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Basic Genetic Genealogy for paper trail genealogists

George Valko your speaker...

15 year paper trail genealogist, 4 year genetic genealogist, published 5 family history books, group administrator of two genetic genealogy surname projects at Family Tree DNA, member of NGS, AHSGR, CSAGSI, ISOGG. George's main areas of research are Volga Germans and Slovaks who came to Chicago.

What is genetic genealogy...

A useful tool to be used in addition to paper trail genealogy.

Traces everyone's maternal line back thousands of years via mtDNA

Traces men's paternal line back thousands of years via yDNA

Is perfect for determine who you are NOT related to, genetically.

Suggests or confirms a common ancestor; but NOT which common ancestor and suggests a time frame with probability (time frame) for the common ancestor.

DNA test... (for both yDNA & mtDNA)

Buccal cheek scrape; no pain, no blood, no needles; multi-samples.

Genetic genealogy tests and tests for health purposes and criminal cases ARE DIFFERENT TESTS.

Test results can be private.

Be prepared for surprises; i.e. no genetic match OR unexpected genetic matches.

<http://www.familytreedna.com/glossary.html#P>

A

Aboriginal = Relating to a group of people indigenous to a geographic region; the original inhabitants of a region.

Adenine = The "A" of the four bases that make up DNA. The other bases are thymine (T), guanine (G) and cytosine (C). Adenine always pairs with thymine.

Admixture = Of mixed ancestry or mixed origins.

Allele

A DNA sequence that repeats at a certain locus or place. The allele value is the number of times the sequence repeats. Pronounced uh-LEEL.

Amplification

See **DNA amplification**.

Ancestral signature

The oldest known or hypothesized haplotype for a particular lineage. See also: **modal haplotype**.

Anthrogenealogy

The study of human origins, recent and distant, using DNA testing and genealogical methods.

Anthropology

The study of humans, particularly in terms of origin and culture.

Atlantic Modal Haplotype (AMH)

See **Western Atlantic Modal Haplotype**.

Autosomal DNA

The non-sex chromosomes. Humans have 23 pairs of chromosomes: the first 22 pairs are autosomal DNA and the 23rd pair consists of the sex chromosomes (the X- and Y- chromosomes).

B**Base**

The unit or building block of DNA. Adenine (A), cytosine (C), guanine (G), and thymine (T) are the four bases in DNA. The order of bases is the sequence of DNA.

Base pair

Two bases bonded together and attached to one of the strands in the DNA double helix. Adenine always pairs with thymine, and guanine always pairs with cytosine.

Buccal cell

A type of cell found in cheek tissue inside the mouth.

C**Cambridge Reference Sequence (CRS)**

The mitochondrial DNA first sequenced in 1981 which is used as a basis for comparison with mtDNA test results.

Catalyst

A substance which starts or speeds up a chemical reaction without being affected by that reaction.

Chromosome

A structure found in the nucleus of a cell that contains genetic material. Humans have 23 pairs of chromosomes; 22 pairs of autosomes and one pair of sex chromosomes.

Coding region

A region of DNA which contains genes.

Cohanim Modal Haplotype

The Y-DNA haplotype most commonly found among males with an oral tradition of Cohen ancestry.

Cohen

The Hebrew word for priest which refers to a direct male descendant of Aaron, the brother of Moses; plural: Cohanim.

Combined DNA Index System (CODIS)

The CODIS system uses marker locations in the autosomal DNA. CODIS test results are maintained in an FBI database which is used to identify people and solve crimes.

Complementary sequences

Opposing strands of DNA which bond together to form the double helix. The bases always complement one another with adenine and thymine pairing together and cytosine and guanine pairing together.

Convergence

The process of two unrelated or less related lineages changing over time to resemble one another.

