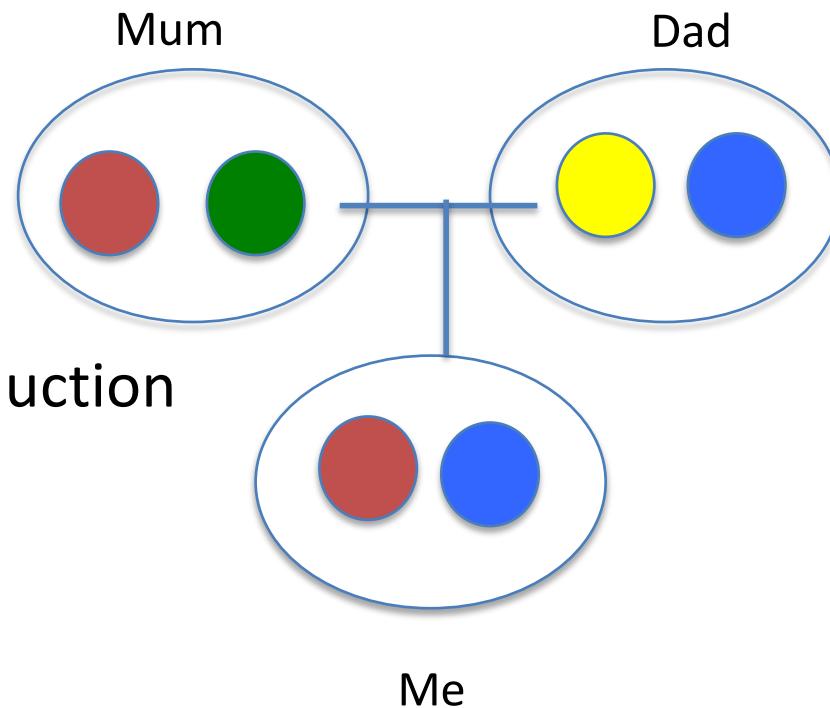


# Lecture 2: EVE102

# Hardy Weinberg Expectations



In adults just before reproduction

Frequency of A =  $p$

Frequency of a =  $q$

What's the frequency of AA homozygotes?

What do I have to assume?

A

a

- SNP A-4213906    p=0.27                q=0.73
- Freq. of AA
- Freq. of Aa
- Freq. of aa
- Actual genotype frequencies.
- SNP A-4213906    0.07                0.40                0.53

What if genotype frequencies were not in Hardy Weinberg at their HW proportions?

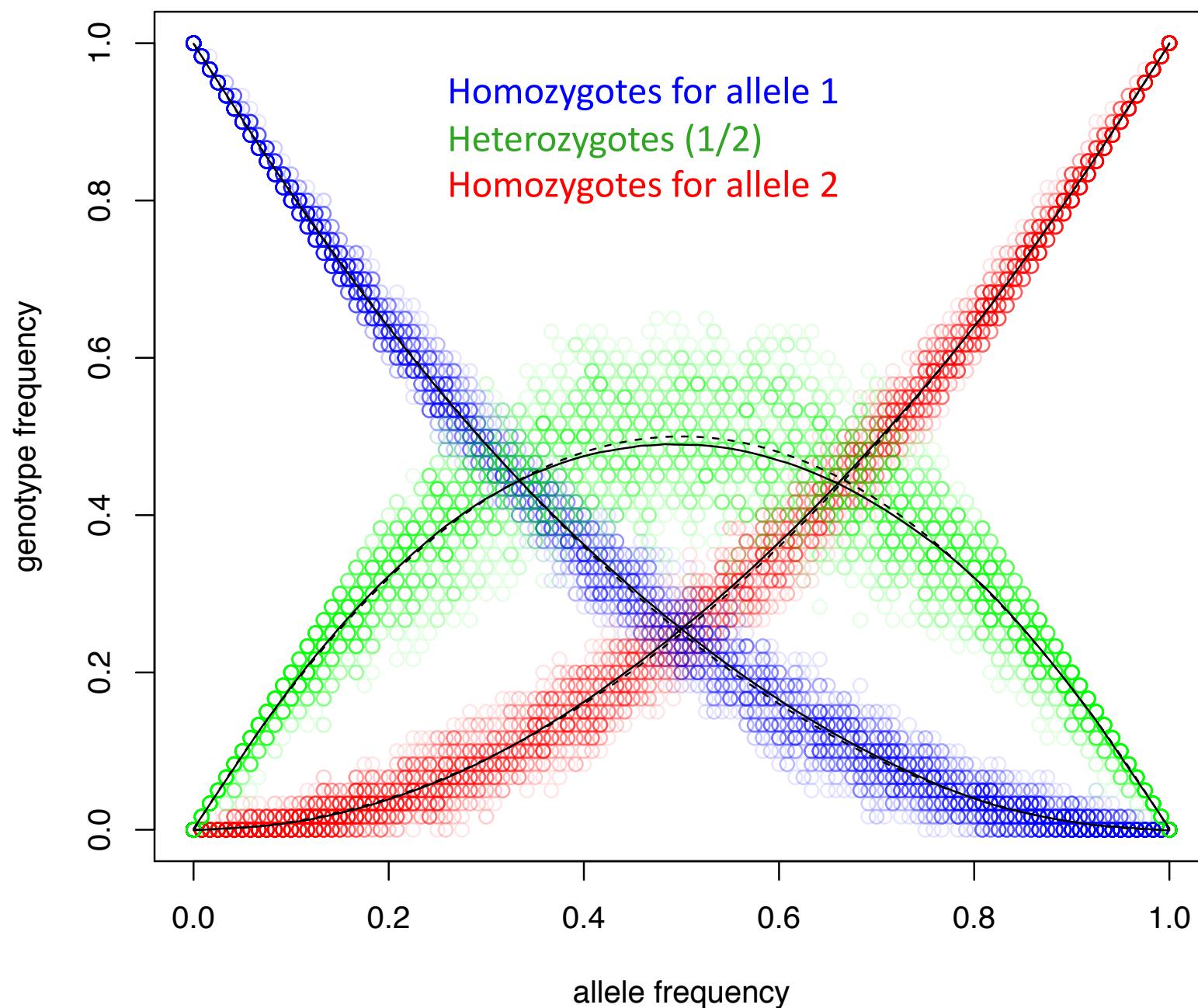
AA	Aa	aa
0.2	0.14	.66

- $p=f_{AA} + \frac{1}{2} f_{Aa} = 0.2 + .14/2 = 0.27$        $q=f_{aa} + \frac{1}{2} f_{Aa} = 0.73$
- Freq. of AA =  $p^2 = 0.27^2 = 0.079$
- Freq. of Aa =  $2pq = 2 \times 0.27 \times 0.73 = 0.394$
- Freq. of aa =  $q^2 = 0.73^2 = 0.533$

Genotype frequencies returned to Hardy Weinberg Proportions within one generation  
Of random mating!

Therefore the Hardy Weinberg expectations/proportions are an equilibrium if nothing interesting happens in popgen.

