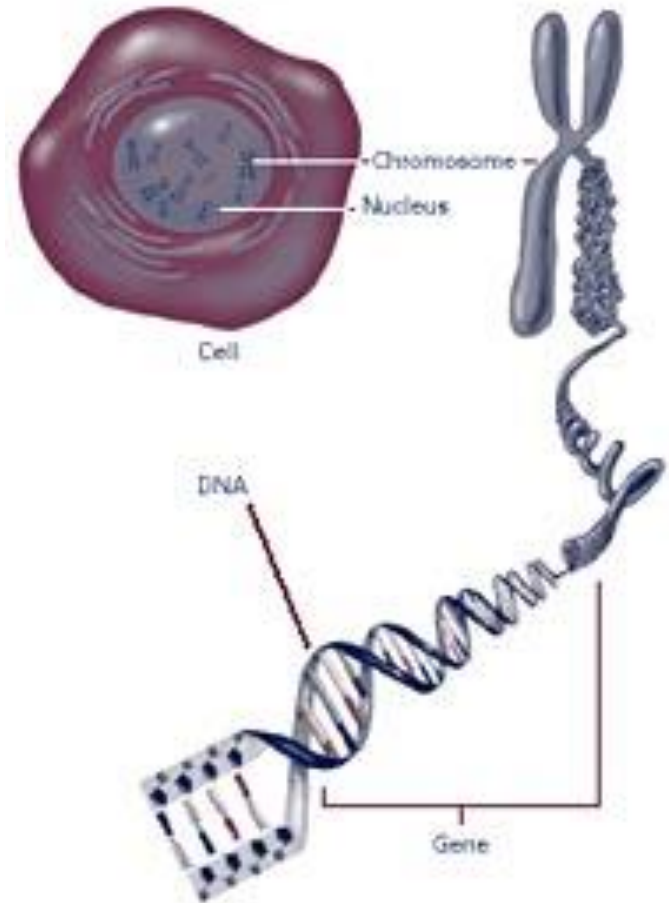




# DNA Testing

February 16, 2018





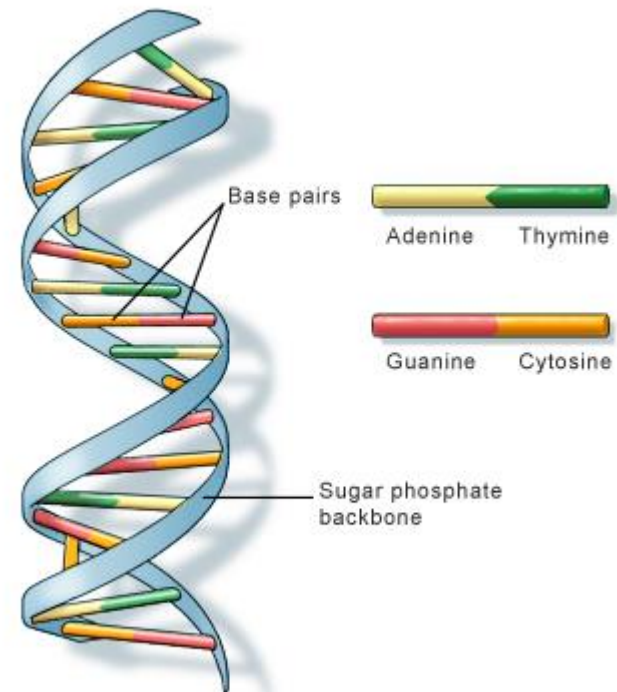
# What Is DNA?