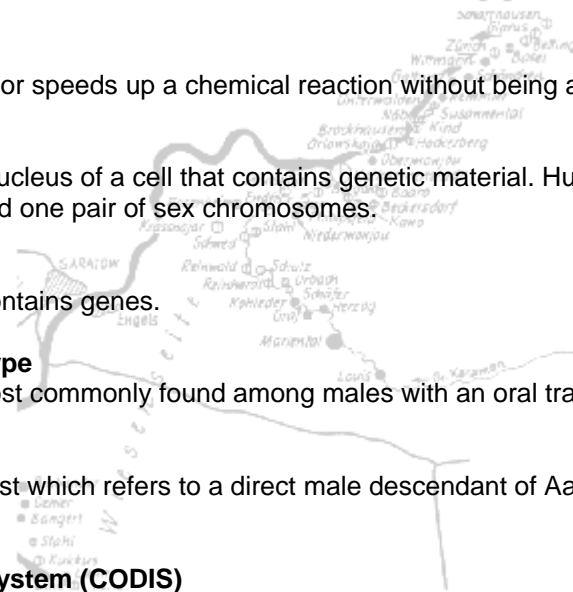
Cross-over

See **recombination**.

Cytosine

The "C" of the four bases that make up DNA. The other bases are adenine (A), guanine (G), and thymine (T). Cytosine always pairs with guanine.

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are no differences in the results being compared against one another (exact match).

Genetics

The study of genes and heredity; the study of DNA.

Genome

The entire complement of genetic material in a chromosome set. The human genome is composed of 46 chromosomes, with a total of 3 billion base pairs.

Genotype

The genetic makeup of an individual organism.

Glacial maximum

The scientific term for the peak of an ice age.

Guanine

The "G" of the four bases that make up DNA. The other bases are adenine (A), cytosine (C), and thymine (T). Guanine always pairs with cytosine.

H

Haplogroup

A genetic population group associated with early human migrations and which can today be associated with a geographic region. It is important to note that even though female and male haplogroups may have the same letters, their definitions are different.

Haplotype

One person's set of values for the markers that have been tested. Two individuals that match exactly on all markers have the same haplotype.

Heredity

The transmission of genetic material from parents to offspring.

Human Genome Organization (HUGO)

The entity to which, among other things, scientists submit new markers for DYS number assignment.

Hypervariable Region (HVR)

One of two regions commonly used in mitochondrial DNA tests which does not code for any known function.

J

Junk DNA

Stretches of DNA that do not code for genes, also called non-coding DNA. Most of the genome consists of non-coding DNA, and was therefore long thought to be 'junk.' Scientists have found that in addition to containing markers that are helpful for genetic genealogy, parts of these non-coding regions have regulatory and other functions.

L

Loci

The plural of **locus**, or marker. Locus and loci are Latin words for location(s).

M

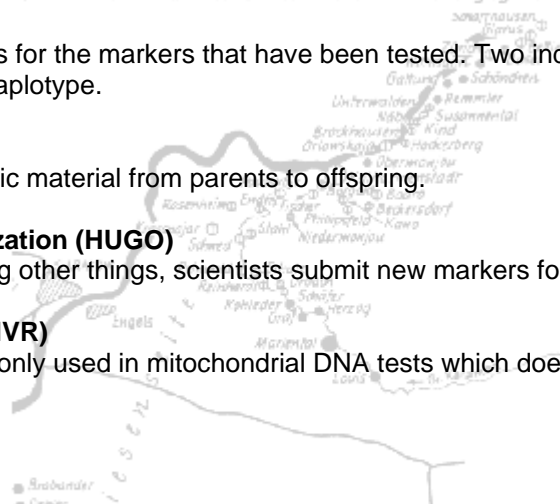
Marker

A physical location (locus) on the chromosome. Family Tree DNA offers 3 levels of Y-DNA testing: 12-marker, 25-marker and 37-marker.

Meiosis

The stage in which sperm and egg cells are formed. It is during this process that the autosomal chromosomes recombine and mutations occur.

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and the other from the father break and trade segments with one another.

Replication

See **DNA replication**.

Restriction enzyme

A protein that recognizes a certain sequence of DNA and cuts the DNA at that site.

Restriction Fragment Length Polymorphism (RFLP)

See **single nucleotide polymorphism**.

S

Sequencing

See **DNA sequencing**.

Sex chromosome

The X- or Y-chromosome. Normally males have one X and one Y and females have two Xs.

Short Tandem Repeat (STR)

A short DNA motif (pattern) repeated in tandem. ATGC repeated eleven times would give the marker a value or allele of 11.

Single Nucleotide Polymorphism (SNP)

A change in the DNA that happens when a single nucleotide (A, T, G, or C) in the genome sequence is altered. A person has many SNPs that together create a unique DNA pattern for that individual.

Surname

A last name or family name traditionally passed down from father to son.

