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Chapter 14

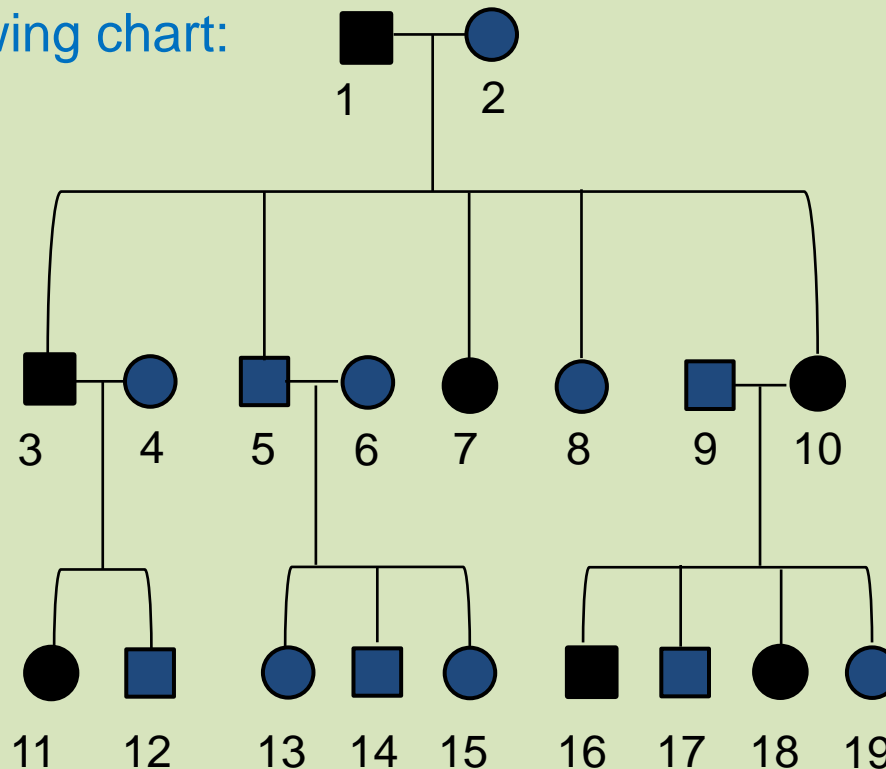
Genetic Variation & Selective Breeding





Review

1. Name three major human genetic patterns of inheritance and give examples of each.
2. What is heterozygous advantage related to genetic disorders?
3. Define affected and carrier related to genetic disorders?
4. What genetic tool maps chromosomes for study of disorders?
5. Decipher the following chart:





Review

1. Name three major human genetic patterns of inheritance and give examples of each.
 - **Autosomal Recessive Inheritance** (Sickle Cell, Phenylketonuria, Cystic Fibrosis)
 - **Autosomal Dominant Inheritance** (Huntington's, Achondroplasia, Aneuploidy)
 - **Sex-linked inheritance** (e.g. hemophilia, pattern baldness)
2. What is heterozygous advantage related to genetic disorders?
 - The condition of the heterozygous form being **protective** against some disease or illness
 - This is the protection that the heterozygous condition can give to people who are **carriers of a recessive allele**.
3. Define affected and carrier related to genetic disorders?
 - **Affected** persons have a genetic disease or condition.
 - **Carriers** do not have a genetic disease, but carry the gene which causes the disease.
 - Carriers have **one normal/dominant** allele + **one recessive** allele for a disease.
4. What genetic tool maps chromosomes for study of disorders?
 - karyotype

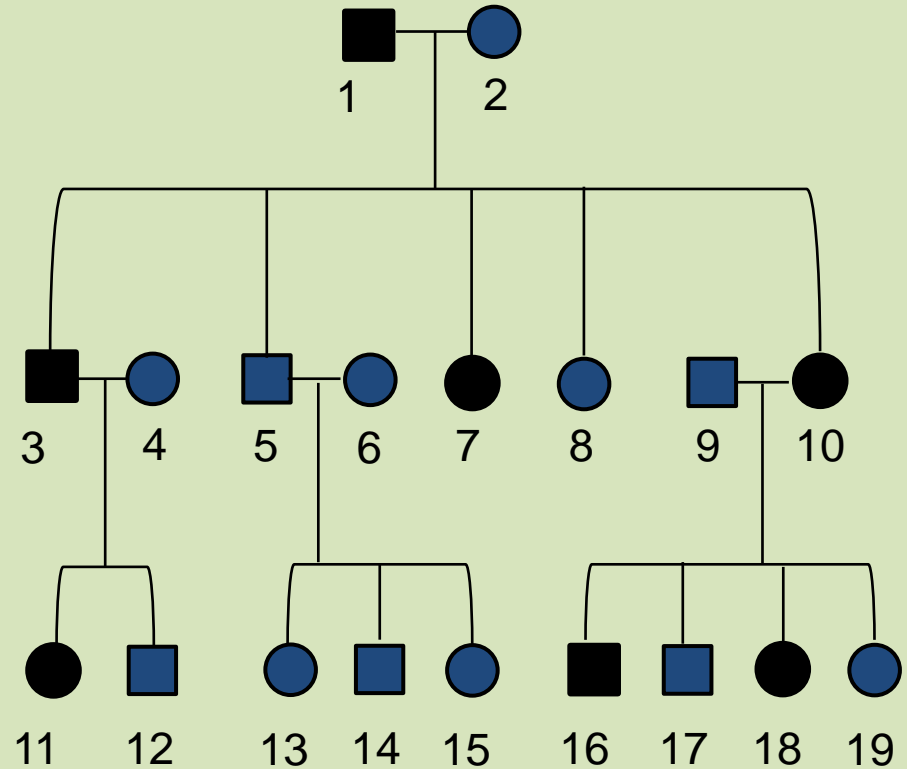


Review

Decipher the following chart:

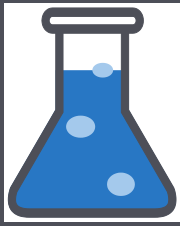
- Generation: row
- **Males**: squares
- **Females**: circles
- Horizontal lines between circle and square: **matings**
- Vertical lines: **offspring** of a mating

Different colors or shading are used to show individuals with and without a trait.





Lesson Objectives



By the end of this lesson, you should be able to:

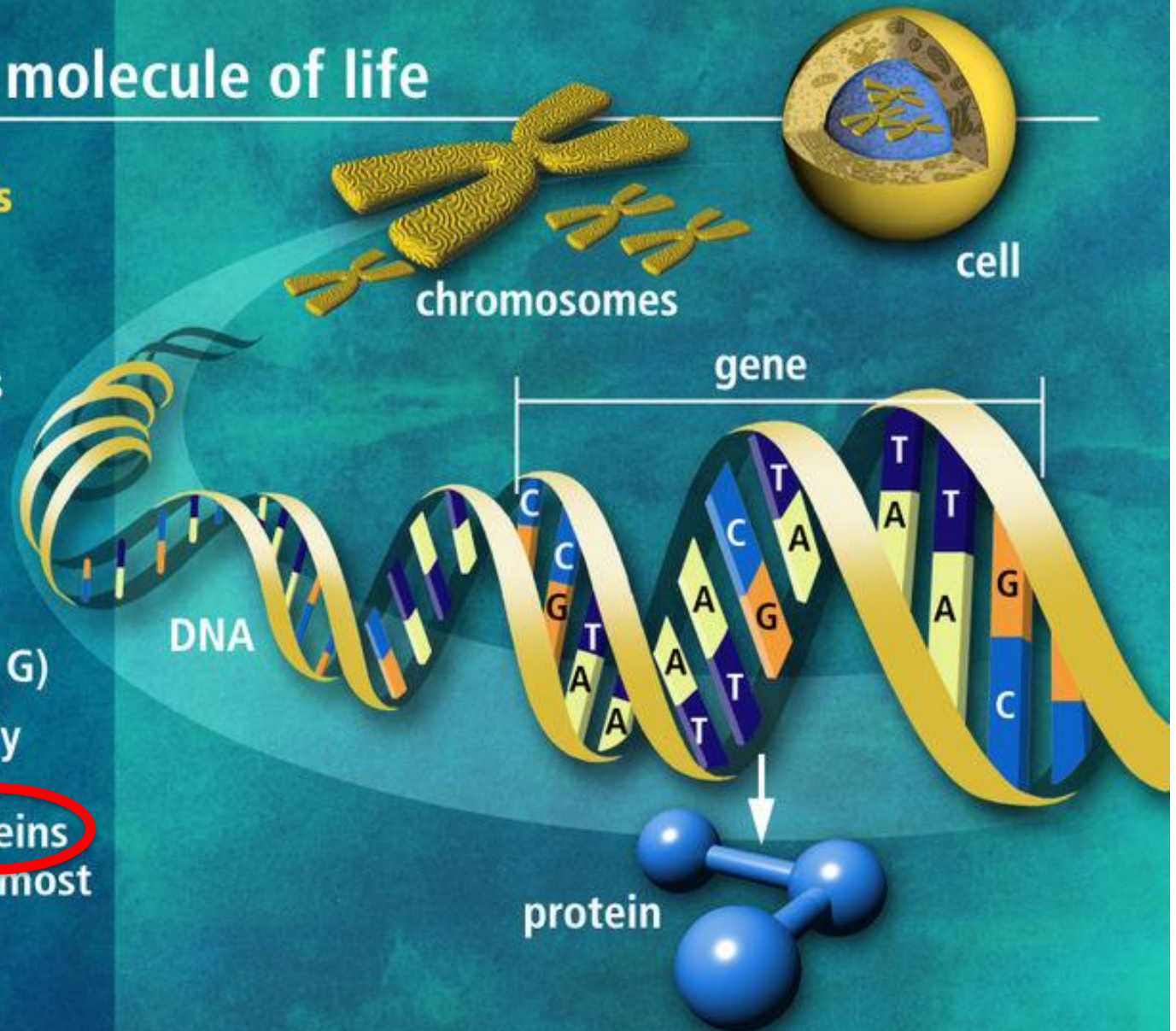
- Explain how traits are expressed and understand conditions that lead to mutations.
- Define mutation and distinguish non-genetic mutations from gene mutations.**
- Define and identify mutations that involve chromosome number and structure.
- Describe mutations in somatic cells versus sex cells.**
- Understand how genetics is used to produce desired traits in an organism (selective breeding; applied genetics).
- Science Practice: Bug Karyotype Lab**

