Natural Selection Genetics and Inheritance

4/11/2023

Review from last week

- What is evolution?
  - Evolution is the **change in allele frequencies** that occurs over time within a population

- Postulates of Natural Selection
  1. There is a struggle for existence.
  2. There is variation in features related to survival and reproduction.
  3. This variation is passed from generation to generation.

Review from last week

- **Darwin's Difficulties Explaining Variation**
  - Is there blending inheritance?

  (how the traits are inherited)
  - How is variation maintained if natural selection eliminates it?
  - Where do novel forms come from if they are not already present in the original population?

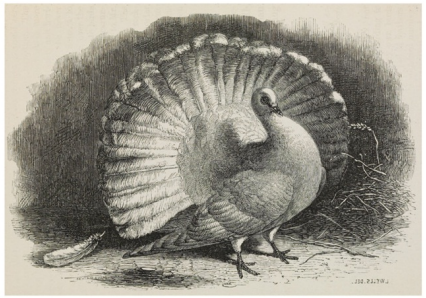
Artificial Selection

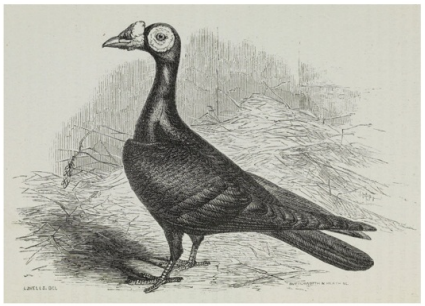
- Humans have domesticated and selectively bred plants and animals.
- Rapid changes have been achieved in few generations.
- Dogs and pigeons are good examples.

FIGURE 1.23

[a]

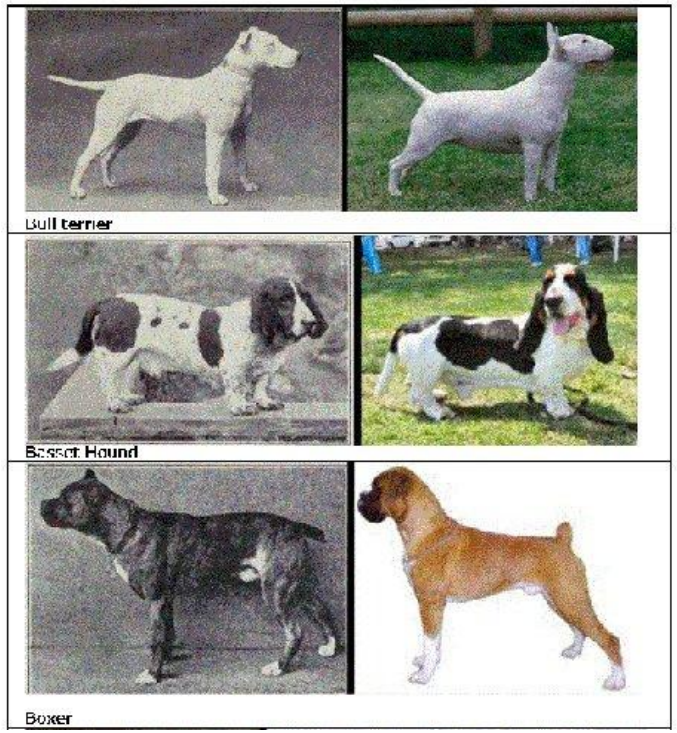
CreativeNature_nl/iStock/Getty Images Plus



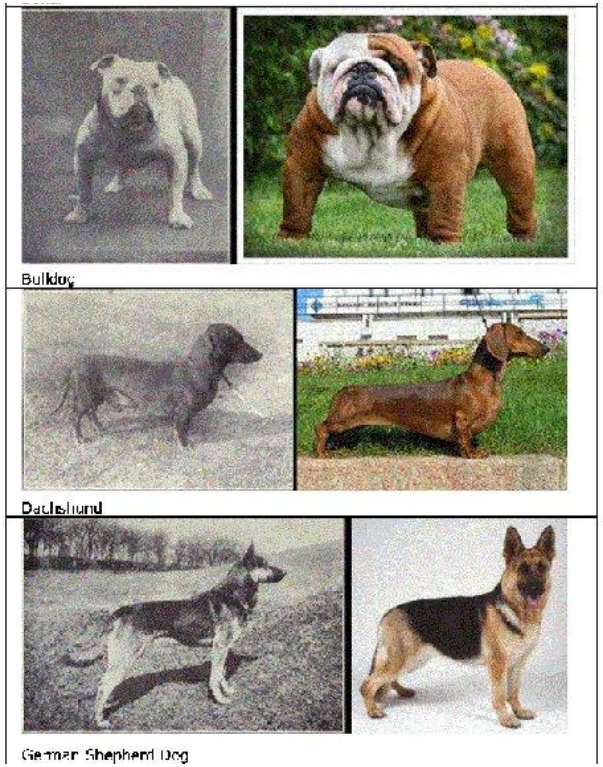


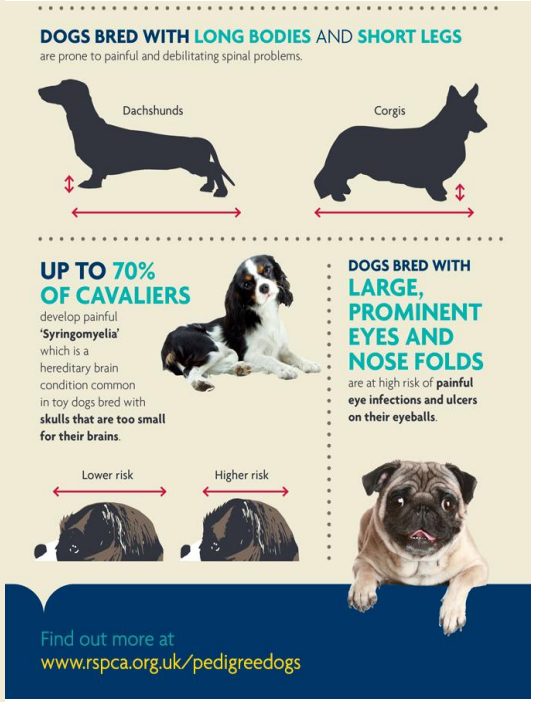
100 Years of Breed “Improvement”

1915 2012



1915 2012





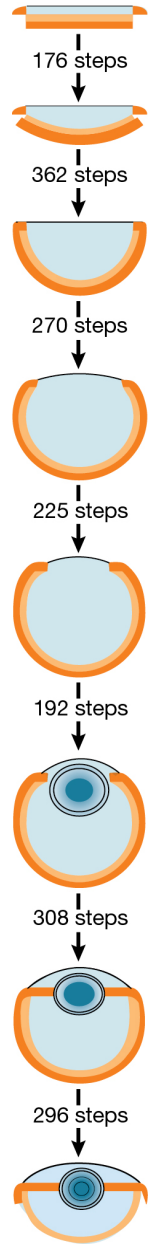
Rates of Evolutionary Change

- Minnows
  - Rapid evolution of placenta-like structure
- Eye
  - Small, incremental steps of 1% change
  - Eye evolves in 1,800 steps (took about 1million years)

FIGURE 1.21

Bill Radke/USFWS

FIGURE 1.22



Mechanisms of Heritability

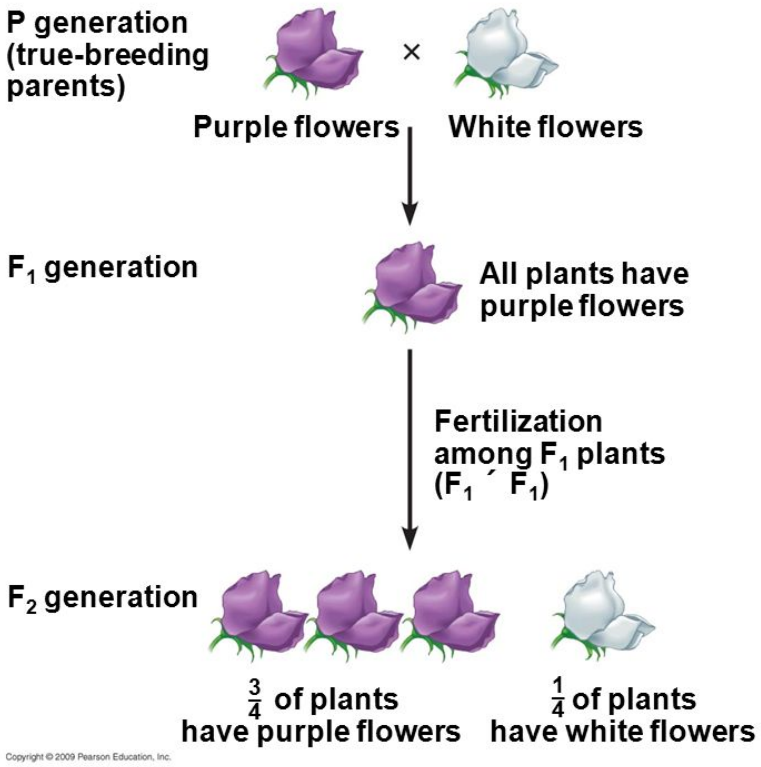
- Discovered laws of inheritance
- Bred garden pea plants (1856–1863)
- Worked with traits with two variants

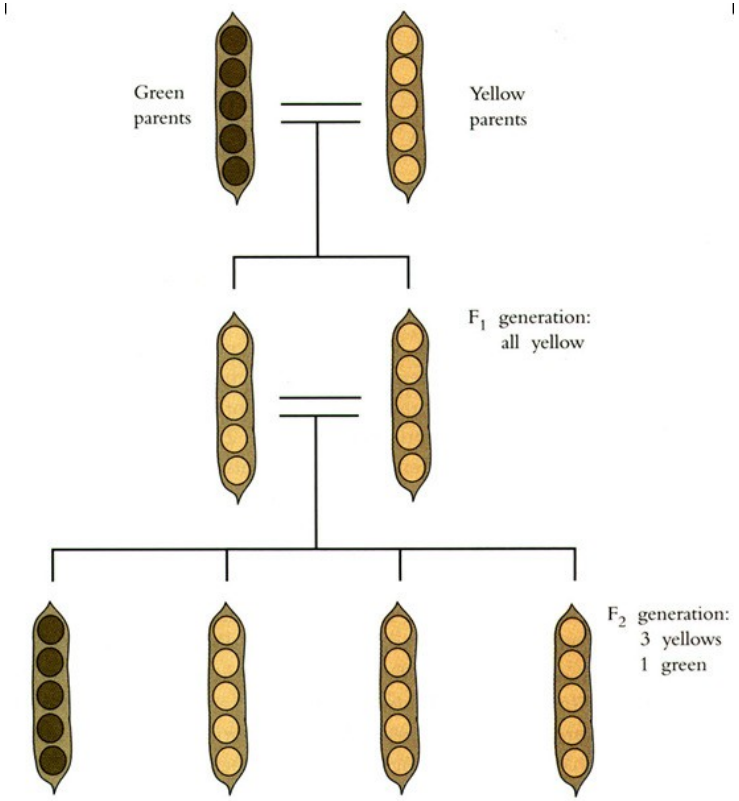
Gregor Mendel
(Austrian Monk)

Seed		Flower	Pod		Stem	
Form	Cotyledon	Color	Form	Color	Place	Size
						
Round	Yellow	White	Full	Green	Axial pods	Tall
						
Wrinkled	Green	Violet	Constricted	Yellow	Terminal pods	Short
1	2	3	4	5	6	7

Crossbreeding Experiments

- "particles" inherited from each parent
- maintains variation





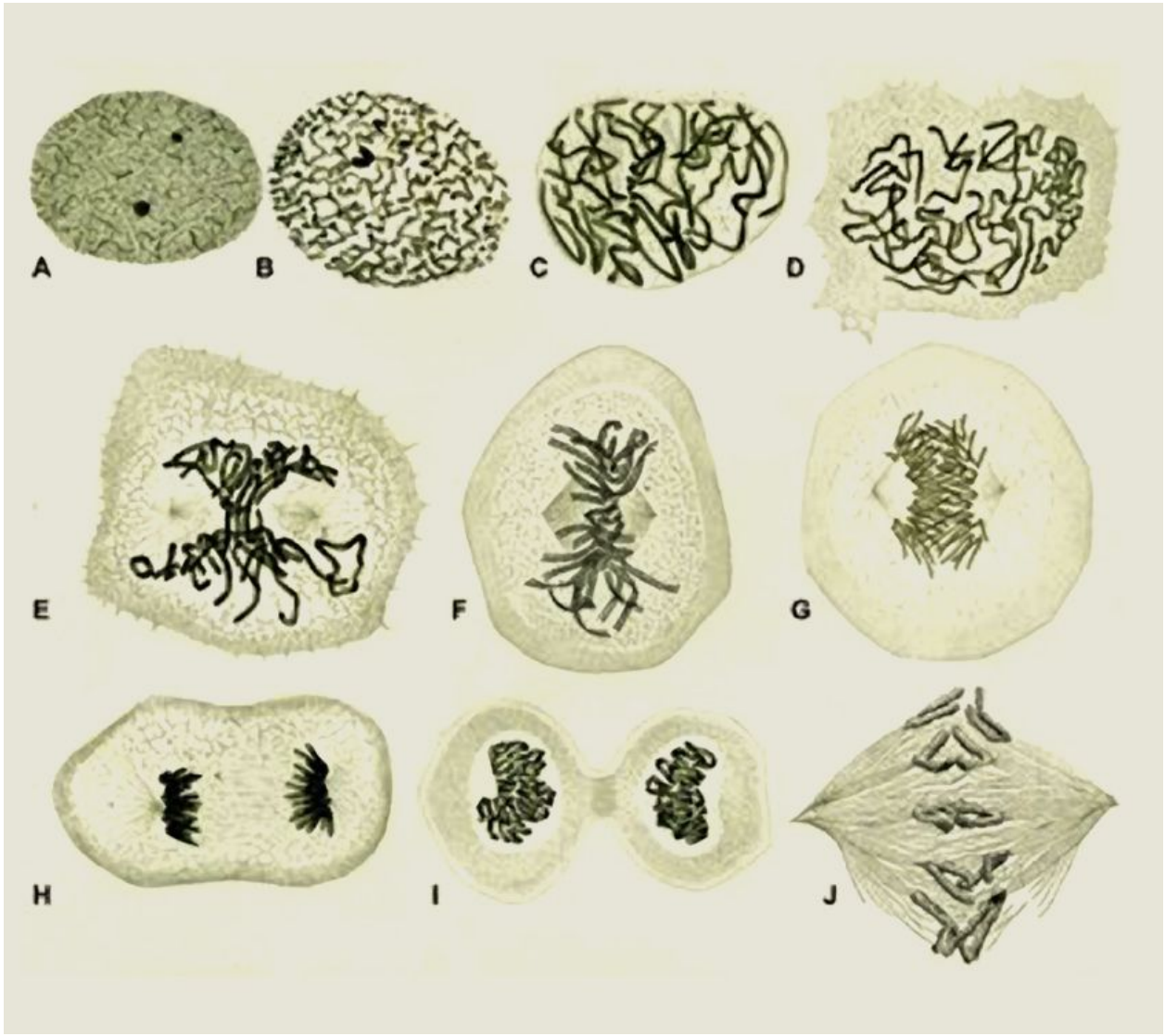
What are the “particles” inherited from each parent?
DNA – A History

- 1900s – Mendel’s work resurfaces, spurs more research

Gregor Mendel

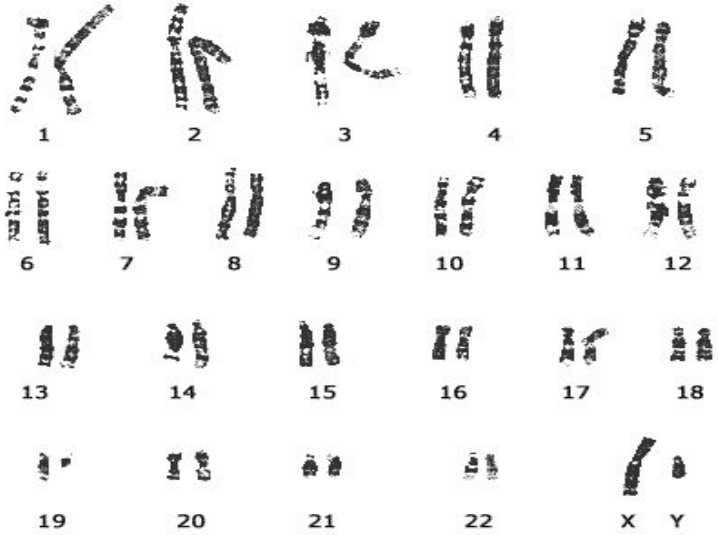
DNA – A History

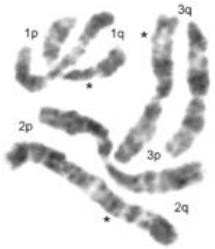
- 1882 – Walther Flemming
  - Discovered a fibrous structure inside the nucleus of cells (salamander embryos)
  - Called it **chromatin**
    - **Chromosomes**
    - Identified the process through which it separates during cell division: **mitosis**



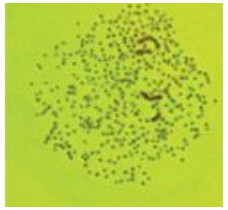
What are Chromosomes?

