

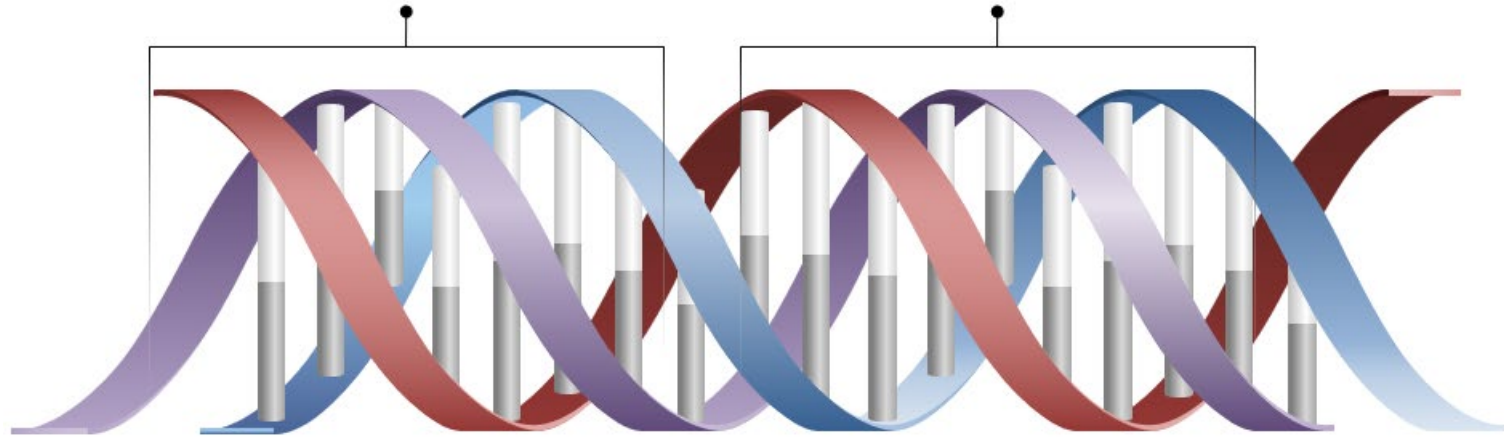


Overview of DNA and Genetic Genealogy

OLLI

February 14, 2020

Photo Credit: Tim Jeffries



Introduction to DNA and Genetic Genealogy

Presenter: Kathleen Reed
Hamilton County Genealogical Society

What Is DNA?



DNA, or deoxynucleic acid, is a sequence of chemicals that defines each human being as unique (with the exception of identical twins). It is in the shape of a double helix.

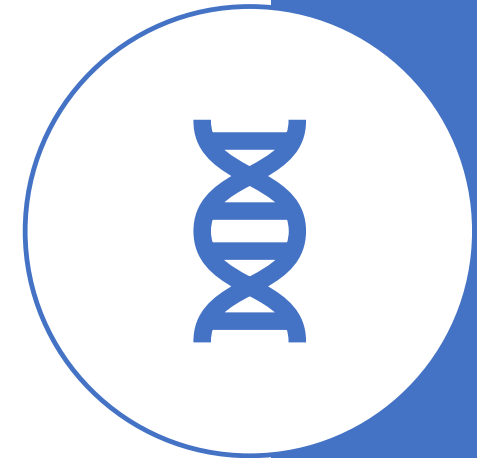
The spirals are held together by four nucleotides or base pairs: adenine (A), guanine (G), cytosine (C) and thymine (T). **A** always pairs chemically with **G** and **C** always pairs with **T**.



DNA, used in conjunction with genealogical research, is known as “genetic genealogy.”

What genetic genealogy is . . .

- It is another tool in the genealogist's toolbox.
- It can be used to both rule in and rule out genetic relationships.
- It can be used to identify “deep” ancestry.
- Some, but not all, types of testing can reveal health information.





What genetic genealogy is not . . .

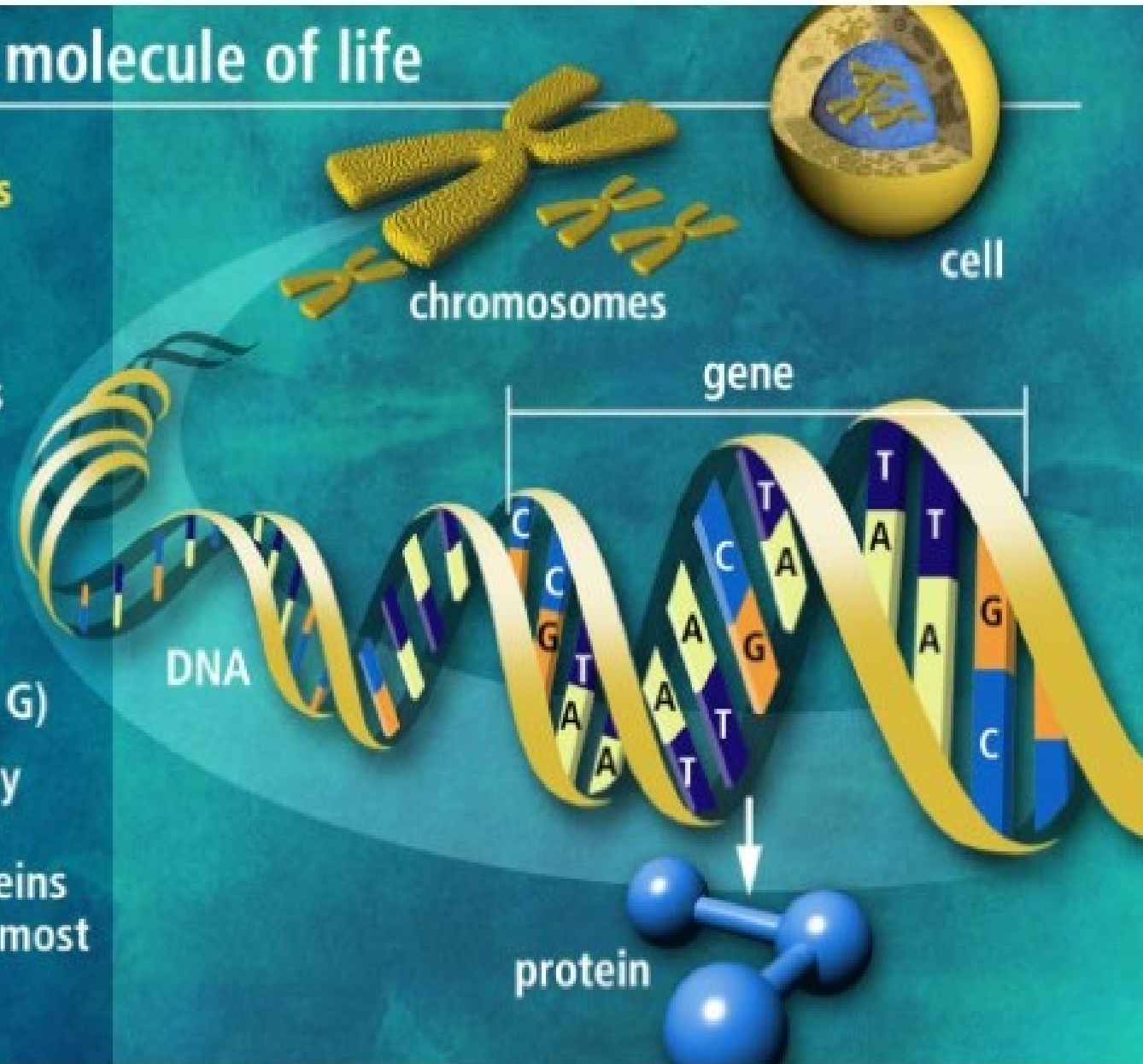
- It is not a replacement for traditional genealogical research
- It does not include a family tree
- It cannot directly tell you the degree of relationship
- It cannot tell you who your great-grandmother was or where she was born.

DNA the molecule of life

Trillions of cells

Each cell:

- 46 human chromosomes
- 2 meters of DNA
- 3 billion DNA subunits (the bases: A, T, C, G)
- Approximately 30,000 genes code for proteins that perform most life functions



There are four types of DNA



Autosomal DNA (atDNA)



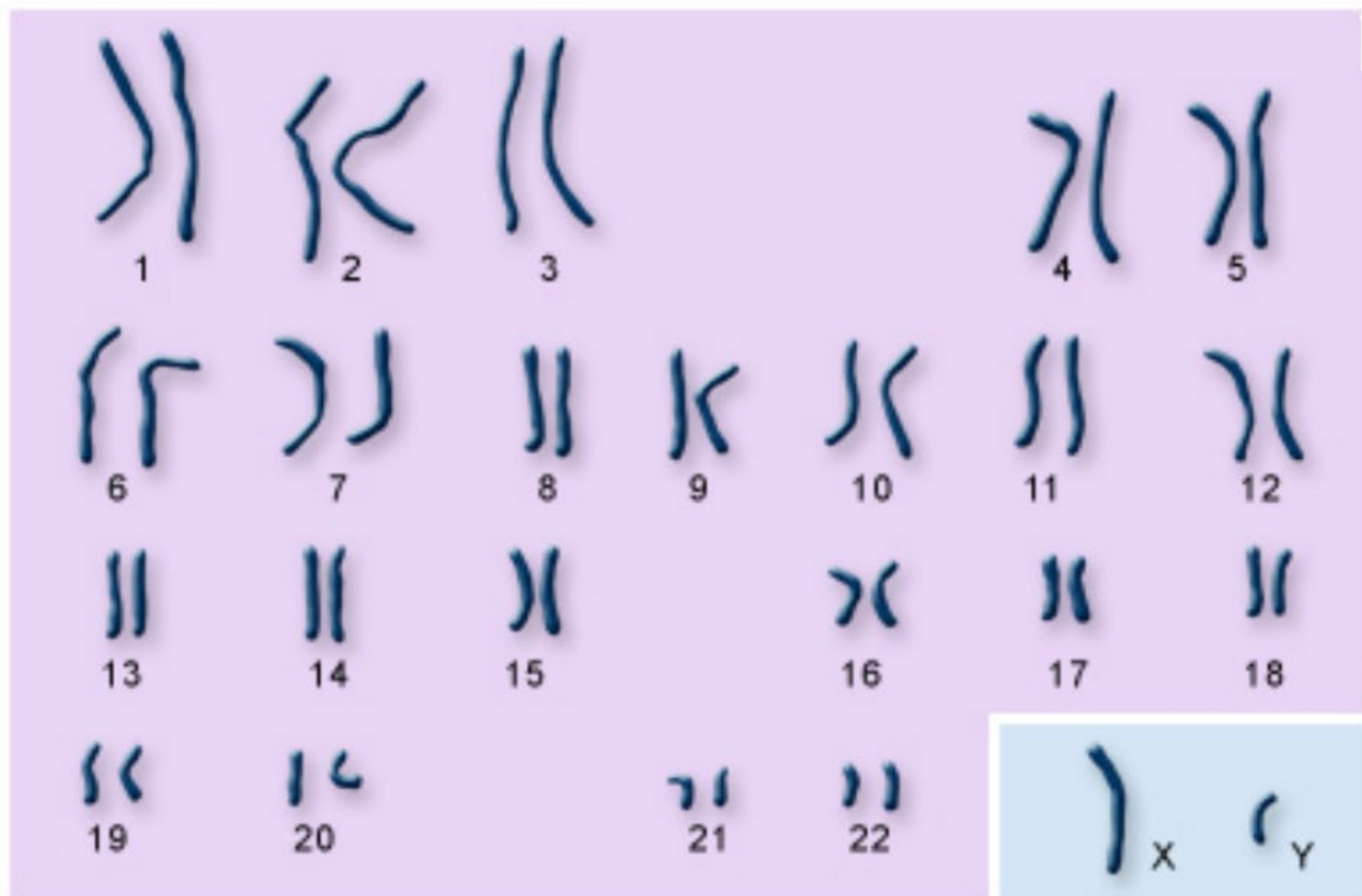
Mitochondrial DNA (mtDNA) (passed in a direct line from a mother to her children)



Y-DNA (passed in a direct line down the paternal line)

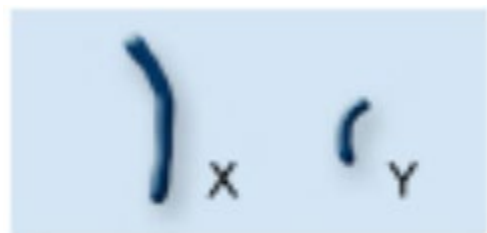


X-DNA (found on the X chromosome)

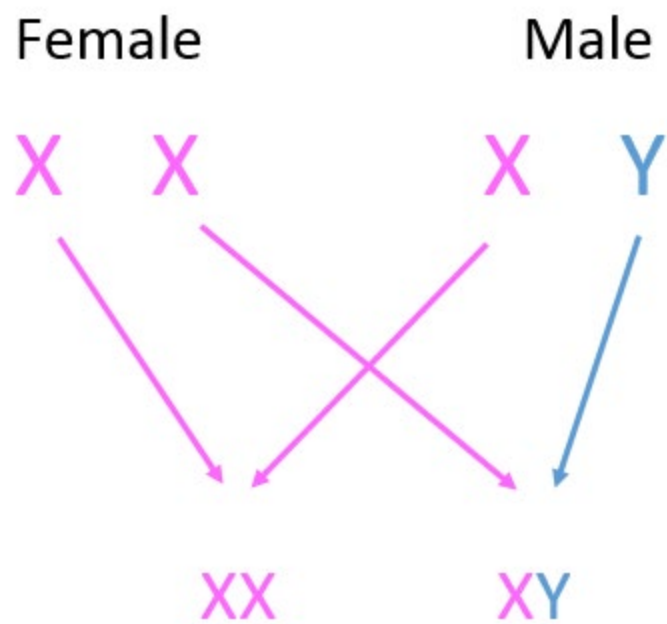


autosomes

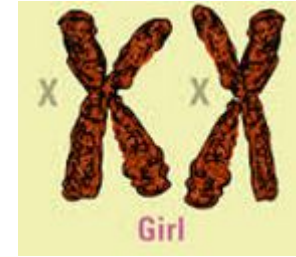
sex chromosomes



sex chromosomes



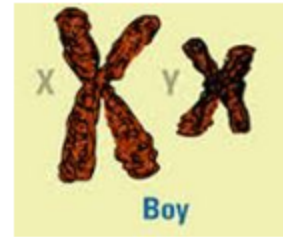
Females



The 23rd chromosome in a female is composed of two X-chromosomes, one inherited from the mother and one from the father. Like the members of the other 22 chromosome pairs, in the female, the 23rd pair form a matched set, can exchange genetic material, and either member can be passed to the next generation. Sons inherit one X-chromosome from their mother.



Males



In the male, the 23rd pair of chromosomes contains an X-chromosome inherited from the mother and a Y from the father. Because the Y-chromosome is non-recombinant and is handed down by a father only to his sons, it can be used in tracing the exclusively male line of a family. This makes it useful for “surname” studies.