T

Thymine

The "T" of the four bases that make up DNA. The other bases are adenine (A), cytosine (C), and guanine (G). Thymine always pairs with adenine.

Time to the Most Recent Common Ancestor (TMRCA)

The amount of time or number of generations since individuals have shared a common ancestor. Since mutations occur at random, the estimate of the TMRCA is not an exact number (i.e., 7 generations), but rather a probability distribution. As more information is compared, the TMRCA estimate becomes more refined.

Transmission event

The passage of genetic material from one generation to the next.

U

Unique Event Polymorphism (UEP)

See **single nucleotide polymorphism**.

W

Western Atlantic Modal Haplotype (WAMH)

The most common Y-DNA haplotypes found in Europe's most common Y-DNA haplogroup, R1b.

X

X-chromosome

One of the two sex chromosomes, X and Y. X is the sex chromosome that is present in both sexes: singly in males and doubly in females.

Y

Y-chromosome

One of the two sex chromosomes, X and Y. The Y-chromosome passes down from father to son. Females do not receive it. The fact that the Y-chromosome goes down the paternal line makes it valuable for genealogy studies, since it typically follows a surname line.

Y-DNA phylogenetic tree

A graphic representation of the Y-DNA haplogroups according to the YCC classification. Haplogroup names and major clades are labeled and mutation names are given along the branches of the trees.

<http://www.isogg.org/course/glossary.htm>

A

Administrator - Also known as a 'Project Administrator', 'Group Project Administrator', 'Project Manager', 'Coordinator' and 'Co-Coordinator'. A volunteer who establishes a DNA study with one or multiple commercial DNA testing companies.

Admixture DNA - The non-gender chromosomes that mix or recombine. Also known as [autosomal DNA](#).

Allele - (pronounced *UH-lee*) - The scientific result for a [marker](#).

Ancestral haplotype - The method of deducing the MRCA's [haplotype](#) by comparing matching descendants' haplotypes, and eliminating the [mutations](#). A minimum of three lines, as distantly related as possible, is recommended for deducing the ancestral haplotype.

Ancestral state - Refers to the state of a [SNP](#) that has [mutated](#) and is shared by the most people. Example: A negative result on a SNP means it is *ancestral*, a positive result means it is [derived](#).

atDNA - Acronym for [Autosomal DNA](#).

Autosomal DNA - The non-gender chromosomes that mix or recombine. Also known as admixture DNA.

Anthrogenealogy - A term coined by Family Tree DNA combining the words 'anthro' and 'genealogy' in reference to utilizing DNA to trace one's heritage far beyond recorded documentation.

C

CODIS - Acronym for **C**ombined **D**NA **I**ndex **S**ystem - The FBI's [autosomal](#) DNA database for profiles of criminal offenders.

<http://www.fbi.gov/hq/lab/codis/index1.htm>

CRS - Acronym for **C**ambridge **R**eference **S**equences. The first mitochondrial DNA to be fully sequenced at Cambridge University in 1981. The anonymous donor of the CRS was [haplogroup](#) 'H'. Mitochondrial results are determined based upon where the results differ from the CRS.

D

Derived state - Refers to the state of a [SNP](#) that has mutated, usually in one man, from the [ancestral state](#) and created a new [haplogroup](#) or [sub-clade](#) of a haplogroup. A positive SNP result is derived, a negative SNP result is [ancestral](#).

DNAF - Acronym for **D**NA-**F**ingerprint - a commercial DNA testing company.

DNAH - Acronym for **D**NA **H**eritage - a commercial DNA testing company.

DNA-NEWBIE - A *DNA-Newbie* is someone who is new to the field of genetic genealogy. It is also the name of a Yahoo mailing list forum sponsored by the International Society of Genetic Genealogy.

<http://groups.yahoo.com/group/DNA-NEWBIE/>

DYS - Acronym for **D**NA **Y**-chromosome **S**egment - The assigned number of a marker on a segment of the Y-chromosome. Example: DYS# 393

FTDNATiP - Acronym for **F**amily **T**ree **DNA** **T**ime **P**redictor - A program created to calculate the time to the [MRCA](#) using mutation rates specific to each marker.

E

EA - Acronym for **E**thno**A**ncestry - a commercial DNA testing company.

F

FTDNA - Acronym for **F**amily **T**ree **DNA** - a commercial DNA testing company.