- ‘Bundles’ of DNA
- **46** total in humans (**23 homologous** (=matching **pairs**)
  - 22 pairs are **somatic chromosomes**
  - 1 pair are the **sex chromosomes** (XY or XX)
- Species varies in # of chromosomes (e.g. chimpanzees have 48)

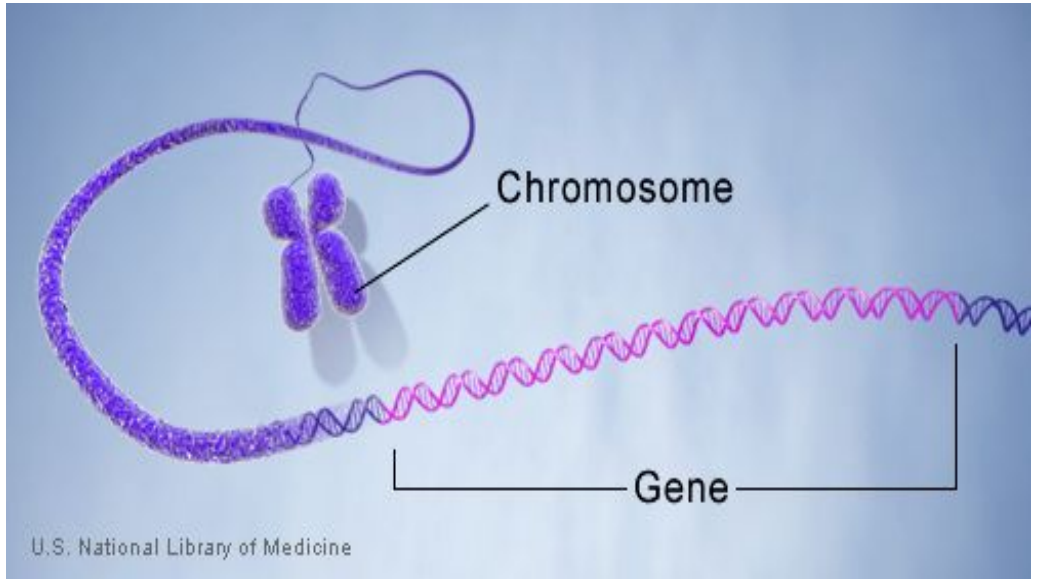




Yellow Fever Mosquito (2n=6)



Atlas blue butterfly (2n=448-452)



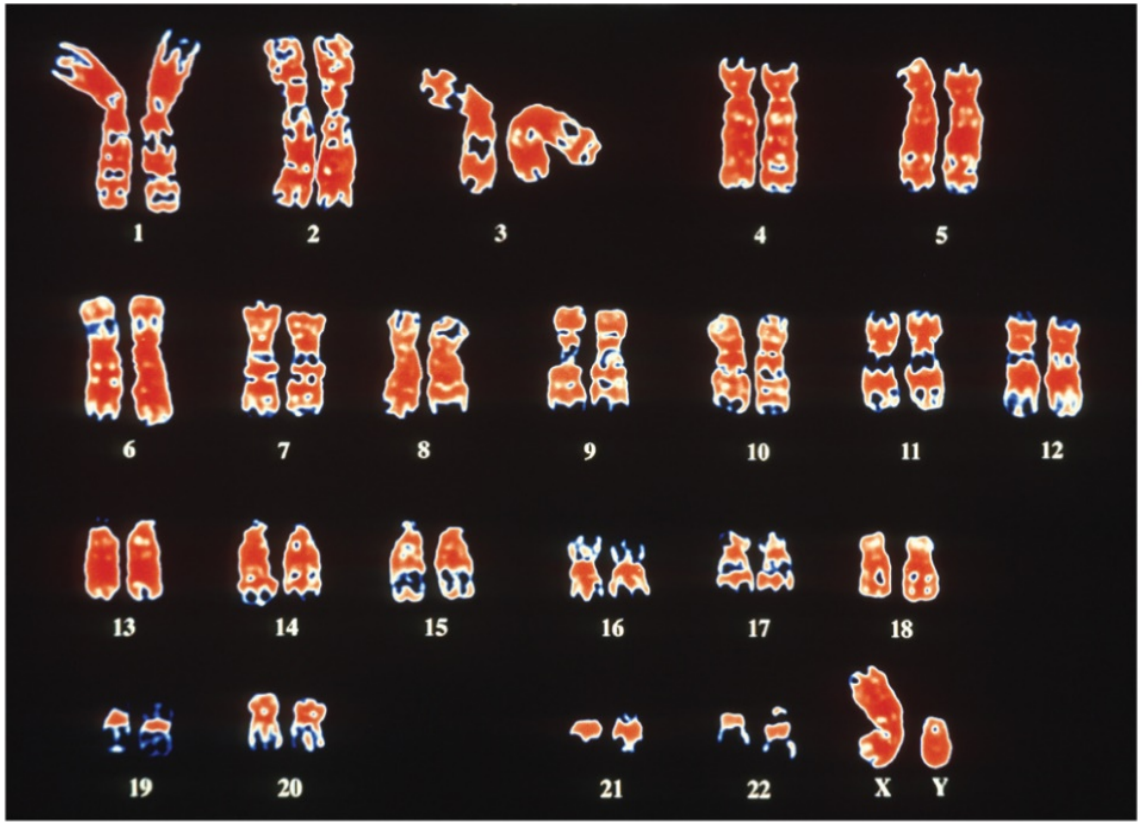
DNA – A History

- Late 1900s – Walter Sutton & Theodor Boveri
  - Worked independently of each other
  - Grasshopper chromosomes
  - Roundworm embryos
  - Genetic material that is passed from parent to child is in the chromosomes
    - **Chromosome Theory of Inheritance**

Cell Division and the Role of Chromosomes in Inheritance

- Karyotype = the picture of an individual's stained chromosomes, arranged in homologous pairs, laid out from largest to smallest
  - Homologous pairs= set of matching chromosomes

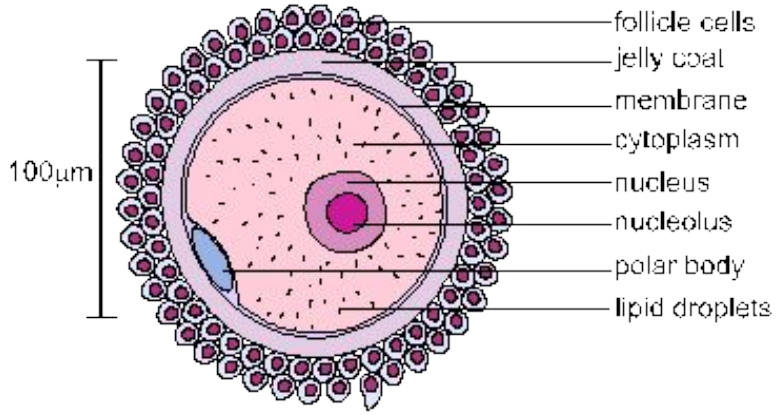
- Understanding chromosomes was key to understanding inheritance, which in turn is key in understanding evolution



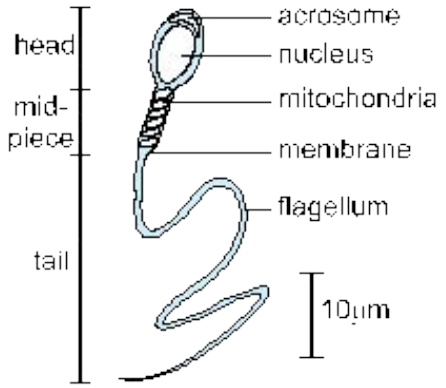
Cell Division

- Mitosis: “Ordinary” or somatic cell division (all cells other than egg and sperm)

- Meiosis: Sex cell division producing gametes
  - Produces haploid cells
  - Variation can be introduced through mutation and recombination



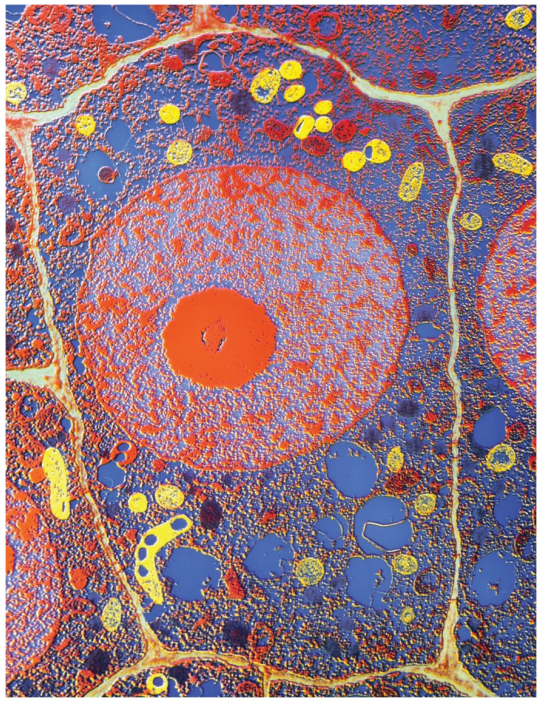
A Human Ovum

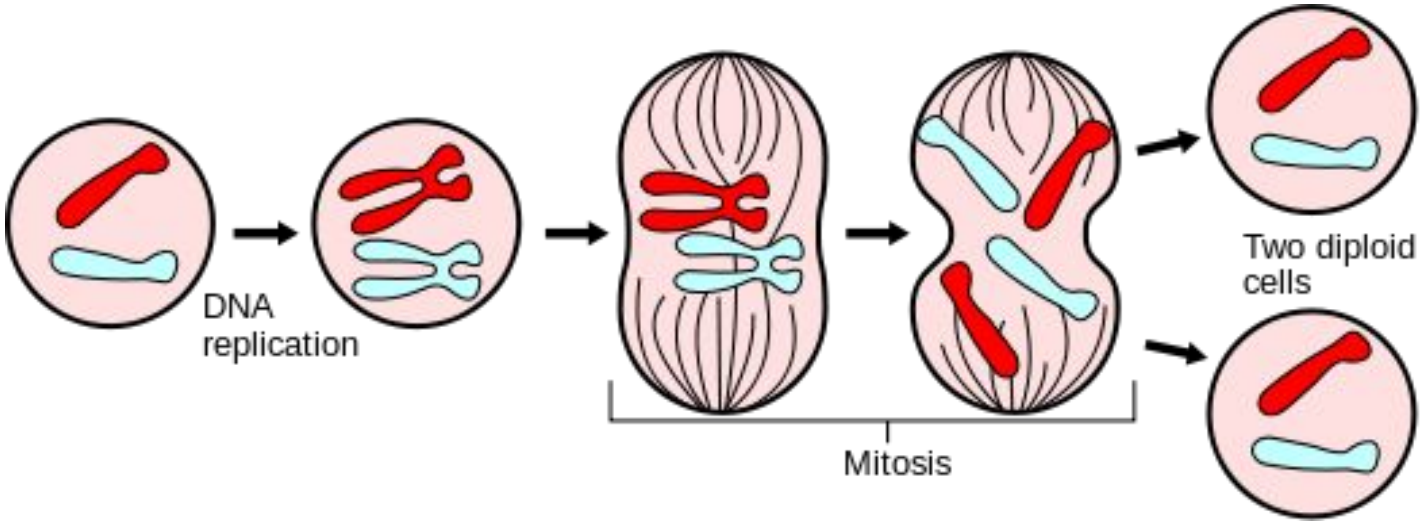


A Human Sperm

Chromosomes and Mitosis

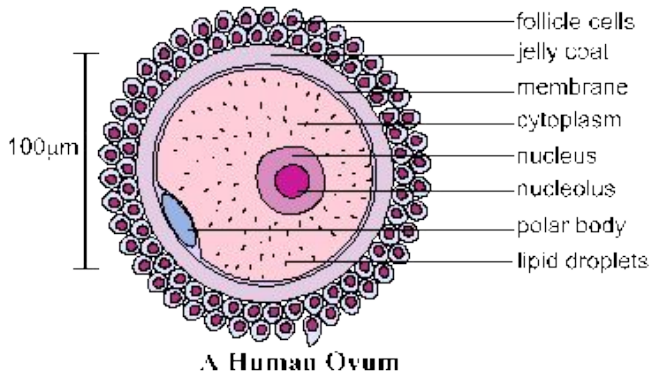
- "Ordinary" or somatic cell division (all cells other than egg and sperm)
  - Diploid (paired) chromosomes
  - Chromosomes replicate before cell division, otherwise each time there was a division there would only be half the genetic material



