A DNA Genealogy Primer

How to break genealogical brick walls with genetic science

Joseph T. Richardson <<u>joseph.t.richardson@gmail.com</u>> 7 Jan 2024 Presented for the Morgan County (Alabama) Genealogy Society

Outline

- DNA Basics
- Types of DNA Tests
 - Y-DNA
 - mtDNA
 - Autosomal DNA

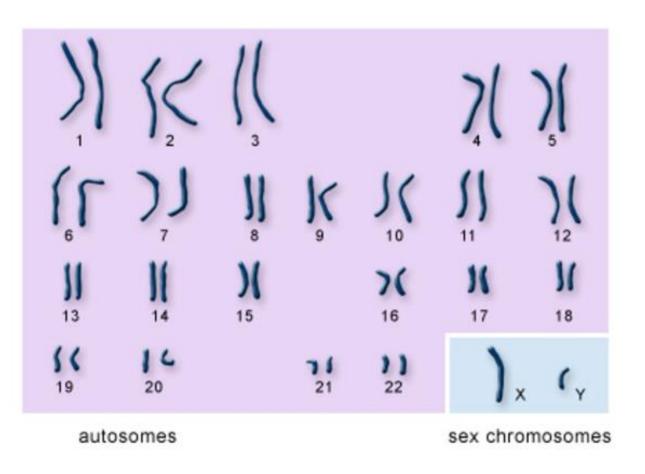


DNA Basics

- DNA (Deoxy-riboNucleic Acid): complex organic molecule that encodes sequences of proteins
- All living things have DNA
- Bundled into *chromosomes*
- Humans generally have 23 pairs of chromosomes (46 in all)

Sex chromosomes and Autosomes

- One pair out of the 23 pair are called *sex chromosomes*
 - Sex chromosomes define male and female characteristics
 - Males: X from mother, Y from father (XY)
 - Females: X from mother, X from father (XX)
- Remaining 22 pair: *autosomes*



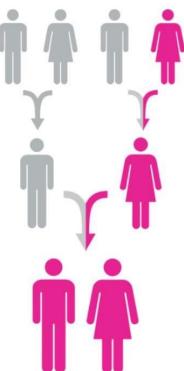
Types of DNA tests

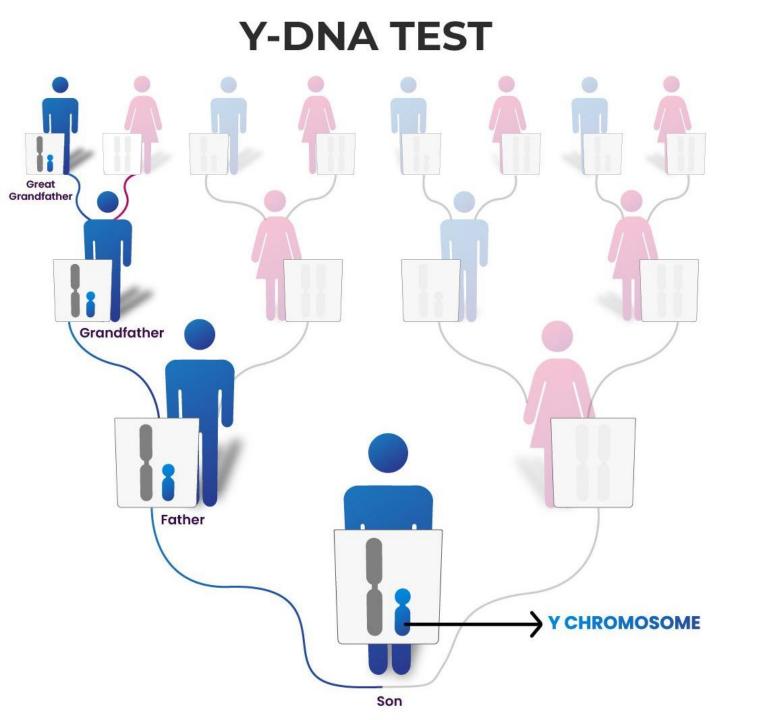
- Y-DNA
- Autosomal DNA
- Mitochondrial DNA (mtDNA)

Y-Chromosone DNA is inherited from your male lineage

Autosomal DNA is inherited from all of your ancestors.

Mitocondrial DNA is inherited from your female lineage

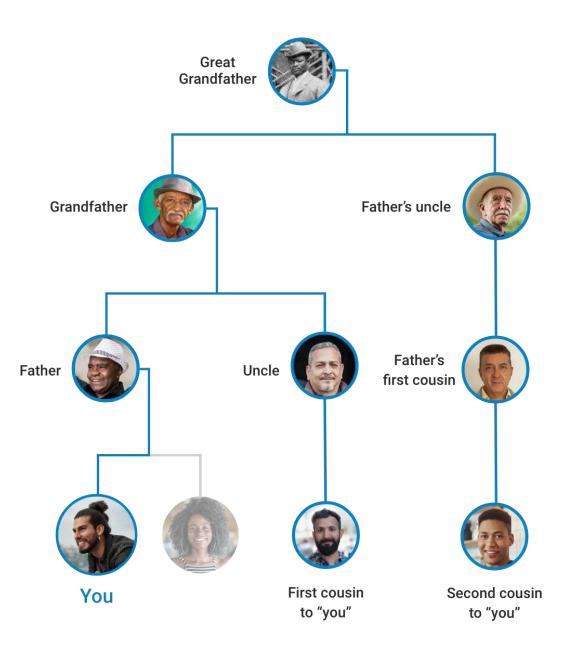




Y-DNA

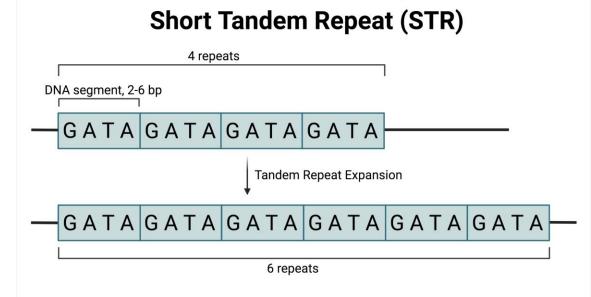
- Studies Y-chromosome (male sex chromosome)
- Handed down from father to son mostly intact
- Matches ancestors from many generations back, and their descendants
- Males have the same Ychromosome their distant ancestors had

