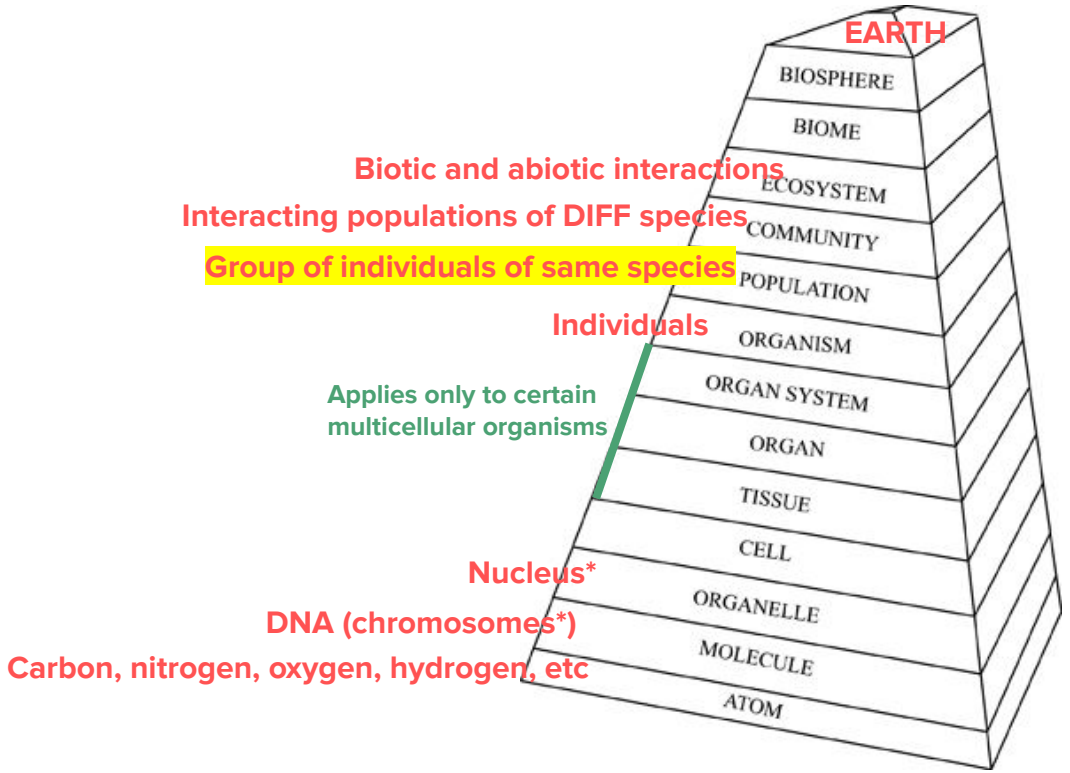
Introduction to genetics and biological variation

Introduction to Evolution and Scientific Inquiry
Dr. Spielman, Spring 2020

A systems view of biological organization



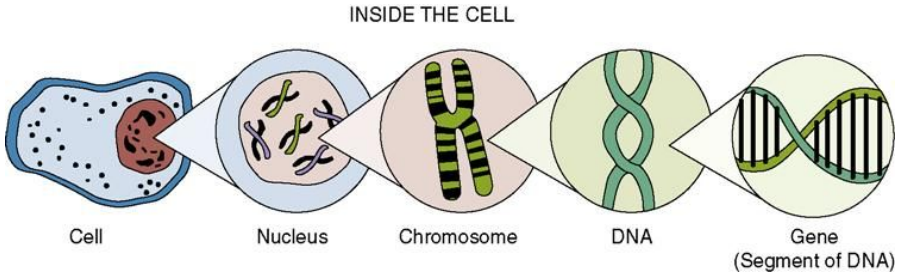
What is evolution?

