DNA Testing and Genetic Genealogy

U3A Family History group presentation by Allan Ferrier and Peter Heilbrunn February 21st 2018

Genetic Genealogy Basics
SUMMARY of the TALK

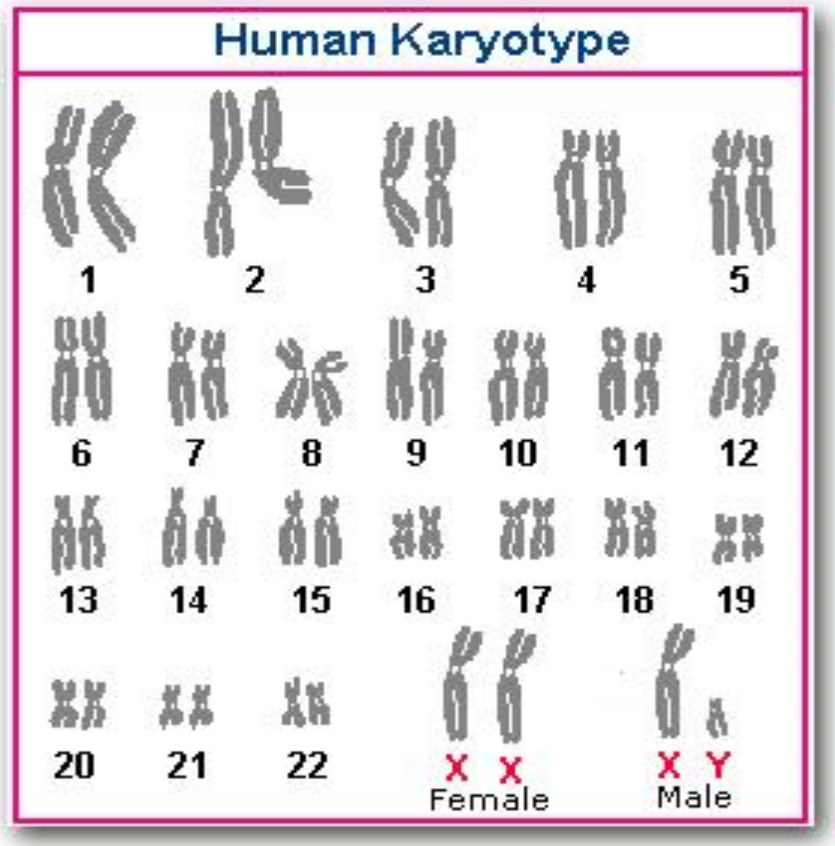
- What is DNA
- How do Genealogists use DNA
- What does DNA testing give you
- Who does DNA testing
- Autosomal testing and its limitations
- What is DNA matching
- Acknowledgements and resources
- On-line examples of testing results.

Genetic Genealogy Basics
What is DNA

- The cell, the basic unit of life, uses genetic material called DNA, to control the vast majority of its functions.
- A small percentage of DNA comprises genes, short segments of DNA that are used as blueprints to create a protein or an RNA molecule-
- A molecule of DNA is composed of a string of smaller units called nucleotides. These are the building blocks and come in 4 types that pair up in specific ways – A, C,G & T
- Together two intertwined molecules interact to form a single double helix structure called a chromosome
- A normal human cell has 92 long molecules of DNA that pair up to form 46 double stranded chromosomes.
- Each of these, in turn forms a chromosome pair with another similar but not identical chromosome to create 23

Genetic Genealogy Basics
What is DNA 2

- This is a human Karyogram arranged in number sequence from longest to shortest. To make a Karyogram, researchers stain chromosomes with a special chemical and then take a photograph. The chromosomes are then digitally rearranged into pairs and organised in to specific numbered sequence.
- Outside the cell's nucleus is the Mitochondrial DNA



