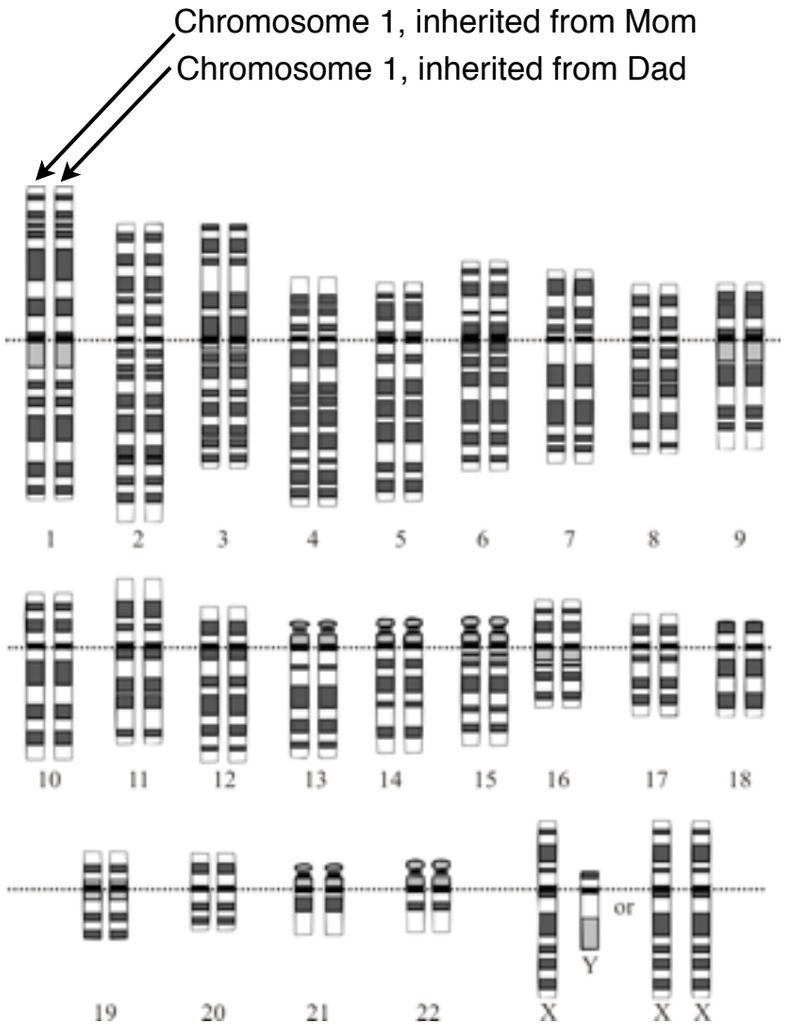
The genome: where genotypes live



Human chromosomes

23 pairs, 46 total
22 pairs are “autosomes”
1 pair are “sex chromosomes”

Genome is the entire DNA sequence of an individual; all chromosomes

Human genome is 3 billion nt

“nt” = nucleotides long
similarly: “bp”

Most bacterial genomes are a few million nt. Most viral genomes are tens of thousands of nt. This plant’s genome is about 150 billion nt.

Paris japonica

Pictures: http://en.wikipedia.org/wiki/Chromosome, http://en.wikipedia.org/wiki/Paris_japonica

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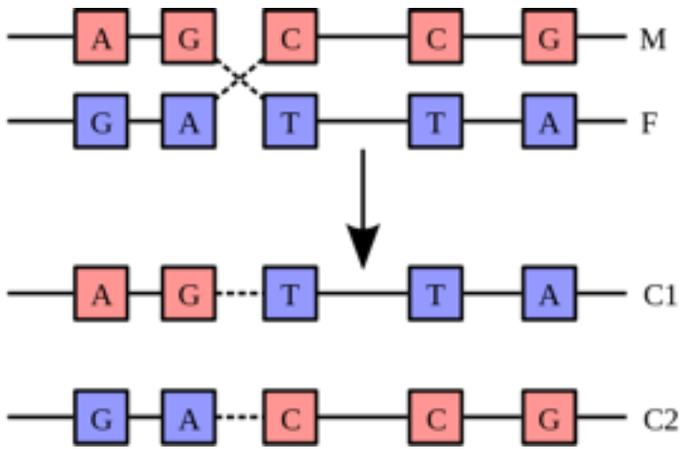
Evolution: why *these* genotypes?