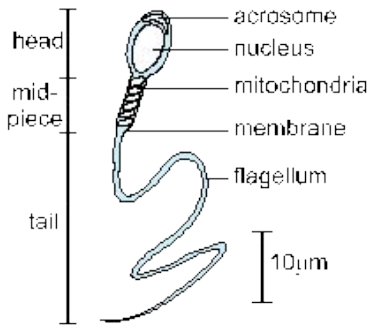


Chromosomes and Meiosis

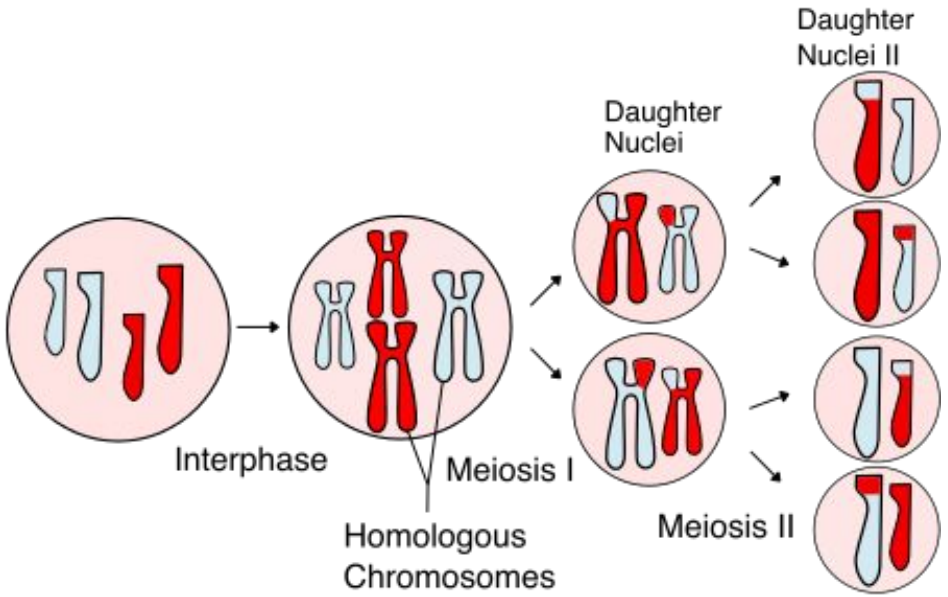
- Sex cell division producing gametes
- Haploid (not paired) chromosomes
  - Halves the number of chromosomes so that when there is a union of egg and sperm the total chromosome number remains the same

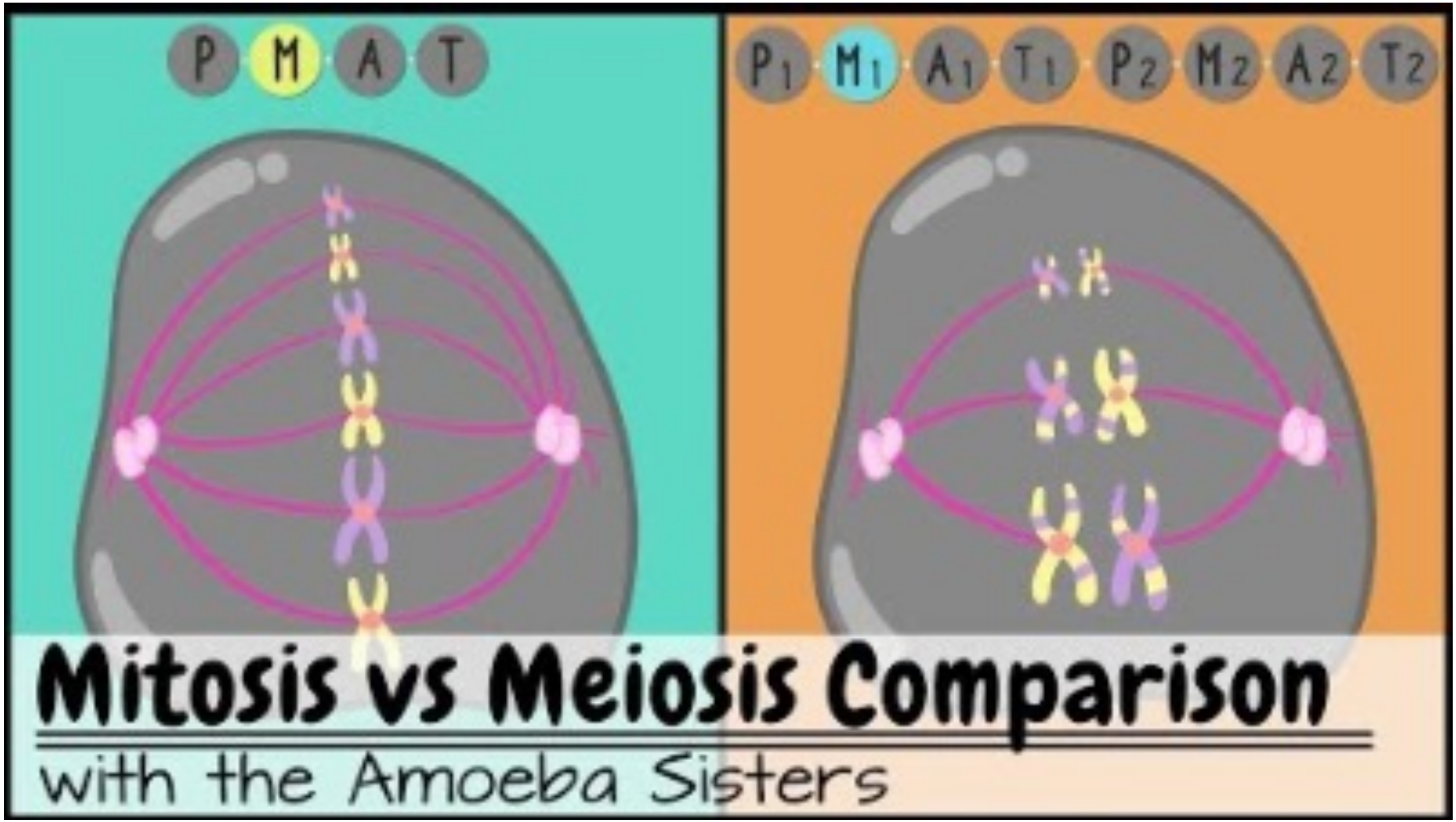


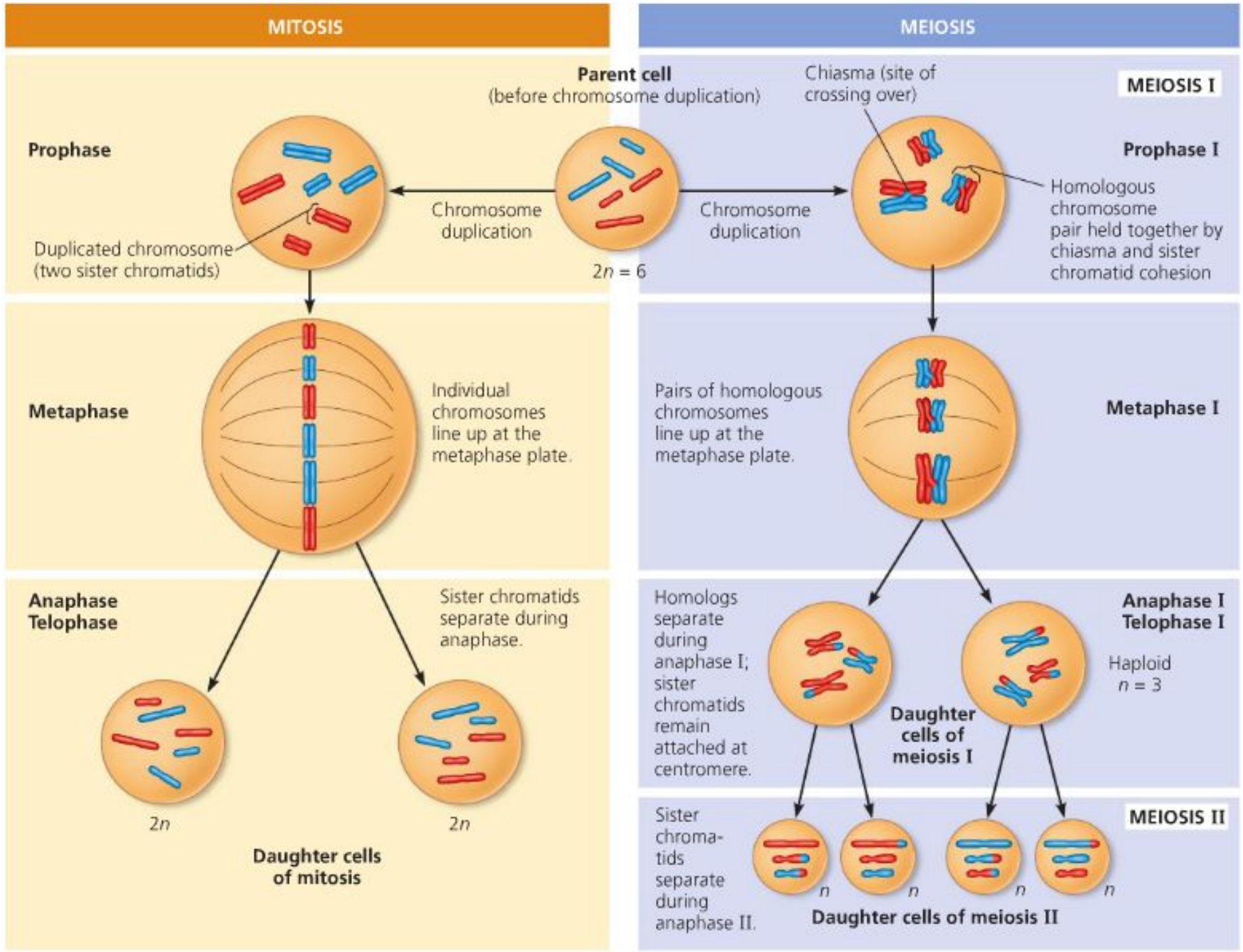
A Human Ovum



A Human Sperm

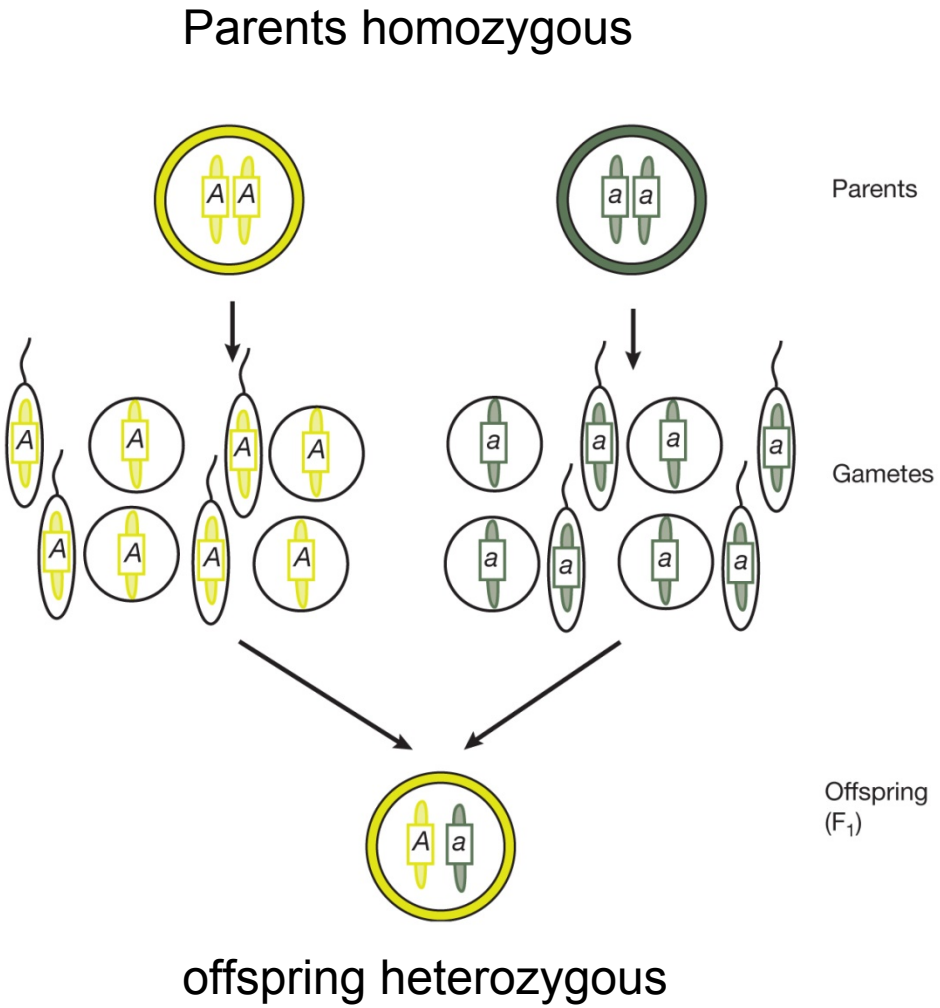



P M A T
P1 M1 A1 T1 P2 M2 A2 T2
Mitosis vs Meiosis Comparison
with the Amoeba Sisters


MITOSIS
MEIOSIS
Parent cell
(before chromosome duplication)
Chiasma (site of crossing over)
MEIOSIS I
Prophase
Prophase I
Chromosome duplication
Chromosome duplication
Duplicated chromosome
(two sister chromatids)
2n = 6
Homologous chromosome pair held together by chiasma and sister chromatid cohesion
Metaphase
Individual chromosomes line up at the metaphase plate.
Pairs of homologous chromosomes line up at the metaphase plate.
Metaphase I
Anaphase
Telophase
Sister chromatids separate during anaphase.
Homologs separate during anaphase I; sister chromatids remain attached at centromere.
Anaphase I
Telophase I
Haploid
n = 3
Daughter cells of meiosis I
2n
2n
Daughter cells of mitosis
Sister chroma-tids separate during anaphase II.
MEIOSIS II
n
n
n
n
Daughter cells of meiosis II

Mendel's Experimental Results

- Gene = a section of DNA on a chromosome that codes for a particular trait
- Alleles = alternate forms of a gene
  - If alleles are the same: homozygous
  - If alleles are different: heterozygous



Mendel's Experimental Results

- Genotype = specific alleles that an organism has for a trait (i.e. the genetic code, we use short form to illustrate)

- Phenotype = the physical expression an organism has for a trait (i.e. what you see)

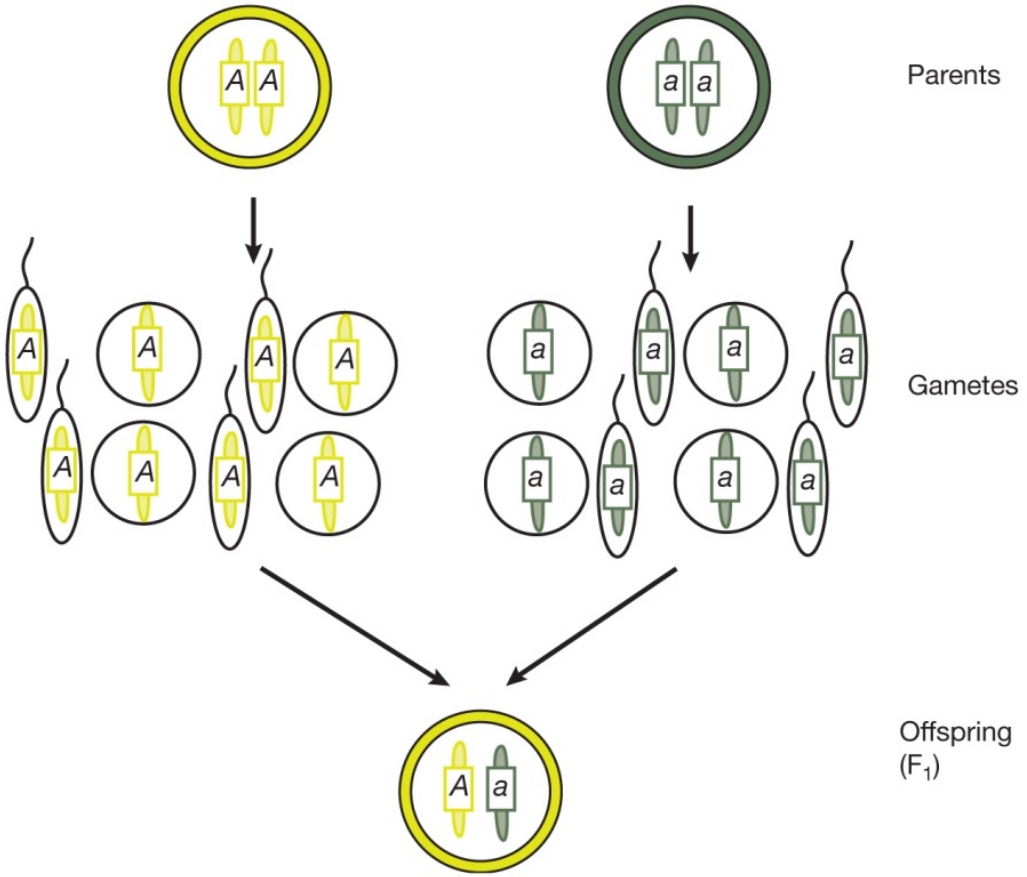
TABLE 2.1

Genotype	Phenotype
AA	Yellow
Aa	Yellow
aa	Green

The relationship between genotype and phenotype in Mendel's experiment on pea color.

