

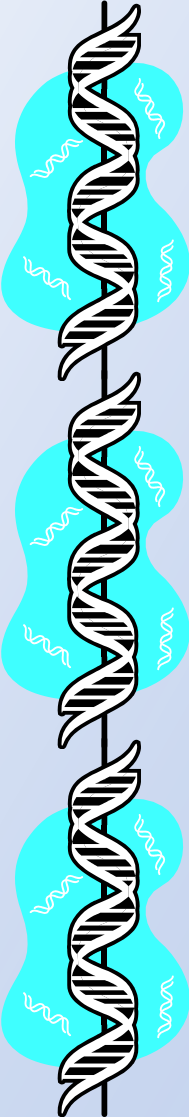


DNA and Genealogy: The Basics

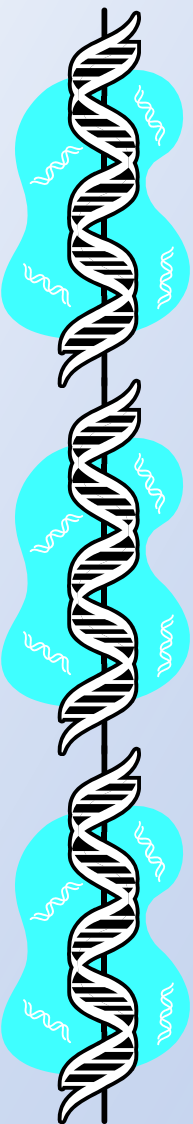
Presented to the
Pinellas Genealogy Society
on October 20, 2007 by Ed Deming

What will be covered?

- Some Terminology
- Types of DNA Testing
- Basic migration maps
- What kind of genealogy questions can be answered? What can't?
- Who are candidates for testing?
- Where can I get my DNA tested?
- Online databases and tools
- A real Y-DNA example

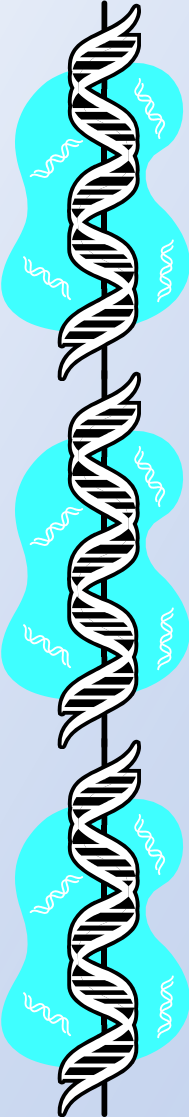


Some Terminology

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- **Allele** – variant form of a gene
 - **Genetic marker** – a DNA sequence with known characteristics that can be tested for comparison purposes
 - **Haplogroup** – defined by mutations on Y-DNA or mtDNA; members link to the original appearance of the mutation; frequently geographically related
 - **Haplotype** – an individual's complete set of test results; a single difference delineates a distinct haplotype
 - **Loci** – positions where particular genetic markers are located

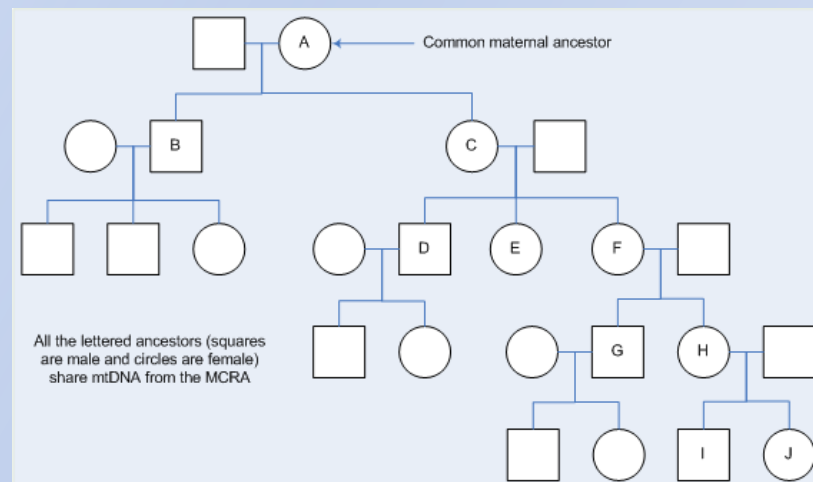
Terminology continued

- **Most Recent Common Ancestor (MRCA)** – the shared ancestor of two or more haplotypes who represents their closest link
- **Mutation rate** – the frequency at which genetic change occurs for specific markers; used in calculations of MRCA or generation distance of MRCA
- **Phylogenetic network/tree diagram** – a pictorial representation of the relationships among individuals and haplogroups sharing a common ancestor



