

Forensic Genetics

Module 16 – Session 8

Relatedness

- Incorporating Relatives
 - Identity By Descent
- Applications of Relatedness
 - Paternity Testing
 - Missing Persons
 - Familial Searching
 - Genealogical Searching

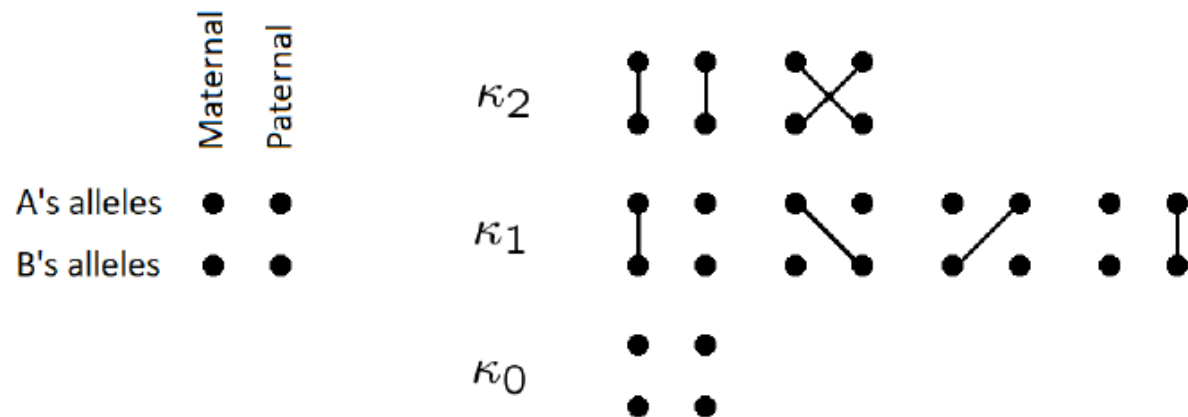
Identity By Descent

- Relatives are similar because they share alleles that are *identical by descent* (IBD).
- IBD alleles are copies of the same allelic type inherited through a common ancestor (and ignores mutation).
- A pedigree or relationship determines IBD probabilities, which determine probabilities of joint genotypes.

IBD Coefficients

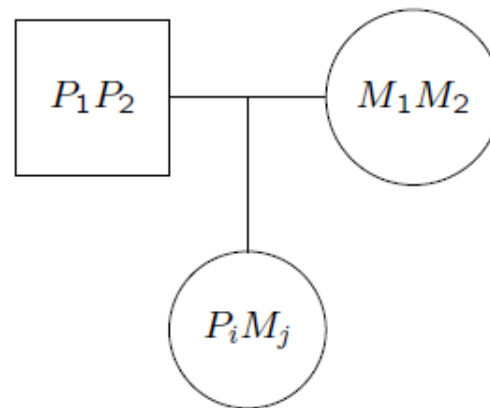
For *non-inbred* relatives, there are three IBD classes. We write κ_i to denote the IBD probabilities:

$$\kappa_i = \Pr(i \text{ alleles IBD})$$



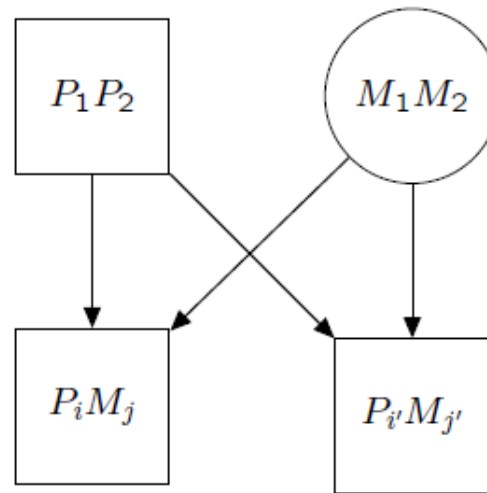
IBD for Parent-Offspring

- Mendel's law states that one of the two alleles from a parent will be passed down to a child;
- Both alleles have equal probability $\frac{1}{2}$ of being passed down.



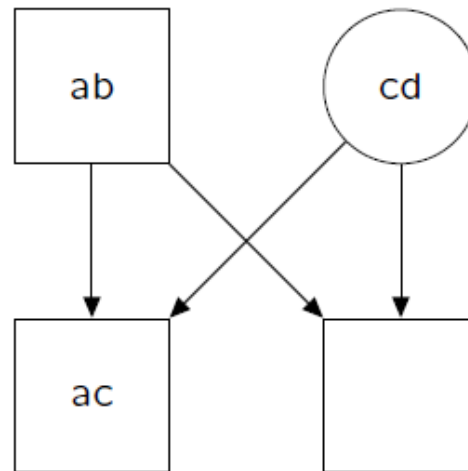
IBD for Siblings

What about siblings?



