

Introduction to Genetic Genealogy

Brad Larkin Surname DNA Journal http://www.surnamedna.com

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Topics

- Type of DNA used in Genetic Genealogy
- SNP vs. STR Measurement
- DNA Relationship Grouping
- Identifying Geographical Origins



Genetic Genealogy

- Genealogy
 - First genealogy society (NEHGS) 1845
 - Who were our ancestors?
 - Where Do We Come From?
- Genetic Genealogy
 - Family Tree DNA founded in 2000
 - Use of genealogical DNA testing to determine the level and type of relationship between individuals.



Types of DNA

Three main types of DNA testing from a genealogy perspective

- 1. Y-Chromosome
- 2. Mitochondrial (MtDNA, from the mother's egg cell)
- 3. Autosomal (chromosomes 1-22)



Butler, J.M. (2005) Forensic DNA Typing, 2nd Edition, Figure 2.3, @Elsevier Science/Academic Press

Chromosome Fit for Genealogy

	Autosomal	Y-Chromosome	Mitochondrial
Recombination - Mixing	Yes	No	No
# Coding Genes	~ 30,000	86	37
# Markers Initial Test	708,093	37	1,120
Mutation Rate	0.5 bp/gen =	µ = 0.0041bp/gen	0.48 bp/MY =
	354,047 per generation	1 change per 165 years	1 change per 1,860 years



Autosomal (passed on in part, from all ancestors)



Y-Chromosome

(passed on complete,

but only by sons)



Mitochondrial (passed on complete, but only by daughters)

SNP vs. STR Measurement

- SNP = Single Nucleotide Polymorphism
- Mutation in a single base pair at a specific position
- Expressed a 'positive' when different from all other human beings.
 - e.g. *position rs1019875*
 - Person1 TATCCT = -
 - Person2 TACCCT = +
- Analogous to 'Trunk and Branches of the Tree'



- STR = Single Tandem Repeat
- Repeating patterns of multiple base pairs
- Allele Count = number repetitions of a particular pattern

 Analogous to 'Leaves on the Tree'



DNA Relationship Grouping

Haplogroup

- Major branches of human tree
 - (e.g. 40 branches for all humanity)
- Always measured with SNP mutations
 - As the number of SNPs identified grows, subordinate Haplogroups are identified.
 - e.g. M222 with haplogroup R = R-M222

Haplotype

- Sub branches of a haplogroup
- e.g. 40 branches from within R-M222
- Typically identified with STR patterns
 - May later be confirmed to share an SNP

			e 142 d 148	R1a1d R1a1e R1b*
		M18		R1b1*
M123 - 7241		1000-		Ribia
P231, P233		M73		R1515*
P234. P236			The second s	R16161
P238, P242			* M37	R1b1b2*
P206, P204 A	343		a	R1b1b2a
b			b	R1b1b2b
			C M133	R1b1b2c
1 (I (I (I (I (I (I (I (I (I (4 100		d 5/072027	R1b1b2d
	1/23	P297	e M222 = USPSY + 3636	R1b1b2e
and the second second		2 10.20	9 190	R1b1b2f
			1 2	R1b1b2g*
144			g 0106 1 0198	R1b1b2g1
			2 1107	R1b1b2g2
		A 13	1	R1b1b2h*
		300	h 0152 1 M126	R1b1b2h1
		-	2 M160	A1b1b2h2
2	10	C M335		Ribic
- Miles, P249, P2	67			R2

Autosomal Match Measurement

- Total and Longest Blocks of shared segments
 - express in the units of CentiMorgans (cM).

Known Relationship	Length of All Shared Blocks (cM)	Longest Block (cM)
Son	3,382	145
Mother	3,380	144
Grandmother	1,757	123

• In more distant relationships, a single shared segment can be though of as a 'match'.



Shared Segment Measurement

- A specific portion of DNA that two or more people have in common
 - IBD = Inherited by Descent
 - DNA Segment inherited by more than one descendant of a common ancestor
- Described by:
 - The Chromosome
 - The Starting Position
 - count of base pairs from beginning of the chromosome to the point at which shared segment begins
 - The Length
 - e.g. Chromosome 20;
 position 1,115,665;
 50 cM



Can a DNA Test Tell Where You Came From?

- Yes if your genetic relatives have already been tested and know their geographic origin.
- Interpretive results improve as more sampling is done in ancestral areas.
 - More Matches



Effective Geographic Identification Using Genetic Genealogy

Identification of geographic origin depends on two factors:

1. MATCHES with GEOGRAPHY Having genetic matches whose geographic origin is known with precision.



2. RESOLUTION

Sufficient resolution in the DNA tests to indicate that the time-to-most-recent-common-ancestor (TMRCA) is proportionate to geographic movements.

A match of 33 on a 37-marker Y STR test ~ 400 years.

How You Can Participate

- Get a male relative to take a Y-37 STR DNA test.
- Recruit DNA participants with known ancestral origins.
 - DNA Testing Instructional video on <u>YouTube.com</u>
- Link Autosomal Shared Segments to Ancestral Geography
- Sponsor Ancestral Parish Sample DNA Testing



Information Links

• Genetic Homeland



- Searchable list of castles, surnames, DNA results, and place names plotted on maps
- <u>www.genetichomeland.com</u>
- FTDNA FAQ
 - http://www.familytreedna.com/faq/
- Surname DNA Journal
 - http://www.surnamedna.com
- Your Genetic Genealogist
 - http://www.yourgeneticgenealogist.com
- DNAeXplained
 - http://dna-explained.com