- Double helix – “ladder” structure where the rungs are molecules called “nucleotides” or bases. DNA contains only four of these nucleotides – A, G, C, T
- The sequence that these nucleotides occurs will determine what protein is made. These proteins then will contribute to “you” – all of your genetic and “phenotypic” characteristics
- By reading the sequence of these nucleotides and comparing to sequences commonly found in geographic areas, your ethnicity can be predicted



# Human DNA

- The entire DNA or genome within a human contains about 6 billion nucleotides
- 99% are identical in all humans; the 1% difference results in individuals

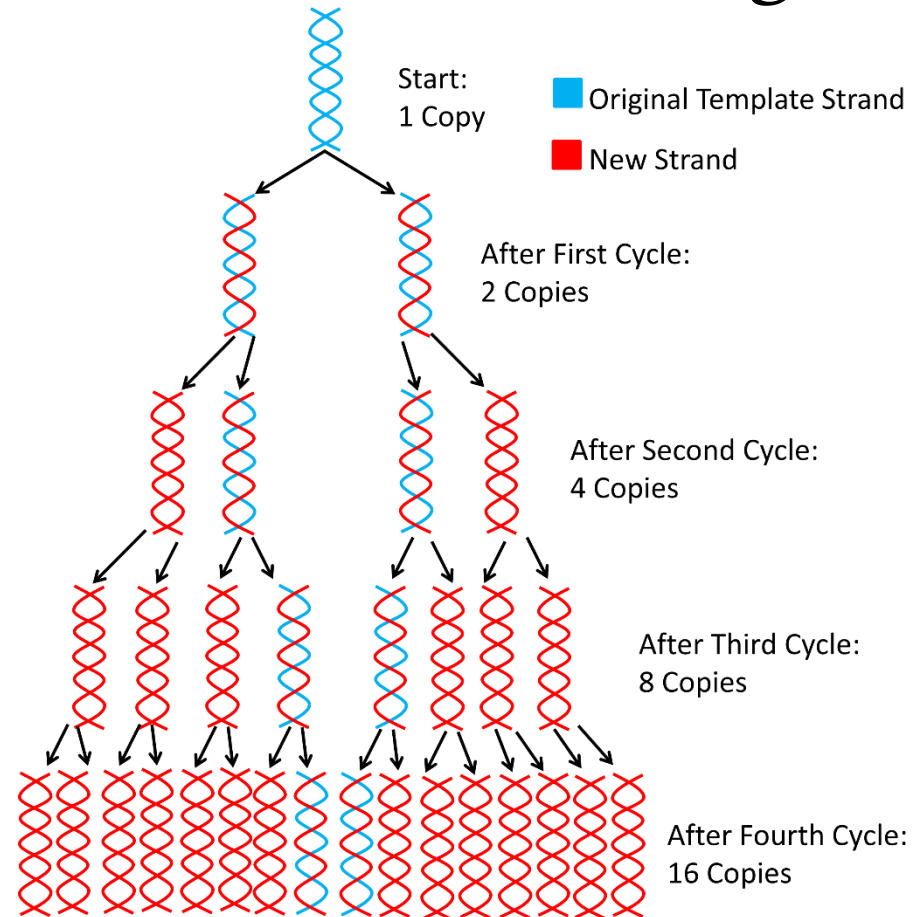


U.S. National Library of Medicine



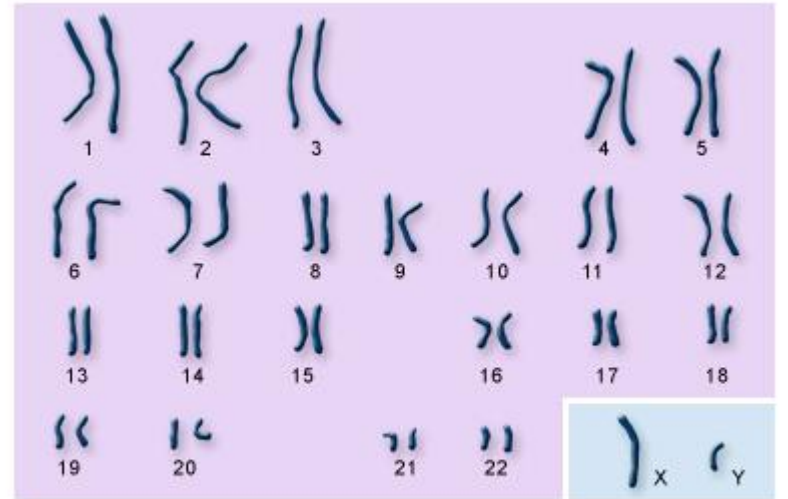
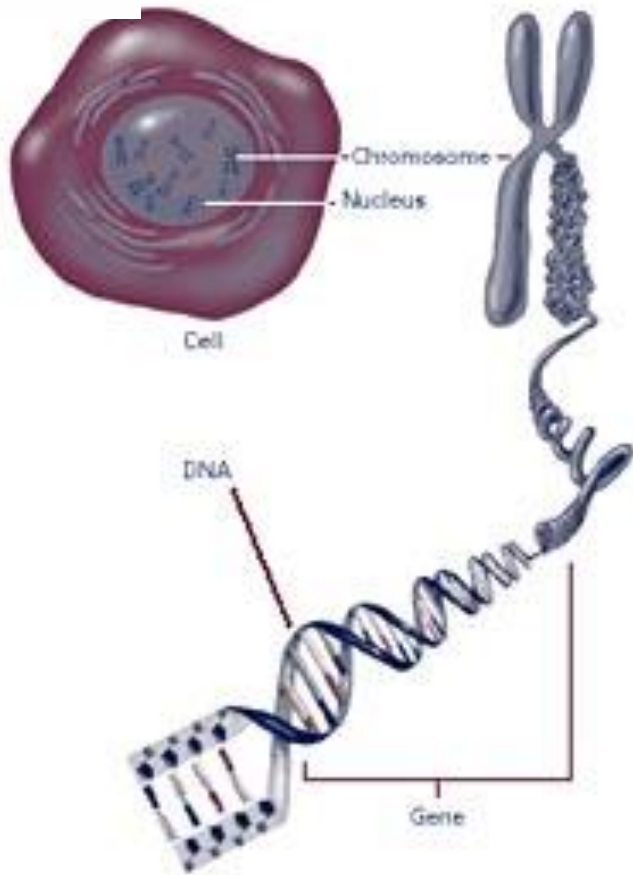
