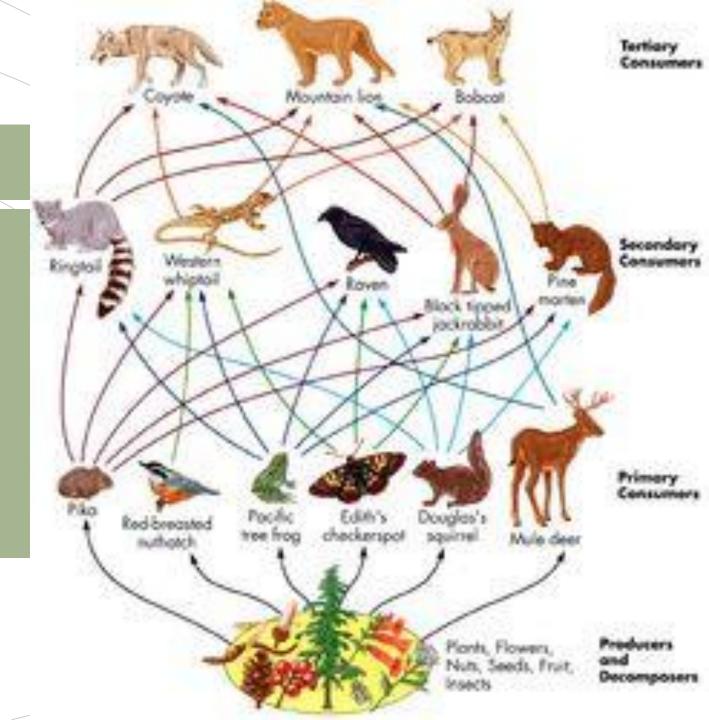


# **Evolution: Genetics**

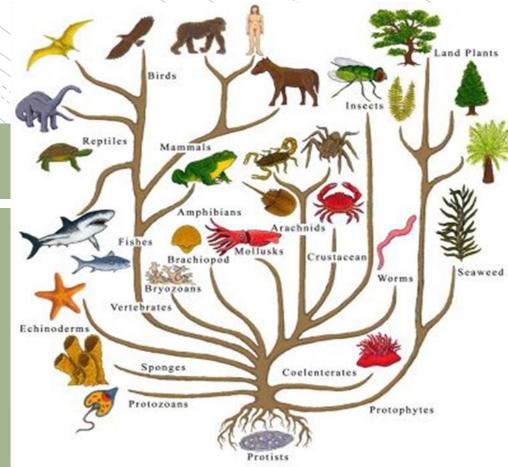
### **Class Questions:**

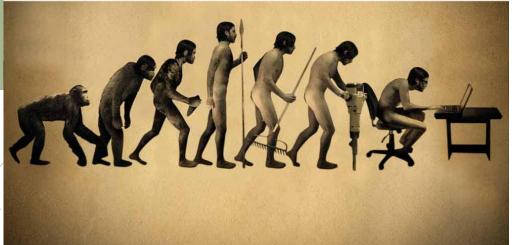
- -What is **Evolution**?
- How and why does evolution occur?
- -What major evolutionary event sparked multicellular life?
- -Which species experience Evolution?



### **Class Questions:**

- -Where did life start?
- -Do all warm blooded animals live on land?
- -What significant research on animals and on DNA itself has given us new insight into evolution?



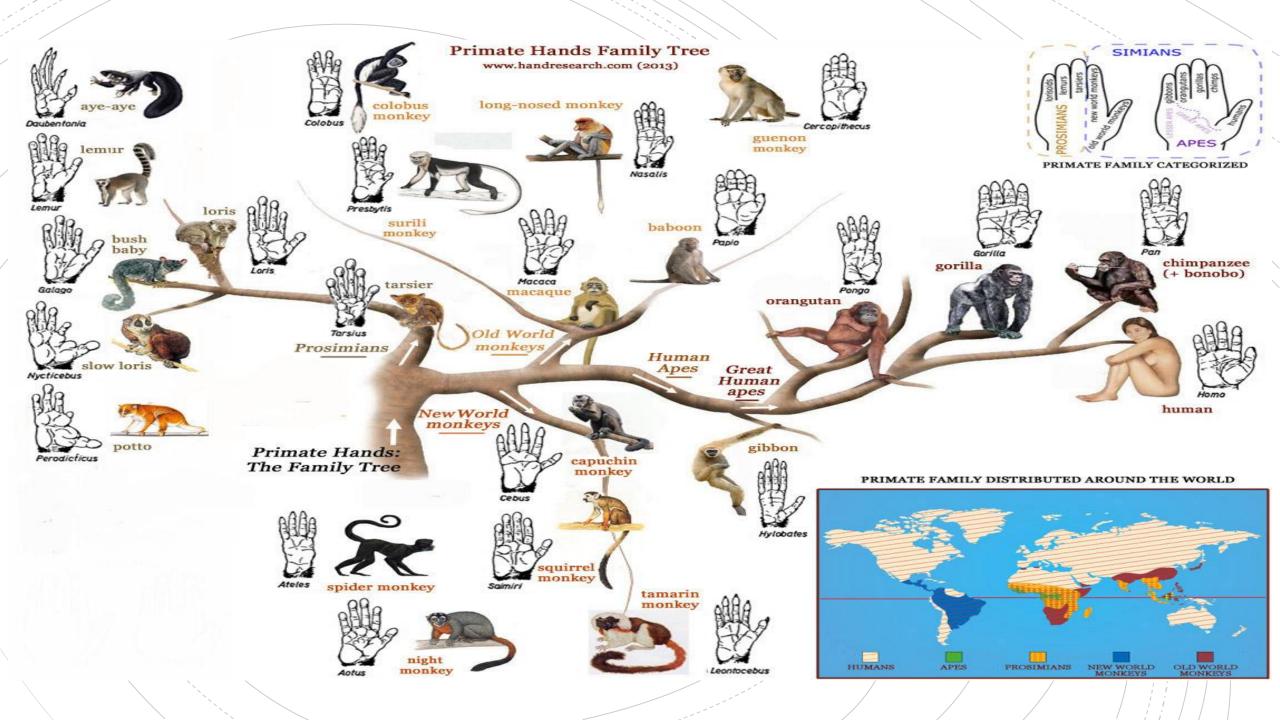








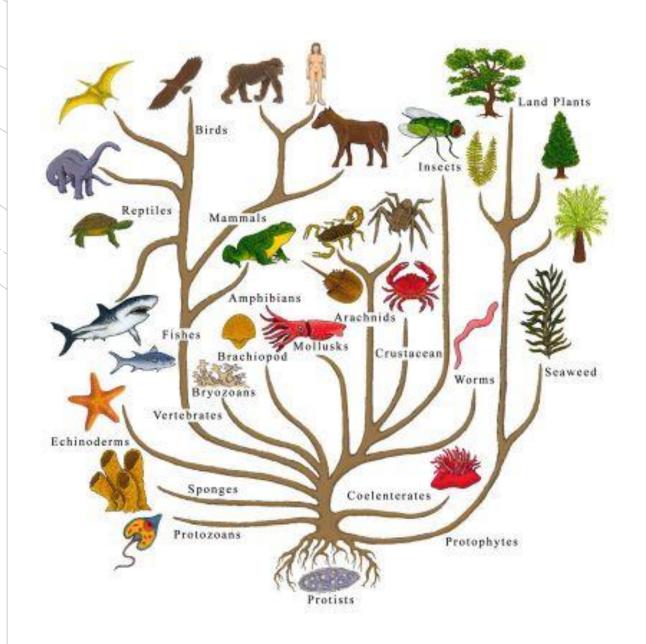
Earth
3.6 billion years old
[As we know it/ [with liquid water]

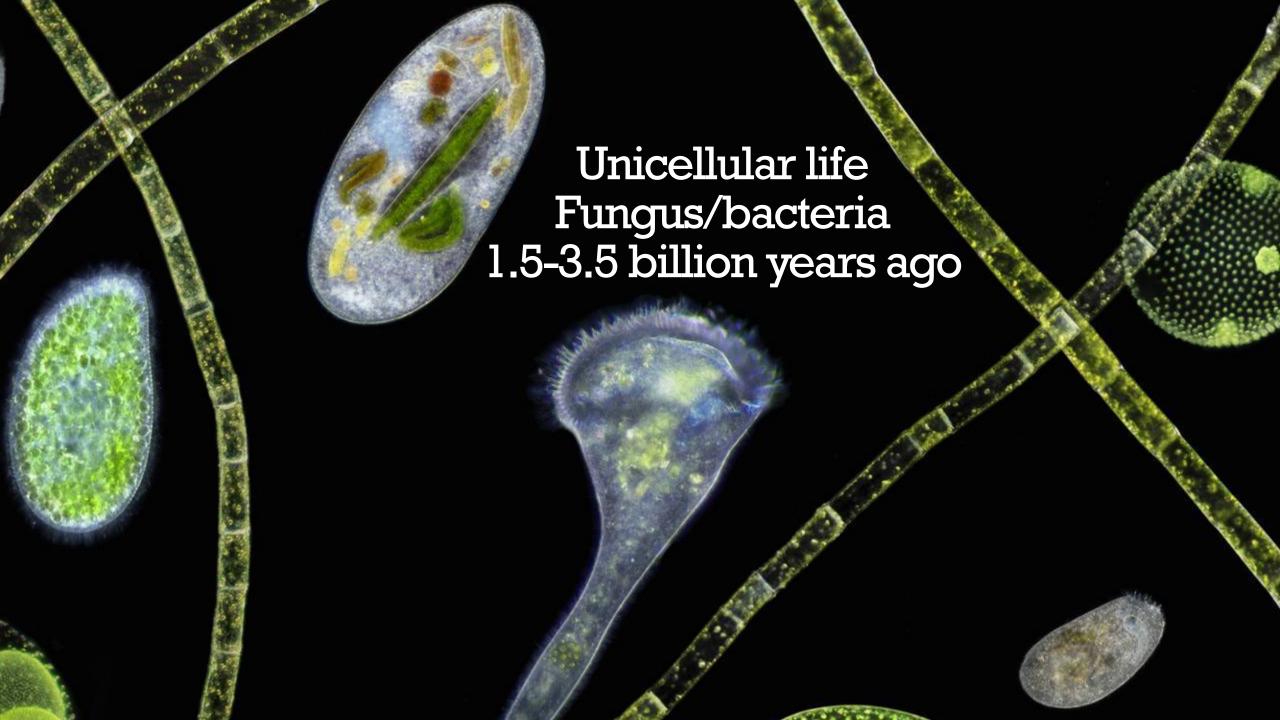


### The Tree of Life

All species on earth evolved from prokaryotic organisms.

Humans are apart of this tree – more current model than the famous 'line' of monkeys/apes merging into humans.







### The Origin of Life:

The Ocean!

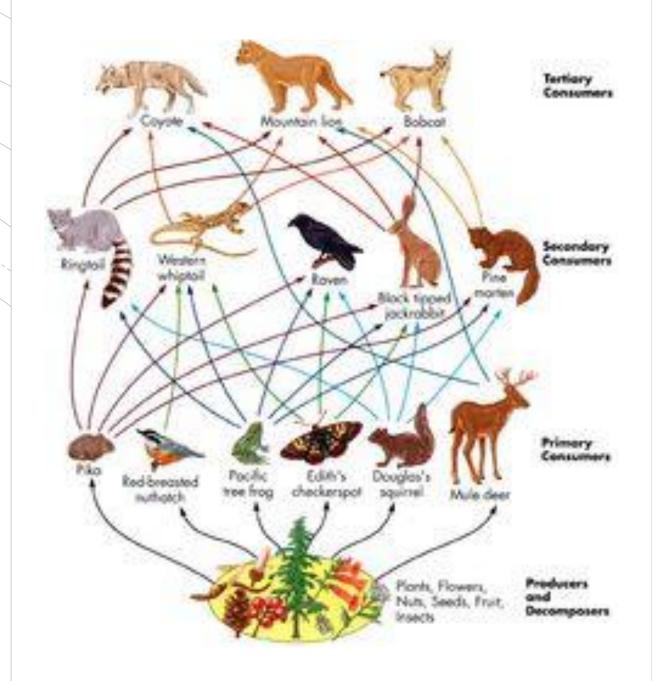




The Cambrian Explosion
-Multicellular life
800 million years ago

### **Animal Evolution**

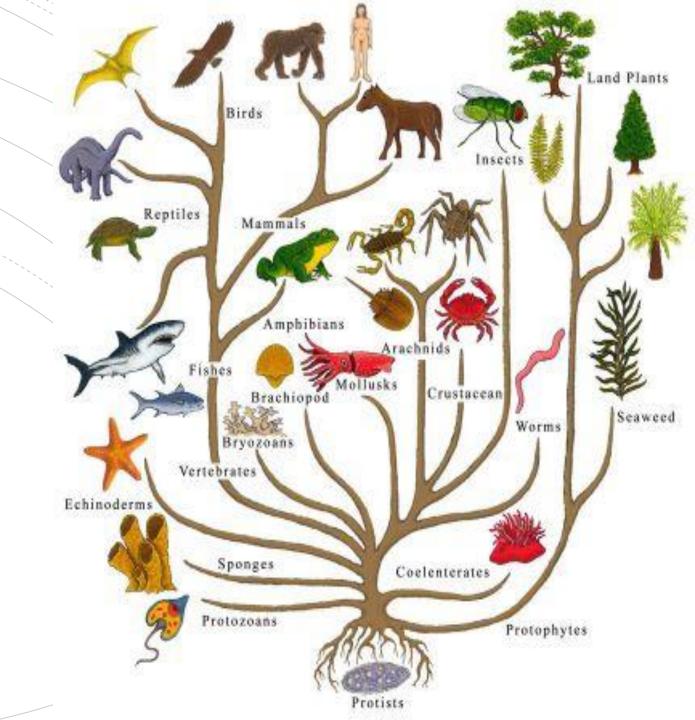
- Transformation of species over many generations that leads to species-change
- Reproductive descent with modification /inheritance of traits through parental lineage



### Macro-evolution:

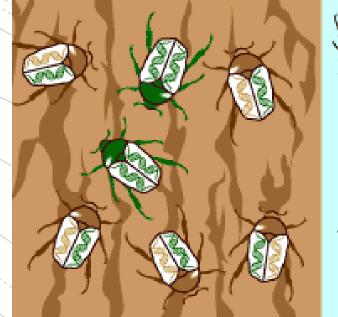
**Species:** "Group of related organisms whose members can interbreed to produce offspring that can live and reproduce" (Kottak 2017)

**Speciation:** "the formation of new species" (Kottak 2017)

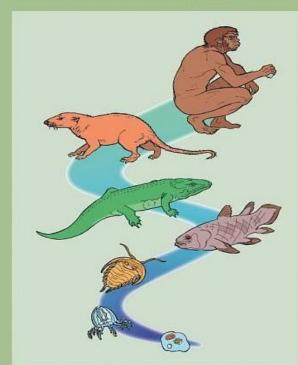


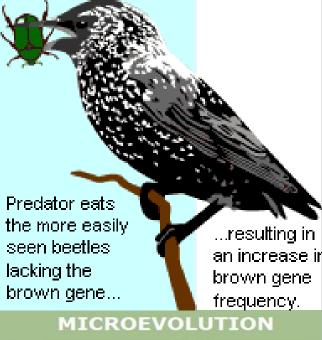
### Micro-evolution:

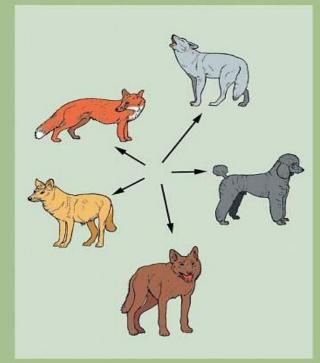
Microevolution: evolutionary shifts in a single species that occur over a few generations / during a relatively short time span.