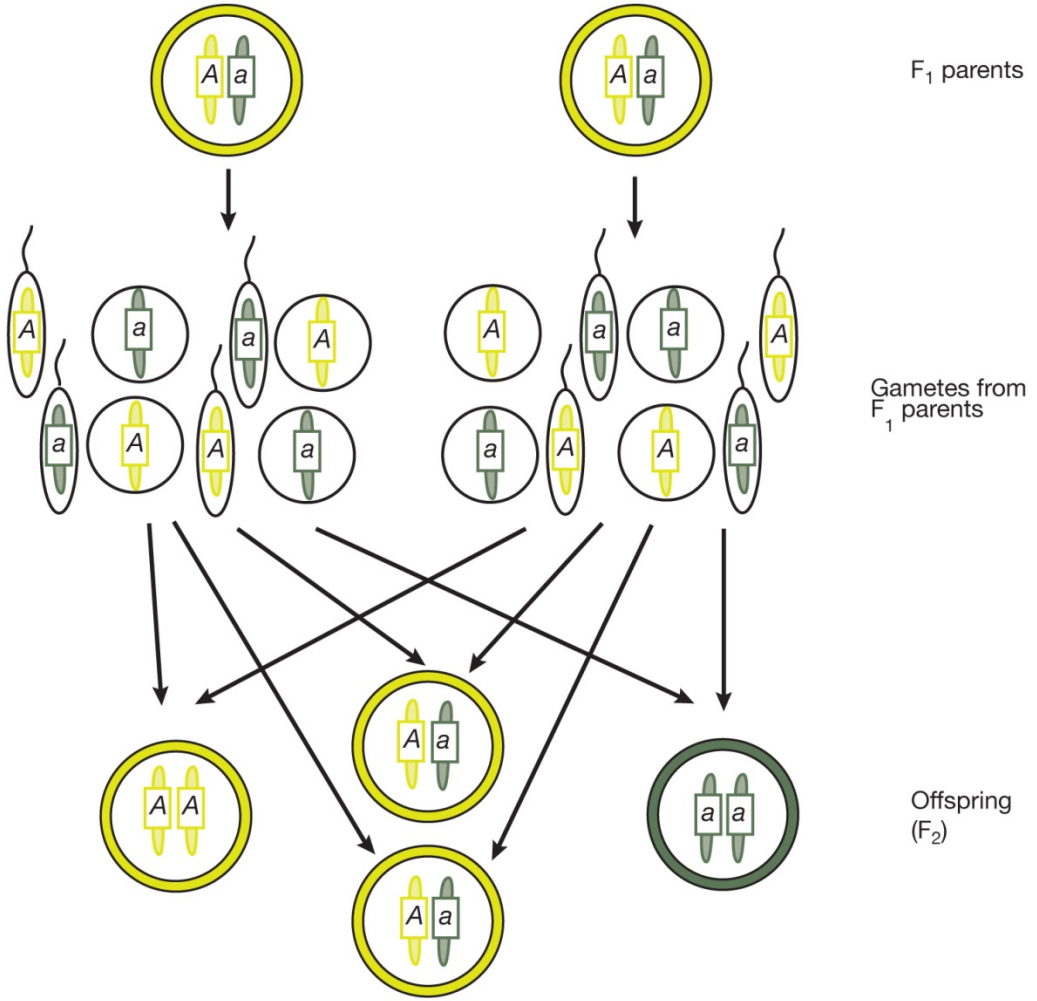
Mendel's Experimental Results

- Dominant allele masks the effects of other alleles for traits
  - Yellow is dominant in this example (A)
- Recessive allele is masked by other alleles for the trait
  - Green is recessive in this example (a)

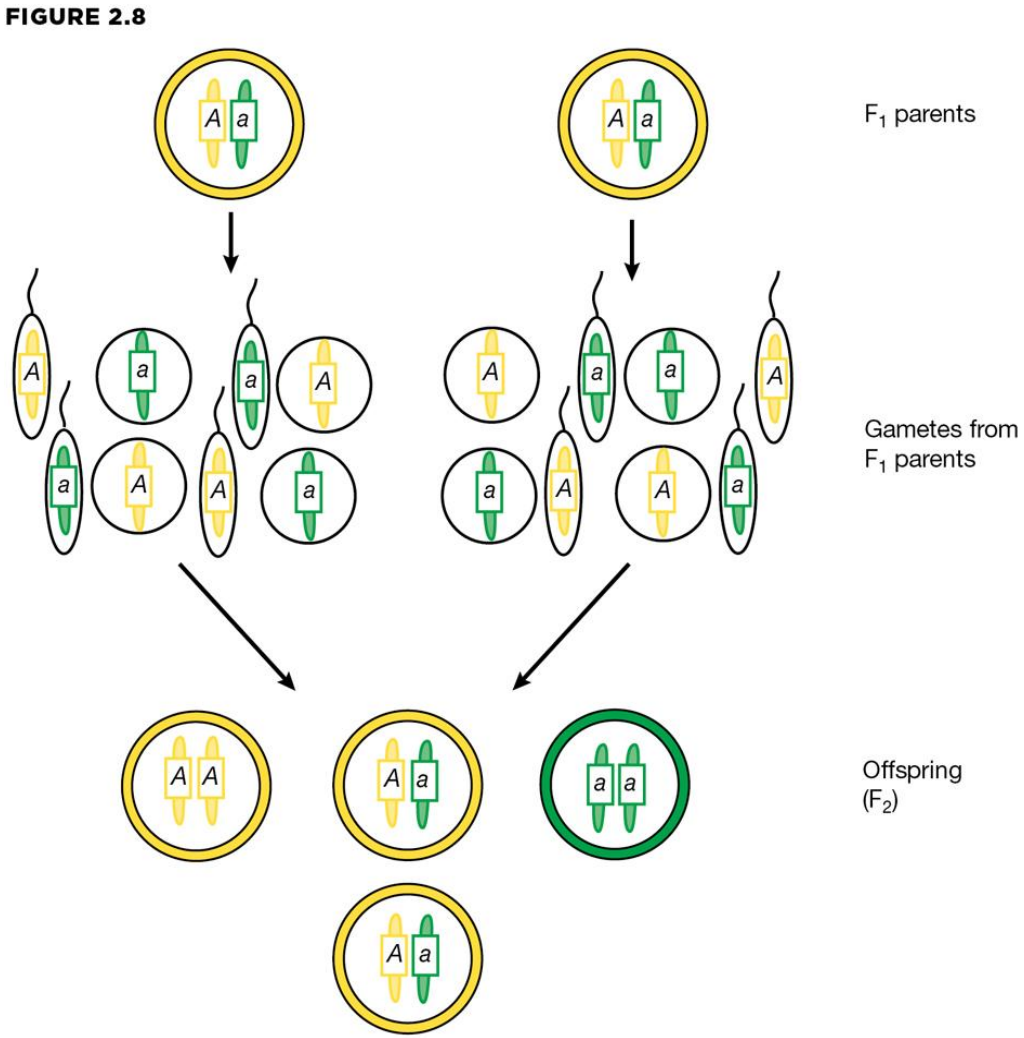


Mendel's Experimental Results

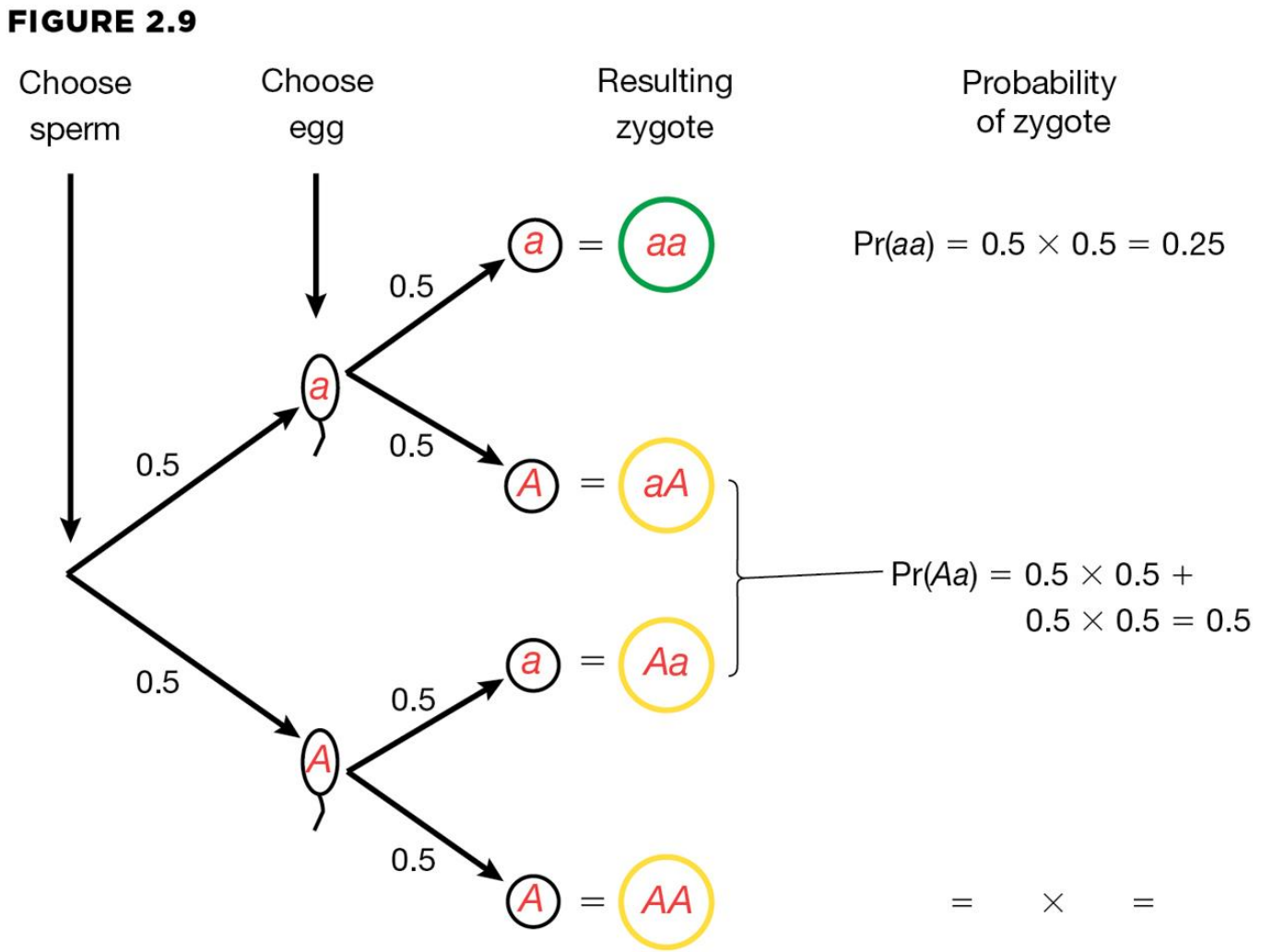
- What happens if we cross heterozygotes?
- What cell division process happens between F1 and gametes from F1?



