- 11:30–12:15 Exercises II
- 12:15–12:30 Summary and discussion

Lunch break 12:30 - 14:00

Afternoon session – Pedigree analysis: Advanced

- 14:00–15:00 **Relatedness inference and pedigree reconstruction** (MDV)
- 15:00–15:45 Exercises III
- 15:45–16:00 *Coffee break*
- 16:00–17:00 **Disaster victim identification** (TE)
- 17:00–17:45 Exercises IV
- 17:45–18:00 Summary and discussion

Files needed for exercises: [data-isfg26.zip](#)



Lecture 1: Pedigrees and measures of relatedness

Magnus Dehli Vigeland

ISFG Congress Montreal 2026

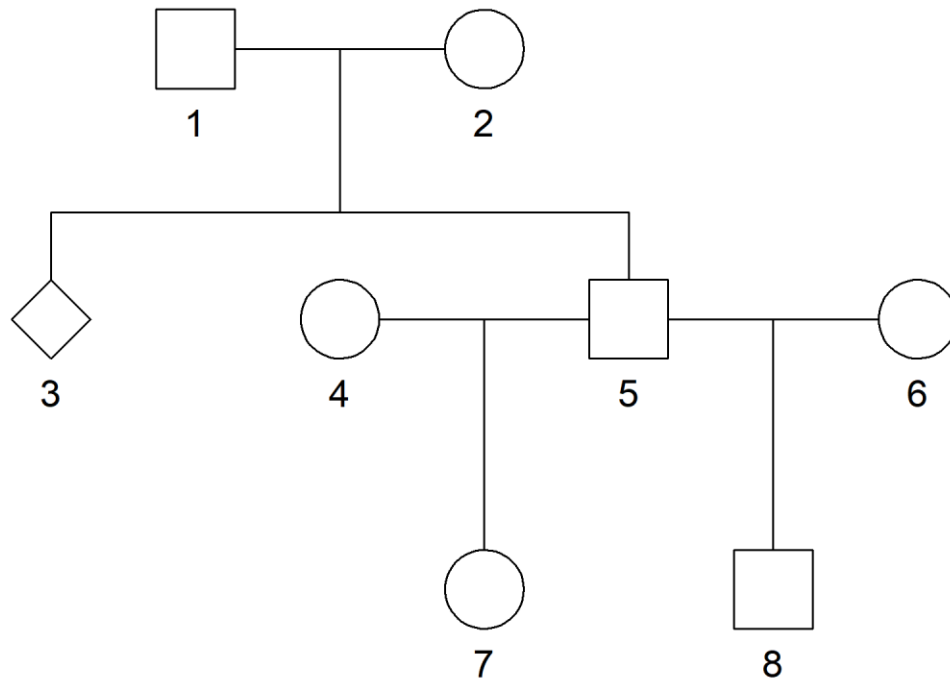
Pedigree workshop

Outline

- Part I: *Pedigrees*
 - Conventions and terminology
 - QuickPed
- Part II: *Measures of relatedness*
 - Identity by descent (IBD)
 - Kinship/inbreeding coefficients
 - IBD triangle
 - A word of caution: Degree of relatedness
- Part III: *Realised relatedness*
 - IBD segments
 - Simulation tool: ibdsim2