### **Human Origins: Timeline**

250 million years ago – No birds or mammals on Earth

**2,300,000 years old -** Genus: Homo

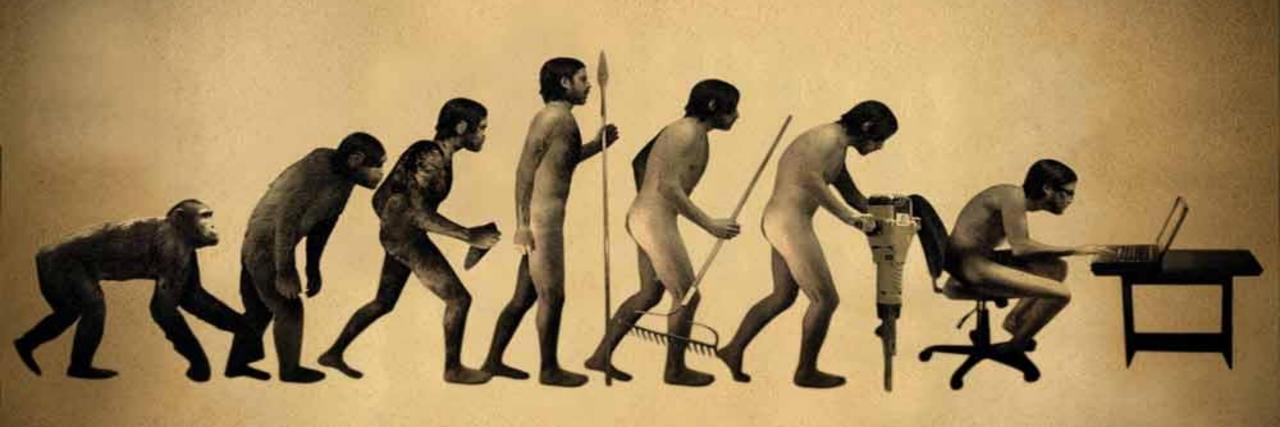
**200,000 years old** – Homosapiens

**120,000-130,000 years** – Symbolic thought/language appears

**74,000 years ago** – <igration outside Africa to Indonesia [volcanic winter]

**12,000 years ago**— Agriculture

**12,000-14,000 years ago** – Settling of the future Americas [via Bering Strait]



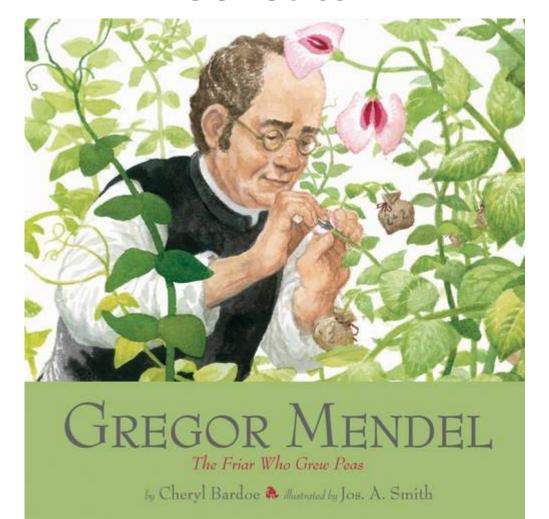
Evolution = only humans? [Nope!]
Progress? [Questionable!]

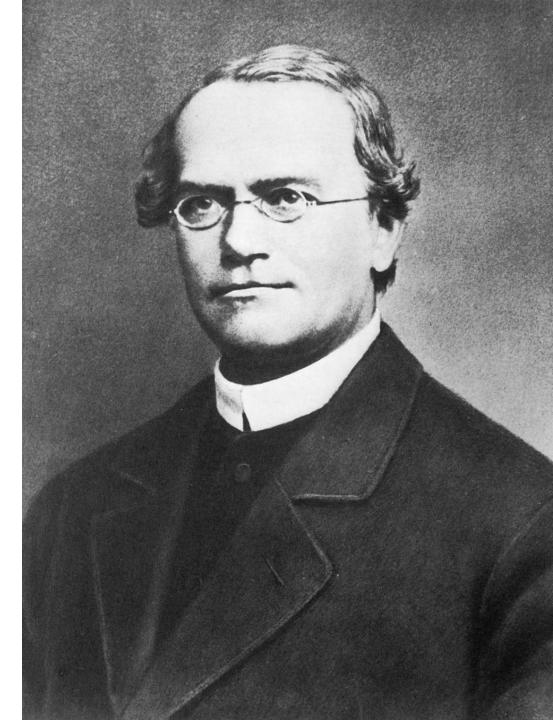
### Early Genetics Research

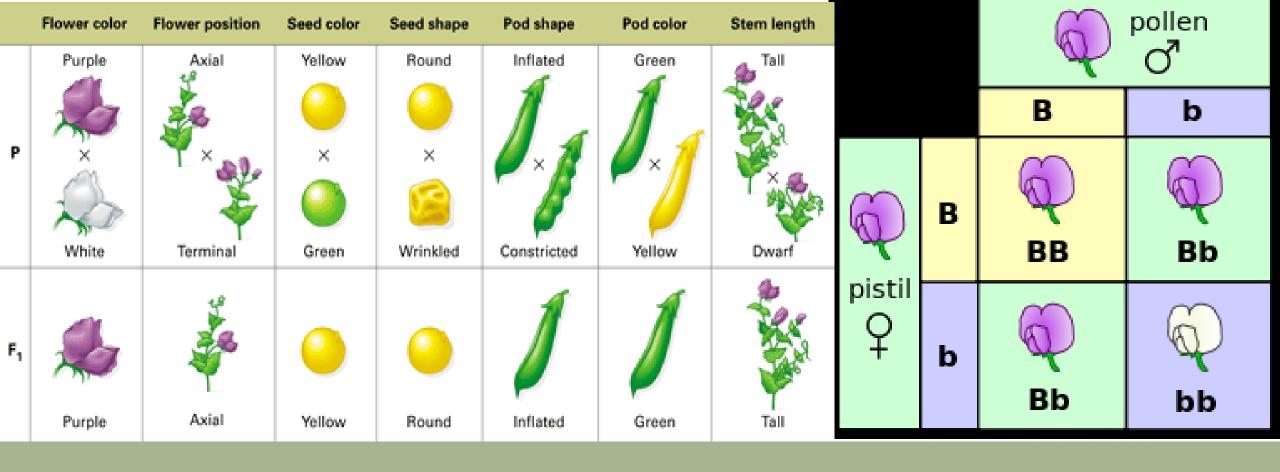
Mendel and Darwin, observing the natural world

1822 - 1884

# Gregor Mendel "The Father of Modern Genetics"

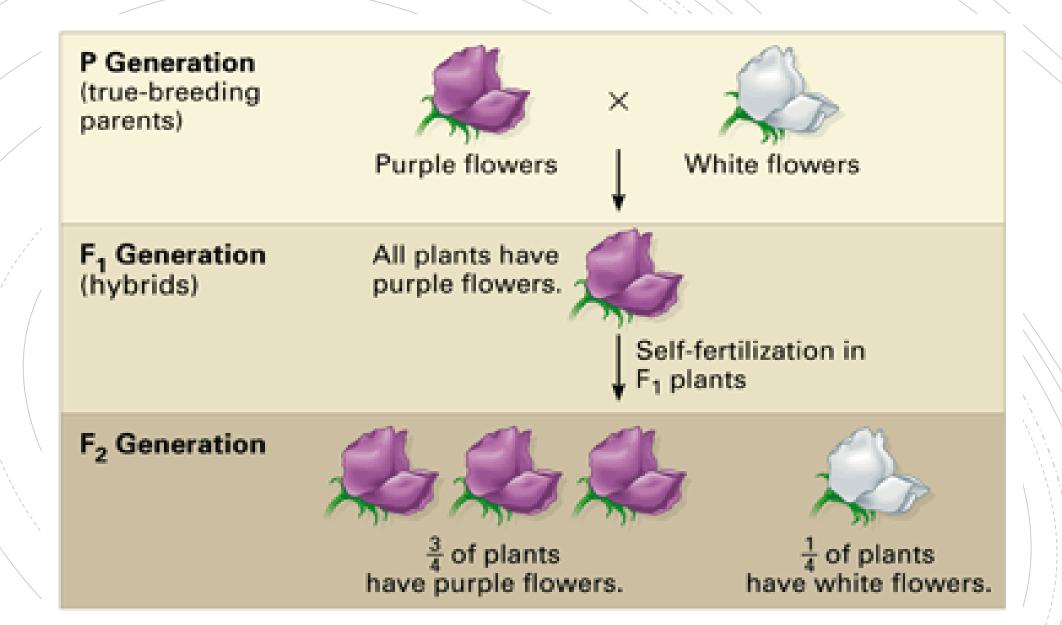




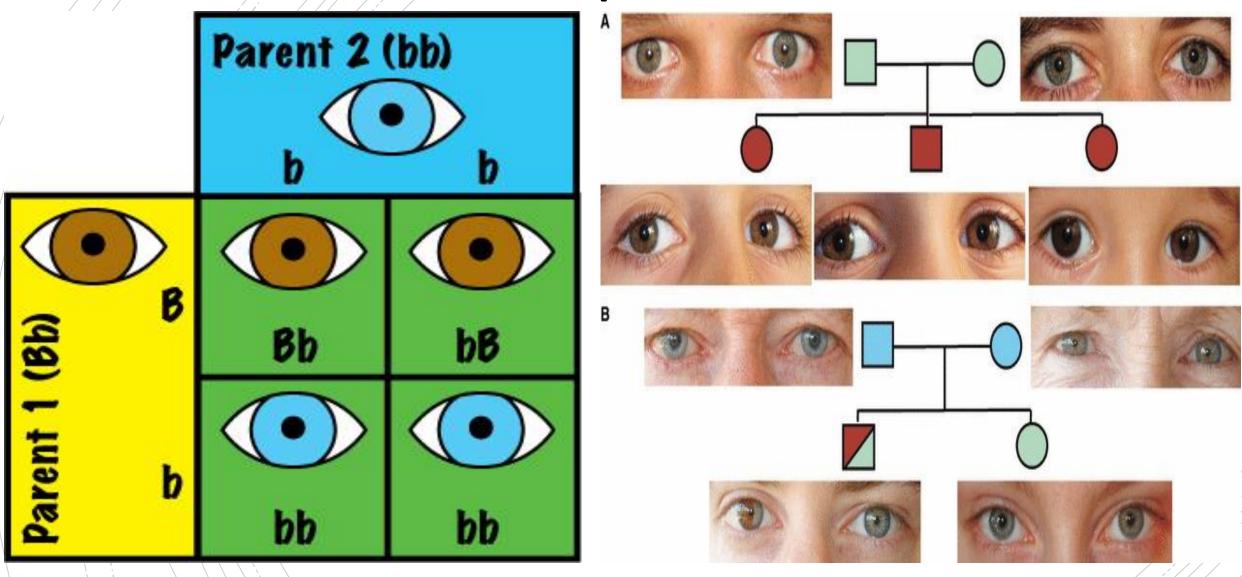


### Mendel's Experiments

### Mendel's Experiments



### **Mendel's Experiments**

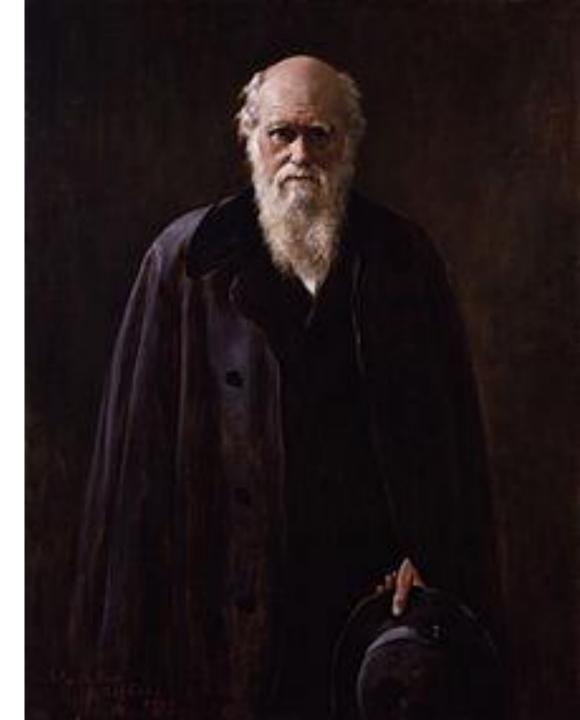


# English Naturalist Charles Darwin 1809-1882 "The Theory of Evolution"

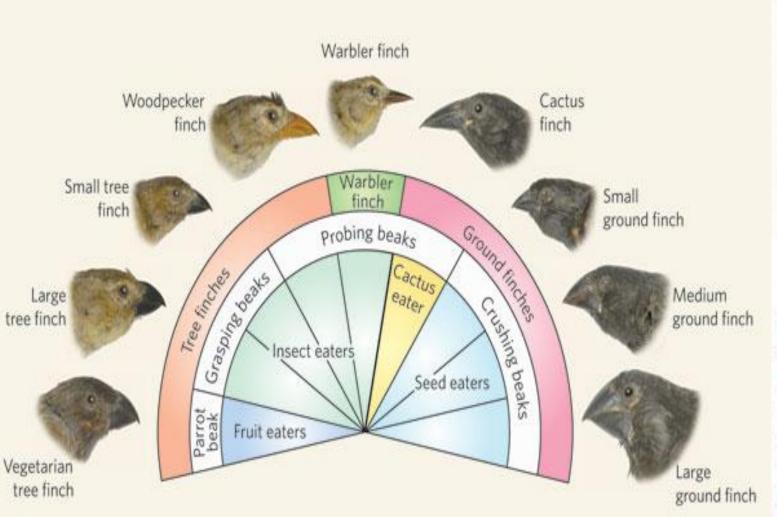
**Famous for work on Natural Selection** 

