DNA Testing 101

An Introduction to the Various Tests Available and their Application to Genealogy Presented by Bob Mallett

Ottawa TMG Users Group

ottawa-tmg-ug.ca

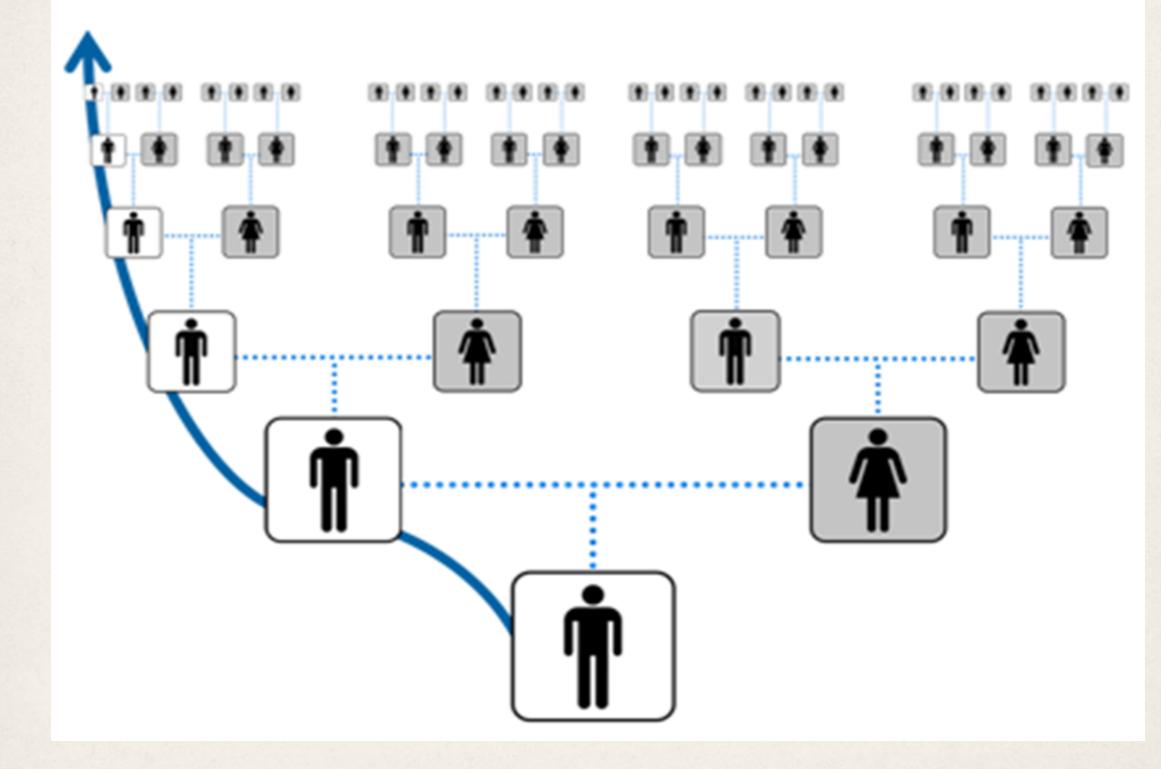
Introduction

There are 4 main categories of tests, each with its own application:

- 1. Y-STR Male line only, father to son
- 2. Y-SNP or Deep Clade Male line only, but (typically) deeper ancestry
- 3. Mithochondrial (mtDNA) Female line only mother to child (son or daughter)

4. Autosomal – All lines

Y-Adam



STRs (Short Tandem Repeats) are short sequences of DNA that are repeated numerous times at a given location on a particular chromosome. Mutations occur from time to time and the number of repeats can increase or decrease.

We simply count the number of repeats at a given location (a marker).

The actual count has no particular significance. What's important is how the counts compare to another's result.

Several levels of the test are available, ranging from 12 to 111 markers . The more markers the better, providing a more refined result. 37 markers is the norm now for an entry level test.