# The Science Behind DNA Testing

- The DNA is isolated from your sample (saliva, cheek cells, etc.) and millions of copies are made, using a method called 'polymerase chain reaction', or PCR.
- PCR uses a naturally occurring enzyme to copy a specific stretch of DNA over and over again.





# Testing DNA “Snippets”



autosomes

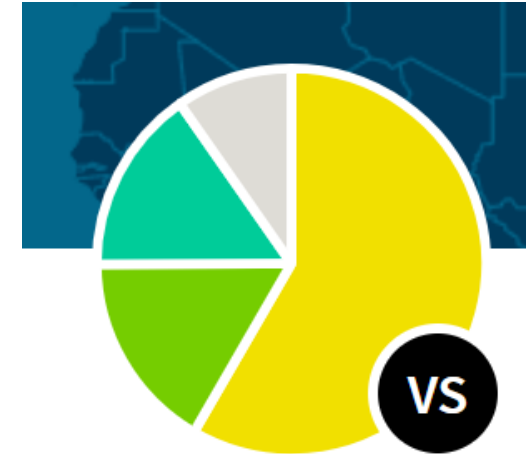
sex chromosomes

U.S. National Library of Medicine



## Why Send in Your DNA for Testing?





- Ancestry will send you a pie chart with your predicted ethnicity percentages
- Ancestry will continually search for relatives based on shared DNA and send you email alerts
- But – don't trade in your lederhosen yet for a kilt – your ethnicity % are estimates
- Each company uses different databases and “best guesses” as to your background (more info to follow)



Ethnicity Estimate	
Scandinavia	58%
Sweden	
Europe West	17%
Ireland/Scotland/Wales	15%
Low Confidence Regions	
Great Britain	4%
Europe South	3%
Iberian Peninsula	1%