Organisms reproduce, offspring *inherit* genotype from parents

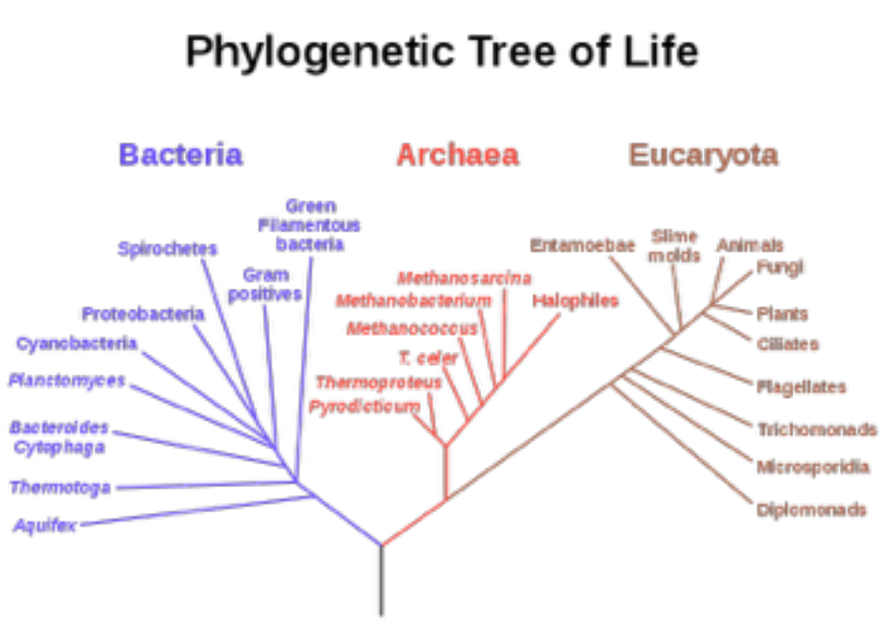
Random *mutation* changes genotypes and *recombination* shuffles chunks of genotypes together in new combinations



http://en.wikipedia.org/wiki/Genetic_recombination

Natural *selection* favors phenotypes that reproduce more

Over time, this yields the variety of life on Earth. Incredibly, all organisms share a common ancestor.



http://en.wikipedia.org/wiki/Evolutionary_tree

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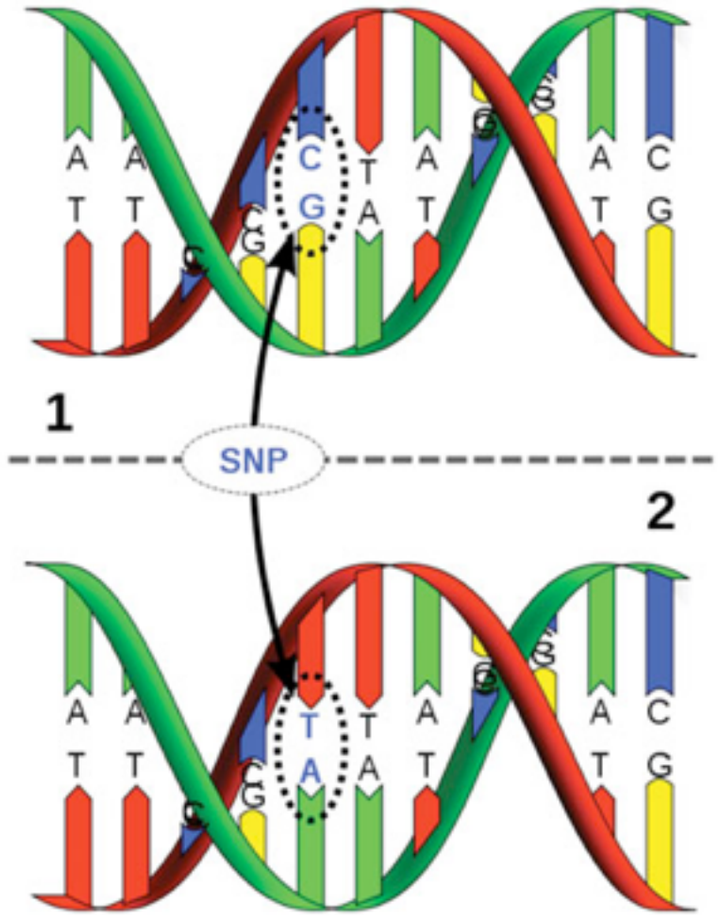
The genome: variation