Y-DNA Inheritance



Y-DNA: STRs and Matches

- Short Tandem Repeat (STR): Common Y-DNA variation
- Men with same or close number of STRs will match
- Genetic Distance (GD): Number of STR differences (steps) between men



Each column a number of STR repeats

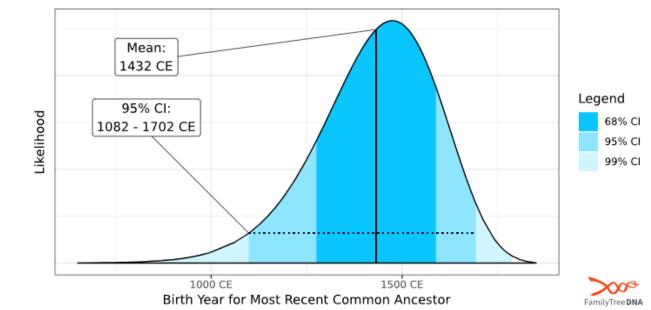
R1b-U106 >	Z307 > DF98 England-PA-MD (Dutton/Warburton Group))																					
MIN				13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	24	14	19	30	15-15-15-16
MAX				13	24	14	12	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	31	15-16-16-17
MODE				13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15-16-16
331088	Richard Dutton, b. 1785 andd. 1857	England	R-DF98	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15- <mark>15</mark> -16
102173	Thomas s/o PA John	Unknown Origin	R-M269	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15-16-16
12715	Thomas s/o PA John	England	R-U106	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15-16-16
998787	Samuel Dutton b. c.1736 Penn., d. Green Co. Ky.	United States	R-FGC13478	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15-16-16
N49453	William Dutton b. 1851 London	United Kingdom	R-DF98	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15-16- 17
316047	John Dutton of Overton (PA)	England	R-DF98	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-16-16-16
414284	George Dutton, b.1745 Cheshire, d.1831 Flintshire	England	R-FGC13446	13	24	14	11	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	31	15-15-16-16
75848	Thomas Dutton,b.1660,Cheshire UK;d.1717,MD USA	England	R-FGC13444	13	24	14	12	11-14	12	12	12	13	13	29	17 9	-10	11	11	24	14	19	30	15-15-16-16
318507	Thomas Dutton, b.1660 Cheshire UK; d.1717 Maryland	United Kingdom	R-DF98	13	24	14	12	11-14	12	12	12	13	13	29	17 9	-10	11	11	24	14	19	30	15-15-16-16
159028	Thomas Duttonb.1660, Cheshire UK; d.1717, MD USA	England	R-FGC13444	13	24	14	12	11-14	12	12	12	13	13	29	17 9	-10	11	11	25	14	19	30	15-15-16-16

of STRs Genetic **Tested** Distance **STR Results** Parke 19 멉 10 BIG Y-700 Y-DNA111 Markers Tested Genetic Big Y STR Paternal Country of Paternal Earliest Known Y Haplogroup Distance Differences Origin Ancestor 1 to 700 R-FTC74761 Onknown Origin 8 steps 10 of 657 Roger... Shows list of men with closest Genetic Link on Family Tree

- Shows list of men with closest Genetic Distance, with their Earliest Known Paternal Ancestor
- Genetic Distance used to make statistical prediction of Time to Most Recent Common Ancestor (TMRCA)
- E 🖻 Match date: June 26 2022 Parks 멉 **=** ē FAMILY FINDER Y-DNA111 Markers Tested Paternal Earliest Known Genetic Big Y STR Y Haplogroup Paternal Country of Distance Differences Origin Ancestor R-M269 1 to 111 🕂 England Not Available Samuel Parks b. 1759 (Roger... 8 steps Link on Family Tree Match date: June 26 2022 19 Park 멉 E 🖻 MTFULL SEQUENCE Y-DNA111 Markers Tested Paternal Earliest Known Genetic Big Y STR Y Haplogroup Paternal Country of Differences Distance Origin Ancestor R-M269 1 to 111 🕂 England Not Available 9 steps Roger>Roger... Link on Family Tree Match date: June 26 2022 Park 멉 := Ē MTFULL SEQUENCE Y-DNA111 FAMILY FINDER Markers Tested Paternal Earliest Known Genetic Big Y STR Y Haplogroup Paternal Country of Differences Distance Origin Ancestor R-M269 1 to 111 Not Available United States James Park b. 1798 and d. B... 9 steps Link on Family Tree Match date: June 26 2022



