

# DNA Testing: The Very, Very, Basics

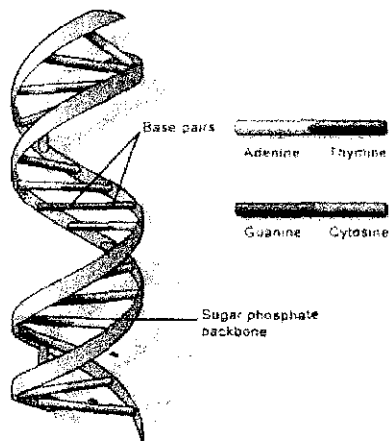
Thomas H. Shawker, M.D.

## Genetic Testing for Genealogy (Genetic Genealogy)

1. Genetic tests, two types currently in use:
  - a. The Y-chromosome for paternal ancestors
  - b. mtDNA for maternal ancestors
2. Using the information: Relationships between individuals, Family reconstruction, Surname societies, Ethnic origins
3. Genetic Testing Laboratories:
  - Oxford Ancestors: <http://www.oxfordancestors.com> (10 Y markers)
  - Relative Genetics: <http://www.relativegenetics.com> (18, 26, 43 Y markers)
  - Family Tree DNA: <http://www.familytreedna.com> (12, 37, 67 Y markers)
  - DNA Heritage: <http://www.dnaheritage.com/> (43 Y markers)
4. Securing the Specimen – cheek swab, swabs into containers and mailed to labs

## DNA

1. Coiled Helix Molecule (“spiral ladder”) with alternating sequence of 4 bases: A,G,C,T
2. Two types of “ladder” rungs: T-C and G-A; these bases are always paired
3. DNA: in chromosomes within cell nucleus & in cytoplasm in mitochondria (mtDNA)
4. Chromosomes:
  - a. 23 Pairs of chromosomes in the cell nucleus
  - b. One of each pair comes from the father, the other comes from the mother
  - c. 22 non-sex chromosomes; 1 pair of sex chromosomes (XX = female; XY = male)
  - d. For genealogy relationships – only using Y-chromosome at this point in time



## **Y-Chromosome**

1. The "male" chromosome, analyzed for genealogy
2. All males are XY – The "Y" comes from the father, and his father, and his father, etc.
3. Y-chromosome: Testing specific segments of the non-functional portion

### **Timen Stiddem Society** (<http://homepages.rootsweb.com/~tstiddem/Pages/dna.html>)

1. Dr. Timen Stiddem, an early physician in Delaware with descendants
2. Four sons with male descendants, fifth son "daughtered out" in the 1800's
3. Y chromosome testing of 11 current 10th and 11th generation individuals
4. By genealogy, 5 of the 11 descended from 3 sons Lucas, Adam, Benedictus; the other 6 descended from Adam's grandson Adam
5. Results of DNA testing:
  - a. 5 descended from 3 sons Lucas, Adam, Benedictus, Y-chromosomes are all the same
  - b. 6 descended from Adam's grandson Adam- were found to have a different Y-chromosome from first 5 individuals (non-paternity event)

### **Non-paternity Event** (A supposed father and son found not to be biologically related)

1. Affairs outside of marriage
2. Bridal pregnancy where the groom is not the father
3. A son taking his stepfather's name
4. Unrecorded adoption
5. Improper conclusion from genealogical evidence

### **Beal Surname DNA Project** (<http://home.earthlink.net/~bealldna/project/overview.html>)

1. Y-chromosome 25 Markers results on 58 Beal's (5 Oct, 2002)
2. Match individuals with genealogical pedigrees; group individuals by test results

### **Understanding Y-Chromosome Results**

1. The more markers tested the better; consider 37 or 43 marker test
2. Comparing 2 individuals - 42/43 or better with good genealogy = probably identical ancestor, but don't know exactly who or when; there is a 50% chance that the most recent common ancestor was no longer than 4 generations

### **Mitochondrial DNA (mtDNA)**

1. DNA outside nucleus, a circular molecule of 16,569 bases sequentially numbered
2. Passed from mother to ALL children (for males: first generation only)
3. mtDNA establishes maternal line as only passed by a female to her children
4. Analyze the non-functioning control region, HVR1 and HVR2 (hypervariable regions 1 and 2); not gene containing area
5. Cambridge reference sequence (CRS): sequence of bases used as the standard
6. Individual mtDNA testing reports out where there is a difference in the tested sample and this reference sample DNA (CRS)

## mtDNA Uses

1. To assign daughters, born from different wives, to the correct mothers
2. To prove a female was adopted into a household
3. To determine if women of unknown parents or uncertain birth names were siblings
4. Confirm documented maternal lineage
5. Reveal a female line as originating from an ethnic group

## DNA Markers

1. Change in DNA length, STR ("Stirs") Short Tandem Repeats; used for Y chromosome analysis of "junk" DNA or non-functioning region of DNA molecule; a count of repeats of a short series of the same bases; a STR sequence can frequently be used to estimate haplogroup
2. Change in DNA sequence, SNP ("Snip") Single Nucleotide Polymorphism; a change in a single DNA base; used for mtDNA analysis and compared to CRS; used to determine both Y chromosome and mtDNA haplogroups
3. Haplotype: an individual's series of markers; STR's for Y chromosome; SNP's for mtDNA at HVR 1 & 2; this is the result an individual receives from a DNA test

## Ethnicity: Irish Modal Haplotype (IMH)

Amer. J. Human Genetics 78:334, 2006

1. One in 12 NW Irishmen descend from single early medieval man, Niall
2. "Niall of the Nine Hostages" kingship of Ireland from 7 to 11 century AD

## Haplogroups

1. Y chromosome & mtDNA originated thousands of years ago)
2. Both Y Chromosome & mtDNA haplogroups defined by SNP's

## Further Reading

On-Line Tutorials: <http://www.dnaheritage.com/tutorial1.asp>

[http://en.wikipedia.org/wiki/Genetic\\_genealogy](http://en.wikipedia.org/wiki/Genetic_genealogy) ; <http://www.kerchner.com/dna-info.htm>

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