IBD for Siblings

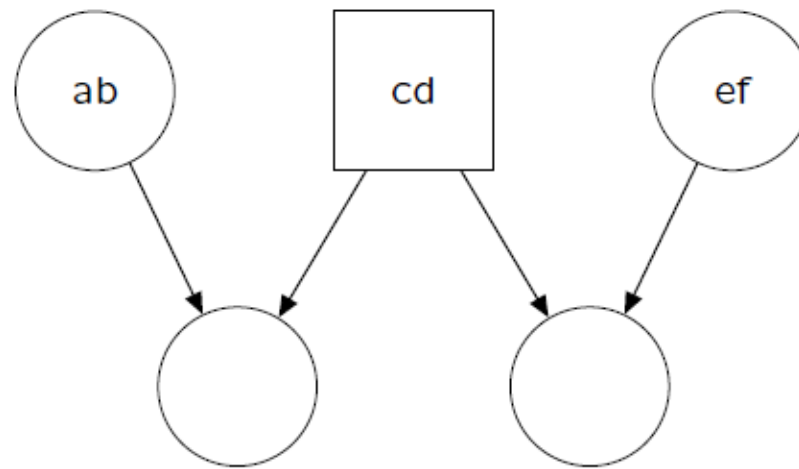
They share either both, one or none of the alleles IBD.



		Alleles IBD		
Sib 1	Sib 2	0	1	2
ac	ac	✓		
	bc		✓	
	ad		✓	
	bd			✓
Total		1/4	1/2	1/4

IBD for Half-Sibs

What are the IBD coefficients for half-sibs?



IBD Coefficients

The following table shows IBD probabilities for common relationships:

Relationship	κ_0	κ_1	κ_2
Unrelated	1	0	0
Parent/child	0	1	0
Identical twins	0	0	1
Siblings	1/4	1/2	1/4
Half-sibs	1/2	1/2	0
First cousins	3/4	1/4	0

These IBD probabilities give the expected relatedness between individuals (the realized relatedness is variable).

Match Probabilities for Relatives

Combining the terms leads to the overall single-locus match probability for relatives:

$$\kappa_2 + \kappa_1 M_1 + \kappa_0 M_2,$$

which yields a standard match probability of M_2 for unrelated individuals.

LR – Relatives

Consider a simple single-source crime scene sample with genotype $G_C = AA$, and a suspect that matches at that locus. Calculate the LR, using $p_A = 4\%$, and alternative hypotheses:

- The DNA in the sample came from an unrelated individual;
- The DNA in the sample came from a half-brother of the suspect;

LR – Relatives

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- $$LR = \frac{\Pr(AA|AA,H_p)}{\Pr(AA|AA,H_d)} = \frac{1}{p_A^2} = 625;$$

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- $LR = \frac{1}{\kappa_0 M_2 + \kappa_1 M_1 + \kappa_2} = \frac{1}{0.5p_A^2 + 0.5p_A} \approx 48;$