Y-DNA Is Passed From Father to Son

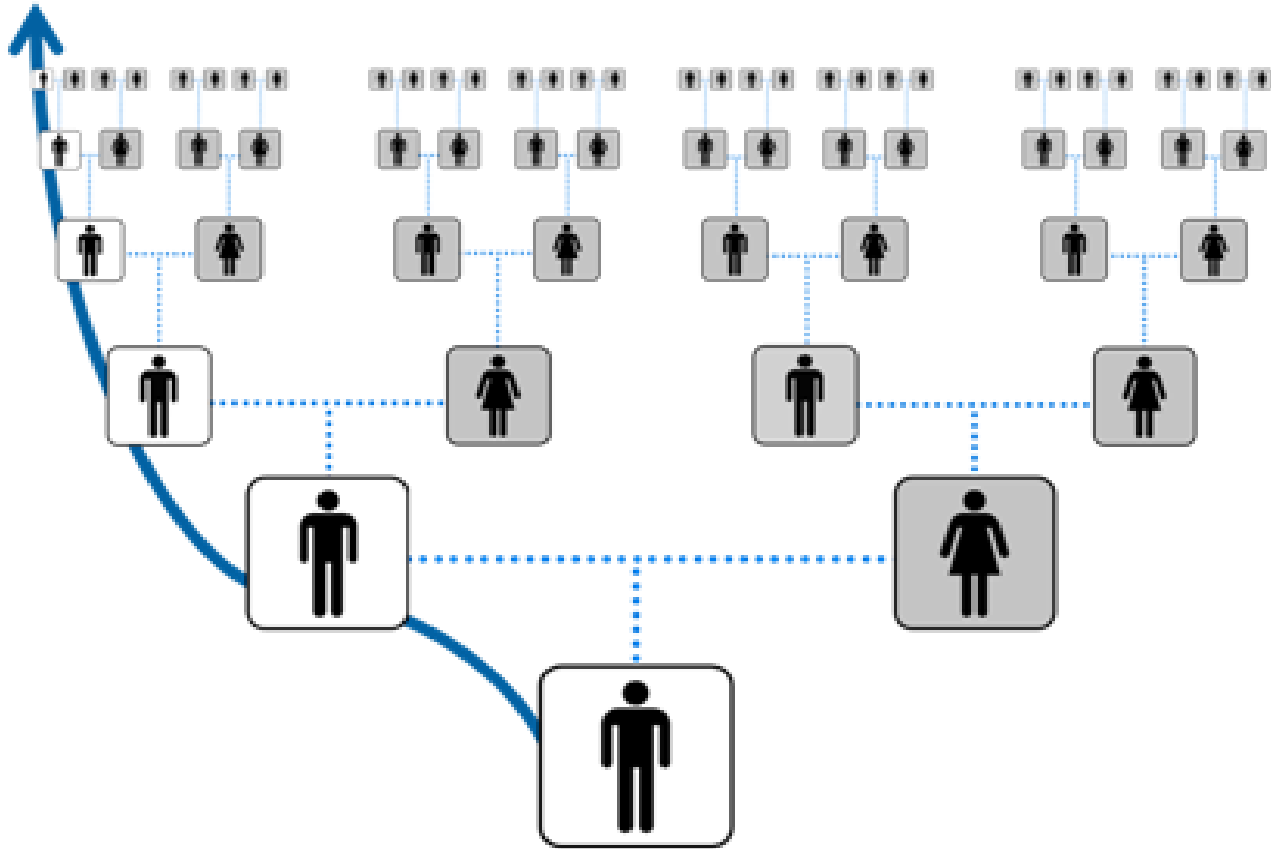
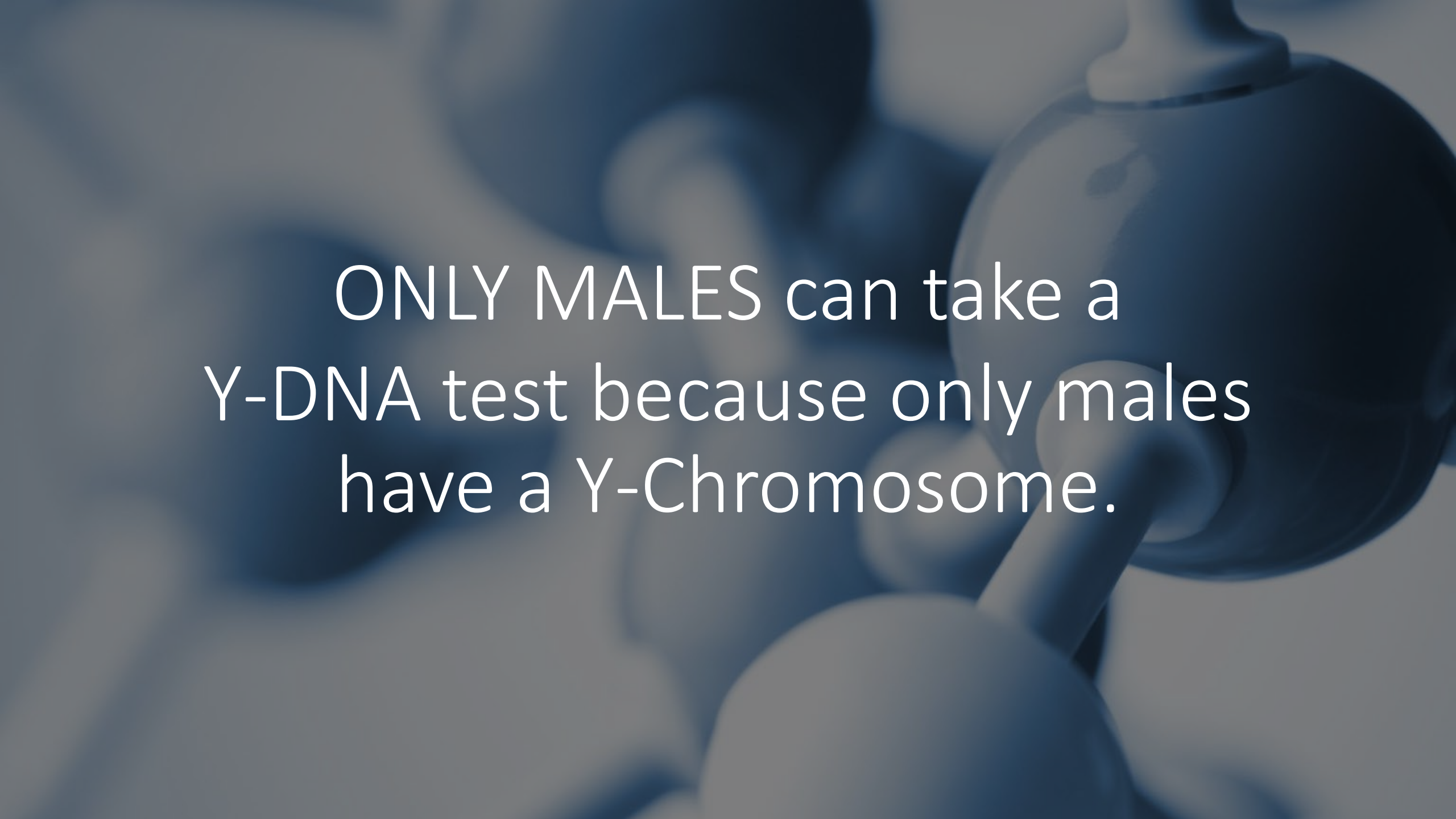
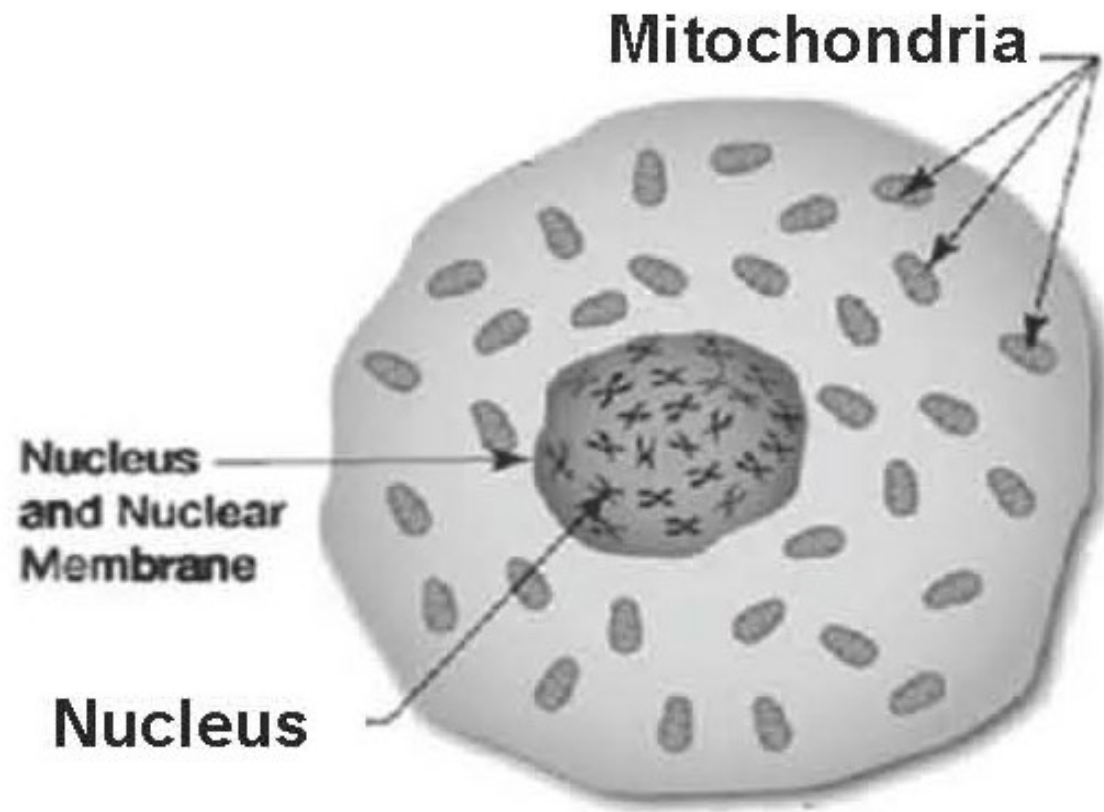


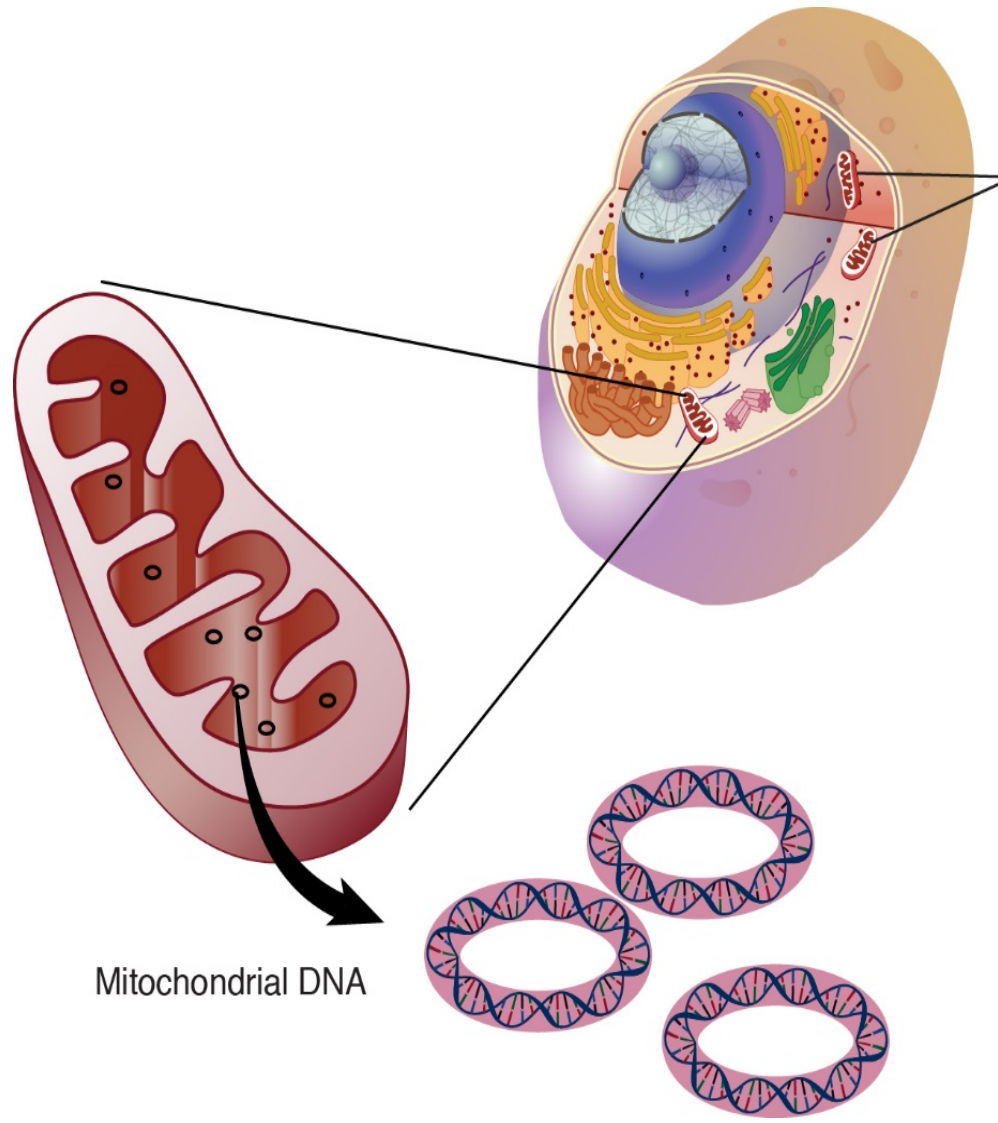
Image from familytreedna: Used with permission



ONLY MALES can take a
Y-DNA test because only males
have a Y-Chromosome.

Kinds of DNA: mitochondrial DNA (mtDNA) and nuclear DNA (Y-DNA and autosomal)





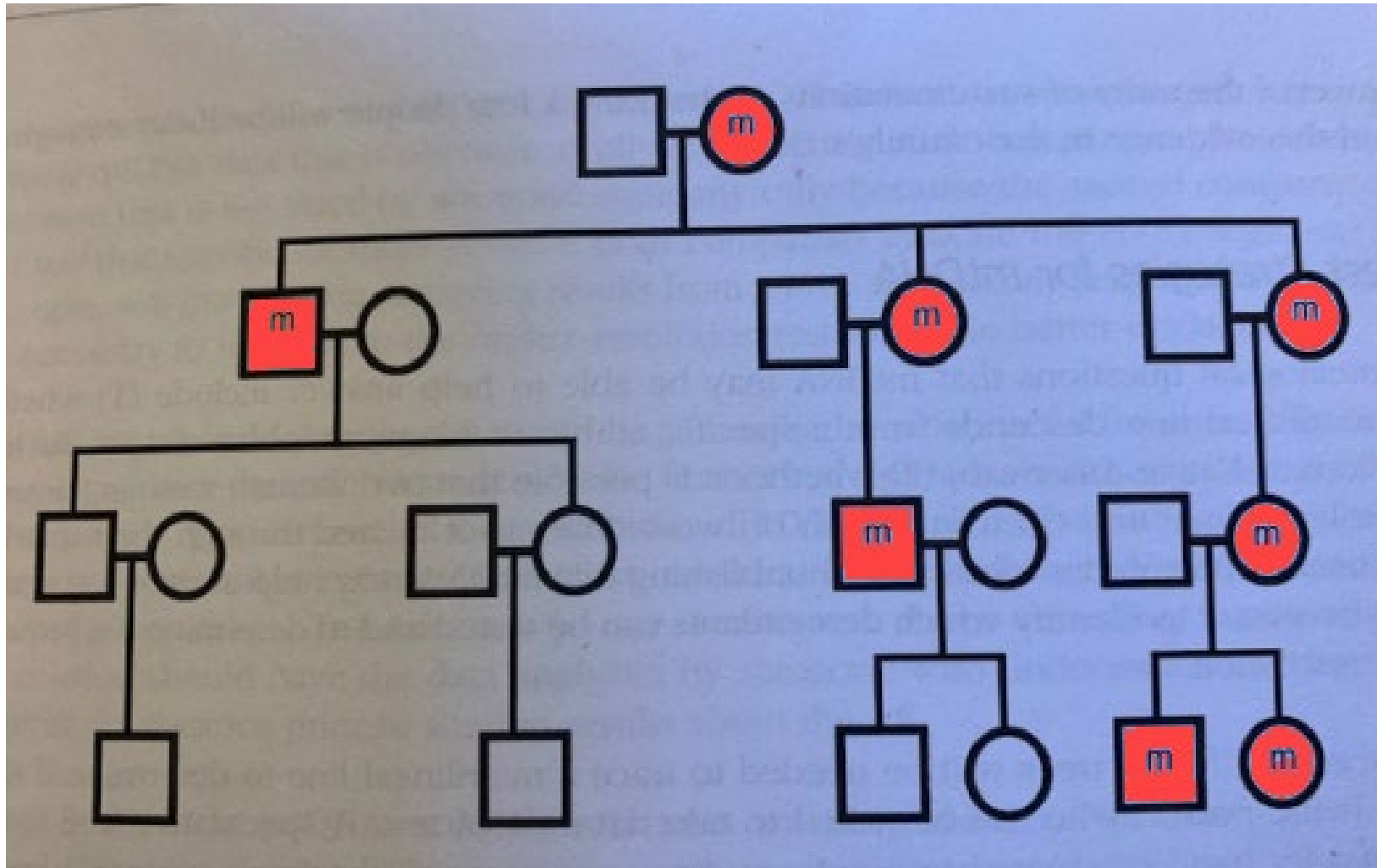
Mitochondria

Mitochondrial DNA is the small circular chromosome found inside mitochondria. The mitochondria are organelles found in cells that are the sites of energy production. The mitochondria, and thus mitochondrial DNA, are passed from mother to offspring.