Estimates of relatedness between 2 individuals are based on the number of mismatches (genetic distance), and the mutation rates of markers, using a complicated algorithm.

saturday, battom, line is that there can't be many mismatches.

Results from the Mallett Surname Y-DNA Study:

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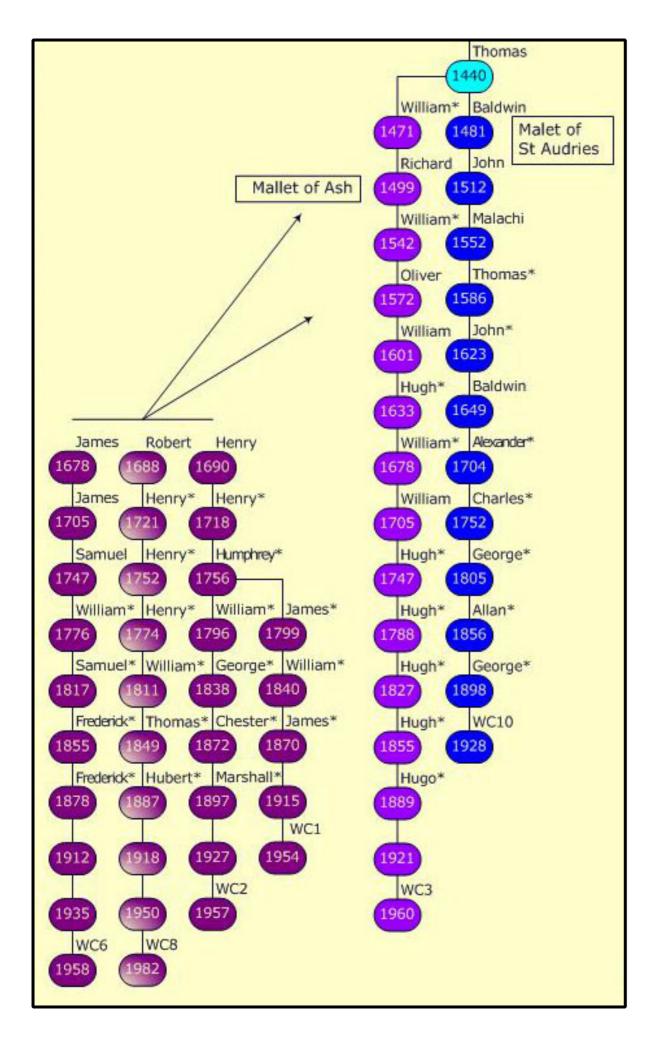
Probability of a relationship:

Genetic Distance = 1:

- 4 generations is 58.69%
- 8 generations is 88.85%
- 12 generations is 97.40%
- 16 generations is 99.44%
- 20 generations is 99.88%
- 24 generations is 99.98%

Genetic Distance = 6:

- 4 generations is 0.21%
- 8 generations is 5.17%
- 12 generations is 21.88%
- 16 generations is 46.07%
- 20 generations is 68.25%
- 24 generations is 83.57%



Devon / Cornwall and Mallet of Ash

The accompanying graphic illustrates the probable connection between three North Devon lines and the Mallet of Ash line suggested by the conventional research and bolstered by the DNA results.

The average genetic distance of the 3 families on the left compared to Mallet of Ash is 3, giving the following probabilities of relatedness:

...4 generations is 22.18%.

...8 generations is 66.49%.