G

GAP - Acronym for the **G**roup **A**dministrator **P**age - This is a webpage in which a DNA Project Administrator utilizes functions such as creating a public website, generating a FTDNATiP report, etc. to assist project participants in coordinating results.

GEDCOM - Acronym for **G**enealogical **D**ata **C**ommunications - A plain text program created for exchanging genealogical data between different genealogical programs. Family Tree DNA's 'My FTDNA' page, [Y-Search](#) and [Mitosearch](#) all contain a feature to upload a GEDCOM for pedigree comparisons to matches.

Genetic Genealogy - The latest tool for genealogists utilizing DNA to aid genealogical research.

Genographic Project - (pronounced *GENE-o-graphic*) - A five-year scientific genetics project launched in 2005 to study and map human migratory patterns. Along with testing indigenous populations, the project offers a 12-marker Y-chromosome or HVR1 mtDNA test for public participation with a portion of the proceeds benefiting the indigenous communities. The project is sponsored by National Geographic, IBM and the Waitt Foundation. Genographic participants have the option to join Family Tree DNA's database once their DNA results are in.

H

Haplogroup - A group of similar haplotypes that share a common ancestor with a [SNP](#) mutation. Because a haplogroup consists of similar haplotypes, this is what makes it possible to predict a haplogroup. A SNP test confirms a haplogroup. Haplogroups are assigned letters of the alphabet, and refinements consist of additional number and letter combinations, **Example:** R1b1. Y-chromosome and mitochondrial DNA haplogroups have different haplogroup designations. Haplogroups pertain to your deep ancestral origins dating back thousands of years.

Haplotype - The term for the set of numbers that consists of your [Y-chromosome](#) or [mitochondrial](#) DNA results. Haplotypes are also known as signatures.

HVR - Acronym for **H**yper **V**ariable **R**egion - The sections of [non-coding mitochondrial](#) DNA that are used for genealogical DNA testing.

I

ISOGG - Acronym for the **I**nternational **S**ociety **o**f **G**enetic **G**enealogy, a free society founded in 2005 for the promotion and education of genetic genealogy.

J

Junk DNA - Slang term usually used in referring to the [non-coding](#) region of DNA on the [Y-chromosome](#). For more about: <http://www.psrast.org/junkdna.htm>

JoGG - The **J**ournal **o**f **G**enetic **G**enealogy - An online journal published quarterly with articles and features pertaining to [genetic genealogy](#) and [anthrogenealogy](#).
<http://www.jogg.info/>

M

Marker - A type of [non-coding](#) Y-chromosome DNA. The numbers designating your individual DNA segment numbers you receive with [Y-chromosome](#) DNA results. Example: 393=13. This means at marker #393, your [allele](#) value is 13.

Mitochondrial DNA - Energy releasing [organelles](#) located in the cytoplasm of cells, which contain their own DNA. Mitochondrial DNA is passed from mother to child, but only females continue to pass on their maternal mitochondria to their children.

Mitosearch - A free public database sponsored by Family Tree DNA where [mitochondrial](#) DNA results from any testing facility may be uploaded and compared.
<http://www.mitosearch.org/>

MRCA - Acronym for **M**ost **R**ecent **C**ommon **A**ncessor.

mtDNA - Acronym for [mitochondrial DNA](#).

Mutation - A change in the DNA that occurs at random. *Mutation* is a scientific term that often connotes a negative connotation as a result of 1950's 'B' movies, but in [genetic genealogy](#), a mutation is usually beneficial. Mutations on the [Y-chromosome](#) are often used for distinguishing different ancestral lines.

N

Non-coding DNA - Also referred to as "[Junk DNA](#)", non-coding DNA has no known biological function.

Nuclear DNA - DNA of chromosomes found in the nucleus of the cell.

Null - A *null* is a value of zero on a marker. Nulls can occur due to missing genetic material on a marker, or a [SNP](#) can sometimes cause a null result. Several [YSTR](#) markers have been identified in certain families to have null results: [439](#), [448](#)

O

OA - Acronym for **O**xford **A**ncestors - a commercial DNA testing company.

Organelle - A cell structure with specialized functions.

<http://en.wikipedia.org/wiki/Organelle>

P

Phylo-tree - Shortened term for **P**hylogenetic **T**ree - Most often used in reference to the available online diagrams connecting all [Y-chromosome haplogroups](#). This term is also applied to DNA project diagrams created by [Project Administrators](#) utilizing specialized software.