Mitochondrial DNA

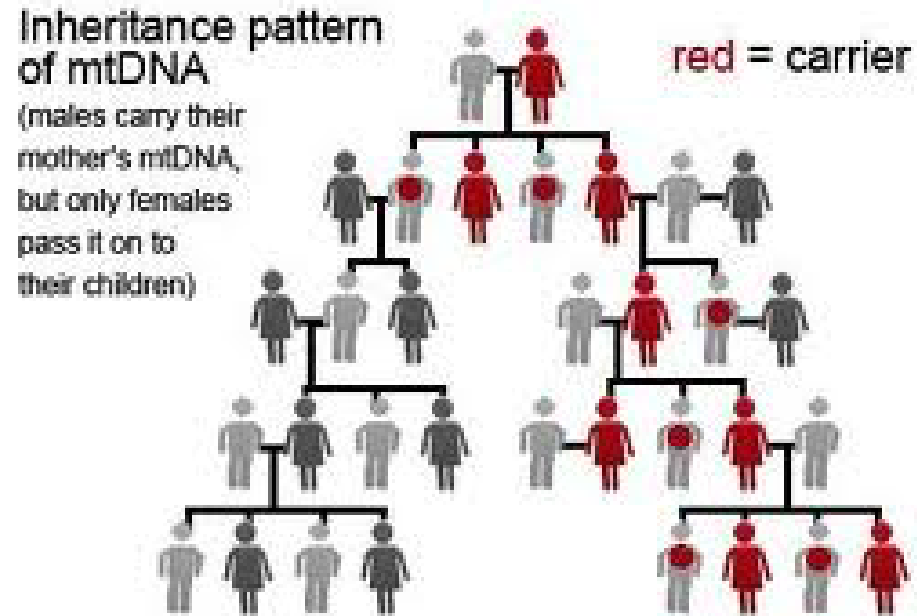
Everyone can take a Mitochondrial DNA Test because mtDNA is inherited from the mother. However, only females can pass mtDNA onto their descendants.

Mitochondrial DNA Inheritance Pattern



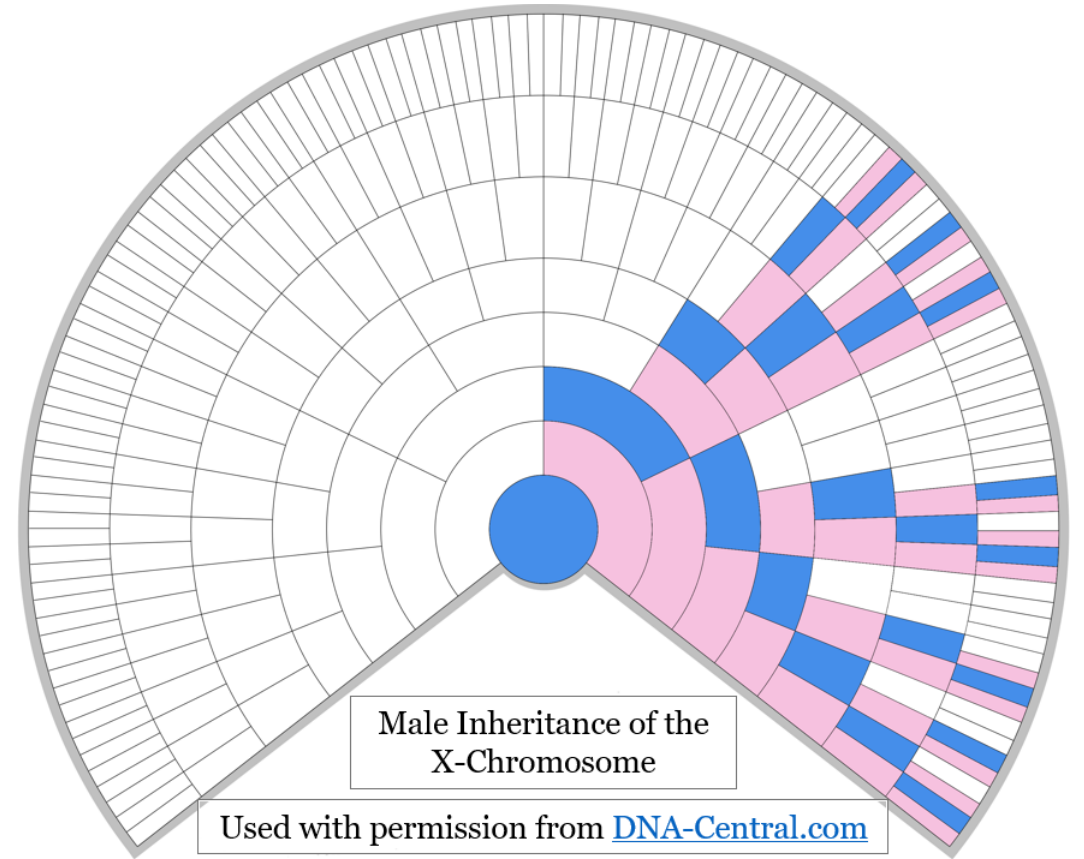
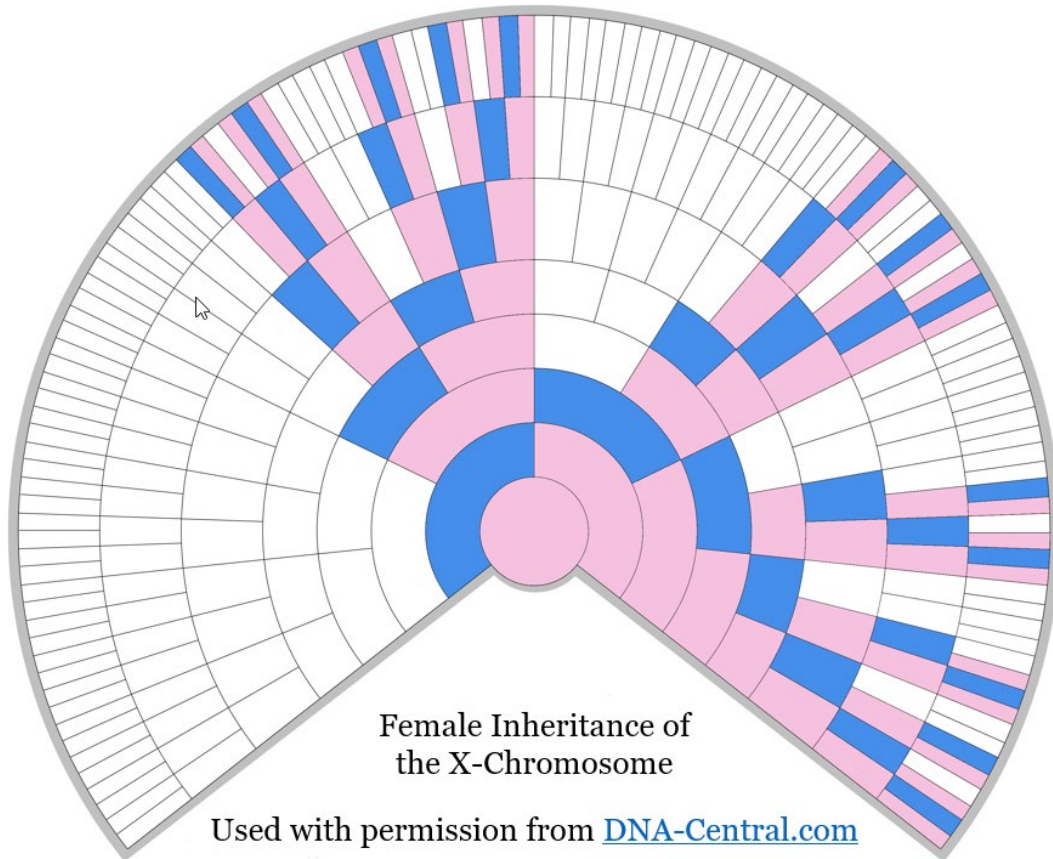
Adapted from Genetic Genealogy in Practice

Mitochondrial (mtDNA) Inheritance



Poster Credit: genebase.com

X-DNA Inheritance



Why is autosomal DNA
important in genetic
genealogy?

Autosomal DNA

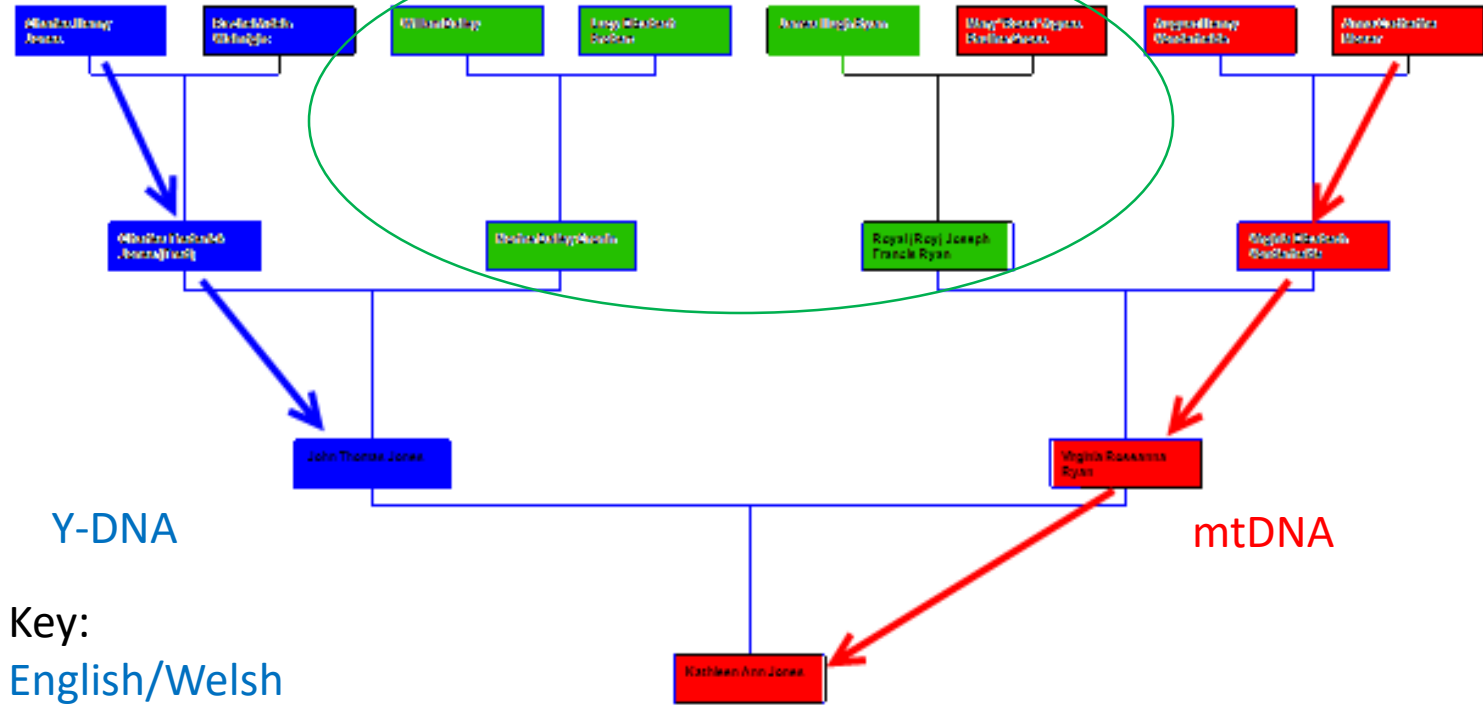
Autosomal DNA is nuclear DNA that includes the 22 pairs of chromosomes that are not involved in the determination of sex. Autosomal DNA is inherited from both parents, who in turn, inherited it from their parents. It can be used to study all lines of a family. It is not passed intact from generation to generation. During meiosis, pairs of chromosomes can exchange sections of DNA through recombination.