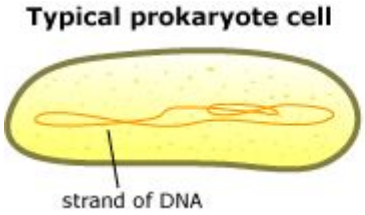
Biological evolution is *heritable* change in *populations* over *time* (generations)

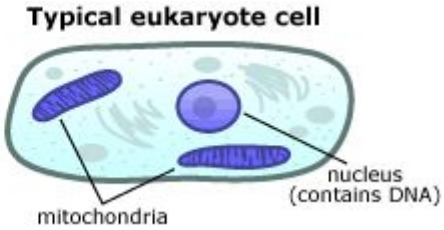
- Heritable: evolving trait requires a genetic* basis
- Populations: individuals do not evolve
- Time: evolution occurs over multiple *generations*, not within a generation

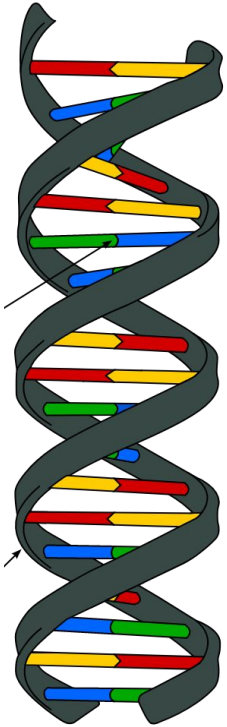
AKA: Biological evolution is change in *allele frequencies* in *populations* over *generations*

The genetic basis of variation: A brief overview

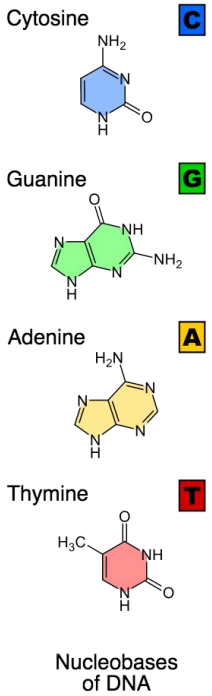








DNA
Deoxyribonucleic acid



Nucleobases of DNA

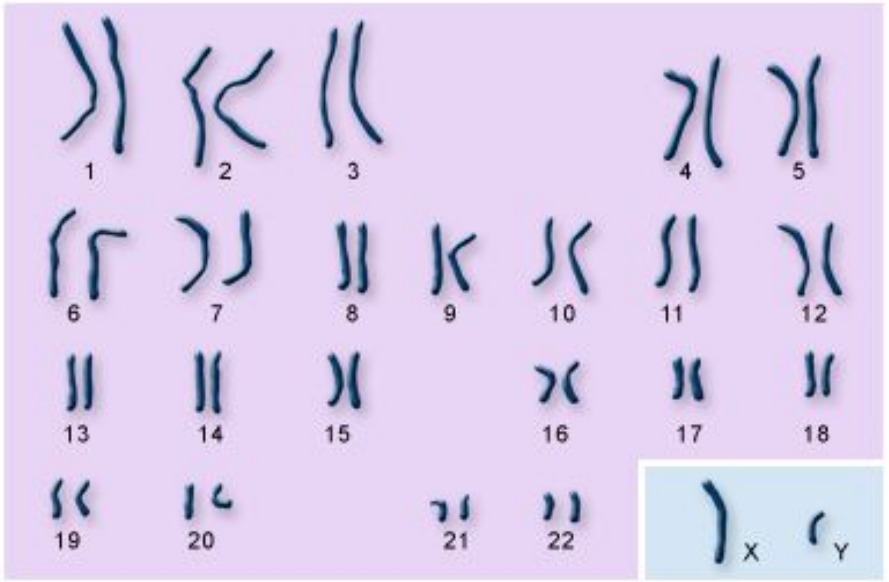
Humans have 23 *pairs* of chromosomes

One copy from mom, one copy from dad = 23*2 = 46 total

We are **diploid** (most *animals* are)

There are roughly 3 billion nucleotides in the human genome

Across the chromosomes, there are roughly 20,000 genes

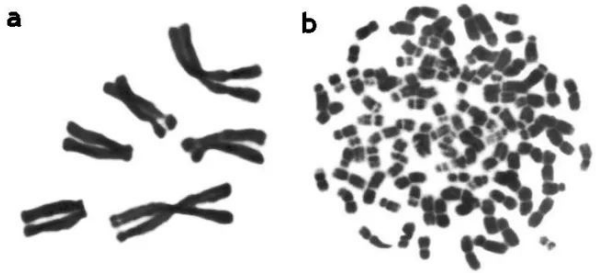


U.S. National Library of Medicine

Chromosomes are VERY variable ("karyotype")