Genetic Genealogy Basics
How do Genealogist use DNA

Four types of DNA are used for genetic genealogy: mtDNA, Y-DNA, atDNA and X-DNA

1. Mitochondrial DNA (mtDNA) is a small circular piece of DNA found in the cell's energy factory, the mitochondria. This is the only DNA not found in the cells nucleus. It is passed exclusively from mother to child. A mtDNA test reveals information about a person's direct maternal line.
2. Y-chromosomal DNA (Y-DNA) focuses on the unique male Y chromosome. This is passed exclusively from father to son
3. Autosomal DNA (atDNA) is composed of pairs of chromosomes found in the nucleus of the cell. Humans have 23 pairs of chromosomes of which 22 are autosomes. One copy of each chromosome is inherited from the mother and one copy from the father. Thus an atDNA test reveals information about both parental lines.
4. X-chromosomal DNA (X-DNA) focuses on the X chromosome, one of the 2 sex chromosomes that determine gender. Women have 2 X chromosomes, one from their father and one from the mother.; men have one X chromosome from their mother. For men, the X-DNA test reveals information about maternal lines. For women, it reveals information about both maternal & paternal lines.

Genetic Genealogy Basics
What does DNA testing give you

- **Relationship tests** – These tests are used to establish potential genetic connections between two or more people. This is a basic paternity/maternity test, but this group contains much more than that, including relations such as uncle/aunt, twin, grandparents, siblings, family reconstruction, male/female lineage, etc.
- **Ancestry tests** – They provide a (pre)historic picture of your genetic ancestry, allowing you to see regions around the world with the highest concentration of your DNA throughout history, look at genetic and ethnic maps, determine percentages of residue Neanderthal DNA in your genome, as well as identifying unknown relatives and connect with them if you wish.
- **Genomic tests** – These tests are used to determine potential "risk-carriers" within your genome; genes able to cause genetic disorders.. Many chronic diseases have a genetic component, so uncovering your family history can be important in this respect.

Genetic Genealogy Basics
Who does DNA testing

Within the past few years, millions of people around the world have taken a DNA test for genealogical purposes. There are now 4 major testing companies to choose from;
23and ME
Ancestry DNA
Family Tree DNA
My Heritage DNA
A good comparison of these companies can be found on
https://isogg.org/wiki/Autosomal_DNA_testing_comparison_chart

In addition there is a free service called GEDmatch
https://www.gedmatch.com/login1.php where you can upload your results from the above.

Genetic Genealogy Basics
Autosomal DNA testing

As explained earlier there are 4 test each with their own purpose.

The cheapest and most commonly offered is the **Autosomal test**.

Unlike the other 3 types, atDNA is inherited equally from both parents. Accordingly an individual gets one chromosome in each pair from Mum & Dad.

The amount of ancestral DNA reduces as one goes back a generation

Genetic Genealogy Basics
Limitations of Autosomal testing

Dilution of atDNA

- Parents 50%
- Grandparents 25%
- G Grandparents 12.5
- GG Grandparents 6.25

Odds of matching a relative in a DNA database

Relationship	Odds
1st Cousin or closer	100%
2nd Cousin	99.9%
3rd Cousin	Ca 90%
4th Cousin	Ca 50%
5th Cousin	Ca 15%

What is DNA Matching

Results – Not full Genome Sequencing about aprox 700k into SNPs which are know to vary between individuals