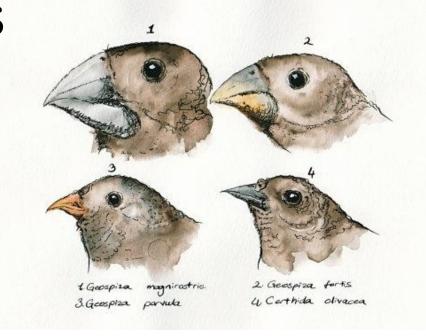


Above – with son, William 1842

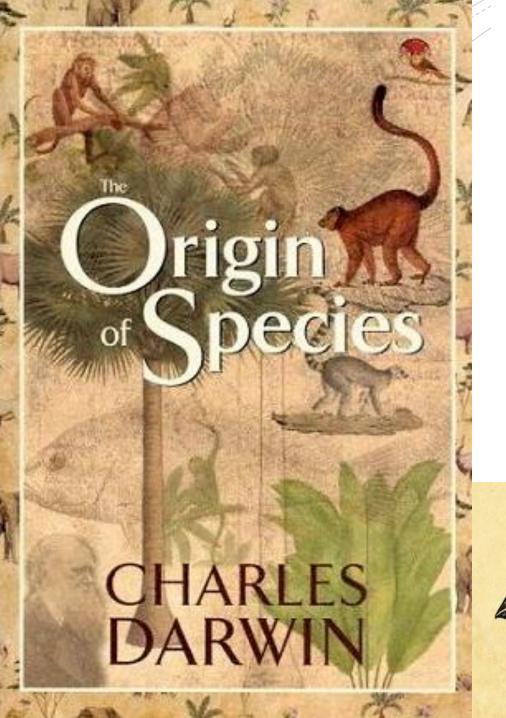


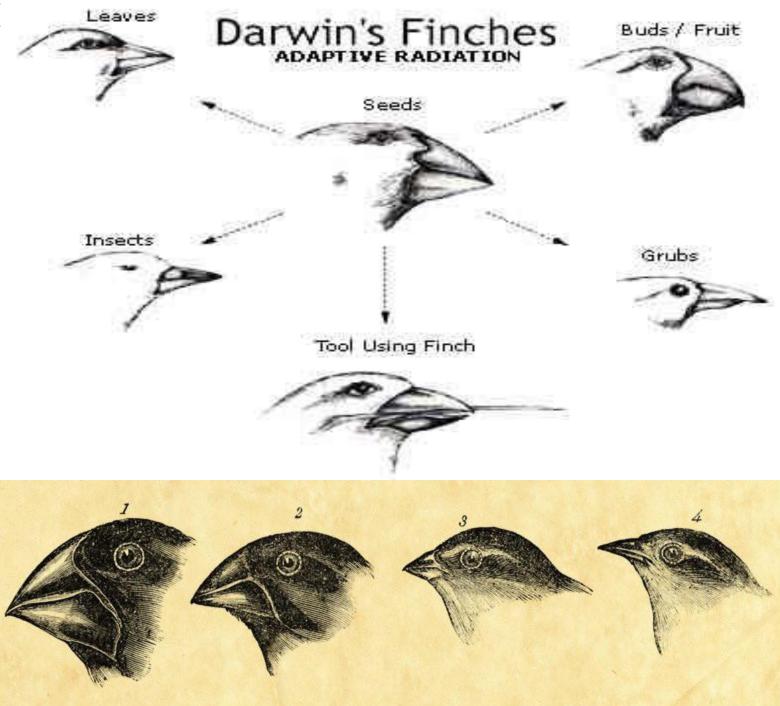
Darwin's Finches

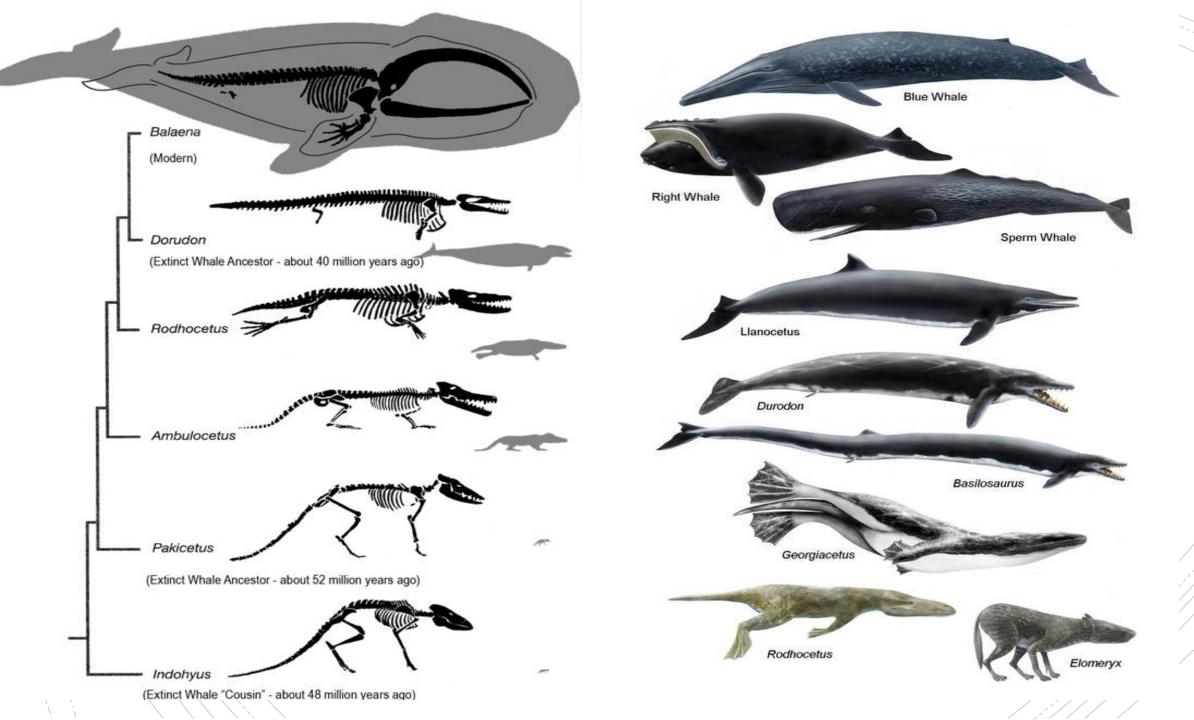




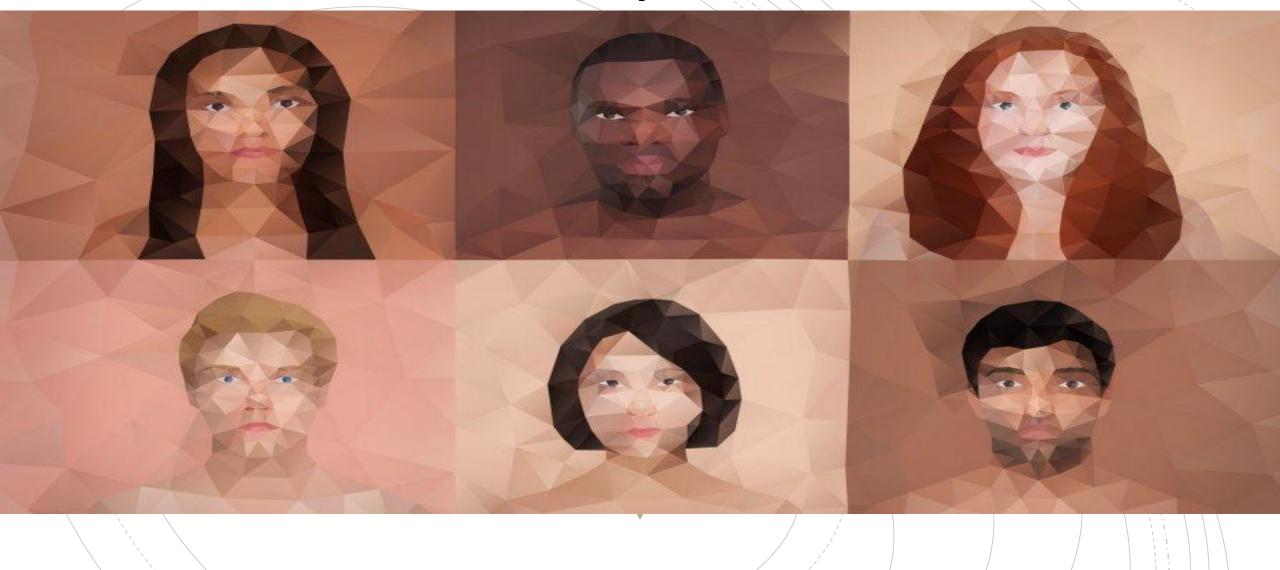




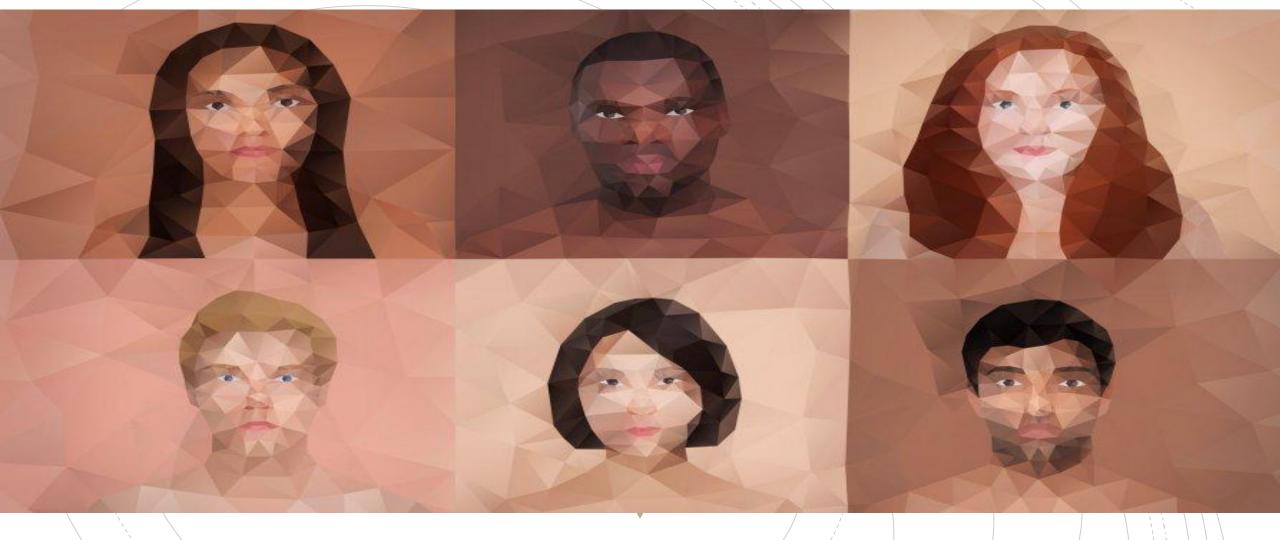




### Central Question: Why is there Human Variation?



### Central Question: Why is there Human Variation?



Answer: Caused by processes of heredity [mutation/gene flow/genetic drift/natural selection] and adaptation [Genetic; Developmental; Physiological; Behavioral; and cultural]

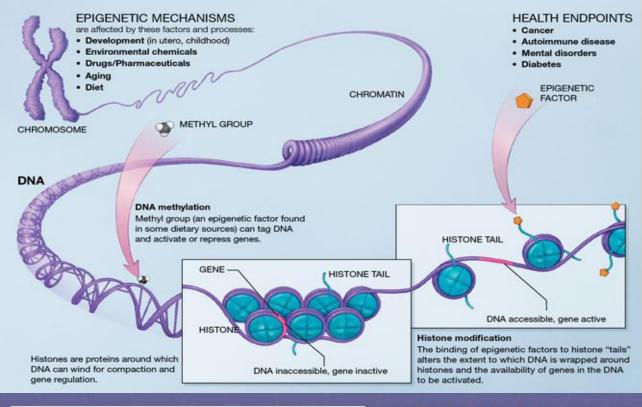
## Genetics and Epigenetics: Topics

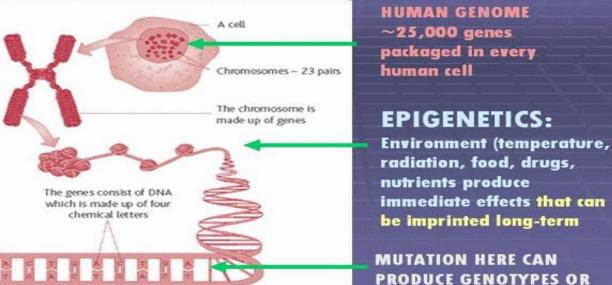
Genes and the Human Body

#### **Genetics**

Genetics is the study of heredity in living organisms through a focus on molecular structures such as genes, and the causes and results of genetic variation.