# Which Company Should You Choose?

Company	Features
	<ul style="list-style-type: none"><li>42+ ethnic regions, some of which are exclusive to MyHeritage</li><li>More focused ethnicity specifics than other main-market solutions</li><li>Great experiential results including animations and soundtrack</li><li>Easy integration with other MyHeritage features (some of which require additional payment)</li><li>8 billion historical documents, 300 million photographs, 34 million family trees. 2.1 billion names</li></ul>
	<ul style="list-style-type: none"><li>Autosomal DNA testing</li><li>“Shared Ancestor Hints” find intriguing connections for you</li><li>Specific results: 150 ethnic regions, 5 times more than the next leading DNA test</li><li>Connect with 90,000,000 family trees and billions of historical records</li><li>DNA samples are stored forever, so you never have to re-test</li><li>Create “DNA Circles” of probable relatives</li></ul>
	<ul style="list-style-type: none"><li>Highly-detailed results broken down to regions within countries</li><li>Tracing ethnic heritage back 10 generations</li><li>Y, mt and autosomal testing</li><li>Interactive map to follow your ancestors migrations</li><li>Highly-advanced testing that implements the know-how of over 100 expert scientists</li><li>Provides raw data results which can be used on other genealogy sites</li></ul>
	<ul style="list-style-type: none"><li>Inheritance tracing</li><li>Ancestry percentages to the 0.1%:</li><li>Chromosome breakdown of ancestry: See which pieces of your DNA come from 31 populations worldwide</li><li>Compare DNA Relatives' segments</li><li>Opt-in health and ancestry option</li></ul>



- 23andMe **reference datasets** include genomes from 10,418 people who were carefully chosen to reflect populations that existed before transcontinental travel and migration were common (at least 500 years ago).
- Most of the reference individuals are 23andMe customers who have consented to participate in research. When a 23andMe research participant tells us that they have four grandparents all born in the same country — and the country isn't a colonial nation like the US, Canada, or Australia — that person becomes a candidate for inclusion in the reference data.



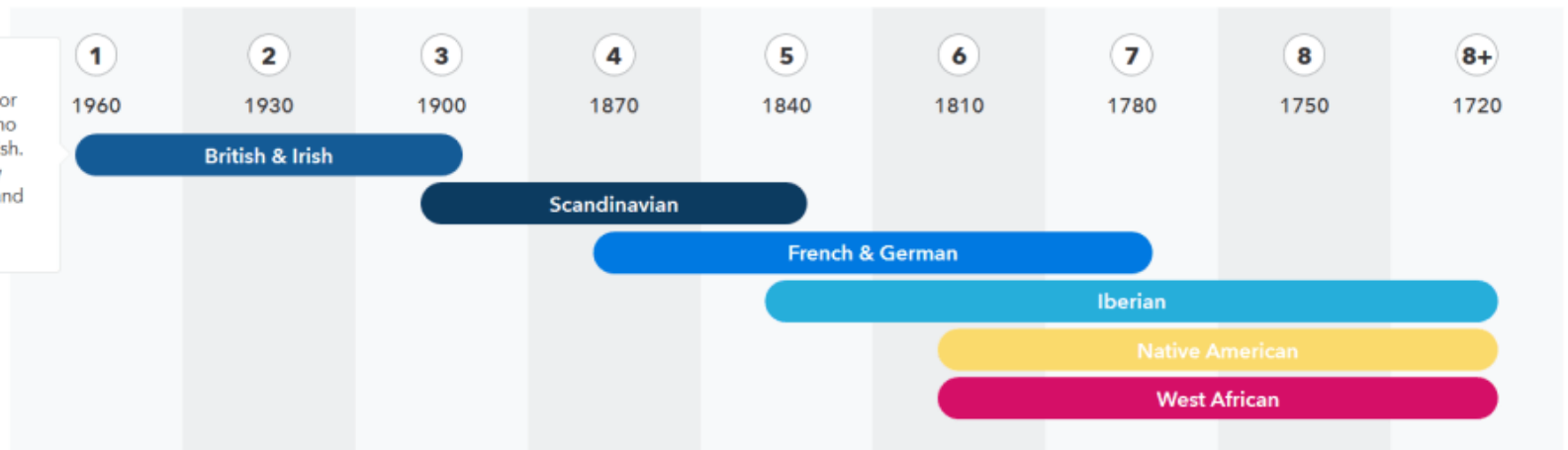


# 23andMe Unique Features

## Your Ancestry Timeline

How many generations ago was your most recent ancestor for each population?