Vertical Pedigree Chart for Kathleen Ann Jones

Father to Son

Mother to Daughter



Y-DNA

mtDNA

Key:

English/Welsh

Irish

German

Disadvantage of Autosomal DNA

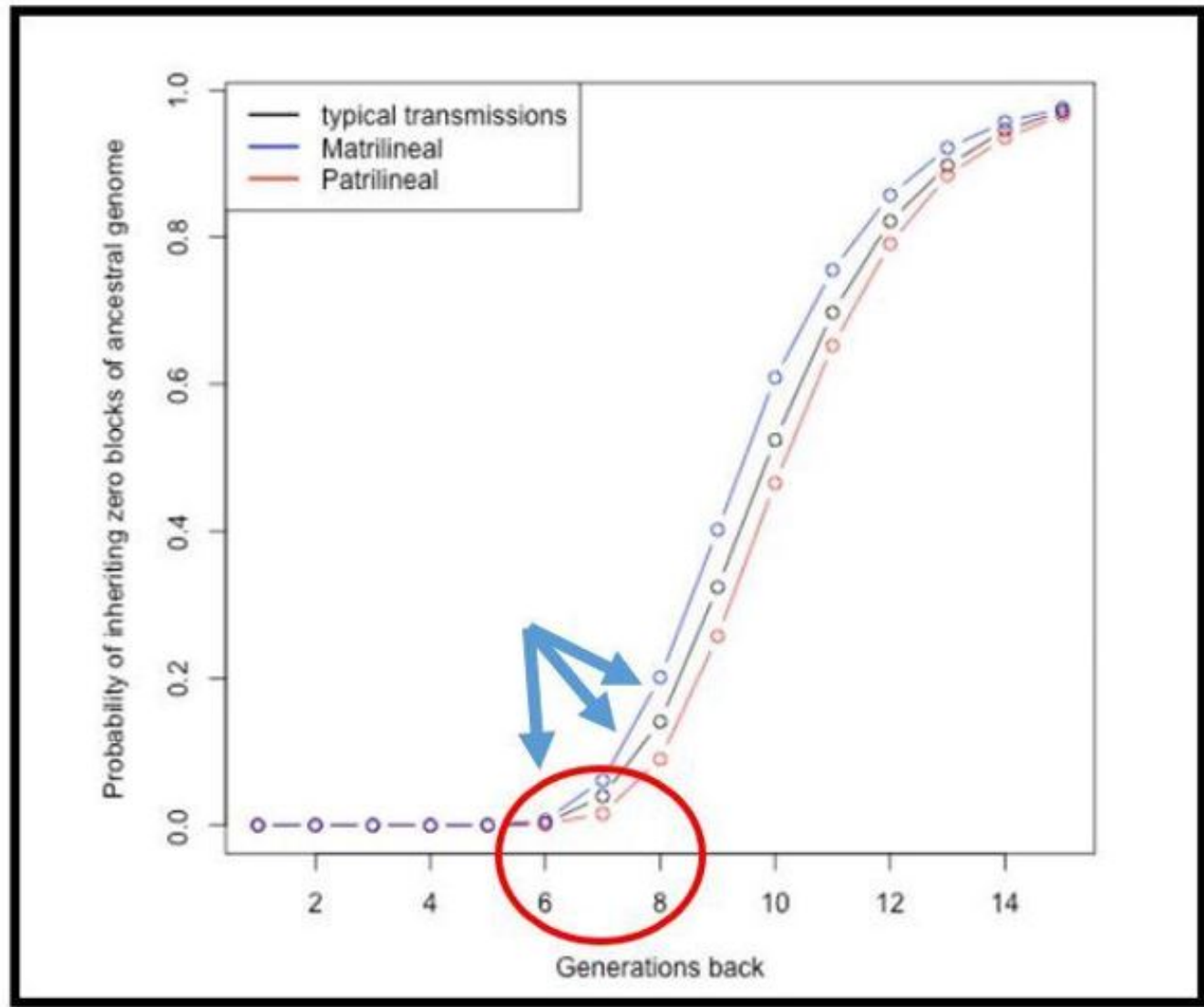


Figure 1: The probability of inheriting zero blocks of DNA from your ancestors, Image Graham Coop, 2013

How do you determine that you are “related” to someone else?



Once your DNA is analyzed, it is compared to everyone else in the database.



Segments of DNA that match someone else’s DNA are identified and measured.



These segment lengths are measured in centimorgans (cM).










The longer the segment, the more likely that you share an ancestor with the match.



Some programs enable you to graphically visualize your matches.

2-d Chromosome with Kit T5

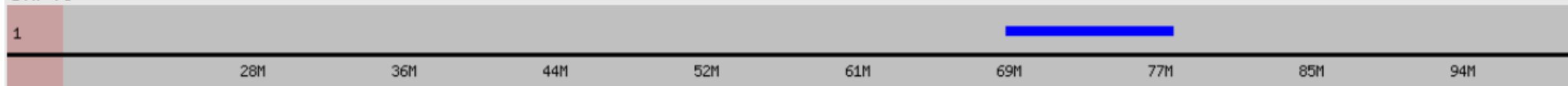
LEGEND

Length	> 100 cM	50-100 cM	20-50 cM	10-20 cM	5-10 cM	< 5 cM	Centromere
Color							

Chr 15

Match ID	Name	Matching segments on Chromosome 15	Overlap with previous match
1	Jo [redacted] (452)	69900146 - 78960529 (8.118 cM)	root

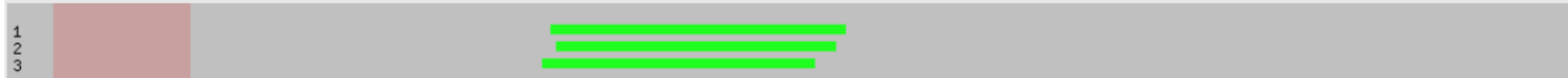
Chr 15

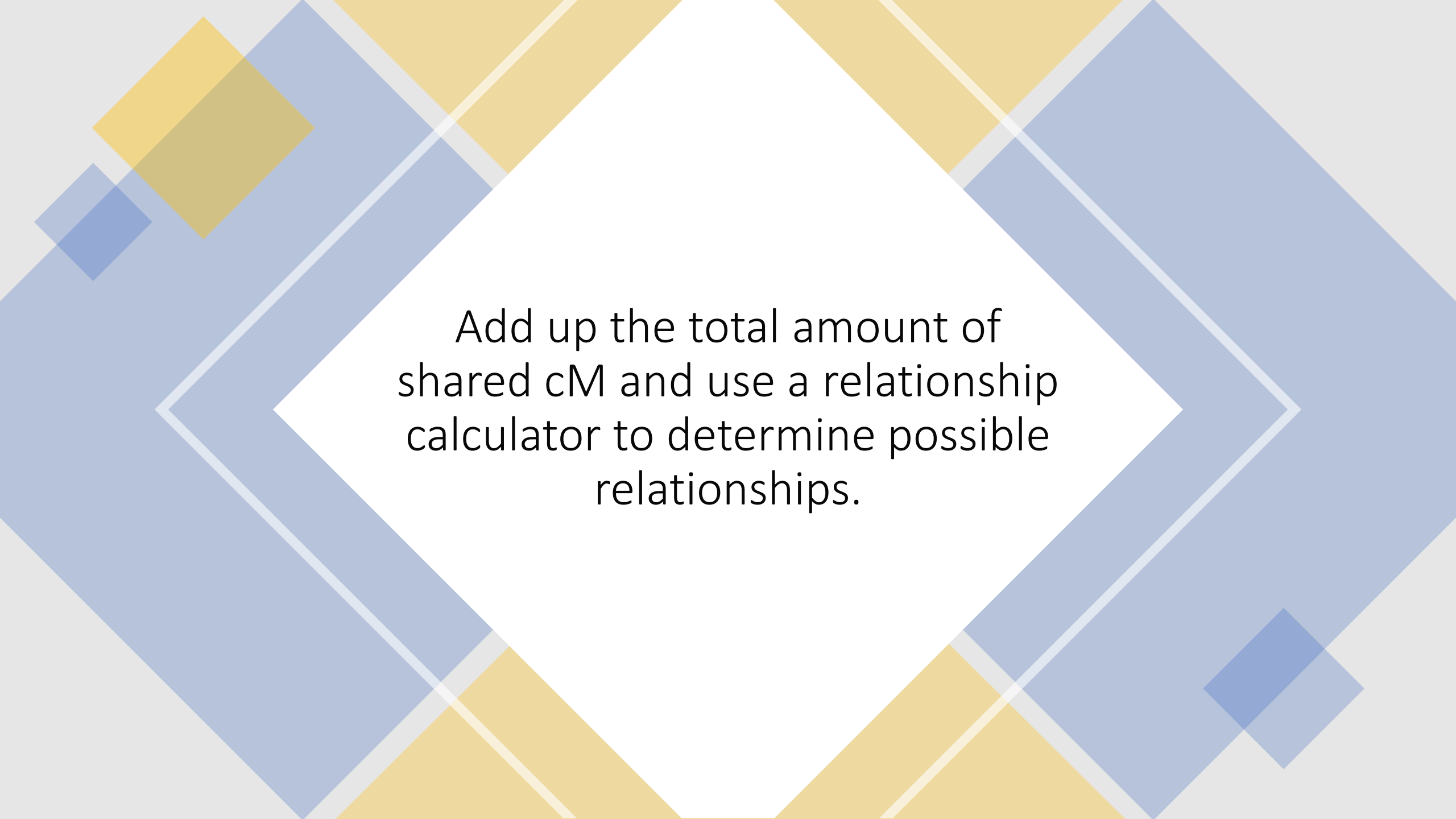


Chr 21

Match ID	Name	Matching segments on Chromosome 21	Overlap with previous match
1	Fra [redacted] (81)	22181904 - 29441100 (12.277 cM)	root
2	J [redacted] (2)	22325822 - 29174612 (11.476 cM)	22325822 - 29174612
3	[redacted] ()	21983892 - 28659242 (11.074 cM)	22325822 - 28659242

Chr 21





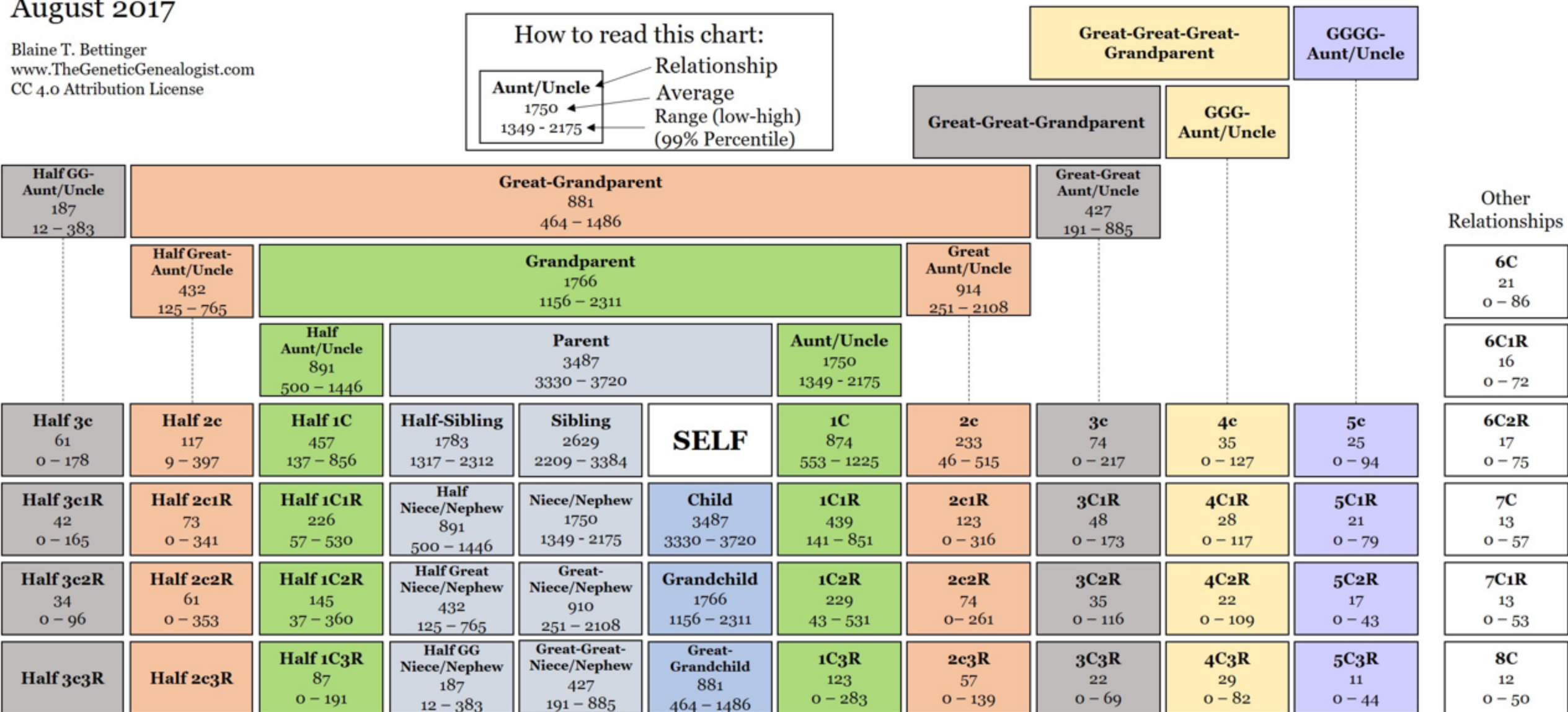
Add up the total amount of shared cM and use a relationship calculator to determine possible relationships.

August 2017

Blaine T. Bettinger
www.TheGeneticGenealogist.com
CC 4.0 Attribution License

How to read this chart:

Aunt/Uncle	Relationship
1750	Average
1349 - 2175	Range (low-high) (99% Percentile)



Minimum was automatically set to 0 cM for relationships more distant than Half 2C, and averages were determined only for submissions in which DNA was shared

Gedmatch summarizes a comparison between two kits with:

- 1) the **largest** segment,
- 2) the **total** segments,
- 3) an estimate of the Most Recent Common Ancestor (MRCA) and
- 4) The number of **shared** segments.

Largest segment = 40.7 cM

Total Half-Match segments (HIR) = 163.1 cM (4.548 Pct)

Estimated number of generations to MRCA = 3.2

7 shared segments found for this comparison.

290598 SNPs used for this comparison.

53.072 Pct SNPs are full identical

Comparison took 0.285 seconds.

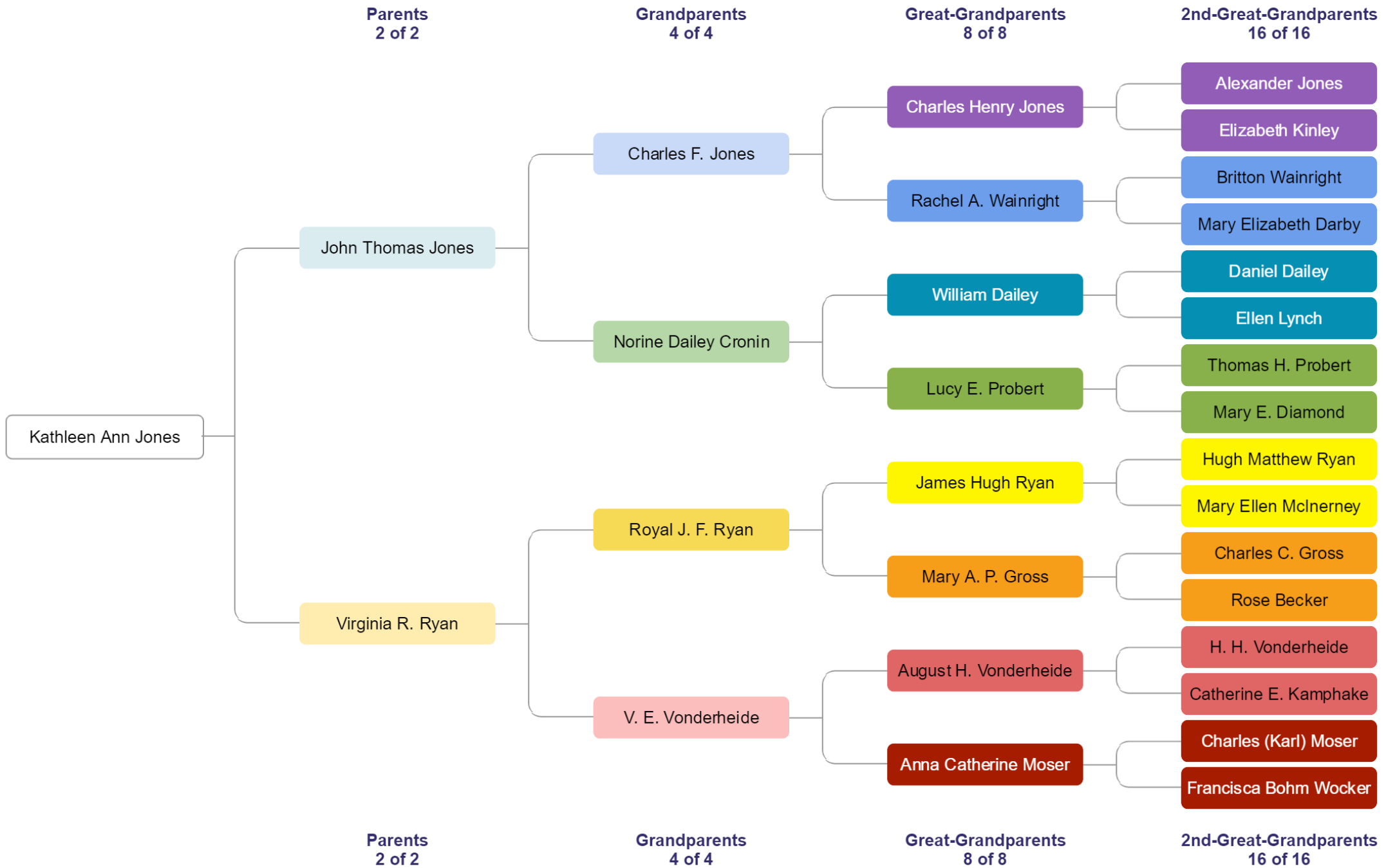
CPU time used: 0.044 cpu seconds.