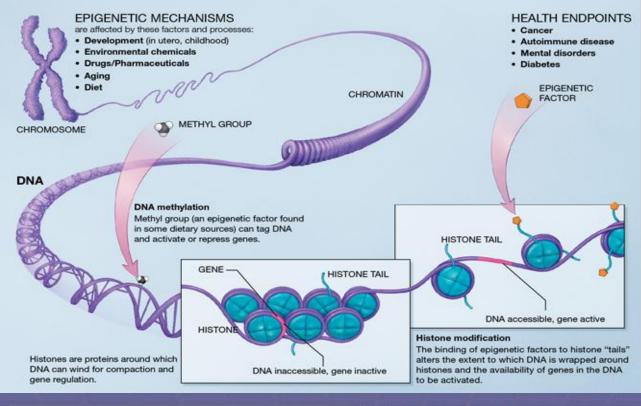
RARE GENETIC DISORDERS

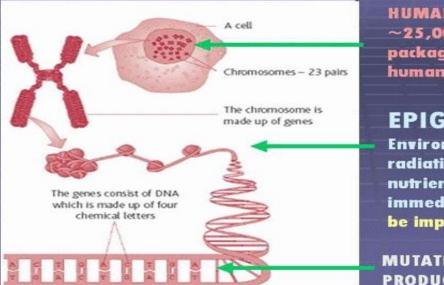
### **Epigenetics**

**Epigenetics** is the study of the external modifications to DNA that turn genes "on" or "off."

Modifications do not change DNA sequence, but determine which proteins are transcribed and thus which trait appears.







HUMAN GENOME
~25,000 genes
packaged in every
human cell

#### **EPIGENETICS:**

Environment (temperature, radiation, food, drugs, nutrients produce immediate effects that can be imprinted long-term

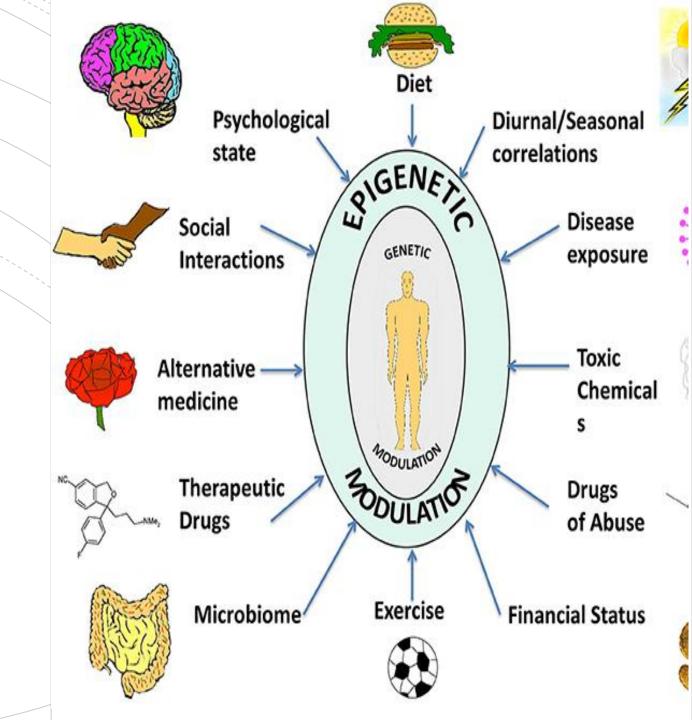
MUTATION HERE CAN PRODUCE GENOTYPES OR RARE GENETIC DISORDERS

### **Epigenetics**

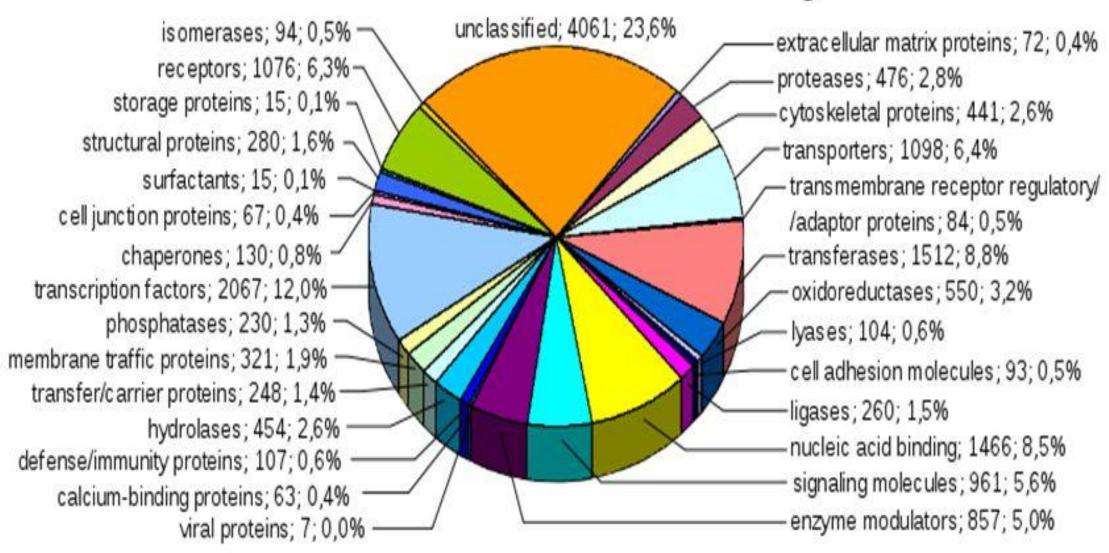
**Epigenetics** is the study of the external modifications to DNA that turn genes "on" or "off."

Epigenetics also studies how RNA, DNA, and genes are affected by environmental factors in ways that can be passed down across generations.





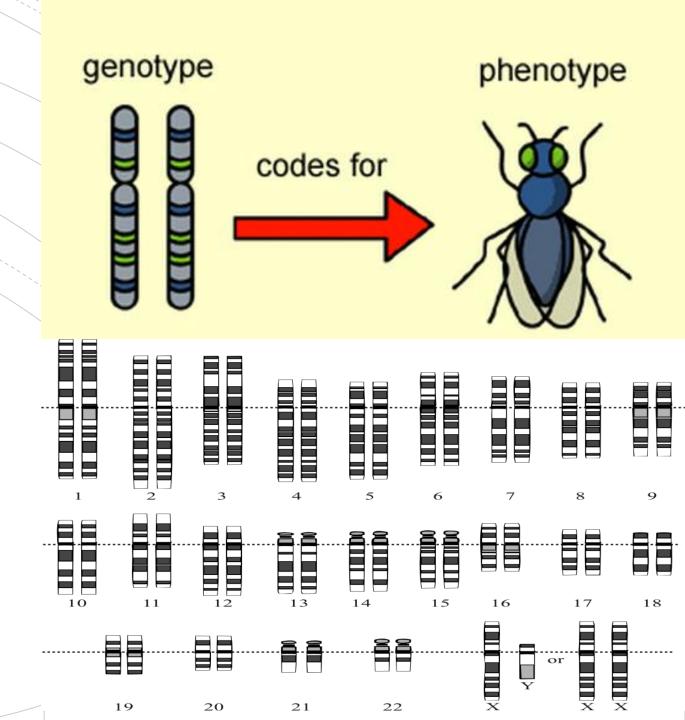
### Human Genome Project



### Genotype

Genotype the complete genetic identity / unique genome of an individual organism or group of organisms, with reference to a single trait, set of traits, or an entire complex of traits; the sum total of heritable genes transmitted from parent to offspring.

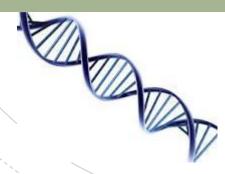




### Phenotype

**Phenotype** is set of observable physical or biochemical characteristics of an individual resulting from the interaction of the genetic makeup [genotype] with the environment.

Examples: Stature, eye color, skin color, hair color.













a. Short lingers: SS or Ss





t. Long lingers: ss



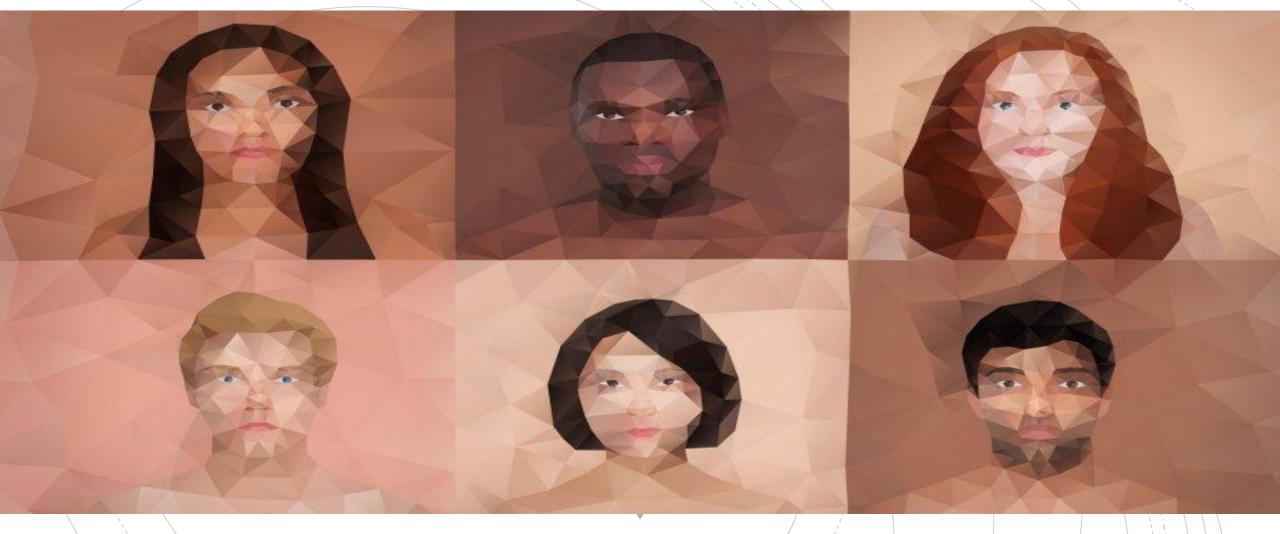


Genotype=bb Recessive=b

Genotype = Bb or BB Dominant =B



### **Human Variation**



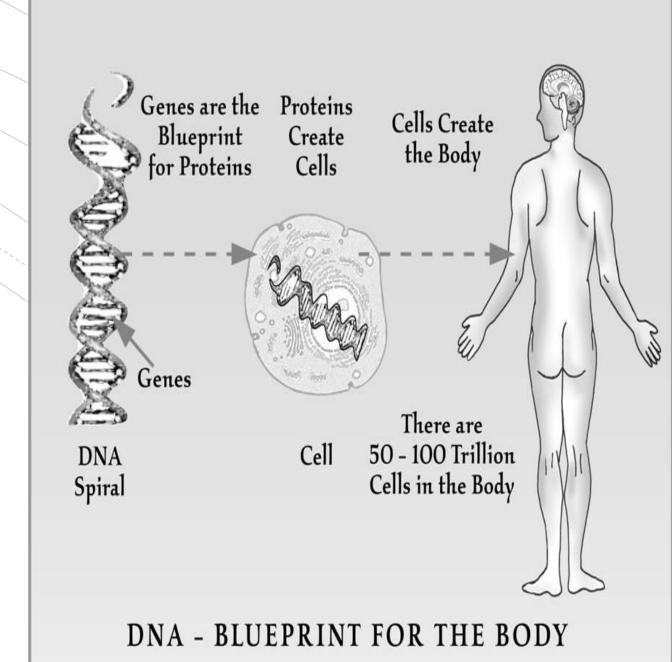
Differences in genotype and phenotype amongst a population or amongst populations. Caused by factors of heredity [recombination, mutation/gene flow/genetic drift/natural selection] as well as differential adaptation strategies in different organisms/species.

### **Human Body**

Contains 37.2 trillion cells

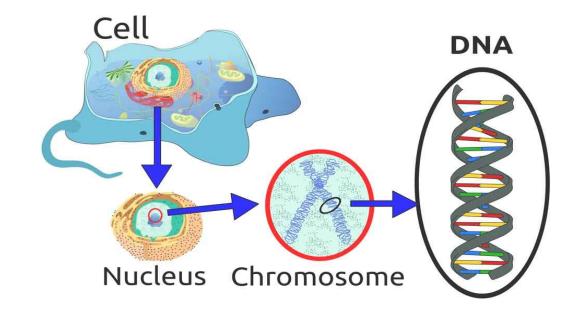
19,000-30,000 genes

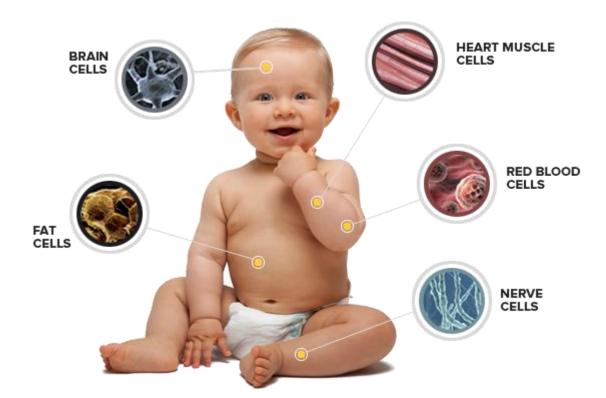
46 chromosomes = 23 chromatid pairs



# **DNA** Coding

Within the nucleus of our cells, there are **chromosomes** on which sit **DNA** molecules.



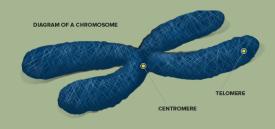


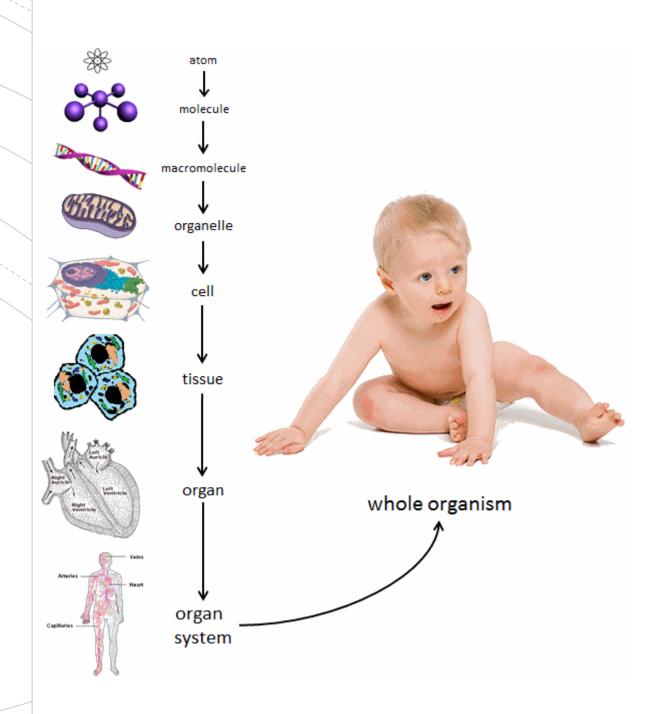
# **Human Body**

46 chromosomes = 23 chromatid pairs

22 pairs of Autosomes

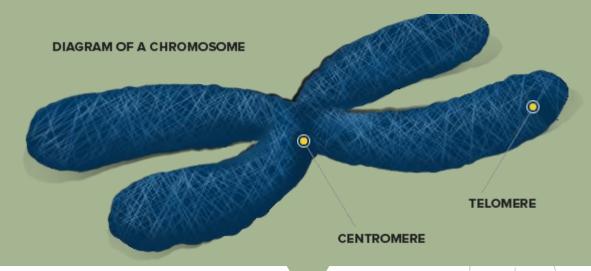
1 pair of sex chromosomes





### Chromosomes

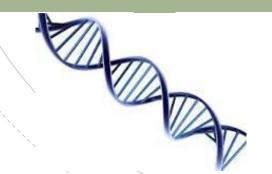
**Chromosomes** are basic genetic units, arranged in matching pairs.