# The Empirical Relationship between Genotype and allele Frequencies in a European population

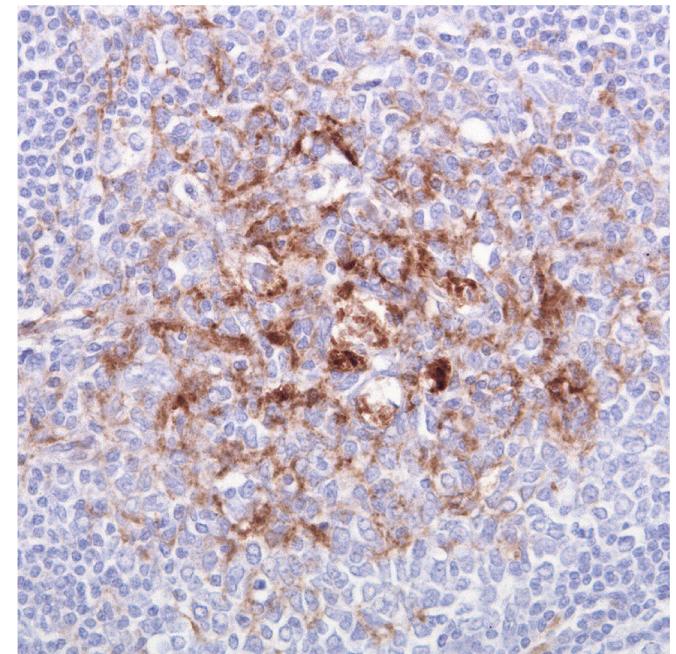


# Kuru outbreak in the Fore people

The Fore people of Papua New Guinea practiced ritual funereal cannibalism (till 1950s)

coding polymorphism (Methionine/Valine) at codon 129 of *PRNP*. Homozygotes for either allele develop prion disease at a higher rate.

Met/Met	Met/Val	Val/Val
4	23	3

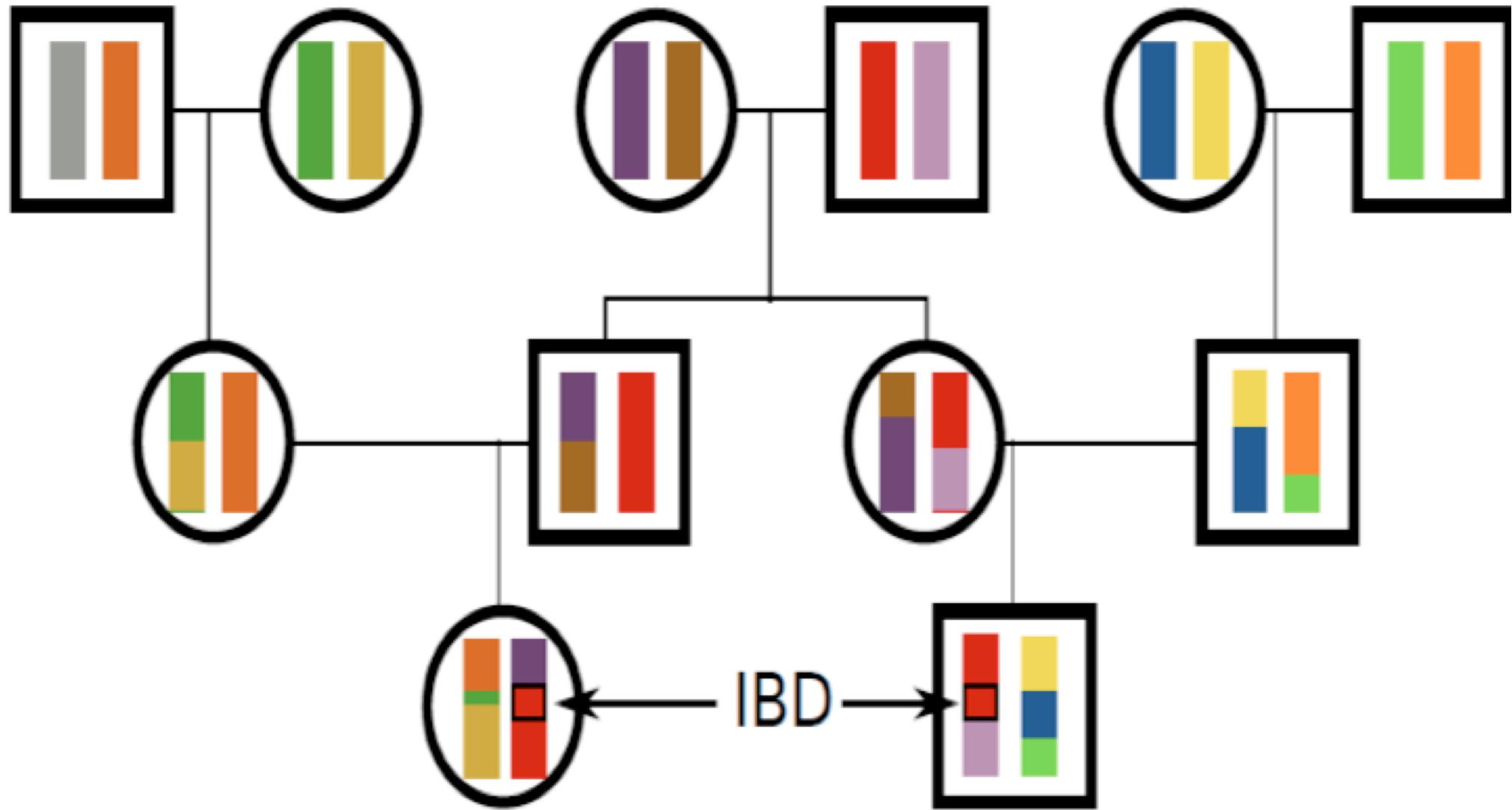


[https://en.wikipedia.org/wiki/Creutzfeldt%20Jak\\_disease#/media/File:VCJD\\_Tonsil.jpg](https://en.wikipedia.org/wiki/Creutzfeldt%20Jak_disease#/media/File:VCJD_Tonsil.jpg)

Chi-squared statistic significance =0.0034

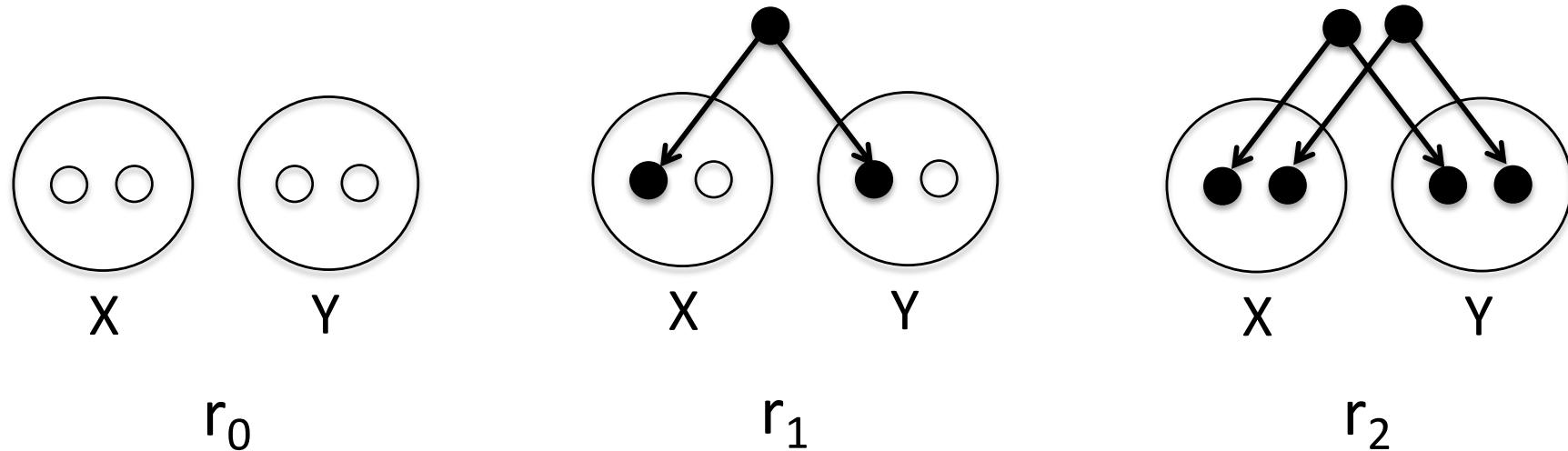


Frequency of red hair = 0.01



Identity by descent: an allele (or region of chromosome)  
Shared between individuals due to a recent ancestor