Types of DNA Testing in Genealogy

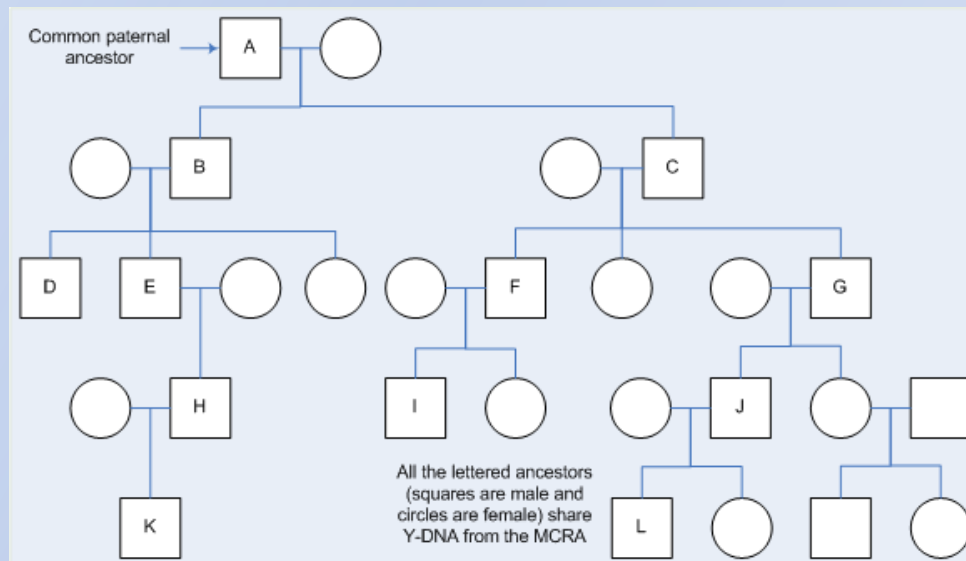
- Mitochondrial DNA (mtDNA)
 - Available for both males and females
 - Changes very slowly (a haplotype could remain unchanged for thousands of years)
 - If you share the same mtDNA you share a common maternal ancestor
 - Could be applied to old hair samples



Types of DNA Testing in Genealogy

(continued)

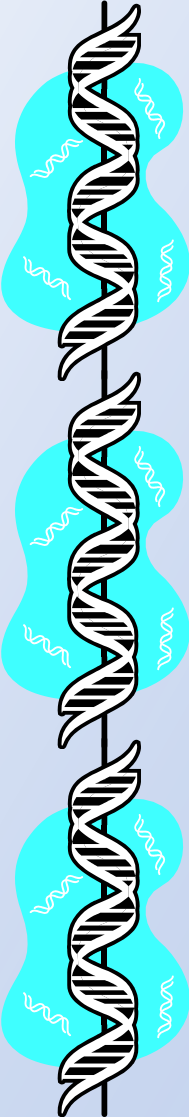
- Y chromosome DNA (Y-DNA)
 - Available only for males
 - Shared markers used to determine a probability for a mean time to common paternal ancestor
 - Changes slowly (a haplotype could remain unchanged for hundreds of years)



Types of DNA Testing in Genealogy

(continued)

- Geographical (paternal or maternal)
 - mtDNA or Y-DNA testing can be used
 - Smaller number of loci can be tested
 - Seven daughters of Eve; or ancestral clans



Haplogroups and Migration Patterns

Tracing Human History Through Genetic Mutations

By examining DNA patterns that are inherited maternally or paternally, scientists can trace human lineages back to the original branches, or sons and daughters, of a genetic Adam and an Eve.