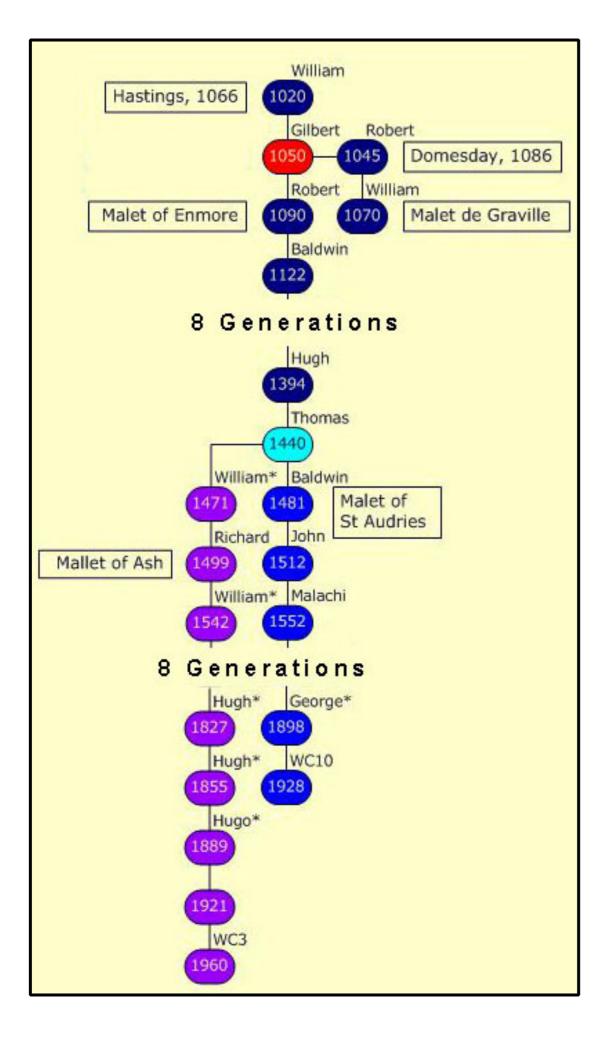
...12 generations is 89.97%.

...16 generations is 97.56%.

...20 generations is

99.48%.

Recently a family of Malletts born in the mid 1600's was found in Bratton Clovelly, a village not far from the others, that provides a hard link for at least one of the families, and may be the missing link for all of them.



Malet of Enmore and St Audries (Somerset, England) Mallet of Ash (Devon, England)

The Malets first appear in Somerset in the mid 12th century. It is assumed that they are related to the Domesday Malets, but this has never been proven beyond doubt.

The first Malet in Somerset in connection with this line was Robert c 1090 at Enmore, in Somerset The Enmore line died out, but two spinoff branches continued; one was located at St Audries (West Quantoxhead) Somerset, the other in Iddesleigh, North Devon (Mallet of Ash). Both families are extant in the male line. Their probability of relatedness is as follows (GD of 6):

...4 generations is 0.89%.

-8 generations is 14.77%.
- ...12 generations is 45.23%.
- ...16 generations is 73.27%.

...20 generations is 89.43%.

Several other Mallett families have been identified that are genetically related to one another, but not to the other family groups, not even within the last 1000 years.

Some are tantalisingly close, but others are so far apart that there is absolutely no possibility of a relationship.

Many individuals have tested who match no-one else in the study.

There may have been a genetic relationship between at least some of the families at one time, but the link may have been broken because of an "NPE" (Non Paternal Event) or "Not the Parent Expected" ③.

Conventional research might identify such an event, or it may be so far back in time that we will never know.

SNP = Single Nucleotide Polymorphism:

"Variation in the nucleotide allele at a certain nucleotide position in the human genome. When the change occurs it is called a polymorphism, and polymorphisms accumulate over time. A polymorphism can be very common (found in a significant fraction of global or localized populations) or very rare (found in a single individual). Common variations are used to track the evolution of the human genome over time (population genetics) and can be graphically represented in a haplogroup or phylogenetic tree."

(From ISOGG Y-DNA Glossary).

ISOGG = International Society of Genetic Genealogy

Unlike STRs, which can mutate at any time, once a SNP occurs, it remains – hence its value for population genetics and for genealogy.

Thousands of SNPs have been identified, and more are being discovered all the time as more people take the "Big Y" test at FTDNA, or "Geno 2.0" at National Geographic (there are other tests too).

Once a SNP has been identified it is placed on the Y-DNA Haplogroup tree at ISOGG and FTDNA (ISOGG is the most up to date). The tree begins with "Y-Chromosome Adam", who is thought to have lived about 60,000 years ago. R-M269, the dominant haplogroup in Western Europe, is thought to have occurred 4000 to 10,000 years ago (Wikipedia: Haplogroup R1b).