The average number of alleles shared identical by descent between relatives

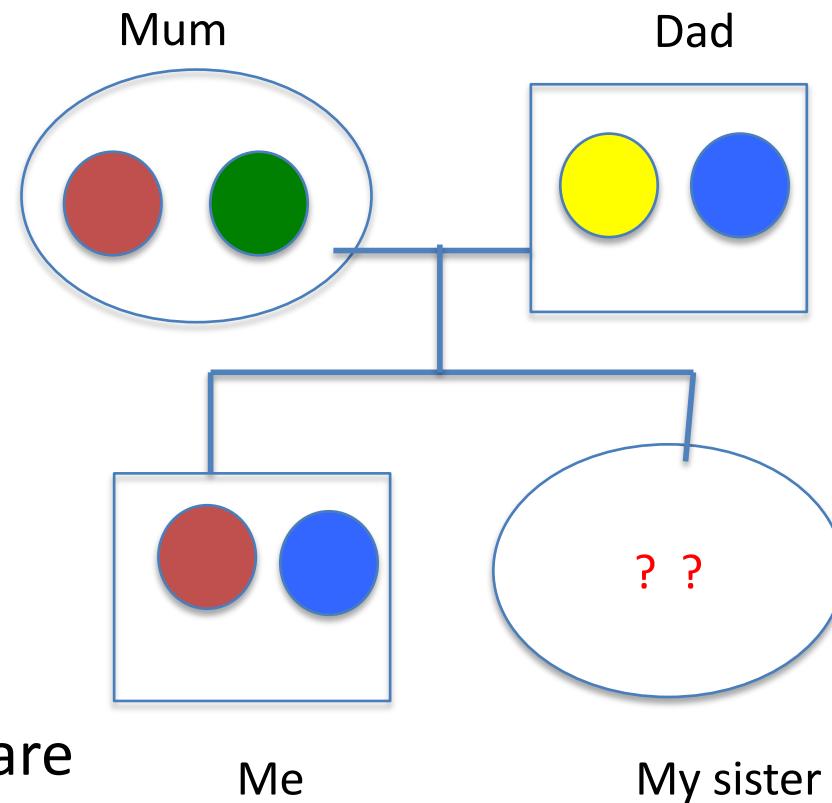


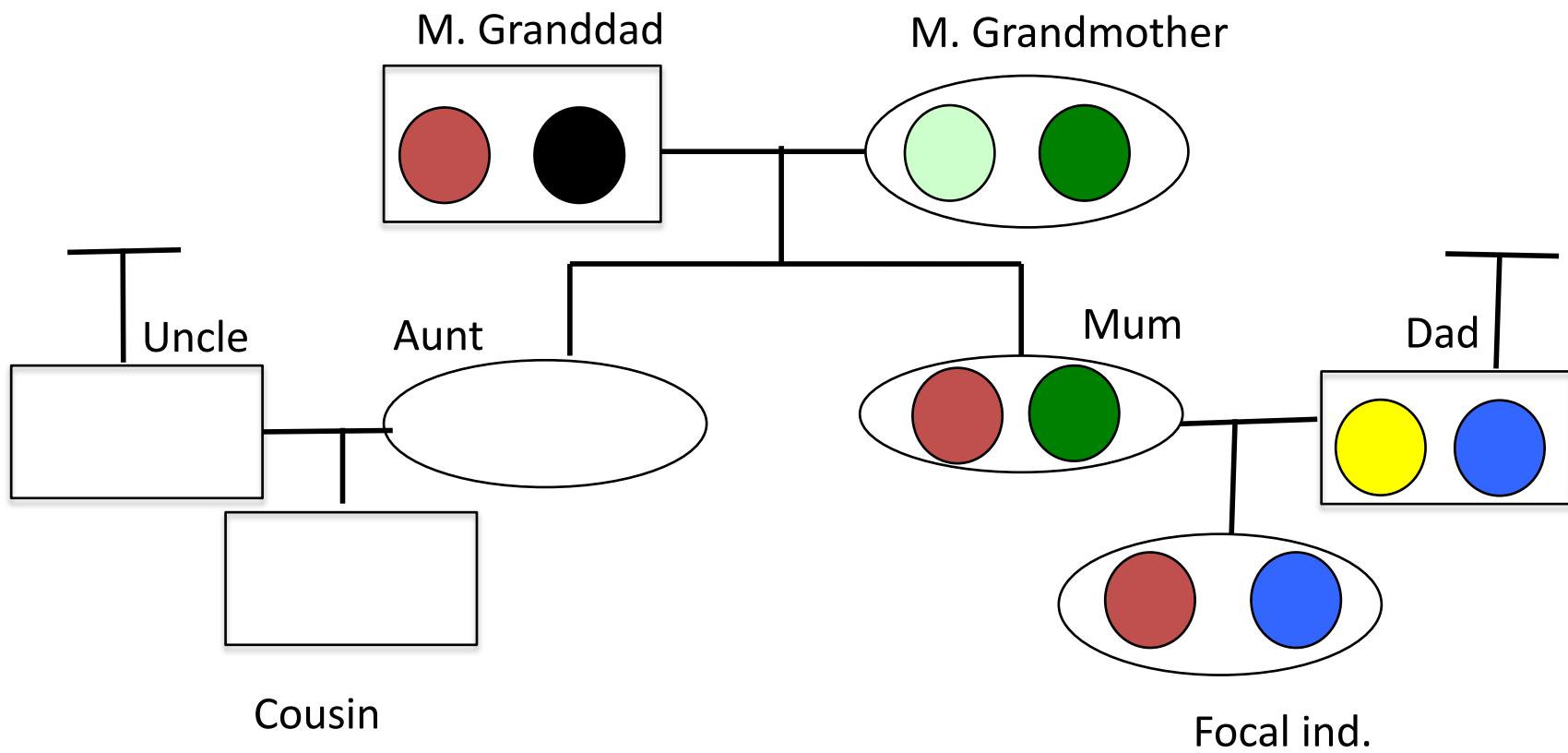
# Oh brother what genotype are thou?