You most likely had a parent, grandparent, or great-grandparent who was 100% British & Irish. This person was likely born between 1900 and 1960.





# Ancestry.com

- Largest and most diverse database compared to other testing kits: connect with 90,000,000 family trees and billions of historical records
- Over 6 million users
- Offers information both on the distant past, as well as more recent relatives and your family tree
- Pricing is one of the lowest on the market (\$79)
- Will search for matches for you and email the connections to you
- One con: does not provide health information



# Ancestry DNA Testing

- Ancestry uses microarray-based autosomal DNA testing that surveys the entire genome at over 700,000 sites
- Autosomal DNA simply means the DNA found in a chromosome inherited from both your father and mother, and contains bits of genetic sequences from past generations.
- Close relatives share large fragments from a common ancestor, while the matched fragments get smaller from more distant ancestors



## What Kinds of DNA Tests Do Genealogy Companies Use?

- The Y chromosome test allows you to follow only the paternal line. Males carry one Y chromosome that determines gender. If XX, then the child is female; is XY, the child is male. The Y test follows the inherited Y chromosome sequences. This test can go back as far as 25 generations.
- The mitochondrial (mt) DNA test follows the maternal line. Only females pass on the small amount of DNA found in the mitochondria of cells. This test also goes back 25 generations.
- The autosomal DNA test looks at locations on all chromosomes that companies have decided are best “ethnic” matches. Therefore, this test follows both maternal and paternal. But, this test can only go back as far as 5-6 generations.



# Autosomal DNA Testing

- The chance that an autosomal DNA test will accurately detect a relative decreases with the distance of the relationship. For example, most autosomal DNA ancestry tests predict an accuracy rate of 90–98% when detecting a match with a 3rd cousin, but around a 45–50% chance of detecting a match with a fourth cousin.
- The amount of autosomal DNA shared with a relative decreases with each successive generation.
  - 50% - parents and siblings
  - 25% - grandparents, half-siblings, aunts/uncles, double first cousins
  - 12.5% - first cousins
  - 6.25% - first cousins, once removed
  - 3.125% - second cousins, first cousins twice removed



## Confidence Levels in Ancestry

- The confidence score for your DNA matches is based on the amount and location of the DNA that you share with that potential match. A high confidence score means that its almost certain that the DNA sequence you share with a match is identical because it was inherited from a recent common ancestor.
- The confidence score is related not to whether you are 4<sup>th</sup> cousins or not, but that you are related through a common ancestor (discuss more on the website demonstration).
- The chance of a match decreases with each successive generation
  - 90-98% when detecting a match with a third cousin
  - 45-50% with a fourth cousin



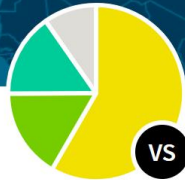
# Ancestry DNA Results



Hello, virginia

This test is shown to matches as shephev  Linked to Virginia Johnson

## DNA Story ?



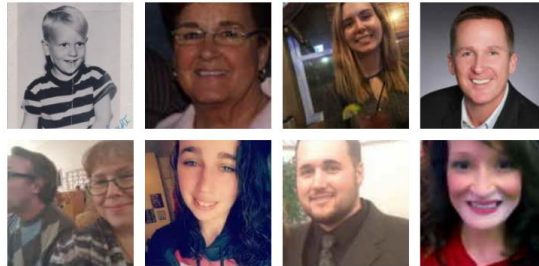
### Ethnicity Estimate




- 58% Scandinavia
- 17% Europe West
- + 6 Other regions

Discover the places, history, and cultures that shaped who you are today—using just your DNA.