Indian muntjac

smallest # in mammals



Viscacha rat

largest # in mammals

Siberian Roe deer

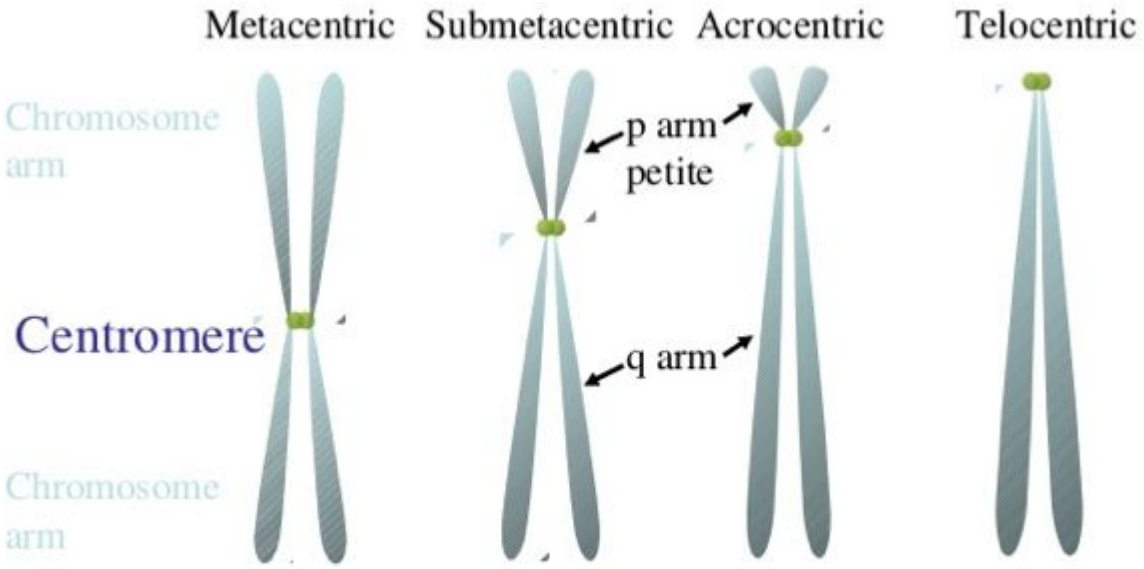
has weird transient "B" chromosomes

Transcaucasian mole vole female ("X" is not universal!)

Images during metaphase (DNA replication)