Part I: Pedigrees

Conventions and terminology

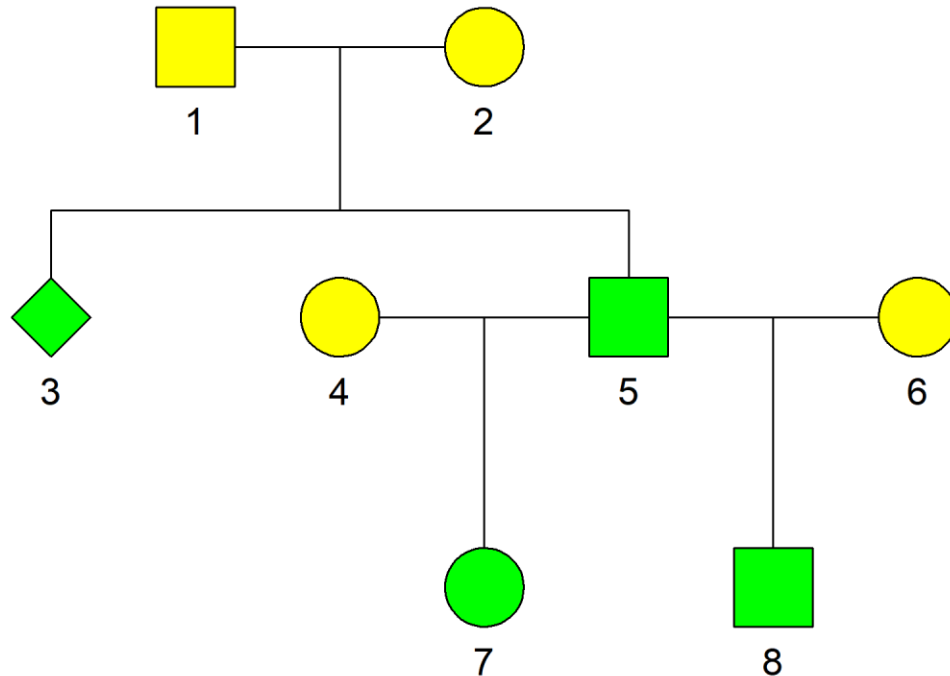


□ = male

○ = female

◇ = unknown

Conventions and terminology



□ = male

○ = female

◇ = unknown

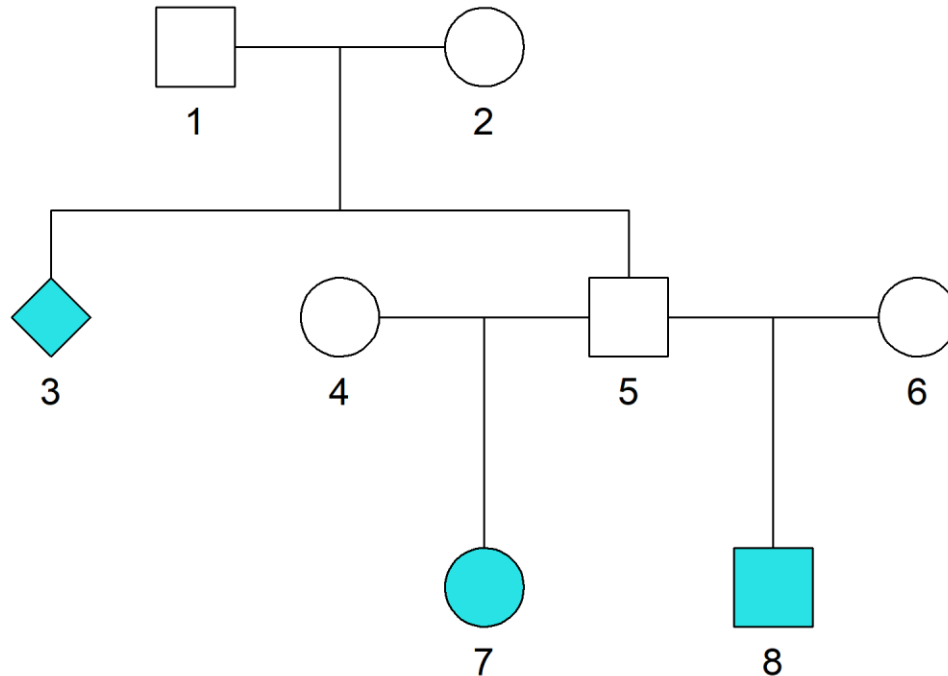
Founders

No parents included
in the pedigree

Nonfounders

Parents are included

Conventions and terminology



□ = male

○ = female

◇ = unknown

Founders

No parents included
in the pedigree

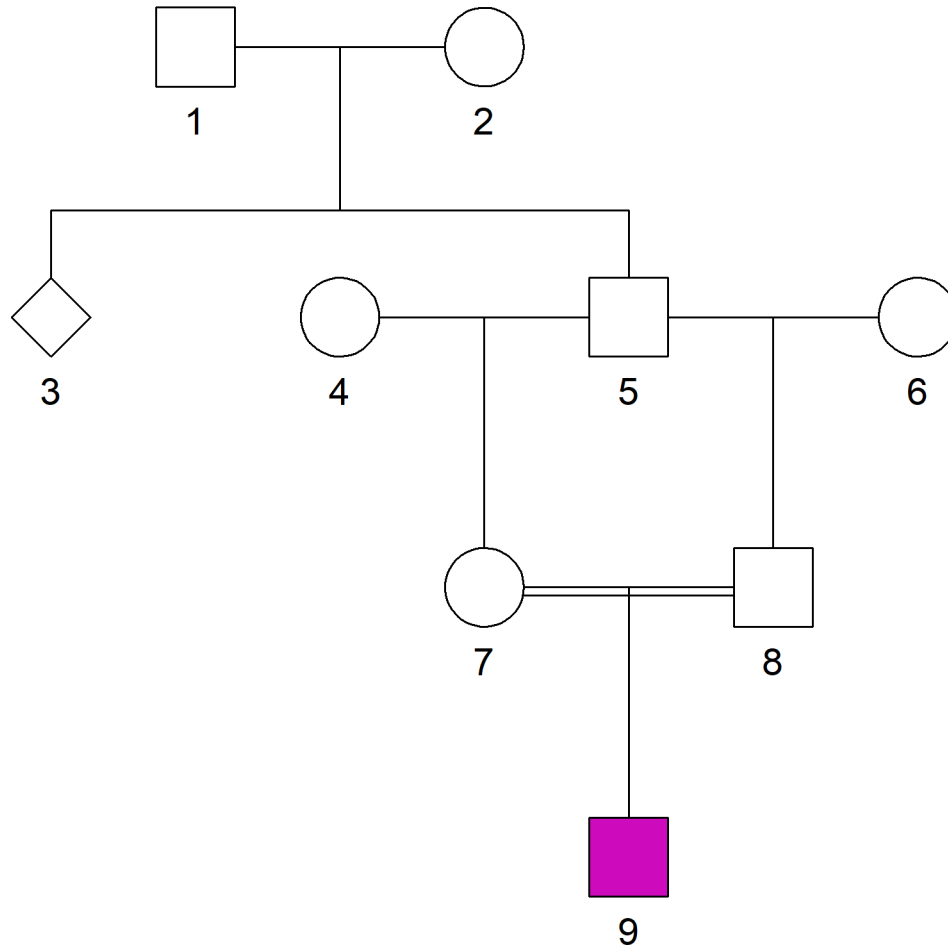
Nonfounders

Parents are included

Leaves

No children included

Conventions and terminology



□ = male

○ = female

◇ = unknown

Founders

No parents included
in the pedigree

Nonfounders

Parents are included

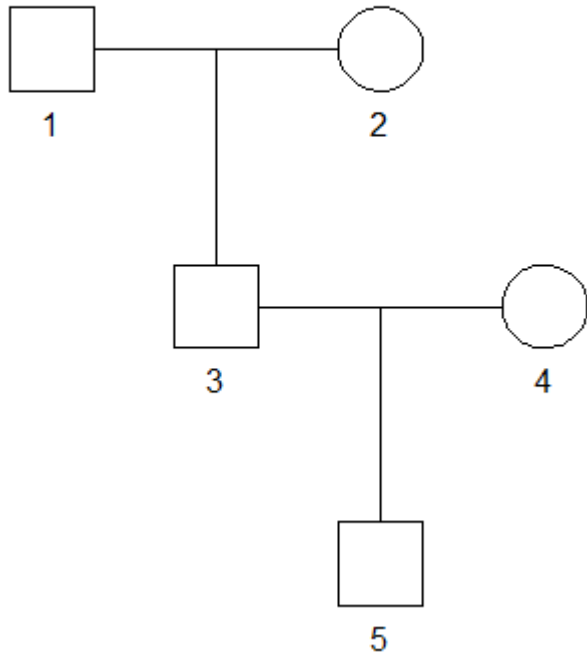
Leaves

No children included

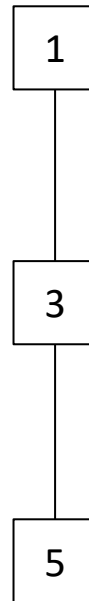
Inbred

Children of related
parents

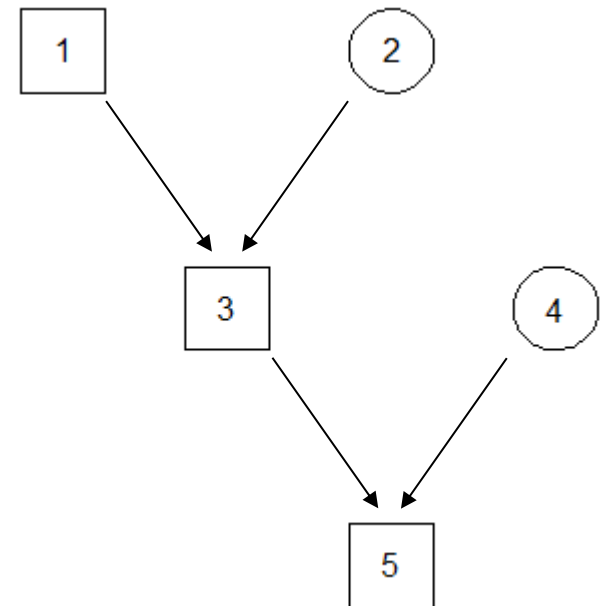
Alternative ways of drawing pedigrees



Standard



Simplified

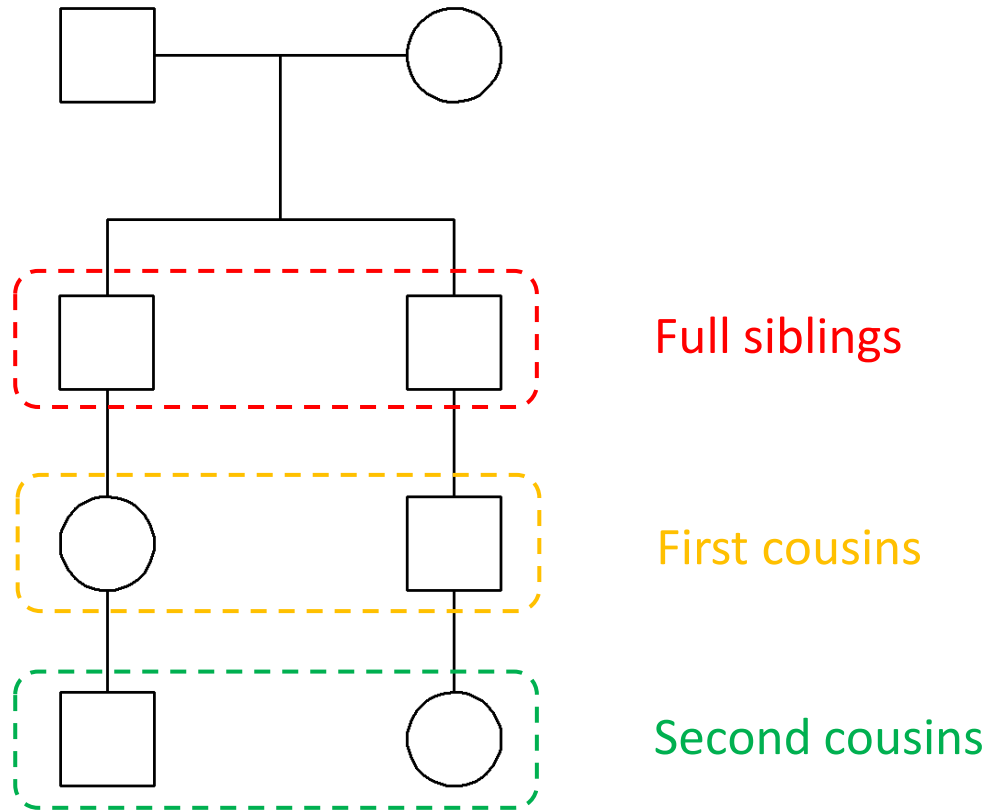


Directed acyclic graph

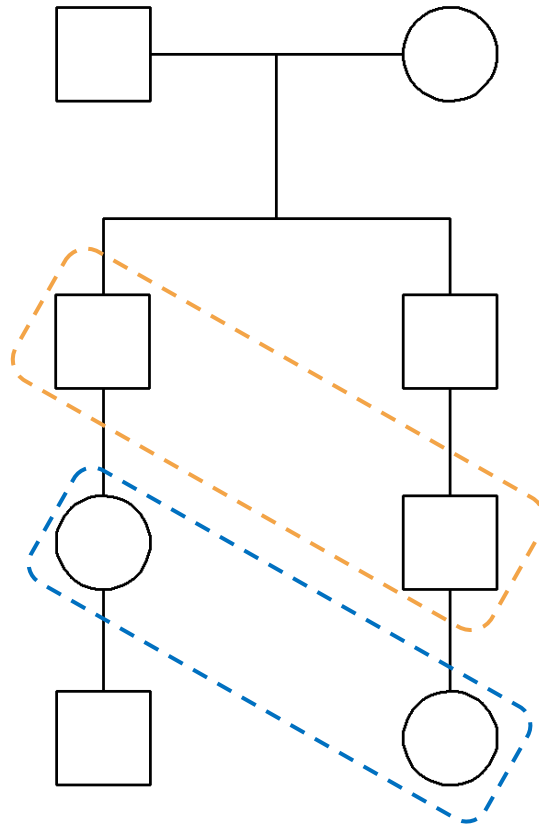
Some common relationships

(and some less common...)

Cousin relationships



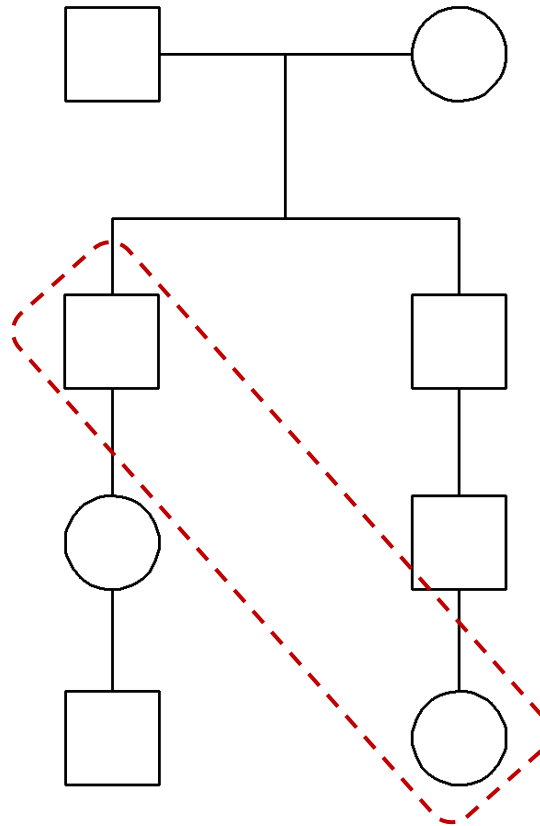
Cousin relationships



Uncle - nephew
(*avuncular*)

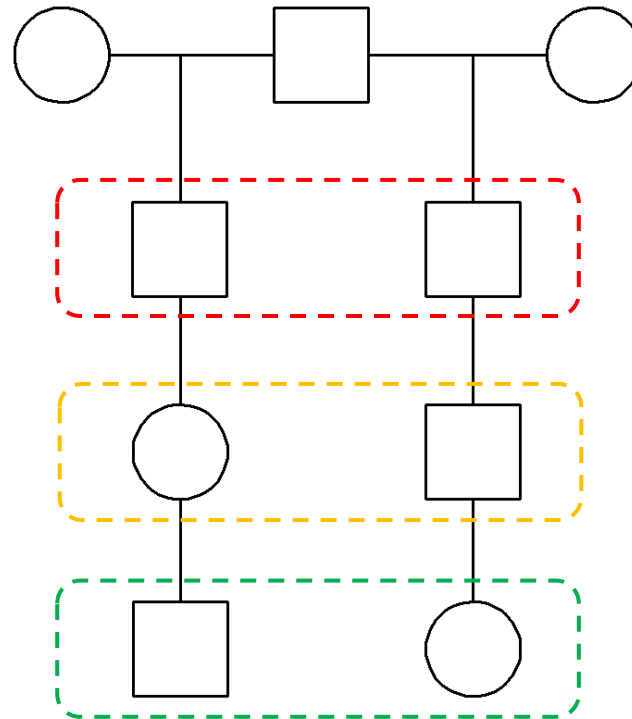
First cousins
once removed

Cousin relationships



Great-uncle
(or sometimes: *granduncle*)

Half cousin relationships

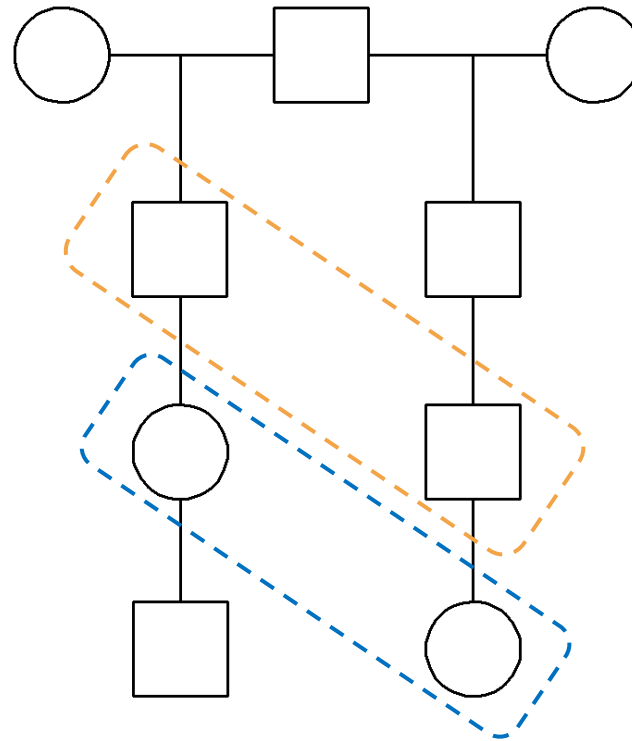


Half siblings

Half first cousins

Half second cousins

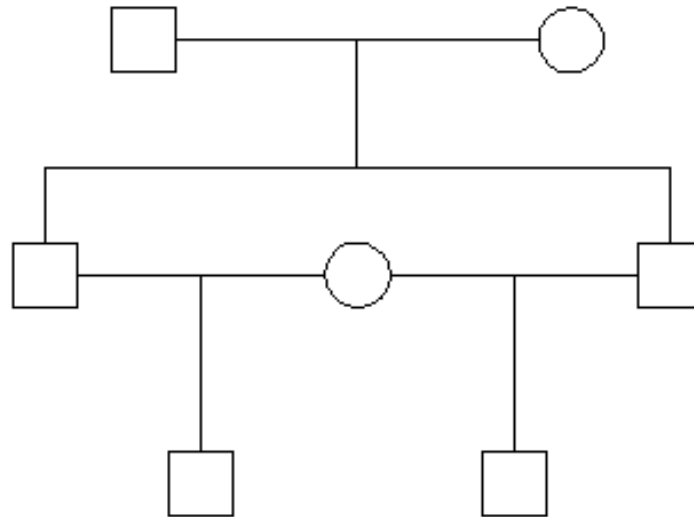
Half cousin relationships



Half-uncle - half-nephew
(*half avuncular*)

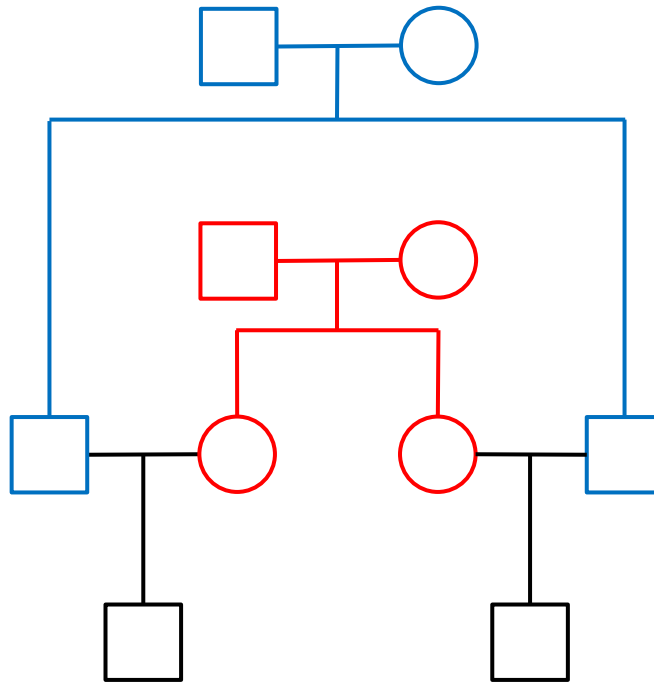
Half first cousins
once removed

Double relationships



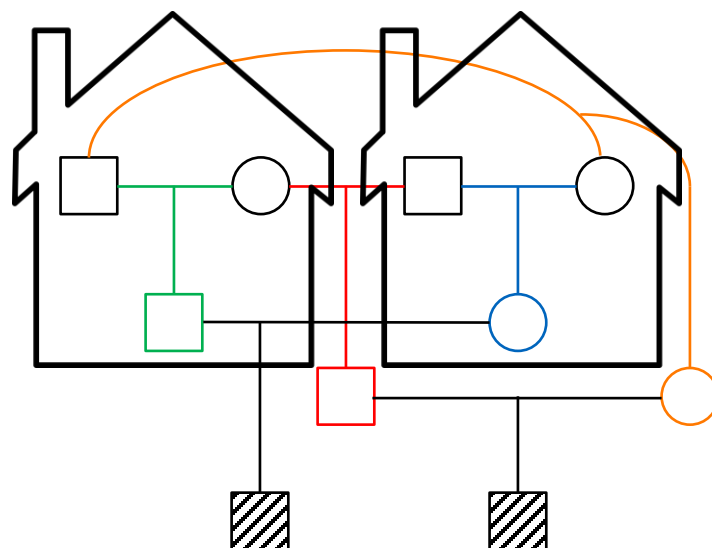
First cousins + half siblings = $3/4$ siblings

Double relationships



Double first cousins

The connoisseur's favourite



Quadruple half first cousins!



What software
exists to create
pedigrees?

It depends!

- medical genetics
- forensic genetics
- animal pedigrees
- amateur genealogy

In this course:

- QuickPed



QuickPed: An Interactive Pedigree Creator

New app design!

Discover the **new features**
Or stay with the old version: QuickPed3

Purpose: QuickPed lets you rapidly create attractive pedigree plots, save them as images or text files, and analyse the relationships within them.

Instructions: Choose a suitable start pedigree and modify it by clicking on individuals and using appropriate buttons. For example, to add a male child, select the parent(s) and press the  icon. Check out the [online user manual](#) for various tips and tricks, including an introduction to relatedness coefficients.

Citation: If you use QuickPed in a publication, please cite this paper: Vigeland MD (2022). QuickPed: an online tool for drawing pedigrees and analysing relatedness. *BMC Bioinformatics*, 23. DOI:[10.1186/s12859-022-04759-y](https://doi.org/10.1186/s12859-022-04759-y).

Quick start

Built-in pedigree

Trio

or

Load a ped file



or

Random pedigree

Reset all

Modify

Add



Sex



Style



Fill



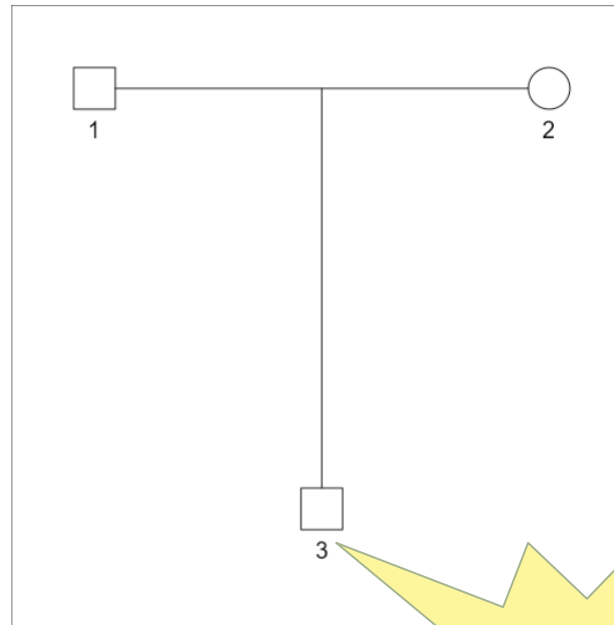
Twins

MZ / DZ

Remove



Undo



Double-click on an individual to add text

Labels

1, 2, 3, ..

I-1, I-2, ..

☒ Show all ☐ Hide all

1

2

3

Update

Plot settings

Width

430

Height

430

Cex

1,4

Symbols

1

Margins

3

Other options (beta)

☐ Straight legs

☐ Arrows

R code

 PNG

 PDF

Relationships



4

DEMO!

Ped file

Include

☒ Headers

☐ Family ID

☐ Affection status

 Save ped file

Part II: Measures of relatedness

Possible answers

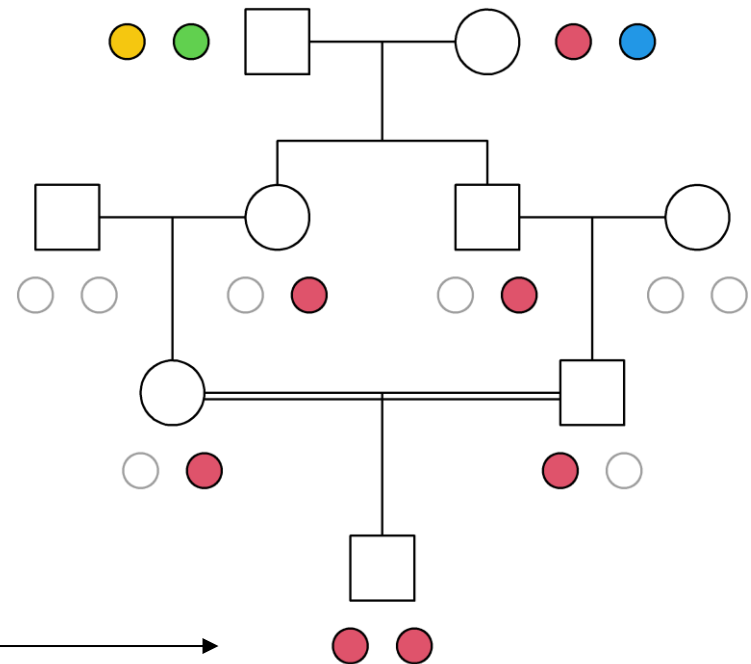
- being connected by family
- having a common ancestor...
(not too far back)
- sharing DNA ...
(more than unrelated people)

To make this precise, we need
some terminology!