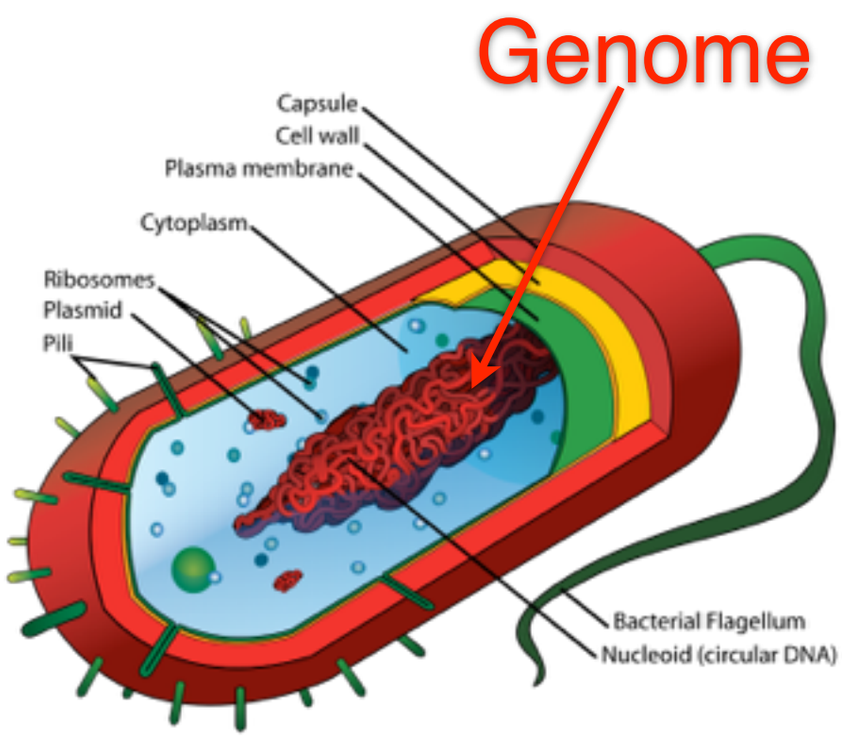
Two unrelated humans have genomes that are ~99.8% similar by sequence. There are about 3-4 million differences. Most are small, e.g. Single Nucleotide Polymorphisms