Since the advent of "Big Y" and other tests, many downstream SNPs have been discovered, and it is thought that at least some of them will fall within a genealogical timeframe, making SNP testing useful to Genealogists as well as Population Geneticists.

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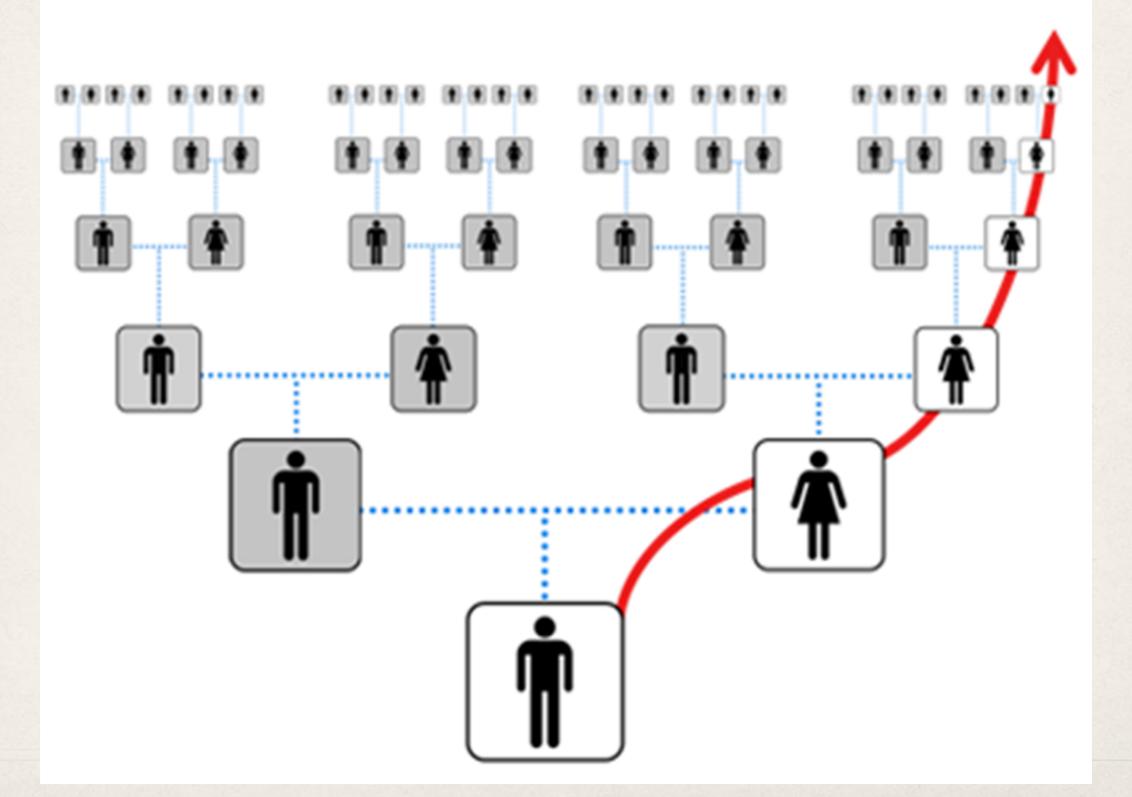
Individual SNPs can be tested for about \$30.00. But how does one know which SNP to test?

There exists a community of people trying to make SNP predictions based on STR values, and they are working with FTDNA to group likely SNPs together into "panels", much like the 1 to 12, or 23 to 15 marker panels for STR testing, so that one can pursue SNP testing at a reasonable cost.

We're not there yet. At the moment it's the proverbial needle in a haystack.