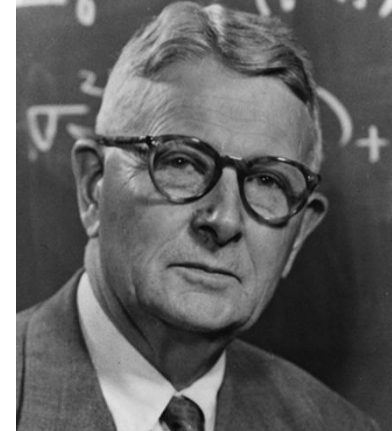
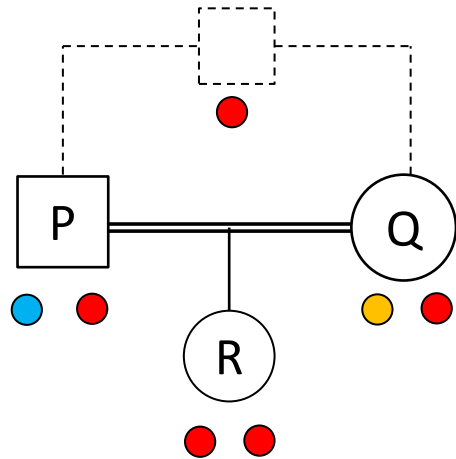
IBD and autozygosity

- identity by descent (IBD)
 - when alleles have a common origin in the given pedigree
- autozygous
 - homozygous; alleles are IBD



Inbreeding coefficient
 $f = Pr(\text{autozygosity})$

Coefficient of kinship/inbreeding



Sewall Wright
(1889 - 1988)

- Wright (1921): The kinship coefficient φ

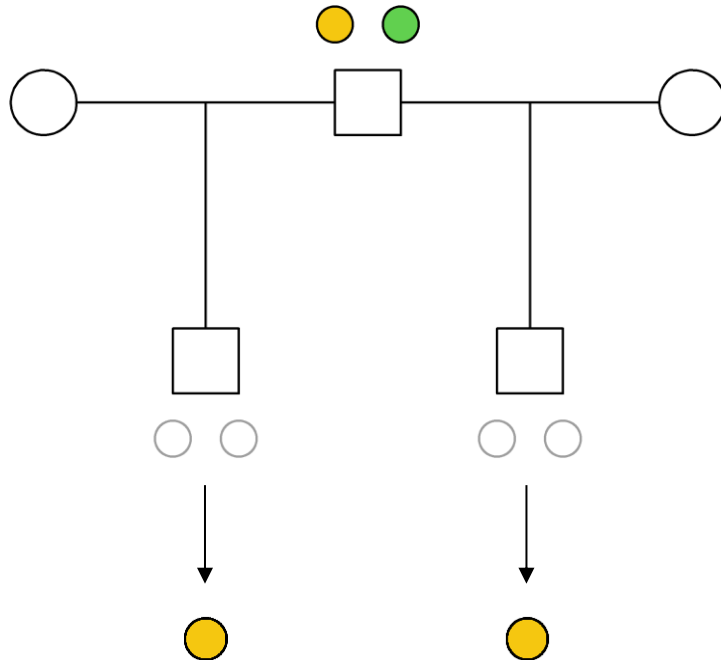
$$\varphi_{P,Q} = \Pr(\text{P and Q emit IBD alleles})$$

$$= \Pr(\text{R is autozygous})$$

$$= f_R$$

Kinship of parents = inbreeding of child

Example 1: Kinship coefficient of half siblings



Kinship coefficient

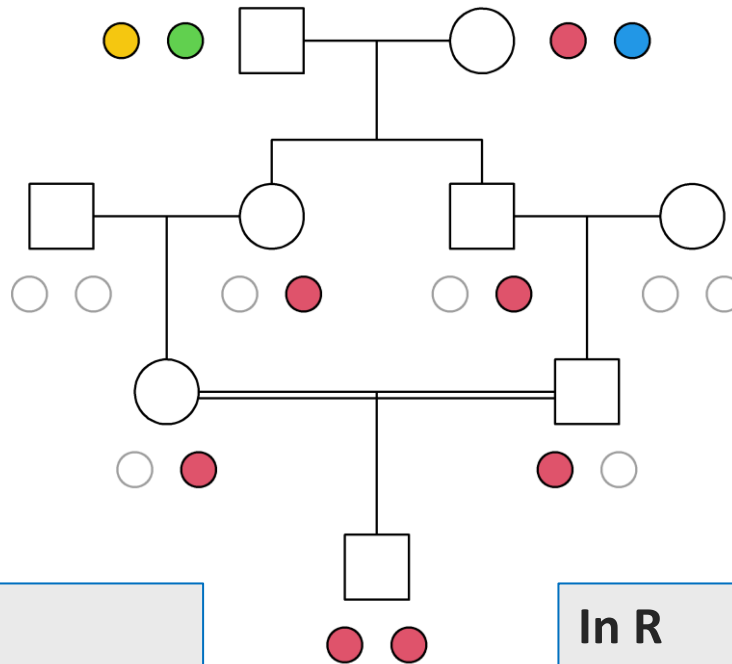
$$\begin{aligned}\varphi &= P(\text{yellow circle from both}) \cdot 2 \\ &= 0.5^4 \cdot 2 \\ &= 1/8\end{aligned}$$

↑
green

Example 2: Inbreeding coefficient

Wright's path formula:

$$\varphi_{P,Q} = \sum_A \sum_v \left(\frac{1}{2}\right)^{|v|+1} (1 + f_A)$$



By hand

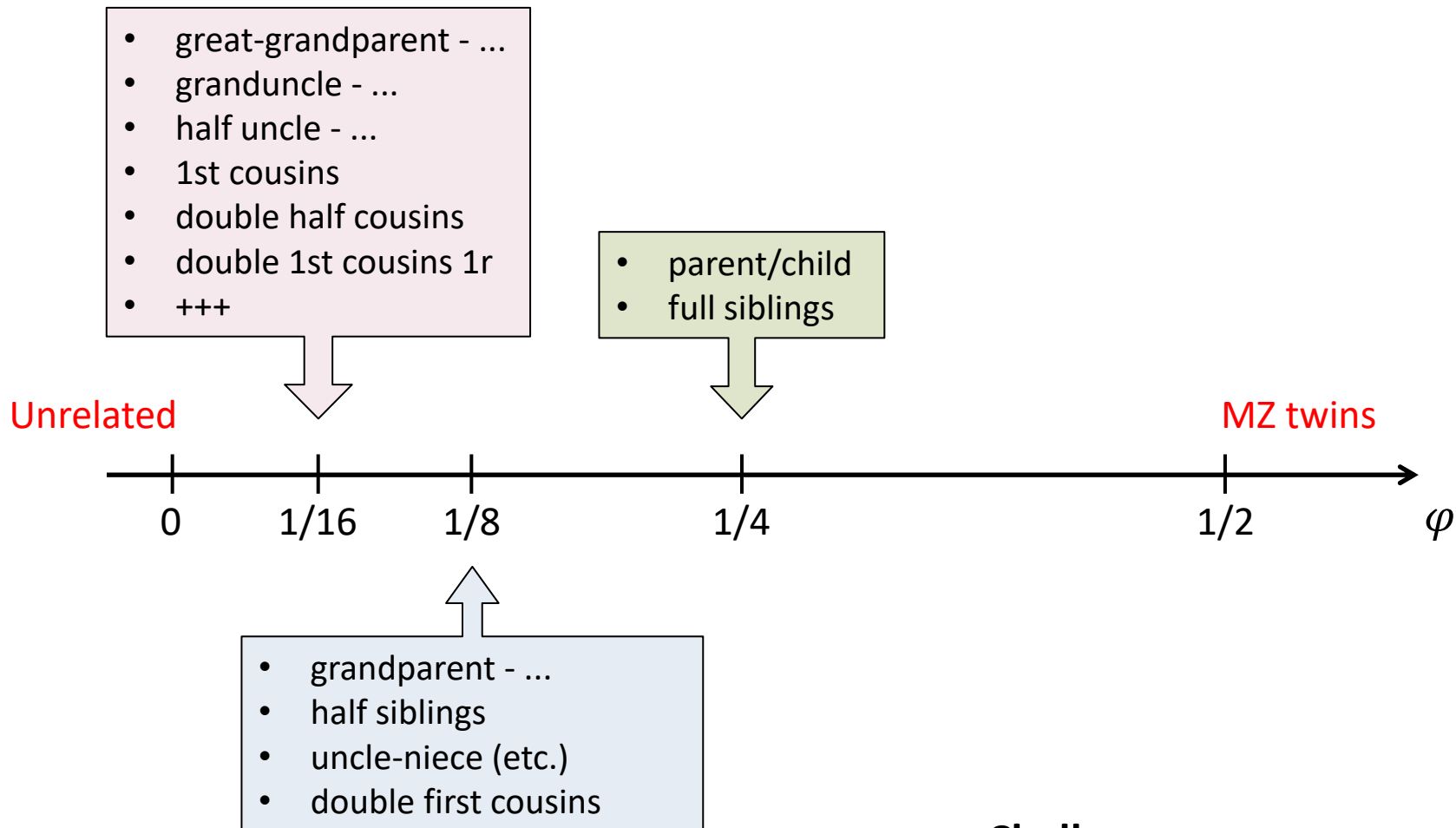
$$\begin{aligned} f &= P(\text{red autozygous}) \cdot 4 \\ &= 0.5^6 \cdot 4 \\ &= 1/16 \end{aligned}$$

↑
other colors

In R

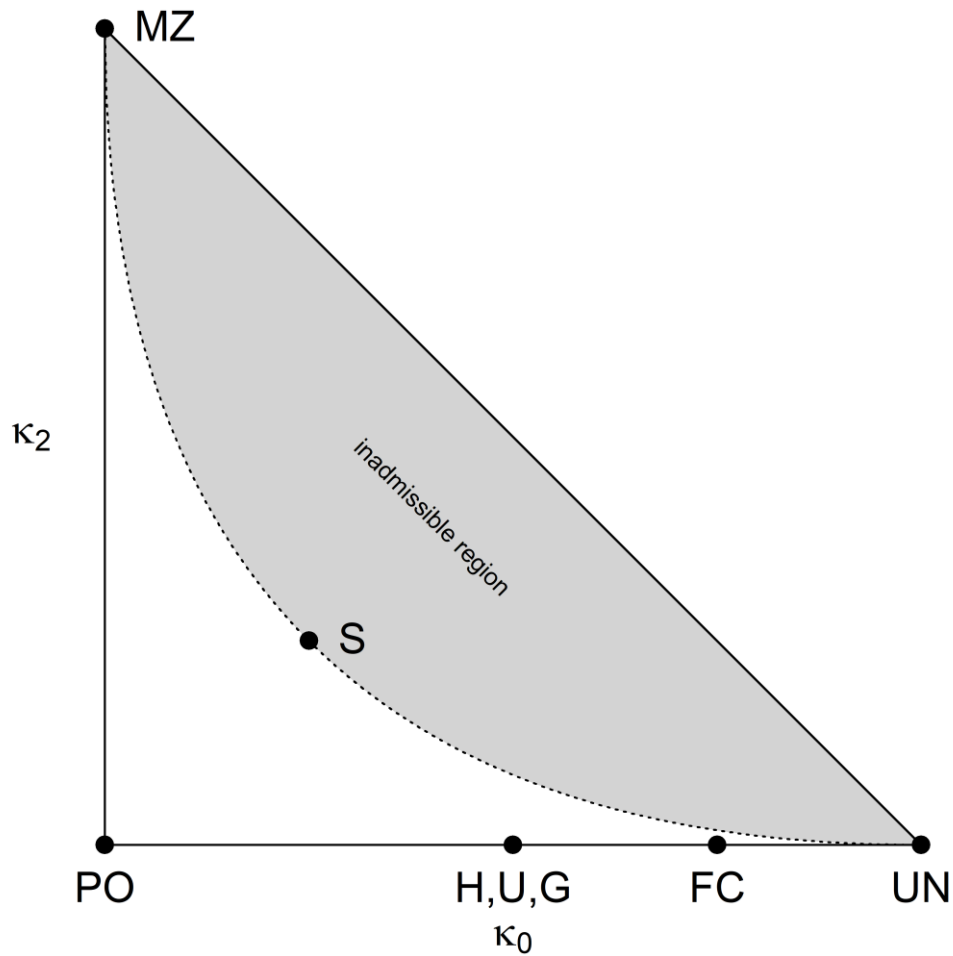
```
> library(pedsuite)

> x = cousinPed(1, child = T)
> inbreeding(x, ids = 9)
[1] 0.0625
```



Challenge:
Many relationships have
the same kinship!

The IBD triangle



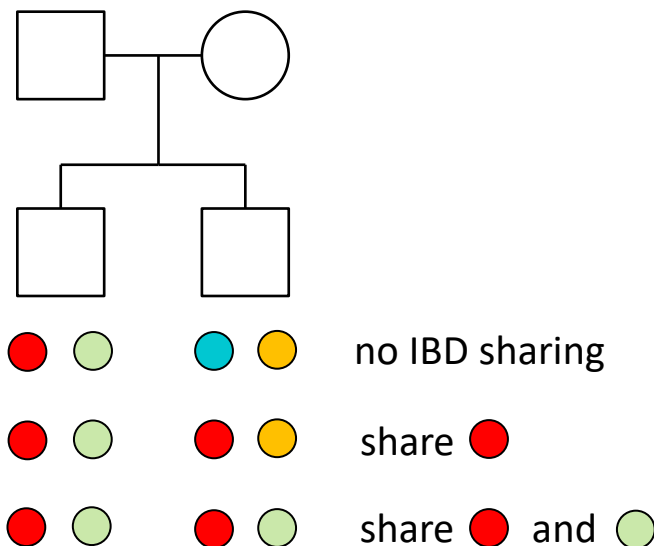
Charles Cotterman
(1914-1989)



Elisabeth Thompson
(1949 -)

IBD coefficients

- Summary so far:
 - Related individuals share alleles by descent (IBD)
 - Kinship coefficient measures the overall amount of IBD sharing
- Natural generalisation:
 - How *many* alleles are IBD in each locus?