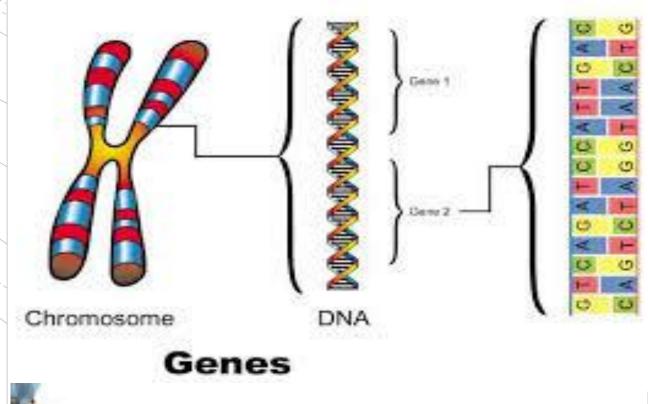


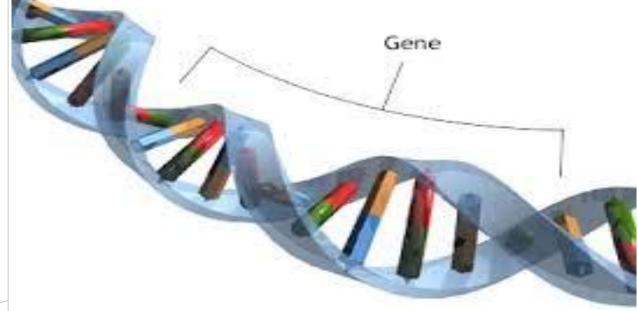
Humans: 46 chromosomes – 23 pairs

### Gene:

Gene: the location on a chromosome that helps determines a particular trait of the organism



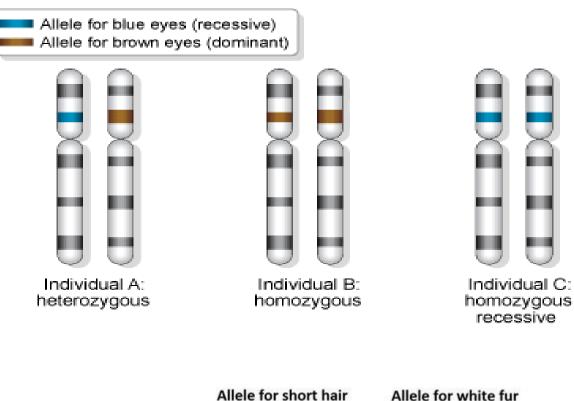


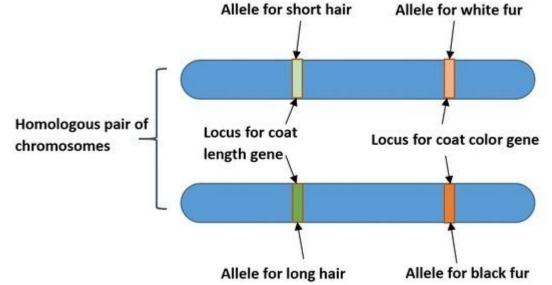


### Allele

Allele- a variant/alternative form of a gene that occupies a given locus (position) on a chromosome

Active in DNA coding.



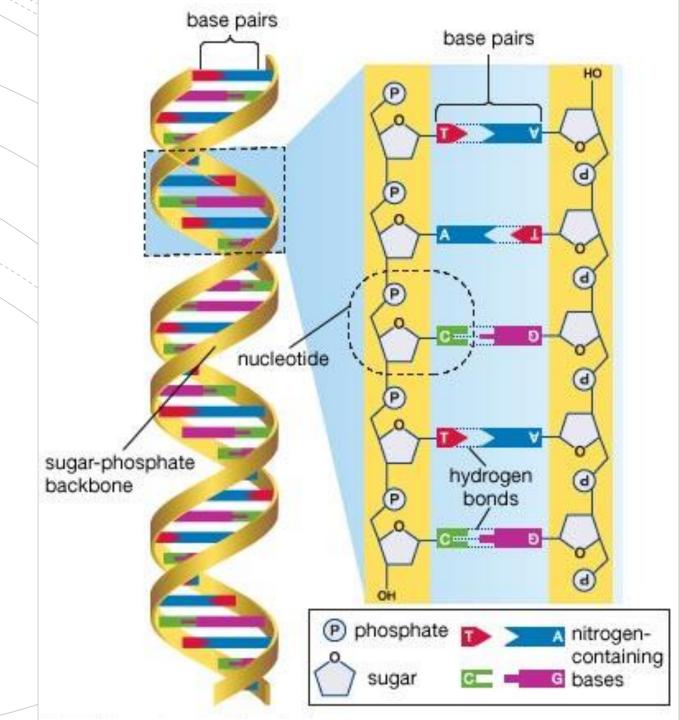


### **DNA**

DNA (deoxyribonucleic acid):

"a molecule that carries genetic instructions used in the growth, development, functioning and reproduction of all known living creatures"; basic hereditary units that lead to inheritance of traits from parent lineages in reproduction



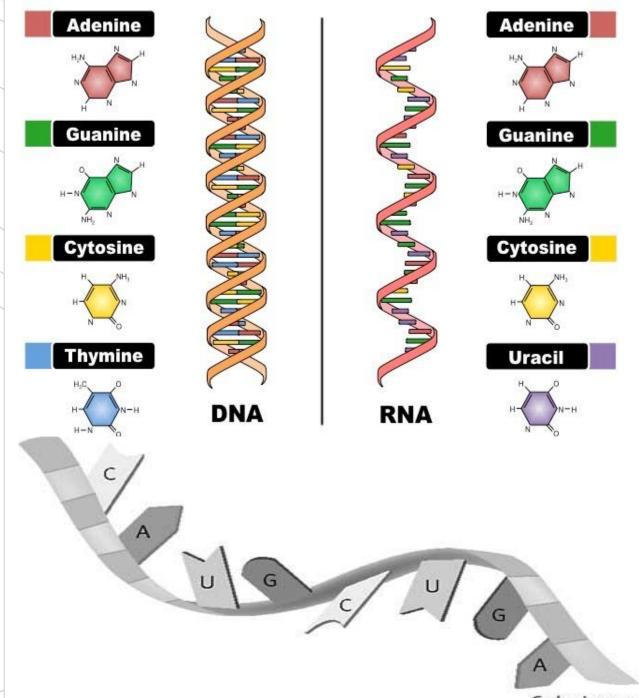


### **RNA**

Ribonucleic acid (RNA) is a polymeric molecule implicated in various biological roles in coding, decoding, regulation, and expression of genes.

Both RNA and DNA are nucleic acids containing complementary base pairs of nucleotides

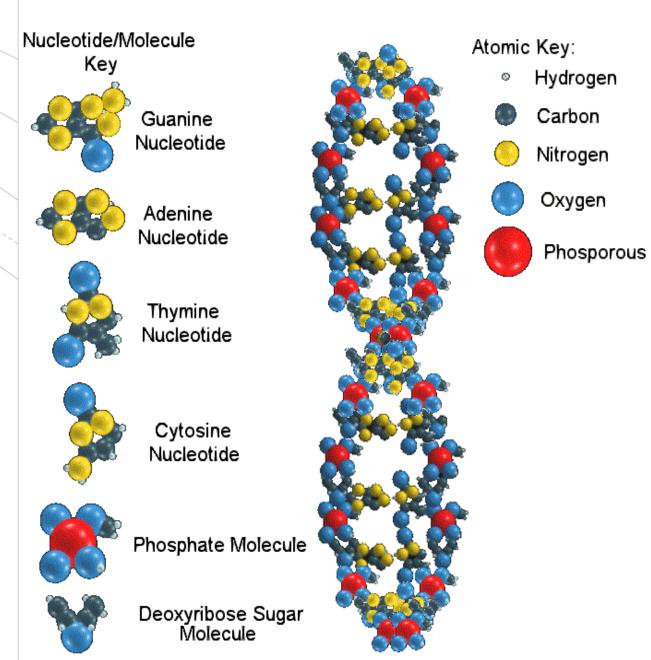




### DNA Polymer

### **Nucleotide**

building blocks of nucleic acids which form complementary pairs of chemical structures in DNA and RNA [Guanine; Adenine; Thymine; Cytosine]; Composed of 3 parts: five-sided sugar. \* phosphate group. \* nitrogenous base (nitrogen containing)



### DNA Polymer

### **Nucleotide**

The four main nucleobases found in the nucleic acids DNA and RNA:

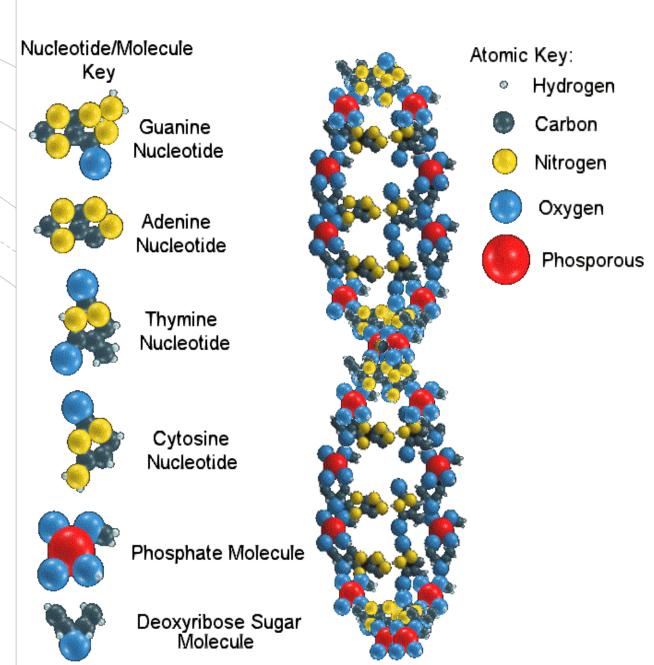
Guanine

Adenine

Thymine

Cytosine

+Uracil [RNA only]



### DNA Polymer

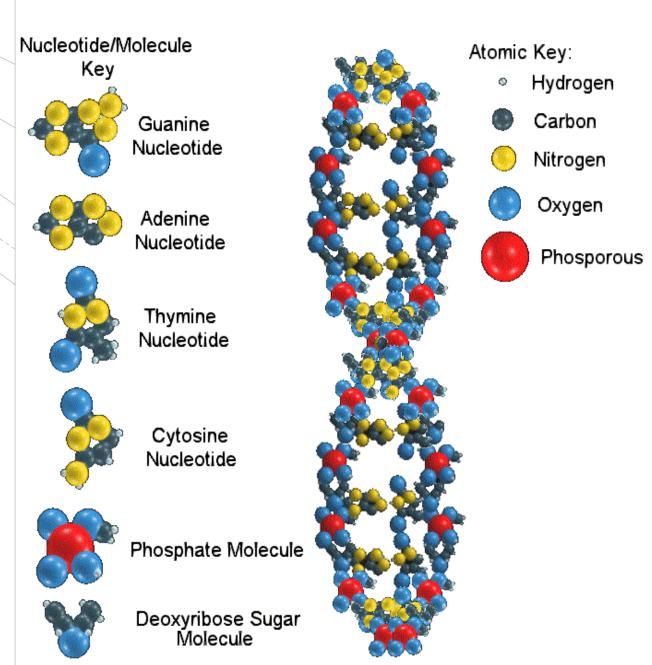
### **Nucleotide**

Guanine – paired with Cytosine [G/C]; purine derivatives

Adenine – binds to Thymine in DNA; Uracil in RNA; purine derivatives

Thymine – binds to Adenine in DNA; pyrimidine derivatives

Cytosine – Pairs with Guanine in DNA; pyrimidine derivatives



### **Dominant Allele**

Dominant trait/allele—
coordinate/trait on gene that
masks another allele/ is brought
to full expression and causes
another potential trait to be
recessive.



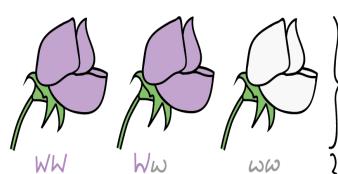


# Table C-1: Expression of Dominant and Recessive Alleles

Alleles Present	Allele Expressed
Dominant, Dominant	Dominant
Dominant, Recessive	Dominant
Recessive, Recessive	Recessive

W-dominant purple allele

w - recessive white allele



Phenotype flower color

Genotypepair of alleles

### **Recessive Allele**

Recessive trait/allelecoordinate/trait on gene masked by another allele/ is not brought to full expression but remains in the genetic code



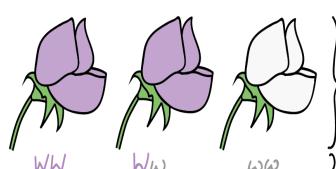


# Table C-1: Expression of Dominant and Recessive Alleles

Alleles Present	Allele Expressed
Dominant, Dominant	Dominant
Dominant, Recessive	Dominant
Recessive, Recessive	Recessive

W-dominant purple allele

w - recessive white allele

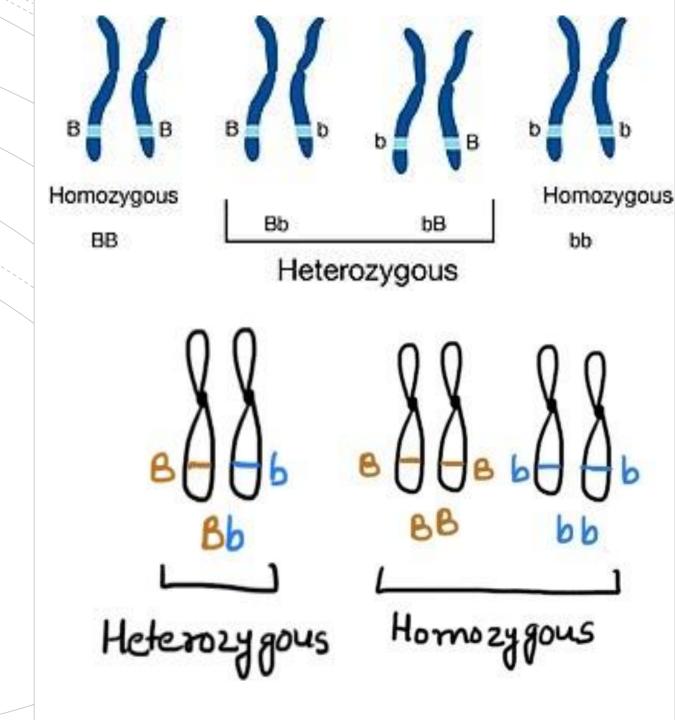


Phenotype flower color

Genotype pair of alleles

# Heterozygous

When a gene locus of a diploid organism contains two different alleles of a gene, it is heterozygous.