DNA the molecule of life

Trillions of cells

Each cell:

- 46 human chromosomes
- 2 meters of DNA
- 3 billion DNA subunits (the bases: A, T, C, G)
- Approximately 30,000 genes code for **proteins** that perform most life functions

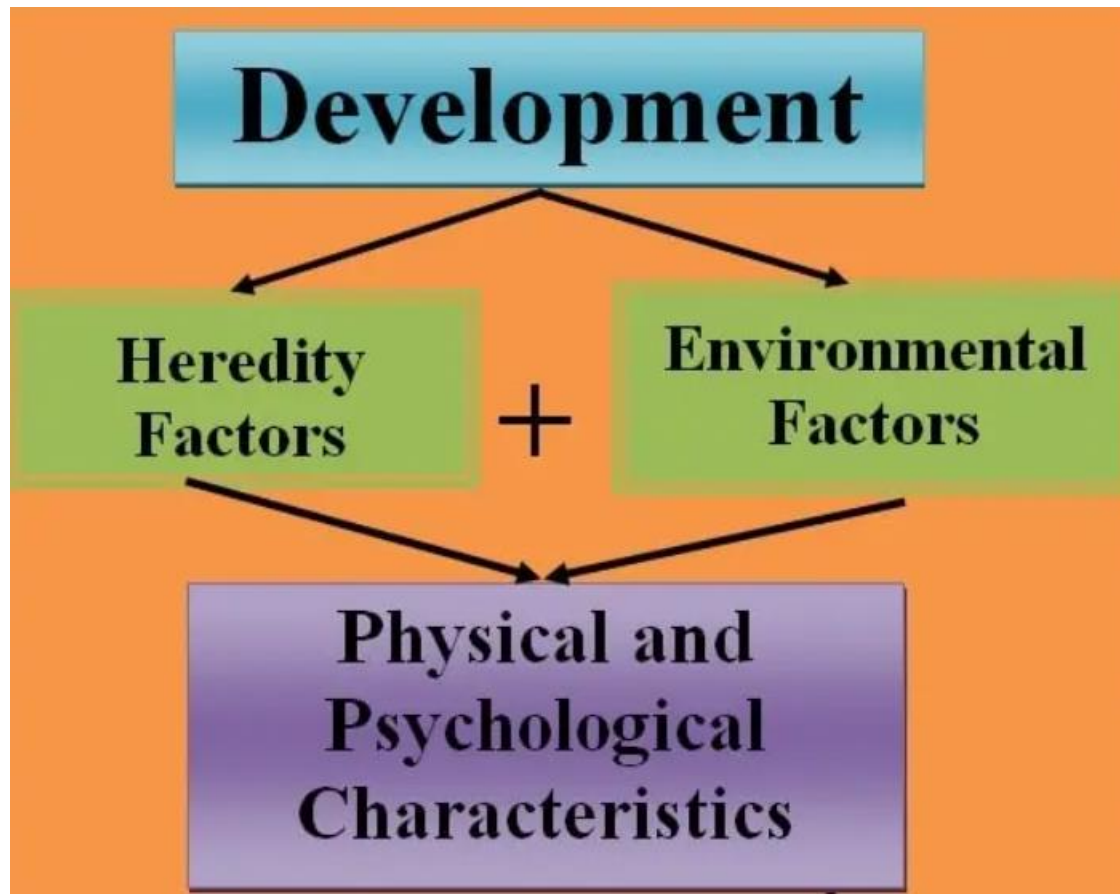


Genes control Phenotypic Traits through the Synthesis of Proteins

- DNA specifies Traits by dictating Protein Synthesis.
- Proteins are the links between **genotype** and **phenotype**.
- The **molecular chain of command** is from
 - **DNA** in the nucleus to **mRNA**,
 - **RNA** in the cytoplasm to ribosomes,
 - where **amino acids** → **protein**.

Expression of Traits

- **Genes** only determine **potential capacities**.
- **Environmental factors** play a large role in **genetic expression**.



Genetic Variation

- **Normal** and **Abnormal** types of genetic variation exist.
- Variation is critical for the survival of a population (e.g. adaptation).
- Occurs as a result of different alleles for a trait.



Normal Genetic Variation

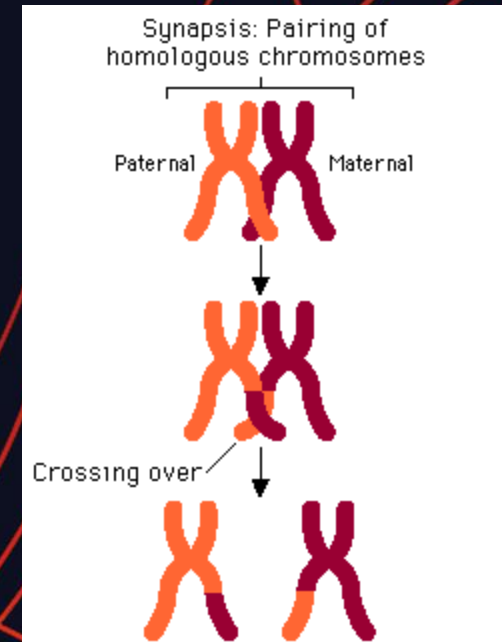
Occurs in **Meiosis** by

✓ **Independent Assortment**

✓ **Crossing Over**

when homologous chromosomes exchange pieces of the chromosomes as they are twisted around one another.

✓ **This results in an equal swap of the genes involved.**



ABNORMAL Genetic Variation

Mutation

- May occur in **somatic cells** (are not passed to offspring).
- May occur in **gametes (germ cells)** (eggs and sperm) and be passed to offspring.
- Can affect **one nucleotide only** or **larger segments of chromosomes**.



Mutation

- *A mutation is any change in structure or genetic material.*
- *Any change in a cell can be considered a mutation and may NOT be inherited or passed on to offspring.*
 - *Tumors, warts, moles.*
- *A **gene mutation** is a change in the DNA and will be inherited or passed onto offspring. However, many mutations involved recessive traits and are masked.*

Gene Mutations

- Changes in the **normal nucleotide sequence** of a gene.
- Almost all cause harm or death to the organism.
- They arise from a number of different mechanisms.



Types of Gene Mutations

1) Change in Chromosome Number

- Aneuploidy

- Non-disjunction
- Having more or less chromosomes

- Polyploidy

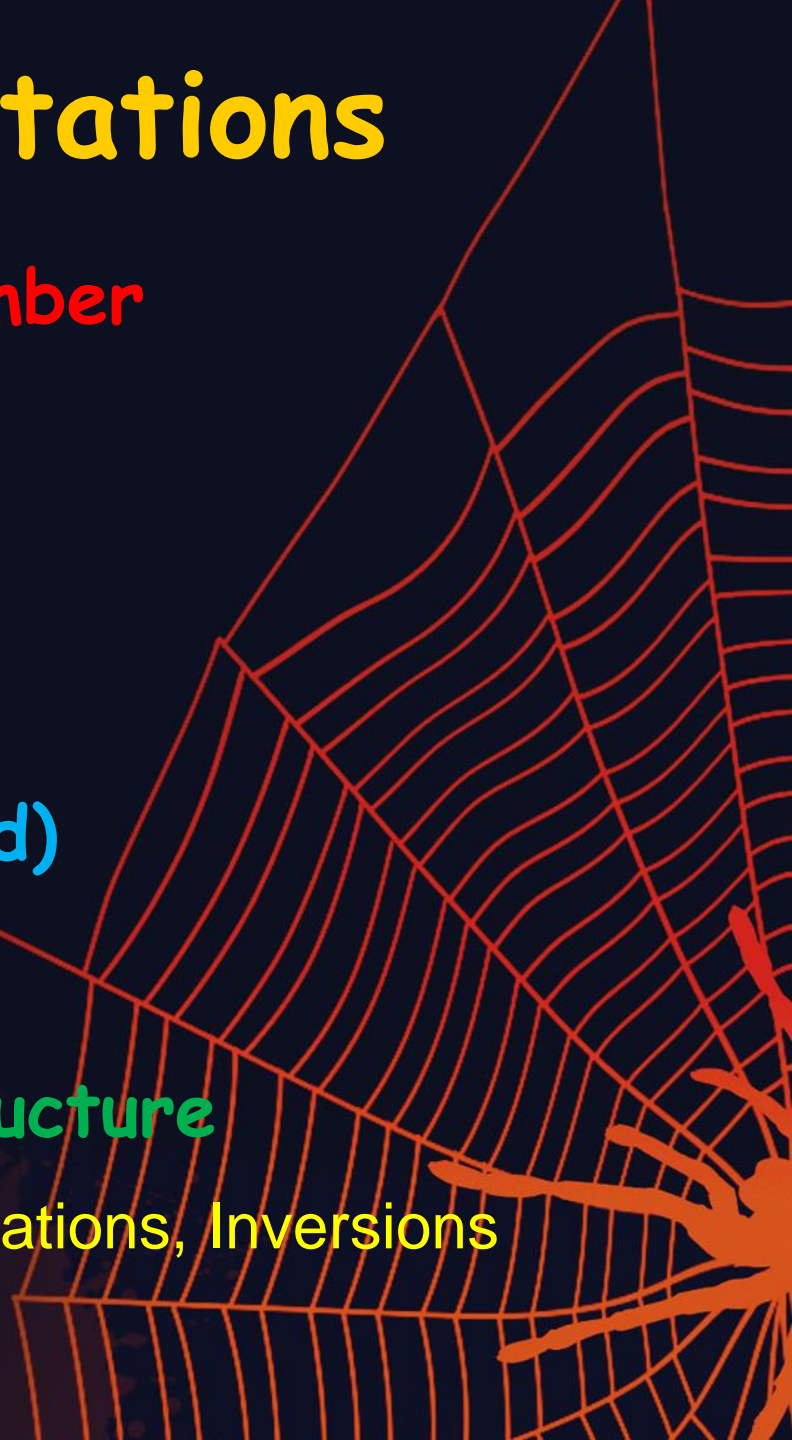
- Multiples of the $2n$ (diploid) number