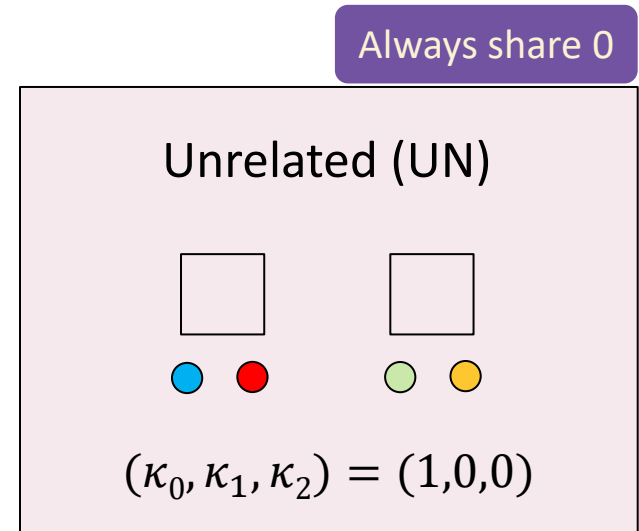
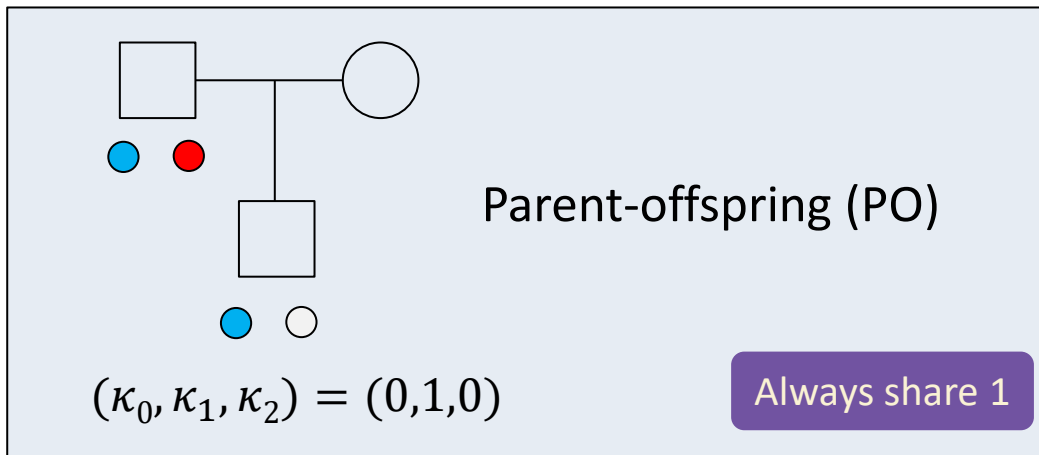
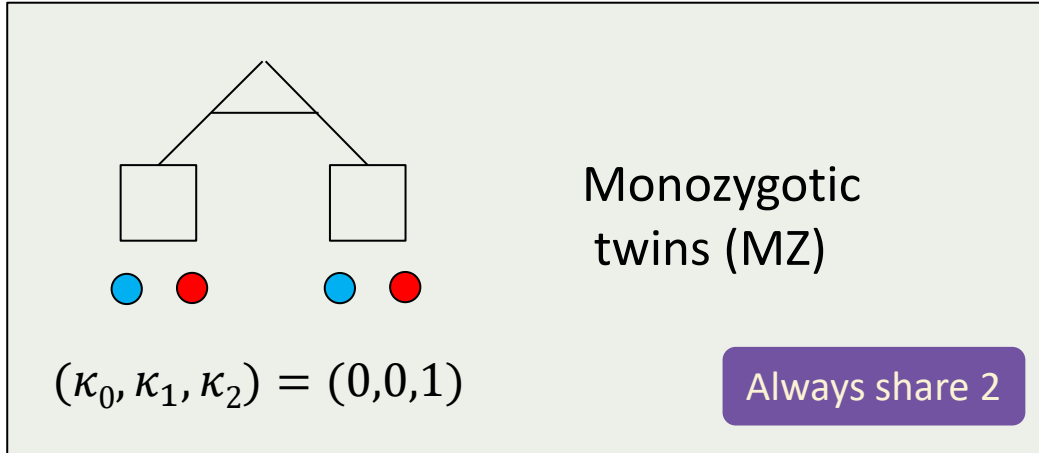


Definition

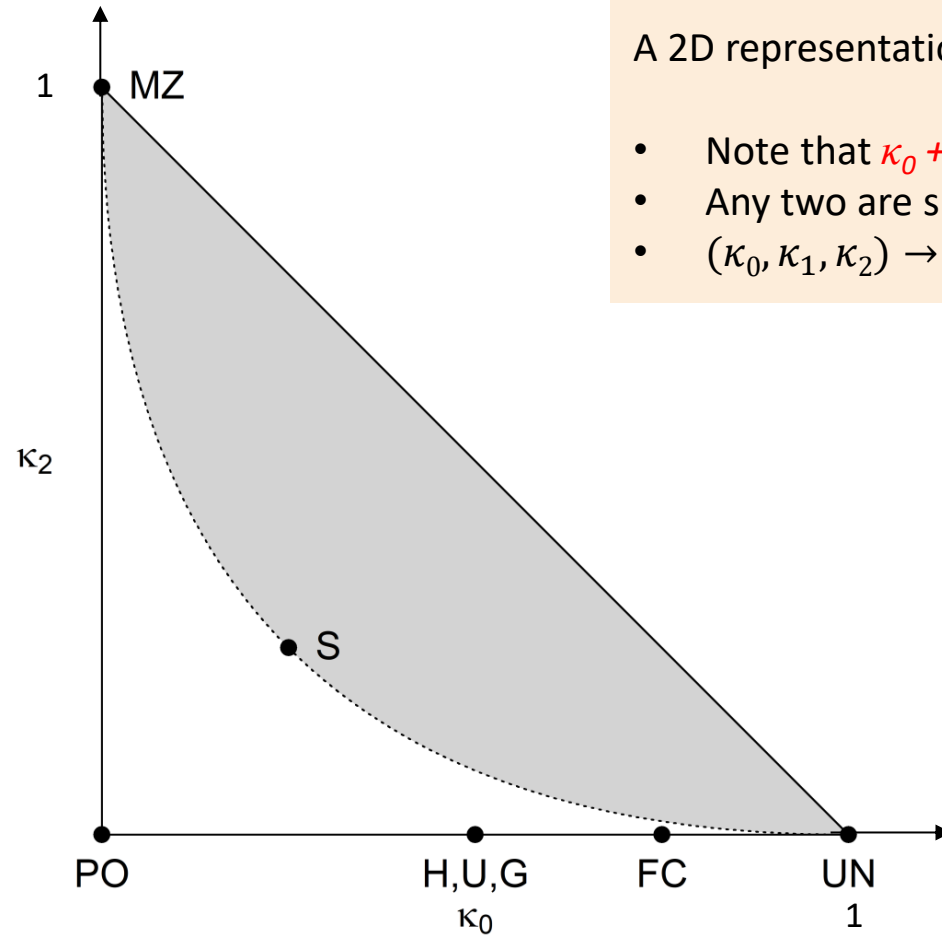
- $\kappa_0 = Pr(0 \text{ alleles IBD})$
- $\kappa_1 = Pr(1 \text{ allele IBD})$
- $\kappa_2 = Pr(2 \text{ alleles IBD})$

(at random autosomal locus)

Three trivial relationships



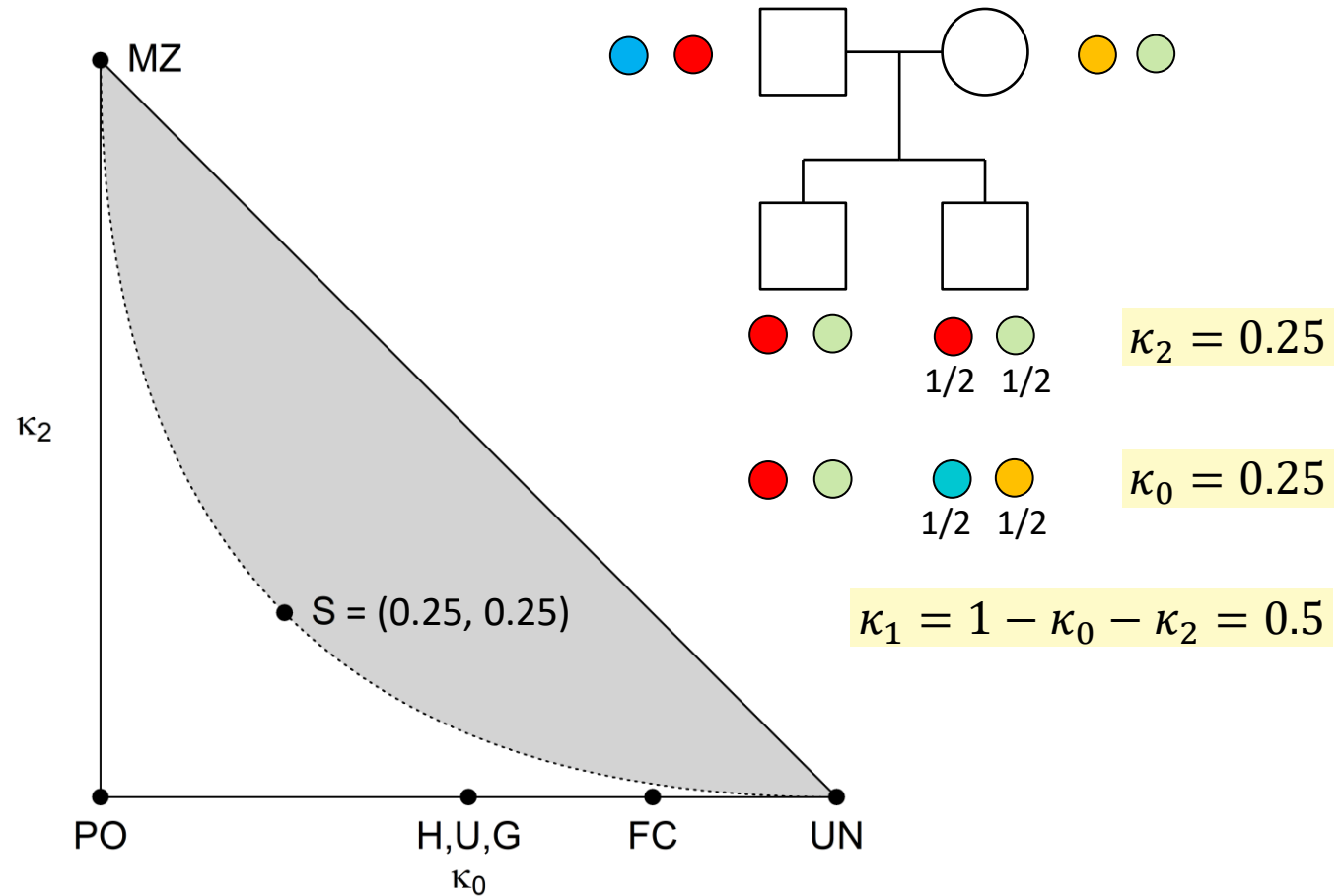
The relatedness triangle



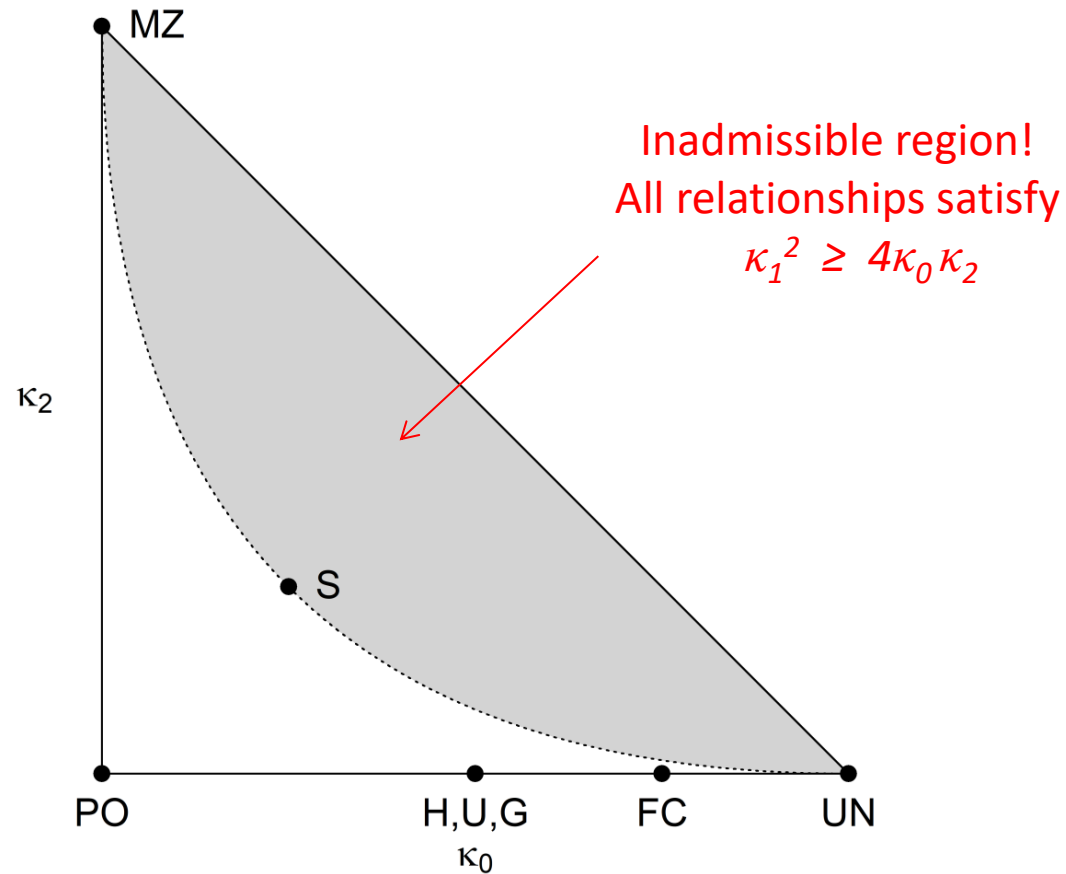
A 2D representation of $(\kappa_0, \kappa_1, \kappa_2)$

- Note that $\kappa_0 + \kappa_1 + \kappa_2 = 1$
- Any two are sufficient
- $(\kappa_0, \kappa_1, \kappa_2) \rightarrow (\kappa_0, \kappa_2)$