DISCOVER YOUR DNA STORY

## DNA Matches ?



-  42 Shared Ancestor Hints
-  0 Starred matches
-  352 4th cousins or closer

VIEW ALL DNA MATCHES

## DNA Circles 5 ?



Confidence: Weak

### Hardy Keel DNA Circle

4th Great-Grandfather

25 MEMBERS

VIEW ALL DNA CIRCLES



# DNA Test of Siblings

**DNA Story** ?

**Ethnicity Estimate**

- 47% Great Britain
- 23% Scandinavia
- + 5 Other regions

Discover the places, history, and cultures that shaped who you are today—using just your DNA.

[DISCOVER YOUR DNA STORY](#)

17% Germany  
23% Scandinavia  
9% Ireland/Scotland/Wales

**DNA Story** ?

**Ethnicity Estimate**

- 35% Great Britain
- 29% Europe West
- + 3 Other regions

Discover the places, history, and cultures that shaped who you are today—using just your DNA.

[DISCOVER YOUR DNA STORY](#)

29% Germany  
21% Scandinavia  
14% Ireland/Scotland/Wales





# Mother-Daughter Results



Gisele Grayson  
Ancestry Composition

## Ancestry Composition

Your genome tells the unique story of your ancestry: where your ancestors lived, when they contributed to your family tree, and how their DNA was passed down to you through your parents. For more information about your results, see [Frequently Asked Questions](#).



Gisele Grayson		100%
<b>European</b>	<b>99.6%</b>	
Northwestern European	90.5%	
French & German	50.2%	
British & Irish	23.3%	
Broadly Northwestern European	17.0%	
Southern European	6.1%	
Iberian	2.4%	
Italian	1.6%	
Broadly Southern European	2.0%	
Broadly European	3.0%	
<b>South Asian</b>	<b>&lt; 0.1%</b>	
Broadly South Asian	< 0.1%	
<b>Sub-Saharan African</b>	<b>&lt; 0.1%</b>	
East African	< 0.1%	
<b>East Asian &amp; Native American</b>	<b>&lt; 0.1%</b>	
Broadly East Asian & Native American	< 0.1%	
<b>Unassigned</b>	<b>0.3%</b>	



Carmen Grayson  
Ancestry Composition

## Ancestry Composition

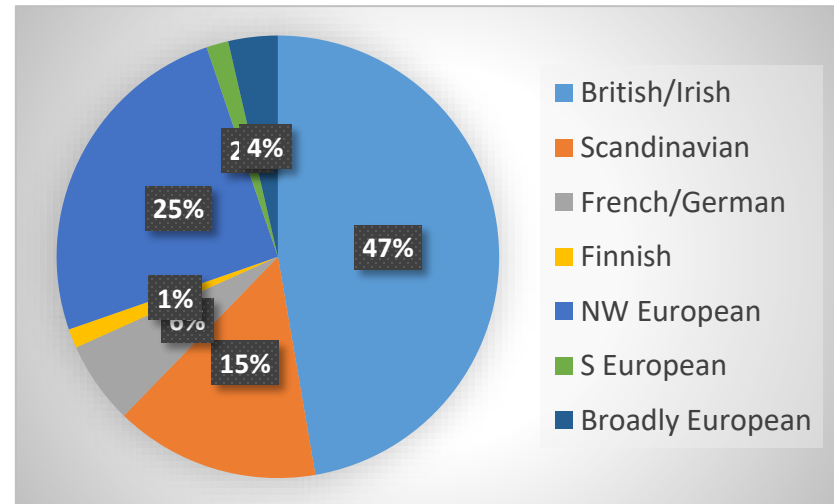
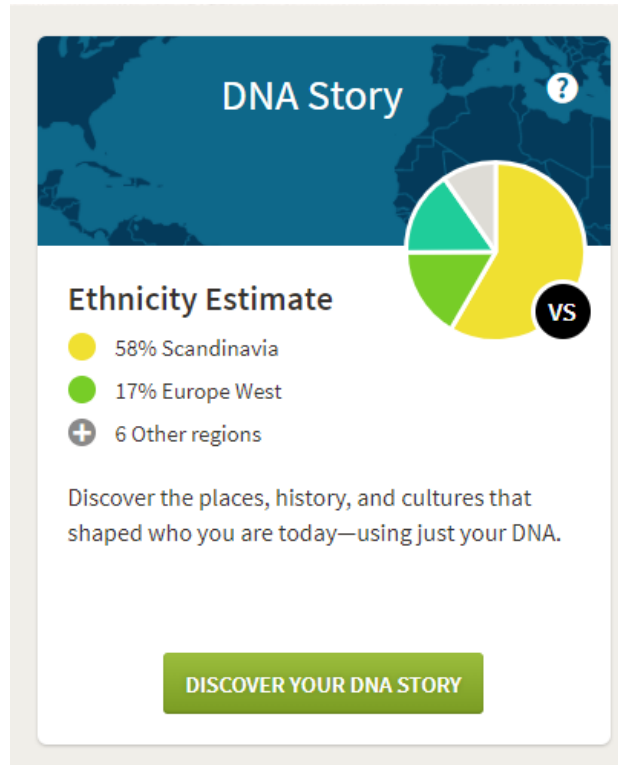
Your genome tells the unique story of your ancestry: where your ancestors lived, when they contributed to your family tree, and how their DNA was passed down to you through your parents. For more information about your results, see [Frequently Asked Questions](#).