Mithochondrial

Mithochondrial Eve



Mithochondrial (mtDNA)

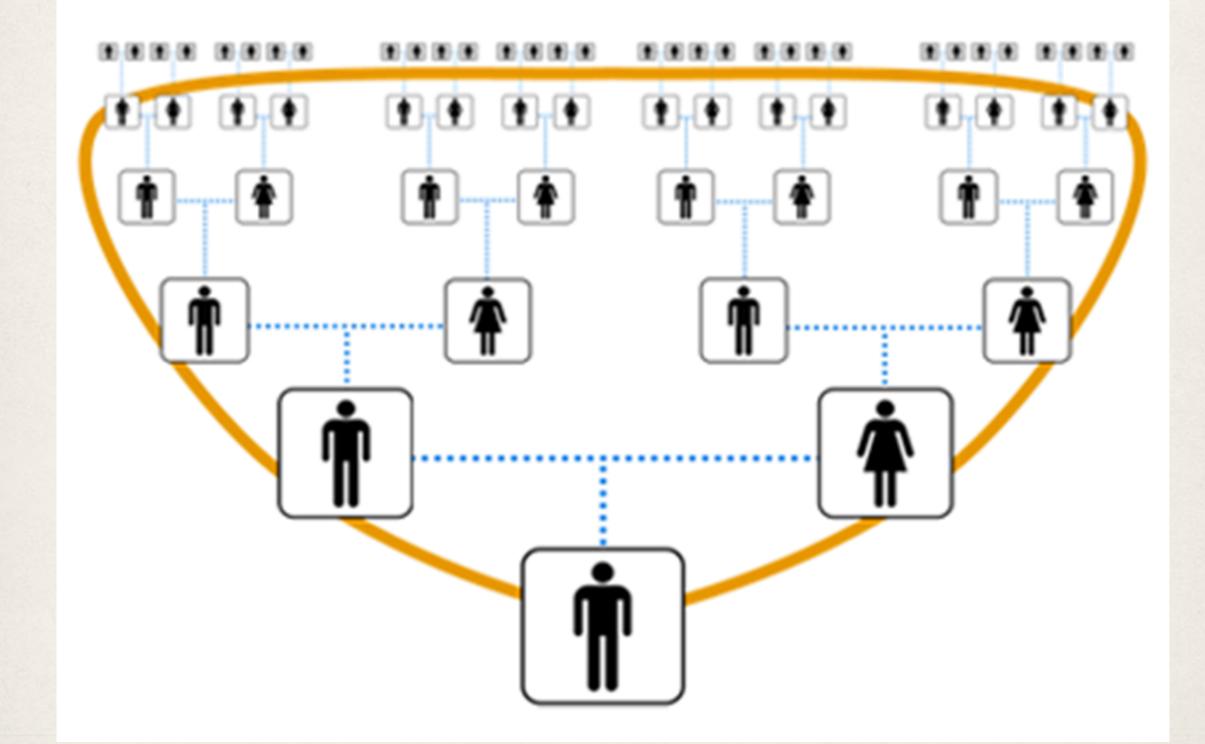
FTDNA currently offers 3 levels of mtDNA testing:

- 1. mtDNA
- 2. mtDNAPlus
- 3. mtFullSequence

Only mtFullSequence offers a probability of relationship at a high enough level to be really useful within genealogical time (95% confidence at 22 generations).