Genetic vs. Genealogical Tree

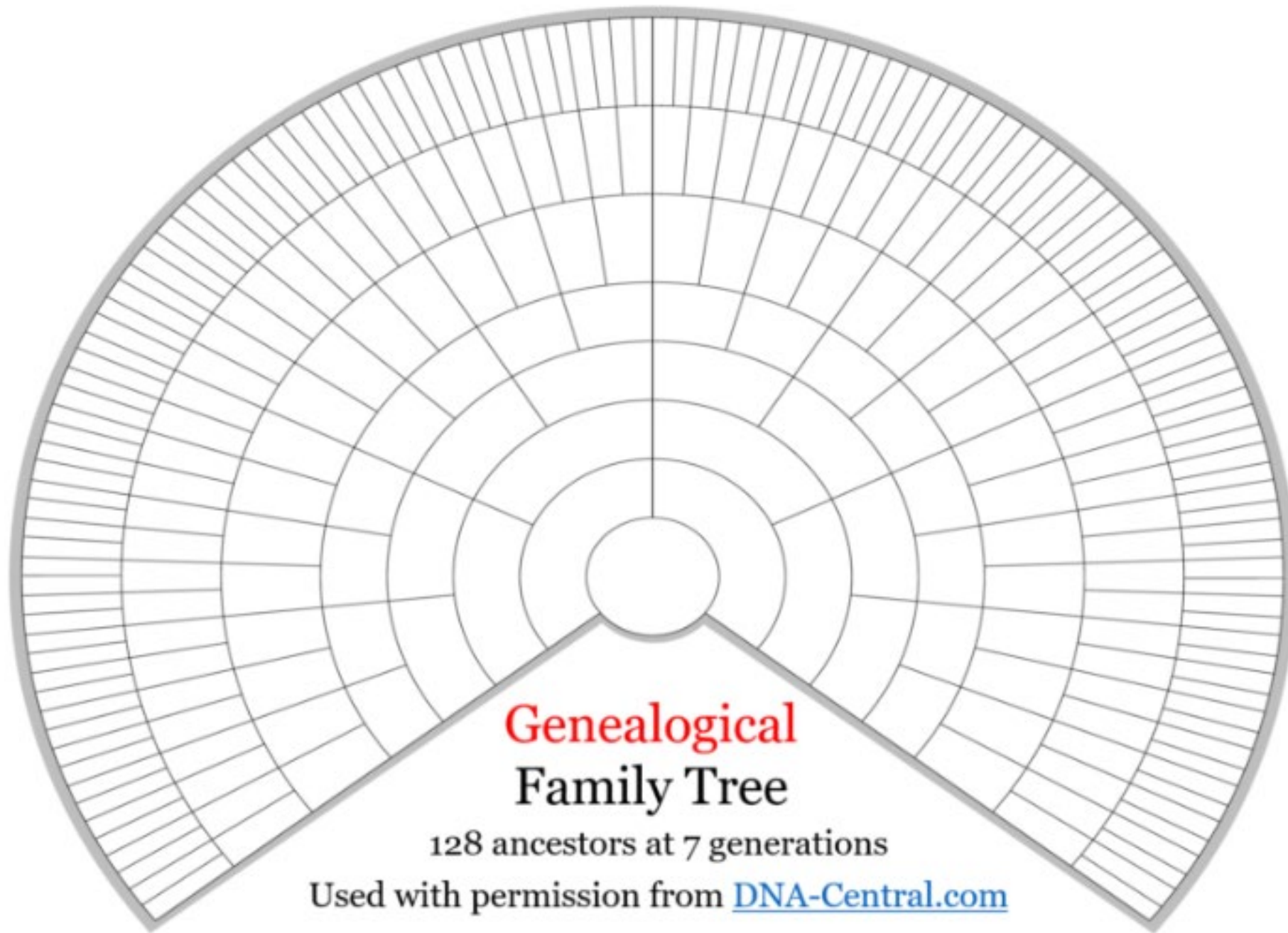
Generation	Number of ancestors in each generation	Average percentage of DNA received from ancestors in each generation
You	1	
Parents	2	50%
Grandparents	4	25%
G-Grandparents	8	12.5%
2X-G-Grandparents	16	6.25%
3X-G-Grandparents	32	3.125%
4X-G-Grandparents	64	1.56%
5X-G-Grandparents	128	0.78%

At some point, people no longer receive DNA from *every* ancestor in each generation. In addition, even siblings who each inherit about 50% of their DNA from each parent inherit *different* DNA. This also explains how siblings can have different ethnicity estimates. Siblings have the same genealogical tree, but different genetic trees.



Each of us has two trees:

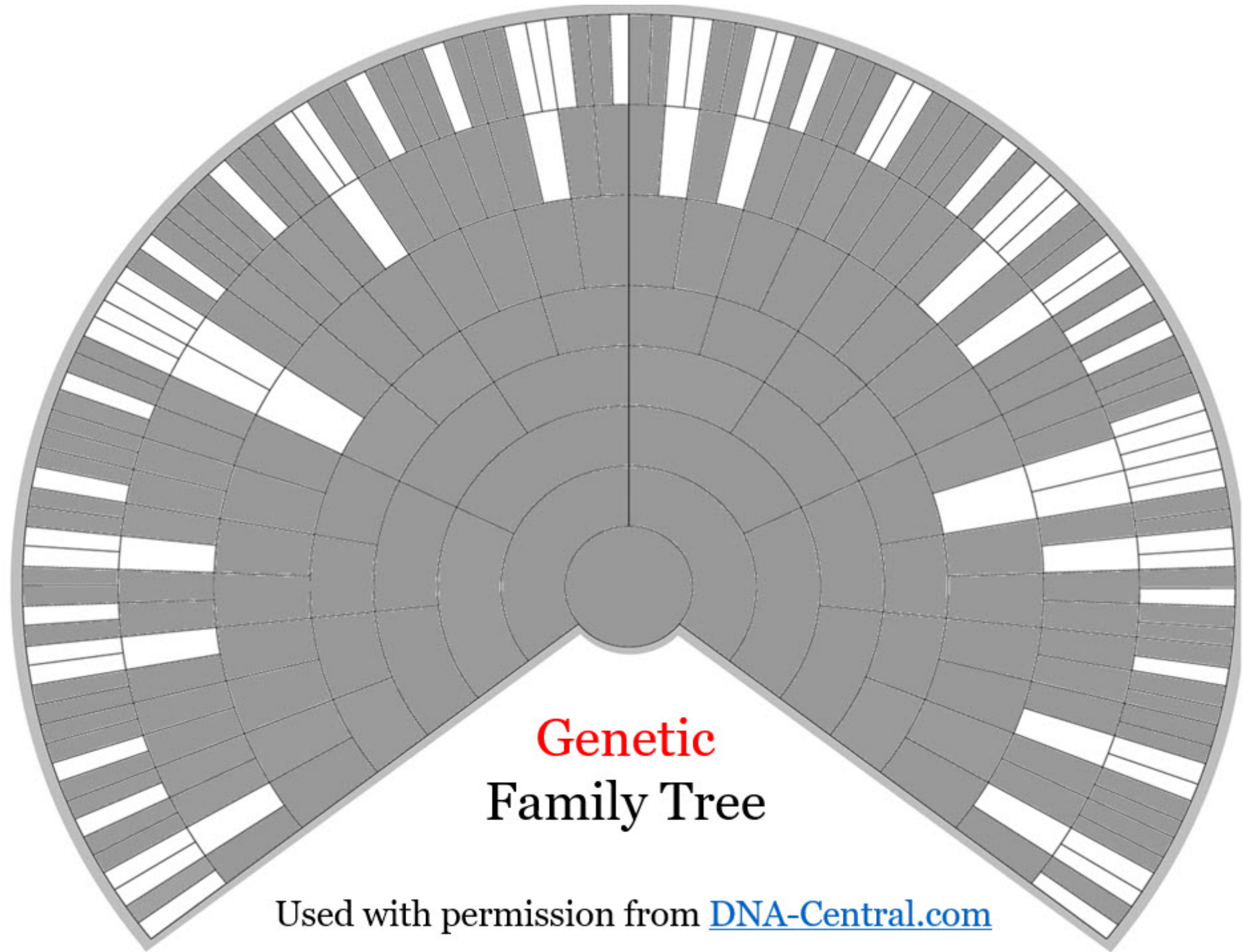
- Genealogical family tree – includes all our ancestors, known and unknown.
- Genetic family tree – only includes the subset of ancestors from whom we inherited DNA.



**Genealogical
Family Tree**

128 ancestors at 7 generations

Used with permission from DNA-Central.com



Genetic
Family Tree

Used with permission from DNA-Central.com

Ethnicity Estimates



Comment on facebook:

“My daughter received 23andMe and Ancestry.com for Christmas, she did both kits at same time and sent them in and they came back with totally different results. I think it’s all a scam.”



So is it?

Ethnicity and Admixture

- In the science of genetic genealogy, ethnicity estimates are the least reliable aspect of the science.
- All three companies can reliably estimate your ethnicity at the continental level (European, Asian, African).
- The results get more inconsistent at the sub-regional level.

How Are Populations Defined?

Each company defines populations according to their own definition.

For instance, if you are Greek, 23andme will categorize your DNA as “Balkan.” Ancestry.com would categorize the same DNA as “Italy/Greece.”

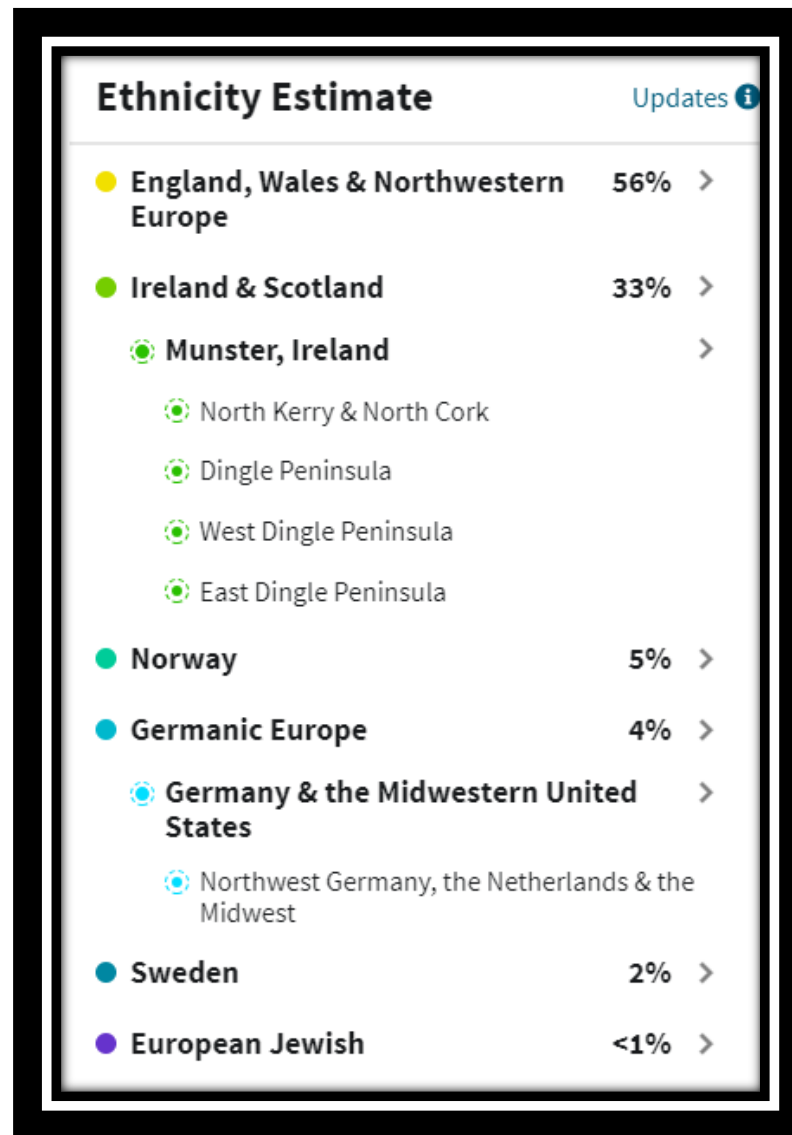
Factors in Inconsistencies

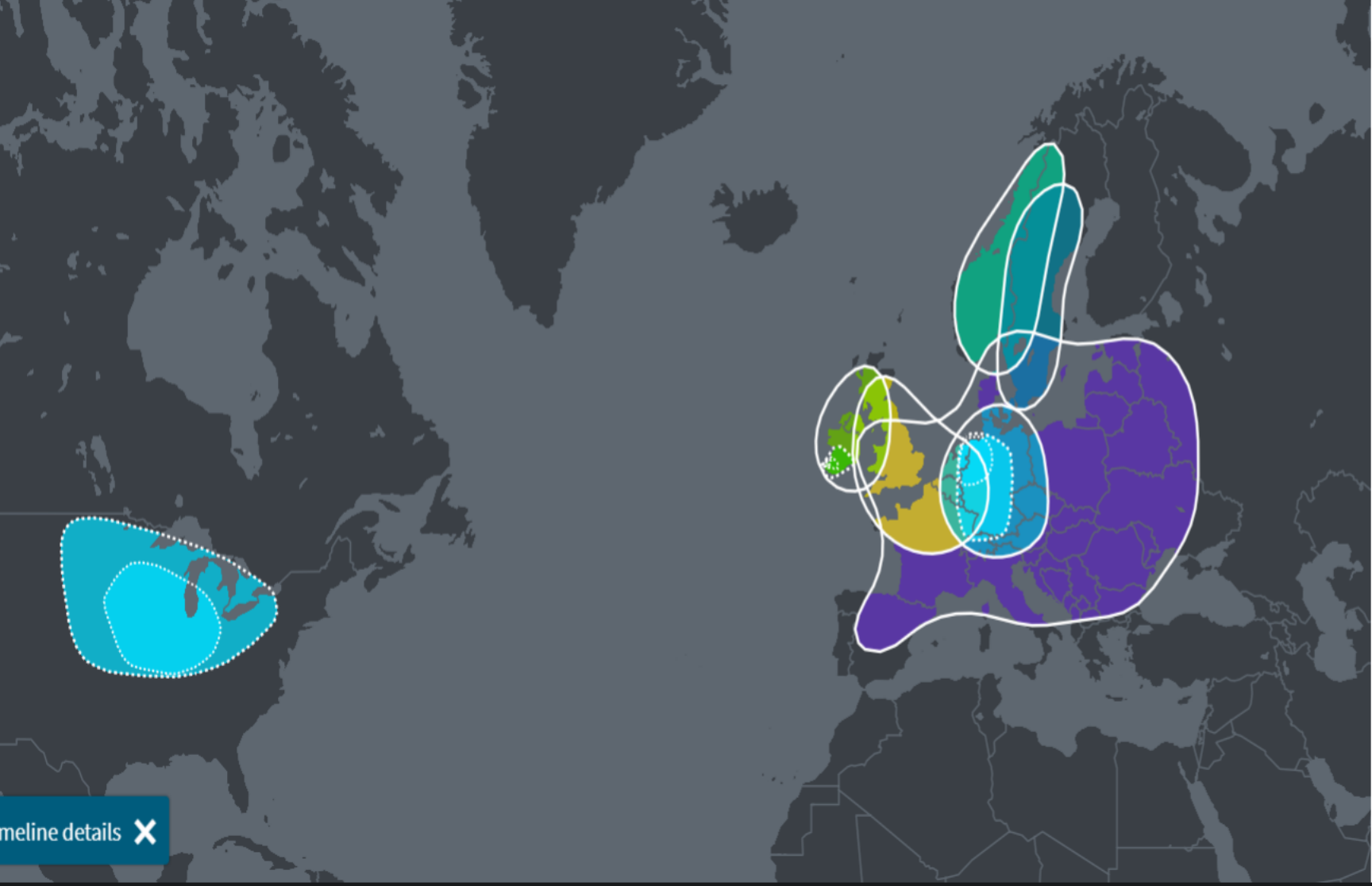
- Each company uses **different** reference samples made up of proprietary data and publicly available data.
- Proprietary data often includes self-reported information from customers who report that all four of their grandparents are from the same country.
- Self-reported results are extensively analyzed and some data are “thrown out” as “outliers” which differ significantly from others in the population.