Y-111: Genetic Distance 8

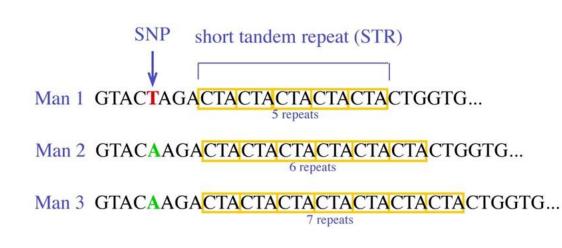


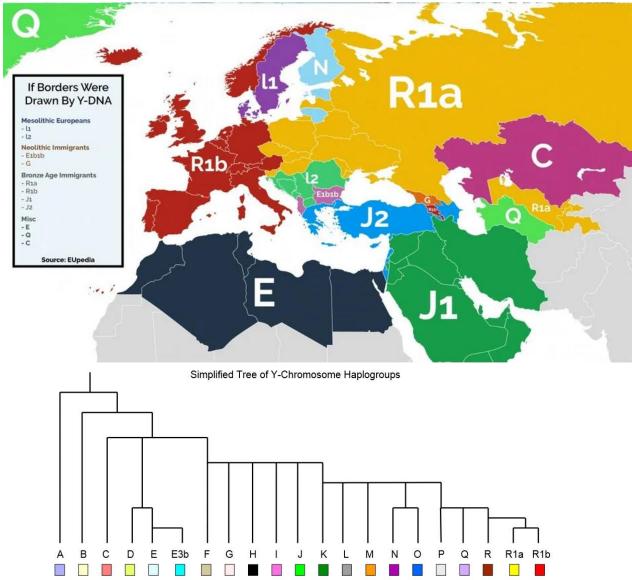
Y-111: Genetic Distance 8

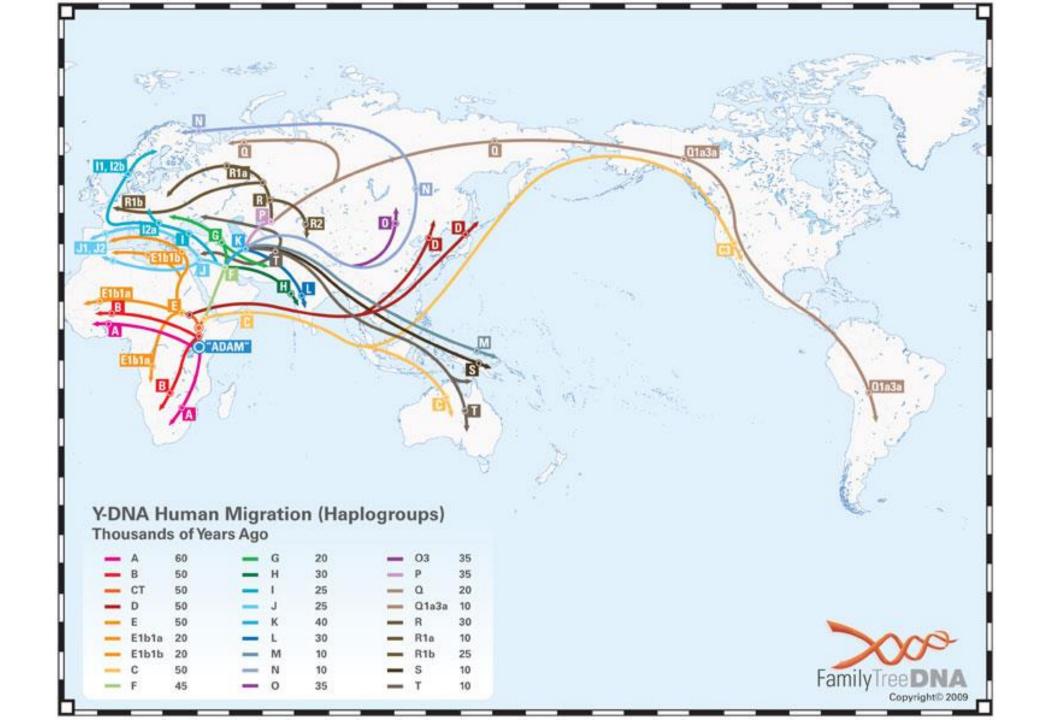
Statistic*	Years Before Present	Calendar Date
99% CI	1176 - 227	847 - 1796 CE
95% CI	941 - 321	1082 - 1702 CE
68% CI	747 - 435	1276 - 1588 CE
Mean	591	1432 CE

Y-DNA: SNPs and Haplogroups

- Single Nucleotide Polymorphism (SNP): Much rarer variation
- Men with same SNPs grouped into haplogroups
- Used to study whole population groups, human migrations, as well as families
- More definite matching

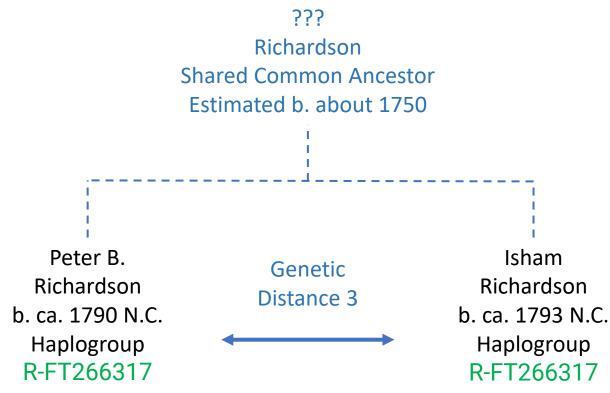






Y-DNA: Genealogical Applications (1)

- Matching other men means a shared common paternal ancestor
- Locate potential ancestors when paper trail is cold

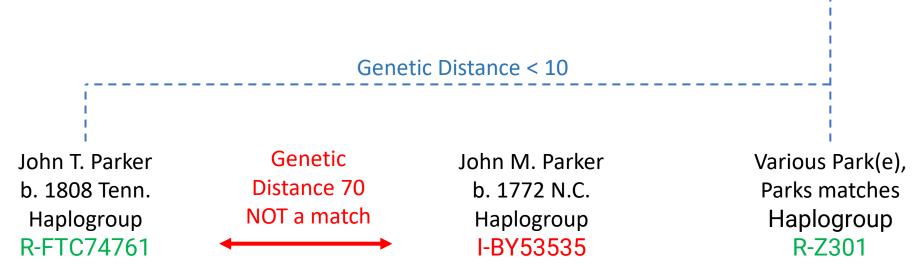


Y-DNA: Genealogical Applications (2)