Identity by descent

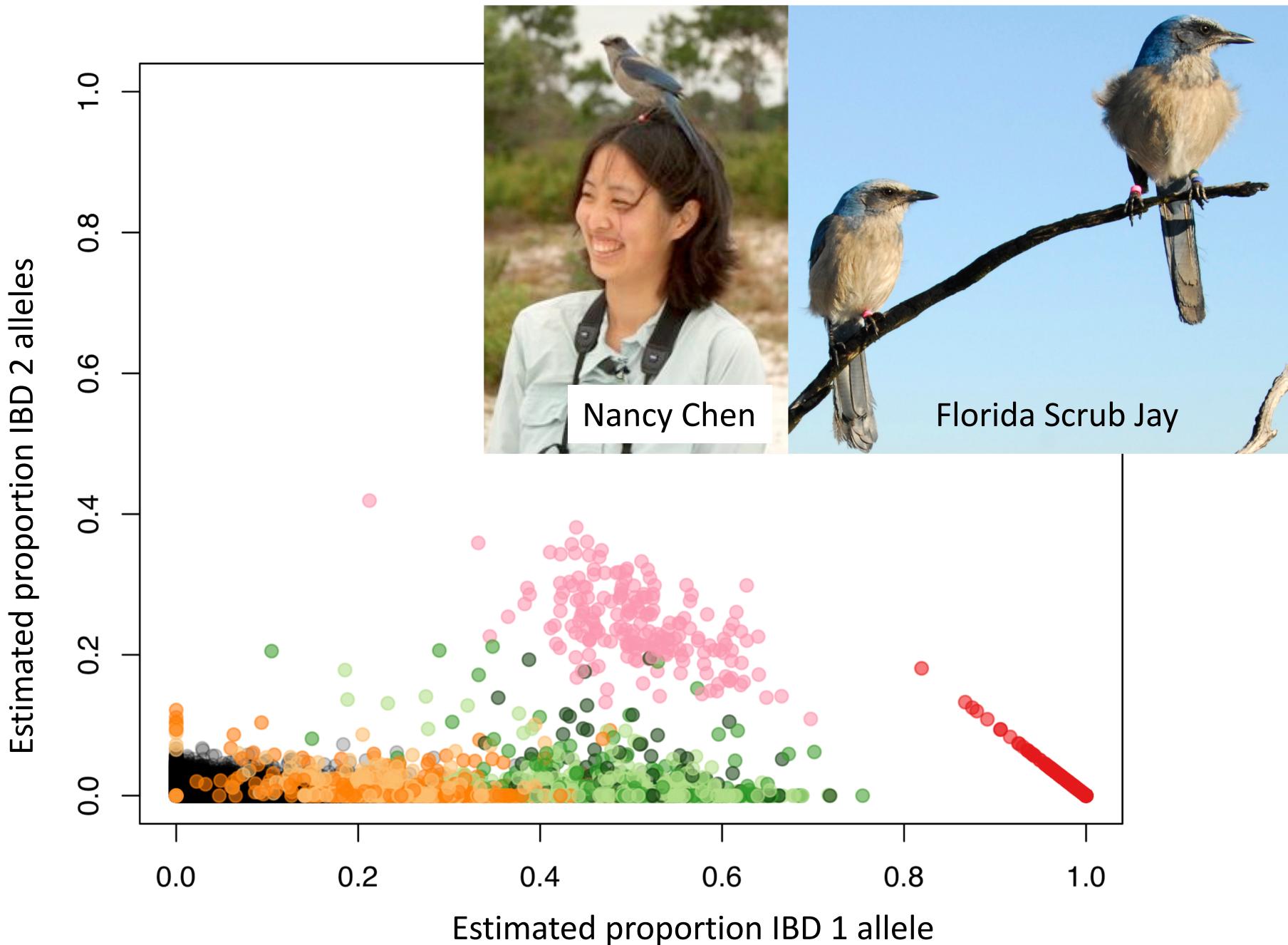
What is the probability  
I share 0,1,or 2 alleles  
with my Mum ibd?

What's the probability that I share  
0,1,2 alleles with my sister ibd?