2) Somatic Cells (not inherited)

vs Sex Cells (inherited)

3) Change in Chromosome Structure

- Deletions, Translocations, Duplications, Inversions
- Point Mutations



Point Mutations

- Substitutions

- "Silent"
- "Missense" - new protein (Amino Acid Substitutions)
- "Nonsense" - stop codon

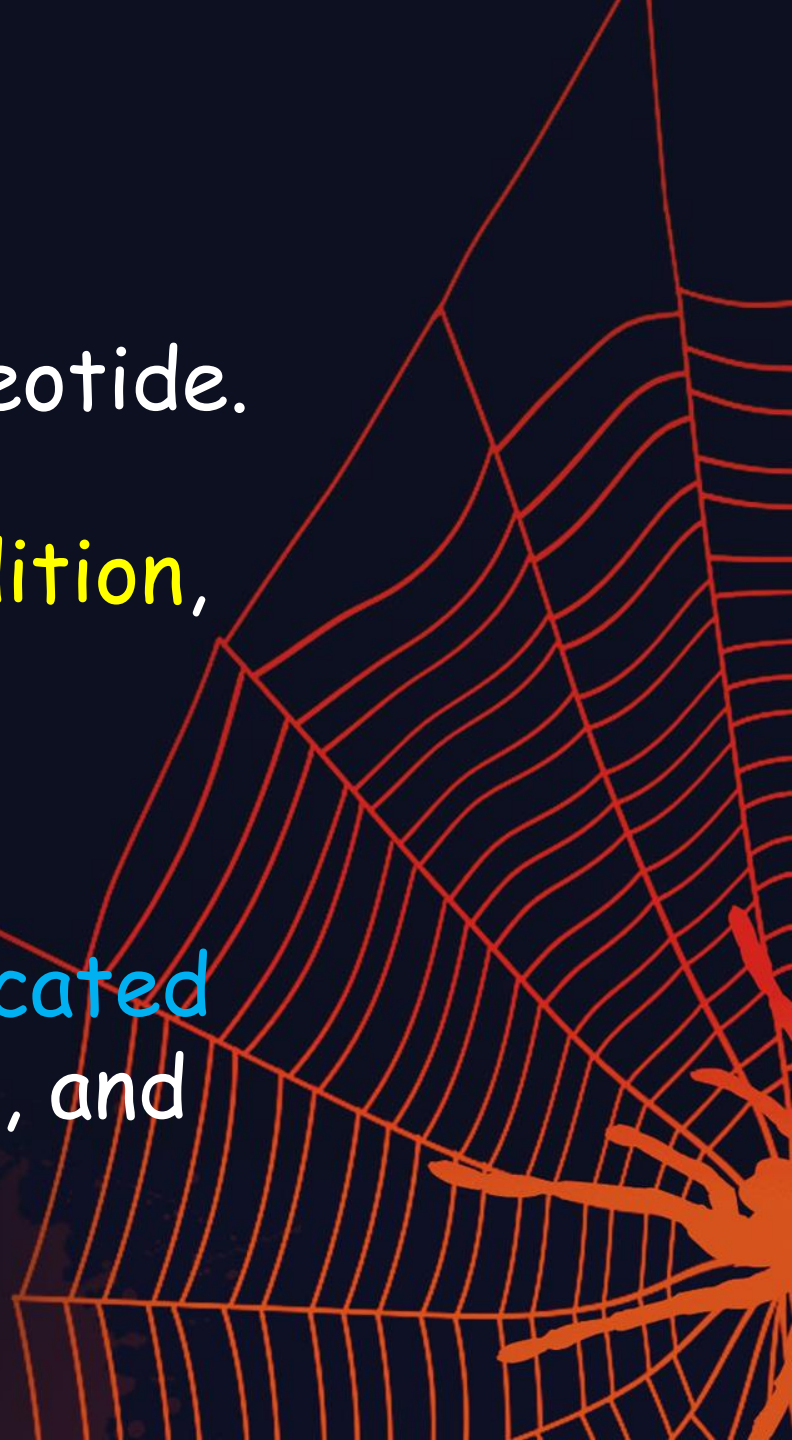
- Additions and Deletions

- Triplet Repeats
- Frameshift Mutations



Point Mutation

- Change of a **single** nucleotide.
- Includes the **deletion, addition,** or **substitution** of **ONE** nucleotide in a gene.
- Occurs when **DNA is replicated** during mitosis and meiosis, and a mistake occurs.



Point Mutations: Substitutions

- An incorrect nucleotide is inserted into the DNA instead of the correct one.

- Example:

A-T-A-G-G-G-C

A-T-A-A-G-G-C



Point Mutations: Substitutions

Silent Mutation

- A **substitution** mutation that does not cause any observable change in the protein or the function of the protein that the gene codes for.



Point Mutations: Substitutions

Silent Mutation

Example:

- ✓ Both AAA and AAG code for phenylalanine.
- ✓ If the third A in AAA undergoes a substitution mutation so that A is changed to a G, then it still codes for phenylalanine.

Point Mutations: Substitutions

Silent Mutation

Example:

- ✓ A substitution mutation can occur that alters a group of nucleotides in such a way that the amino acid is changed.
- ✓ But, if the new amino acid has similar properties to the original amino acid, then the protein may still function normally.

Point Mutation: Substitutions

Missense Mutation

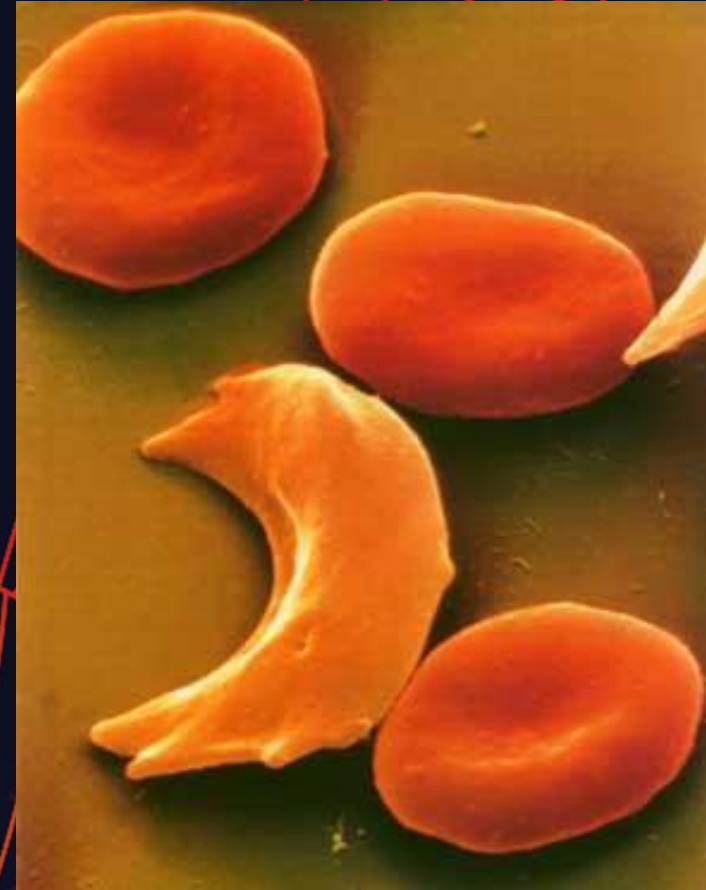
- Caused by **Substitutions**
- It changes the group of nucleotides and results in the **insertion of the incorrect amino acid into the protein** during protein synthesis.
- The results could be severe or silent.