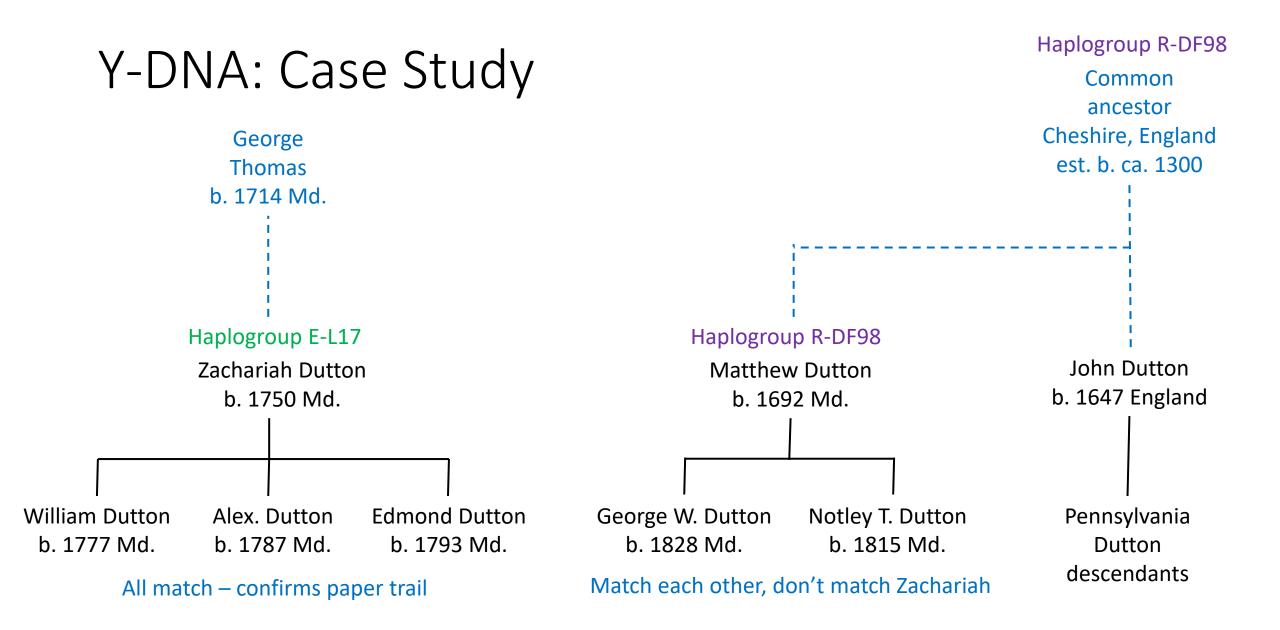
Not matching other men means you do not share a common paternal ancestor
 Roger Parke

b. 1648 England

• Prove or disprove hypotheses



No common paternal ancestor for thousands of years



Y-DNA: Ordering a Test



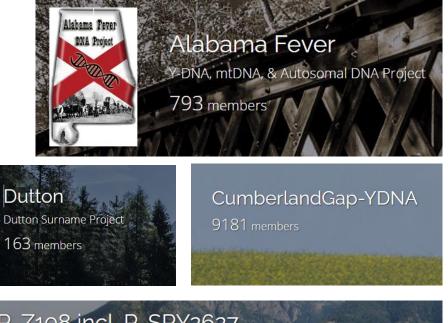
 Only major genealogy DNA provider offering Y-DNA testing is Family Tree DNA (<u>https://www.familytreedna.com</u>)

 Only genetic males can test 						
 Three levels offered: 						
 Y-37: 37 STRs, basic testing 						
 Y-111: 111 STRs, better testing 						
 Big Y-700: 700+ STRs, 500k+ SNPs 						
 Can upgrade after initial test 						

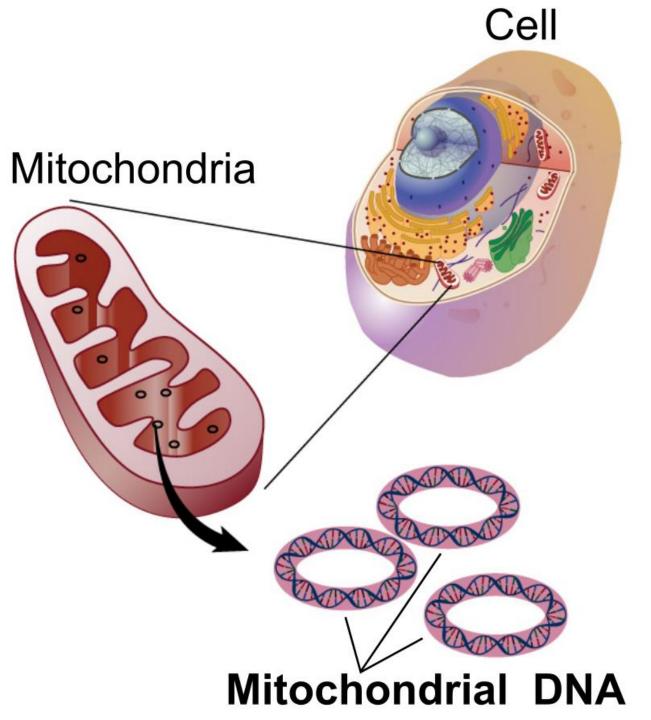
A GOOD START	REFINED MATCHING	GENEAOLOGIST FAVORITE
Y-37	Y-111	Big Y-700
\$119 ^{USD} \$99 ^{USD}	\$249 ^{USD} \$219 ^{USD}	\$449 USD \$399 USD
 37 markers is	Increase your	For community
a great place to start and can confirm close relationships	marker count to 111 to identify closer matches.	research and the most informative results, check out Big Y-700!

Group Projects

- Family Tree DNA offers many group projects for surnames, ethnicities, localities, haplogroups
- Allows experts to help with your Y-DNA
- Allows you to see how your STRs match