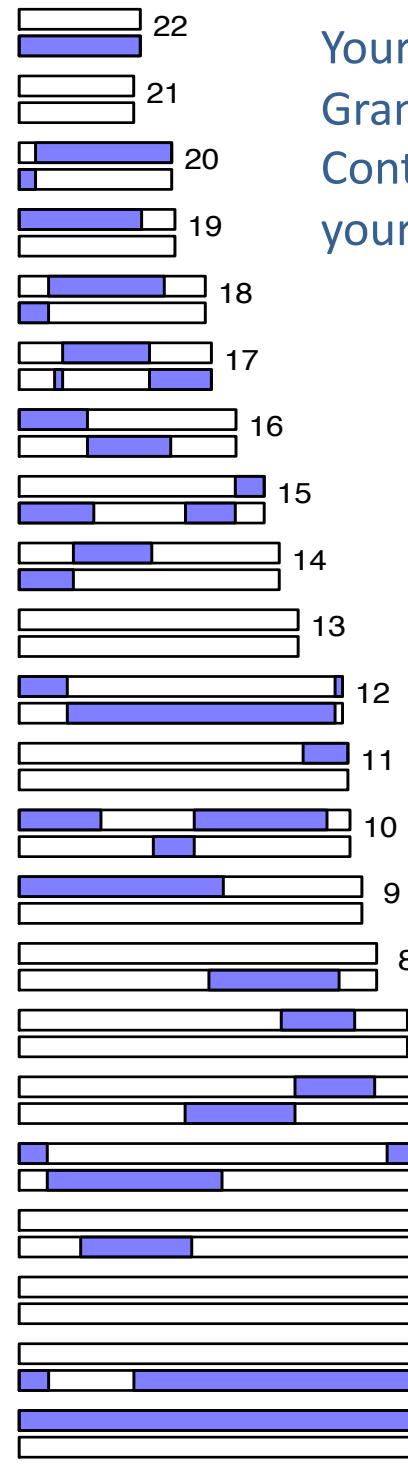


Given a good assessment of allele frequencies we can identify close relatives

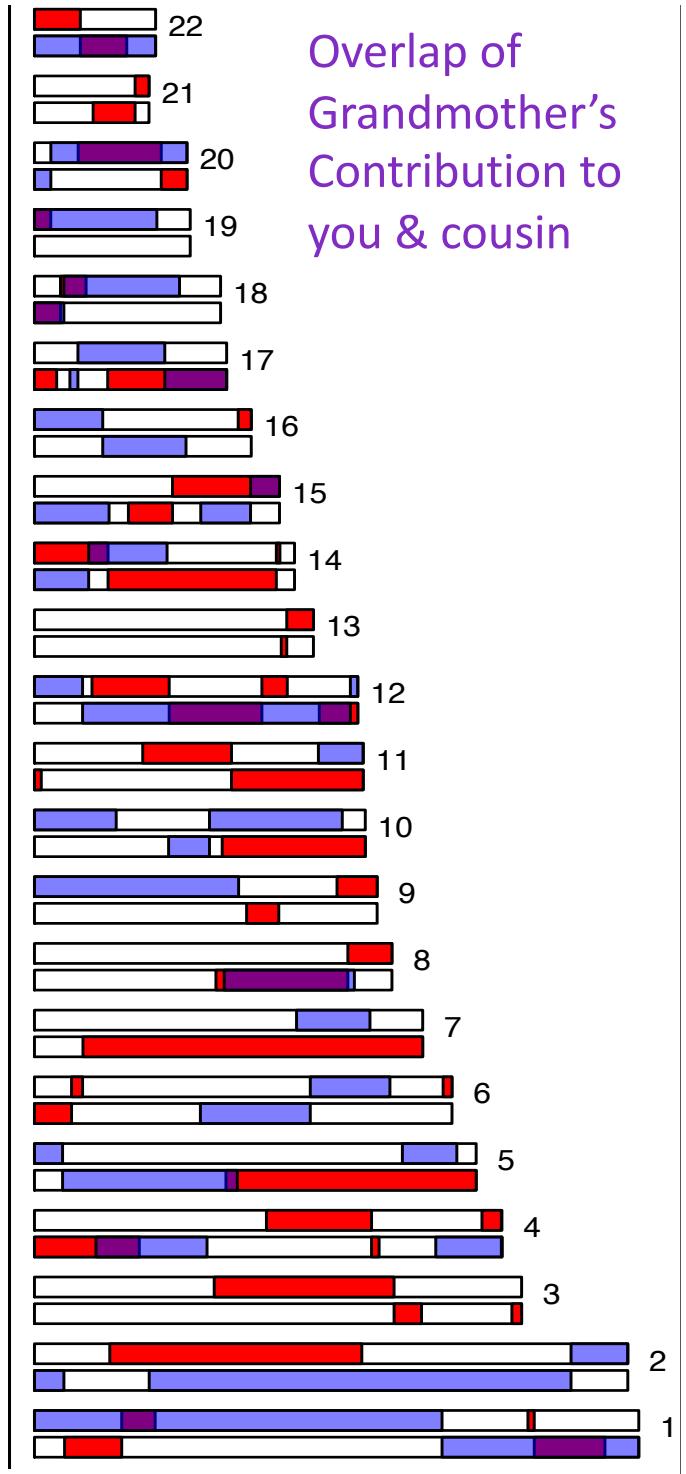




Your maternal  
Grandmother's  
Contribution to you



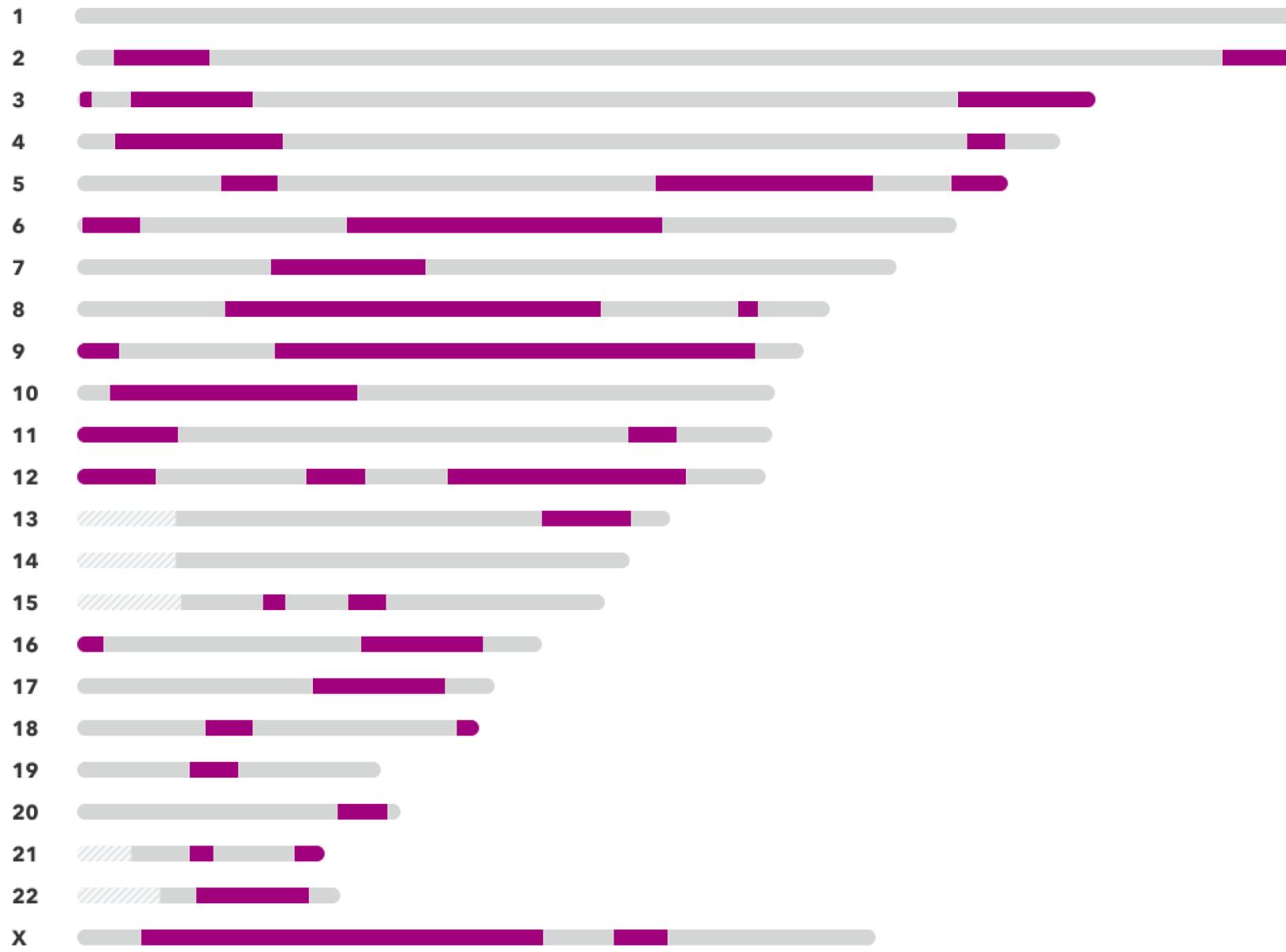
Your maternal  
Grandmother's  
Contribution to  
your cousin