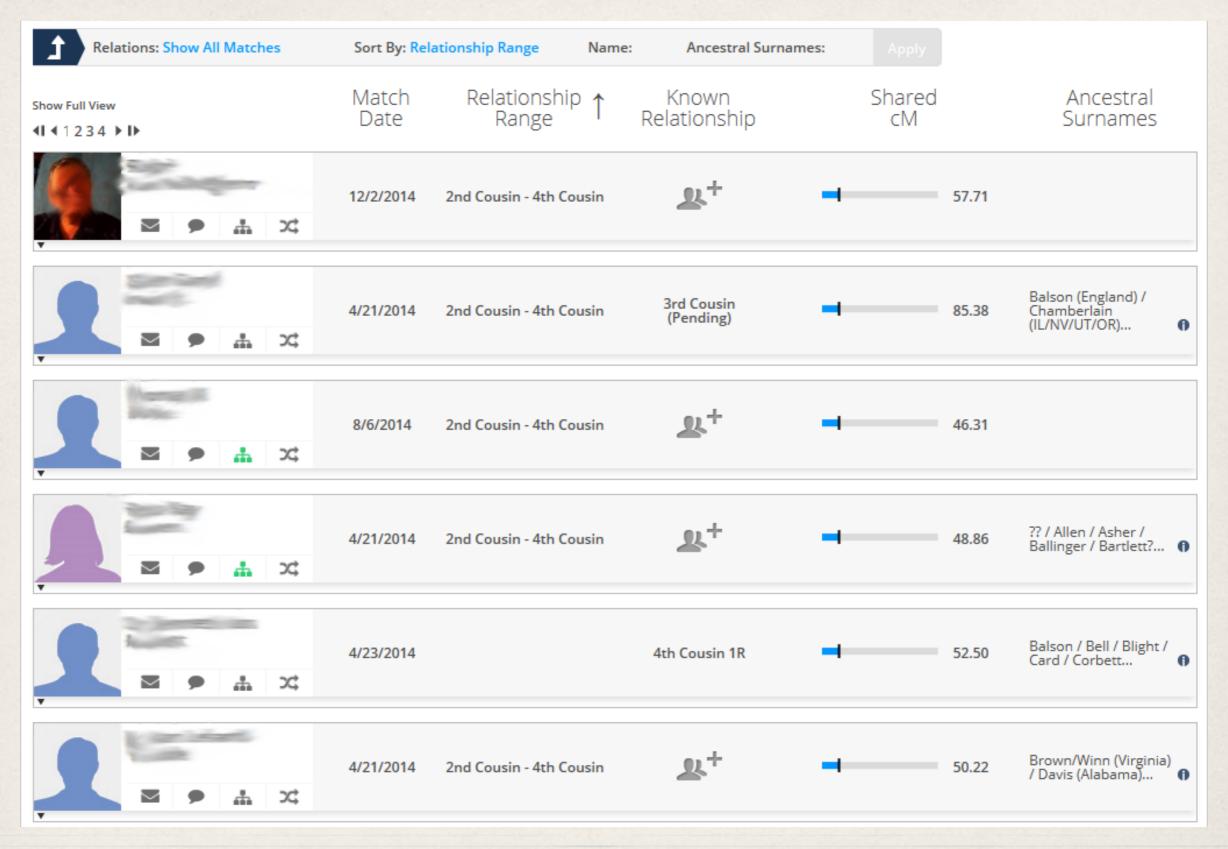
The other 2 tests can still be helpful as an adjunct to paper research, but they tend to point to deep ancestry. mtDNAPlus only allows for a confidence level of 50% at 28 generations. mtDNA only reaches 50% at 52 generations.

An mtDNA test can be interesting to determine one's deep maternal ancestry, e.g. from which of "The Seven Daughters of Eve" does one descend?



Autosomal DNA statistics 50% Mother, father, siblings 25% Grandparents, aunts, uncles, half-siblings, double first cousins 12.5% First cousins 3.125% Second cousins 0.78% Third cousins 0.195% Fourth cousin 0.049% Fifth cousins 0.0122% Sixth cousins 0.003052% Seventh cousins Chances of matching 2nd cousin >99%3rd cousin > 90%4th cousin > 50% 5th cousin > 10% 6th cousin or more distant Remote (typically less than 2%)

Matching segements are measured in Centimorgans (cM).



cM = Centimorgan: the distance between chromosome positions.