# Homozygous

When a gene locus of a diploid organism contains two identical alleles of a gene, it is homozygous.

F = freckles
 f = no freckles
 W = widow's peak
 w = straight hairline







Ff Ww Eggs:



FVI

FW	1 16 FFWV
Fw	1 16 FFW
fW	1 16 <i>Ff W</i> I

FW	Fw	fW	fw
FFWW	1 16 FFWw	1/16 Ff WW	1/16 Ff Ww
1/16 FFWw	1 16 FFww	1 16 Ff Ww	1/16 Ffww
1/16 Ff WW	1 16 <i>Ff Ww</i>	1/16 ffWW	1/16 ff Ww
1 16 <i>Ff Ww</i>	1 16 Ffww	1 16 ff Ww	1 16 ffww

9 16 freckled, widow's peak

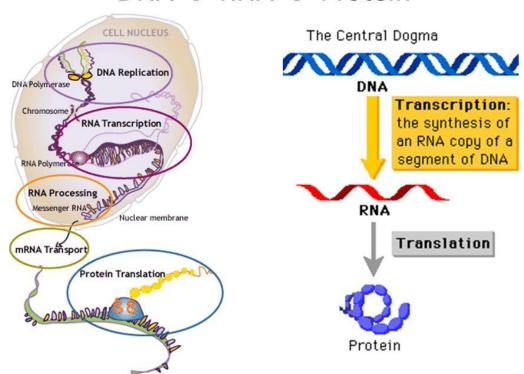
3 freckled, straight hairline

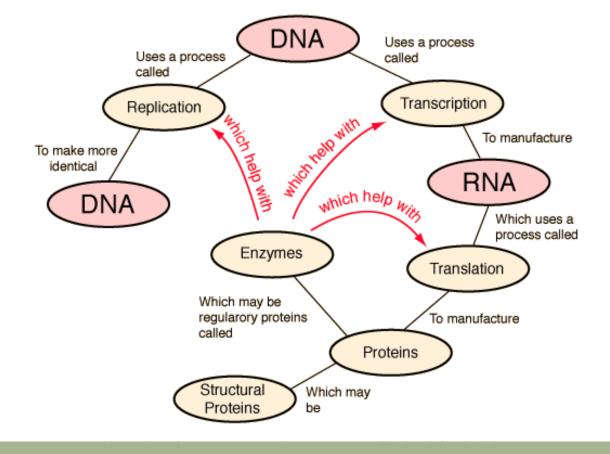
fw

3 no freckles, widow's peak

1/16 no freckles, straight hairline

### DNA $\rightarrow$ RNA $\rightarrow$ Protein



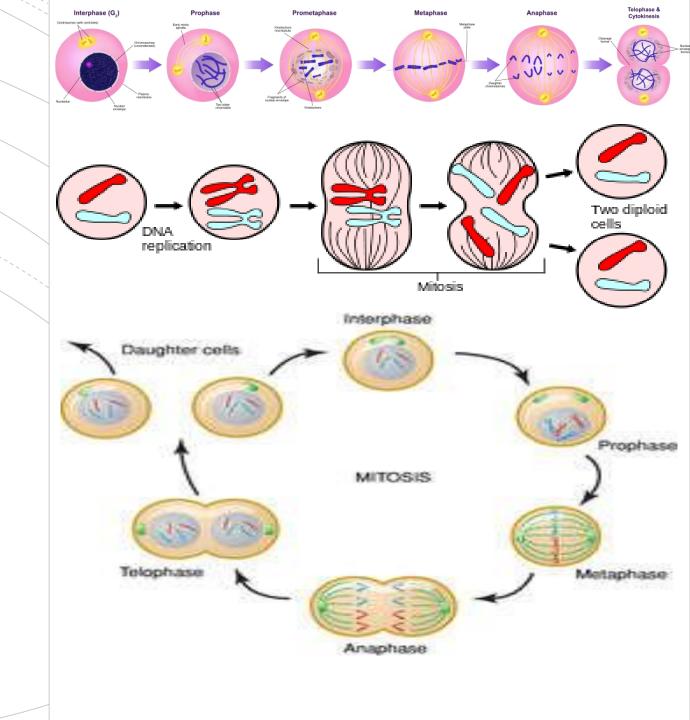


# Protein Coding – DNA Processes

# Methods Human Reproduction at the Molecular Level

### **Mitosis**

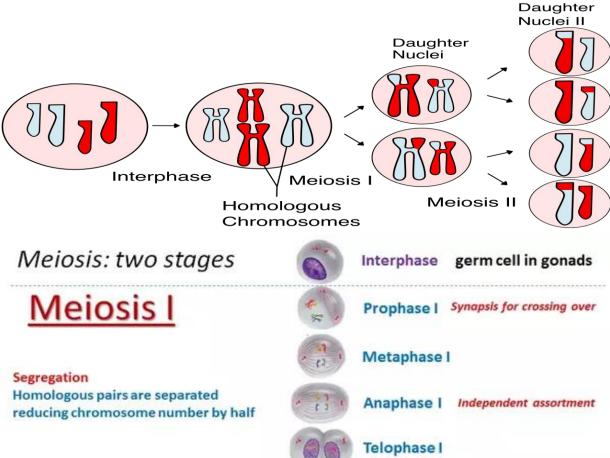
Mitosis: a form of cell division which results in the creation of two daughter cells with the same kind and number of chromosomes as the parent nucleus.



### **Meiosis**

a form of cell division where a single cell divides twice to result in four daughter cells, each with half the number of chromosomes of the parent cell / half of the original genetic information.

[gametes/sex cells; plant spores]





### Meiosis II



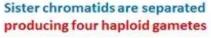


Prophase II





Metaphase II







Anaphase II



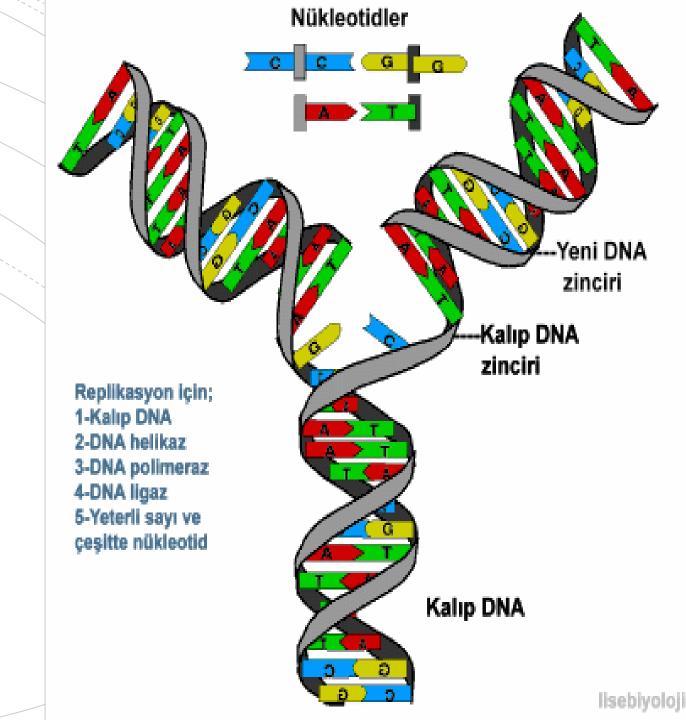






# Replication and Recombination

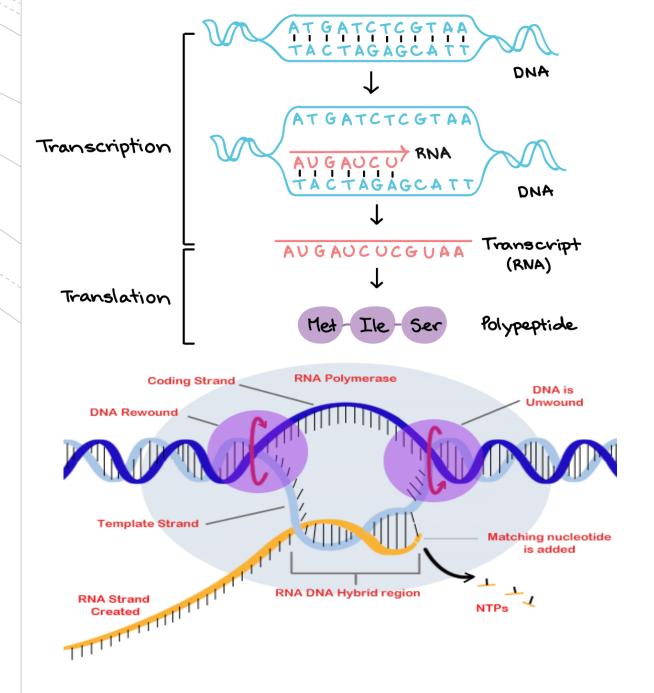
**DNA replication -** biological process of one DNA molecule producing two identical replicas of the original.



# **Transcription**

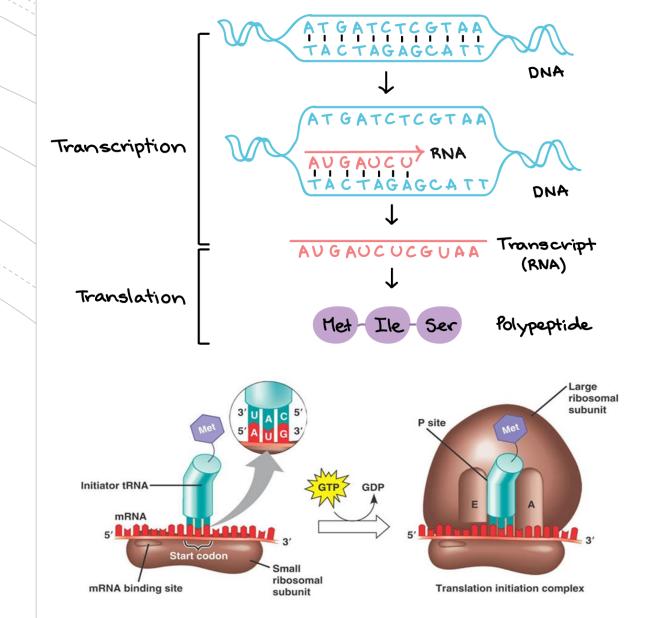
**Transcription** – a process by which a segment of DNA is copied by/onto RNA/ mRNA.

The first phase of gene expression



### **Translation**

messenger RNA (mRNA) is the next stage of gene expression. mRNA is decoded outside the nucleus by a ribosome, which creates a polypeptide (amino acid chain). This polypeptide then folds into an active protein, performing specific functions inside the person's cells.



# Methods, Part II

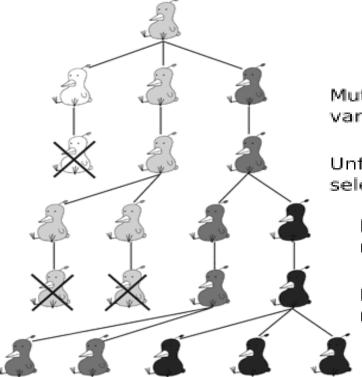
Human Evolution at the Molecular Level

### **Natural Selection:**

Natural Selection is "the process by which those characteristics that allow individuals to have more offspring become more widely spread in the population (selected for) and those that allow individuals to have fewer offspring traits become rarer (Selected against)" (Mielke 2006)

Conditions necessary: Multiplication;

Heritable Variation; Competition



Mutation creates variation

Unfavorable mutations selected against

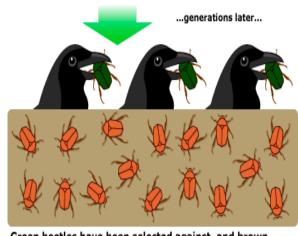
> Reproduction and mutation occur

Favorable mutations more likely to survive

... and reproduce

### Natural selection, in a nutshell:

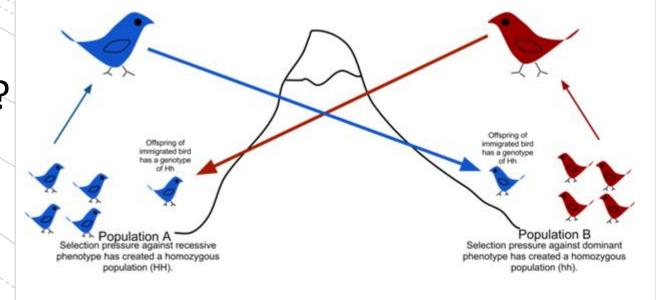


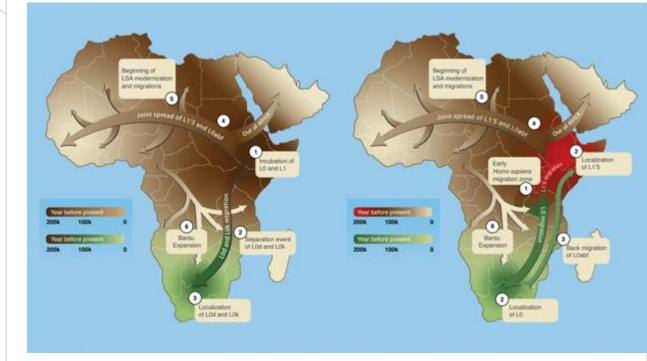


Green beetles have been selected against, and brown beetles have flourished.