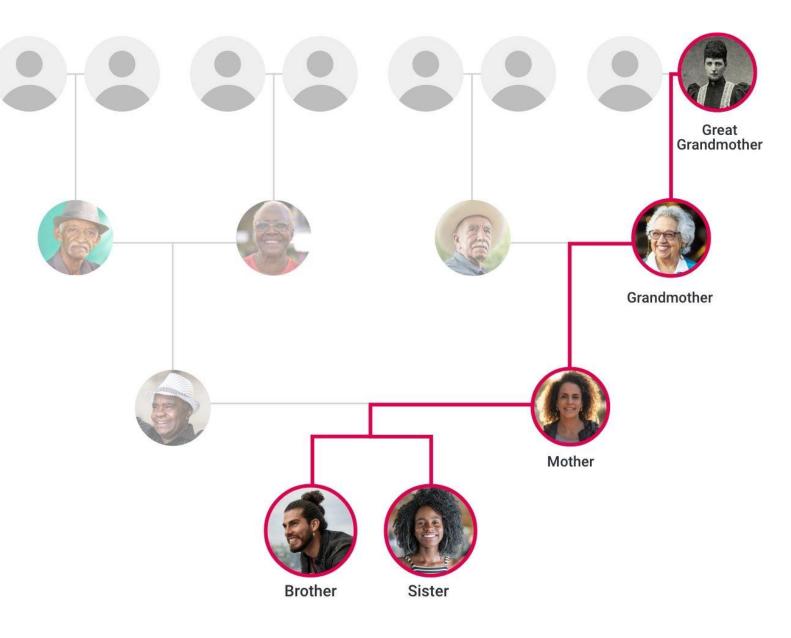
R-Z198 incl. R-SRY2627 R-Z198 and subclades R-ZS312 (incl. R-SRY2627), R-CTS4188 and R-L165 1622 members



Mitochondrial DNA (mtDNA)

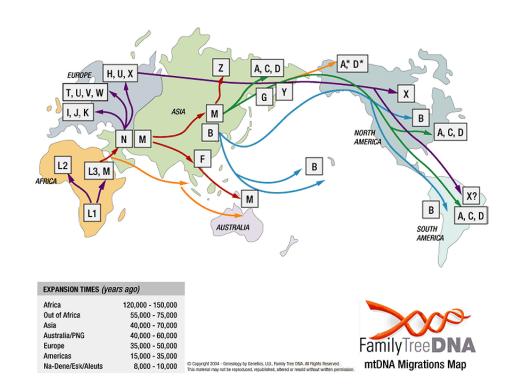
- DNA contained within *mitochondria* of cells rather than nucleus
- Passed from mother to child follows maternal line
- Both men and women can test
- Used in forensics
- Has mutations and haplogroups similar to Y-DNA

Mitochondial DNA Inheritance



Mitochondrial DNA

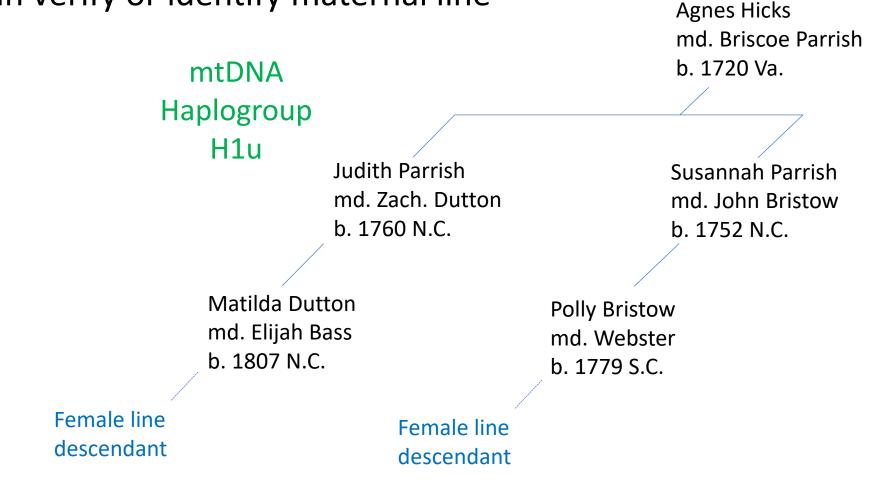
- Like Y-DNA, can chart human migration patterns
- Has Haplogroups, Genetic Distance



						Page: 1 <u>2</u> o
Genetic Distance ()	Name 🕦			Earliest Known Ancestor 🚯	mtDNA Haplogroup ()	Match Date
1	Freeman	🔺 🗐 🚏	FMS FF	Mary Jones, b. 1794 and d. 1882	H1u	12/22/2022
1	Berg	🔺 🗏 🚏	FMS		H1u	7/28/2022
1	Mr. White Jr.	M 🗉 🗗	FMS FF	Elizabeth Price, b. 1828 and d. 1904	H1u	5/23/2022
1	Mr. Cooley	M 🖻 🗗	FMS	Harriet Bennett	H1u	11/30/202
1	Thornton	🞽 🗐 🚏	FMS FF		H1u	8/6/2020
1	Barber	≥ 🗏 🖁	FMS FF		H1u	1/9/2020
1	Moore Pless	M 🗉 🗗	FMS	Martha A. Frost, b 1841 and d. Bef. 1900	H1u	1/9/2020
1	Hamilton	<mark>≌</mark> 閏 맴	FMS		H1u	1/9/2020
1	Brasher	Z 🗉 🖬	FMS FF		H1u	1/9/2020

mtDNA: Genealogical Applications

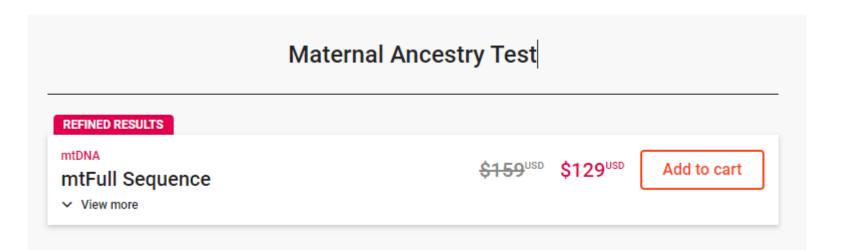
- Difficult to use because of poor documentation of maternal lines
- Can verify or identify maternal line