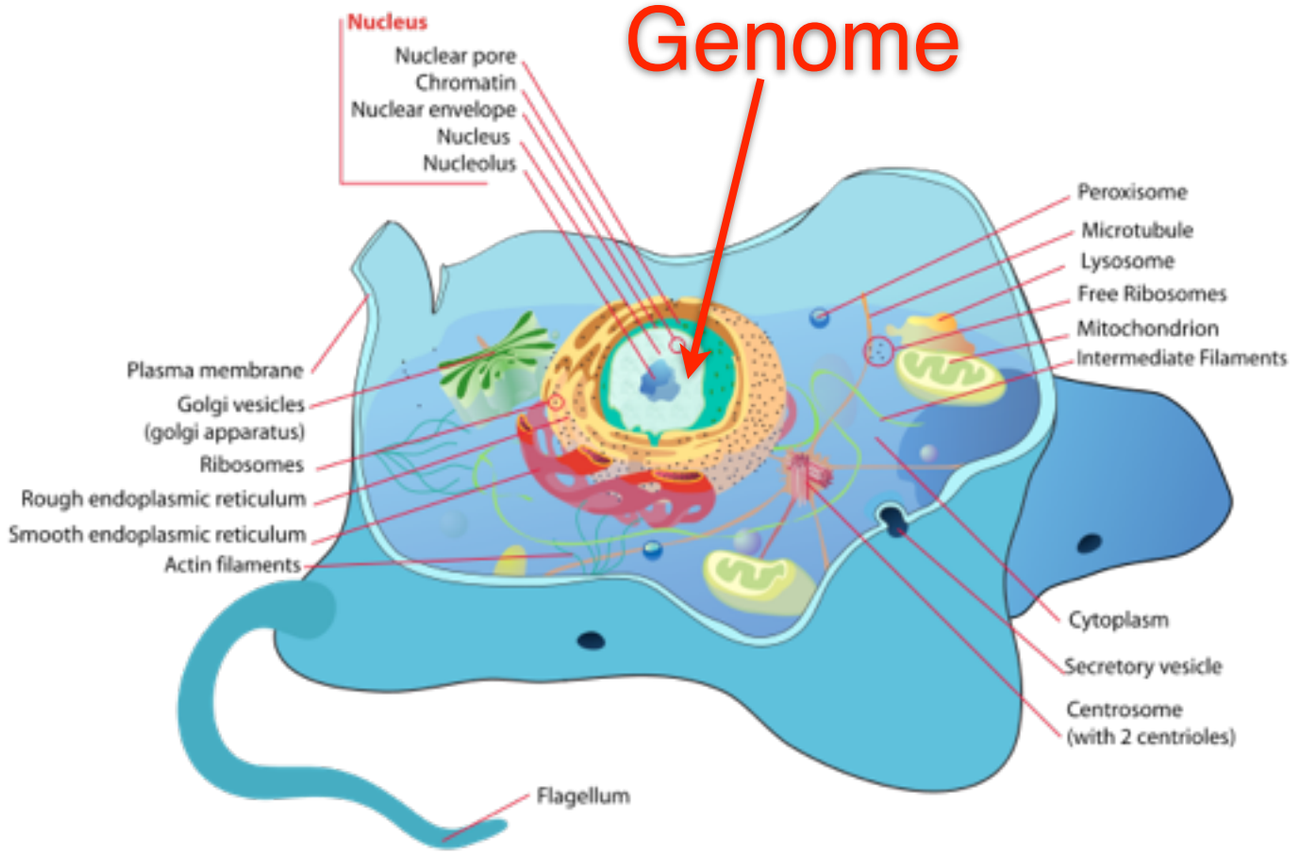


Human and chimpanzee genomes are about 96% similar

Pictures: http://www.dana.org/news/publications/detail.aspx?id=24536, http://en.wikipedia.org/wiki/Chimpanzee

Cells: where genomes live





Prokaryotic cell

A bacterium consists of a single prokaryotic cell

Eukaryotic cell
(pictured: animal cell)