# **Gene Flow [Migration]:**

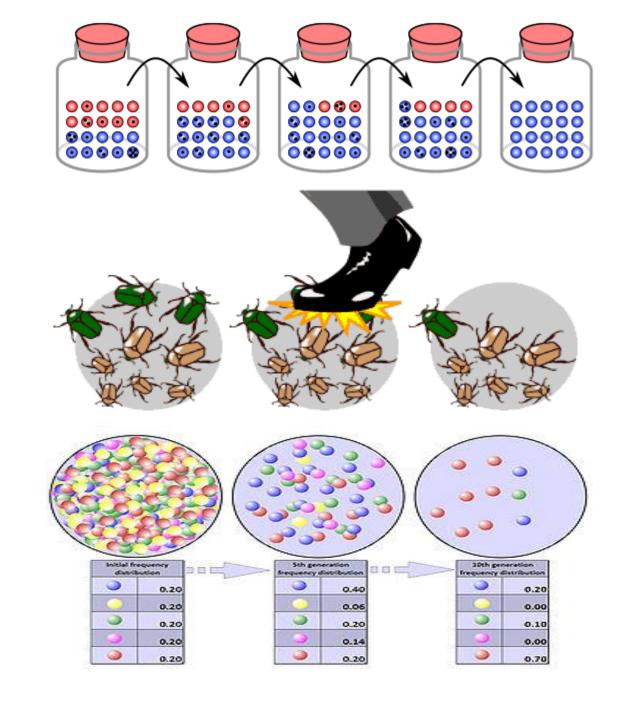
Gene Flow [Migration] is an "exchange of genes that occurs when a given population experiences a sudden expansion caused by in-migration of outsiders from another population of the species" (Mielke 2006)





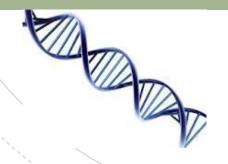
### **Genetic Drift:**

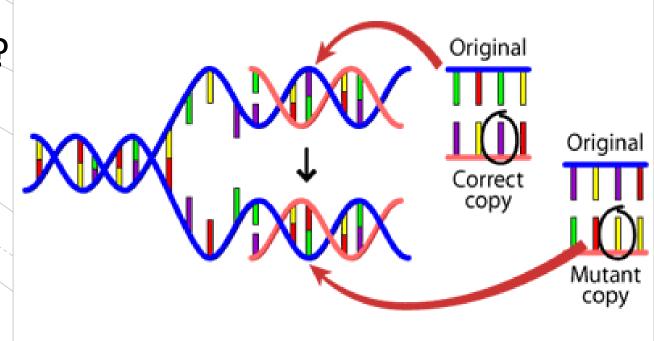
Genetic Drift: "Random changes in gene frequencies from one generation to the next caused by a sudden reduction in population size as a result of disaster disease or the outmigration of a small subgroup from a larger population" (Mielke 2006)



### **Mutation:**

Mutations are permanent alteration of the nucleotide sequence of the genome of an organism, virus, or extrachromosomal DNA or other genetic elements.







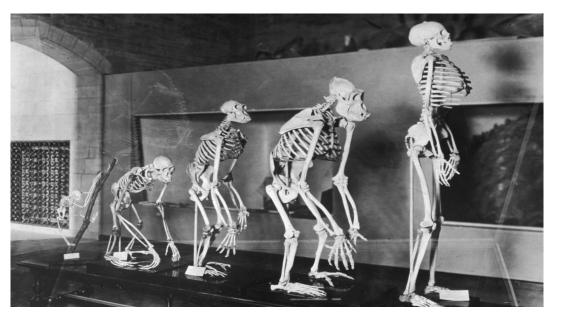


### **Mutation:**

In mutations, time and order of gene-activation is altered to reconfigure old genes in new ways.

•







# **Types of Mutation**

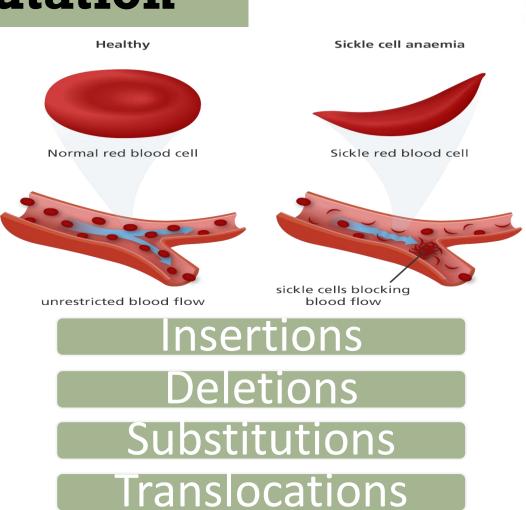


Point Mutation

 change of a single nitrogen base in a DNA sequence; maximum consequence changes in single amino acid in a protein; minimal harm



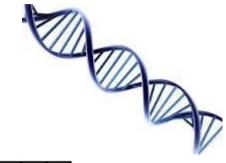
 Change of a single nitrogen base which is either deleted or has an extra one inserted into the middle of DNA sequence; causes the reading frame to shift



Recombinations

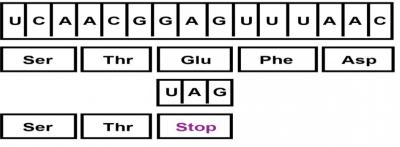
Non-Disjunction

# **Types of Mutation**



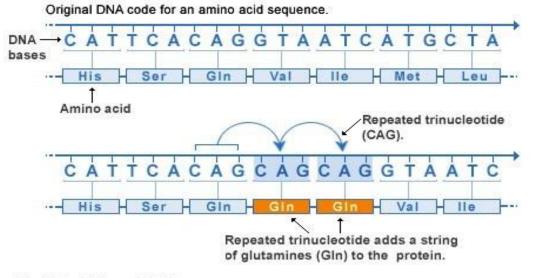
Nonsense mutation

 change in one DNA base pair in which the altered DNA sequence prematurely signals the cell to stop building a protein, which results in a shortened protein that may not function, or may function improperly



Repeat expansion

 Nucleotide repeats are short DNA sequences that are repeated a number of times in a row. Repeat expansion mutation



Duplication

 A duplication consists of a piece of DNA that is abnormally copied one or more times. This type of mutation may alter the function of the resulting protein.

U.S. National Library of Medicine

# **Types of Mutation**



### Insertions

• addition of one or more nucleotide base pairs into a DNA sequence

### Deletions

• changes the number of DNA bases by removing a piece of DNA.

### Substitutions

• An exchange of one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G)

### Translocations

• mutations in which chromosome segments, and the genes they contain, change positions.

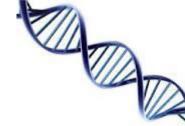
### Recombinations

• exchange of **genetic** material either between multiple chromosomes or between different regions of the same chromosome; can produce offspring with different characteristics than either parent.

# Non-Disjunction

• failure of homologous chromosomes or sister chromatids to separate properly during cell division.

### **Insertion**



Insertion

**Deletion** 

Substitutions

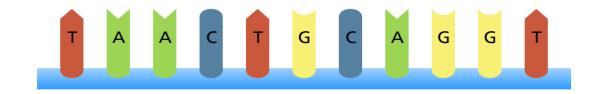
**Translocations** 

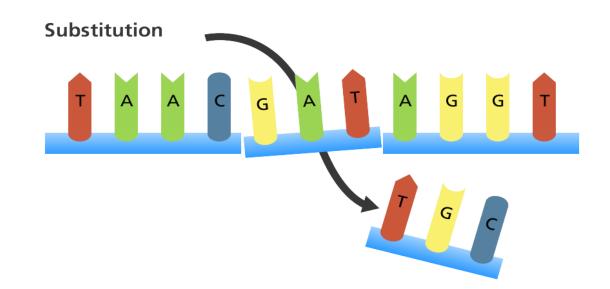
Recombinations

Non-Disjunction

changes the number of DNA bases by removing a piece of DNA.

### **Original sequence**





# **Deletion**

**Deletion with** 

Frameshift



Insertion

changes the number of DNA bases by removing a piece of DNA.

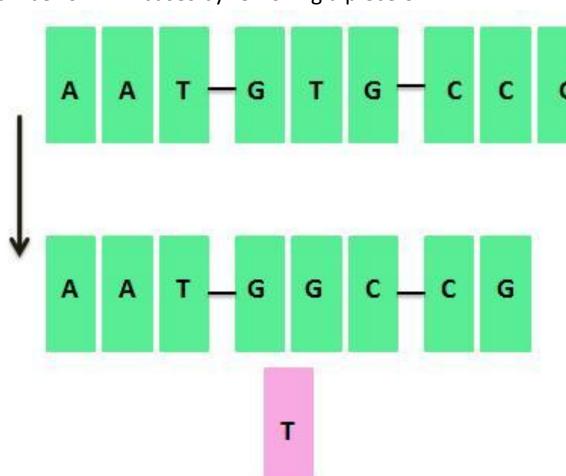
**Deletion** 

Substitutions

**Translocations** 

Recombinations

Non-Disjunction







Insertion

An exchange of one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G)

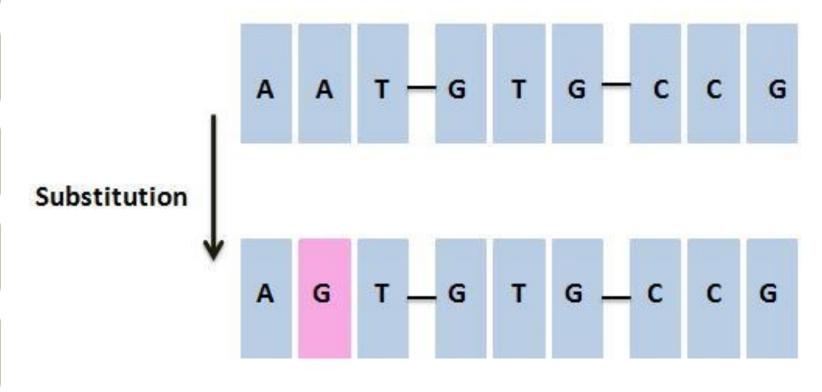
**Deletion** 

**Substitutions** 

**Translocations** 

Recombinations

**Non-Disjunction** 



### **Translocation**



Insertion

mutations in which chromosome segments, and the genes they contain, change positions.

Chromosomes

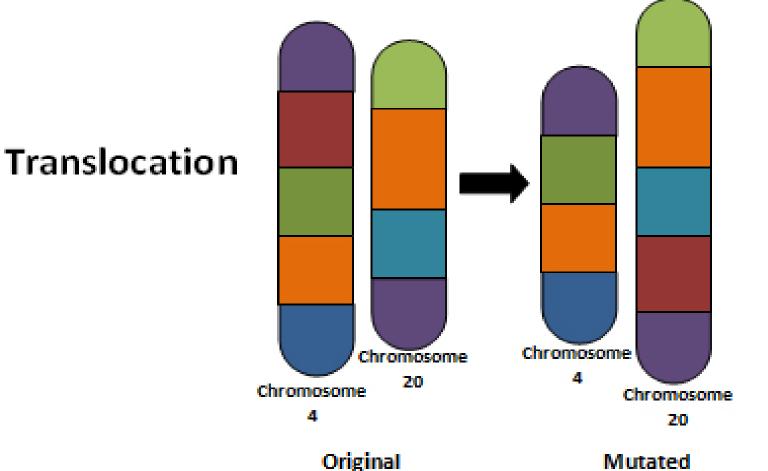
**Deletion** 

**Substitutions** 

**Translocations** 

Recombinations

Non-Disjunction



Chromosomes



### Recombination

Insertion

exchange of **genetic** material either between multiple chromosomes or between different regions of the same chromosome; can produce offspring with different characteristics than either parent.