Family Finder - Chromosome Browser

Feedback Refer Friends & Family Page Tour

Chromosome Browser Tutorial

Optional Views: • Download to Excel (CSV Format)

View this data in a table > Download All Matches to Excel (CSV Format)



Filter: 10 cM Prediction: 2nd to 4th Cousin

Actual Relationship? Orange: 3rd Cousin Blue: 4th Cousin once Removed Green: No Idea Pink: No Clue

Family Finder - Chromosome Browser

Feedback Refer Friends & Family Page Tour

Chromosome Browser Tutorial

Optional Views: • Download to Excel (CSV Format)

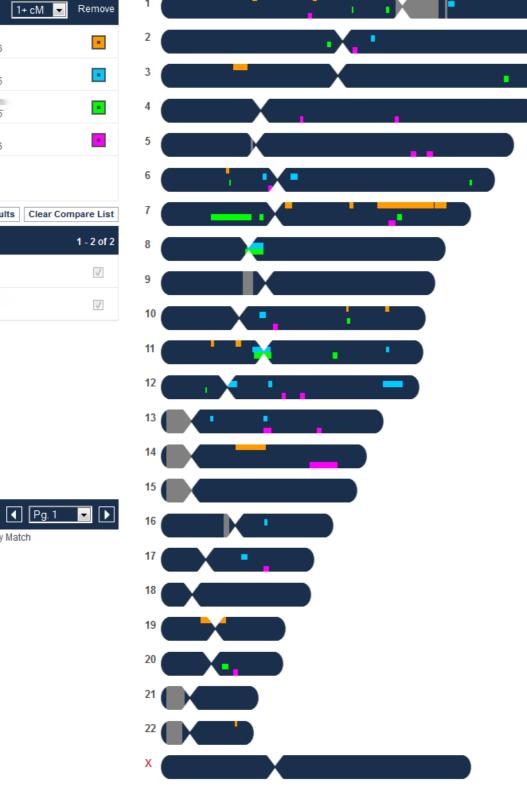
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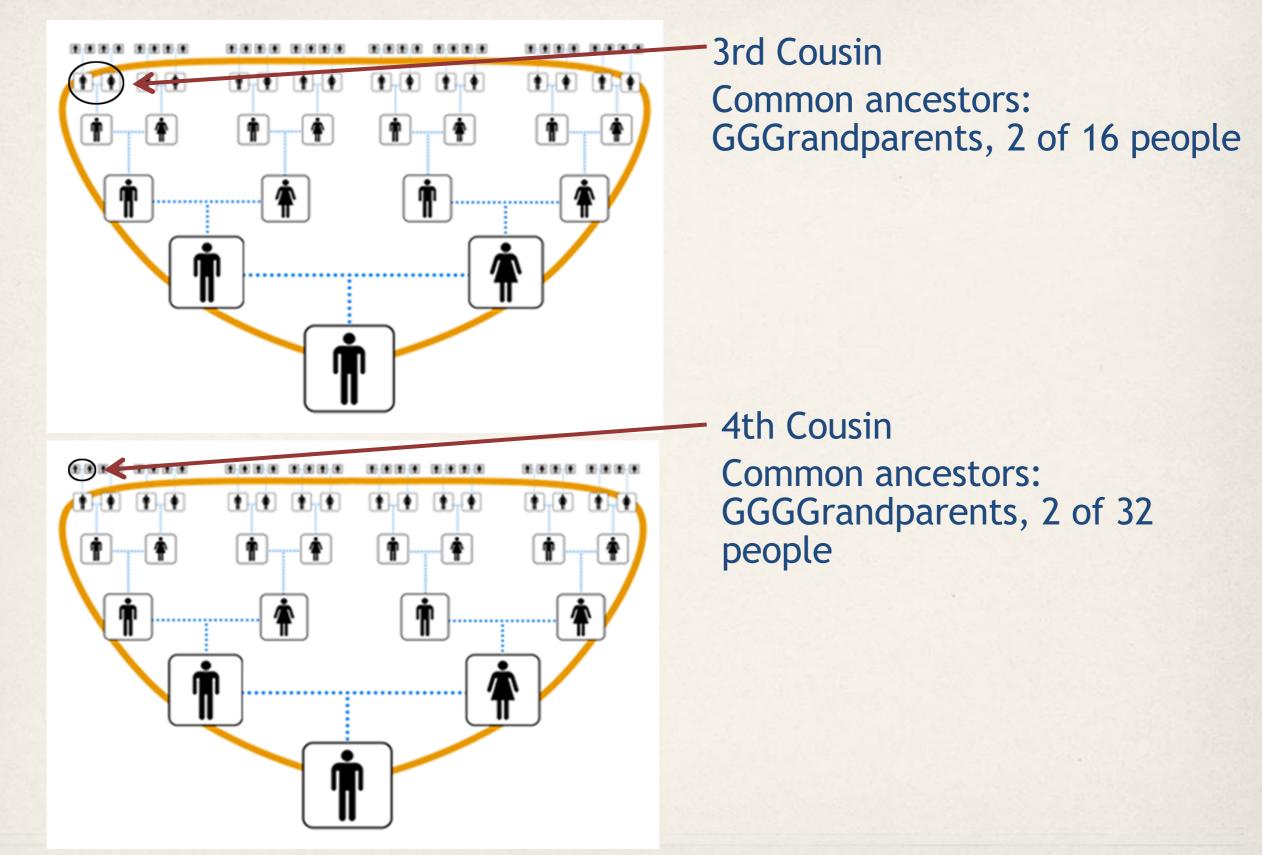
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Actual Relationship? Orange: 3rd Cousin Blue: 4th Cousin once Removed Green: No Idea Pink: No Clue