Europe

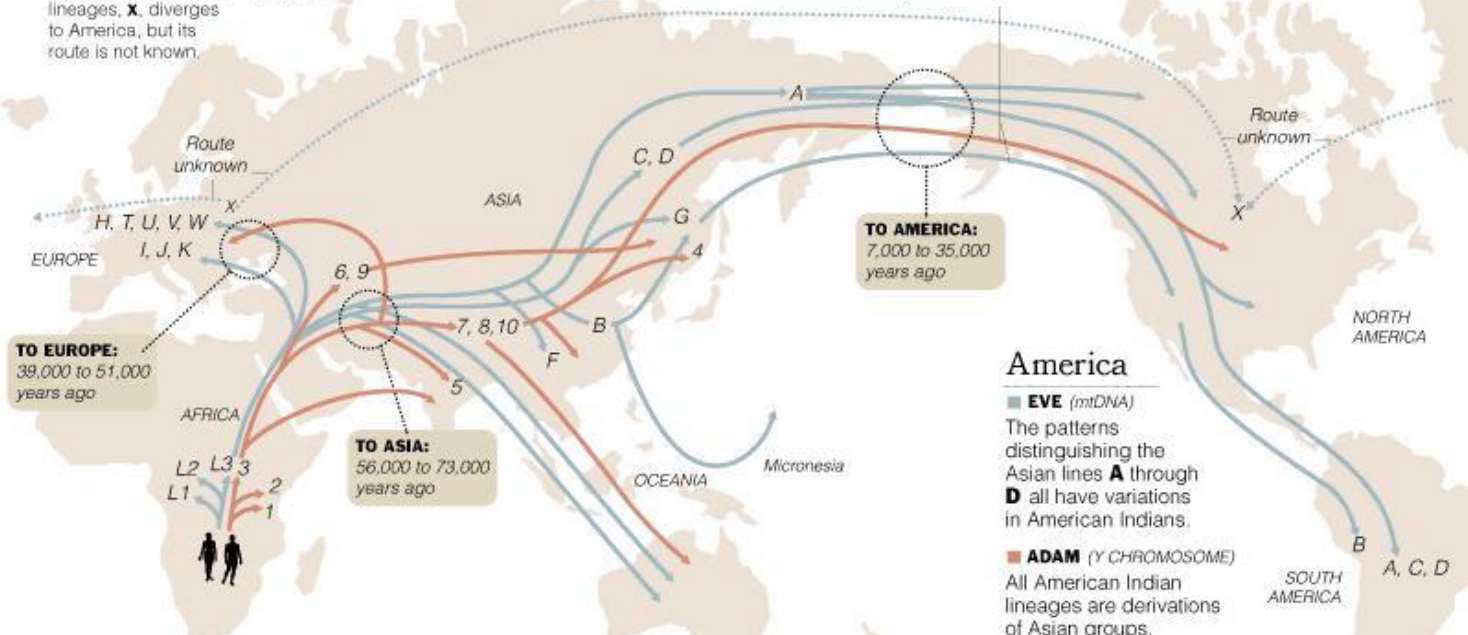
EVE (mtDNA)

The nine European lineages are named **H** through **K**, and **T** through **X**. One of the lineages, **X**, diverges to America, but its route is not known.

ADAM (Y CHROMOSOME)

All European lineages are variations of African and Asian branches.

Men and women certainly colonized the world together; the differences between the routes shown reflect differences in genetic information.



Africa

EVE (mtDNA)

The three African branches are named **L1** through **L3**, and **L3** separates into all the other branches.

ADAM (Y CHROMOSOME)

The three African branches are named **1**, **2** and **3**, and **3** separates into all the other branches.

Asia

EVE (mtDNA)

The six Asian branches are named **A** through **D** and **F** and **G**.

ADAM (Y CHROMOSOME)

The seven Asian branches are **4** through **10**, and these groups branch off into Oceania, Europe and America.

America

EVE (mtDNA)

The patterns distinguishing the Asian lines **A** through **D** all have variations in American Indians.