[Ancestry.com/DNA](https://www.ancestry.com/DNA)

On Ancestry,
click on the
DNA tab and
choose “DNA
Story.”





Ethnicity Estimate

Updates

- **England, Wales & Northwestern Europe** 56% >
- **Ireland & Scotland** 33% >
- **Munster, Ireland** >
 - North Kerry & North Cork
 - Dingle Peninsula
 - West Dingle Peninsula
 - East Dingle Peninsula
- **Norway** 5% >
- **Germanic Europe** 4% >
- **Germany & the Midwestern United States** >
 - Northwest Germany, the Netherlands & the Midwest
- **Sweden** 2% >

Timeline details

- Overview**
- 1700
- 1775
- 1800
- 1825
- 1850
- 1875
- 1900
- 1925

My DNA Story

Solid vs.
Dotted Lines





23andMe

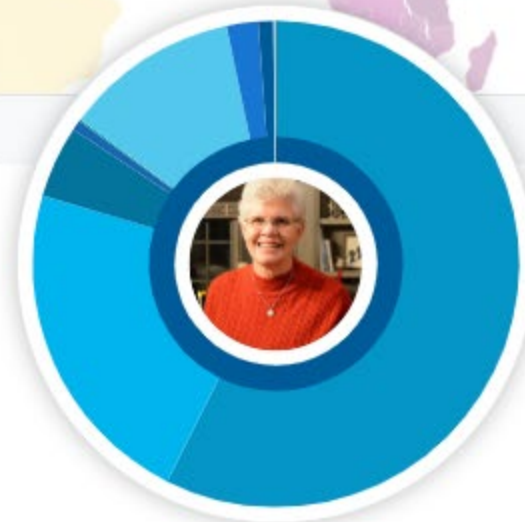
Overview

All Reports

Ancestry Composition

Your DNA tells the story of who you are and how you're connected to populations around the world.

[View your ancestry composition](#)



YOUR ANCESTRY IN DETAIL



NEW

57.4% British & Irish

[View report](#)



NEW

22.1% French & German

[View report](#)



NEW

4.8% Scandinavian

[View report](#)

In the last 200 years, your ancestors may have lived in the following locations.

We found evidence of your recent ancestry in the following regions. Darker regions represent places where you have DNA in common with more people who report ancestry from that particular region. Because these results reflect the ancestries of individuals currently in our reference database, expect to see your results change over time as that database grows. [Read more in Scientific Details.](#)

British & Irish

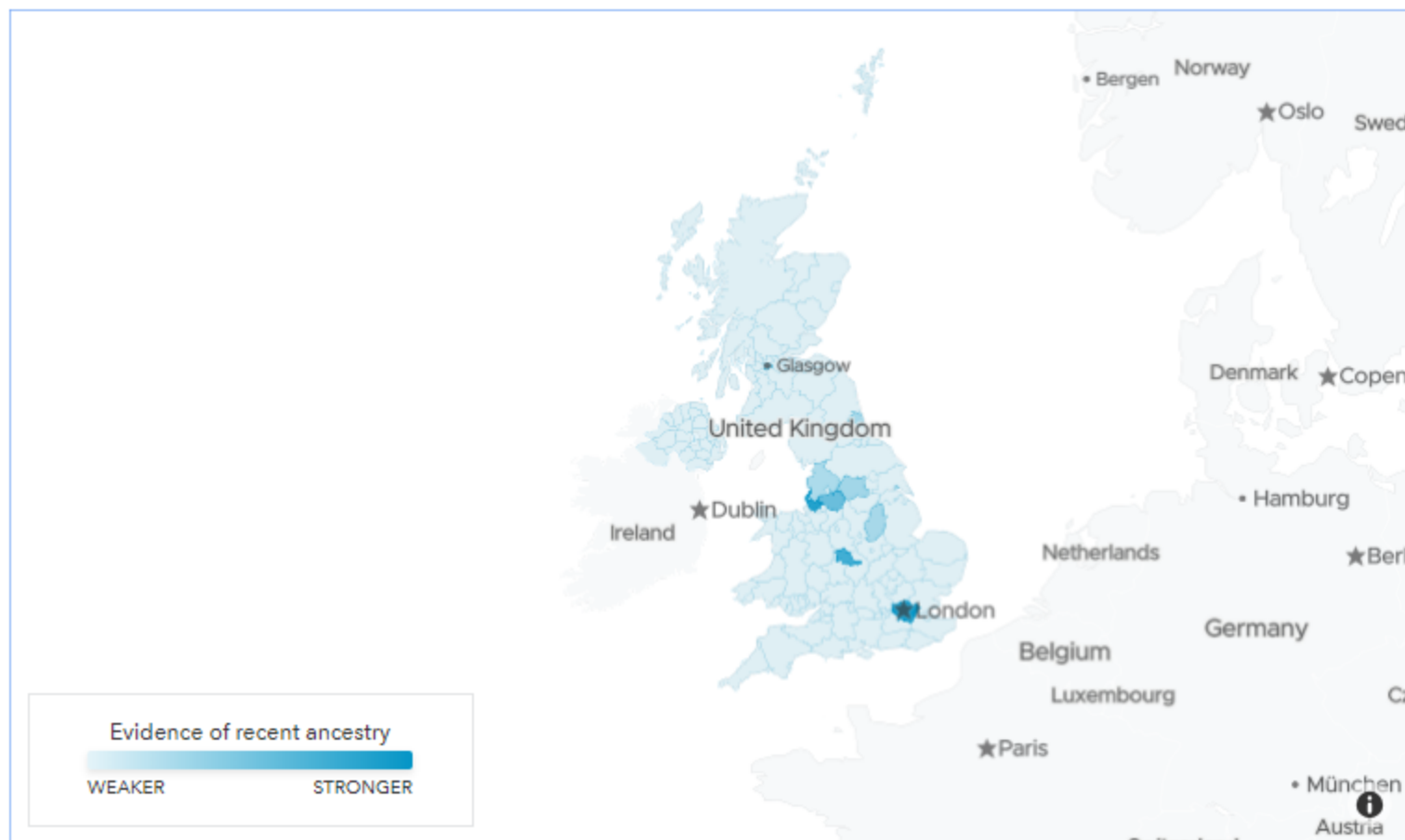
57.4%

United Kingdom

Highly Likely Match ^

United Kingdom has 165 administrative regions, and we found the strongest evidence of your ancestry in the following 10 regions.

1. Greater London
2. Merseyside
3. West Midlands
4. Greater Manchester
5. Glasgow City
6. West Yorkshire
7. Nottinghamshire
8. Tyne and Wear
9. Hull
10. Lancashire



British & Irish

57.4%

United Kingdom

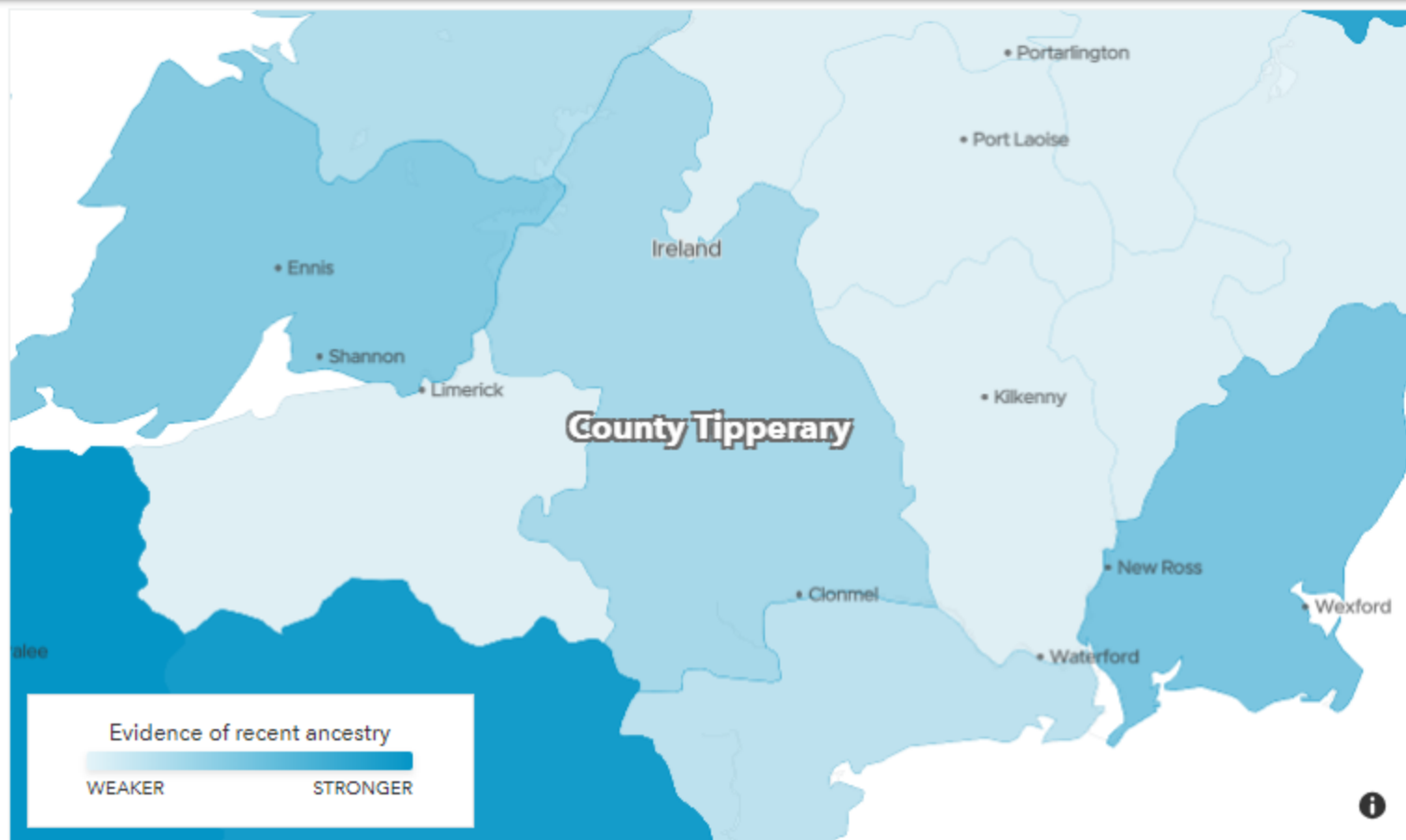
Highly Likely Match ▾

Ireland

Likely Match ▴

Ireland has 26 administrative regions, and we found the strongest evidence of your ancestry in the following 10 regions.

1. County Kerry
2. County Cork
3. County Dublin
4. County Wexford
5. County Clare
- 6. County Tipperary**
7. County Mayo
8. County Galway
9. County Cavan
10. County Waterford



Speculative vs Conservative Estimates

Speculative Estimate		Conservative Estimate	
European	99.8%	European	98.1%
British and Irish	57.4%	British and Irish	14.6%
French and German	22.1%	French and German	2.1%
Scandinavian	4.8%	Scandinavian	N/A
		Broadly Northwestern European	58.4%
		Broadly European	22.8%

General Observations:

1. The more specific you try to get, the more speculative the guess.
2. greater specificity = greater speculation



FamilyTree**DNA**

FTDNA Reference Populations N=24

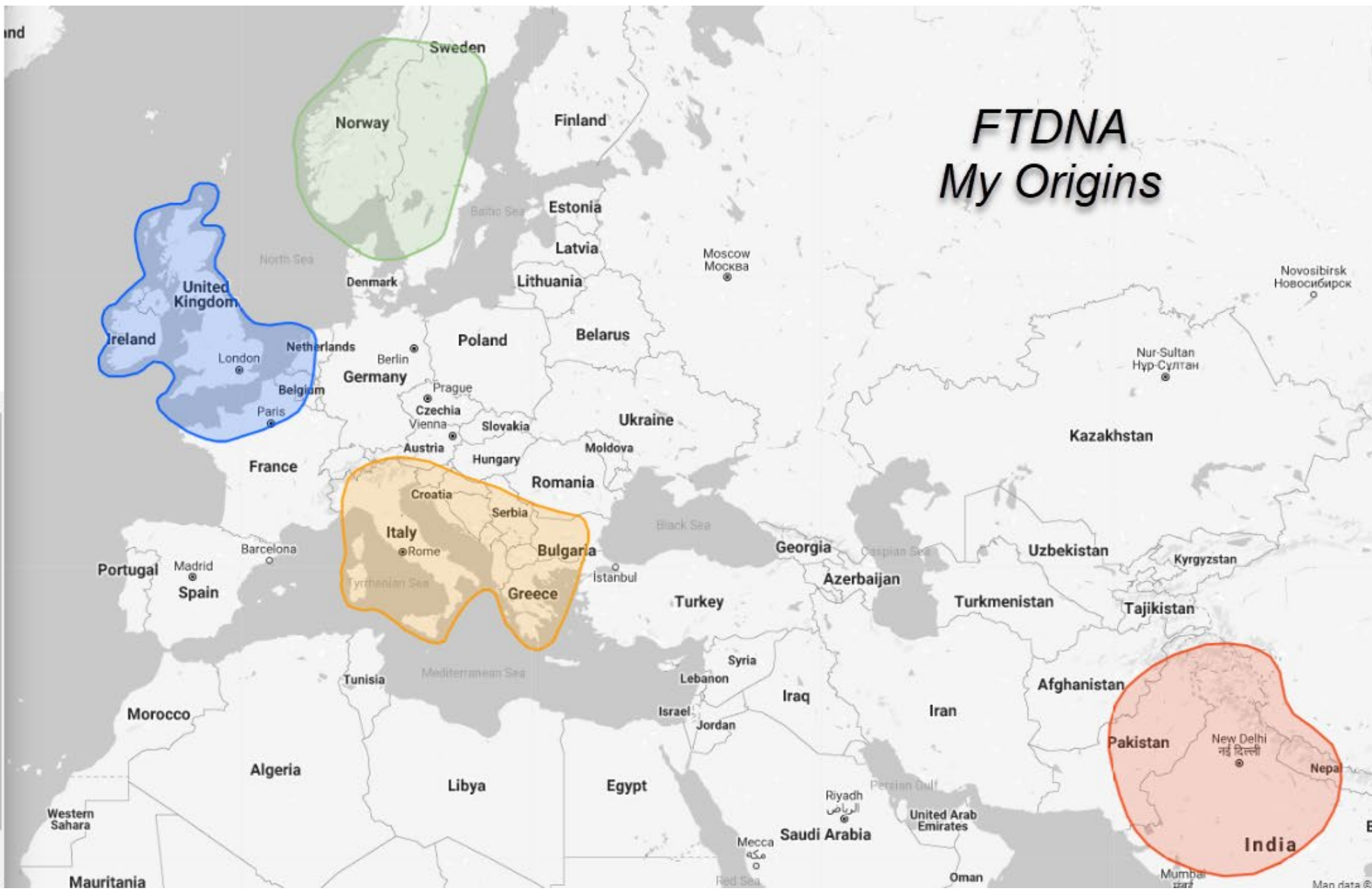
- + Sephardic
- + Ashkenazi
- + North and Central America
- + South America
- + British Isles
- + Scandinavia
- + Finland
- + West and Central Europe
- + Southeast Europe
- + East Europe
- + Iberia
- + West Middle East