https://molecularcytogenetics.biomedcentral.com/articles/10.1186/1755-8166-4-22

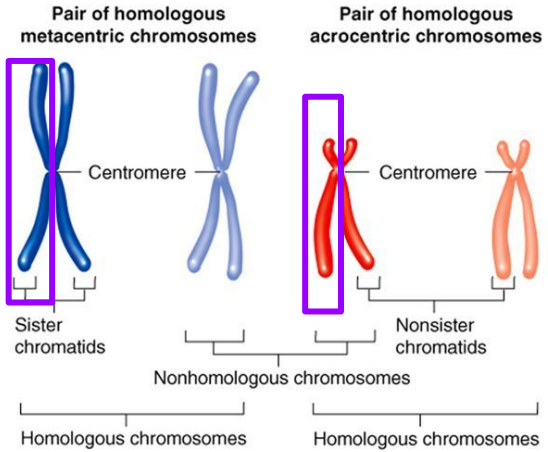
Some fun facts about chromosomes

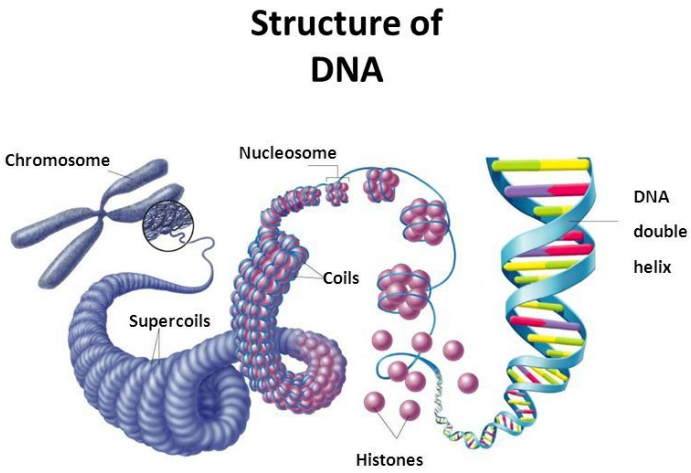


Most human chromosomes are **submetacentric**

One more view…

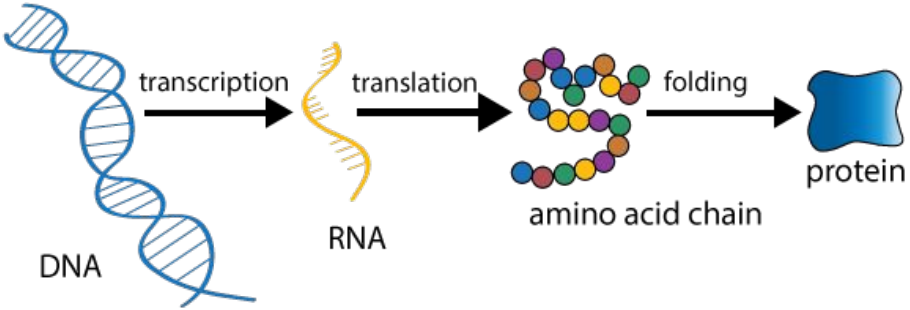
Fig. 4.5
Metaphase chromosome can be classified by centromere position

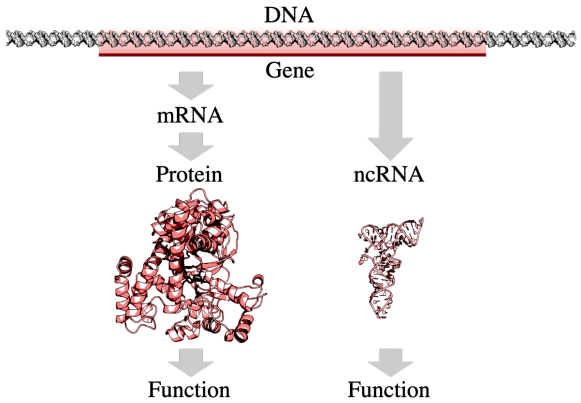




What is a gene?

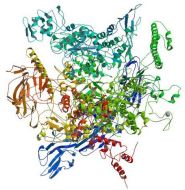
- A **gene** is a stretch of nucleotides in the genome that CODES for something
  - A "blueprint" to make other types of molecules
  - Can code for a **protein** end-product, or an **RNA** end-product (which also has a function!)



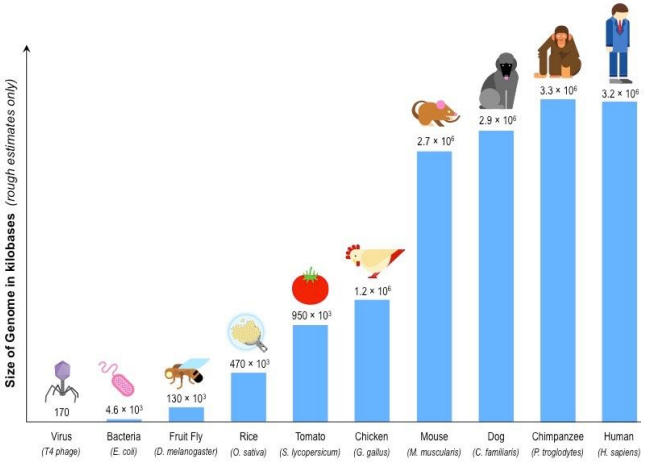


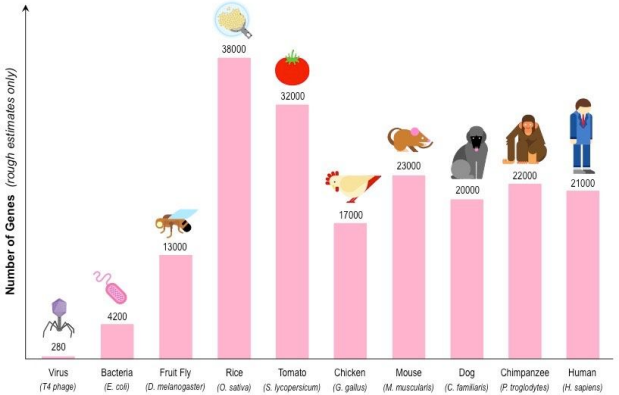
Proteins are small "machines" in cells that carry out the functions of life