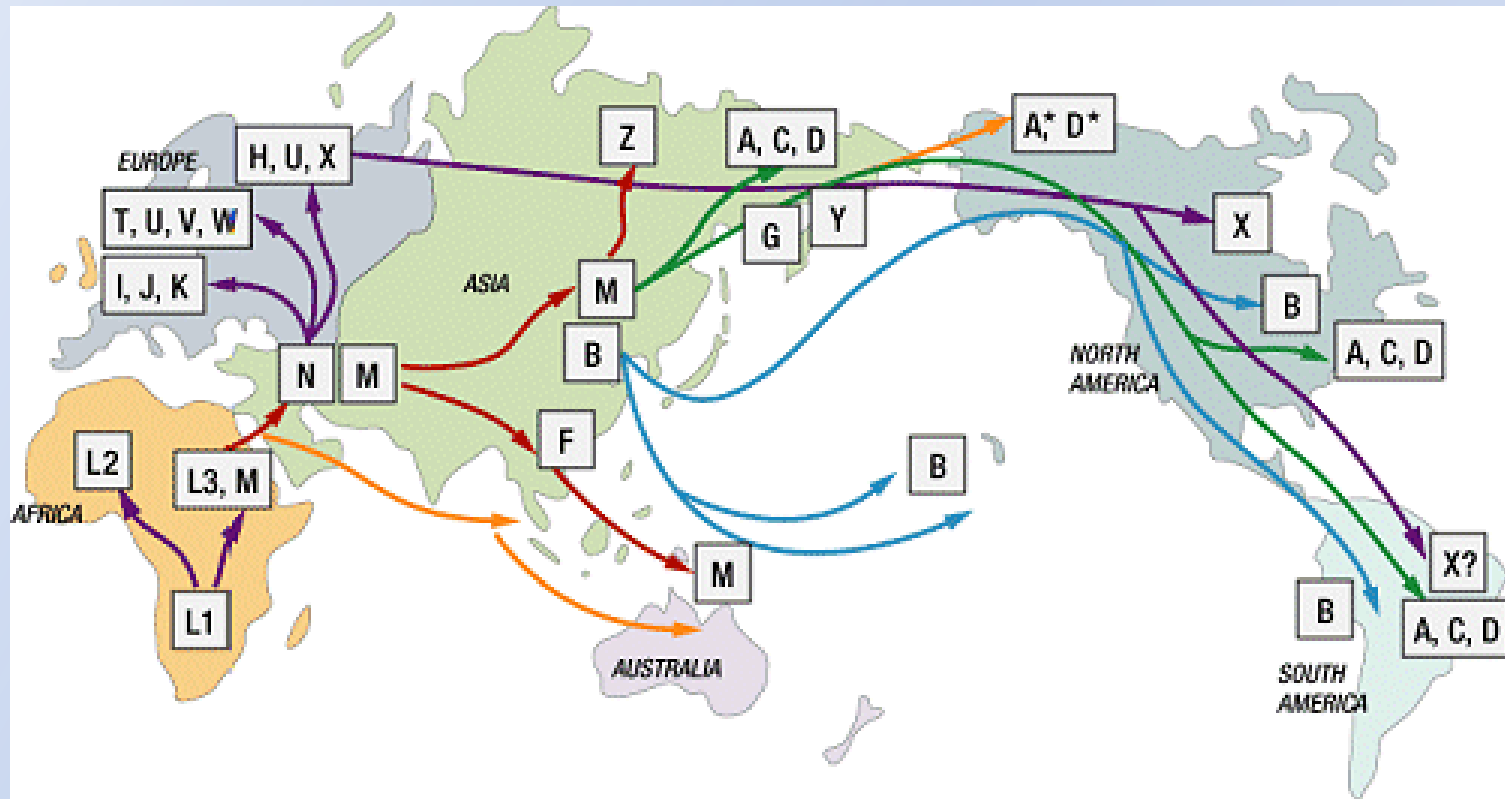
ADAM (Y CHROMOSOME)

All American Indian lineages are derivations of Asian groups.

Sources: Dr. Douglas C. Wallace, Marie T. Lott, Emory University; Dr. Peter A. Underhill, Stanford University; "Genes, Peoples, and Languages," by Dr. Luca Cavalli-Sforza