Point Mutation: Substitutions

Missense Mutation

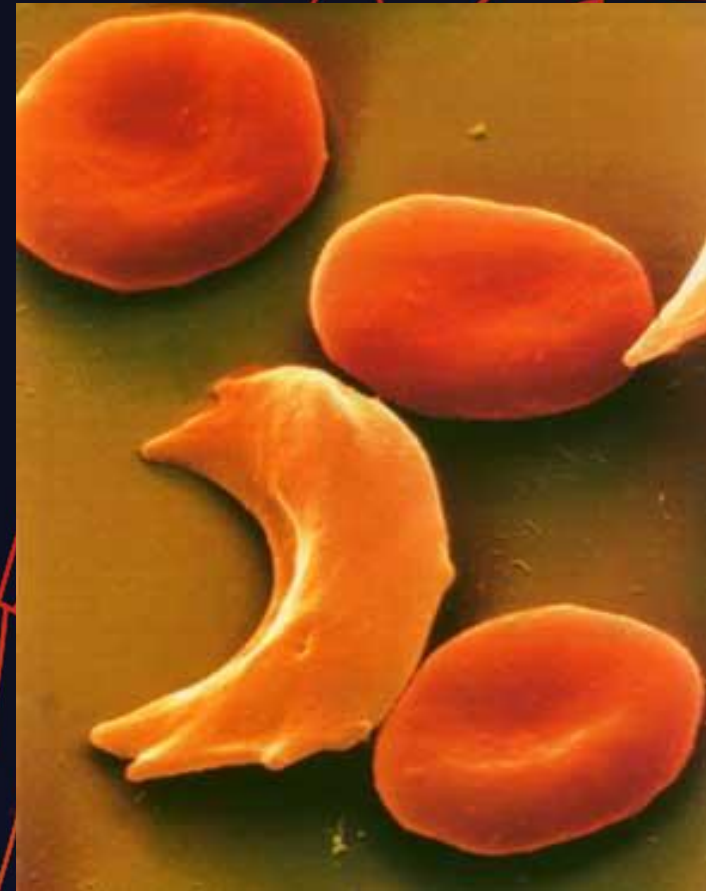
- **Sickle Cell Disease** is the result of one nucleotide substitution.
- Occurs in the **hemoglobin gene**.
- **Hemoglobin** is a protein that carries oxygen to our cells and carbon dioxide away from the cells. Exists inside our red blood cells.



Point Mutation: Substitutions

Missense Mutation

- This mutation changes the "A" in "GAG" to a "T".
- During translation, this results in the amino acid **valine** being inserted into the protein instead of the correct amino acid: **glutamic acid**.
- Because of this "minor" change a completely **dysfunctional** hemoglobin molecule is formed.
- Causes **Sickle Cell Disease**, a very severe blood disease that mainly affects people of African descent.



Point Mutation: Substitutions

Nonsense Mutations

- Caused by **Substitutions**
- It alters an amino acid encoding a group of nucleotides into a **STOP** signaling group.
- When the message is read by the ribosome, **the protein will stop being made ... too early.**
- It will be smaller and minimally functional.

Deletion and Addition Point Mutations

- **Deletions:** occur when a nucleotide is left out of its proper sequence.
- **Additions:** occur when a single nucleotide is added during replication.



Point Mutation - Reading Frame

- Since RNA translates DNA **3 base sequences (codons)**, the exact sequence must be maintained = **READING FRAME**.
- Substitution mutations **DO NOT** alter the reading frame.
- **Deletion and addition** mutations alter the **READING FRAME**.

Point Mutation - Reading Frame →

Frame Shift Mutations

- **FRAME SHIFT MUTATIONS** result from deletion or addition point mutations.
- These are usually more harmful than substitution mutations.



Point Mutation - Reading Frame → Frame Shift Mutations

Normal:

THE DOG RAN AND ATE THE PIG

Mutated:

THE DGR ANA NDA TET HEP IG

This is a deletion mutation ("O" in DOG).

Frameshift Mutation

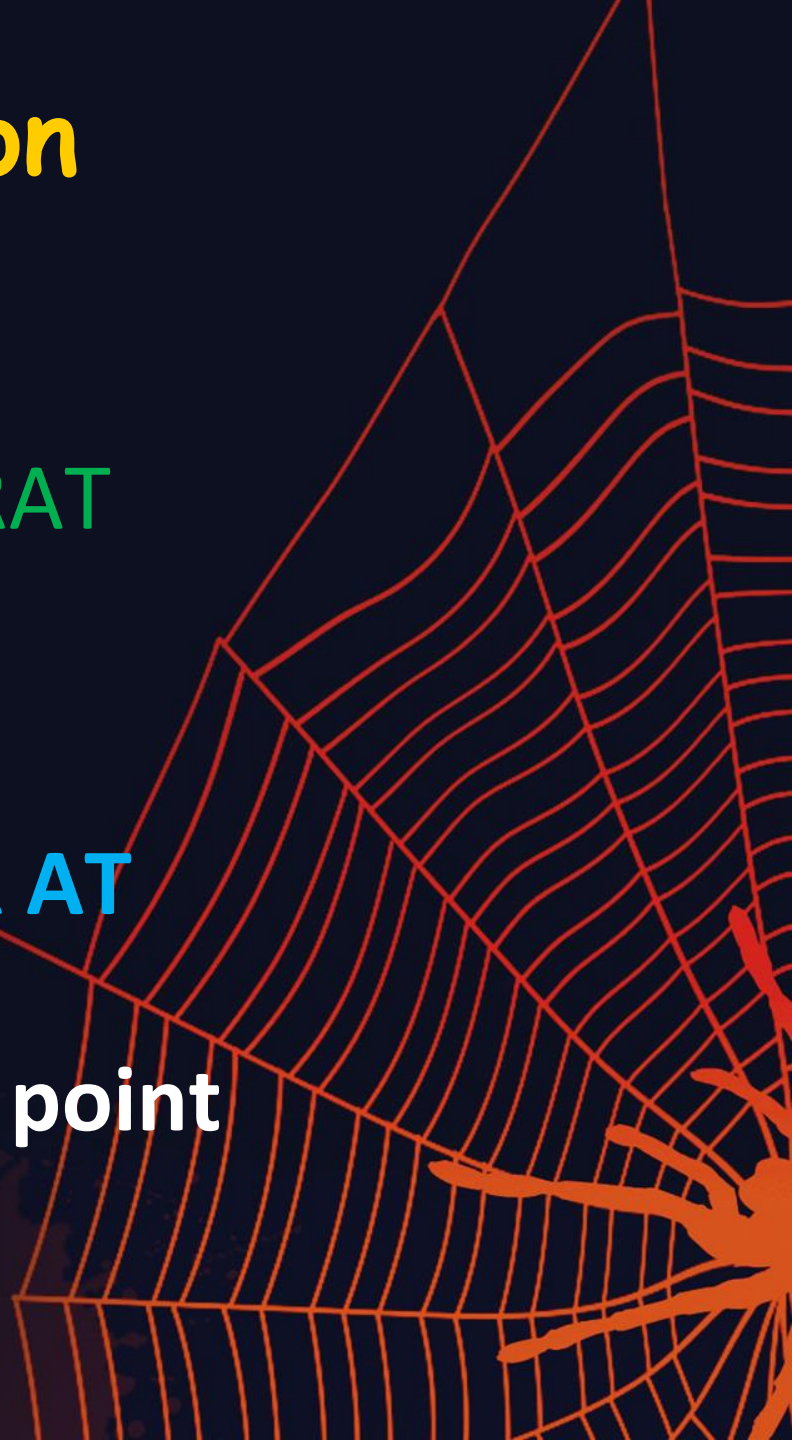
Original:

THE FAT CAT ATE THE WEE RAT

Frame Shift (“a” added):

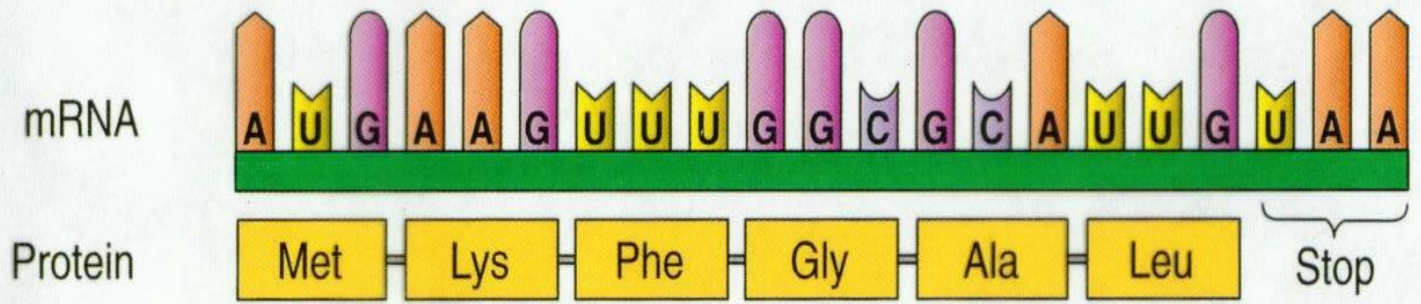
THE FAT CA**A**TET HEW EER AT

This was due to an addition point mutation.