The Island Problem

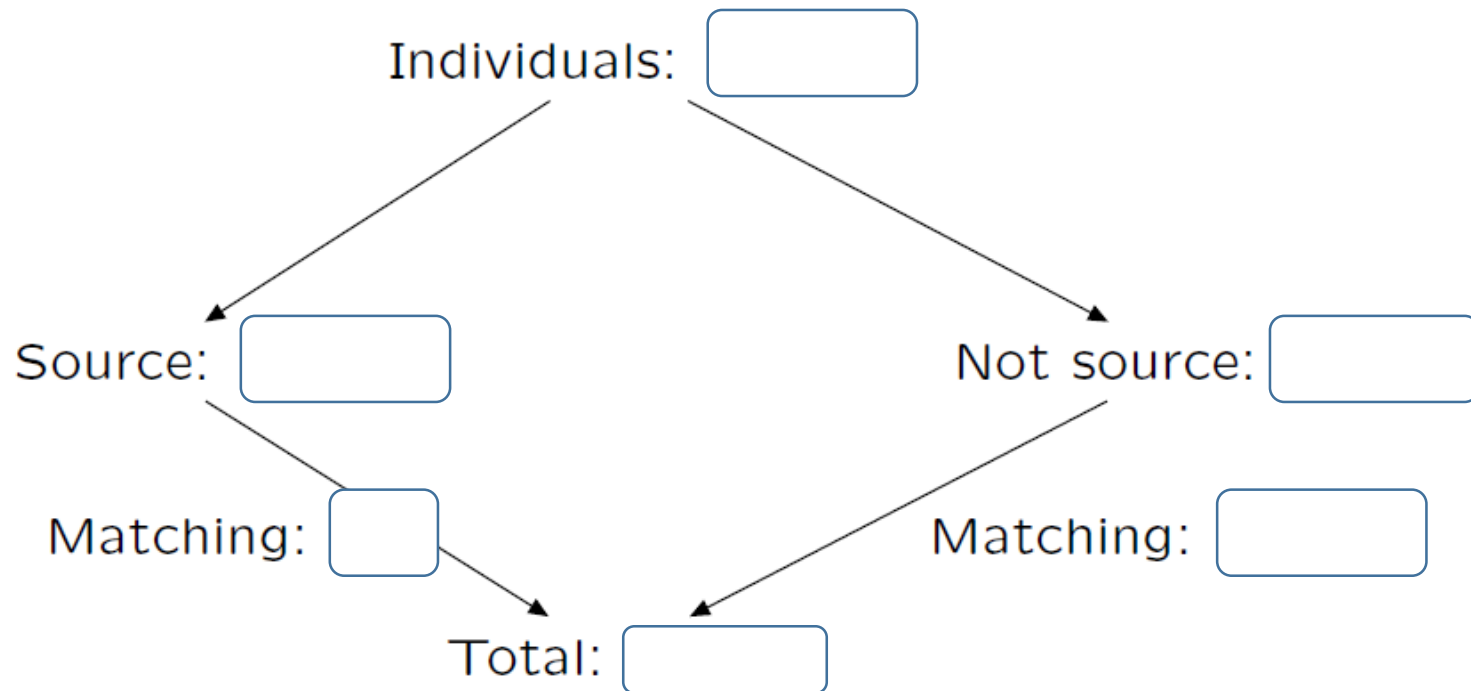
Suppose there is a crime committed on a remote island with a population of size 1001. A suspect Q is found to match the crime scene profile. What is the probability that Q is the source of the profile, assuming that:

- All individuals are equally likely to be the source.
- The DNA profiles of all the other individuals are unknown.
- The match probability for unrelated individuals is 5×10^{-6} .

Source: Weight-of-Evidence for Forensic DNA Profiles (Balding, 2015)