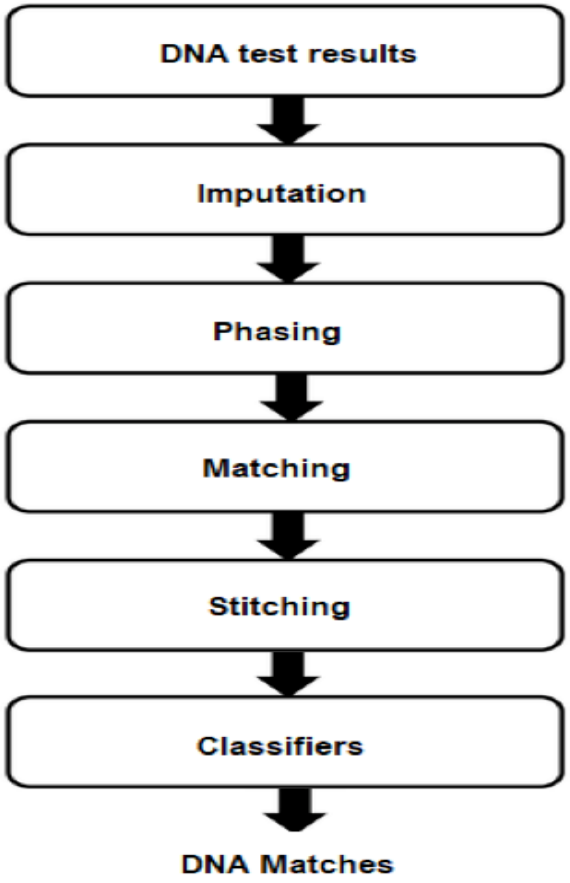
Imputation – Infers SNPs not read in Results

Phasing – Clusters all variants of each parent into separate buckets for each

Matching – Identifies shared segments between each pair of kits held on the database.

Stitching - Adjacent shared segments are the stitched on if they are considered contiguous.

Classifiers – These are statistical algorithms which review the matches, determine the level of confidence and suggests relationships



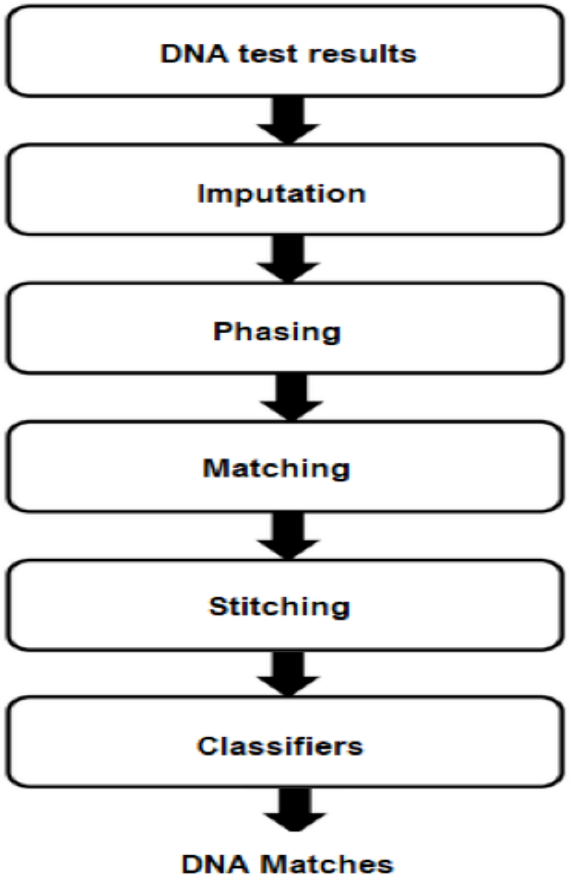
What is DNA Matching

This schematic shows the various stages involved as explained by My Heritage. For a full explanation go to

https://blog.myheritage.com/2018/01/major-updates-and-improvements-to-myheritage-dna-matching/

Additionally here is a video link

https://youtu.be/Z_806nvZF2o



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Sources and Acknowledgements

Acknowledgements

The Family Tree guide to DNA Testing and Genetic Genealogy by Blaine T Bettinger.

MyHeritage Blog https://blog.myheritage.com/

More Resources

Essential reading for genetic genealogists
www.isogg.org/wiki/Wiki_Welcome_Page

Autosomal –DNA Statistics
www.isogg.org/wiki/Autosomal_DNA_statistics

Autosomal webinar -
https://familytreewebinars.com/download.php?webinar_id=680

23and me Blog – https://blog.23andme.com/

Ancestry Blog – https://blogs.ancestry.com/ancestry/category/dna/