Steve Duenes/The New York Times

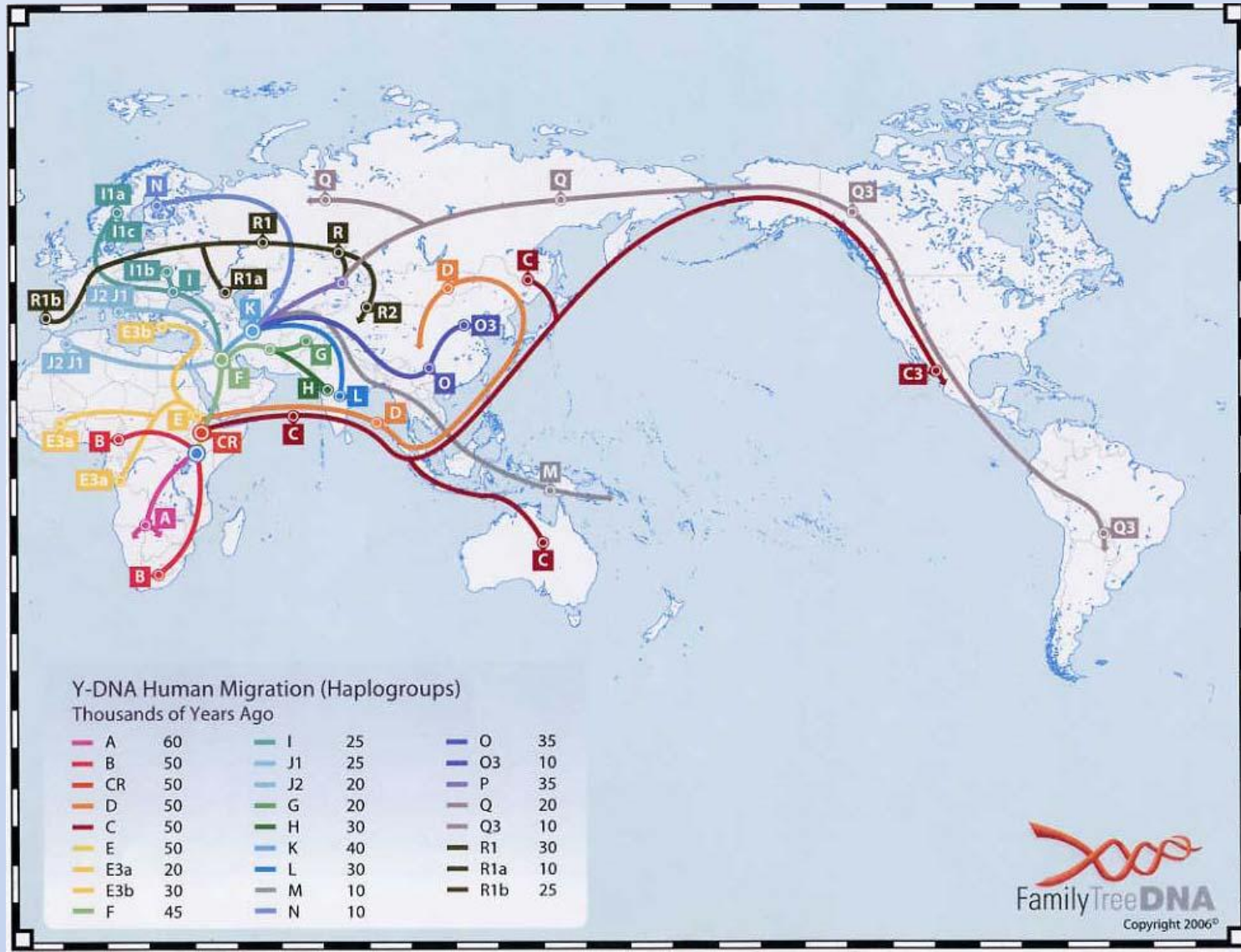
mtDNA Migrations



EXPANSION TIMES (years ago)

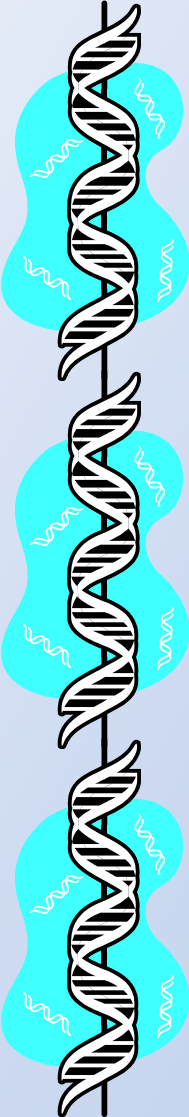
Africa	120,000 - 150,000
Out of Africa	55,000 - 75,000
Asia	40,000 - 70,000
Australia/PNG	40,000 - 60,000
Europe	35,000 - 50,000
Americas	15,000 - 35,000
Na-Dene/Esk/Aleuts	8,000 - 10,000

Y-DNA Migrations

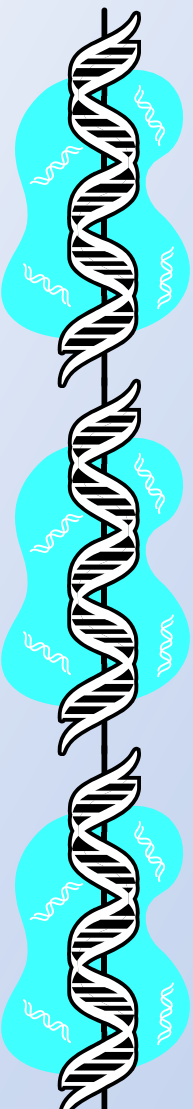


What use is mtDNA in genealogy?