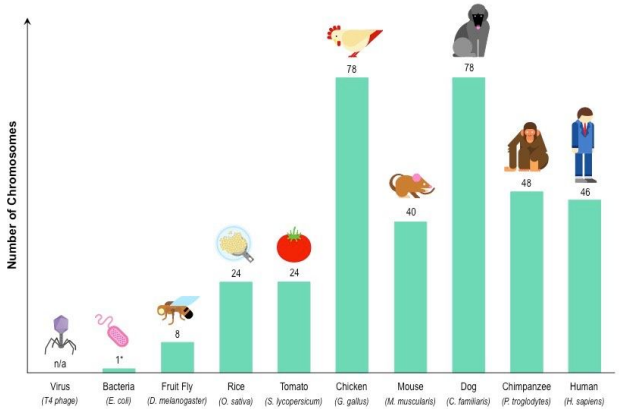
They are not a food group!



There is a **lot** of variation across the tree of life







Don't memorize these values - understand the CONCEPT

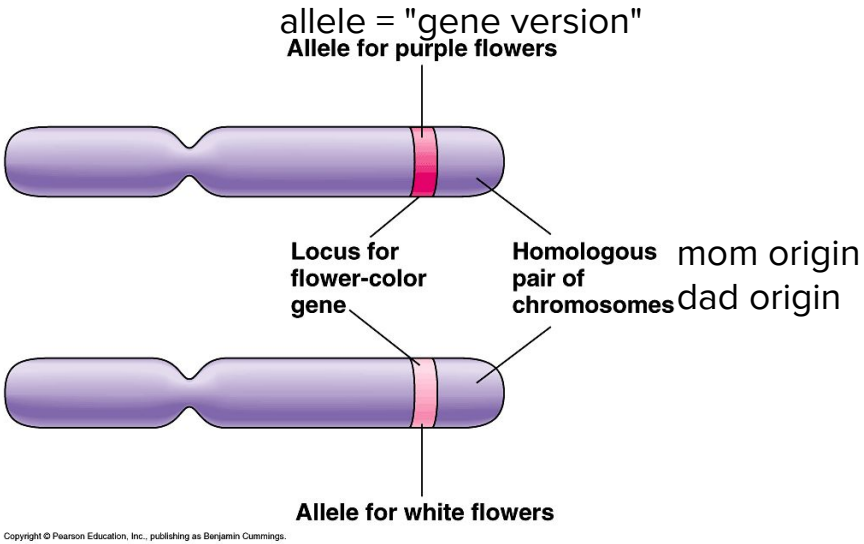
Genes are not magic formulas

THERE IS NO SUCH THING AS "the X gene"

What headlines have you seen for "scientists find the GENE that does THING?"

Instead, an individual's genetics ("genotype") **interact** with the environment to produce an individual's **phenotype**.

The location of a gene on a chromosome is called the **locus**



BUT THERE IS NO REAL SUCH THING AS GENE FOR "FLOWER COLOR"

Genotype → Phenotype is HARD!!!