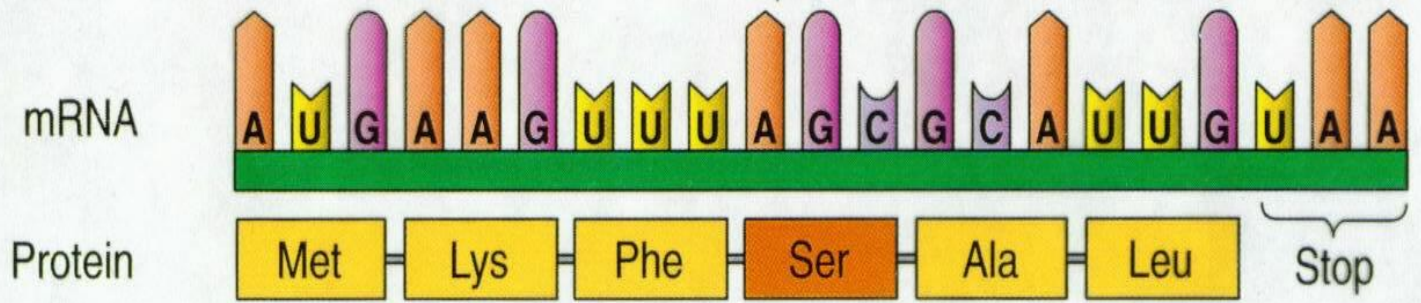




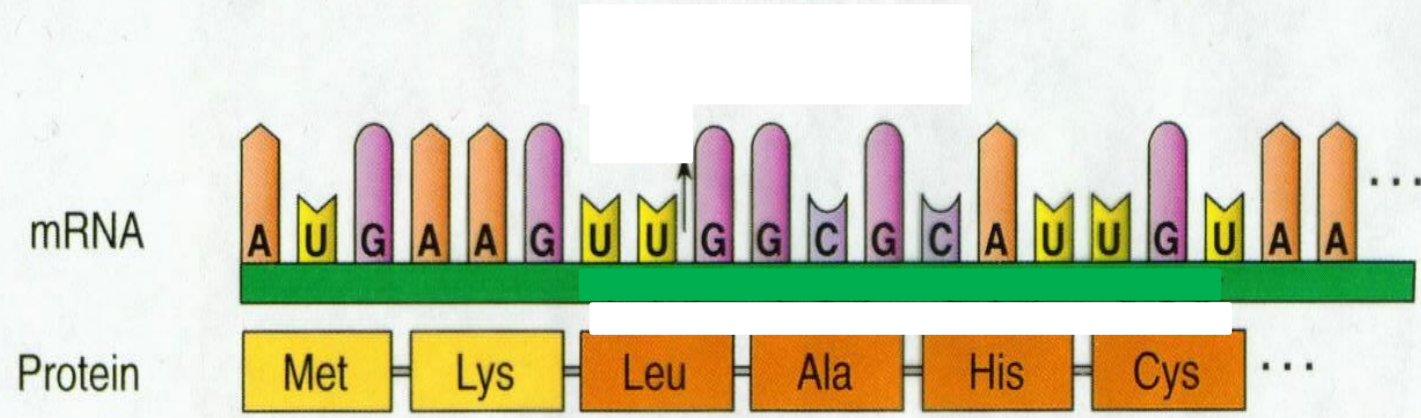
Normal



What type of mutation is shown here?

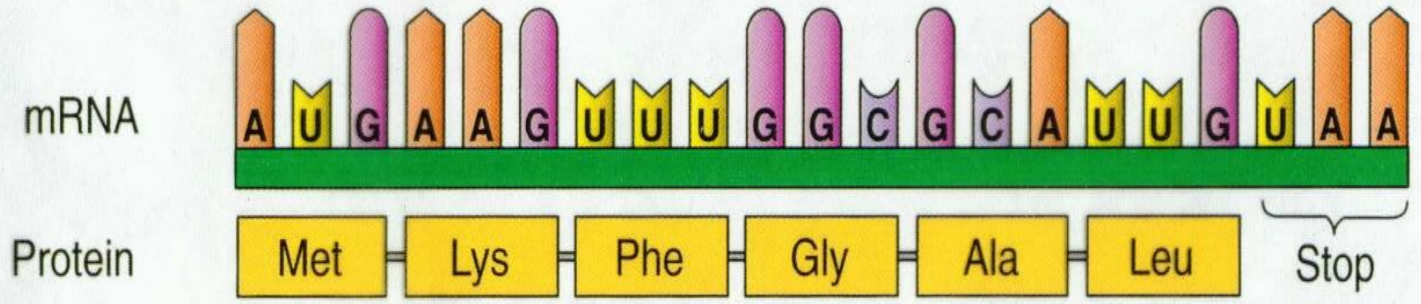


What type of mutation is shown here?