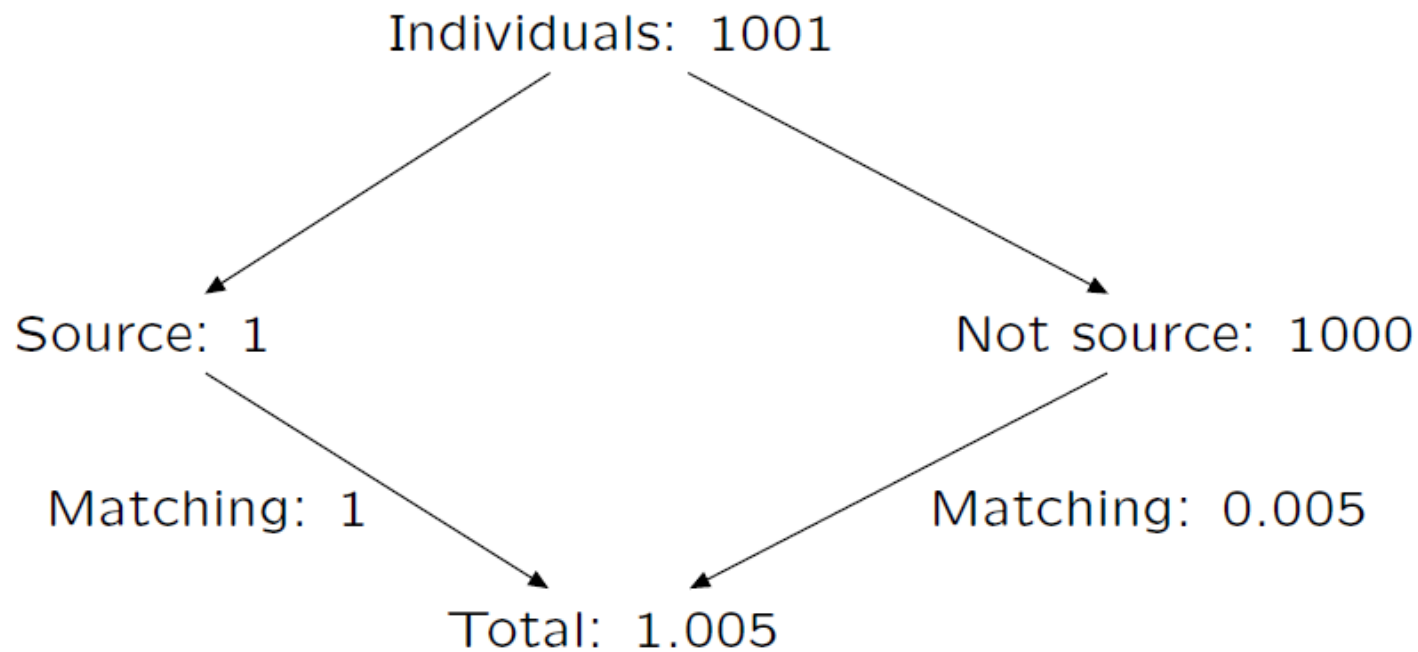
The Island Problem

Assuming Q has no relatives on the island, there is a chance that Q is the source.



The Island Problem

Assuming Q has no relatives on the island, there is a $\frac{1}{1.005} \approx 99.5\%$ chance that Q is the source.



The Island Problem – Relatives

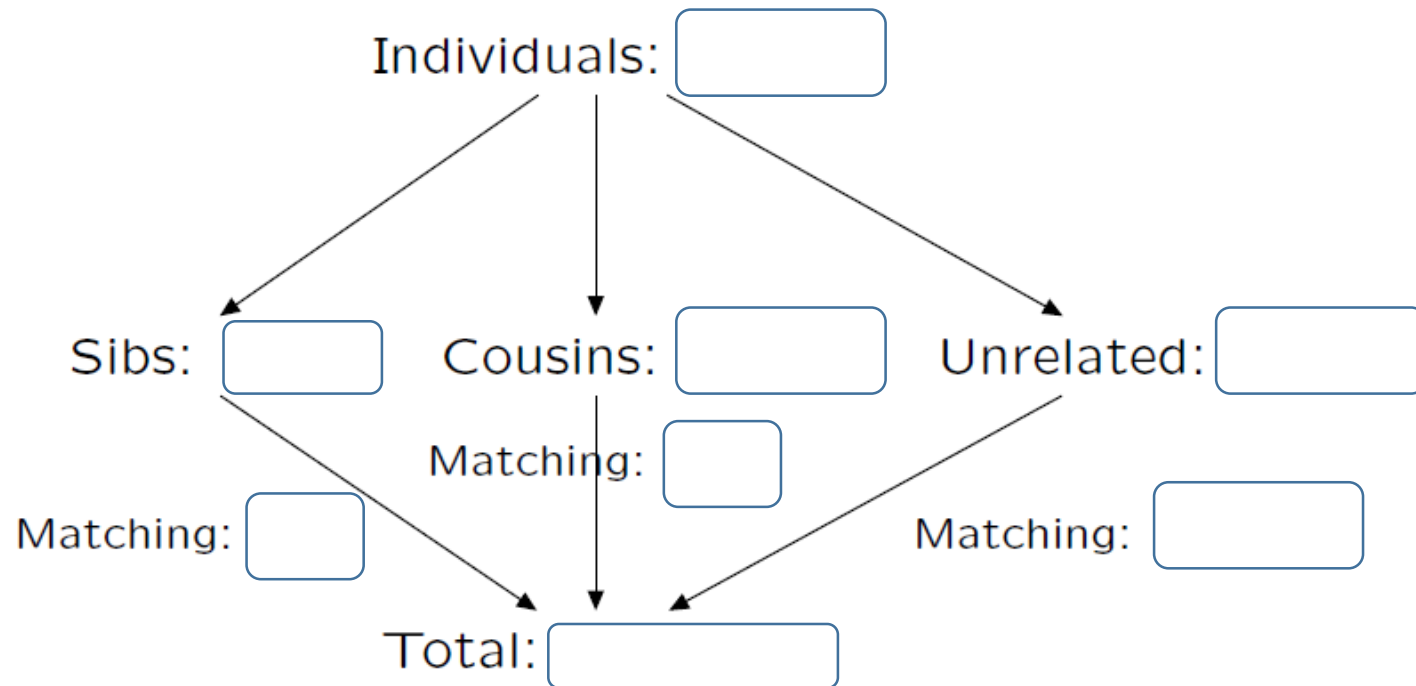
Now suppose that Q has one sibling and 20 cousins on the island, and no other relatives. What is now the probability that Q is the source, using match probabilities of:

- 1 in 1000 for a cousin;
- 1 in 100 for a sibling;
- and 5×10^{-6} for unrelated individuals.