Mendel's Experimental Results

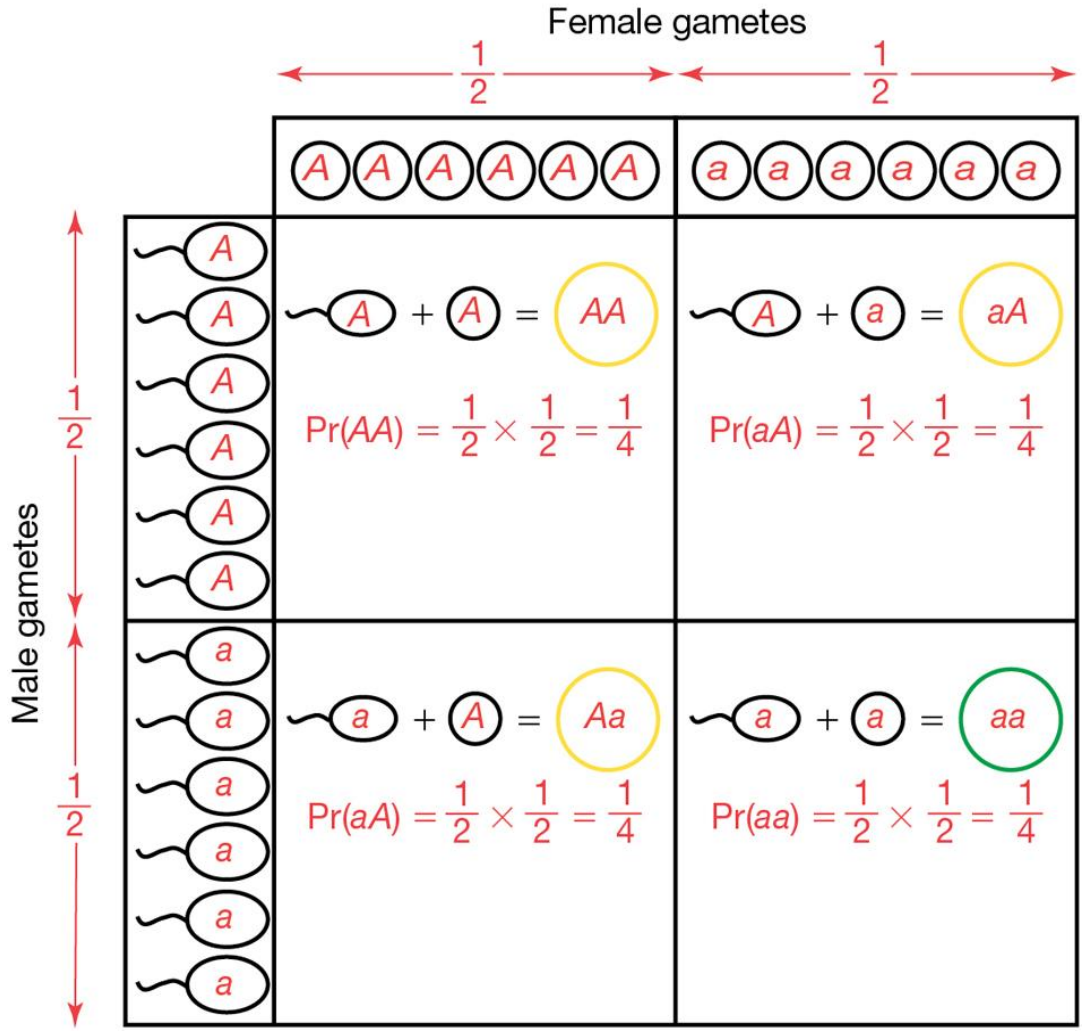


Mendel's Experimental Results: Event Tree



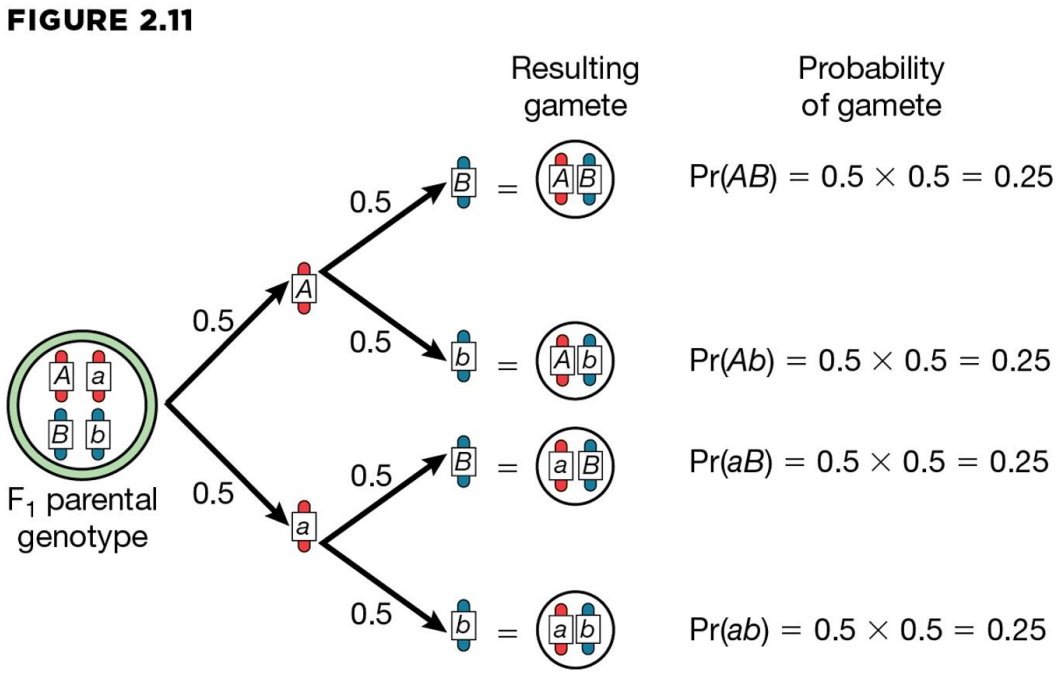
Mendel's Experimental Results: Punnett Square

FIGURE 2.10



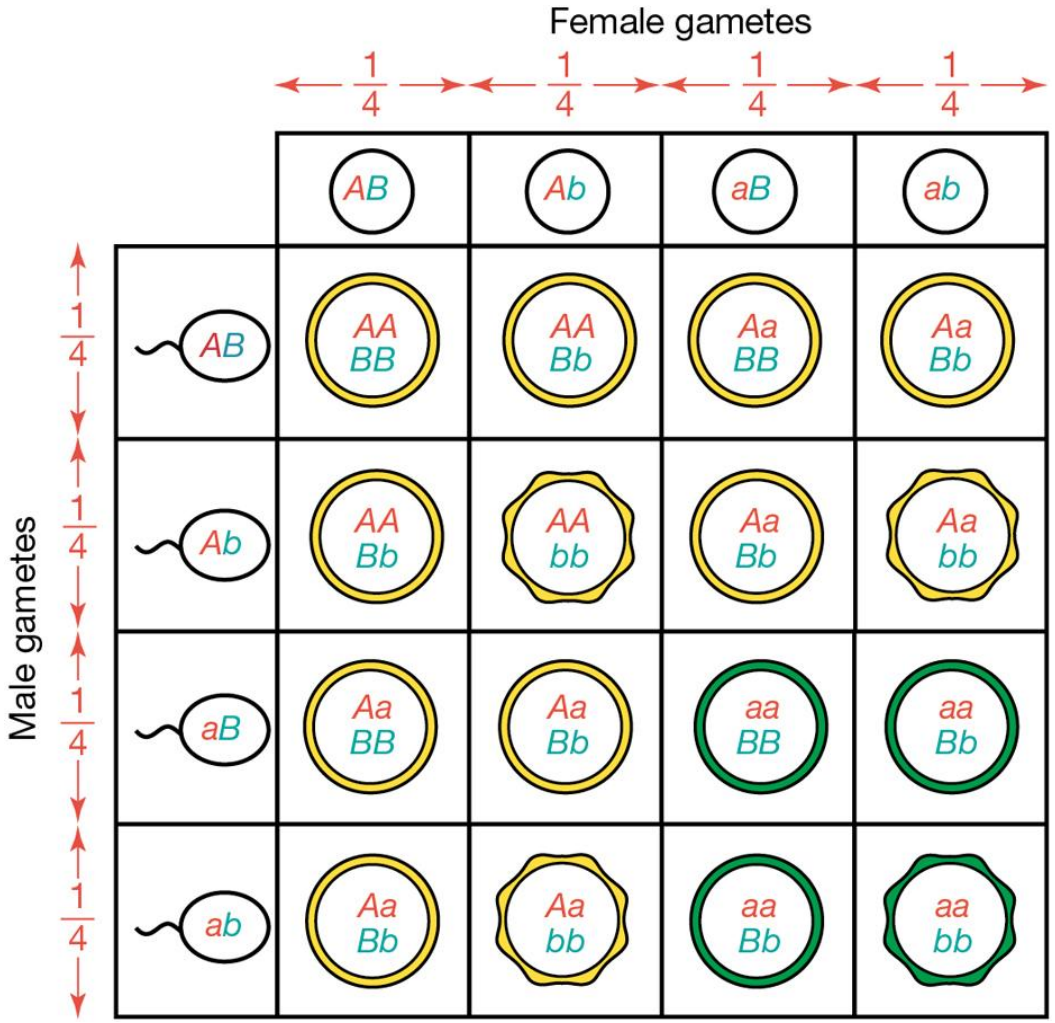
Linkage and Recombination

- Independent assortment of color and texture
- *AABB* × *aabb*
- F_1: *AaBb*
  - Phenotype: yellow and smooth



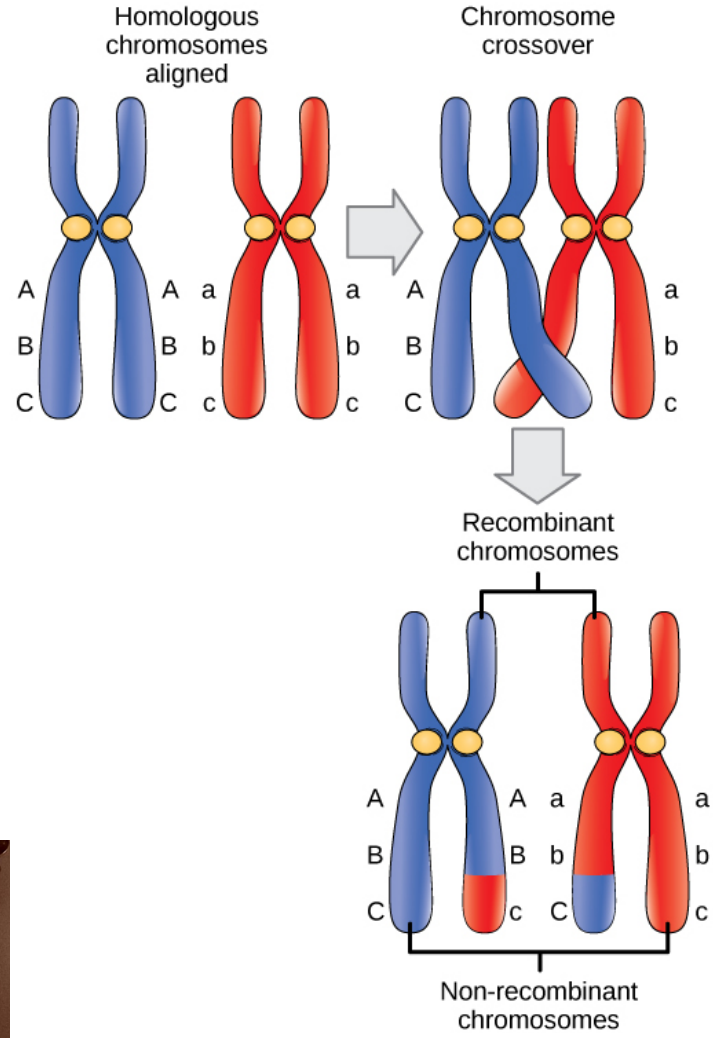
Linkage and Recombination

FIGURE 2.12



Linkage and Recombination

- Recombination: Crossing Over in Meiosis
- Independent assortment of traits in Mendelian traits
  - Genes for texture and color on different chromosomes
- Segregation may not be independent if the loci are physically near one another
- Crossing-over
  - New combinations of genes not present in the parent can result in increased variation!



Linkage and Recombination

FIGURE 2.14

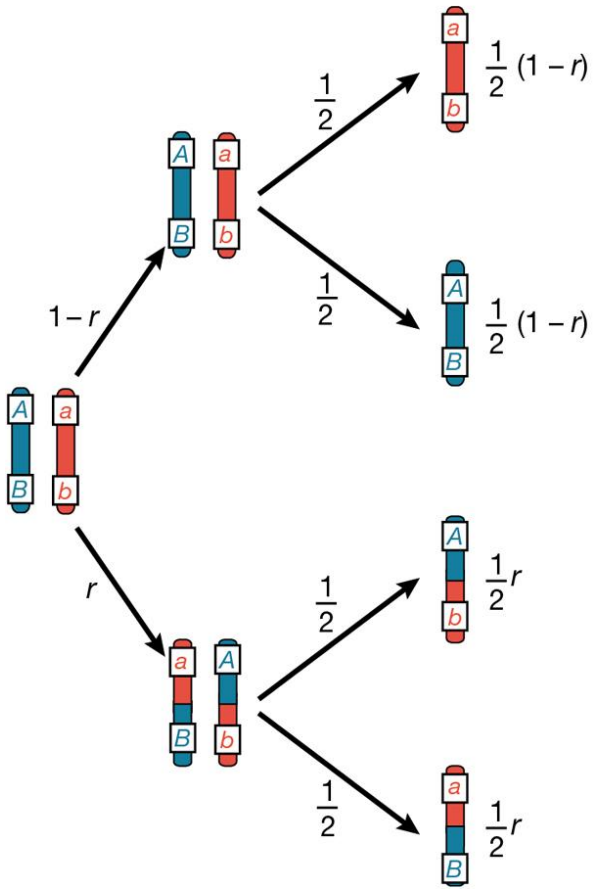
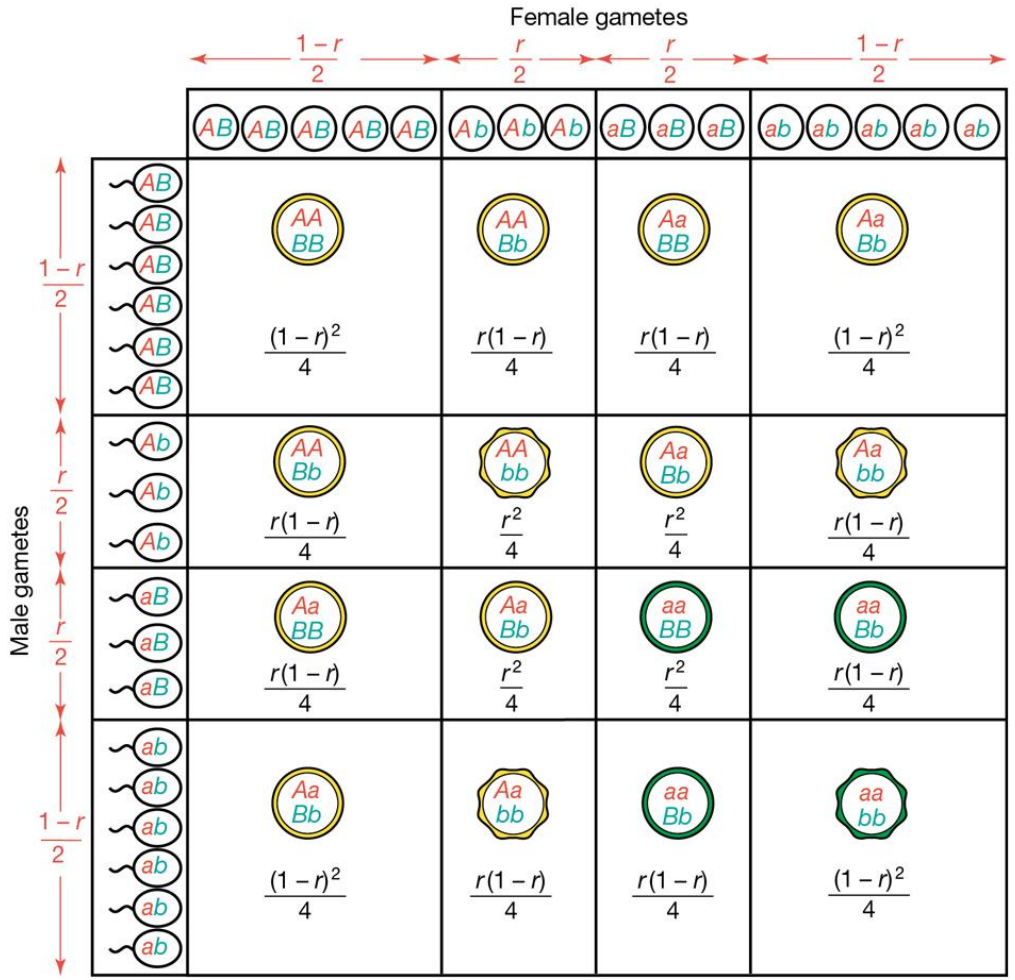
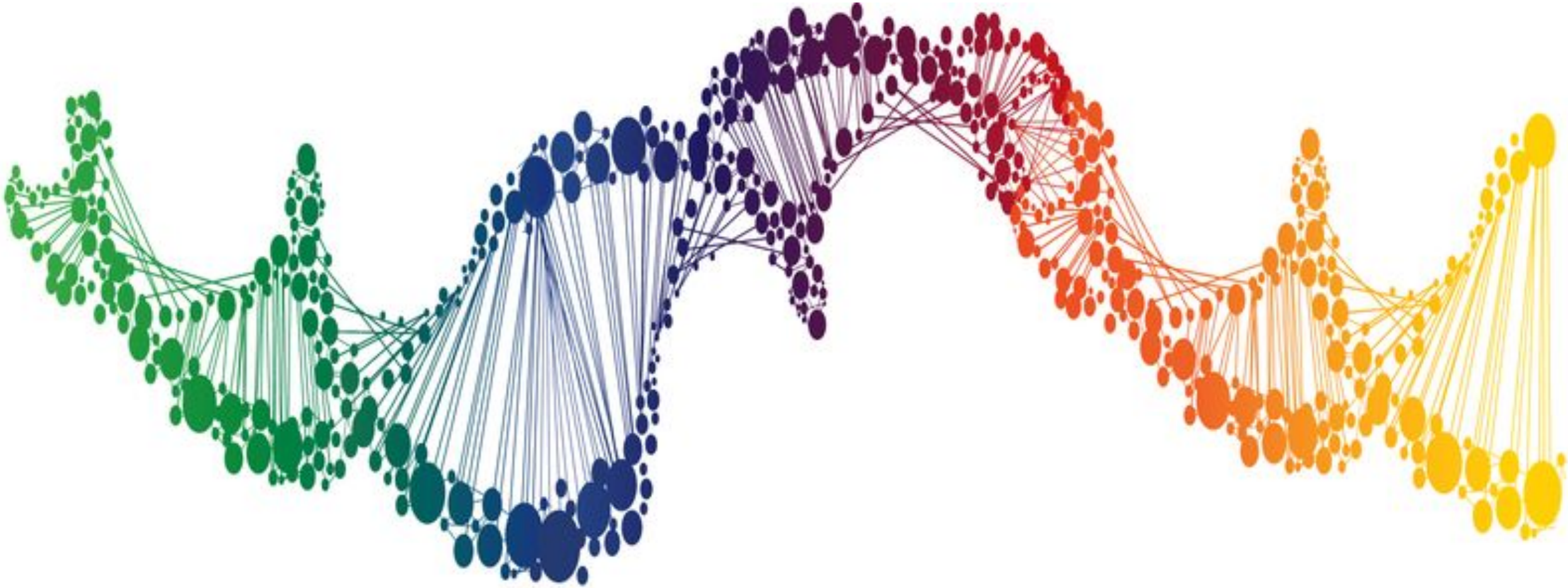