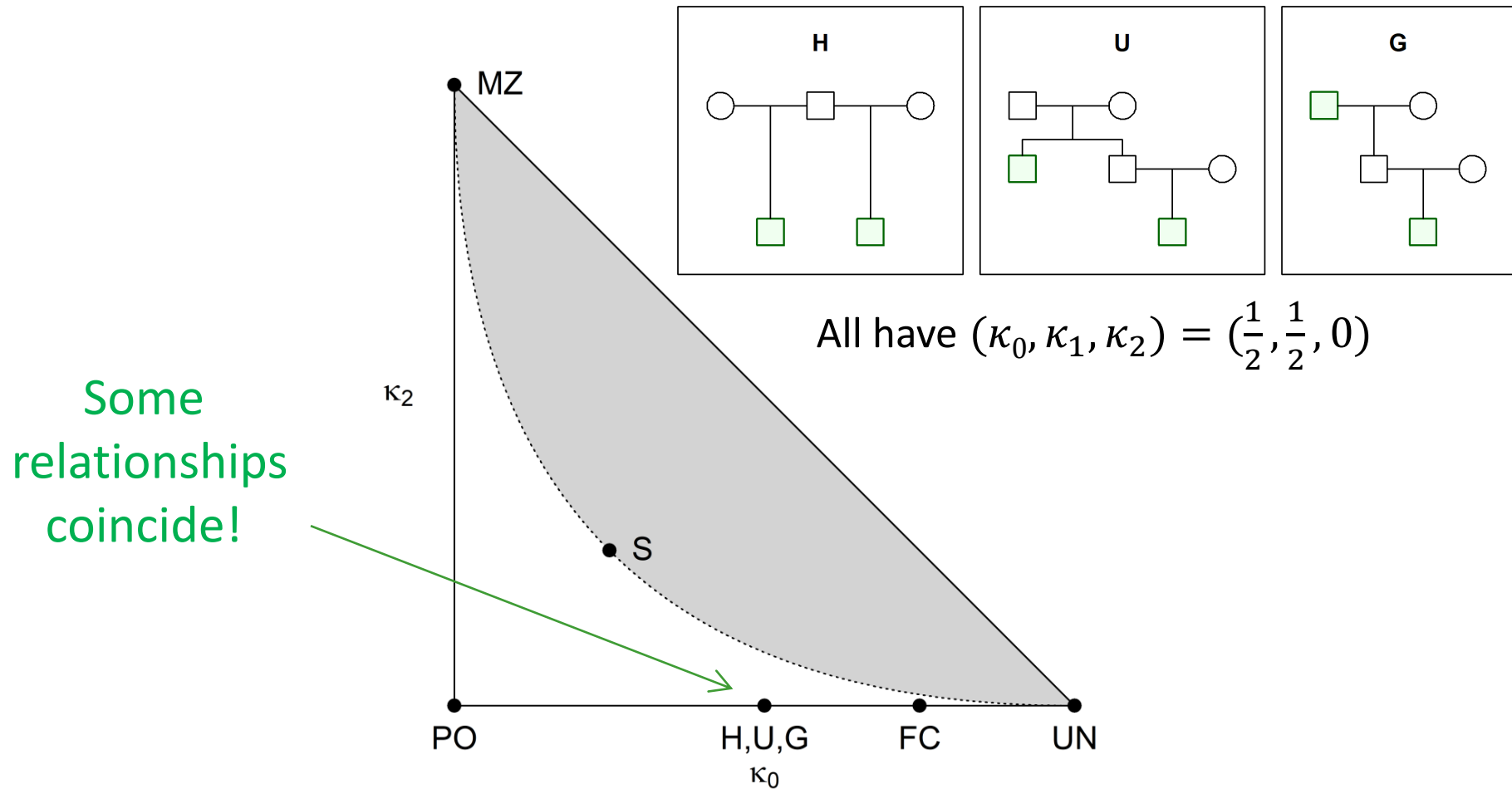
Kappa coefficients of full sibs



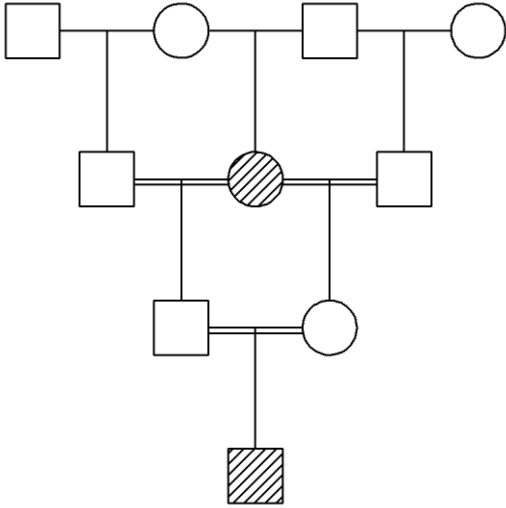
Quirks of the relatedness triangle: 1



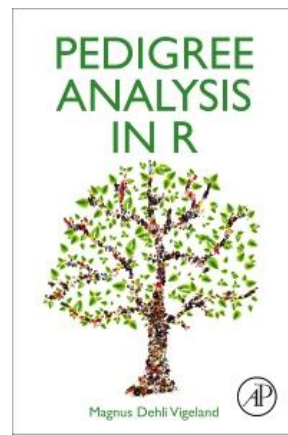
Quirks of the relatedness triangle: 2



A word of caution



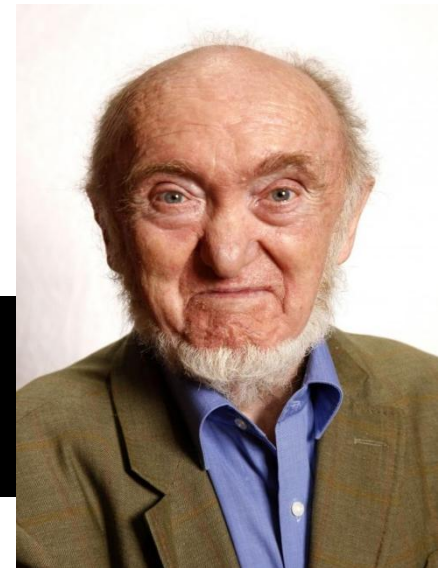
K is only defined for **non-inbred** individuals.
For the whole story, we need 9 coefficients!



Jacquard's identity coefficients

Activity coefficients

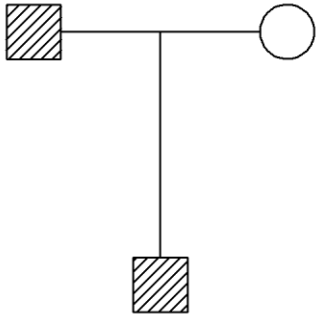
Not suitable for a basic course ...



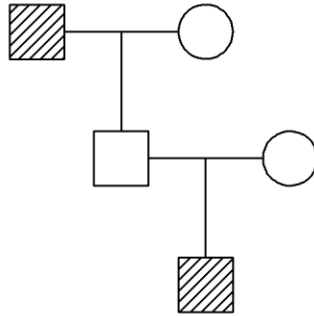
Albert Jacquard
(1925 - 2013)

Another word of caution: *Degree of relatedness*

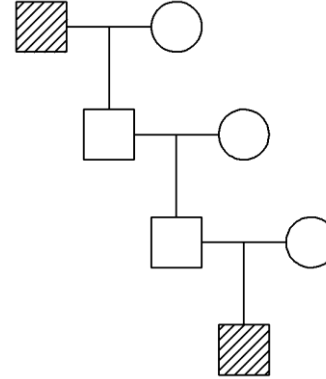
deg = 1



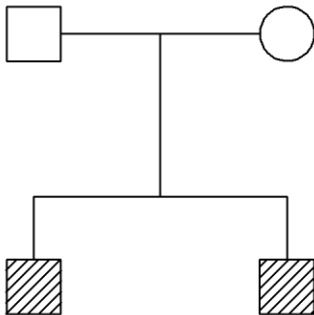
deg = 2



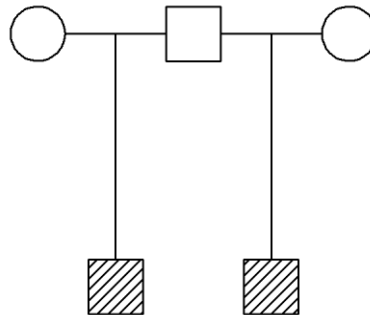
deg = 3



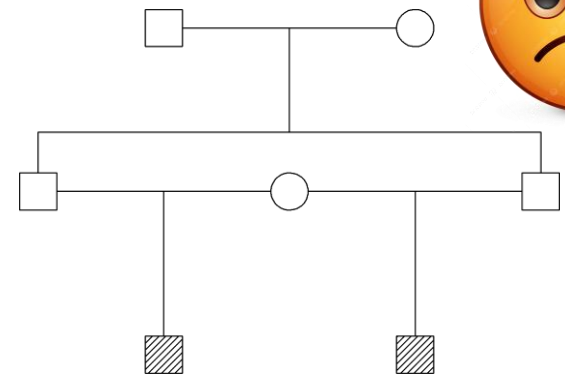
deg = ?



deg = ?



deg = ?



Another word of caution: *Degree of relatedness*

Degree of relationship			Relationship
Genetics	Roman law	Canon law	
0	0	0	self
0	2	1	parent / mother / father / daughter / son [10]
1	1	1	parent / mother / father / daughter / son [10]
1	3	2	parent / mother / father / daughter / son [10]
1	2	2	parent / mother / father / daughter / son [10]
2	2	2	parent / mother / father / daughter / son [10]
2	3	2	parent / mother / father / daughter / son [10]
2	2	2	parent / mother / father / daughter / son [10]
2	4	2	parent / mother / father / daughter / son [10]
3	4	2	parent / mother / father / daughter / son [10]
3	3	3	parent / mother / father / daughter / son [10]
3	4	3	parent / mother / father / daughter / son [10]
3	3	2	parent / mother / father / daughter / son [10]
3	5	3	parent / mother / father / daughter / son [10]
3	6	3	parent / mother / father / daughter / son [10]

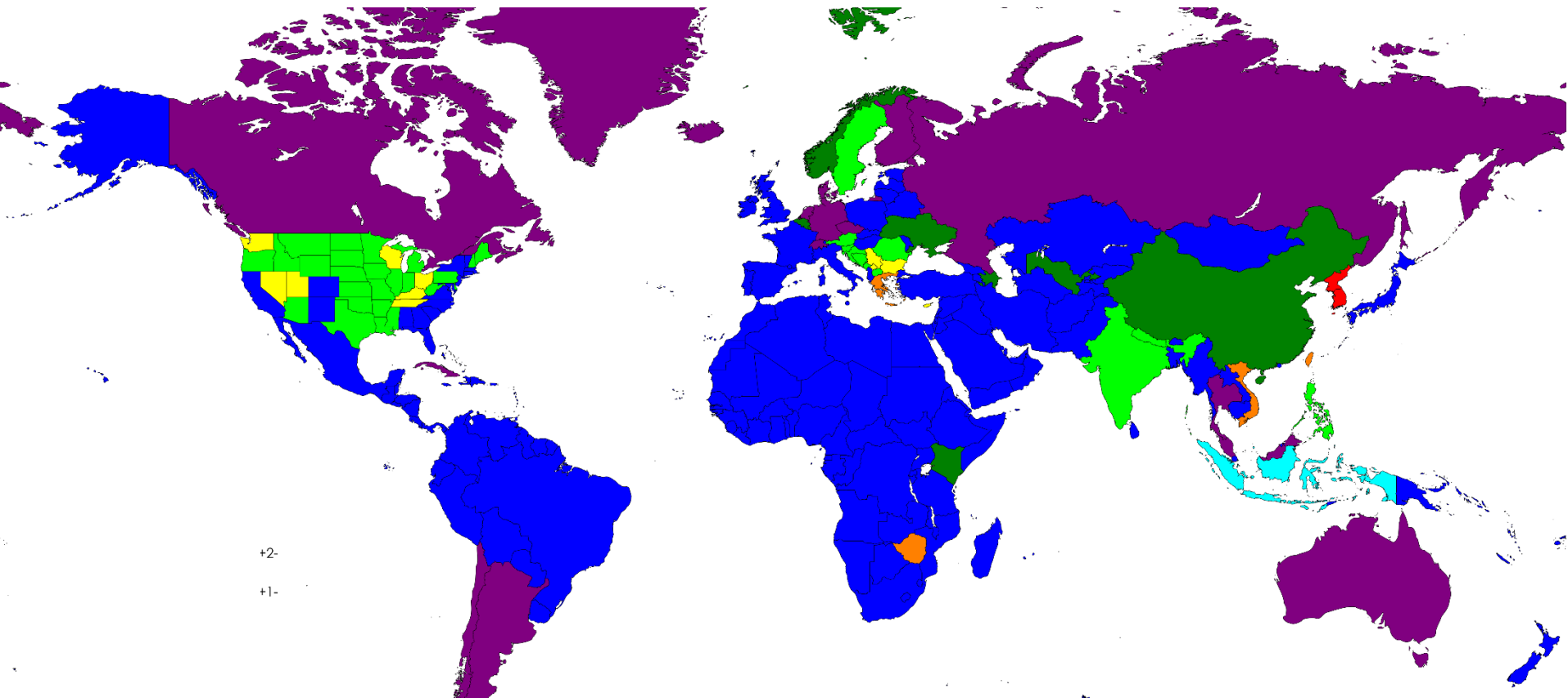


Source: Wikipedia *Coefficient of relationship*

Moral

- If you use the word **degree**, make it very clear what you mean.
 - Give examples!
-
- Here is a good example from Wikipedia 

Prohibited degree of kinship for marriage



+2-

+1-

- Within degree 8 (third cousin)
- Within degree 6 (second cousin)
- Within degree 5 (first cousin once removed)
- Within degree 4 (first cousin/granduncle-grandniece)
- Within degree 3 (uncle-niece), and for degree 4, first cousin is prohibited but granduncle-grandniece is not
- Within degree 3 (uncle-niece), and for degree 4, granduncle-grandniece is prohibited but first cousin is not
- Within degree 3 (uncle-niece), i.e. allow [first cousin marriage](#) and granduncle-grandniece marriage
- Within degree 2 (sibling), i.e. allow [avunculate marriage](#) (uncle-niece marriage)

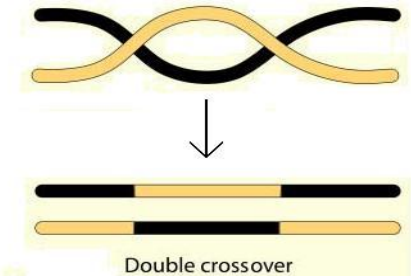
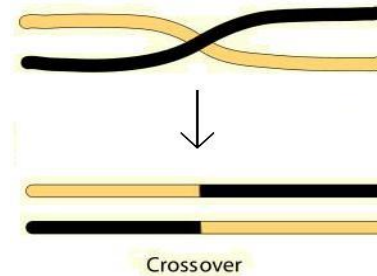
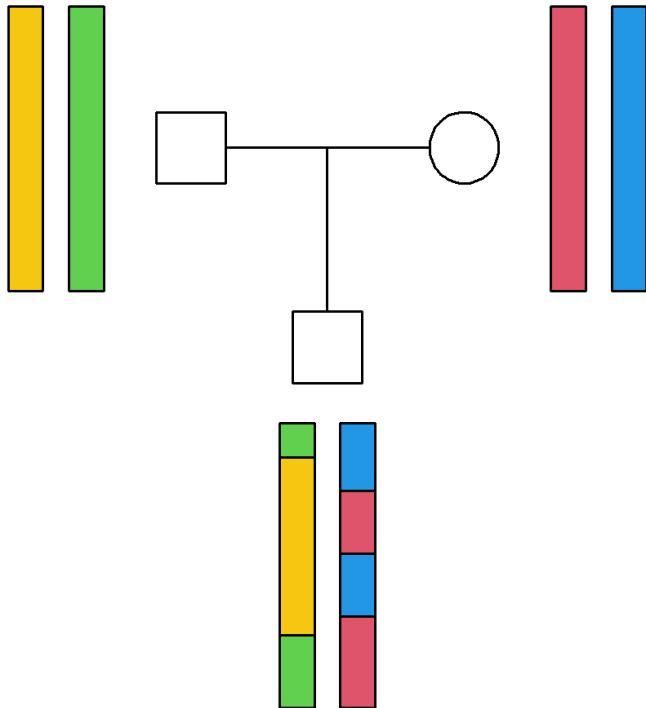
Source: Wikipedia, *Prohibited degree of kinship*