Make up animals, plants, fungi, other eukaryotes

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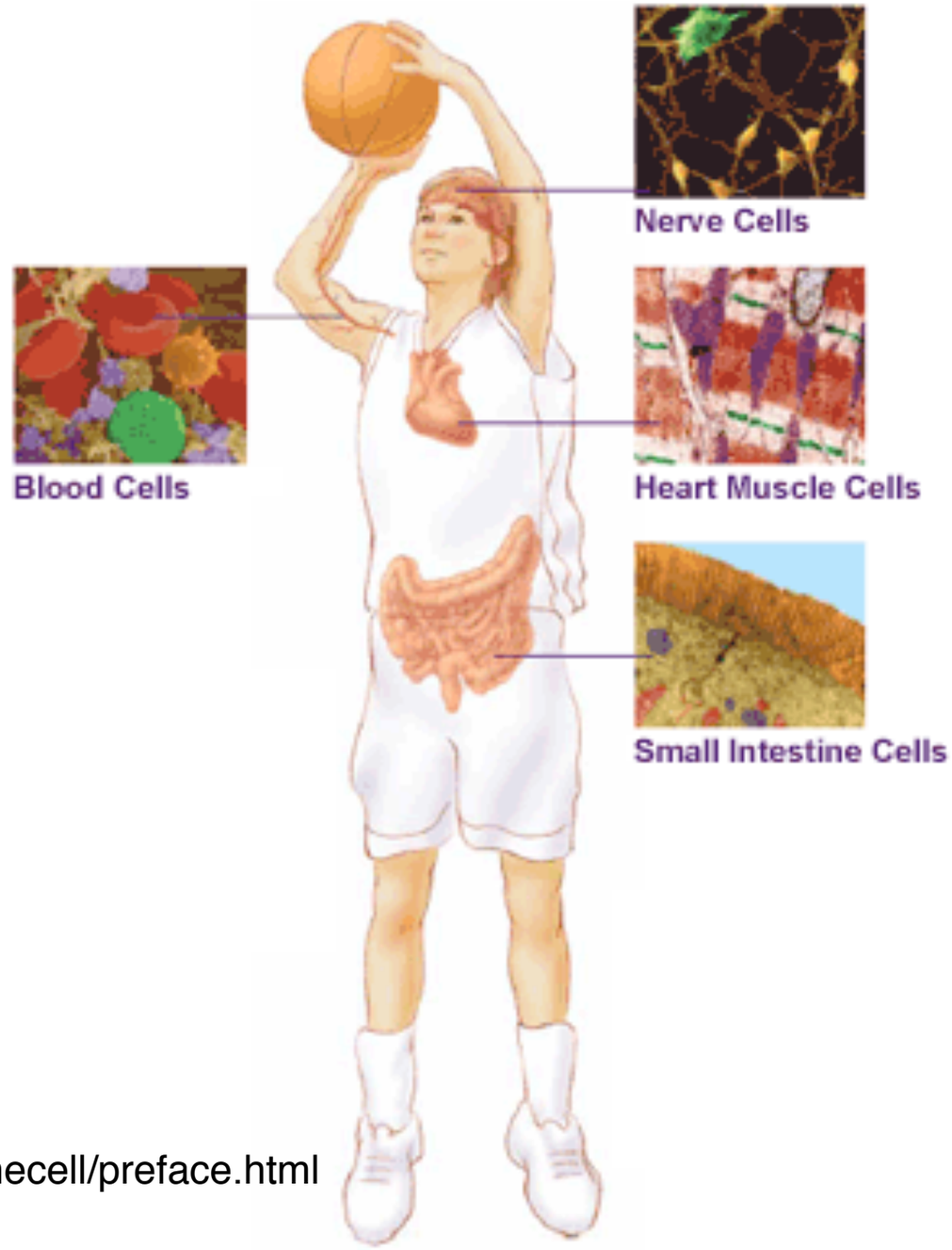
Pictures: http://en.wikipedia.org/wiki/Cell_(biology)