mtDNA: Ordering a Test

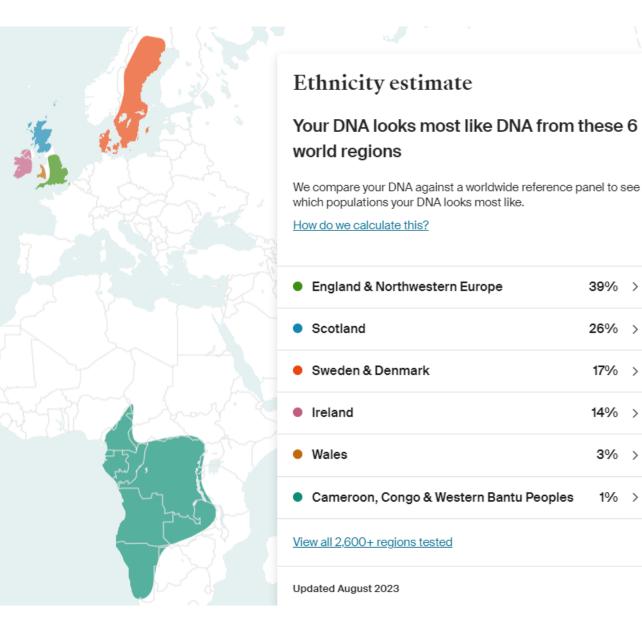


- Only major genealogy DNA provider offering mtDNA testing is Family Tree DNA (<u>https://www.familytreedna.com</u>)
- One test currently being offered
- Can be packaged with other tests



Autosomal DNA

- Compares DNA segments on 22 autosomes (non-sex chromosomes) against other testers
- Most common genealogical DNA test today (Ancestry, 23AndMe, MyHeritage, Family Tree DNA)
- Gives ethnicity estimate



17% >

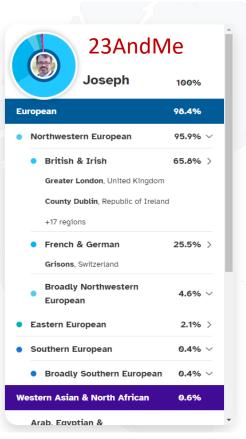
14% >

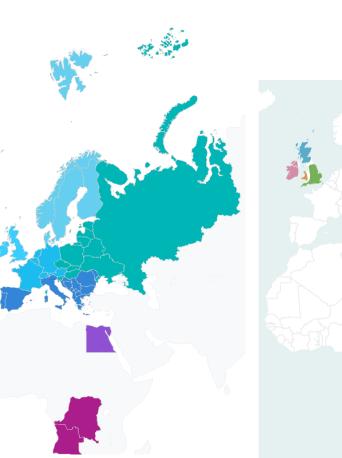
3% >

1% >

Autosomal DNA: Ethnicity Estimate

• Subjective, not precise or genealogically useful; different at each site





myOrigins[®] Version 3

myOrigins®

Joseph Richardson

Western Europe

Scandinavia

Eastern Europe

Magyar 1

Ireland

England, Wales, and Scotland

Europe

Uncover the unique genetic assemblage that has been passed down to you from your ancestors, and see to which of our 90 reference populations from ... Read more

Compare Origins

100%

81%

12%

7%

<1%

Family Tree DNA Family Finder



Ethnicity estimate AncestryDNA

Your DNA looks most like DNA from these 6 world regions

We compare your DNA against a worldwide reference panel to see which populations your DNA looks most like.

How do we calculate this?

England & Northwestern Europe	39%	>
Scotland	26%	>
Sweden & Denmark	17%	>
• Ireland	14%	>
• Wales	3%	>
Cameroon, Congo & Western Bantu Peoples	1%	>
View all 2,600+ regions tested		
Updated August 2023		

20	Thomas Richardson	Father 3,475 cM 50% shared DNA Paternal side	 Public linked tree 6,823 People Common ancestor
	Rosemary Richardson	Mother 3,473 cM 50% shared DNA Maternal side	 Public linked tree 3,010 People Common ancestor
Close Family	y		
	Robert Pierson Richardson	Grandfather 1,748 cM 25% shared DNA Paternal side	 Public linked tree 6,823 People Common ancestor
🕞 My grandf	ather.		
	Sarah Ruth Cook	Grandmother 1,676 cM 24% shared DNA Paternal side	 Public linked tree 6,823 People Common ancestor
🕞 My grandr	mother.		
2	L.D. Managed by cis901	Grandaunt 834 cM 12% shared DNA Maternal side	 Public linked tree 7,389 People Common ancestor
🗊 Lorene Du	utton Bryant, my great-aunt	test by Crystal, her ex-granddaughter-in-l	aw).
	Lorene Dutton	Grandaunt 832 cM 12% shared DNA Maternal side	₽Public linked tree8,596 People▲Common ancestor
E Lorene Du	utton Bryant, my great-aunt.		
8	C.M. Managed by	1st cousin 1x removed 556 cM 8% shared DNA	Public linked tree733 People

Deternal oide

Common encoster.

Autosomal DNA: Matching

- List of DNA matches cousins who share one or more common ancestors
- Shared centiMorgans (cM) or percentage
- Number of shared segments



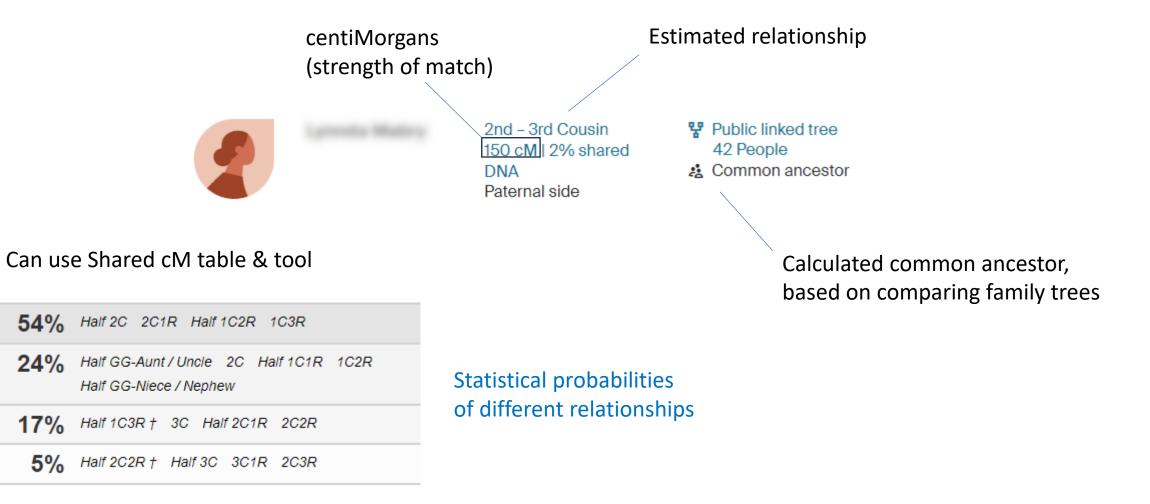
Autosomal DNA – Genealogical Applications

- Discovering unknown ancestors
 - Finding people you share DNA with, with ancestors they have documented but you haven't
 - Projecting unknown common ancestry from matches
- Proving ancestral hypotheses

How am I related to a match?