Part III: Realised relatedness

*Why are some siblings
more alike than others?*

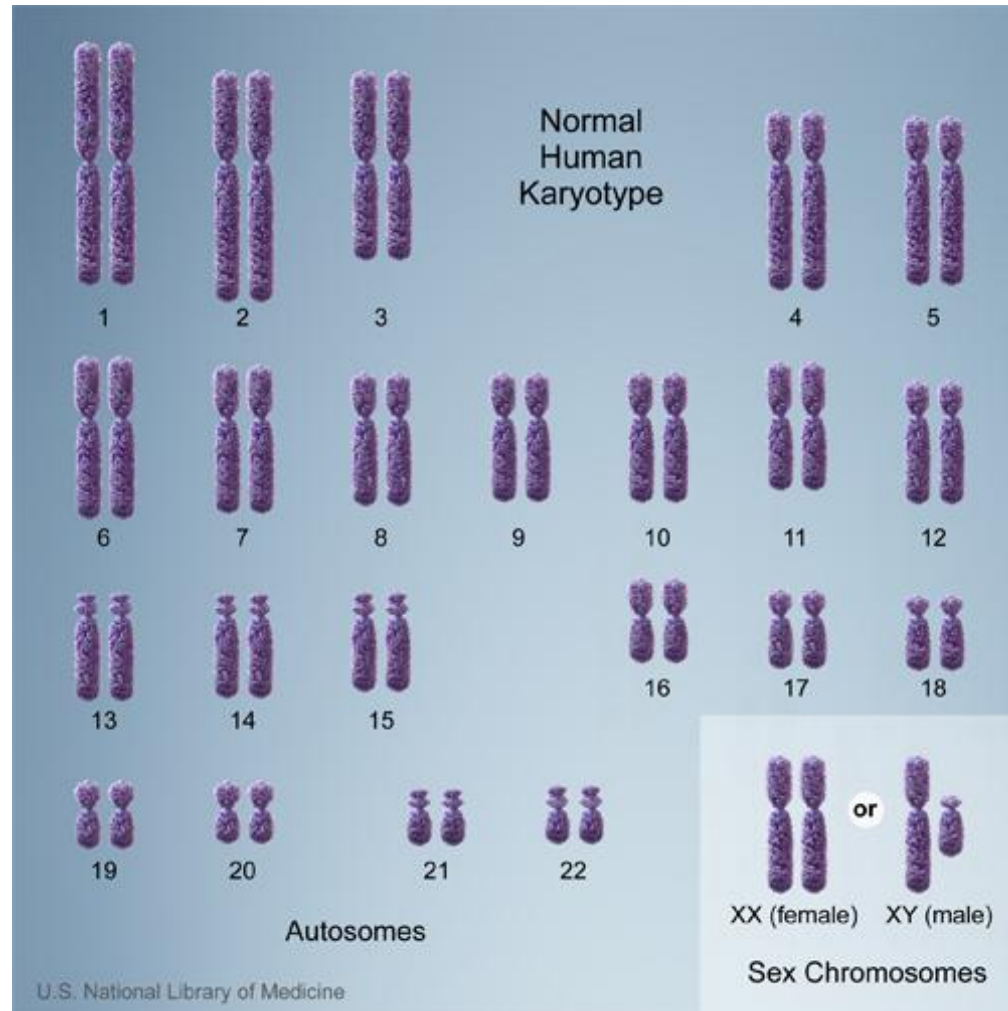


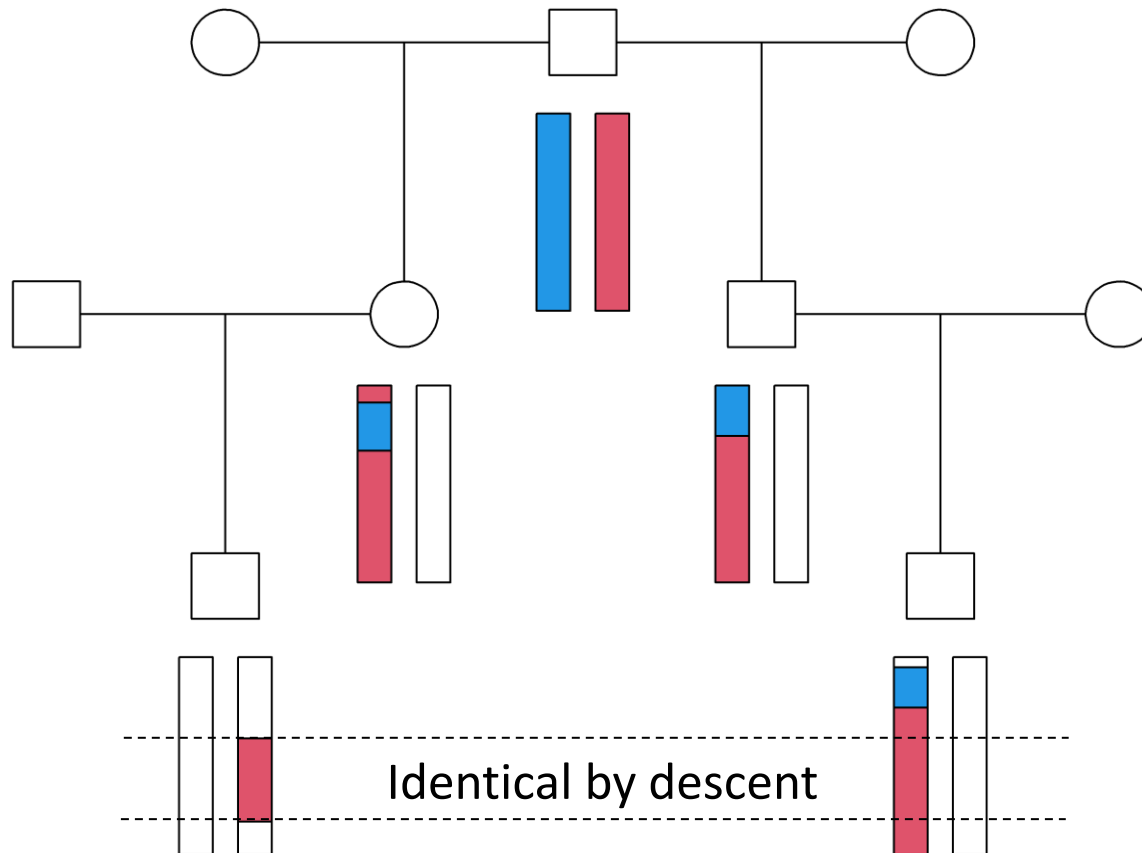
Meiotic recombination



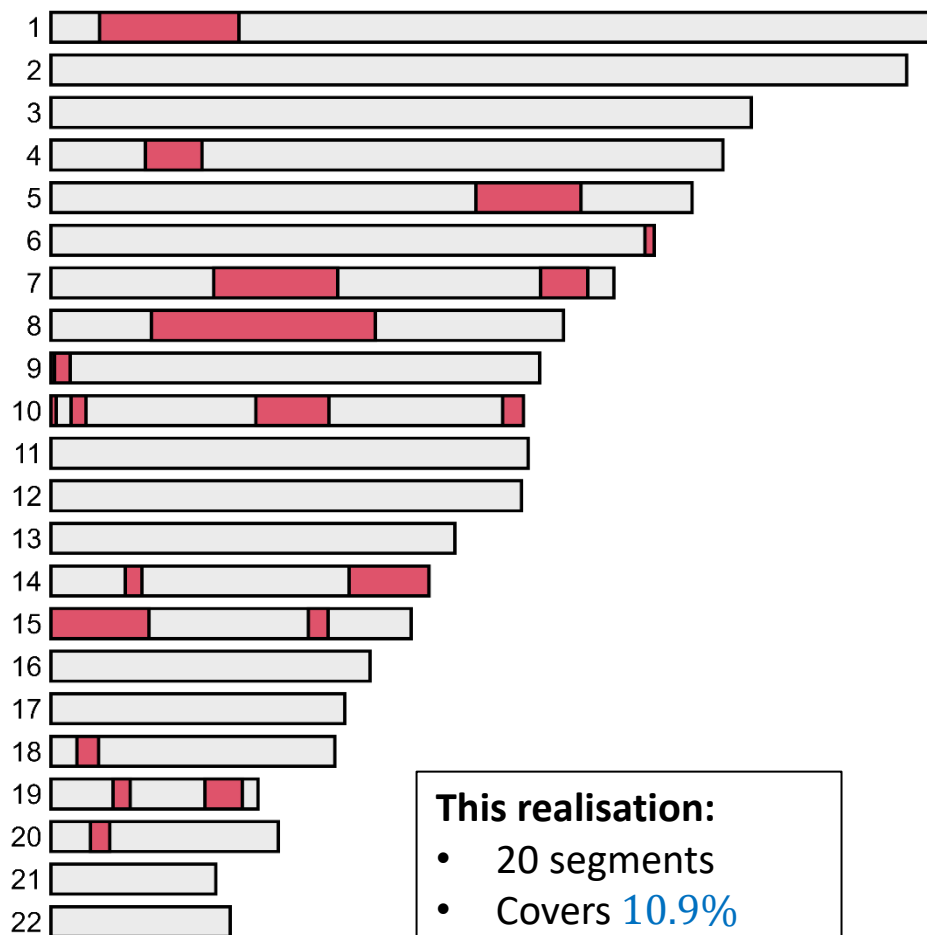
- **Genetic distance** between two loci:
= *average # crossovers per meiosis*
- Units:
 - 1 Morgan (M) = 1 crossover per meiosis
 - 1 centiMorgan (cM) = 0.01 M
- The human genome: Ca 30 Morgan

Rule of thumb: One crossover per chromosome arm



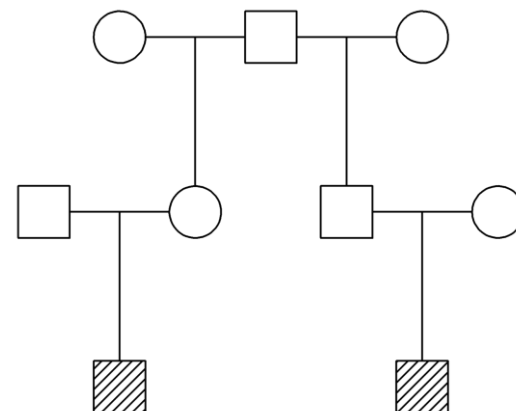


Actual IBD sharing



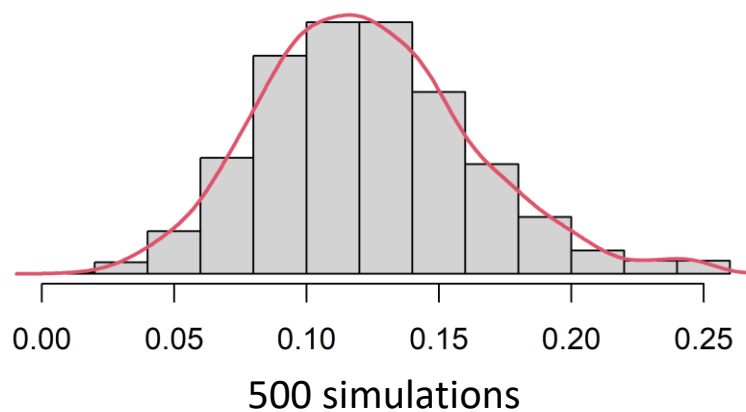
This realisation:

- 20 segments
- Covers 10.9%

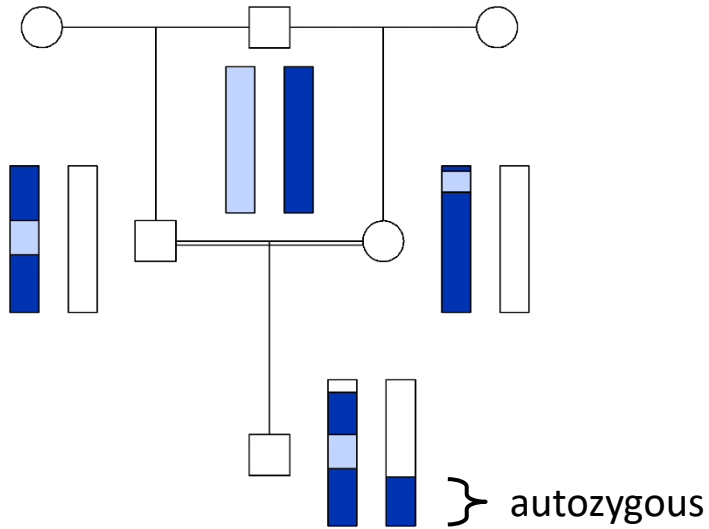
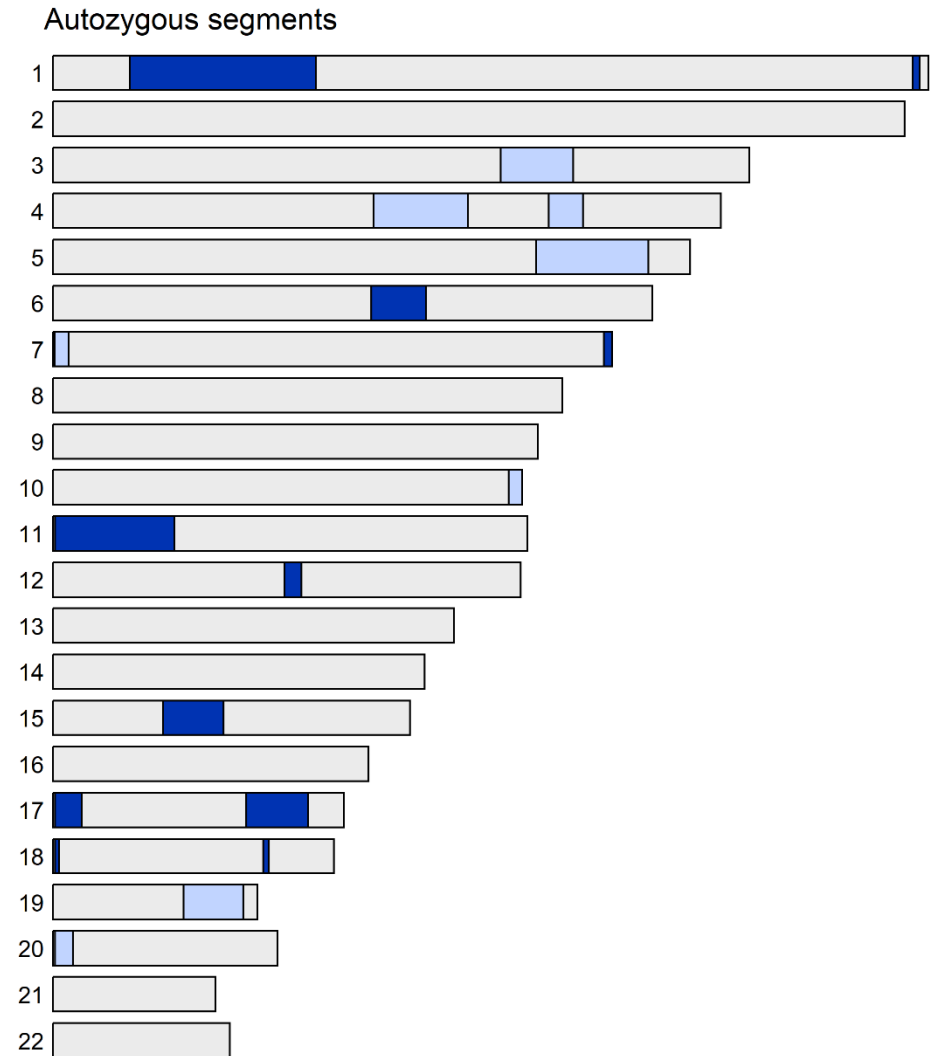
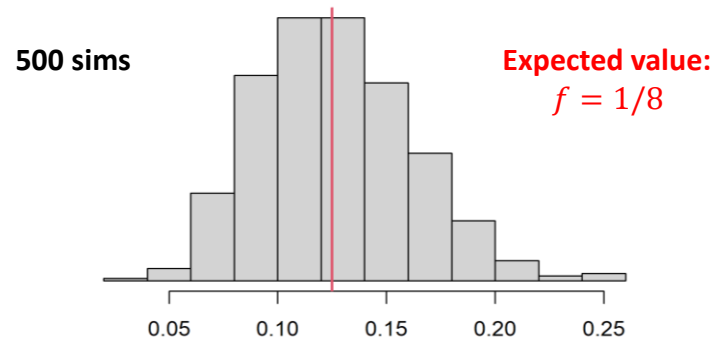