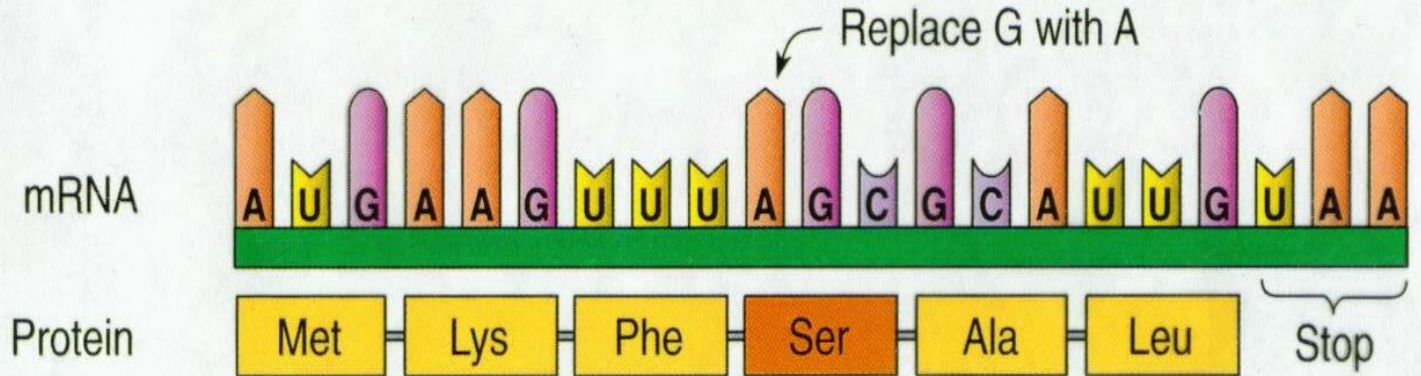




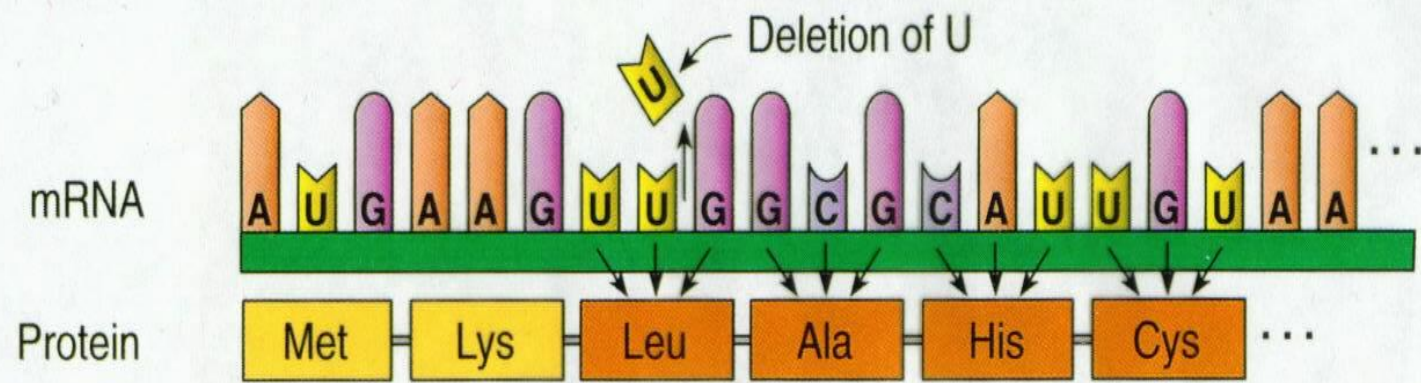
Normal



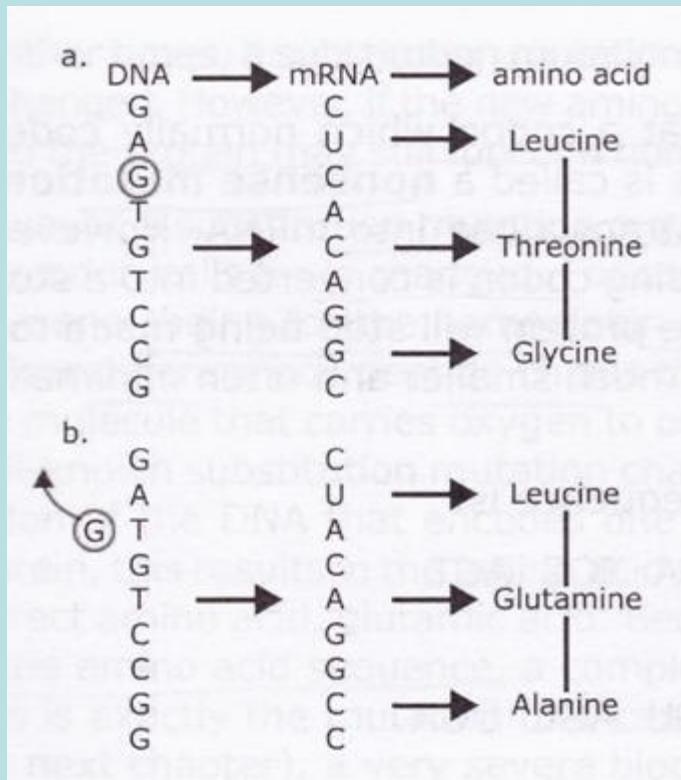
Point mutation



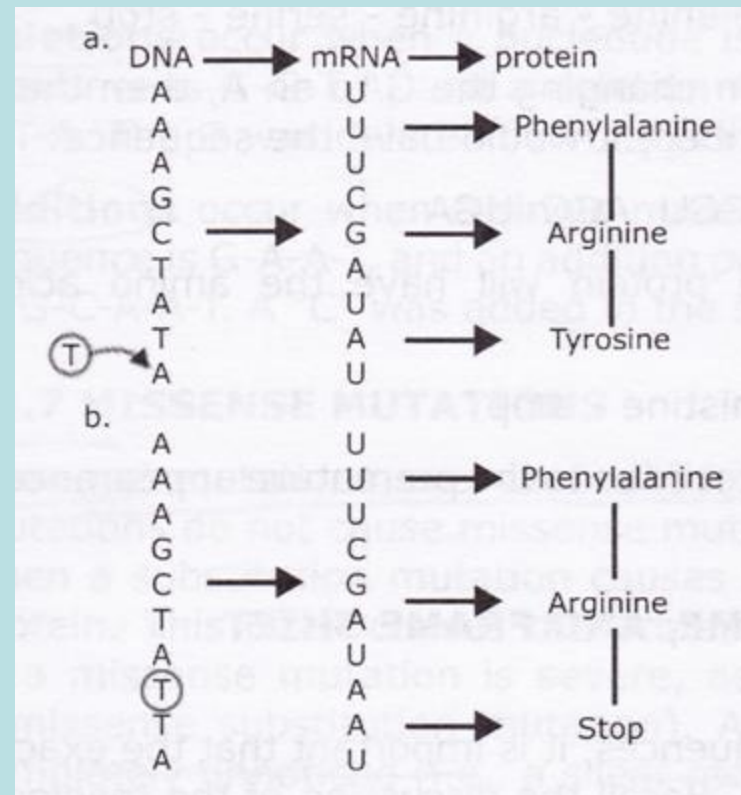
Frameshift mutation



Amino Acid Sequence Changed



Deletion Mutation



Addition Mutation

Chromosome Mutations

May Involve changing the
STRUCTURE of a chromosome

- The **loss or gain** of part of a chromosome.
- Most occur during mitosis and meiosis.



Chromosome Structure Mutations

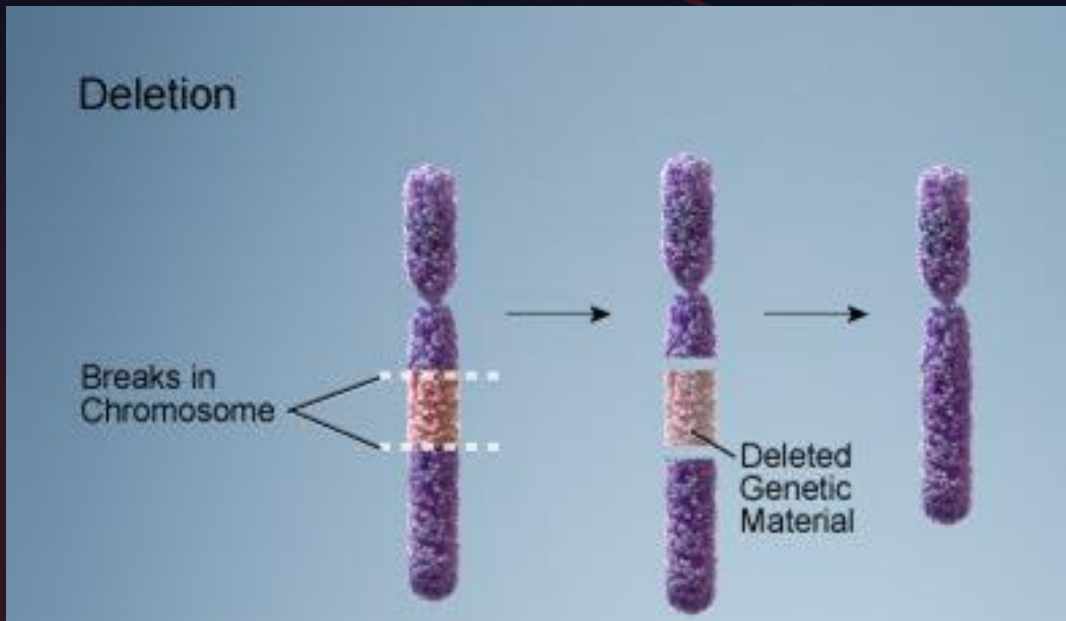
Five types exist:

- Deletion
- Inversion
- Duplication
- Translocation
- Nondisjunction



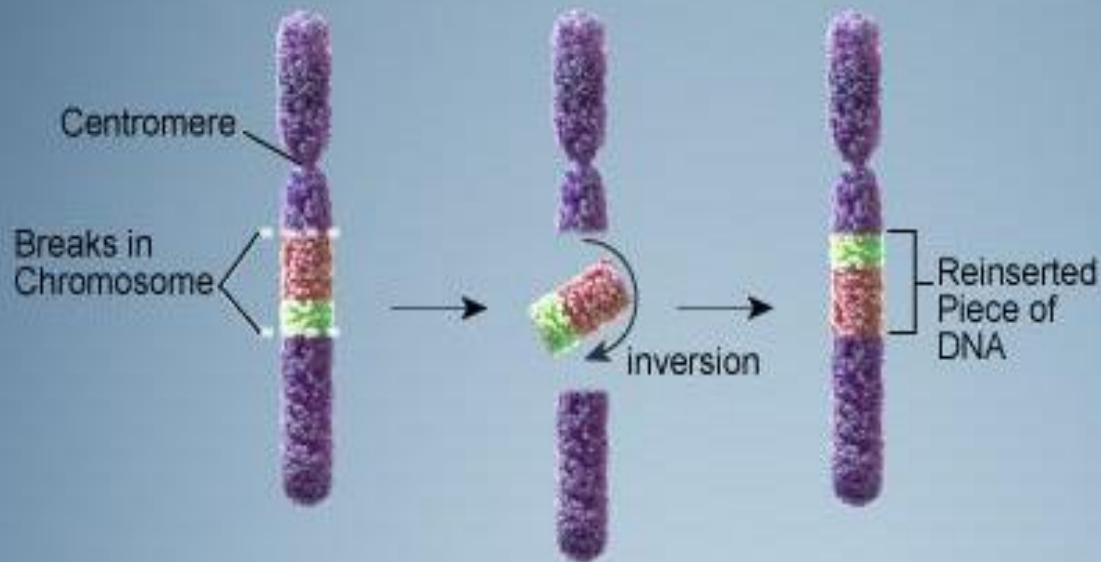
Structural Deletion Mutation

- Due to **breakage**.
- A **piece** of a chromosome is **lost**.
- Usually results in death of organism.



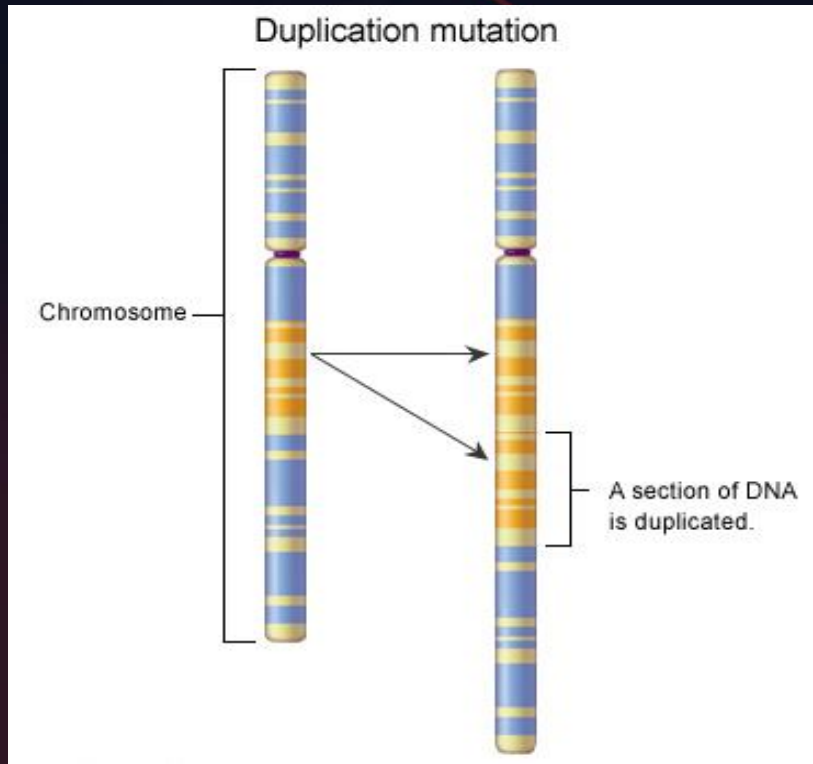
Structural Inversion Mutation

- Chromosome segment **breaks off**.
- Segment flips around **backwards**.
- Segment **reinserts** into the same chromosome.



Structural Duplication Mutation

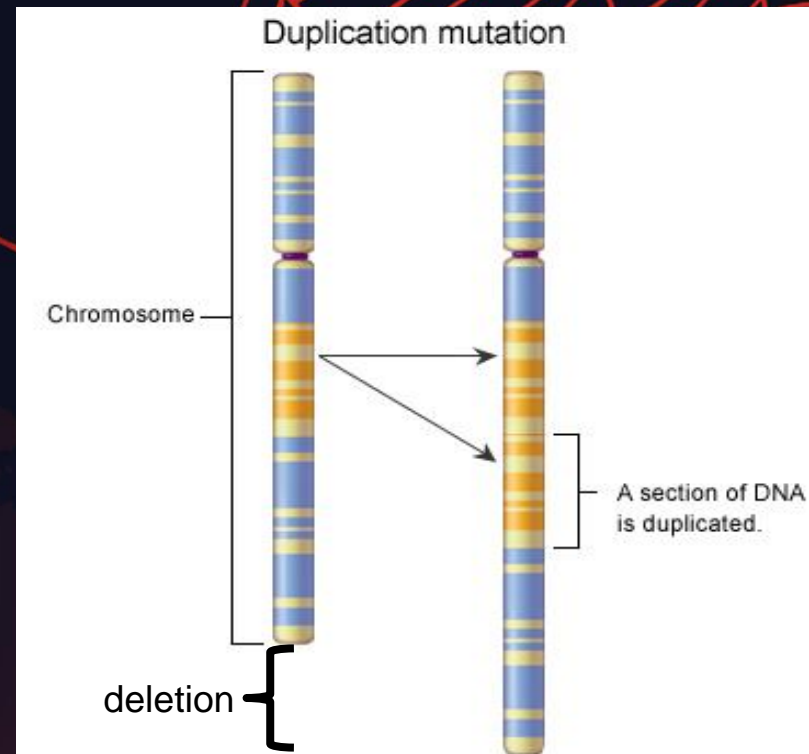
Occurs when a piece of one homologous chromosome breaks off and is inserted into the other homologous chromosome.