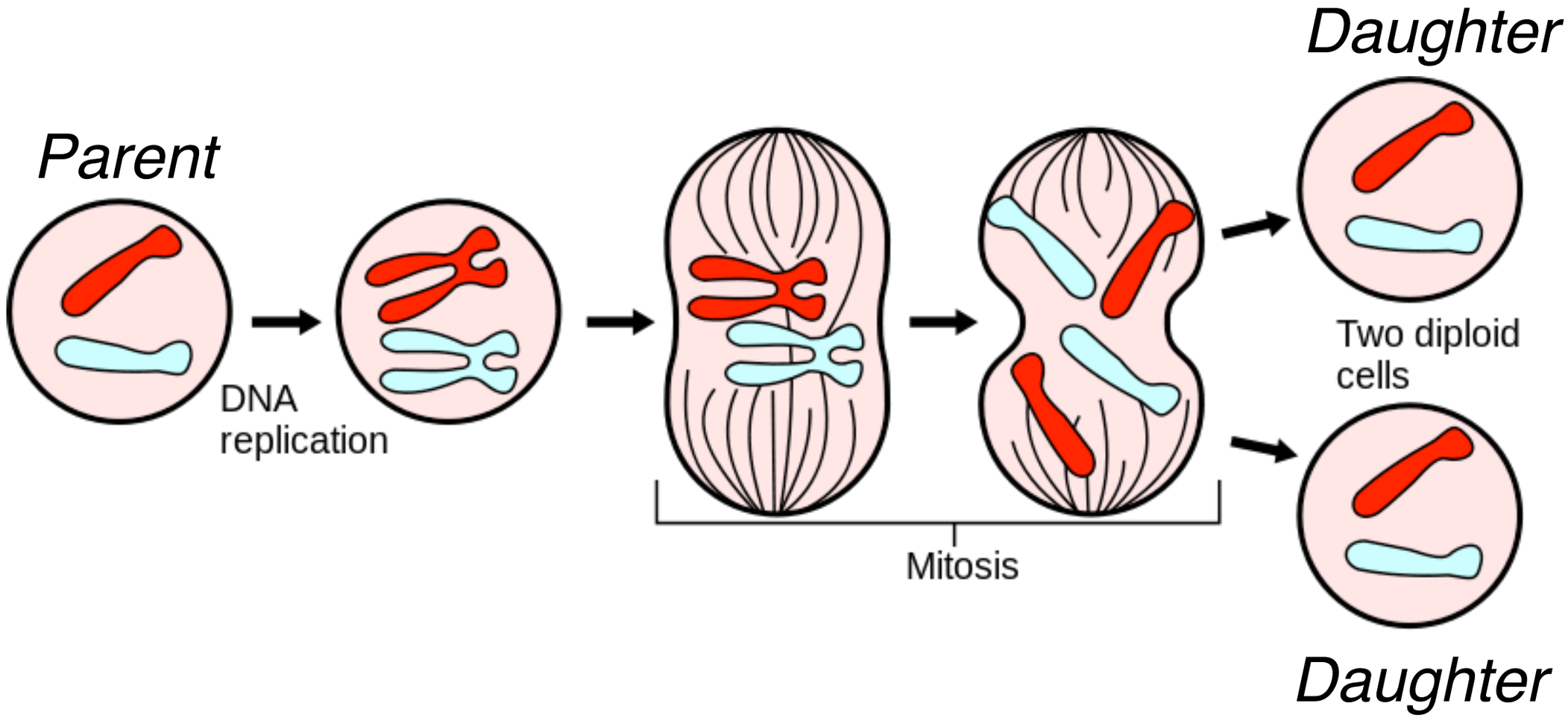
Cells: where genomes live

All the trillions of cells in a person have same genomic DNA in the nucleus



Picture: http://publications.nigms.nih.gov/insidethecell/preface.html

Cells: division



During cell division (*mitosis)*, the genome is copied

Picture: http://en.wikipedia.org/wiki/Mitosis

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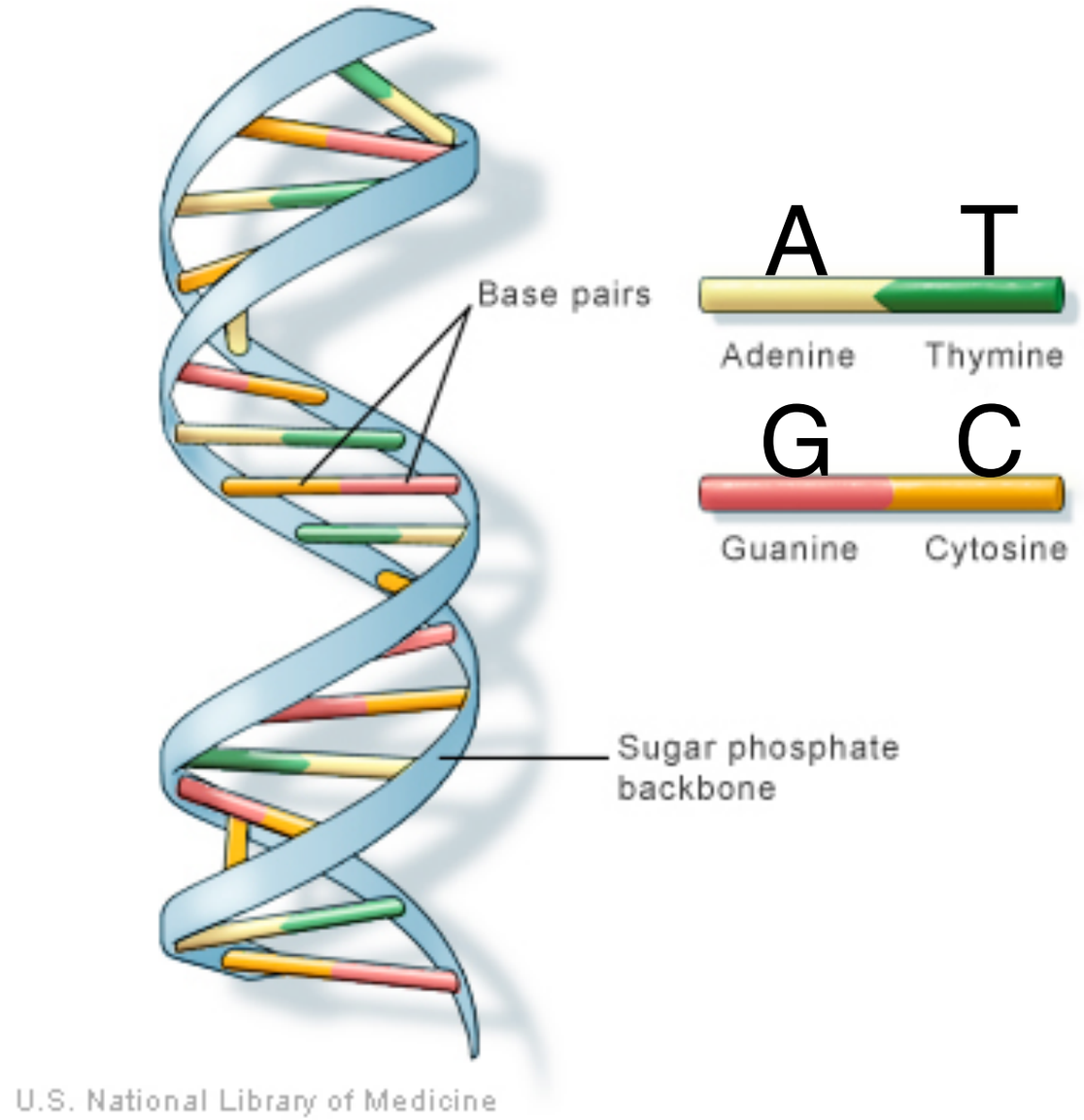
DNA: the genome's molecule



Picture: http://ghr.nlm.nih.gov/handbook/basics/dna

Deoxyribonucleic acid

"Rungs" of DNA double-helix are base pairs. Pair combines two complementary

Complementary pairings: A-T, C-G

Single base also called a "nucleotide"

DNA fragment lengths are measured in "base pairs" (abbreviated bp), "bases" (b) or "nucleotides" (nt)

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Stringizing DNA

DNA has *direction* (a *5' head* and a *3' tail*). When we write a DNA *string*, we follow this convention.

