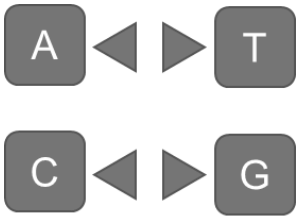
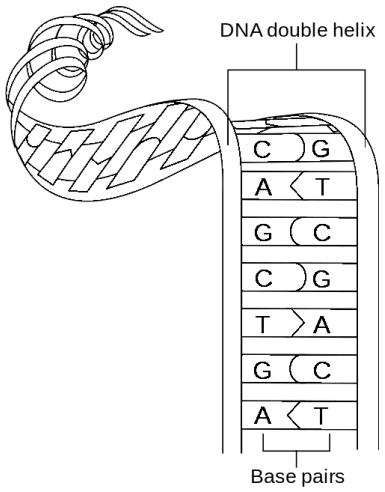
**Through the Generations: Autosomal DNA Inheritance**
New England Regional Genealogical Conference
5 April 2019

## Basic Human Genetics

- Human DNA is composed of approximately 3 billion base pairs, also known as *single nucleotide polymorphisms,* or *SNPs* (pronounced "snips"). Base pairs are bonds between chemicals labeled A, C, G, and T. The chemicals pair as follows:




*Source:* "Diagram Showing a Double Helix of a Chromosome," Cancer Research UK, *Wikimedia Commons* (https://commons.wikimedia.org/wiki/File:Diagram_showing_a_double_helix_of_a_chromosome_CRUK_065.svg : accessed 4 January 2018). Used under license.

- All 3 billion base pairs are organized into packages of DNA called *chromosomes*.
- Humans have 23 pairs of chromosomes in each cell. One pair determines the sex of an individual. The 22 remaining pairs of non-sex chromosomes are called autosomes.
- Sperm and egg cells undergo a process called *recombination*, or *crossing-over*, which results in a shuffling of genetic material between the maternal and paternal chromosome pairs.
- My grandmother and I share a segment on chromosome 1 that spans around 66 megabases. We also share a segment on chromosome 12 of about 66 megabases. However, the segment on chromosome 12 has a relatively higher recombination rate compared to chromosome 1. [See **Figure A**]
- The chromosome 12 segment has a higher probability of being broken during crossing over, and the chromosome 1 segment is more likely to remain intact.
- As a result, a matching segment on chromosome 12 indicates a closer relationship between the two individuals than a segment of comparable length on chromosome 1. This genetic distance is measured in *centiMorgans* (cM). [See **Figure B**]

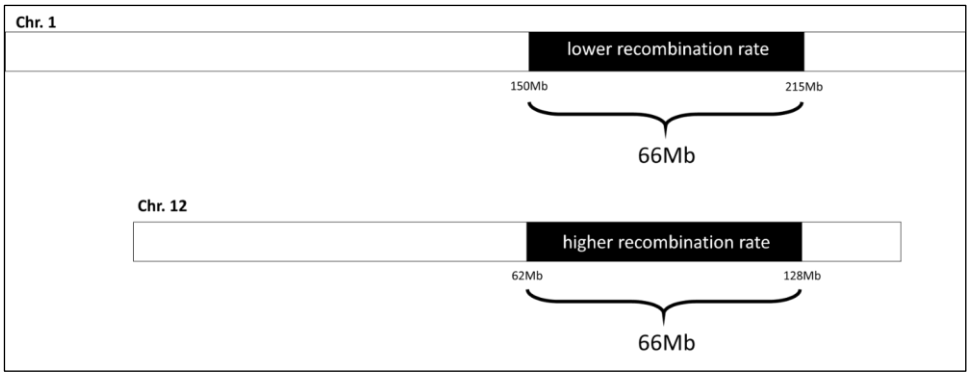



**Figure A**

- CentiMorgans are a unit of recombinant frequency, which means that they take into account how frequently recombination, or crossing over, occurs in a region. The centiMorgan is equal to a 1% chance that a marker at one genetic locus on a chromosome will be separated from a marker at a second locus due to crossing over in a single generation.

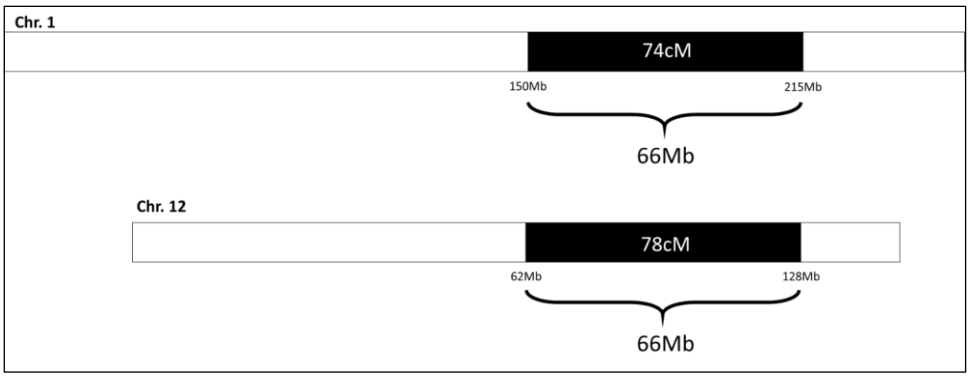



**Figure B**

- The shared cM totals per chromosome provided by a DNA testing company or third-party such as GEDmatch are calculated based on the Human Reference Genome.