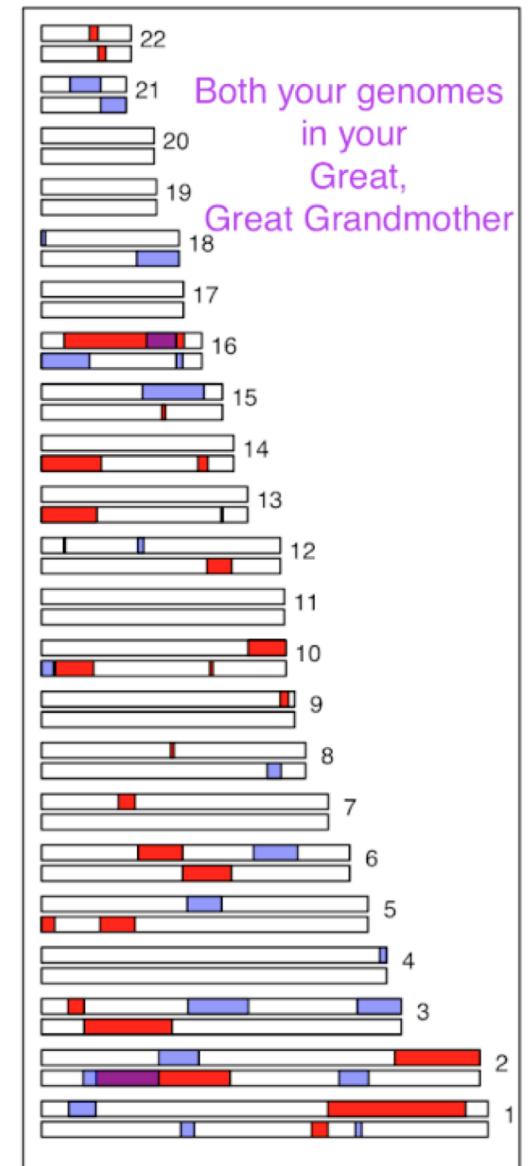
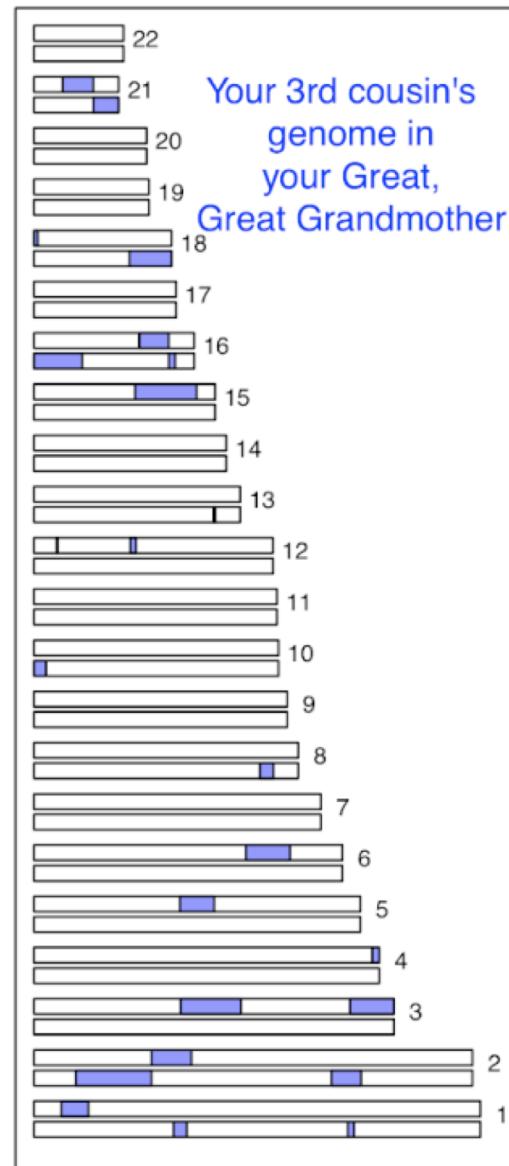
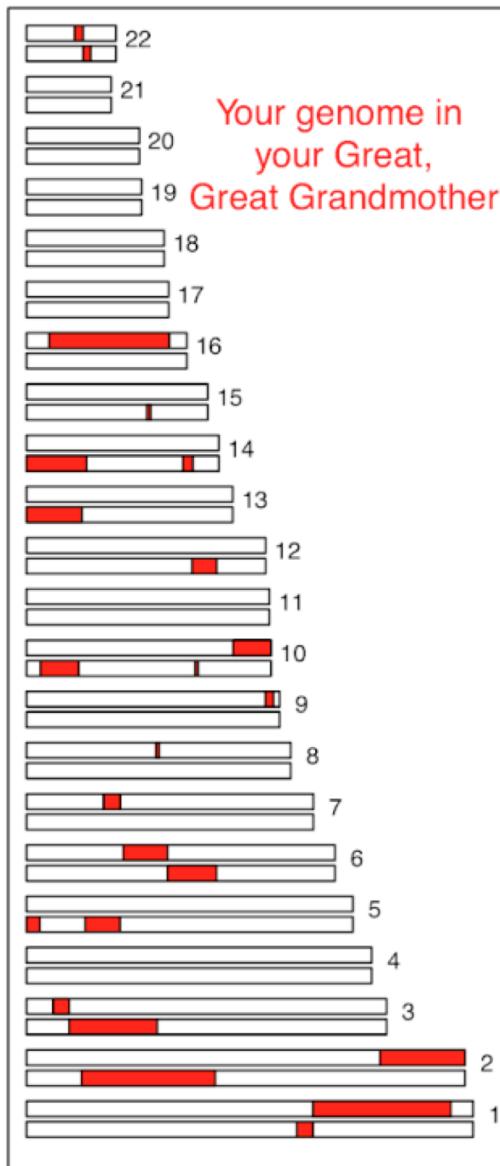


Overlap of  
Grandmother's  
Contribution to  
you & cousin

A person and their 1<sup>st</sup> cousin  
On 23&me

Half identical  
1039 cM  
38 segments





Me and my 3<sup>rd</sup> cousin (?)  
On 23&me

Half identical  
66 cM  
3 segments



# To find alleged Golden State Killer, investigators first found his great-great-great-grandparents

Gedmatch

By Justin Jouvenal April 30  Email the author

To qualify as a 'match' in the genealogical time frame, results must have a largest Autosomal segment that has at least 700 SNPs and be at least 7 cM. It must have BOTH. Results with the largest segment less than 7 cM are highlighted in pink.

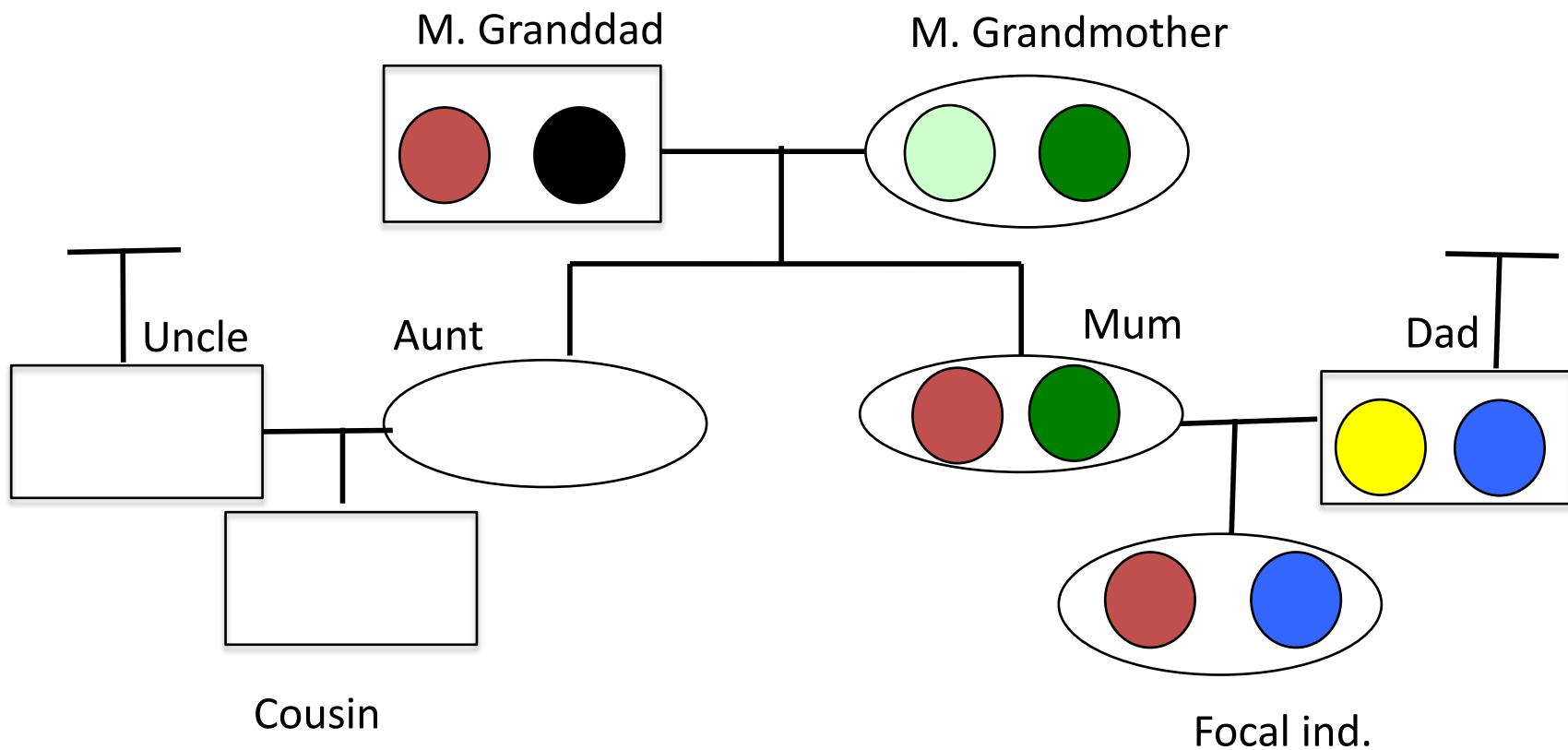
In general, the results shown below use thresholds LESS than 7cM / 700 SNPs.