Structural Duplication Mutation

Since the chromosomes are homologues, the chromosome that donated the segment has undergone a **deletion** mutation.

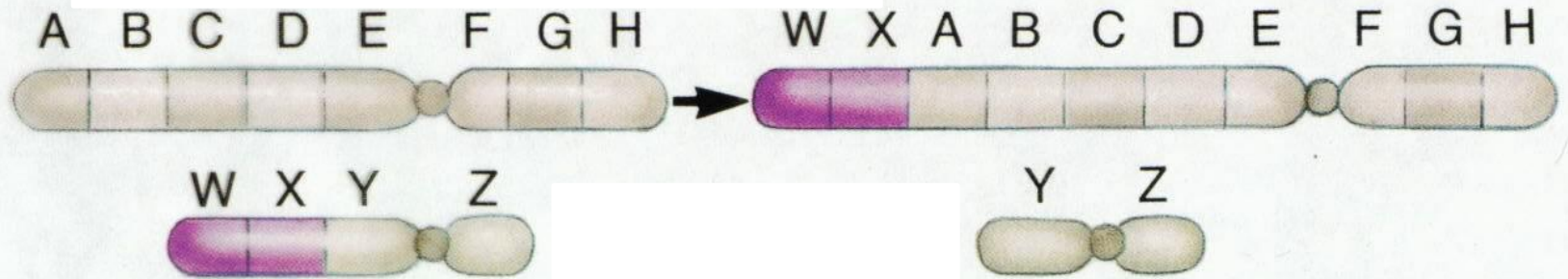
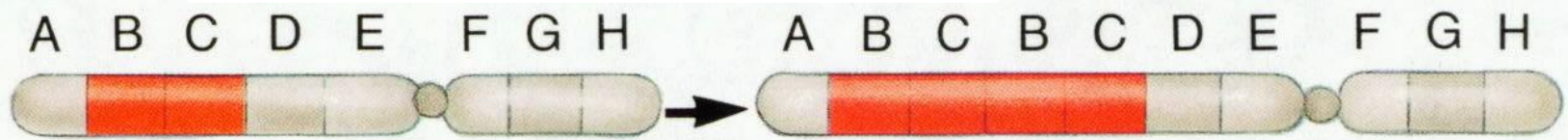
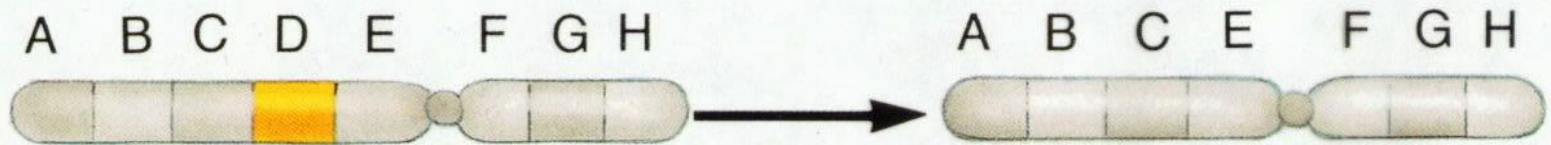
And the chromosome that receives the segment has **DUPLICATION** of genes.



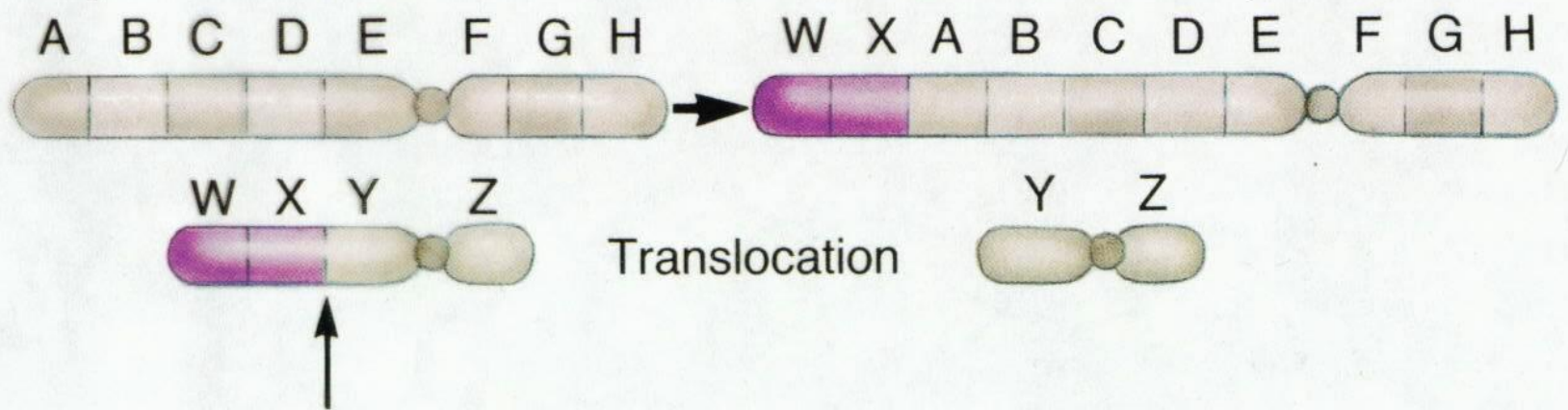
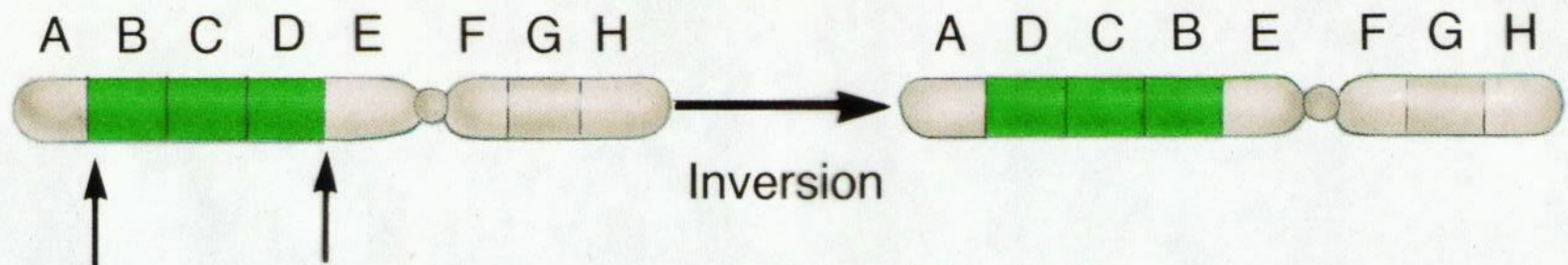
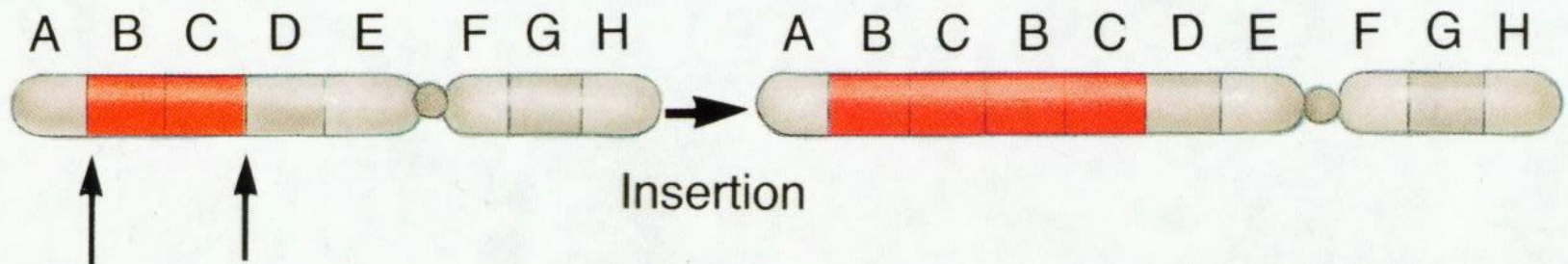
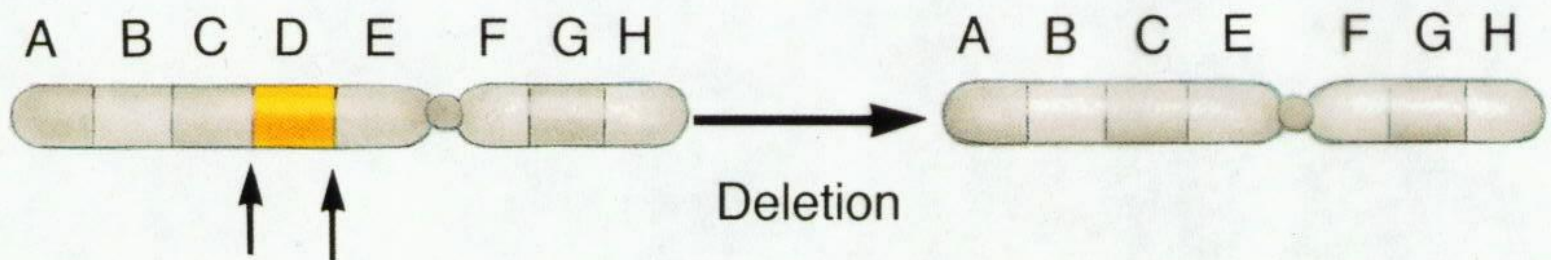
Structural Translocation Mutation

- Involves **two chromosomes** that **aren't homologous**.
- A segment of a chromosome breaks off of one chromosome and inserts into a **non-homologous chromosome**.