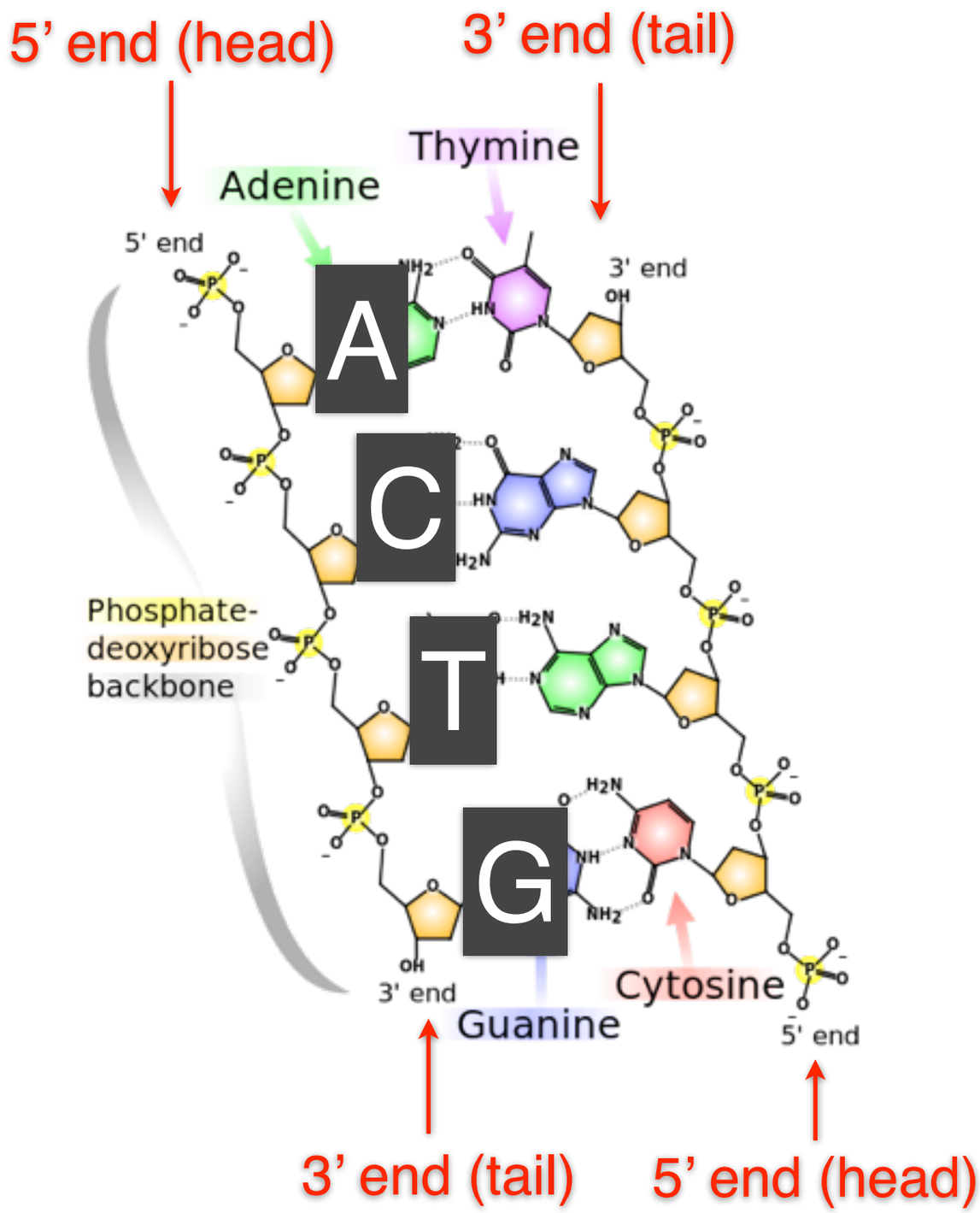
When we write a DNA string, we write just one strand. The other strand is its *reverse complement*.

To get reverse complement, reverse then complement nucleotides (i.e. interchange A/T and C/G)

5' end A C T G *3' end*

reverse complement

5' end C A G T *3' end*



Picture: http://en.wikipedia.org/wiki/DNA

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