•

• Identifying a match by the shared centiMorgans (cM)

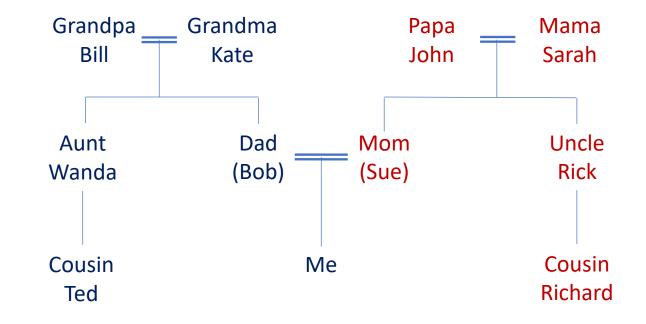


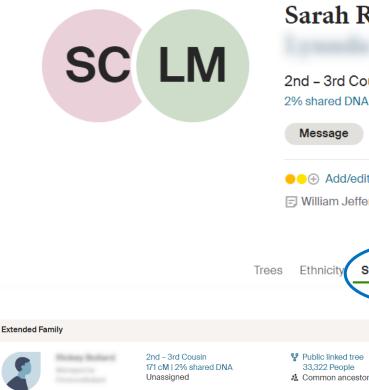
Using Shared Matches: Basic Example

- My Unknown Match, Tom
 - Matches in common:
 - Me
 - Cousin Richard
- Means common ancestors will be on maternal side of family

???

Tom





2nd - 3rd Cousin

2nd – 3rd Cousin

Paternal side

110 cM | 2% shared DNA

Paternal side

William Jefferson Bullard > William Riley Bullard (5 generations from W. J.)

Bullard

127 cM | 2% shared DNA

Sarah Ruth Cook and

2nd - 3rd Cousin | Paternal side 2% shared DNA: 150 cM across 7 segments

●● ① Add/edit groups

Section 2017 Secti

Do you recognize them?

Do you recognize them?

Do you recognize them?

Learn more

Learn more

Learn more

0

0

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Public linked tree

Common ancestor

Public linked tree

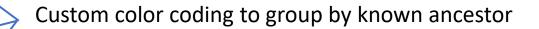
Common ancestor

20 People

747 People

Using Shared Matches

• Using two or more in-commonwith (ICW) matches to identify what family line a match is likely on



E William Jefferson Bullard > John W. R. Bullard

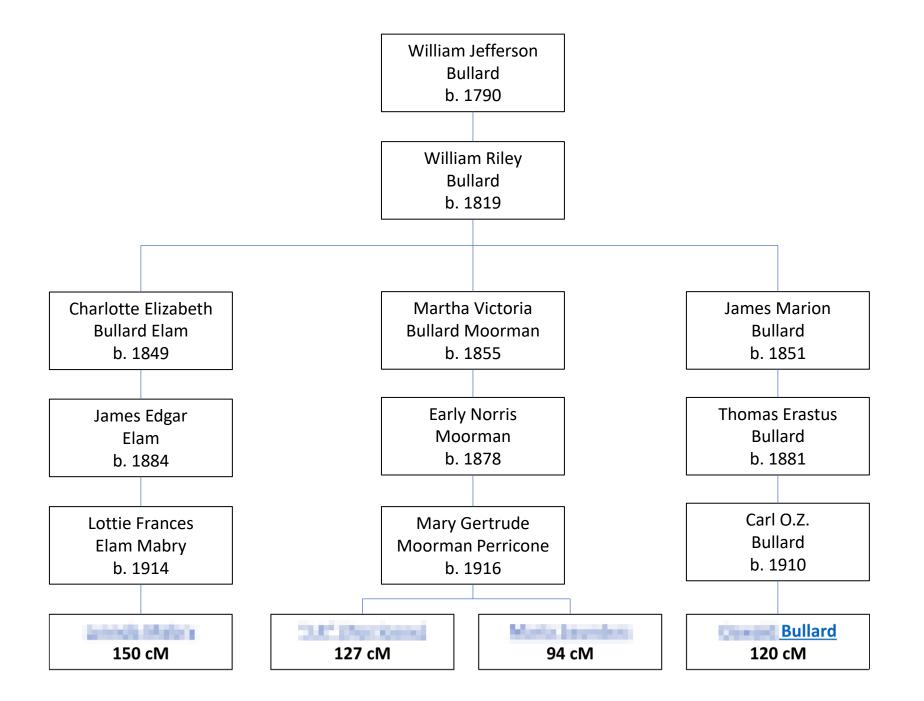
William Riley Bullard > James Marion Bullard

The Shared cM Project – Version 4.0 (March 2020)