FIGURE 2.15



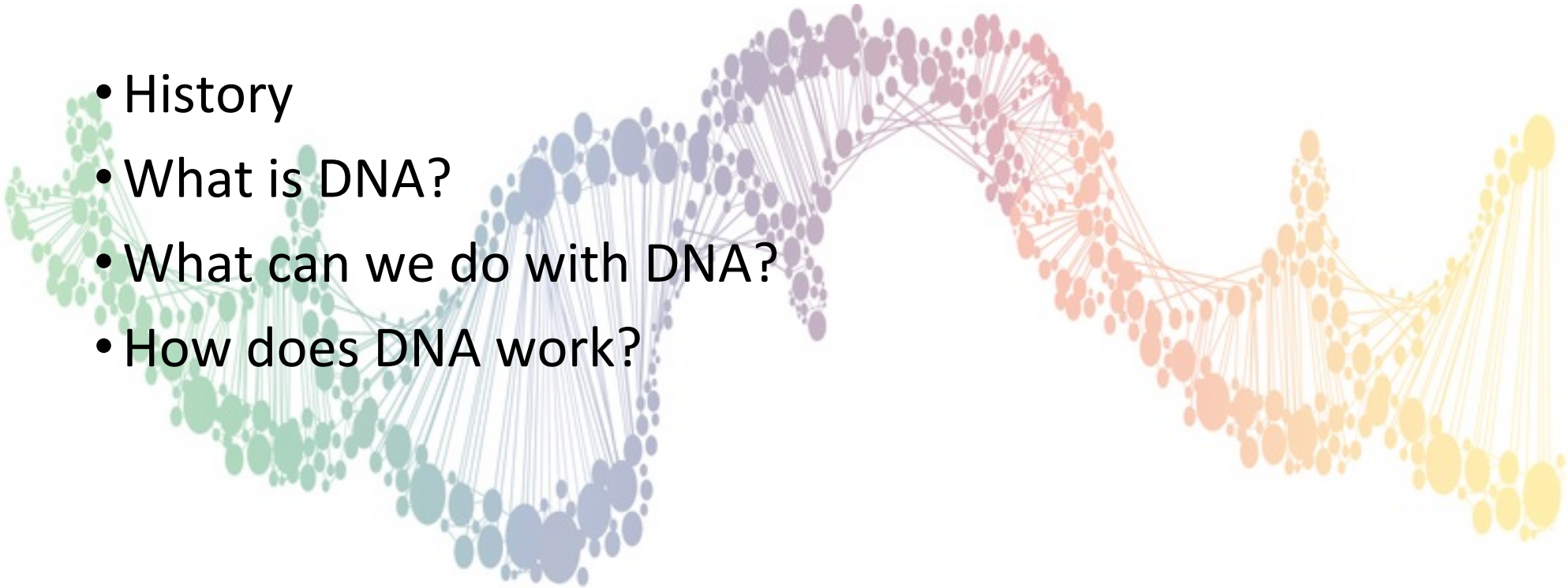
r: Recombination Rate:

The frequency with which a single chromosomal crossing occurs between two genes during meiosis



DNA

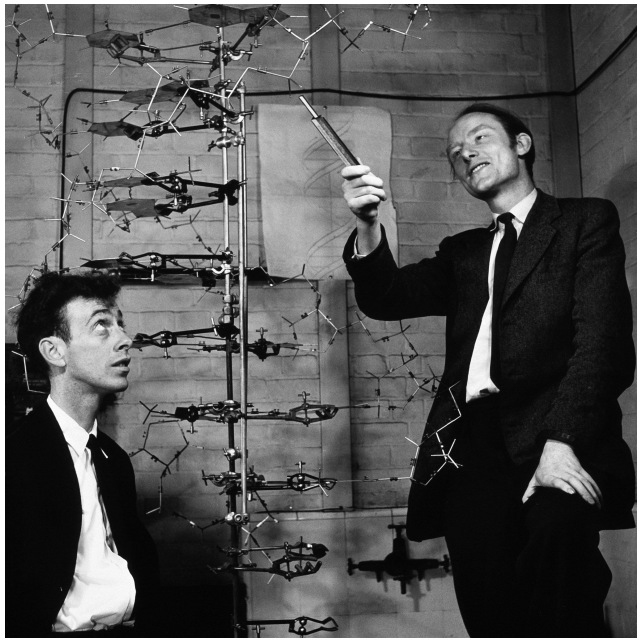
DNA

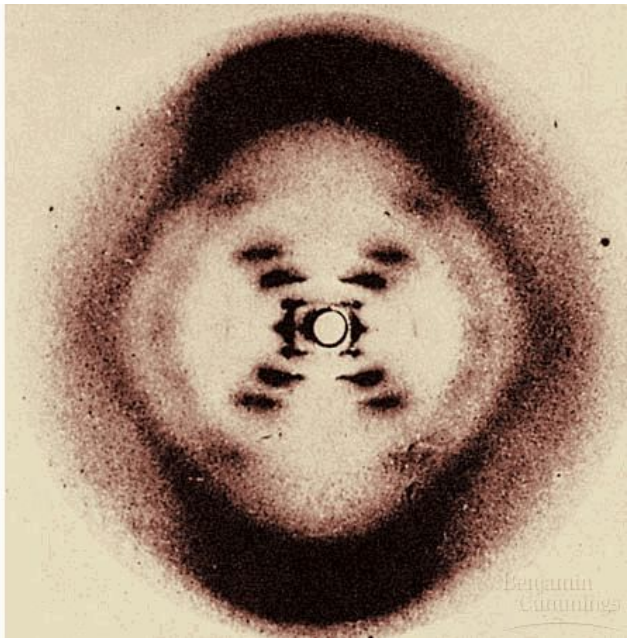


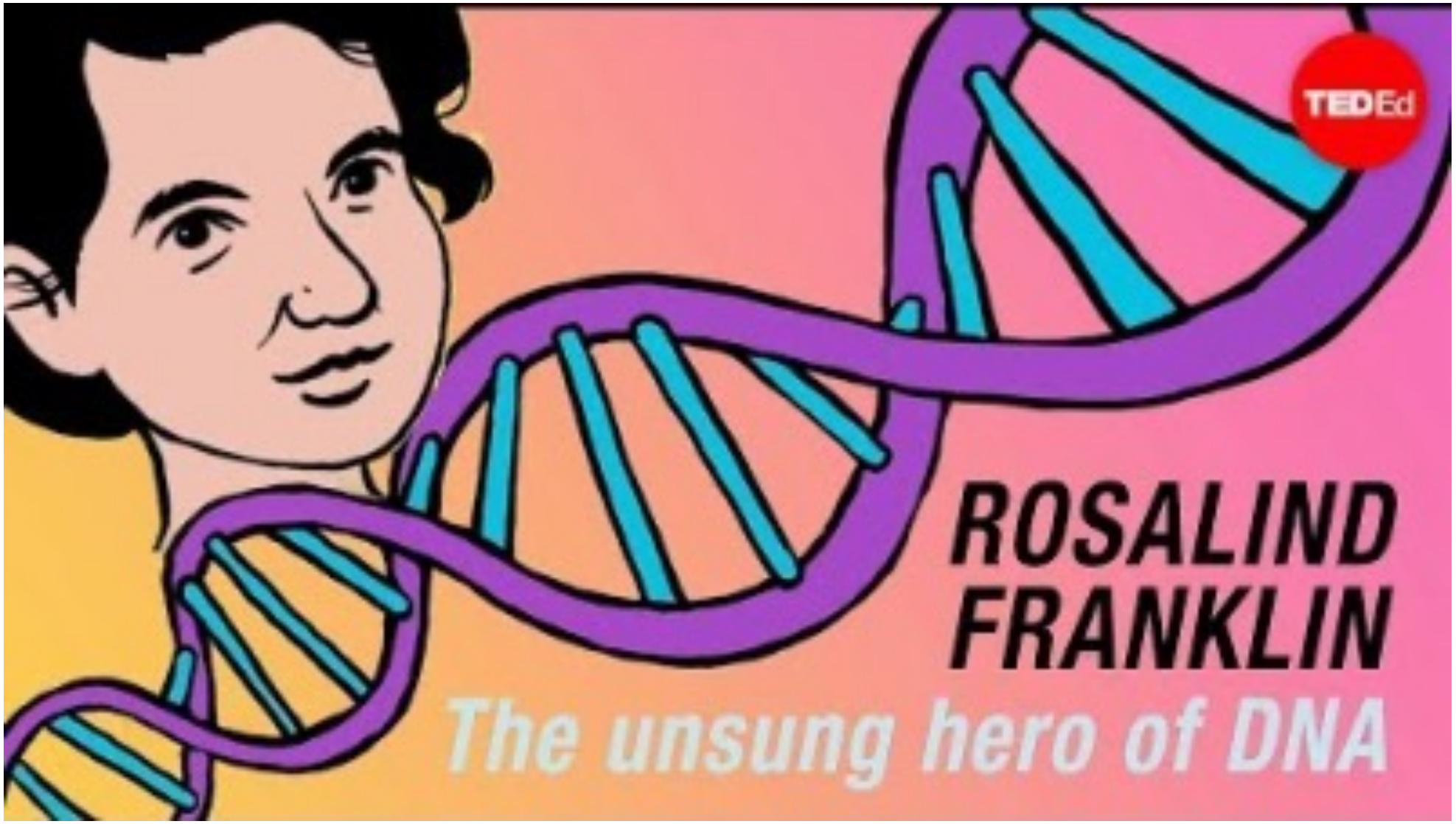
- History
- What is DNA?
- What can we do with DNA?
- How does DNA work?

DNA – A History

- James Watson and Francis Crick published an article in *Nature* in 1953 describing the double-helix structure of DNA (Nobel Prize in 1962)
- Based on the images obtained by Rosalind Franklin using x-rays