Half first cousins

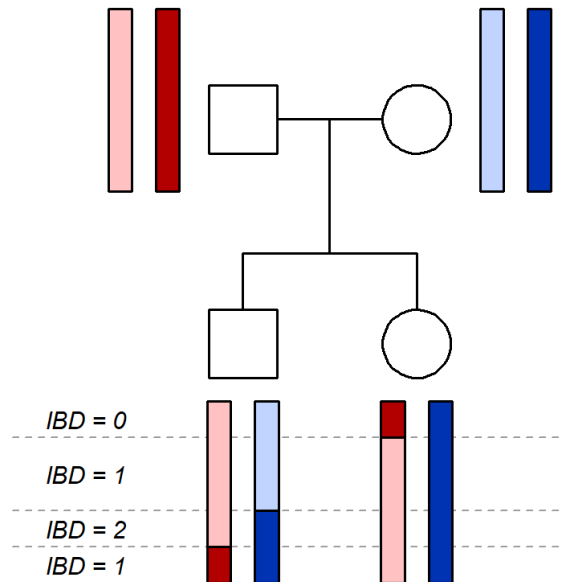
Expected sharing: $\kappa_1 = 1/8 = 12.5\%$



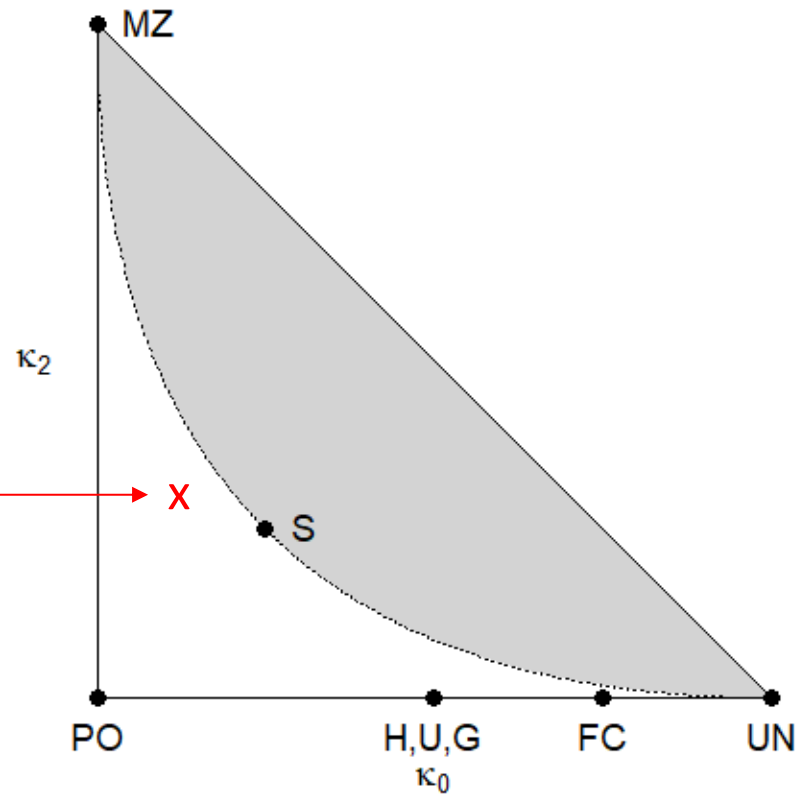
Realised inbreeding

 f_R = autozygous fraction of genome

Realised IBD coefficients



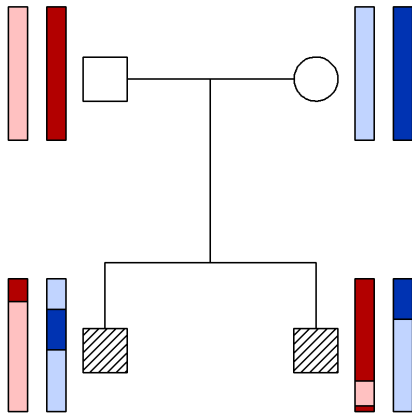
Realised IBD coefficients:
Proportions of genome with IBD = 0, 1, 2