PLEASE verify any result shown on this list with the one-to-one comparison tool before assuming any match is real.

To check the number of SNPs, click on the 'A' on the same line to view the one-to-one comparison detail.

Please DO NOT send emails to anyone on this list without first using the one-to-one utility to verify that it is a legitimate match.  
DO NOT create mass mailing lists from these results.

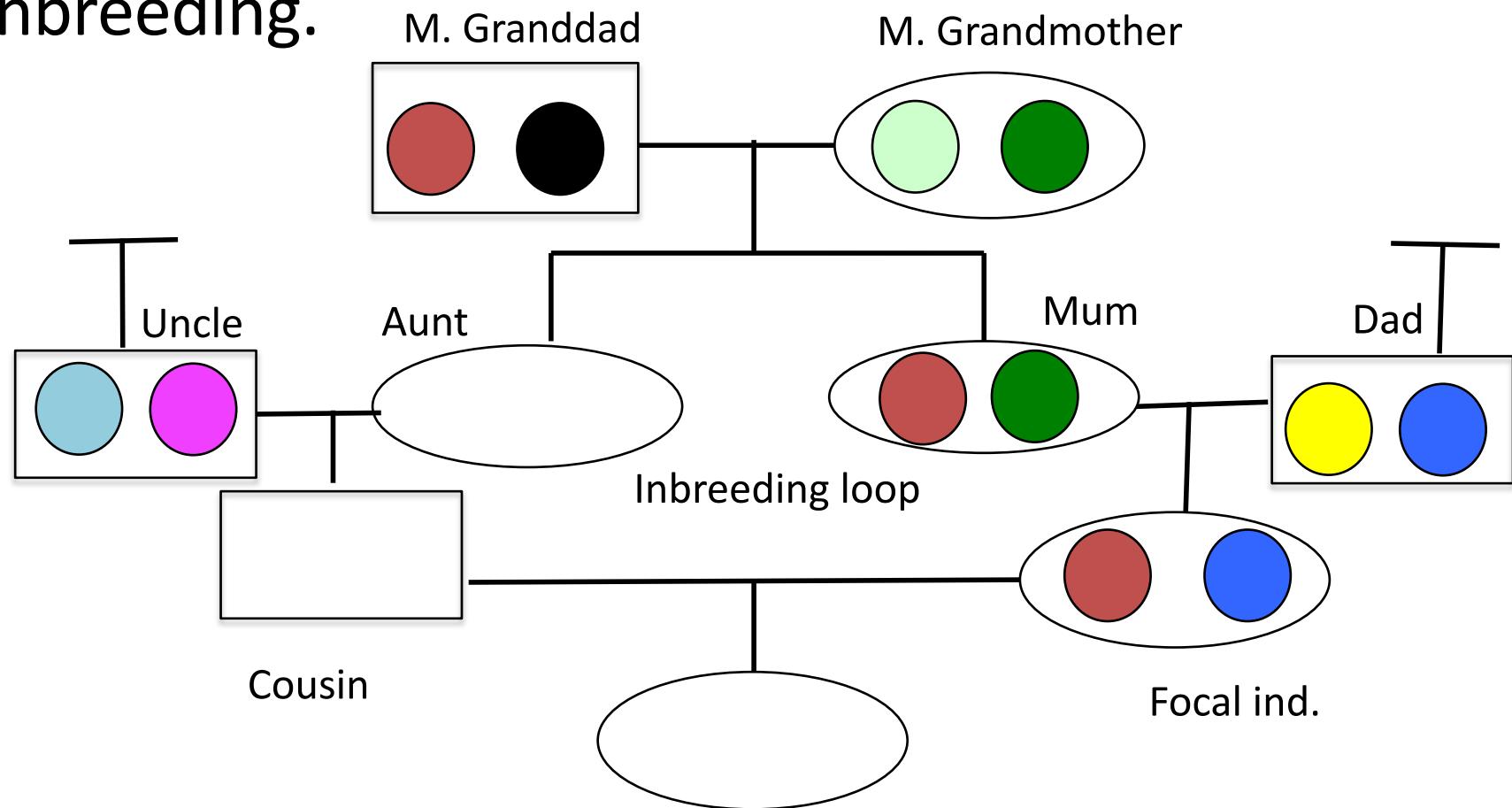
Kit Nbr	Type	List	Select	Sex	GED/WikiTree	Haplogroup		Autosomal			X-DNA			Name (* => alias)	Email	
						Mt	Y	Details	Total cM	largest cM	Gen	Details	Total cM	largest cM		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	F				A	106.7	49.5	3.5	X	6.2	6.2		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M				A	52.3	25.0	4.1	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	F				A	46.5	27.4	4.1	X	6.9	6.9		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M	GED			A	40.1	21.3	4.2	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	F				A	31.5	21.2	4.4	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	F				A	31.1	20.7	4.4	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M	GED			A	29.7	29.7	4.5	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M				A	29.3	29.3	4.5	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M				A	28.7	15.8	4.5	X	0	0		
V4	L	<input type="checkbox"/>	<input type="checkbox"/>	M		H3	I1	A	28.7	28.7	4.5	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	U				A	28.5	22.6	4.5	X	7.3	7.3		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M	GED			A	28.1	17.7	4.5	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	F				A	27.9	15.7	4.5	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	M	GED			A	27.6	27.6	4.5	X	0	0		
F2	L	<input type="checkbox"/>	<input type="checkbox"/>	F				A	27	16.8	4.5	X	11.5	6.4		