**Direct-to-Consumer Tests**

- Humans are over 99% genetically identical. To accomplish their DNA analysis, direct-to-consumer testing companies don't test all 3 billion SNPs. They test somewhere between 700,000 to 1 million SNPs which they've determined are unique to individuals.
- The five major companies (23andMe, AncestryDNA, Family Tree DNA, LivingDNA, MyHeritageDNA) allow users to download a file containing their result at each tested SNP.

**DNA Matches**

- Analyze the segments shared with your DNA matches.
- Locate places along your chromosomes where you have multiple DNA matches in one spot.
- Check to see that all the individuals who you match on any given segment also match one another on that segment. This is called *triangulation* and confirms that you all inherited that segment from the same common ancestral couple.
- If possible, testing another close family member (besides a sibling) will help you identify whether your DNA matches are on the paternal or maternal chromosomes.
- When you identify a set of triangulated matches, compare their family trees (pedigrees) to see if they share any ancestors in common.

## Glossary of Terms

- **autosome**: a non-sex chromosome (humans have 22 pairs of autosomes in each cell)
- **base pair**: bond formed between two nucleotide bases
- **centimorgan** *or* **cM**: a unit of recombinant frequency which relates the physical length of a DNA segment (number of base pairs) to the chromosome's tendency to recombine at that location
- **chromosome**: an organized package of DNA in the nucleus of the cell (humans have 23 pairs of chromosomes in each cell)
- **crossing-over**: exchange of genetic material between maternal and paternal chromosome pairs; also known as *recombination*

- **DNA** *or* **deoxyribonucleic acid**: a double chain of linked nucleotides
- **DTC**: direct-to-consumer
- **endogamy**: intermarriage within a population
- **GPS**: Genealogical Proof Standard
- **megabase**: unit representing 1 million base pairs
- **meiosis**: biological process where chromosomes duplicate, exchange information (recombine), and divide into gametes (sperm or egg cells)
- **MRCA**: most recent common ancestor
- **nucleotide**: the molecules which link together to form DNA (adenine, cytosine, guanine, and thymine)
- **recombination**: exchange of genetic material between maternal and paternal chromosome pairs; also known as *crossing-over*
- **RSID**: a universal reference number assigned to individual SNPs
- **triangulation:** verification that all individuals that match the target kit on any given segment also match one another on that segment; confirms all individuals inherited that segment from the same common ancestral couple

## Selected Resources


Beckins, LLC. *Genome Mate Pro.* Release 2019.04. Software program. https://www.getgmp.com/ : 2019.

Bettinger, Blaine T. "Q&A: Everyone Has Two Family Trees – A Genealogical Tree and a Genetic Tree." *The Genetic Genealogist*, 10 November 2009. https://thegeneticgenealogist.com/2009/11/10/qa-everyone-has-two-family-trees-a-genealogical-tree-and-a-genetic-tree/ : 2019.

———. "August 2017 Update to the Shared cM Project." *The Genetic Genealogist,* 26 August 2017. https://thegeneticgenealogist.com/2017/08/26/august-2017-update-to-the-shared-cm-project/ : 2019.

———. "Mapping Your Chromosomes With DNAPainter." Video tutorial. *YouTube*, 30 October 2017. https://www.youtube.com/watch?v=wyjcJxywTZI : 2018.

Bettinger, Blaine T., and Debbie Parker Wayne. *Genetic Genealogy in Practice.* Arlington, Virginia: National Genealogical Society, 2016.

Estes, Roberta. "Concepts – CentiMorgans, SNPs and Pickin' Crab." *DNAeXplained*, 30 March 2016. https://dna-explained.com/2016/03/30/concepts-centimorgans-snps-and-pickin-crab/ : 2019.

———. "Concepts – DNA Recombination and Crossovers." *DNAeXplained*, 9 November 2017. https://dna-explained.com/2017/11/09/concepts-dna-recombination-and-crossovers/ : 2019.

GEDmatch, Inc. *GEDmatch Genesis: Tools for DNA & Genealogy Research*. https://genesis.gedmatch.com : 2019.

Genetic Genealogy Standards Committee. "Genetic Genealogy Standards." http://www.geneticgenealogystandards.com : 2019.

Griffiths, A.J.F., J.H. Miller, D.T. Suzuki, et al. *An Introduction to Genetic Analysis*. 7th edition. New York: W. H. Freeman, 2000. Web edition. *National Center for Biotechnology Information*. https://www.ncbi.nlm.nih.gov/books/NBK21889/ : 2017. [Book is accessible by the search feature, but cannot be browsed.]

Perl, Jonny. "DNA Painter." Visual analysis tool. DNA Painter. https://dnapainter.com/ : 2019.

———. "The Shared cM Project 3.0 tool v4." Visual analysis tool. *DNA Painter*, August 2017. https://dnapainter.com/tools/sharedcmv4 : 2019.

Schaeffer, Stephen Wade. "Chromosome Behavior and Gene Linkage." Pennsylvania State University. *Biology 110 - Basic Concepts and Biodiversity*. https://wikispaces.psu.edu/display/110Master : 2019.

**Notes:**