Variation in realised IBD coefficients



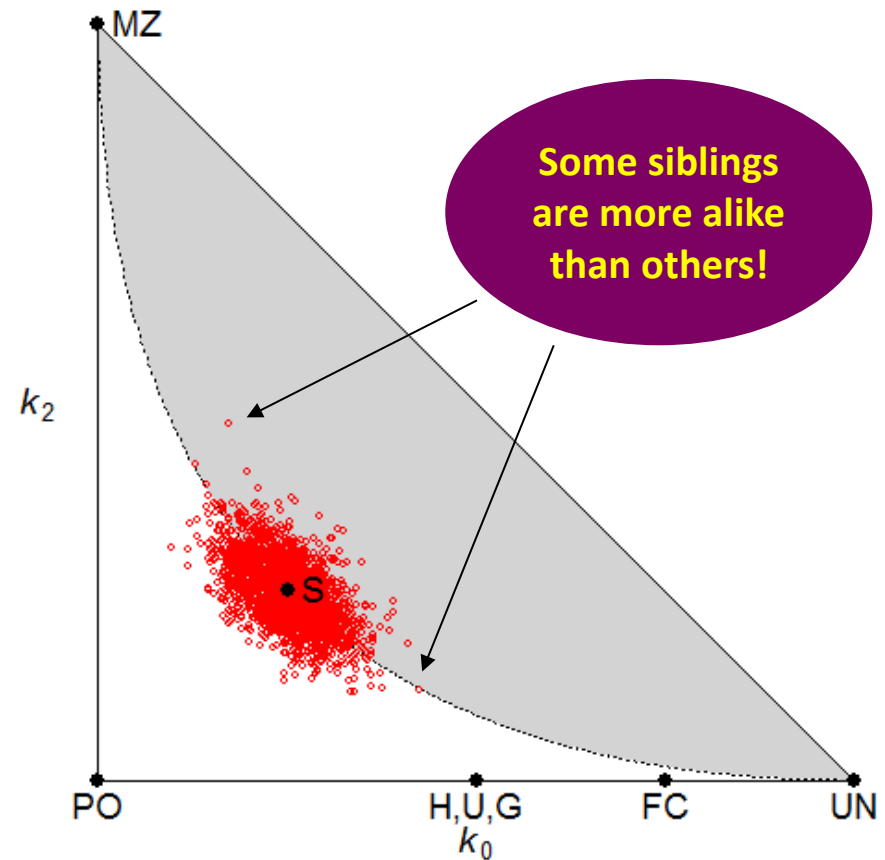
1000 simulations



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

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

> showInTriangle(k)
```



The variation depends on genome length



Human:

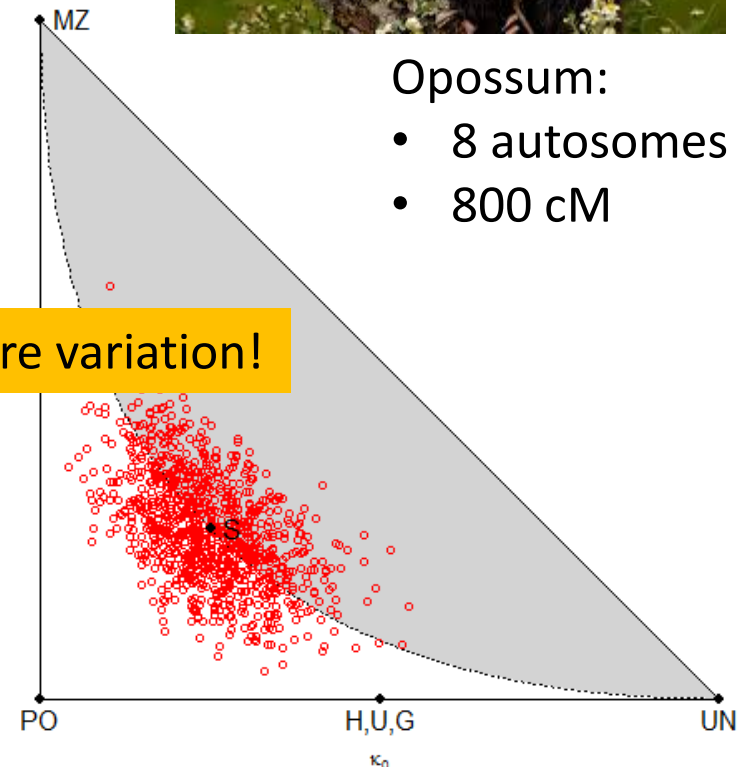
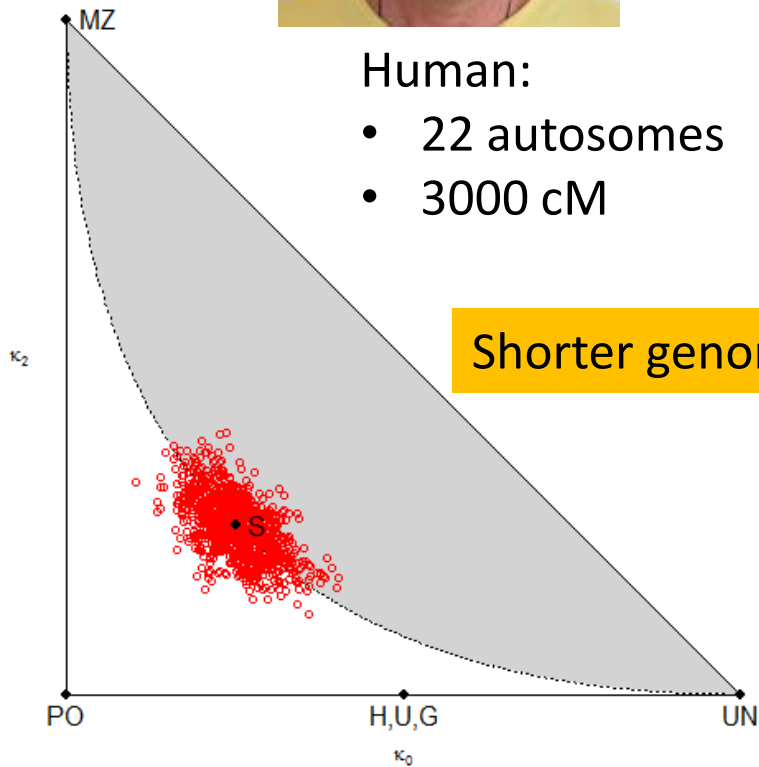
- 22 autosomes
- 3000 cM



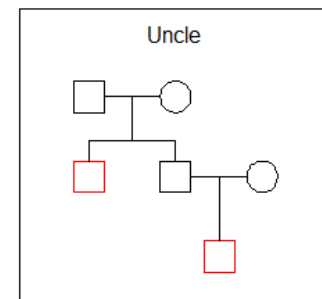
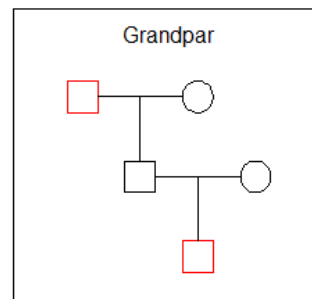
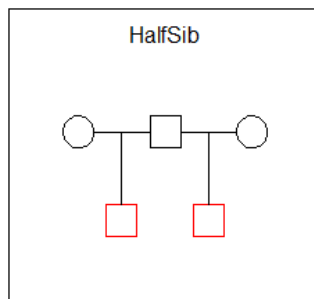
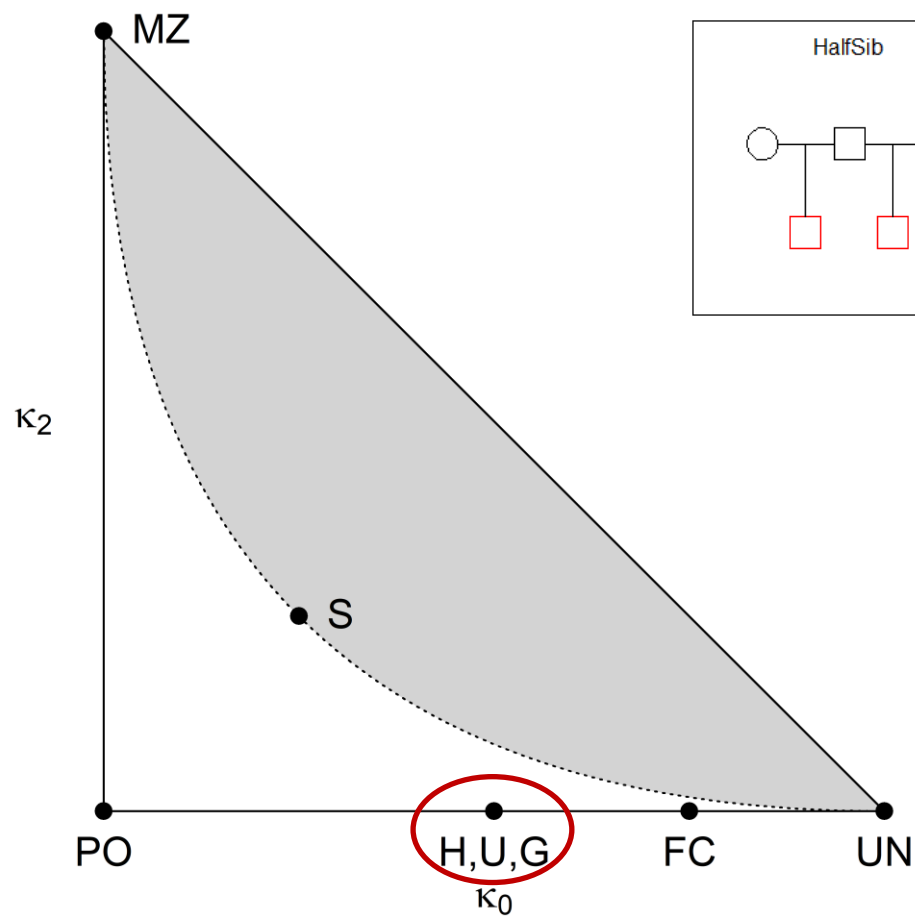
Opossum:

- 8 autosomes
- 800 cM

Shorter genome = more variation!



Recall these "indistinguishable" relationships

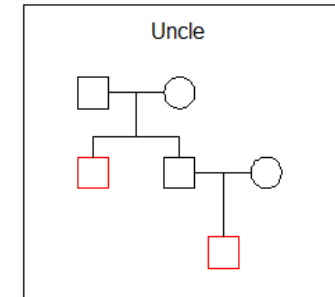
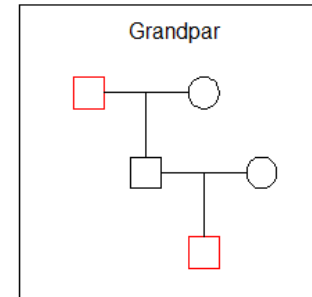
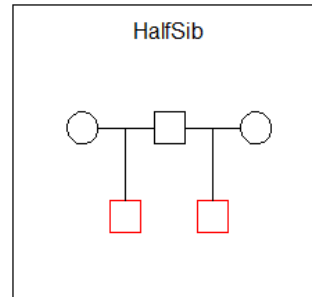
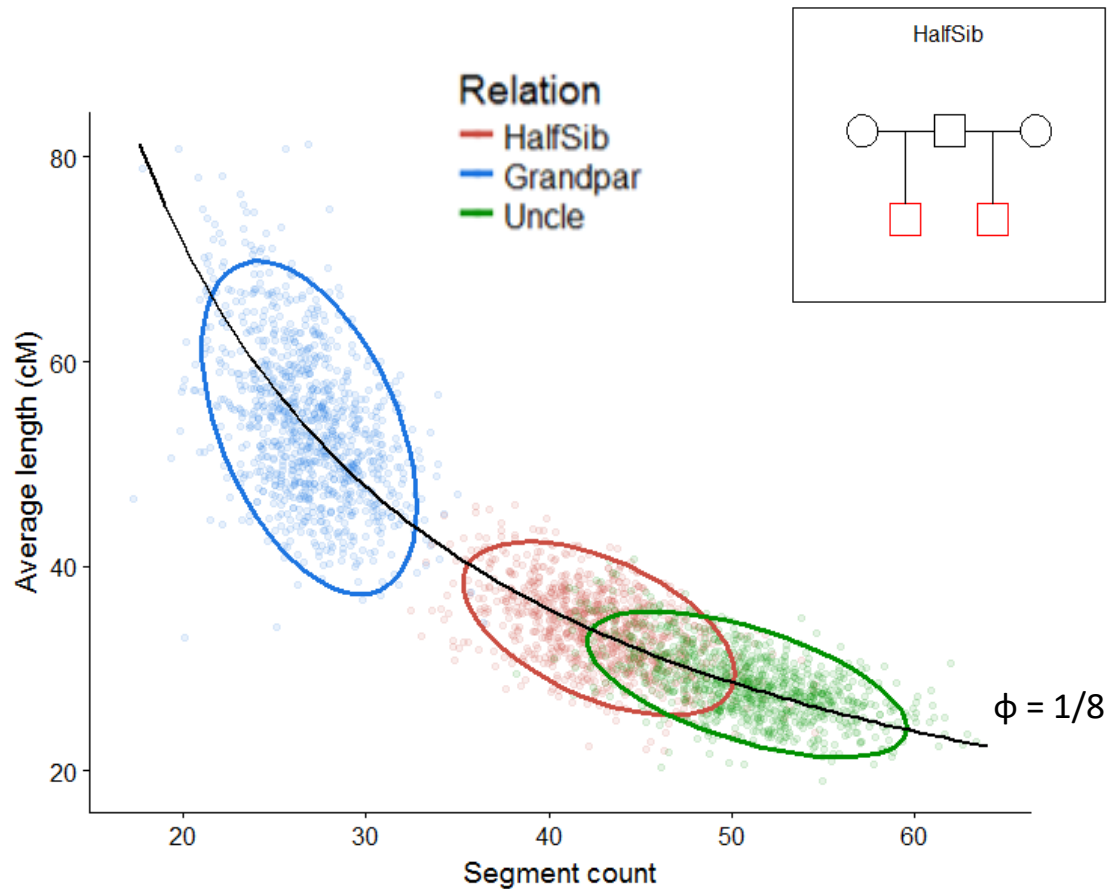
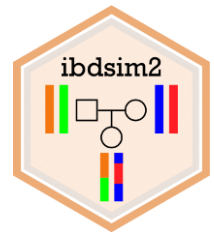


$$\kappa_0 = 0.5$$

$$\kappa_1 = 0.5$$

$$\kappa_2 = 0$$

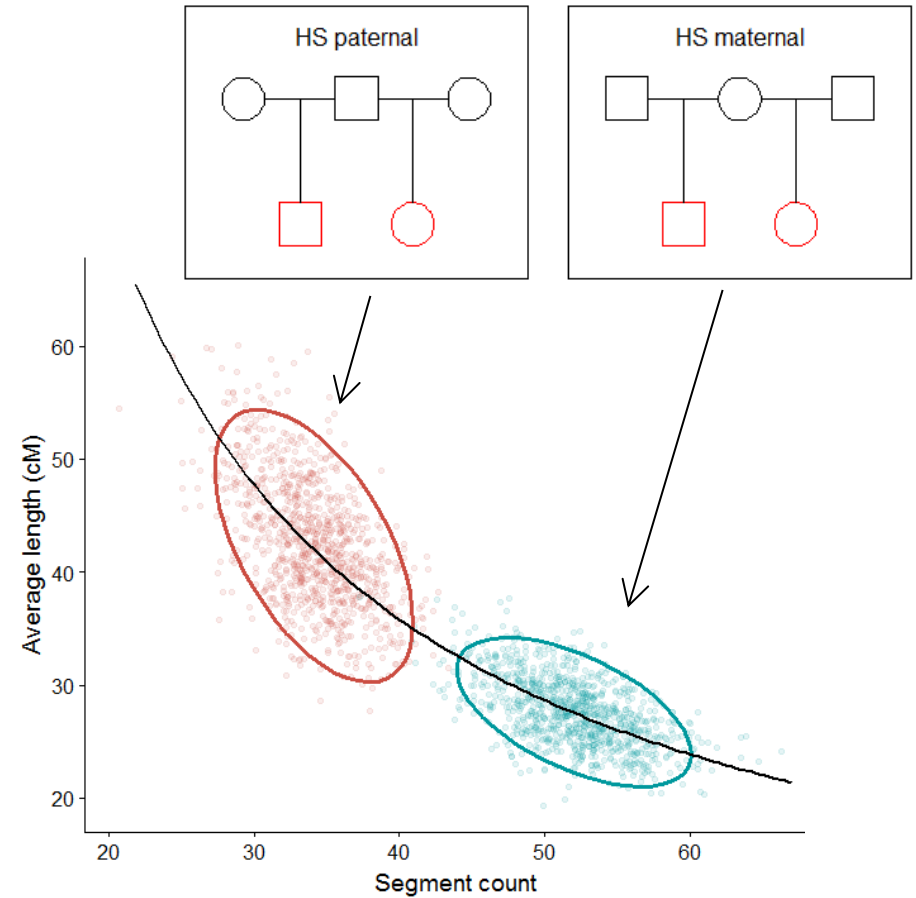
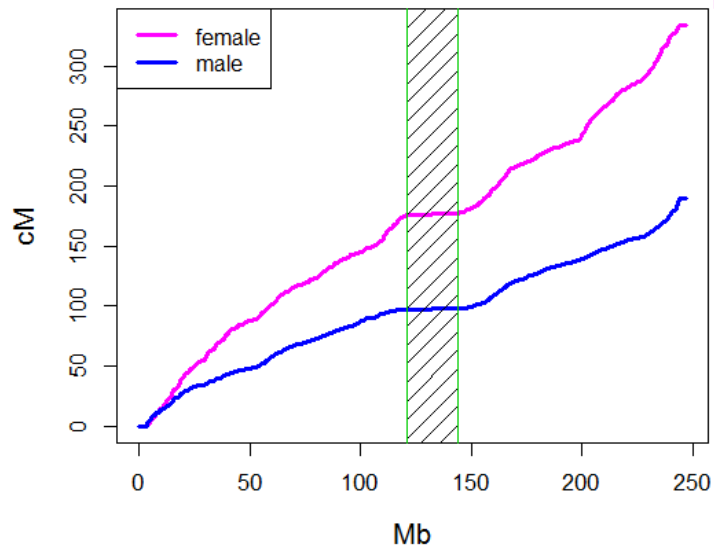
Different IBD segment distributions!

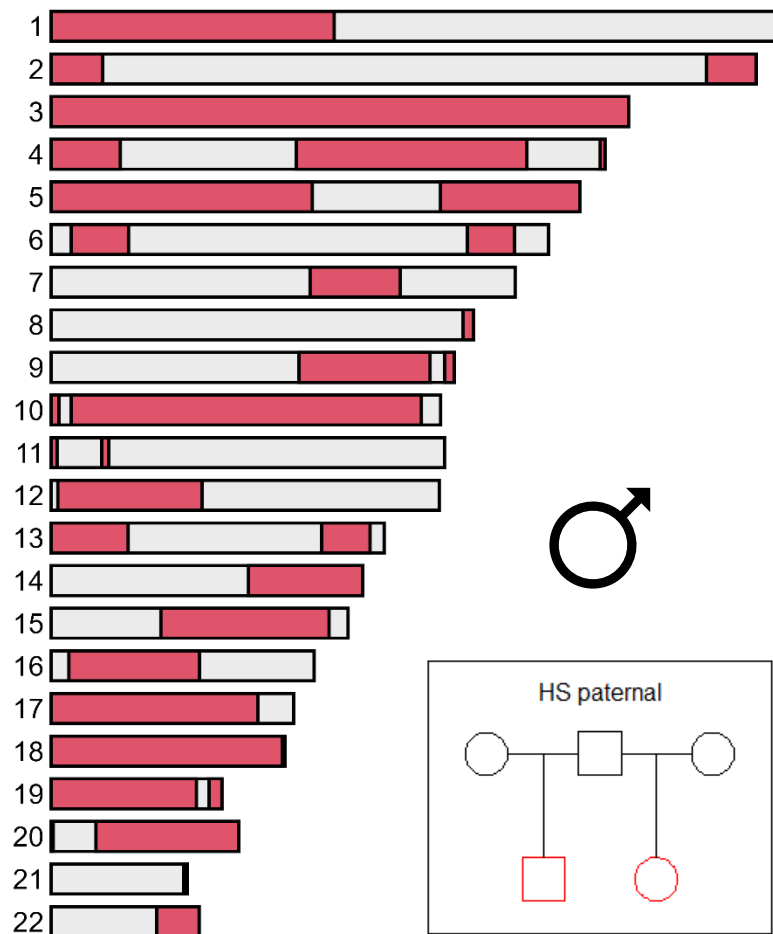


Therefore:
In theory distinguishable
with linked markers!

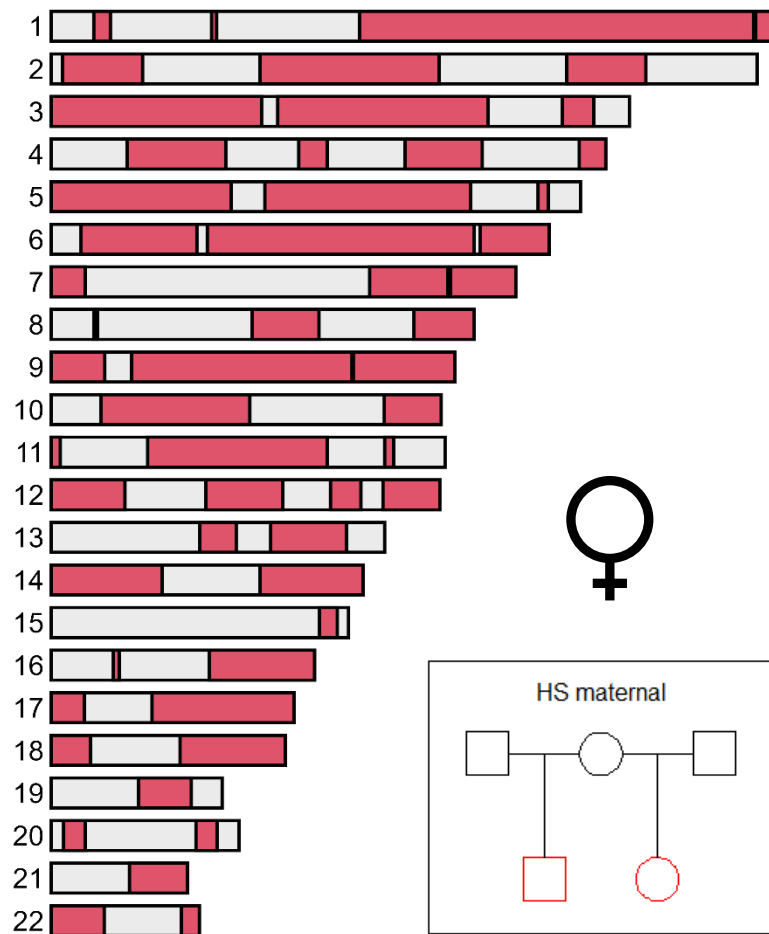
The importance of sex

Genetic map of chromosome 1





48% (32 segments)



55% (50 segments)

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

0

Seed

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

0

Seed

123

Simulate!

Ped 1

Settings

Number of sims

500

Length unit

☒ cM ☐ Mb

Analysis

☒ Sharing ☐ Autozygosity

Download data

Observed data

Total length

Count

Segments

Summary of genetic relatedness

- Pedigree-based measures of relatedness:
 - 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** resulting from recombination
 - females recombine more than males
 - useful variables: segment count & lengths

QuickPed

<https://magnusdv.shinyapps.io/quickped>

ibdsim2

<https://magnusdv.shinyapps.io/ibdsim2-shiny>

Your turn: Exercises!