- Verify or refute matrilineal ancestry
- Determine which of multiple same surname families a female ancestor could come from
- Other?



What use is Y-DNA in genealogy?

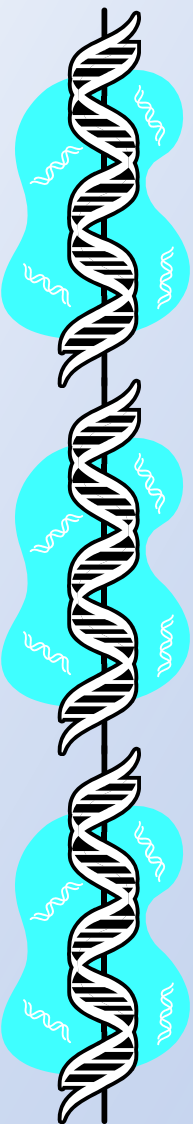
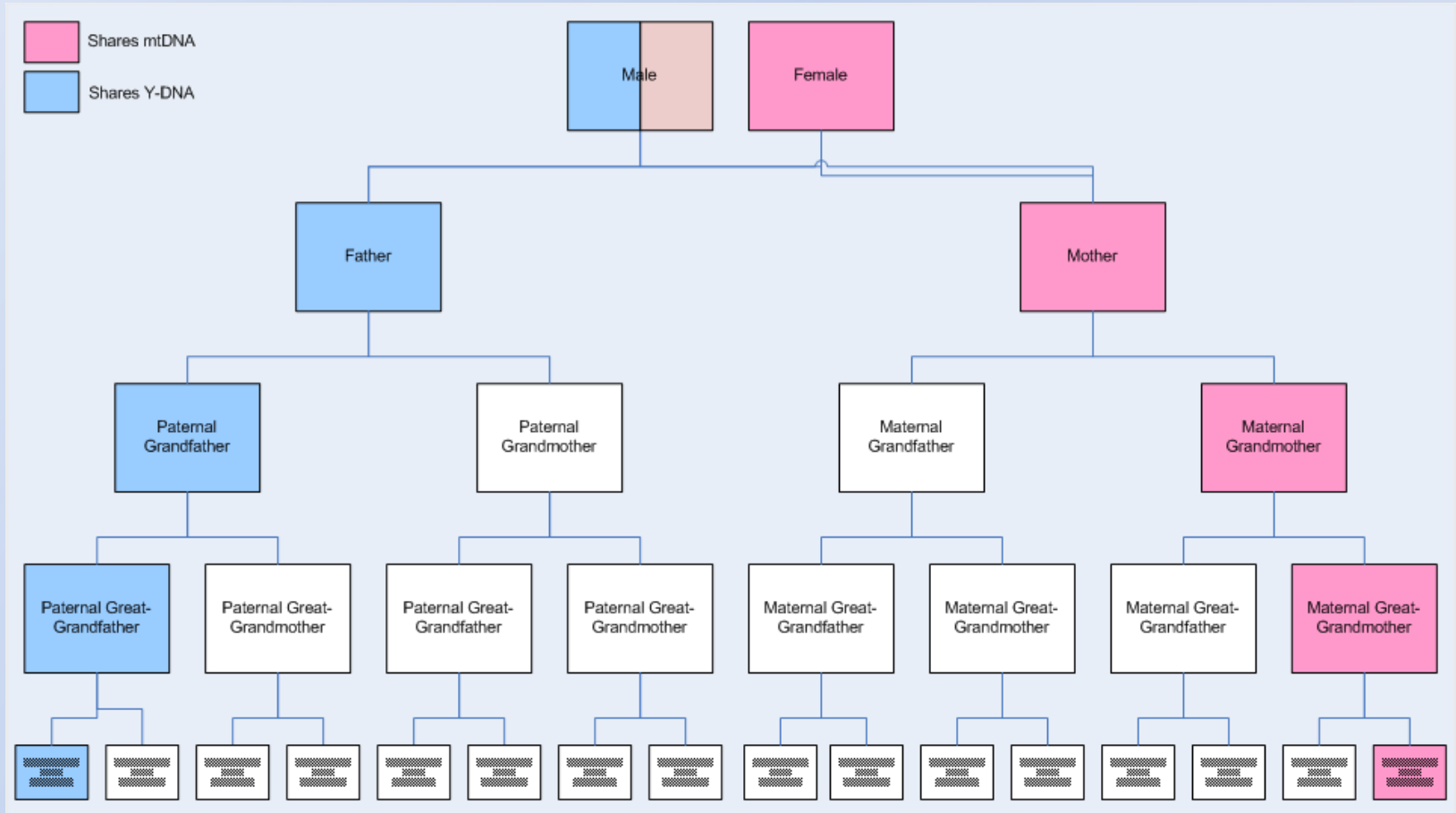
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- Broad surname study – what family groupings exist for a given surname?
 - Are two families with the same surname related?
 - Are related surnames descended from the same family?
 - Verify or refute descent from a given male ancestor