1. Mendelian traits: One allele for one phenotype. *Extremely rare - exceptions!*

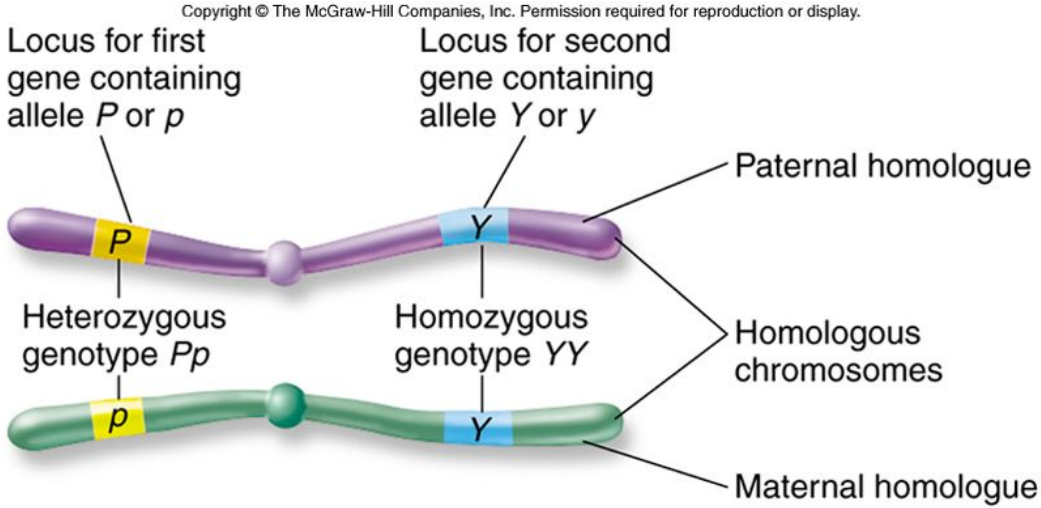
2. *Quantitative traits***: Dozens, hundreds, thousands of genes interact to produce a phenotype

3. Most *variation in traits* is affected by environment nonetheless

Genetic terminology

Homozyote: An individual with the *same* version (allele) of the gene on both chromosomes

Heterozygote: An individual with a *different* allele on each chromosome



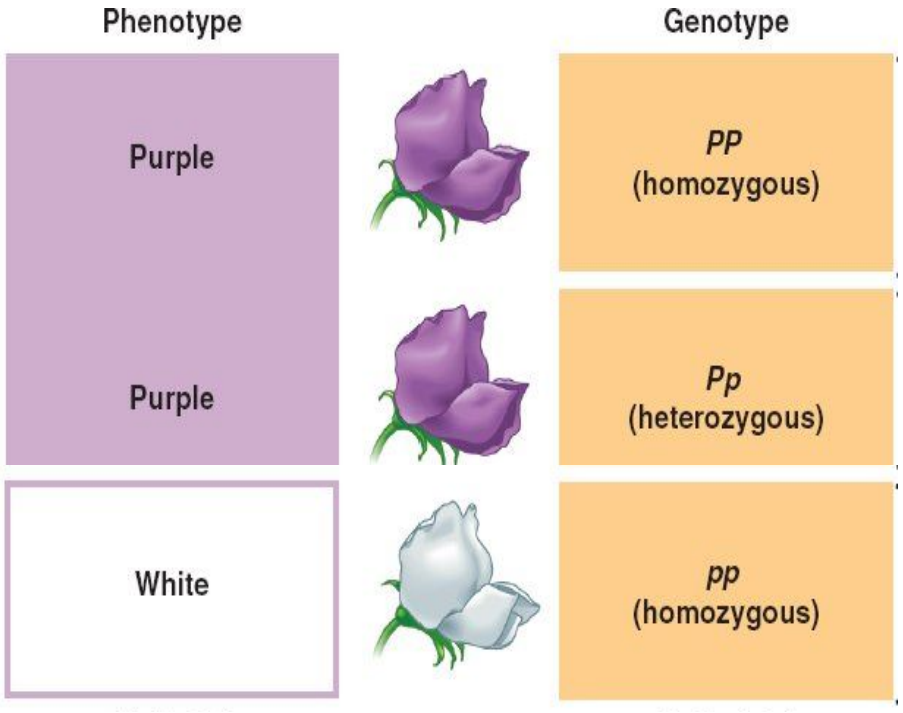
Genetic terminology

Phenotype = physical appearance

Genotype = underlying genetics

Trait itself: The flower has color

Trait variation: What color is it?