Carmen Grayson		100%
<b>European</b>	<b>99.8%</b>	
Northwestern European	66.1%	
French & German	52.1%	
British & Irish	6.3%	
Broadly Northwestern European	7.7%	
Southern European	25.4%	
Italian	11.3%	
Iberian	1.6%	
Broadly Southern European	12.5%	
Eastern European	2.9%	
Broadly European	5.4%	
<b>East Asian &amp; Native American</b>	<b>&lt; 0.1%</b>	
Native American	< 0.1%	
<b>Unassigned</b>	<b>0.1%</b>	



# Mother –Son Results





## Using the Shared DNA Tool in Ancestry

- The Shared Matches tool will show which matches you and any given match on your list share in common. You can use this new tool to help narrow down your matches to a particular side of your family. It's especially helpful if you've had a parent tested because once you have a parent tested, you'll see a new filter at the top of your match list that lets you find the DNA matches that you share with your mom or dad.
- I have had a 1<sup>st</sup> cousin tested, and I can now see all the matches we share. From this list I can possibly narrow down to those matches that might stem from a common ancestor, and not have to search through all the matches.



# Using the Shared Tool

Look at this DNA Match	To focus on this family line
Mom, dad, aunt, uncle, 1st cousin (on maternal or paternal line depending on which side you're interested in)	Maternal or paternal line ( $\frac{1}{2}$ of your tree)
Maternal grandparent or 2nd cousin	Maternal grandparent's line ( $\frac{1}{4}$ of your tree)
Paternal grandparent or 2nd cousin	Paternal grandparent's line ( $\frac{1}{4}$ of your tree)
Maternal great-grandparent or 3rd cousin	Maternal great-grandparent's ( $\frac{1}{8}$ of your tree)
Paternal great-grandparent or 3rd cousin	Paternal great-grandparent's ( $\frac{1}{8}$ of your tree)



# DNA Testing Limitations

- DNA tests analyze less than 1 percent of a person's genome; they will miss most of a person's relatives.
- DNA snippets, or markers, are inconsistent. Sometimes they are passed on and sometimes they are not, and whether they are or aren't is random.
- When a DNA test comes back saying you are 28% Finnish, all it's really saying is that of the DNA analyzed, 28% of it was most similar to that of a completely Finnish person (and what does "completely Finnish mean?").
- People are continuously moving and reproducing with other, diverse people. If anything our DNA is getting more muddled, not more clear.



# Examining the Ancestry DNA site in detail