Source: Weight-of-Evidence for Forensic DNA Profiles (Balding, 2015)

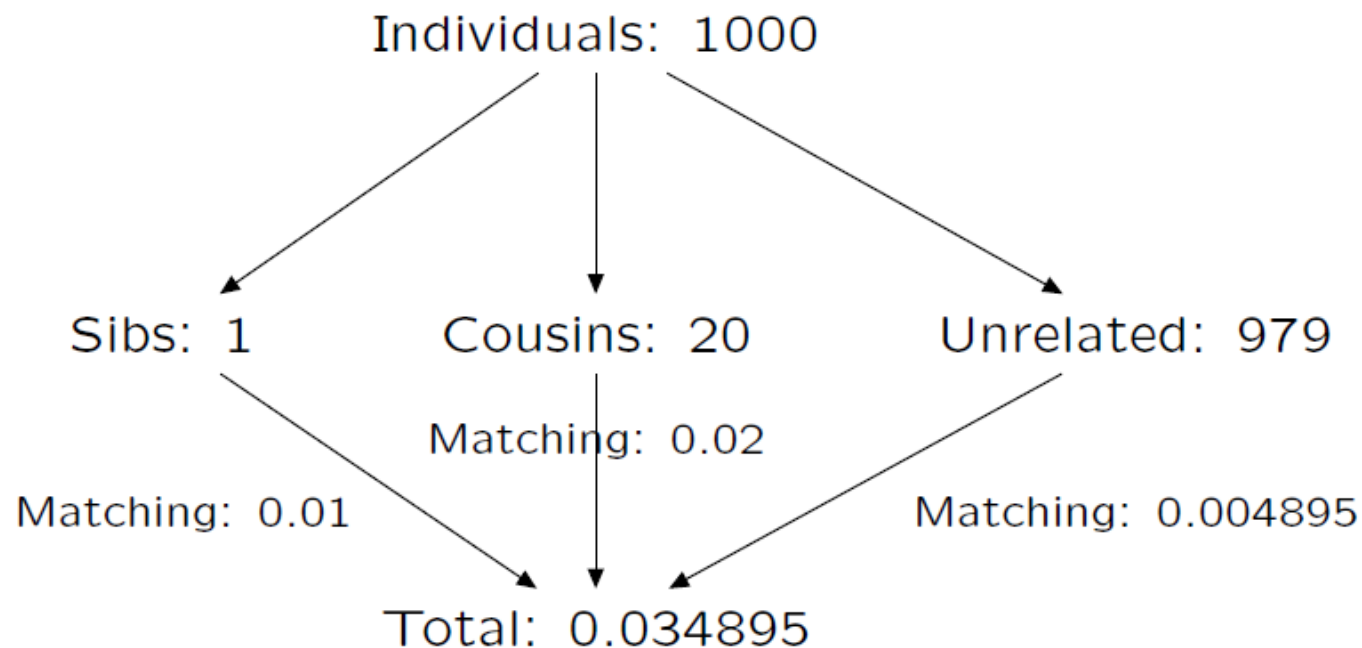
The Island Problem – Relatives

In this case the probability that Q is the source decreases to



The Island Problem – Relatives

In this case the probability that Q is the source decreases to $\frac{1}{1.034895} \approx 96.6\%$.



The Island Problem – Relatives

Note how the LR for unrelated individuals ($LR_U = 200\,000$), the LR for cousins ($LR_C = 1\,000$), and the LR for siblings ($LR_S = 100$), can be combined as a weighted average of the match probabilities:

$$\left(\frac{979}{1000} \times 5 \times 10^{-6} + \frac{20}{1000} \times \frac{1}{1000} + \frac{1}{1000} \times \frac{1}{100} \right)^{-1} \approx 28\,650.$$

With prior odds of $\frac{1}{1000}$, the probability that Q is not the source decreases from $\frac{1}{201} \approx 0.5\%$ to $\frac{1}{29.65} \approx 3.4\%$.