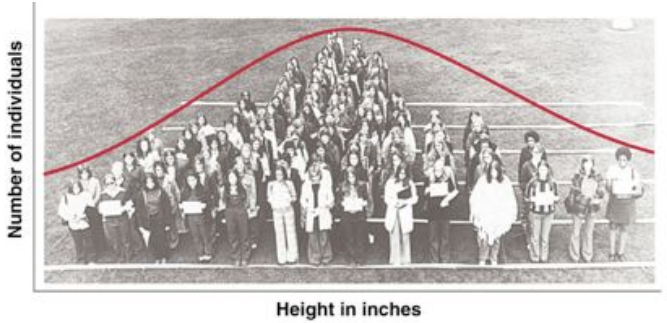


Types of phenotypic variation

Discrete variation is usually caused by a single gene (Mendelian)

"Big A, little a" combinations.
AA: black
Aa: black (with "A" dominant)
aa: white

Continuous variation is usually a complex result of *hundreds or thousands* of interacting genes. <u>The exact genotype is often unknown.</u>



How many alleles does each gene has? Is it always "big A, little a"?

Some genes have **one** allele in a species

Some genes have **dozens or hundreds** of alleles in a species

Brainstorm: Why are there different numbers of alleles across genes? Are more alleles "good", "bad", "neutral"?

What does it really mean to be a different allele?

- We can think of genes as their **DNA sequence:**

...AGGATCGATAGGACACTCGCGGTA...	**"wild type"** (most in species have this sequence)
...AGGATTGATAGGACACTCGCGGTA...	**a single nucleotide difference**
...AGGATAGATAGGACACTCGCGGTG...	**2 nucleotide differences**
...AGGATAGATAGGACACTCGCGGTA...	**some nucleotides are "deleted"**
...AGGATAGATAGGACACTCGCGGTGATAACA...	**some nucleotides are "inserted"**

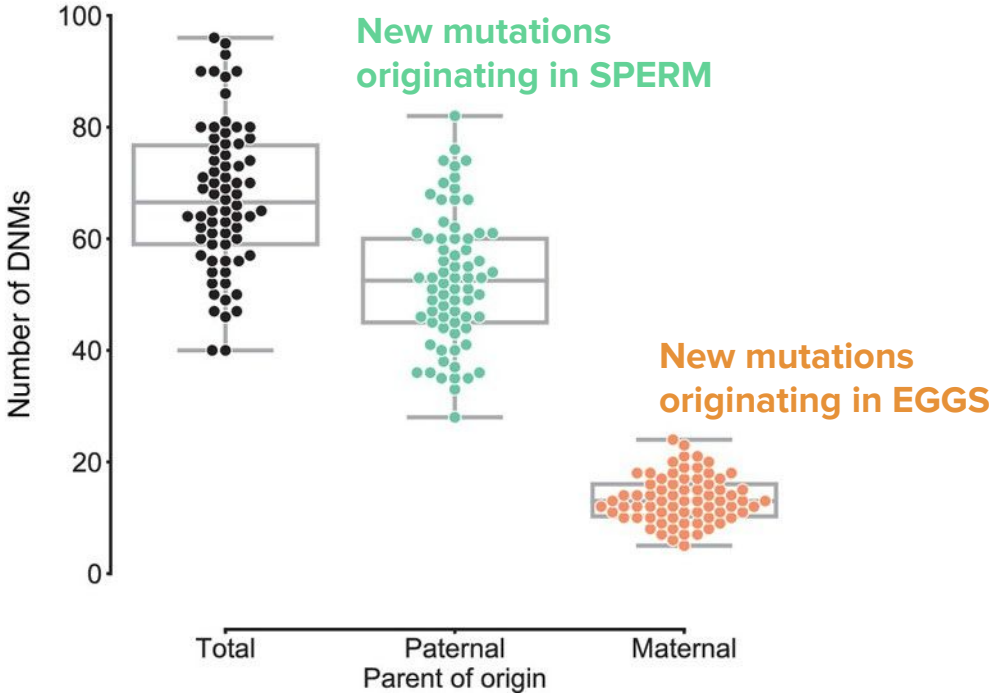
So what are mutations?