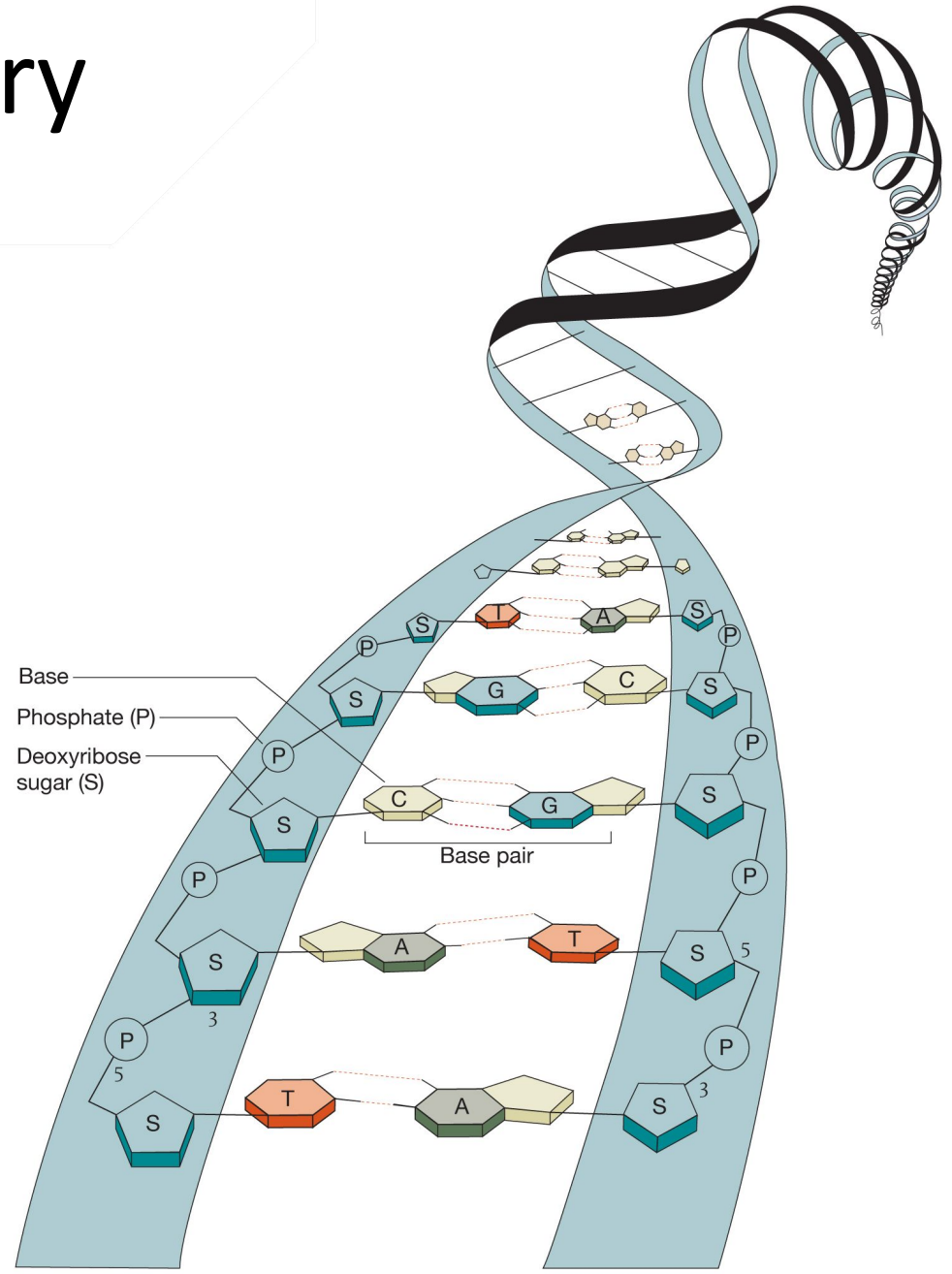





TED-Ed
ROSALIND FRANKLIN
The unsung hero of DNA

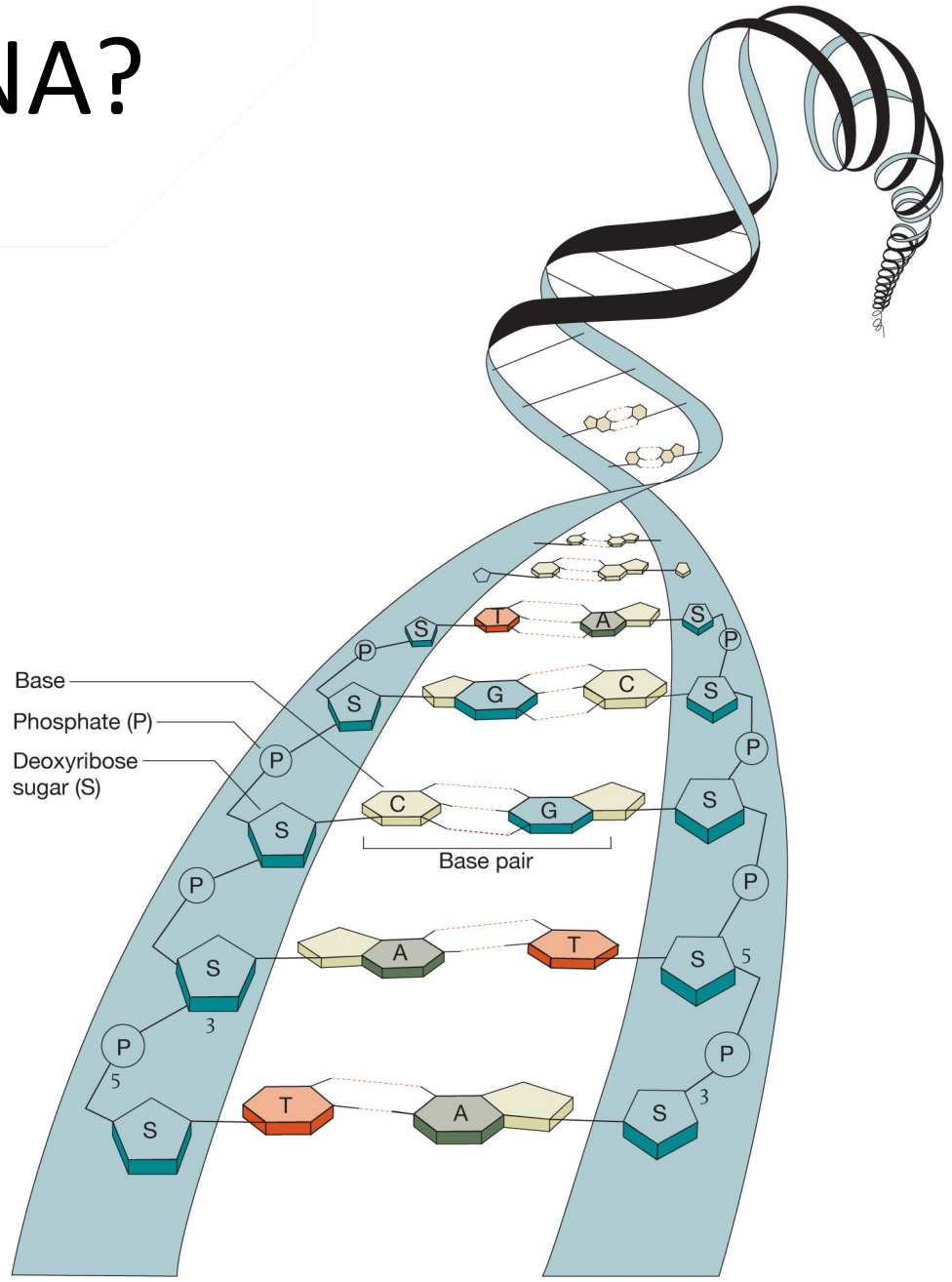
DNA History

- Structure deduced in 1953
  - James Watson, Francis Crick, Rosalind Franklin*
- Understanding structure helps understand function
  - Replication
  - Code for protein formation
- Timing of evolutionary events by comparing DNA of different organisms.
- Causes of evolutionary events



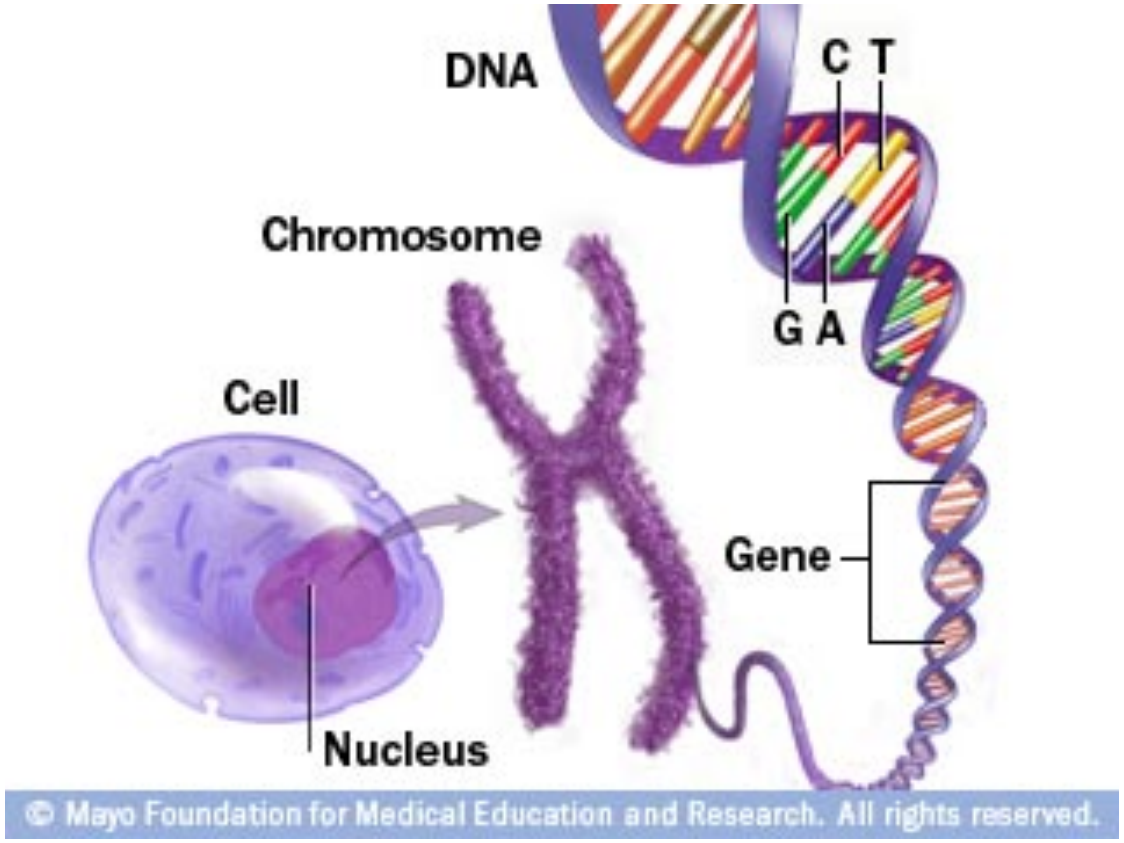
What is DNA?

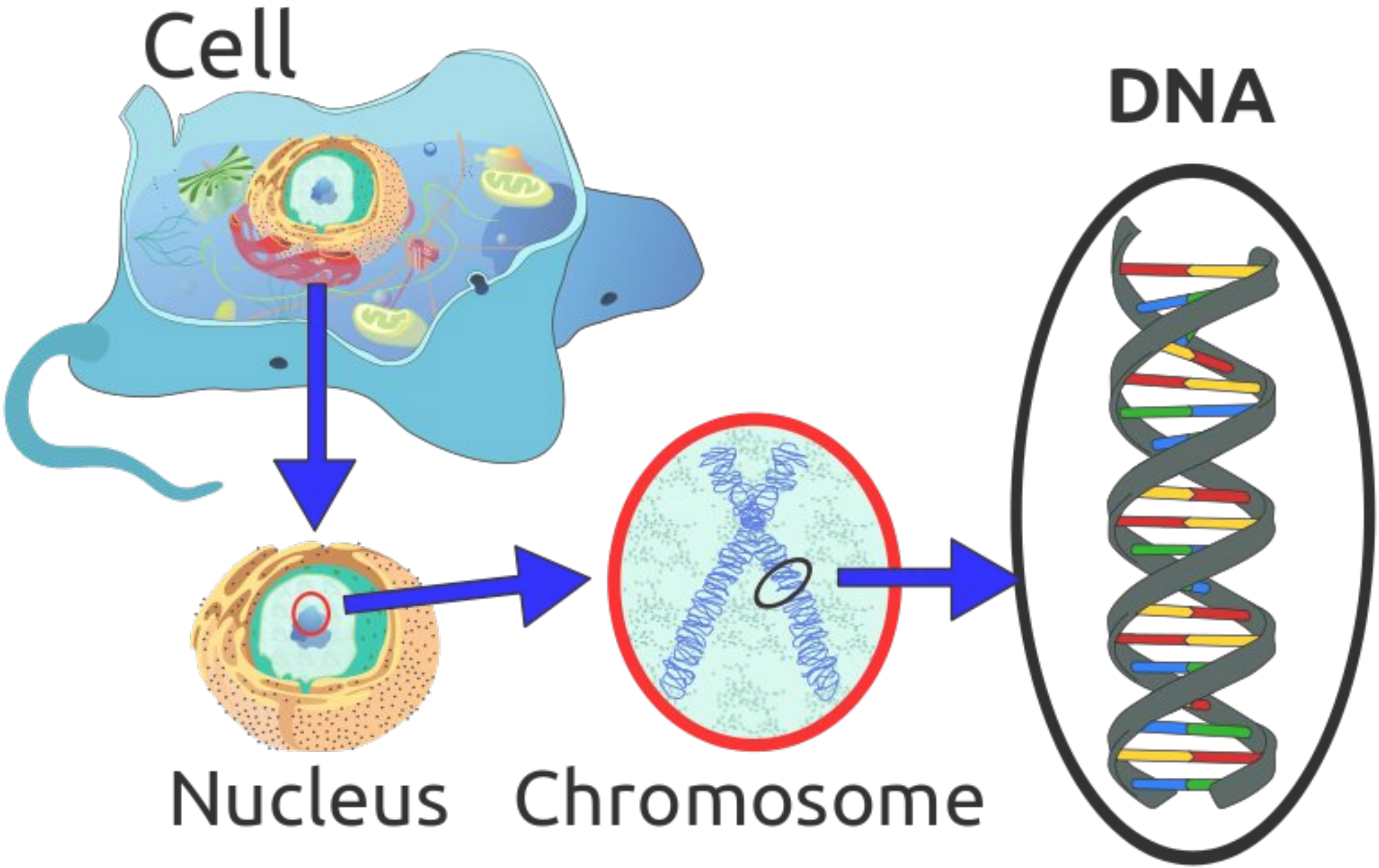
- Deoxyribonucleic acid
- Double helix structure
  - Sugar and phosphate backbone
  - Nucleotide bases
    - Adenine (A)
    - Guanine (G)
    - Cytosine (C)
    - Thymine (T)
    - C always bonds with G
    - A always bonds with T



What are Genes?

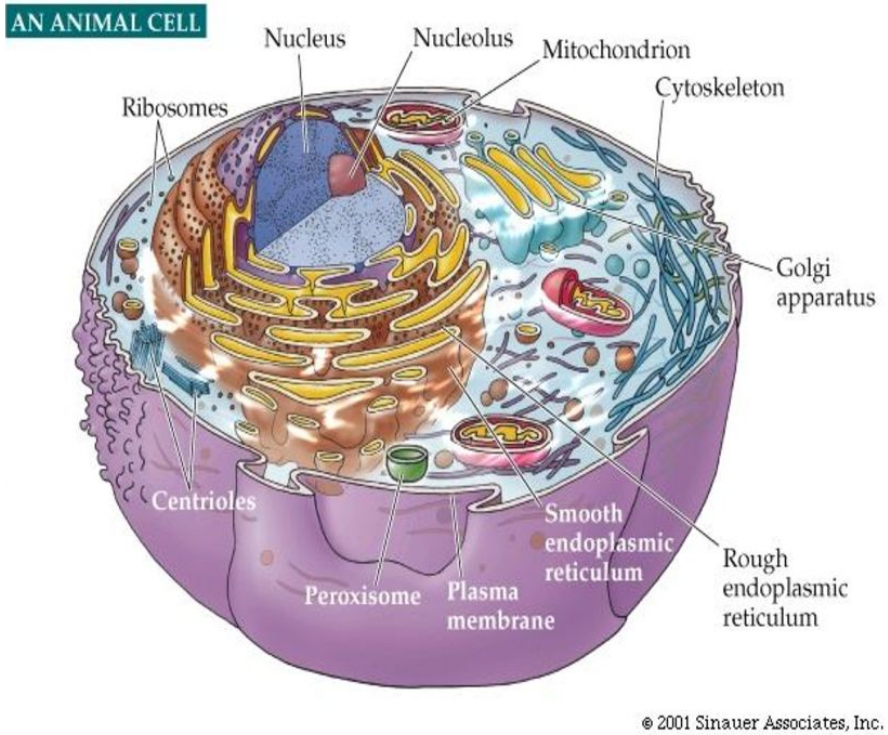
- DNA (some of it) forms genes
- The fundamental unit of heredity
- Sequence of DNA that has a function
- Variants of a gene are called **alleles**
- Humans have 20,000-25,000

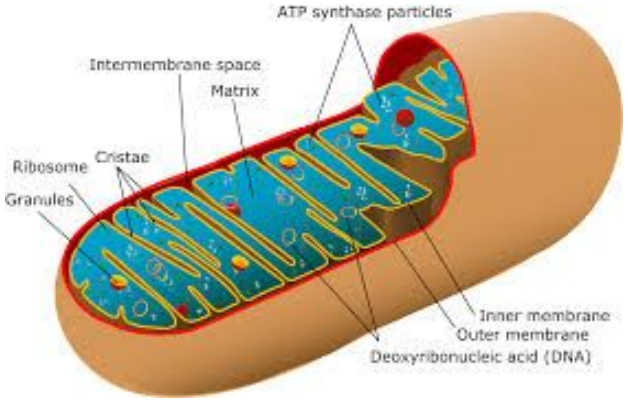



Cell
DNA
Nucleus
Chromosome

Mitochondrial DNA

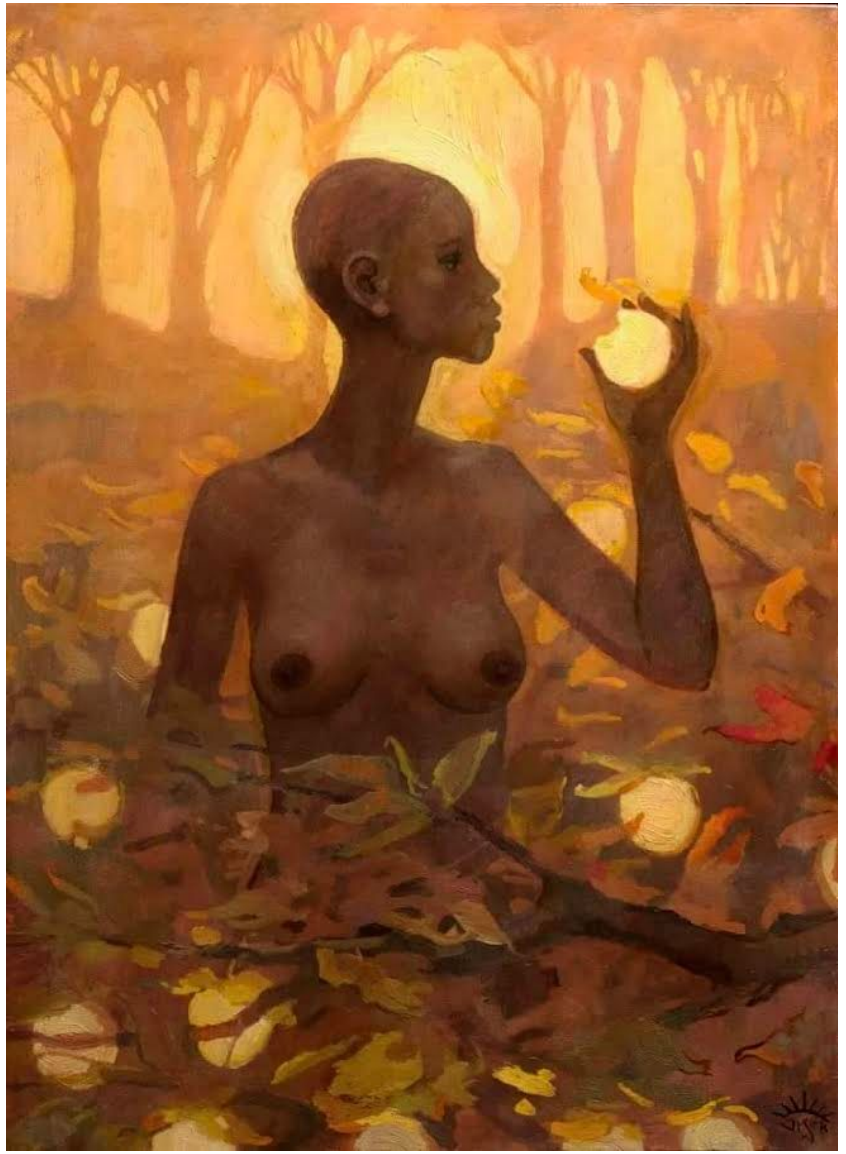
- mtDNA is found in the **mitochondria**
  - first human DNA to be sequenced (only codes for 37 genes)
  - only inherited from mothers, without recombination
  - shorter and more conserved than nuclear DNA