- + East Middle East
- + Asia Minor
- + North Africa
- + East Central Africa
- + South Central Africa
- + West Africa
- + Central Asia
- + South Central Asia
- + Siberia
- + Northeast Asia
- + Southeast Asia
- + Oceania

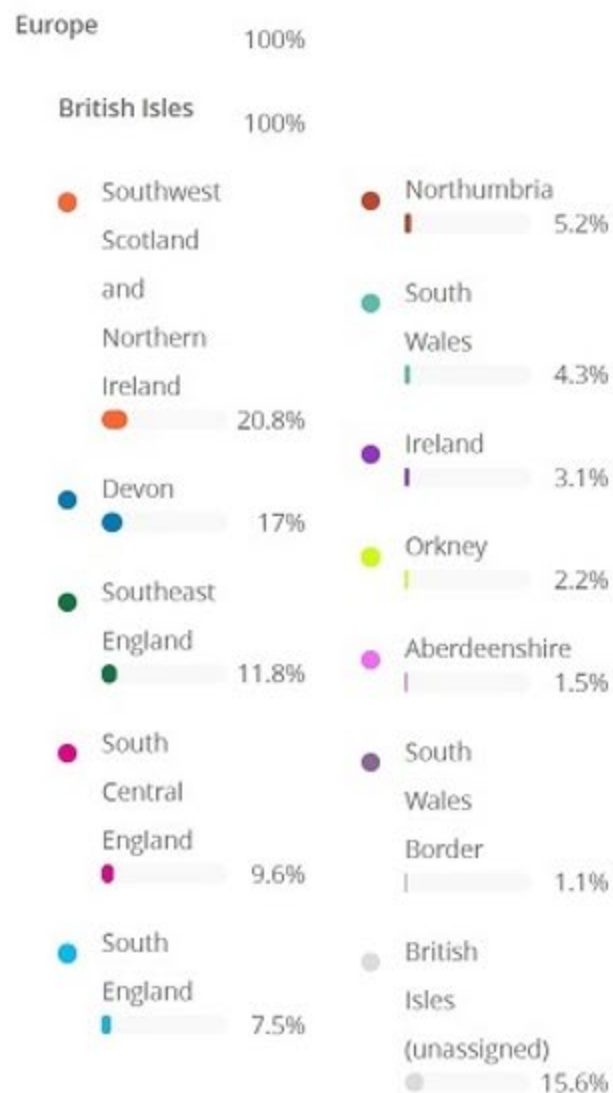
Origins

Uncover the unique genetic assemblage that has been passed down to you from your ancestors, and see which of our 24 reference populations from around the world your autosomal DNA connects to. Our clusters will highlight major historical and genetic events, thus shedding light on the wild complexity of your genetic tapestry. Though we are all unique and distinct, we are also woven from the same fundamental elements. The weight you give to each thread is up to you.

myOrigins	Shared Origins
Kathleen Reed	100%
European	96%
● British Isles	77%
● Scandinavia	14%
● Southeast Europe	5%
Central/South Asian	3%
● Central Asia	3%
Trace Results	
● Sephardic	<2%



If you have UK roots, you may want to try LivingDNA.



Europe 100%

● Great Britain and Ireland 83.6%

● South England 23.1%

● Ireland 18.9%

● Southeast England 13.8%

● East Anglia 6.9%

● Northern Ireland and Southwest Scotland 6.2%

● South Central England 4.6%

● North Wales 3.3%

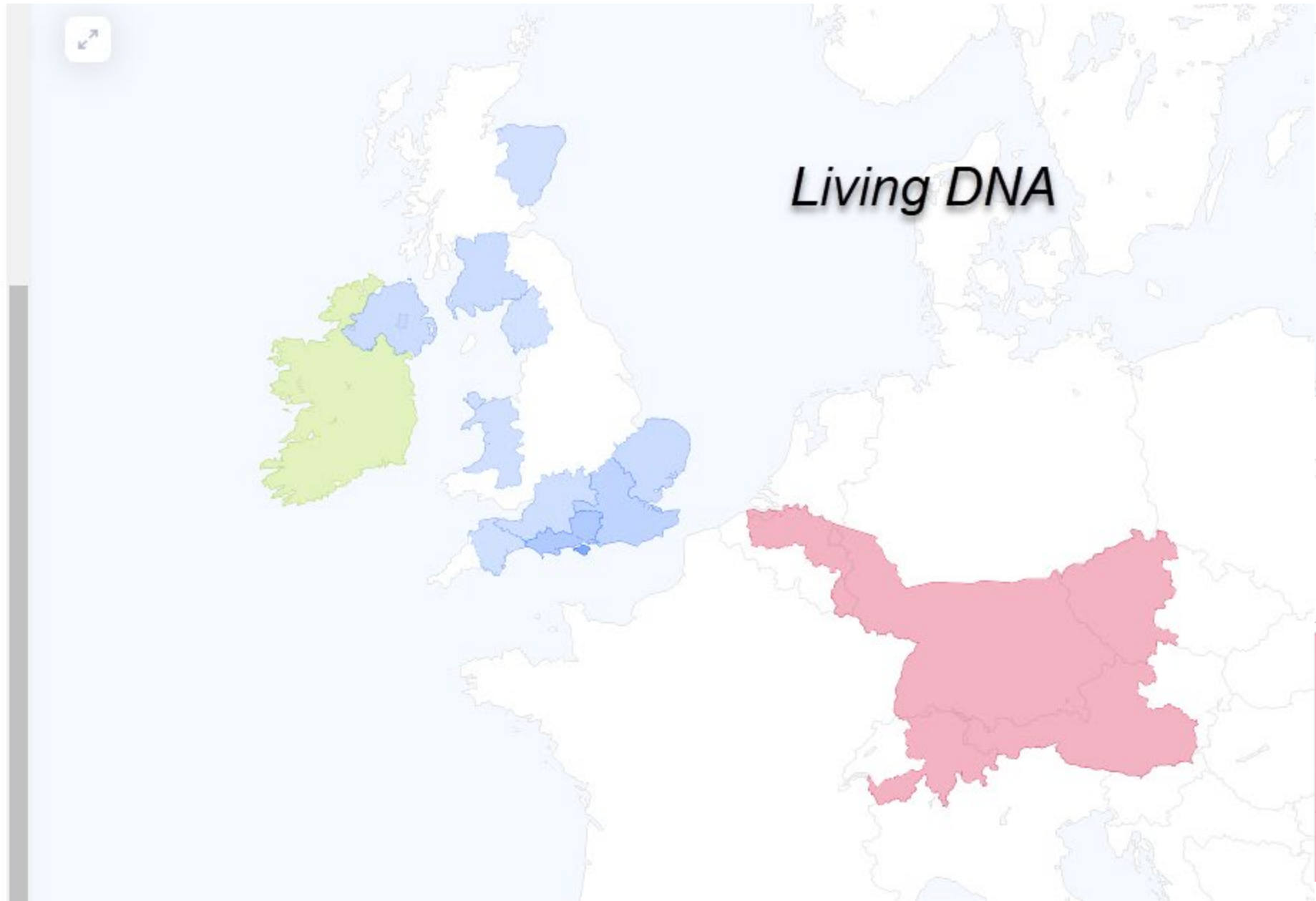
● Devon 3%

● Aberdeenshire 2.2%

● Cumbria 1.4%

● Europe (North and West) 16.4%

● South Germanic 16.4%



Kathleen Reed, you are...

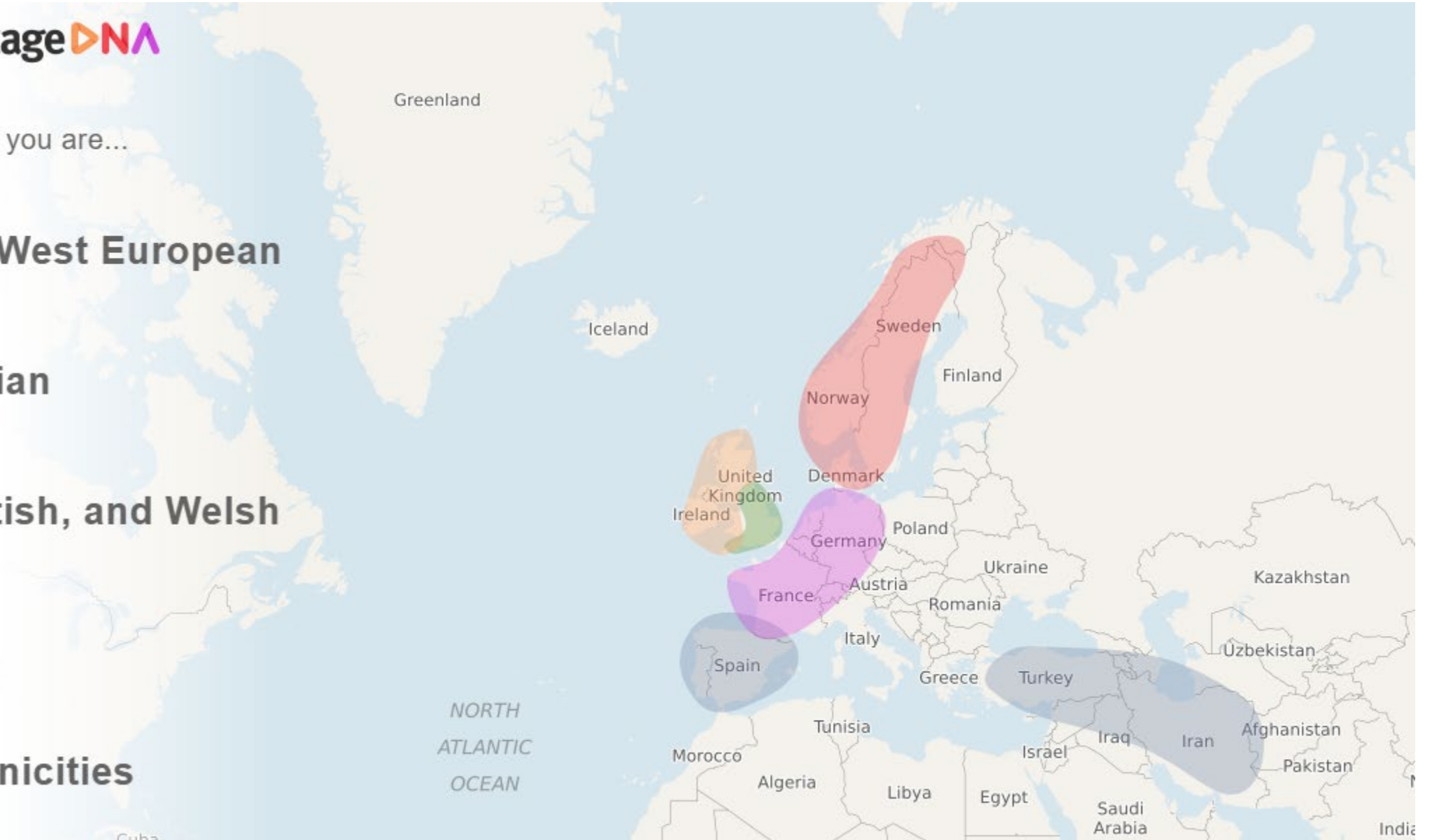
35.9 %
North and West European

28.5 %
Scandinavian

13.9 %
Irish, Scottish, and Welsh

13.1 %
English

8.6 %
2 more ethnicities



All ethnicities

All supported ethnicities

▶ Play Intro

Print

Share

▼ Europe 98.7%

● North and West Europe 91.4%

North and West European 35.9%

Scandinavian 28.5%

Irish, Scottish, and Welsh 13.9%

English 13.1%

Finnish 0.0%

● South Europe 7.3%

Iberian 7.3%

Greek 0.0%

Italian 0.0%

Sardinian 0.0%

● Ashkenazi Jewish 0.0%

Ashkenazi Jewish 0.0%

● East Europe 0.0%

Balkan 0.0%

Baltic 0.0%

East European 0.0%

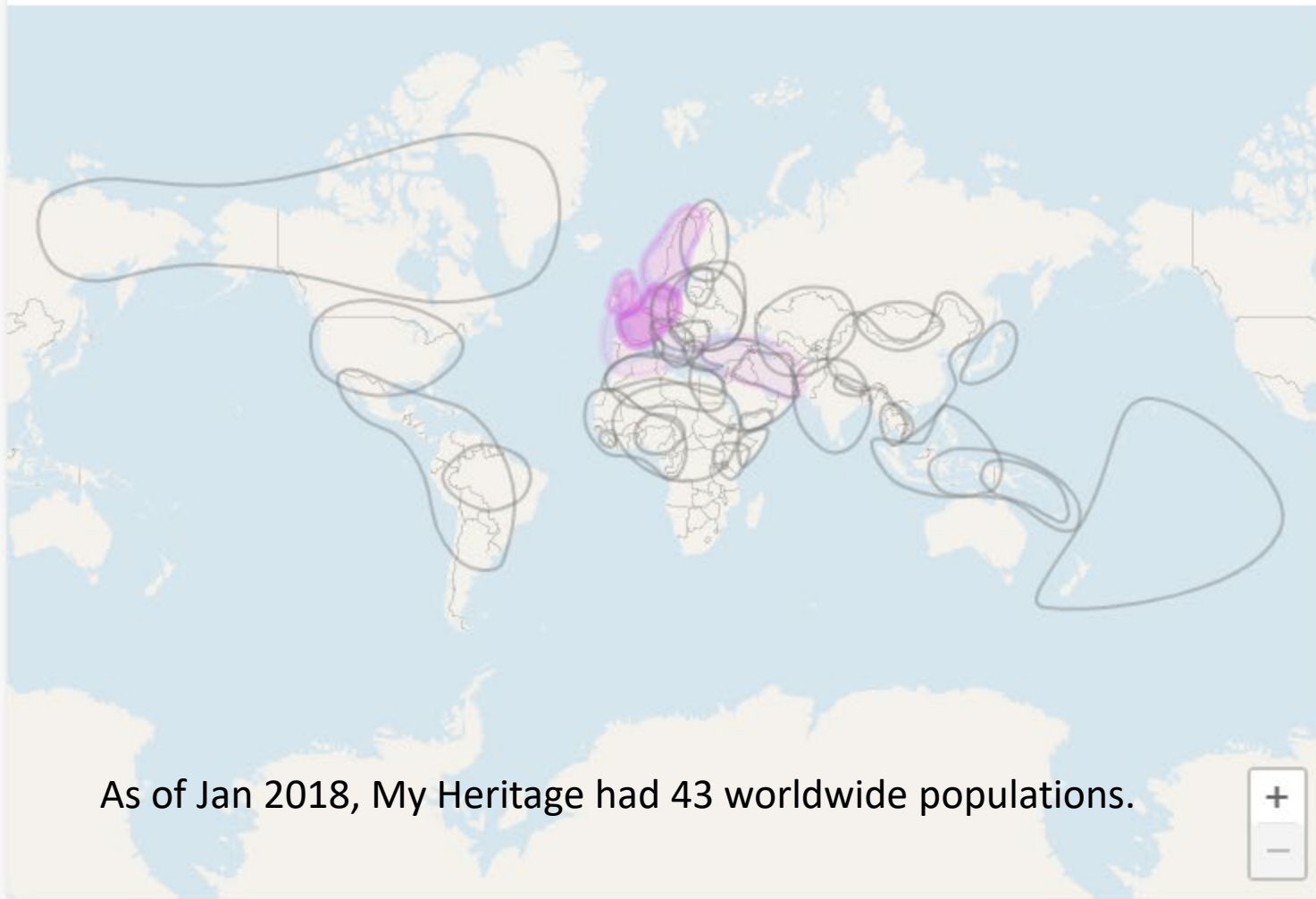
▼ Asia 1.3%

● West Asia 1.3%

West Asian 1.3%


Mizrahi Jewish - Iranian/Iraqi 0.0%

Show events from your family tree ?