What Y-DNA or mtDNA don't tell you

- If you have an inheritable disease or susceptibility
- A certainty of anything. All results are probabilities.

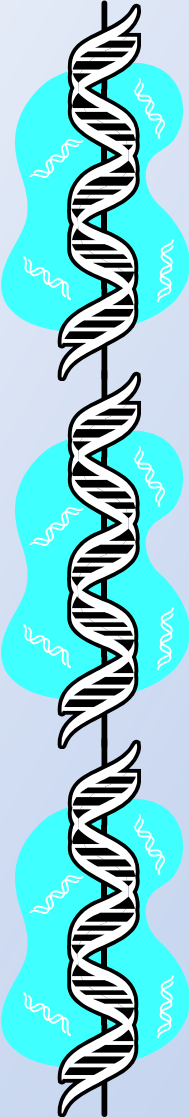


Who are candidates for testing?



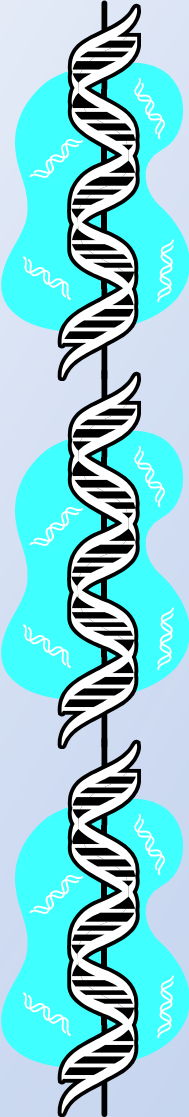
Where can I get my DNA tested?

- Family Tree DNA – www.familytreedna.com
 - 100,000+ Y-DNA; almost 56,000 mtDNA
- DNA Heritage – www.dnaheritage.com
 - Used by about 75,000
- DNA Ancestry (formerly Relative Genetics) – dnaancestry.com
- Oxford Ancestors – www.oxfordancestors.com
 - Low resolution tests to determine haplogroup
- National Geographic Geographic Project – www3.nationalgeographic.com/genographic



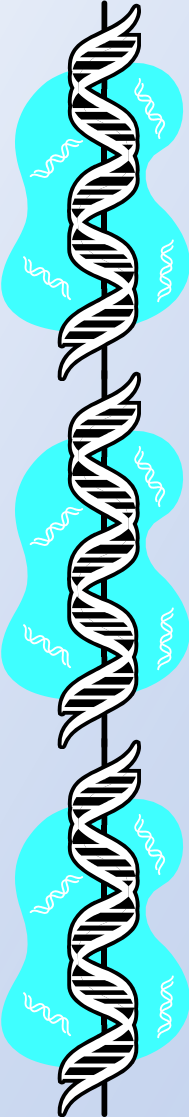
Online databases

- Sorenson Molecular Genealogy Foundation – www.smgf.org (y DNA and mtDNA)
- y search – www.ysearch.org
- Ybase: genealogy by numbers – www.ybase.org
- y Chromosome Haplotype Reference Database – www.yhrd.org
- mito search – www.mitosearch.org