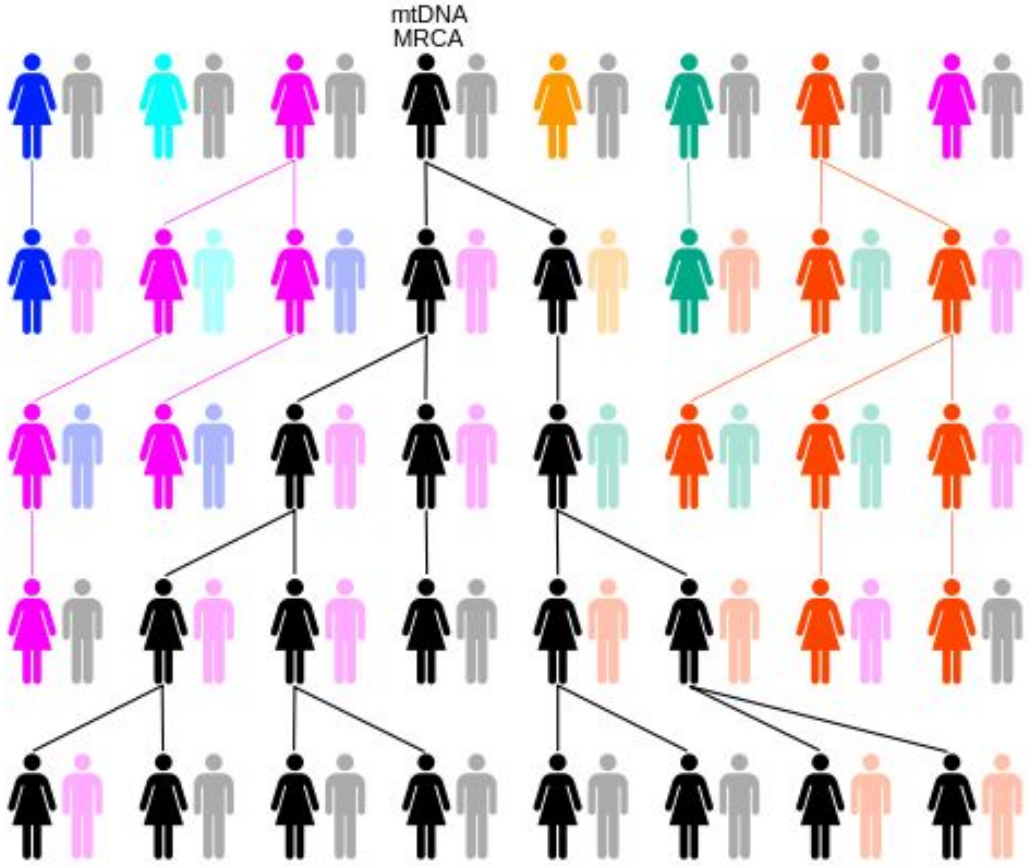
What can we do with DNA?

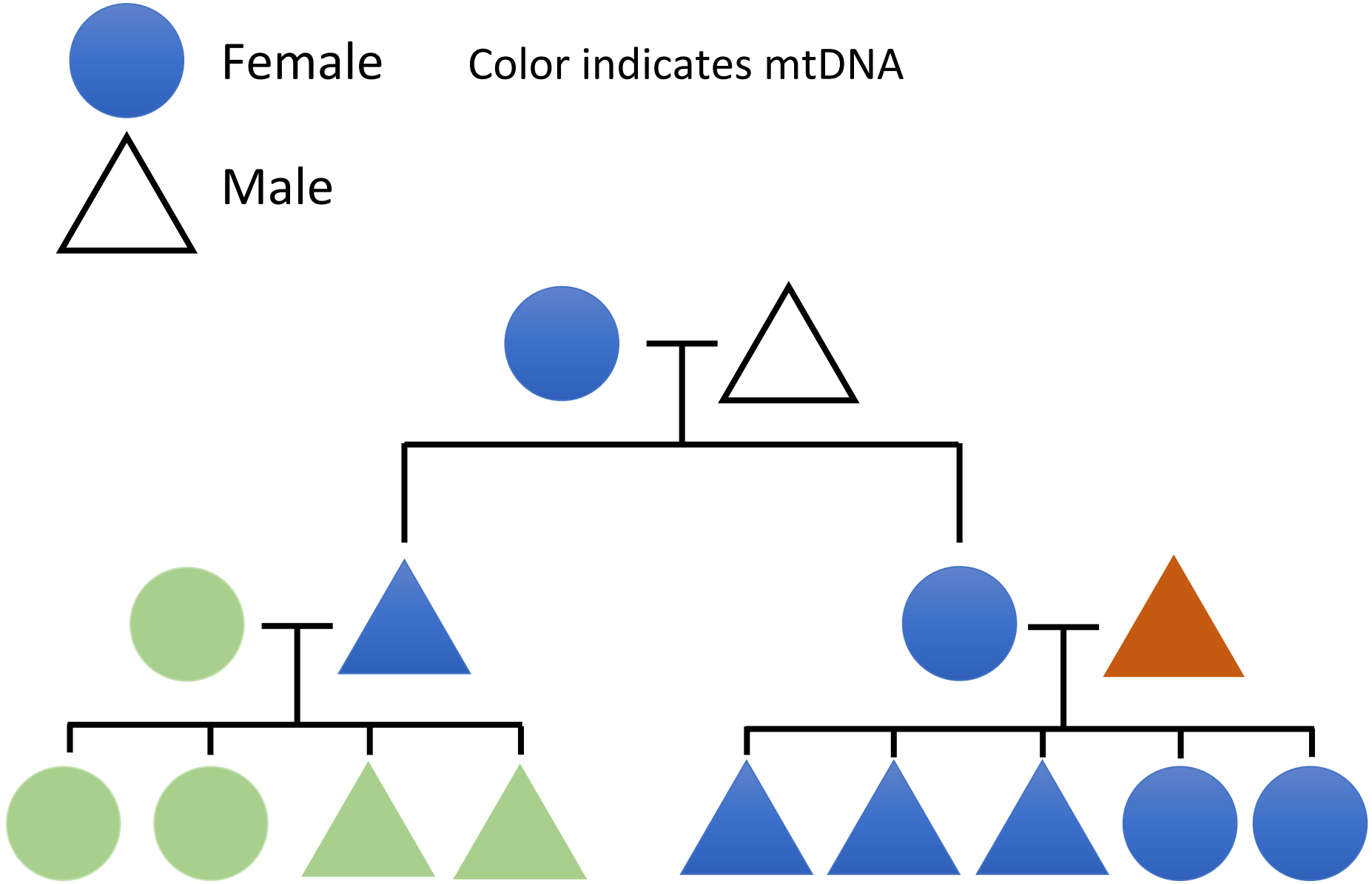
- Estimate age of last common ancestors
  - Mitochondrial Eve
    - Most recent common matrilineal ancestor of all humans
    - 100-200kya in East Africa



Mitochondrial Eve

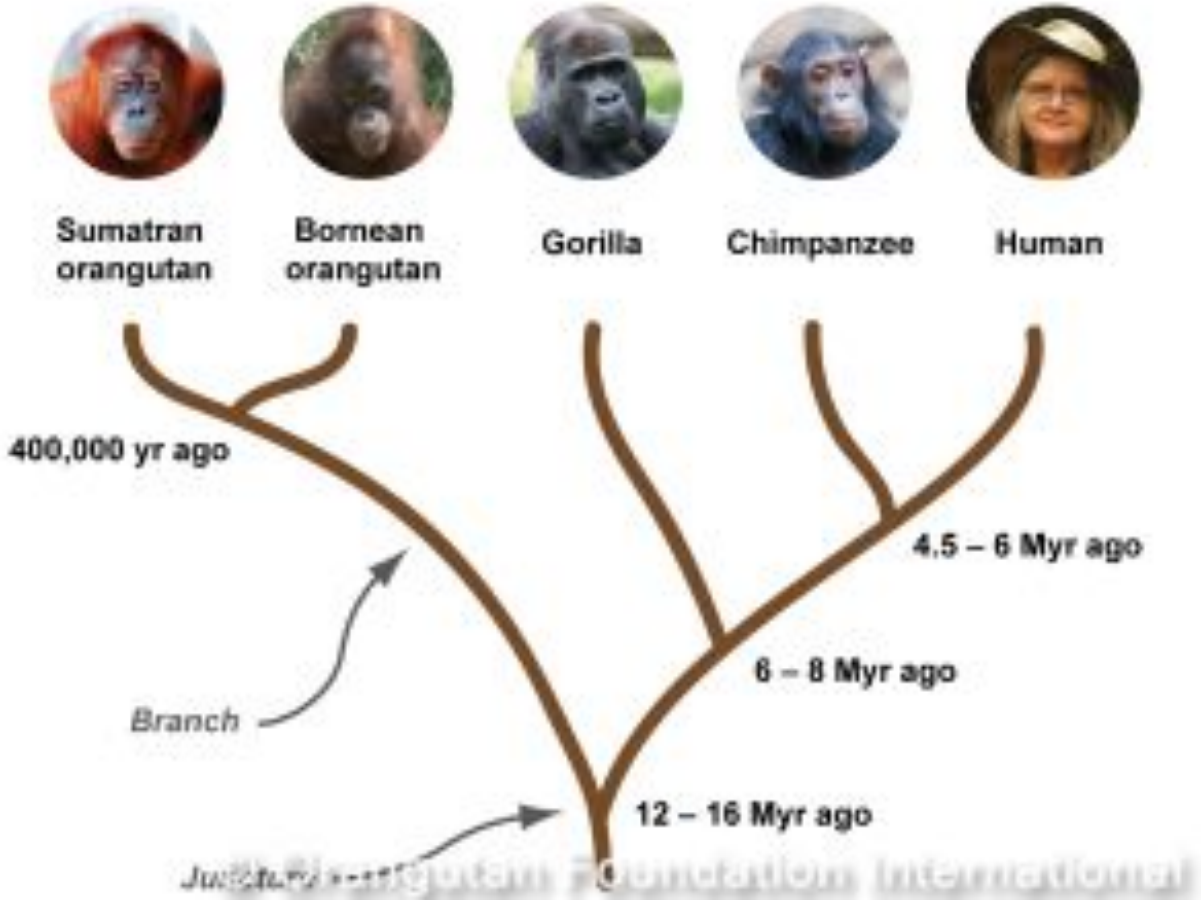
- The most recent ancestor with an unbroken female line.
- Far older the most recent common ancestor of all humans




Female
Color indicates mtDNA
Male

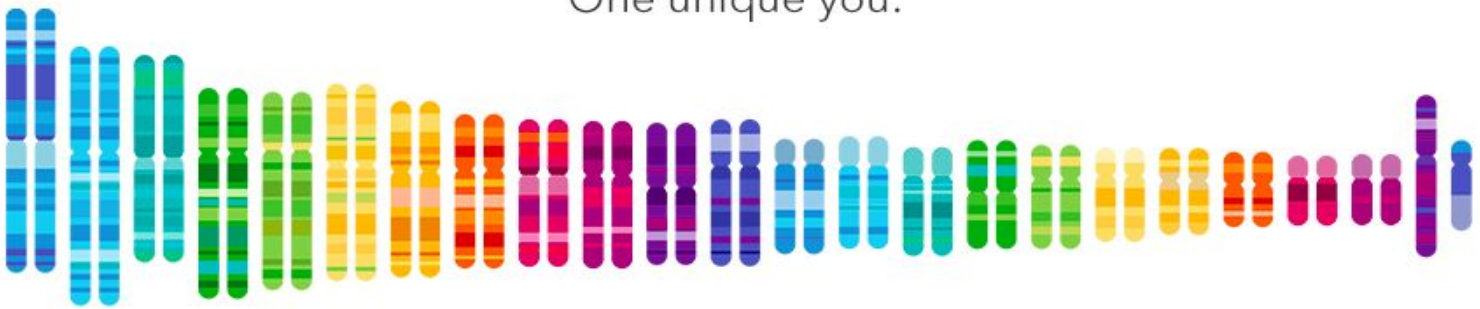
What can we do with DNA?

- Molecular Clock
  - Compare DNA to estimate timing of species divergence
  - Based **neutral sites** (sections of DNA not subject to selection)



23 & Me

23 Pairs of chromosomes.
One unique you.

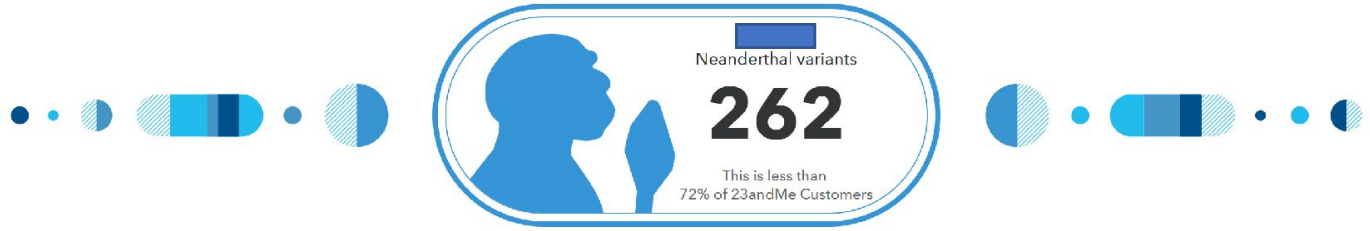


23 & Me

Neanderthal Ancestry

Neanderthals were ancient humans who interbred with modern humans before becoming extinct 40,000 years ago. This report tells you how much of your ancestry can be traced back to Neanderthals.

You have 262 Neanderthal variants.



You have fewer Neanderthal variants than 72% of 23andMe customers.
However, your Neanderthal ancestry accounts for less than 4% of your overall DNA.

23 Pairs of chromosomes.
One unique you.