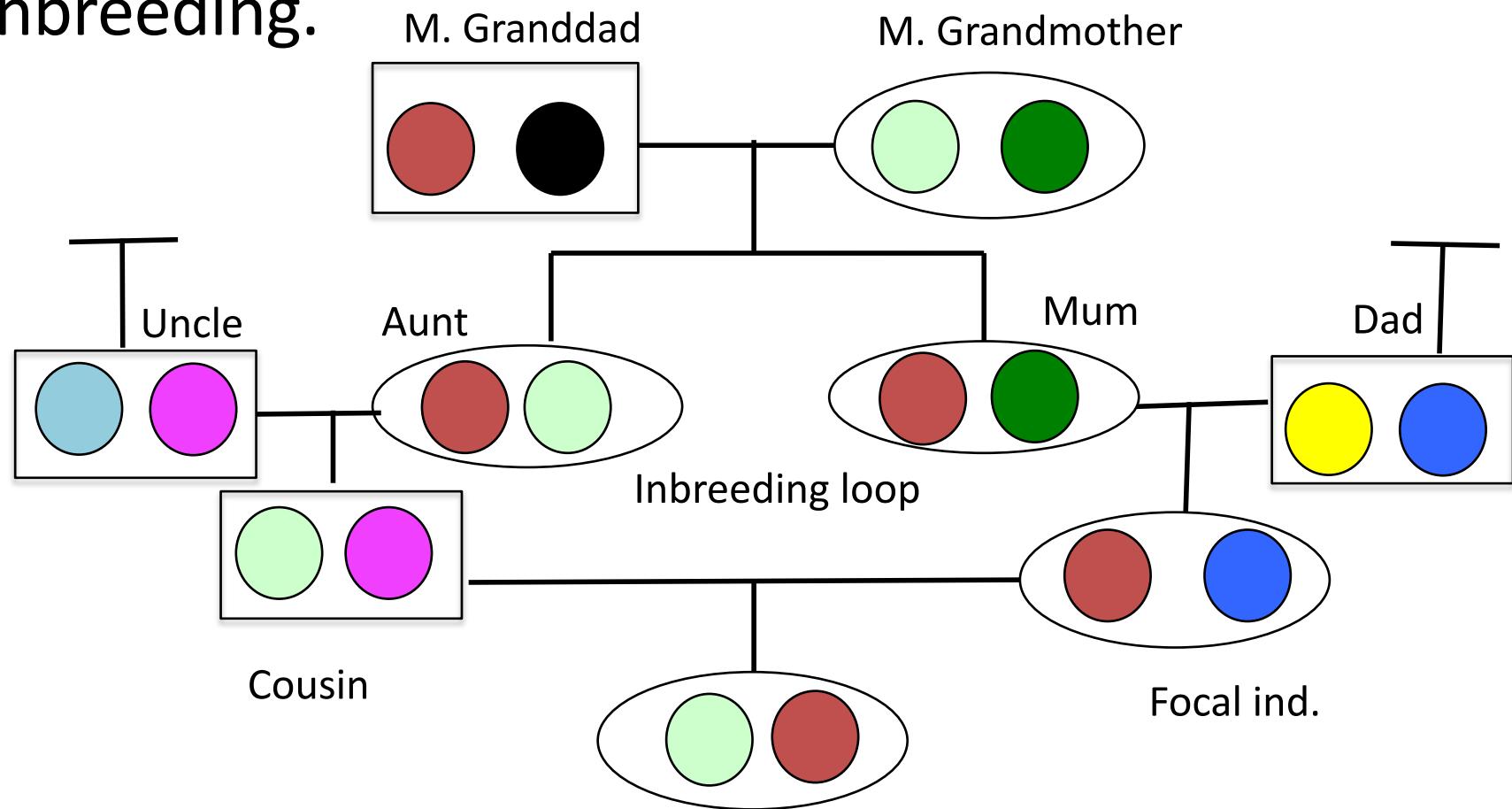
$$f = \text{Coefficient of kinship} = 0 \times r_0 + r_1/4 + r_2/2$$

Notes: Equation 2.3, page 16

# Inbreeding.



# Inbreeding.

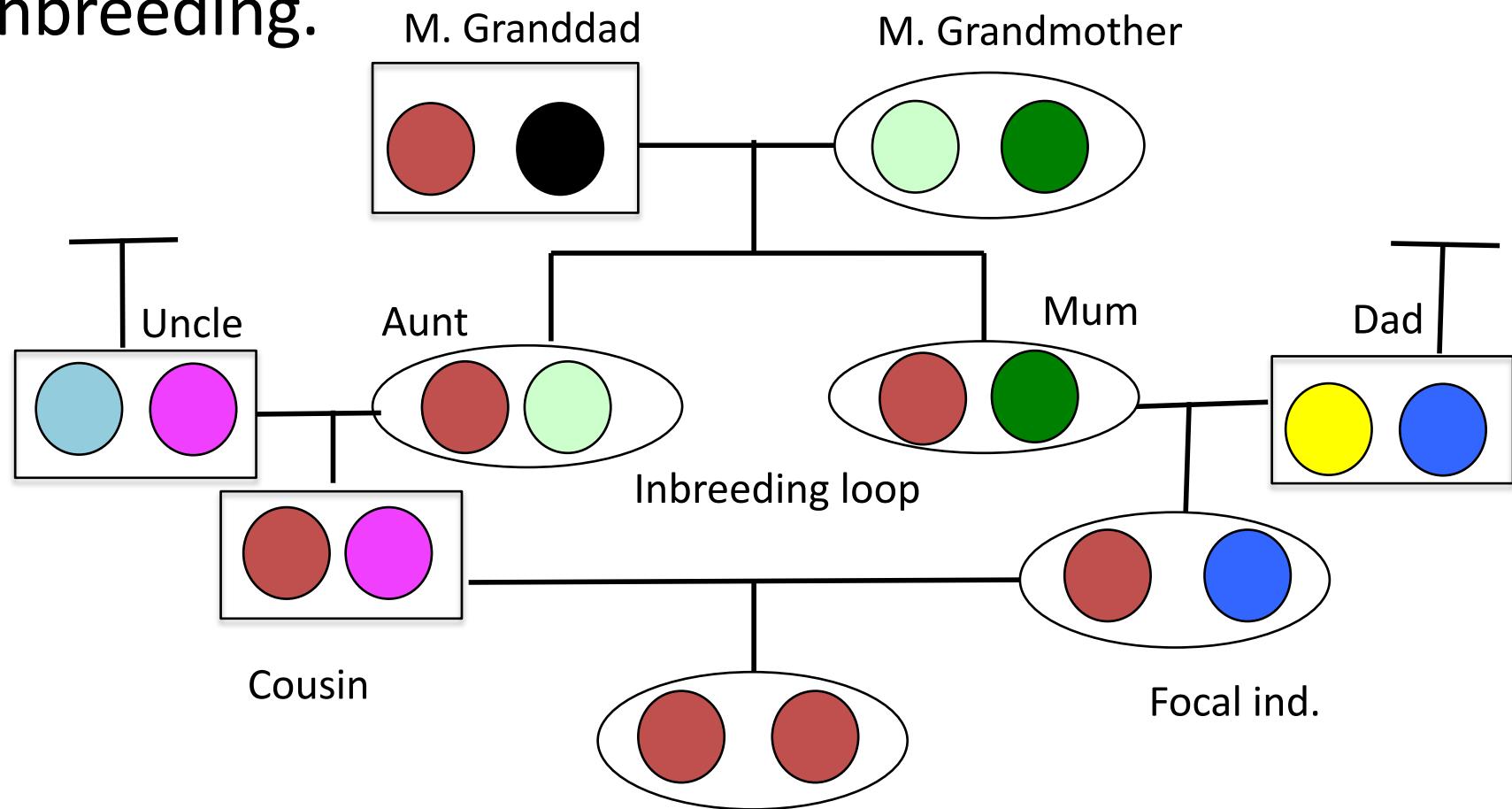


What's the probability that the child of first cousins  
Is heterozygote?

If freq is p

Notes: Equation 2.7 page 20

# Inbreeding.



What's the probability that the child of first cousins  
Is heterozygote?

If freq is p

Notes: Equation 2.7 page 20

Notes: Equation 2.7 and 2.8 page 20

# Generalized HW

Inbreeding coefficient  $F =$   
probability that an individual  
inherits two alleles identical by  
descent at a locus

AA

$$(1-F)p^2+Fp$$

Aa

$$(1-F)2p(1-p)$$

aa

$$(1-F)(1-p)^2+F(1-p)$$

Inbreeding:

Increases proportion of homozygotes  
Decreases proportion of heterozygotes

Note no change in  
allele frequency.  
Only genotype frequency

# The Ancestry of King Charles II of Spain (1661-1700)