- When DNA copies itself (cells divide to make more cells), errors sometimes occur. *These errors are MUTATIONS!*
    - Imagine copying a 3 billion page book by hand. You're going to mess up. Add letters, remove letters
- Sometimes environmental factors (i.e. radiation) cause DNA to change. *These changes are MUTATIONS!*
- Mutations are **random mistakes.**

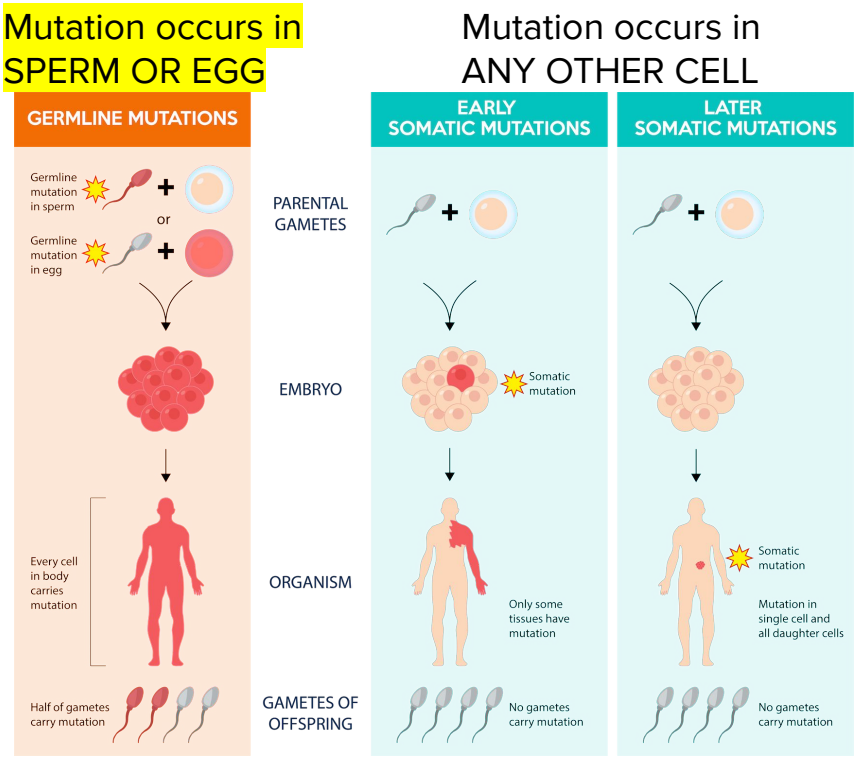
- Mutation is the **raw source of ALL variation** (aka source of all new alleles)
    - Without mutation, there is NO EVOLUTION (keep coming to class for more information!)

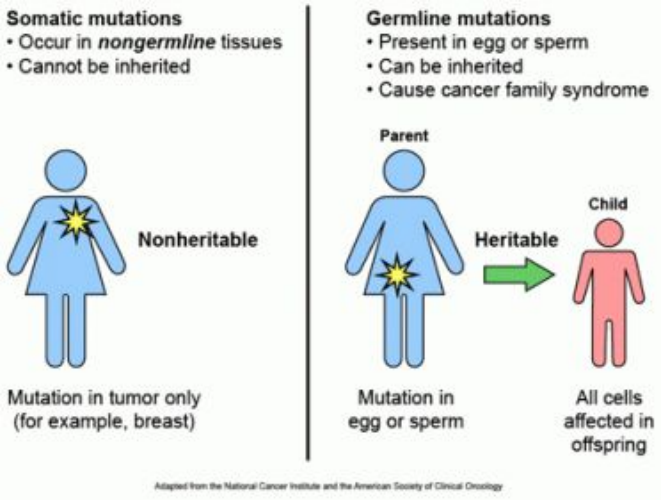
How many **brand new** mutations do YOU have?

DNM = *de novo* mutation
= not in Mom or Dad. Yours!



Somatic vs germline mutations





These "somatic mutations" *can* result in cancer/tumors

What are the potential consequences of a mutation?

- This is a very active area of research!
- There are two competing views of "mutational effects", but it is generally agreed that most random changes are BAD

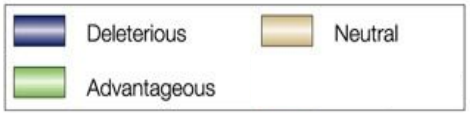
Most mutations are deleterious (bad for organism)

A small proportion are advantageous (helpful for organism)

~Half mutations are deleterious (bad for organism)

~Half mutations are *neutral* (different, but basically "fine")

Very small proportion are advantageous (helpful for organism)



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