What if we were not given any information about the relatives of Q ?

Paternity Testing

Paternity and familial identification can provide evidence in criminal context and during civil litigation. For a paternity case, the two propositions could be:

H_p : The alleged father (AF) is the true father.

H_d : Some other (unrelated) man is the father.

The likelihood ratio is in this case often referred to as the paternity index (PI).

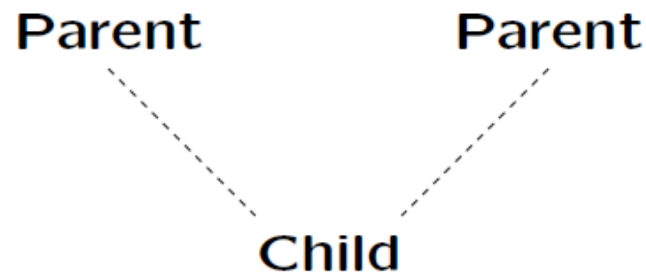
Missing Persons

For a missing person case, the two propositions could be:

H_p : The sample is from the missing person.

H_d : The sample is from some unknown person.

The genetic evidence E may consist of the genotype from a sample that has come from some person X who may be the missing person, together with the genotypes from the parents of the missing person.



Familial Searching

A serial killer nicknamed the Grim Sleeper (due to a 14-year break) was responsible for the death of at least 10 young women in Los Angeles between 1985 and 2007.

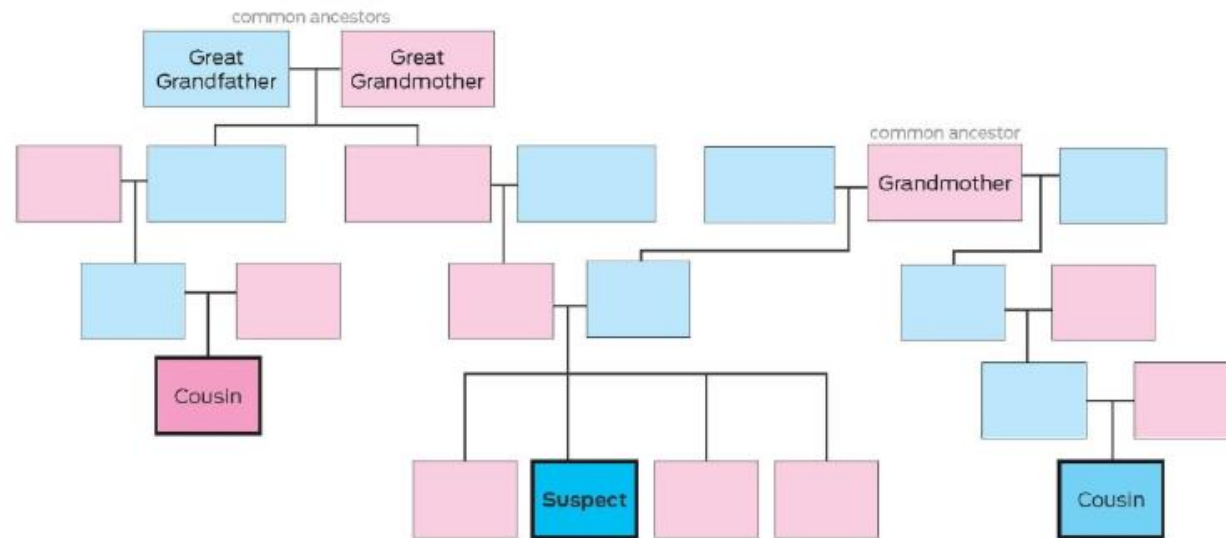
When traditional forensic methods failed, investigators turned to novel partial-match DNA search methods authorized in 2008, eventually leading to a positive result for a recently convicted young man. Together with other evidence this led to the suspicion of the father.

The L.A. police was notified by investigators and got a DNA sample from a discarded piece of pizza. Lonnie Franklin was found to match, leading to an arrest in July 2010 and eventual conviction in May 2016.

Genealogical Searching

Cook/Van Cuylenborg Double Homicide Cold Case

Suspect family tree based on genetic genealogy



If you have information related to this case, please call 425-388-3845



Snohomish County Sheriff's Office

Source: Technique Used to Find Golden State Killer Leads to a Suspect in 1987 Murders (Murphy, 2018).