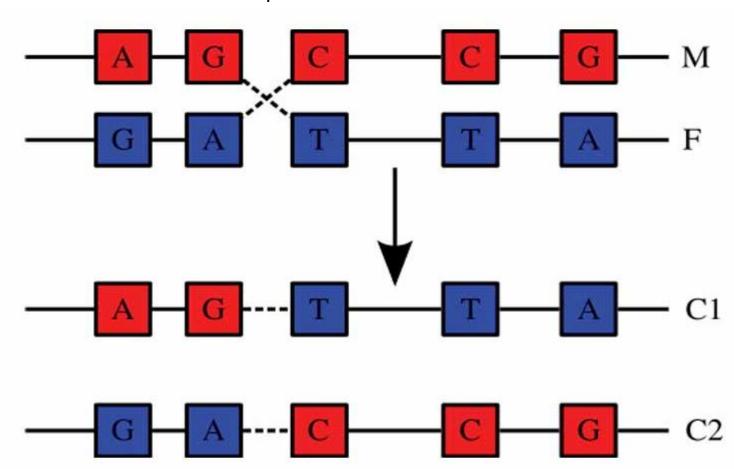
**Deletion** 

**Substitutions** 

**Translocations** 

Recombinations

Non-Disjunction



# Non-Disjunction



to gametes incorrectly

Insertion

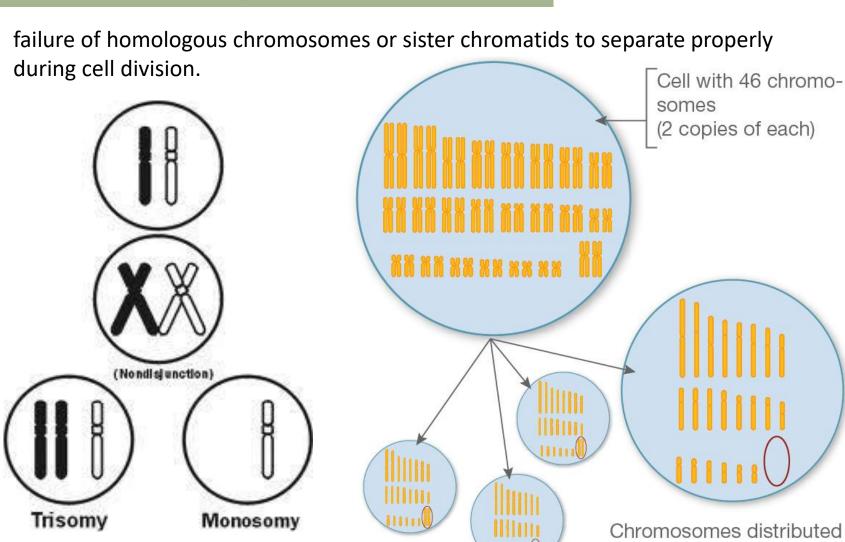
**Deletion** 

**Substitutions** 

**Translocations** 

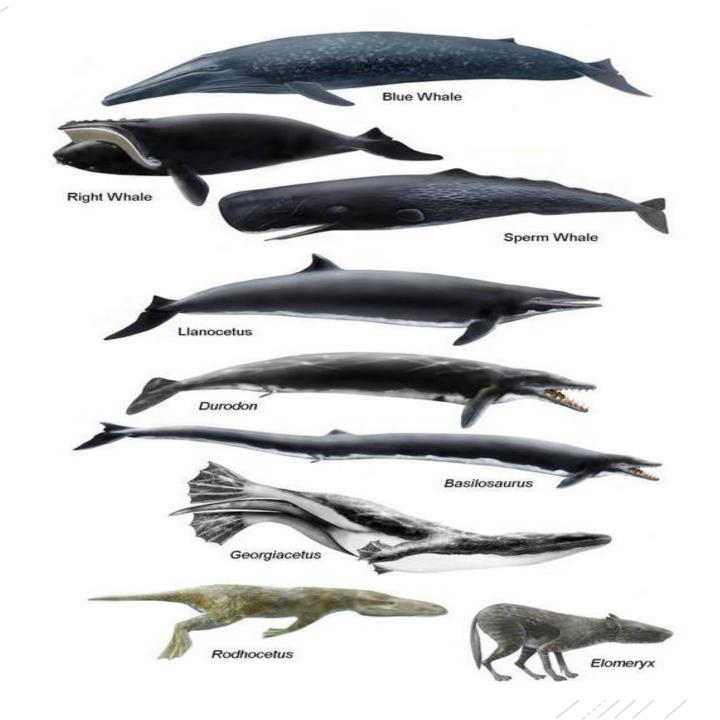
Recombinations

**Non-Disjunction** 



### **Additional Video Resources**

https://www.brightstor m.com/science/biology/ mendelian-genetics/



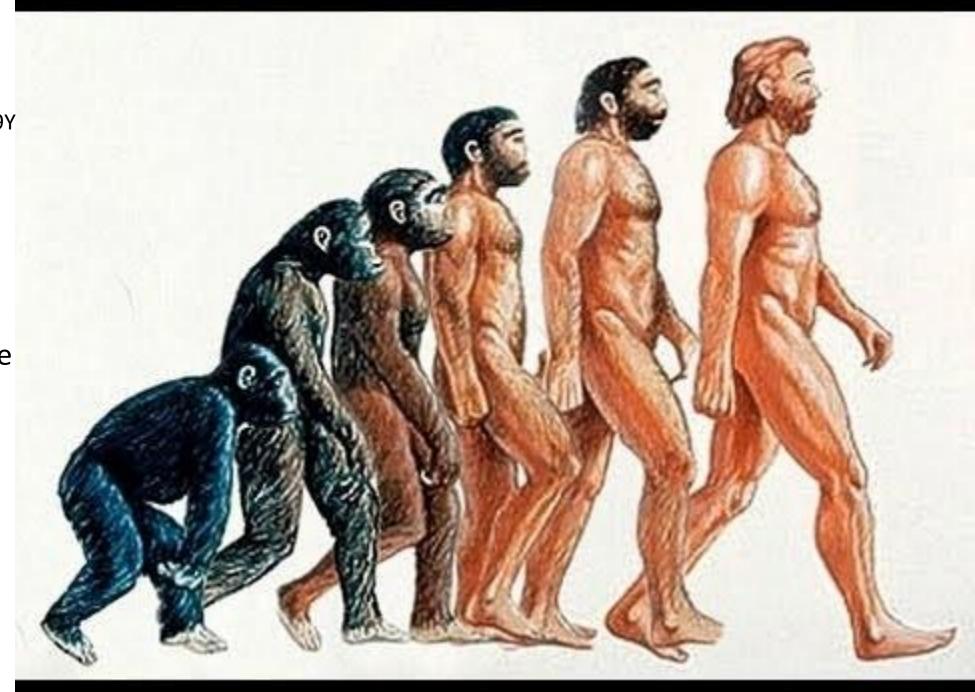
### Evolution [Continued]

https://youtu.be/MsHEAnPX59Y

10:25-12:00 – Cambrian explosion

17:35 –21:05 move from life in ocean to life on land

21:05-24:55 Mammals



### **Evolution Research**

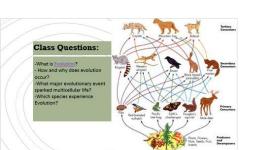
https://youtu.be/NOHzY1fuOz4

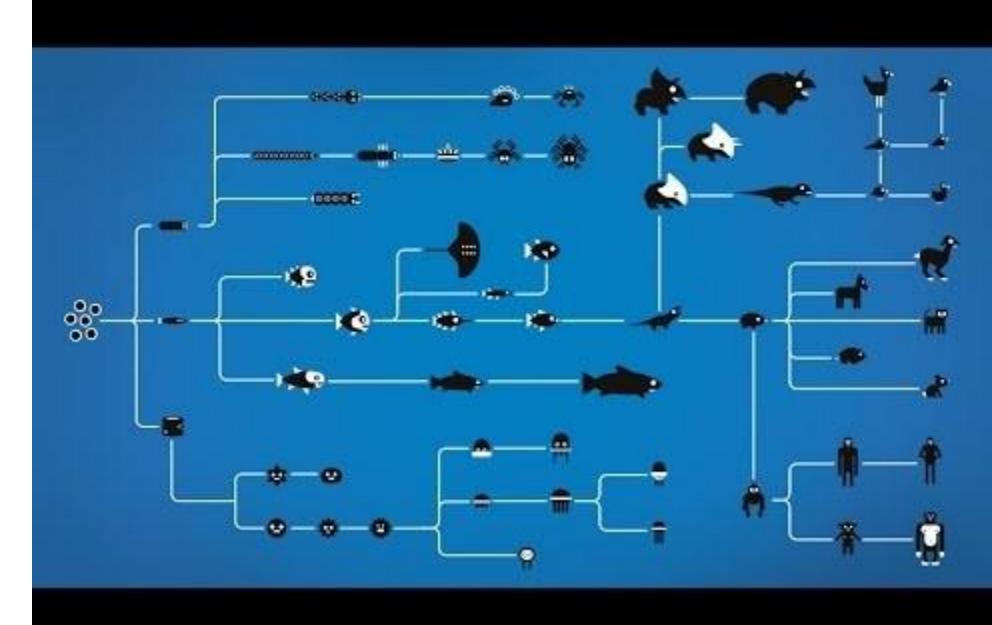
32 mins-mouse- DNA; 39 min- mutation – advantages of animals; 44 embryo studiesmulti species; 233 humans

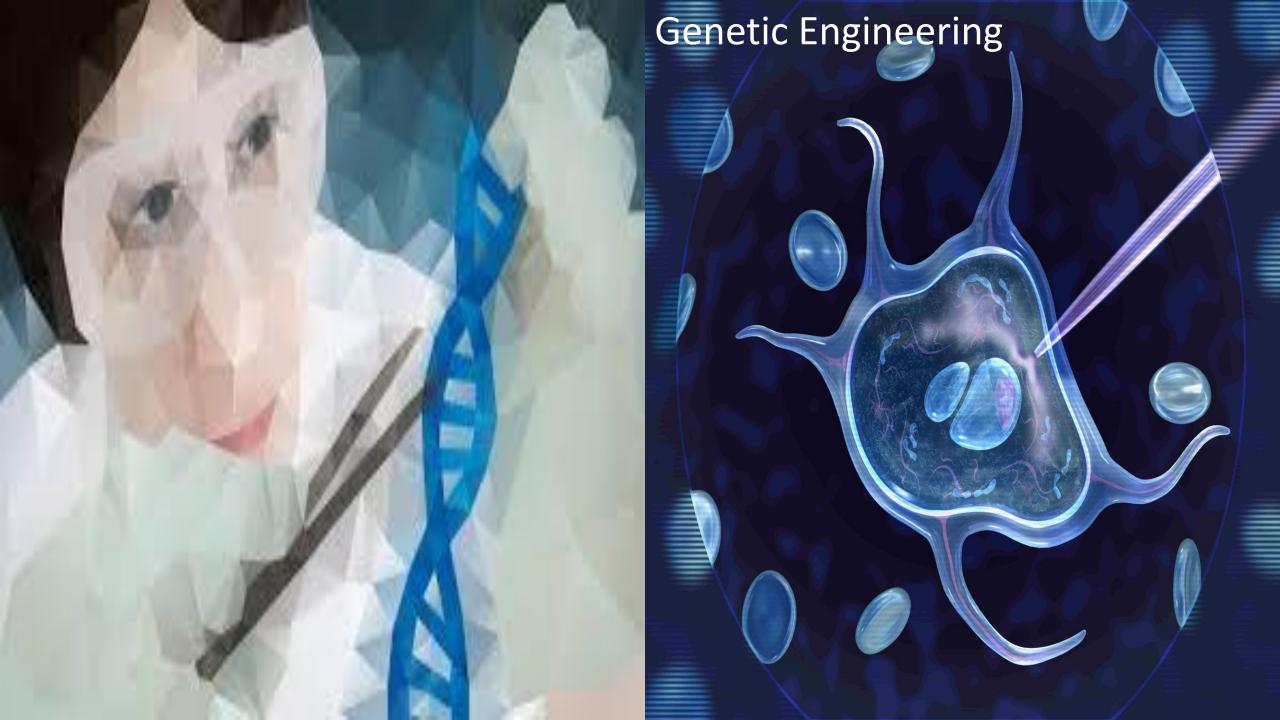


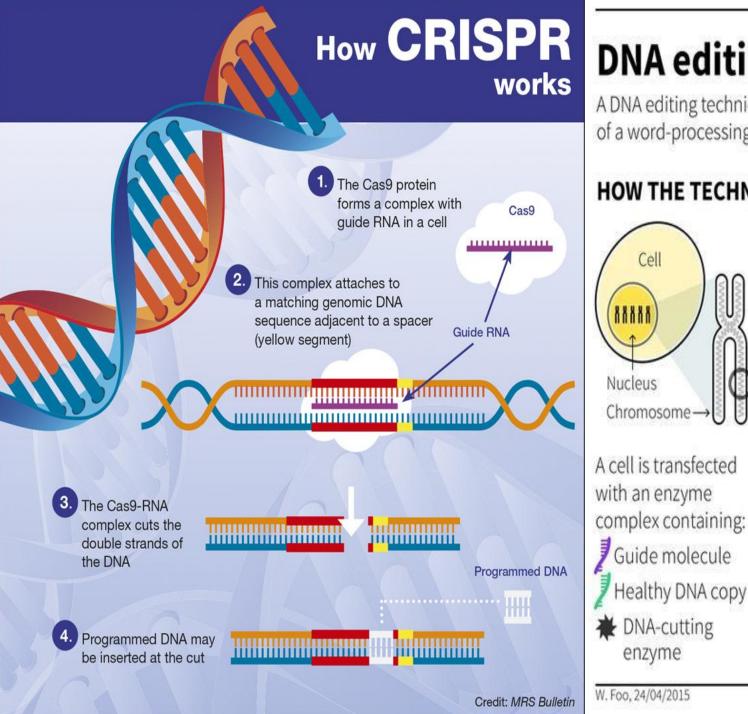
# Evolution Introduction

https://youtu.be/hOfRN0KihOU





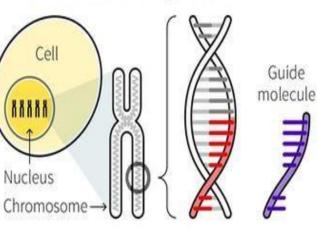




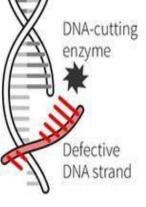
# **DNA** editing

A DNA editing technique, called CRISPR/Cas9, works like a biological version of a word-processing programme's "find and replace" function.

### **HOW THE TECHNIQUE WORKS**



A specially designed synthetic guide molecule finds the target DNA strand.



An enzyme cuts off The defective DNA the target DNA strand is replaced with a healthy copy. strand.

Sources: Reuters; Nature;

Massachusetts Institute of Technology

W. Foo, 24/04/2015

enzyme

Guide molecule

DNA-cutting

Healthy DNA copy



Healthy DNA strand



# Low risk of No baldness-Alzheiners, breast caucer and strokes Perfect Sprinter 20/20 Vision

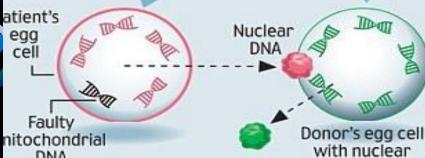
### **MAKING A THREE-PARENT EMBRYO**

Healthy nuclear DNA removed from patient's egg cell, leaving behind faulty mitochondrial DNA.

egg with healthy DNA.



'Reconstructed' egg cell fertilised with sperm in the lab and implanted into patient. Resulting embryo has three genetic parents



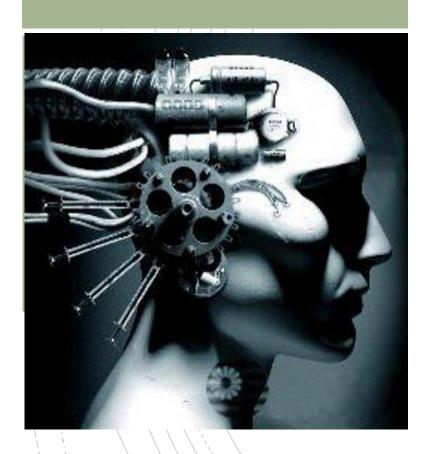






# **Cyborgs**

The integration of genetics and technology; a being with both organic and biomechatronic body parts or using technological adaptations.





http://www.explainthatstuff.com/timeline.html

### **Evolution - Major Historical Events**

- Only watch 10:25-12:00 [Cambrian Explosion, >2 mins] 17:35-21:05 [Move from ocean to life on land >4 mins]; 21:05-24:55 Mammals [>4 mins]
- https://www.youtube.com/watch?v=MsHEAnPX59Y&feature=youtu.be

1:25:00-1:52:00 [27 minutes]

https://www.youtube.com/watch?v=NOHzY1fuOz4&featu
re=youtu.be