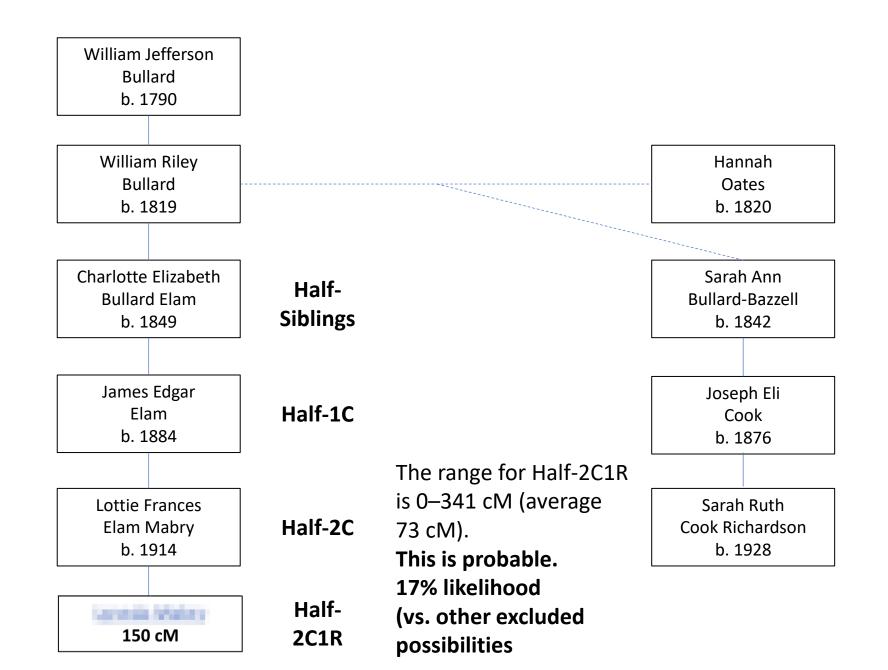
Blaine T. Betting www.TheGenetic CC 4.0 Attributic	IneticGenealogist.com How to read this chart: Great-G							GGGG- Aunt/Uncle			
									GGG- Aunt/Uncle		
Half GG- Aunt/Uncle 208 103 – 284			Gi	reat-Grandpare 887 485 – 1486		Great-Great Aunt/Uncle 420 186 – 713	1C3R 117 25 – 238	2c3R 51 0 - 154	Other Relationships		
Half 1C2R 125 16 – 269	Half Great- Aunt/Uncle 431 184 – 668		Grandparent 1754 984 – 2462				Great Aunt/Uncle 850 330 – 1467	1C2R 221 33 - 471	2c2R 71 0- 244	3C2R 36 0 – 166	6C 18 0 - 71
Half 2c1R 66 0 - 190	Half 1C1R 224 62 - 469	Half Aunt/Uncle 871 492 – 1315		Parent 3485 2376 - 3720		Aunt/Uncle 1741 1201 - 2282	1C1R 433 102 – 980	2c1R 122 14 - 353	3C1R 48 0 - 192	4C1R 28 0 - 126	6C1R 15 0 - 56
Half 3c 48 0 – 168	Half 2c 120 10 - 325	Half 1C 449 156 – 979	Half-Sibling 1759 1160 – 2436	Sibling 2613 1613 - 3488	SELF	1C 866 396 - 1397	2c 229 41 - 592	3c 73 0 - 234	4c 35 0 - 139	5c 25 0 - 117	6C2R 13 0 - 45
Half 3c1R 37 0 - 139	Half 2c1R 66 0 - 190	Half 1C1R 224 62 - 469	Half Niece/Nephew 871 492 - 1315	Niece/Nephew 1740 1201 - 2282	Child 3487 2376 - 3720	1C1R 433 102 – 980	2c1R 122 14 - 353	3C1R 48 0 - 192	4C1R 28 0 - 126	5C1R 21 0 - 80	7 C 14 0 - 57
Half 3c2R 27 0 - 78	Half 2c2R 48 0 - 144	Half 1C2R 125 16 - 269	Half Great Niece/Nephew 431 184 – 668	Great- Niece/Nephew 850 330 – 1467	Grandchild 1754 984 – 2462	1C2R 221 33 - 471	2c2R 71 0- 244	3C2R 36 0 – 166	4C2R 22 0 - 93	5C2R 18 0 - 65	7C1R 12 0 - 50
Half 3c3R	Half 2c3R	Half 1C3R 60 0 - 120	Half GG Niece/Nephew 208 103 - 284	Great-Great Niece/Nephew 420 186 – 713	Great- Grandchild 887 485 – 1486	1C3R 117 25 - 238	2c3R 51 0 – 154	3C3R 27 0 – 98	4C3R 19 0 - 60	5C3R 13 0 - 30	8C 11 0 - 42

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

https://dnapainter.com/tools/sharedcmv4



Discovering the True Father of Sarah Ann (Bullard) Cook





Chromosome Browser

- Graphs what segments of DNA a match shares
- If two or more people share the same segment of DNA, they likely share the same common ancestor

Autosomal DNA: Ordering a Test

Testing Company	AncestryDNA	23AndMe	Family Tree DNA Family Finder	MyHeritage
Pros	Largest database = most people to match with; good family tree tools	Health information	Chromosome browser, group projects	Chromosome browser, good family tree and tools
Cons	No chromosome browser	No tools for family tree sharing or matching	Slow and clunky site; smaller autosomal database	Make you pay for everything
Cost	\$99	\$119 (ancestry only) \$229 (health+ancestry)	\$79	\$89

My recommendation:

- Test with more than one
- If you only do one, do Ancestry and upload to different sites
- If you do Y-DNA or mtDNA with FTDNA, add their autosomal also

Autosomal tips

- DO post your family tree
- DO test your oldest family members
- DO test as many family members as possible
- DO test with, or upload to, as many sites as possible
- DO check your messages regularly



Uploading DNA to Other Sites

Company	GEDmatch	AncestryDNA	23AndMe	Family Tree DNA	MyHeritage
Allows downloads	(does not test)	\checkmark	\checkmark	\checkmark	\checkmark
Allows uploads	\checkmark	no	no	\checkmark	\checkmark

- Allows matching with other sites' database
- I highly recommend GEDmatch (<u>https://www.gedmatch.com</u>) as a site for advanced DNA tools (chromosome browser, etc.) and matching with tests from many different sites.

Advanced Topics

- Chromosome painting and mapping
- Phasing and reverse phasing
- Clustering



Sarah Cook Richardson Full Female

~ 90% / 987 segments painted 🏾 🏾 🕫