R

RAO - Acronym for **R**ecent **A**ncentral **O**rigins (formerly known as **REO** - Recent Ethnic Origins)

The number of matches you have in Family Tree DNA's database as specified by country or region. To access the RAO, Family Tree DNA clients need to click the tab on their "My FTDNA" page.

recLOH - Acronym for **R**ecombinant **L**oss of **H**eterozygosity - When a section of DNA on a marker is missing, that marker is sometimes repaired by another marker filling in the missing DNA with its own material. This is referred to as a "recLOH event" and is usually observed with multi-copy markers like 385a and 385b, and is also common in the 464 set. The recLOH event causes the allele values to match 11-11 instead of the more common, 11-14 that you see in R1b. For more about: [Recombinational Loss of Heterozygosity \(recLOH\)](#)

RG - Acronym for **R**elative **G**enetics - a commercial DNA testing company.

S

SMGF - Acronym for **S**orensen **M**olecular **G**enealogy **F**oundation. Established by James Sorensen as a scientific genealogical DNA database, participants submit a DNA sample along with a four generation pedigree chart. While participation is free, SMGF does not send participants their results. However, many participants are able to "find" themselves in the online results database by matching up their pedigrees. Currently, only [Y-chromosome](#) results appear in the database, and the current wait time for results to appear is between nine months to over two years.

<http://www.smgf.org>

SNP - (pronounced *SNIP*) - Acronym for **S**ingle **N**ucleotide **P**olymorphism. A SNP test confirms your [haplogroup](#) by determining if a SNP has [mutated](#) from its [derived](#) or [ancestral](#) state. A SNP is usually found on a different area of the [Y-chromosome](#) than where the [YSTR](#) markers are. Sometimes, a SNP may cause a [null](#) result on a [marker](#).

STR - Acronym for **S**hort **T**andem **R**epeat - See: [YSTR](#)

Sub-clade - Referring to a "branch" farther down the [phylogenetic tree](#). Example: H3 -> '3' is a sub-clade of [mitochondrial haplogroup](#) 'H'. R1b -> '1b' is a sub-clade of [Y-chromosome](#) haplogroup 'R'. Sub-clade testing is also referred to as *deep clade* testing.

I

TG - Acronym for **T**race **G**enetics - a commercial DNA testing company.

W

Whit's Predictor - The commonly applied nickname to the "Y-Haplogroup Predictor" created by Whit Athey. Enter [Y-chromosome](#) markers into the predictor and it will display percentages for matches to various haplogroups.

<http://www.hprg.com/hapest5/>

X

X-chromosome - The female gender chromosome, if a child receives one X from the father and one X from the mother, the child's gender is female.

Y

Y-chromosome - The male gender chromosome. In other words, only males have a Y-chromosome, which they receive from their father, who received it from his father, and so on. This transmission of the Y-chromosome down the male line is why it is useful for surname testing to determine if two males share a common ancestor.

Y-Base - A free public database sponsored by [DNA Heritage](#) where Y-chromosome DNA results from any testing facility may be uploaded and compared.

<http://www.ybase.org/>

Y-Search - A free public database sponsored by [Family Tree DNA](#) where Y-chromosome DNA results from any testing facility may be uploaded and compared.

<http://www.ysearch.org/>

YSTR - Acronym for **Y**-chromosome **S**hort **T**andem **R**epeat. The number of times the bases repeat that determines the value of the marker. Example: Thirteen repeats of the same bases equals a value of '13'.

<http://www.geneticgenealogydictionary.com/>

Chimera - /chi•me•ra/ - An organism containing two or more genetically distinct cell or tissue lines, i.e., an organism having two or more distinct populations of cells with different nuclear DNA. This can occur from two different egg cells being fertilized by two different sperm cells forming two zygotes with subsequent fusion of the two zygotes and development of a single organism with intermingled cells with different nuclear DNA in various parts of the organism. Compare to Heteroplasmy

Heteroplasmy - /het•er•o•plas•my/ - The presence of a mixture of more than one type of mitochondrial DNA (mtDNA) within a cell's cytoplasm (the plasma material inside the cell but outside the nucleus). Cells contain many hundreds or even thousands of mitochondria organelles with each organelle containing its own mtDNA. Thus it is possible for mutations to affect only some of the copies of the mitochondria, while the remaining ones have the prior, un-mutated type of mtDNA. Unless the population of mutated mtDNA is a significant percentage of the total mtDNA in the cell, the mtDNA maternal line DNA test would detect the dominant population's mtDNA markers. Compare to Chimera.