As of Jan 2018, My Heritage had 43 worldwide populations.





So we've determined that ethnicity estimates will differ from company to company. Some of the factors to consider are:

- The reference populations used by each company.
 - The number of samples collected from each region.
 - The computer algorithm developed by the company to assign customer samples to a region.
- 

Third Party Tools

Gedmatch.com

User Profile (3007):

Name: Kathleen Reed

Email: khreed@cinci.rr.com

Tier 1 Member

Tier 1 Paid Until 2020-02-14

[View/Change/Delete your profile \(password, email, groups\)](#)

[Change EU/Unknown kit status](#)

The number of online users is 129

LEGEND:

(Status indicators shown to the right of each kit below)

✓	Kit has completed all processing and has good status
✎	Click on pencil if you wish to EDIT or DELETE kit profile
?	Unknown Status
Click on blue kit number to go directly to one-to-many results	

Your DNA resources:

T72	y	✓	Jar
T18	y	✓	Eve
M7	y	✓	Wil
M0		✓	Do
M4		✓	J. T
T10		✓	Jea



Information:

- [User Lookup](#) - Find information on your matches.
- [How to use GEDmatch](#)
- [GEDmatch Terms of Service](#)
- [GEDmatch info about you](#)
- [GEDmatch Wiki](#)
- [Useful Videos](#)
- [Support Request](#)

Upload your DNA files:

- [Generic Uploads \(23andme, FTDNA, AncestryDNA, most others\)](#)

DNA Applications:

FREE

- [One-To-Many Beta - give it a try](#)
- [One-To-Many DNA Comparison Result](#)
- [One-to-One Autosomal DNA Comparison](#)
- [One-to-One X-DNA Comparison](#)
- [Admixture \(heritage\)](#)
- [Admixture / Oracle with Population Search](#)
- [People who match both, or 1 of 2 kits](#)
- [DNA File Diagnostic Utility](#)
Analyze DNA file upload for potential problems.
- [Are your parents related?](#)
- [3-D Chromosome Browser](#)
- [Archaic DNA Matches](#)
- [Ancestor Projects NEW](#)

GEDmatch Forums

- [Gedmatch Forums - Starting over!](#)

DNA Painter


DNA PAINTER

Dashboard

Tools

Help

Blog

 Kathleen Reed

WELCOME KATHLEEN ★ THANKS · YOUR SUBSCRIPTION IS ACTIVE UNTIL DECEMBER 29TH 2020 · [VIEW ACCOUNT](#)



ANCESTRAL
TREES



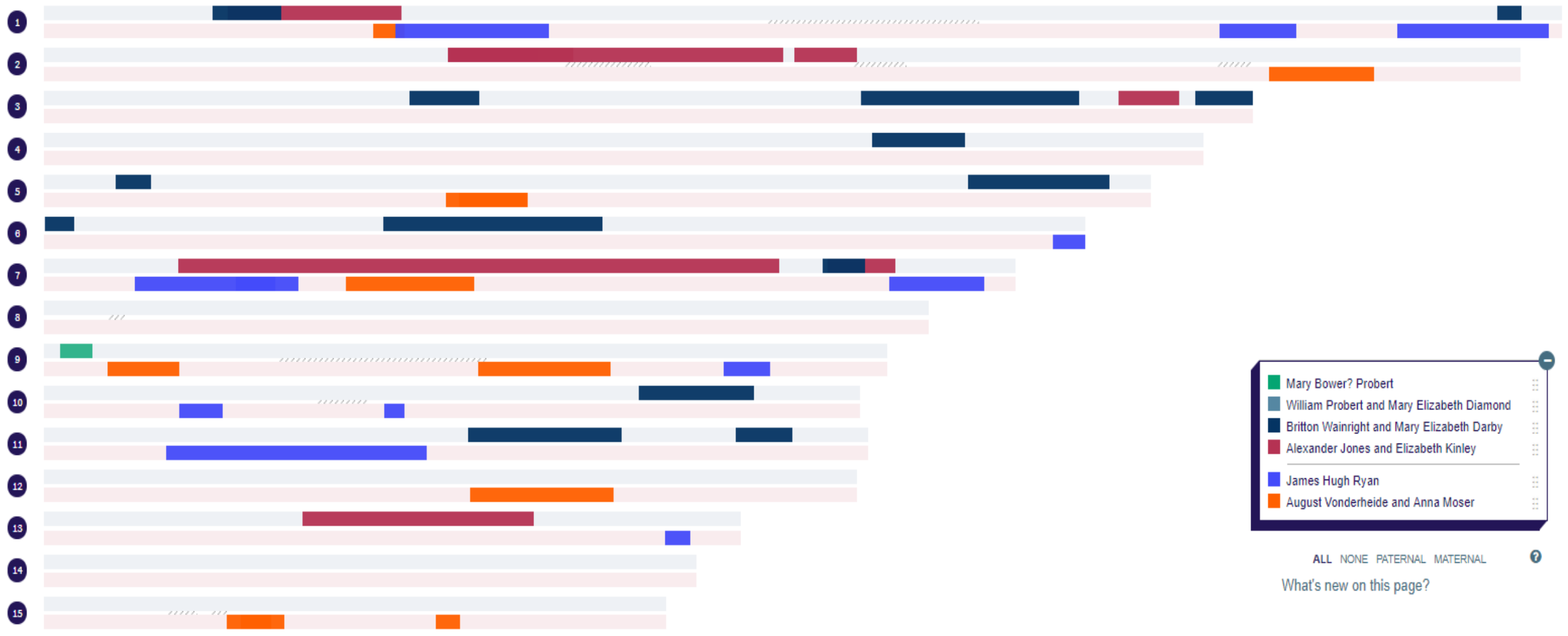
TOOLS AND
WATO



CHROMOSOME
MAPS

Kathleen Reed is the gg-granddaughter of Alexander and Elizabeth Kinley, g-granddaughter of Charles Jones and Rachel Wainright, granddaughter of Charles "Fred" Jones and Rachel Wainright, and daughter of John Thomas Jones and Virginia Ryan.

View segments in a particular group by clicking the group name in the key.

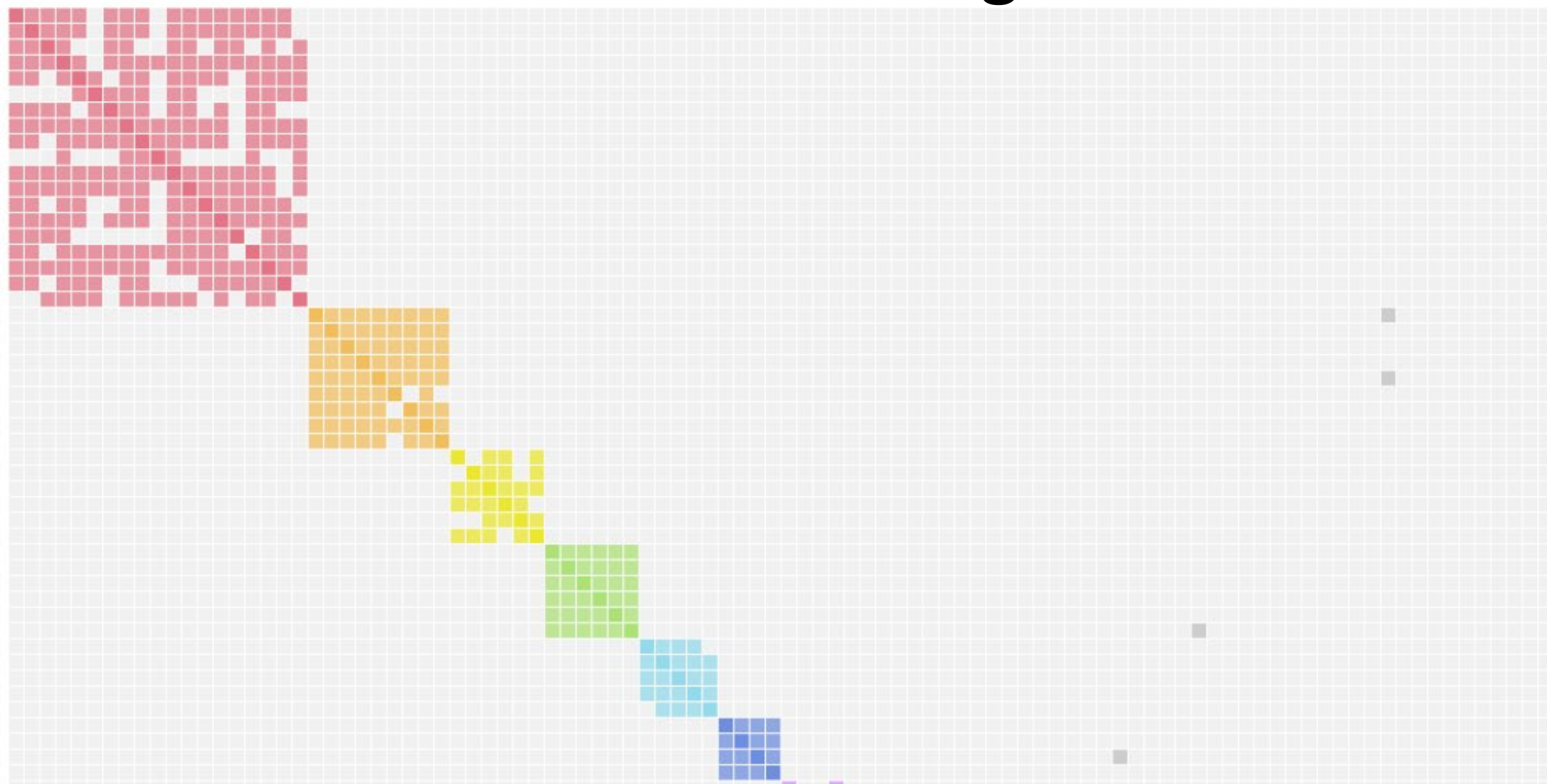


- Mary Bower? Probert
- William Probert and Mary Elizabeth Diamond
- Britton Wainright and Mary Elizabeth Darby
- Alexander Jones and Elizabeth Kinley
- James Hugh Ryan
- August Vonderheide and Anna Moser

ALL NONE PATERNAL MATERNAL

What's new on this page?

Clustering Tool



- Cluster 1: 19 members
- Cluster 2: 9 members
- Cluster 3: 6 members
- Cluster 4: 6 members
- Cluster 5: 5 members
- Cluster 6: 4 members
- Cluster 7: 4 members
- Cluster 8: 4 members
- Cluster 9: 4 members
- Cluster 10: 4 members
- Cluster 11: 4 members
- Cluster 12: 4 members
- Cluster 13: 4 members
- Cluster 14: 3 members
- Cluster 15: 3 members
- Cluster 16: 3 members
- Cluster 17: 3 members
- Cluster 18: 3 members
- Cluster 19: 3 members
- Cluster 20: 3 members



Announcements

What's New at DNA Central
Today?

WHAT'S NEW?



Course Library

Courses to further your DNA
knowledge!

ACCESS NOW



DNA Newsletter

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The best resources for genetic
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CHECK IT OUT



ANC101 - I've Tested at AncestryDNA, Now What?

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MH101 - I've Tested at MyHeritage, Now What?

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23Me101 - I've Tested at 23andMe, Now What?

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Y-DNA Testing 101

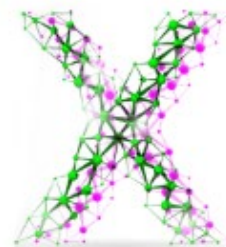
YDNA101 - I've Tested My Y-DNA, Now What?

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Mitochondrial DNA

mtDNA101 - I've Tested my mtDNA, Now What?



X-DNA Testing 101

XDNA101 - Using the X Chromosome



Third-Party Tools

TOOLS101 - Essential Third-Party Tools

Company Comparisons

Company	23andMe	Family Tree DNA's Family Finder test	Ancestry.com's AncestryDNA test	National Geographic Genographic Project Geno 2.0 Next Generation test	MyHeritage
<p>Overall accuracy and sophistication of the biogeographical ancestry analysis rated on a 1-10 scale with 10 being excellent and 1 being poor</p>	<p>Overall rating: 7 The Ancestry Composition feature offers a map view which displays one's ancestral components from various regions of the world as of 500 years ago, a split view for those who also have one or both parents who have been tested by 23andMe, and a breakdown by chromosome. Three settings are available: conservative, standard, and speculative. Overall accuracy is reasonably good, but predictions in Europe are still not optimal, particularly in the speculative mode. Ancestry Finder provides a breakdown of one's ancestry by country.</p>	<p>Overall rating: 3.5 The myOrigins analysis is much improved over FTDNA's previous Population Finder analysis. However, it still lacks specificity, particularly for Europe. No chromosome painting feature is available.</p>	<p>Overall rating: 5 The Genetic Ethnicity Summary consistently overestimates the Central European and Scandinavian ancestral components for people whose ancestors were from the British Isles. The ancestral component from the British Isles is overestimated for people whose ancestors were from continental Europe. The Genetic Community feature indicates which populations or ethnic groups that have existed within the past 150 to 400 years have contributed at least 4% of your autosomal DNA.</p>	<p>Overall rating: 2.5 <i>(Note: This review relates to the former Geno 2.0 test. Results are not yet available for Geno 2.0 Next Generation.)</i> Since a relatively limited number of autosomal SNPs are available for analysis, the biogeographical ancestry analysis is somewhat limited relative to other similar tools, particularly relative to Ancestry Composition. The two closest reference populations are given for each person who is tested. However, these predictions, particularly the second closest reference population, are frequently inaccurate.</p>	<p>Overall rating: 4 The Ethnicity Estimate generally overestimates the English ancestral component for people whose ancestors were from continental Europe.</p>

https://isogg.org/wiki/Autosomal_DNA_testing_comparison_chart



Any

Questions

