

DNA

Deoxyribonucleic acid (DNA) is the chemical inside the nucleus of all cells that carries the genetic instructions for making living organisms. A DNA molecule consists of two strands that wrap around each other to resemble a twisted ladder. The sides are made of sugar and phosphate molecules. The "rungs" are made of nitrogen-containing chemicals called **bases**. Each strand is composed of one sugar molecule, one phosphate molecule, and a **base**. Four different bases are present in DNA - **adenine** (A), **thymine** (T), **cytosine** (C), and **guanine** (G). The particular order of the bases arranged along the sugar - phosphate backbone is called the DNA sequence; the sequence specifies the exact genetic instructions required to create a particular organism with its own unique traits.

Each strand of the DNA molecule is held together at its **base** by a weak bond. The four **bases** pair in a set manner: Adenine (A) pairs with thymine (T), while cytosine (C) pairs with guanine (G). These pairs of bases are known as **Base Pairs (bp)**.

These Base Pairs (bp) are the basis of Y-chromosome testing.¹

Chromosomes

Chromosomes are paired threadlike "packages" of long segments of DNA contained within the nucleus of each cell. In humans there are 23 pairs of chromosomes. In 22 pairs, both members are essentially identical, one deriving from the individual's mother, the other from the father. The 23rd pair is different. In females this pair has two like chromosomes called "X". In males it comprises one "X" and one "Y," two very dissimilar chromosomes. It is these chromosome differences which determine sex.

The Y-Chromosome

Human sex is determined by the X and Y chromosomes. A female has 2 X-Chromosomes and a male has an X and a Y-Chromosome. When a child is conceived it gets one chromosome from its mother and one chromosome from its father. The chromosome from the mother will always be an X, but the chromosome from the father may be either X or Y. If the child gets the X she will be a girl, if the child gets the Y he will be a boy.

This Y-Chromosome has certain unique features:

- The presence of a Y-Chromosome causes maleness. This little chromosome, about 2% of a father's genetic contribution to his sons, programs the early embryo to develop as a male.
- It is transmitted from fathers only to their sons.
- Most of the Y-Chromosome is inherited as an integral unit passed without alteration from father to sons, and to their sons, and so on, unaffected by exchange or any other influence of the X-Chromosome that came from the mother. It is the only nuclear chromosome that escapes the continual reshuffling of parental genes during the process of sex cell production.

It is these unique features that make the Y-Chromosome useful to genealogists.

Testing the Y-Chromosome

The Y-Chromosome has definable segments of DNA with known genetic characteristics. These segments are known as **Markers**. These markers occur at an identifiable physical location on a chromosome known as a **Locus**. Each marker is designated by a number (known as **DYS#**), according to international conventions. You will often find the terms **Marker** and **Locus** used interchangeably, but technically the **Marker** is what is tested and the **Locus** is where the marker is located on the chromosome.

¹ <http://blairdna.com/dna101.html>

Although there are several types of markers used in DNA studies, the Y-Chromosome test uses only one type. The marker used is called a **Short Tandem Repeat (STR)**. STRs are short sequences of DNA, (usually 2, 3, 4, or 5 base pairs long), that are repeated numerous times in a head-tail manner. The 16 base pair sequence of "gatagatagatagata" would represent 4 repeats of the sequence "gata". These repeats are referred to as **Allele**. The variation of the number of repeats of each marker enables discrimination between individuals.²

Tracking your Maternal Line with MtDNA

When we speak casually of DNA, most often we are thinking about the DNA that is found in the 46 chromosomes, inside the nucleus of the cell. The remainder of the cell contents-the cytoplasm-harbors DNA as well, inside small structures called mitochondria, the plural form of mitochondrion. About the size of bacteria, hundreds or even thousands of mitochondria are crammed into one cell. In fact, the very size is a clue to the origins.³

It is from the mitochondrion part of the cell where DNA is drawn to trace the maternal line. Where did the mitochondrion come from? Your mother-----who got it from her mother-----who got it from her mother until the beginning of the first woman on Earth-----**Mitochondrial EVE**.

Curious about Genealogy and Anthropology-FamilyTreeDNA Founder Bennett Greenspan

FamilyTreeDNA Website: <http://www.familytreedna.com/Default.aspx?c=1>

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Genealogist Dr. Spencer Wells talks about Humans Genetics

Spencer Wells is a leading population geneticist and director of the **Genographic Project from National Geographic and IBM**. His fascination with the past has led the scientist, author, and documentary filmmaker to the farthest reaches of the globe in search of human populations who hold the history of humankind in their DNA. By studying humankind's family tree he hopes to close the gaps in our knowledge of human migration.⁴

The Genographic Project Website:

<https://genographic.nationalgeographic.com/genographic/index.html?fs=www5.nationalgeographic.com>

<http://www.youtube.com/watch?v=lxgS9RKwKbU&NR=1>

Reasons to Test for DNA

- Determine if two people are related.
- Determine if two people are descended from a common ancestor.
- Confirm your family tree.
- Find out who with your surname is related.
- Prove or disprove a research theory.
- Find and confirm new individuals in your family tree.

² <http://blairdna.com/dna101.html>

³ Megan Smolenyak Smolenyak and Ann Turner *Trace Your Roots with DNA: Using Genetic Tests to Explore Your Family Tree*, Holtzbrinck Publishers, 2004, pg. 59.

⁴ <http://www.nationalgeographic.com/field/explorers/spencer-wells.html>

Definitions

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).

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.

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.

Deoxyribonucleic acid (DNA)

A chemical consisting of a sequence of hundreds of millions of nucleotides found in the nuclei of cells. It contains the genetic information about an individual and is shaped like a double-stranded helix.

DNA sequencing

The process of determining the exact order of the nucleotide bases in a segment of DNA.

Double helix

The twisted shape DNA forms when its two strands bond together. A double helix looks like a twisting or rotating ladder.

DNA Y-chromosome Segment (DYS)

The "name" of a marker on the Y-chromosome. It is assigned based on a nomenclature system controlled by the HUGO Gene Nomenclature Committee, which assigns DYS numbers to newly discovered markers.

Generation

The number of years between the birth of the parents and the birth of their children. Different studies use different numbers of years per generation. At Family Tree DNA we use 25 years.

Genetic distance

The number of differences, or mutations, between two sets of results. A genetic distance of zero means there are no differences in the results being compared against one another (exact match).

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.

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.

Mitochondria

A specific organelle in the cell that helps it to produce energy.

Mitochondrial DNA (mtDNA)

The genetic material found in mitochondria. It is passed down from females to both sons and daughters, but sons do not pass down their mother's mtDNA to their children.

Most Common Recent Ancestor

The ancestor shared most recently between two individuals.

Mutation

A heritable change that occurs in genetic material. It may lead to a different number of repeats of a certain sequence or a change in one of the bases in a sequence.

Mutation rate

The frequency with which random mutations occur

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.

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-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.⁵

⁵ <http://www.familytreedna.com/glossary.aspx#H>