Saturday, January 3, 2015

Hide 3rd Party Matches

* Indicates Uploaded 3rd Party Match



What about the "Unknown" 2nd to 4th Cousins? Do we have a common ancestor?

The matching DNA could have come from any of the 56 people 3 generations and more back from me. The task of finding the common ancestor among so many possibilities is daunting. How does one go about it?

Research as many lines as possible, as far back as possible.

Look for shared surnames, and particularly shared rare surnames.

Look for shared geographical locations.

Concentrate on matches with the closest predicted relationships (fourth cousins and closer).

Focus on matches with shared segments of 10 cMs or above (20 cMs for Ashkenazi Jews).

The higher threshold for Ashkenazi Jews comes from the fact that they tended to marry within a restricted group. The same thing applies to families from remote areas, such as certain Newfoundland Outports, where a few families intermarried over many generations.

This sort of thing can lead to false positive matches, where segments are considered to be "Identical by State (IBS)", not coming from a single common ancestor.

The term "Identical by Descent (IBD)" is used to describe matching segments that probably do come from a common ancestor.

There are many advanced techniques being developed and used to find common ancestors, too many and too complicated to go into here, but they all require a good deal of time, effort, and additional testing.

Tests are available from 23andMe, FTDNA, and AncestryDNA.

Family Finder - Chrome	osome Browser	Feedback	Refer Friends & Family	Page Tour
Chromosome Browser Tutorial	Optional Views: Download to Excel (CSV Format)	► View this data in a table ► Download All Matches to	Excel (CSV Format)	
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Filter: 1 cM Prediction: Full Siblings

Actual Relationship? Orange: Sister Blue: Sister

Feedback Refer Friends & Family Page Tour

Family Finder - Chromosome Browser

5+ cM 💌

Chromosome Browser Tutorial

Compare List

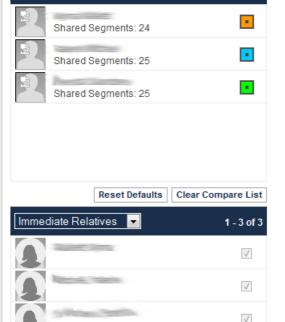
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View this data in a table > Download All Matches to Excel (CSV Format)



Filter: 1 cM Prediction: Parent/Child

Actual Relationship? Orange: Daughter Blue: Daughter Green: Daughter

Saturday, January 3, 2015

Hide 3rd Party Matches

* Indicates Uploaded 3rd Party Match

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Enter the actual values from the test.

This is simply a log of results, attached to a particular individual, but nothing shows up in the person view.

One can enter a "group", but this just seems to be text. Different individuals are not linked by this value.