Glossary Websites:

ISOGG's Y-SNP Tree Glossary of Genetics Terms

<https://www3.nationalgeographic.com/genographic/index.html>

Kerchner's Genetic Genealogy page

<http://www.kerchner.com/table.htm>

National Geographic Genographic Project Genetics Glossary

<https://www3.nationalgeographic.com/genographic/glossary.html>

Family Tree DNA Glossary

<http://www.familytreedna.com/glossary.html>

Genetics/DNA Glossary - Clan Lindsay

http://www.clanlindsay.com/genetic_dna_glossary.htm

DNA Heritage Glossary

<http://www.dnaheritage.com/glossary.asp>

The "Out of Africa Theory"...

Human migration for the past 150,000 years, population expansion, contraction and bottle necks due to ice ages, volcanoes and other natural events.

Genetic genealogy blurs the concepts of ...

Race
Ethnicity
Culture
Family

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FTDNA & other databases...

Over represented for Europeans; England, Scotland, Ireland, Germany.

Under represented for the poor, Asians, Africans, Middle Easterners, Eastern Europe, etc.

Resources...

Genetic Genealogy "genetealogy" testing labs,

African Ancestry <http://www.africanancestry.com/>

Offers Y-DNA and mtDNA tests and compares your results against a proprietary database of African samples. Frequently able to provide a tribal or geographic origin within Africa.

DNAHeritage <http://www.dnaheritage.com/>

UK-based firm which offers Y-DNA tests of up to 43 markers, as well as mtDNA tests. Also offers a customized, per-marker option and sponsors Ybase.org.

DNAPrint Genomics <http://www.dnaprint.com/welcome/home/index.php>

Offers tests that break out your origins in broad, geographic categories (e.g., Indo-European, Native American, etc.). Now offering a more detailed European-centered test.

EthnoAncestry was founded by Dr. James F. Wilson, leading authority on European genetic history. "The Origin of the British" clans. Which one do you belong to?

<http://www.ethnoancestry.com/>

Family Tree DNA <http://www.familytreedna.com/>

Offers a wide variety of Y-DNA, mtDNA and ethnic-oriented tests, as well as handy management tools that make the project manager's job easier. Also sponsor of Ysearch.org.

Gene Tree <http://www.genetree.com/>

Not a genetic genealogy company per se, but offers an interesting collection of "close kin" tests (e.g., siblingship, grandparentage, etc.) for those who run into brick walls early in their research.

GeoGene <http://www.geogene.com/lowres/home.html>

UK-based firm seemingly focused on deep ancestry for both Y-DNA and mtDNA.

Oxford Ancestors <http://www.oxfordancestors.com/> (Bryan Sykes)

UK-based firm that was the first of the genetic genealogy companies to launch. Offers both Y-DNA and mtDNA tests, as well as some colorful ethnic-oriented ones.

Ancestry.com <http://dna.ancestry.com/welcome.aspx>

Roots for Real <http://www.rootsforreal.com/>

A UK-based firm focused on mtDNA.

Bibliography / suggested reading list

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* Wells, Spencer, **Deep Ancestry, Inside the Genographic Project**, ISBN: 0-792262158, Publisher: National Geographic Society, 2006

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 National Genealogical Society Quarterly, Volume 93, No. 4, December 2005, Genealogy and Genetics

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<http://www7.nationalgeographic.com/ngm/0603/feature2/index.html> Geographic Project
<https://www3.nationalgeographic.com/genographic/> National Geographic Genographic Project
<http://www.isogg.org/> International Society of Genetic Genealogists - ISOGG
<http://groups.yahoo.com/group/ISOGG/files/> ISOGG files downloads
<http://www.genetealogy.com/> Megan Smolenyak Smolenyak
<http://www.worldfamilies.net/> World Families
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<https://www.23andme.com/> autosomal DNA
<http://www.decodeme.com/> autosomal DNA
<http://www.navigenics.com/> autosomal DNA

Discussion groups,

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