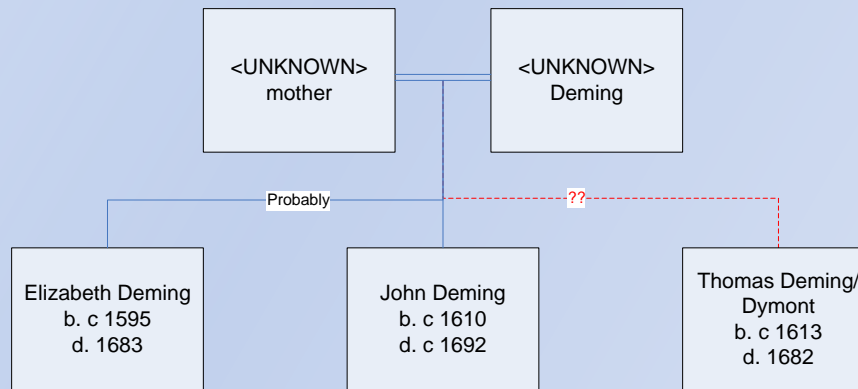
Tools

- Y-Utility: Y-DNA Comparison Utility – www.mymcgee.com/tools/yutility.html
- Haplogroup predictor – <https://home.comcast.net/~hapest5/index.html>
- mtDNAtool: An mtDNA Analysis Utility – <http://freepages.genealogy.rootsweb.com/~glad/dna/mtdnatool.html>



A real Y-DNA example

- The question to be tested using Y-DNA: were John Deming and Thomas Deming/Dymont, both in Wethersfield, CT in the 1640's brothers as some speculate?



A real Y-DNA example (continued)

- An existing surname study for the Diamond Family contained three descendants of Thomas Deming/Dymont

Result comparison

Genetic Distance						
ID	John Deming-1	John Deming-2	John Deming-3	Thomas Dymont-1	Thomas Dymont-2	Thomas Dymont-3
John Deming-1	37	0	1	10	16	10
John Deming-2	0	12	0	5	5	5
John Deming-3	1	0	31	10	12	10
Thomas Dymont-1	10	5	10	25	0	1
Thomas Dymont-2	16	5	12	0	37	1
Thomas Dymont-3	10	5	10	1	1	25
Related	Probably Related		Possibly Related			

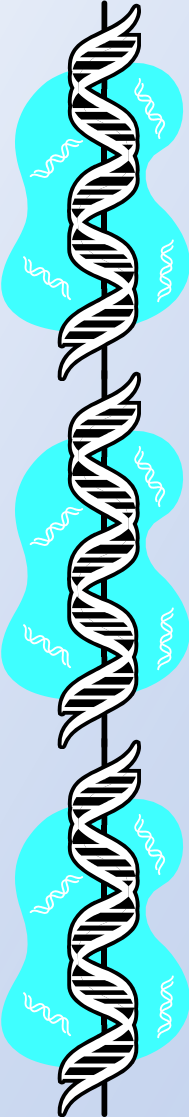
Infinite allele mutation model is used
 Values on the diagonal indicate number of markers tested
 For Family Tree DNA's discussion on interpreting Genetic Distance see:
 - For 12 Marker Tests - www.familytreedna.com/gdrules_12.htm
 - For 25 Marker Tests - www.familytreedna.com/gdrules_25.htm
 - For 37 marker Tests - www.familytreedna.com/gdrules_37.htm

Time to Most Recent Common Ancestor (Years)						
ID	John Deming-1	John Deming-2	John Deming-3	Thomas Dymont-1	Thomas Dymont-2	Thomas Dymont-3
John Deming-1	37	450	360	3570	2850	3570
John Deming-2	450	12	450	4650	4650	4650
John Deming-3	360	450	31	3570	3360	3570
Thomas Dymont-1	3570	4650	3570	25	180	450
Thomas Dymont-2	2850	4650	3360	180	37	450
Thomas Dymont-3	3570	4650	3570	450	450	25
0-270 Years	300-570 Years		600-870 Years		900-1170 Years	

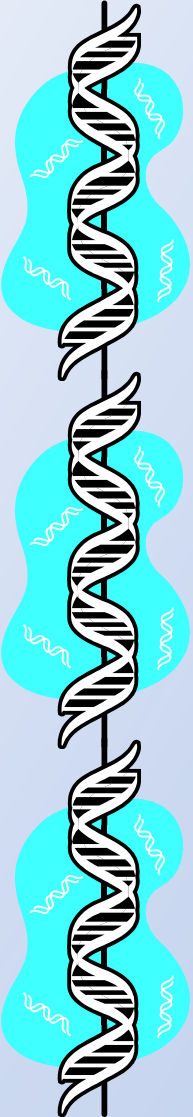
- Infinite allele mutation model is used
 - Average mutation rate varies: 0.0020 to 0.0031 rates derived by Doug McDonald from the Sorenson database
 - Values on the diagonal indicate number of markers tested
 - Probability is 50% that the TMRCA is no longer than indicated
 - Average generation: 30 years

A real Y-DNA example (continued)

- There is very little likelihood that John Deming and Thomas Deming/Dymont were related, except thousands of year ago.



Questions?



An example phylogenetic network tree diagram