What type of mutation is shown in each case?

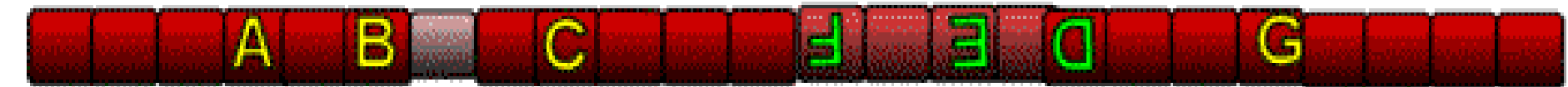
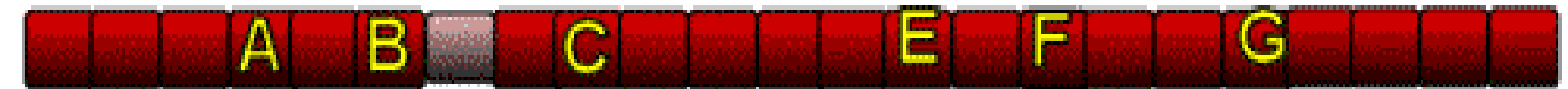


Original Chromosome

What type of mutation is shown in each case?



TRY IT



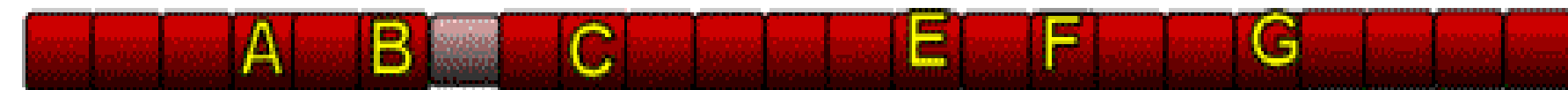
Original Chromosome



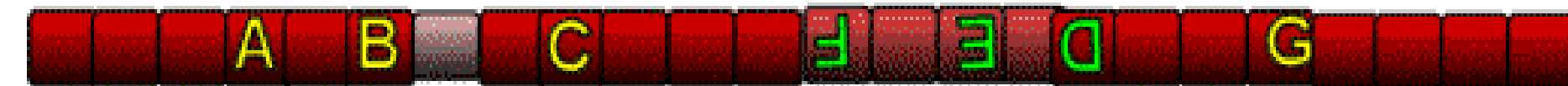
Duplication



Deletion



Inversion



Inversion



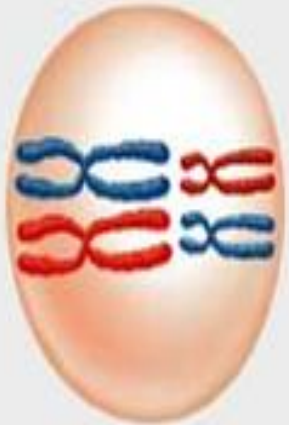
Nondisjunction

Chromosomal Mutation

- Failure of chromosomes to separate during meiosis.
- Causes gamete to have too many or too few chromosomes.
- E.g. Down Syndrome (Trisomy-21)



NONDISJUNCTION



$2n = 4$
 $n = 2$



$n + 1$



$n + 1$



$n - 1$



$n - 1$

1. Meiosis I starts normally. Tetrads line up in middle of cell.

2. Then one set of homologs does *not* separate (= nondisjunction).

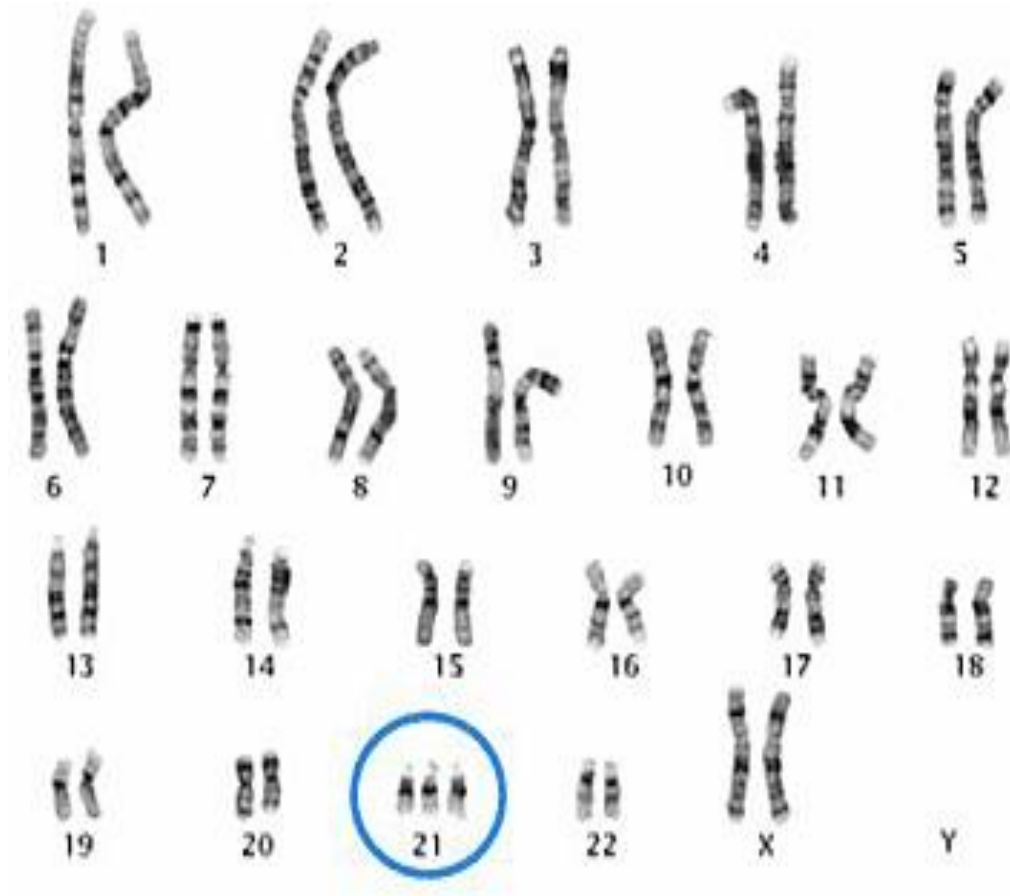
3. Meiosis II occurs normally.

4. All gametes have an abnormal number of chromosomes—either one too many or one too few.

An extra copy of Chromosome 21 Causes Down Syndrome

Trisomy 21

- involves the inheritance of **three copies of chromosome 21**.
- the MOST COMMON human chromosome abnormality.



MUTAGENS

Factors that are known to cause mutations (changes in DNA which lead to cancer).



Cancer

ENVIRONMENTAL CONDITIONS
might trigger a cell to become cancerous.

Radiation

Drugs & Medication

Viruses

Chemicals

Materials (e.g. asbestos)

Cancer

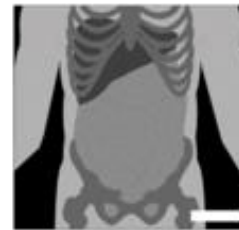
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Carcinogens

Radiation

UV Radiation
both natural sunlight
and tanning beds



X-Rays
medical, dental,
airport security screening

Chemicals

Cigarette Smoke
contains dozens of
mutagenic chemicals



Benzoyl Peroxide
common ingredient
in acne products

**Nitrate and Nitrite
Preservatives**
in hot dogs and
other processed meats

Barbecuing
creates mutagenic
chemicals in foods

Infectious Agents

**Human Papillomavirus
(HPV)**
sexually transmitted virus



Helicobacter pylori
bacteria spread through
contaminated food



How is genetics used to produce desired traits in an organism?



Many-colored carrots

Scientists bred carrots for high levels of the orange pigment beta-carotene, which is high in vitamin A.

Other pigments such as red, yellow, purple, and white are high in other types of nutrients.

Selective Breeding and Society

- The world food supply has been greatly enhanced by selective breeding and genetic engineering.
- Hybridization is very important in producing new varieties of organisms.
- Selective breeding produced many kinds of flowers, pets, and other organisms.



Selective Breeding Is **Artificial Selection**

Artificial selection

- Selective breeding is the same as artificial selection.
- Human intervention in animal or plant reproduction to ensure that certain desirable traits or combinations of traits are passed on to future generations.
- Artificial selection is goal-directed and purposeful.
- **Natural selection** is directed by survival in the environment.



Selective Breeding of Organisms for Food

Plants:

- Faster growth
- Higher yield
- Disease resistance
- Higher nutritional value

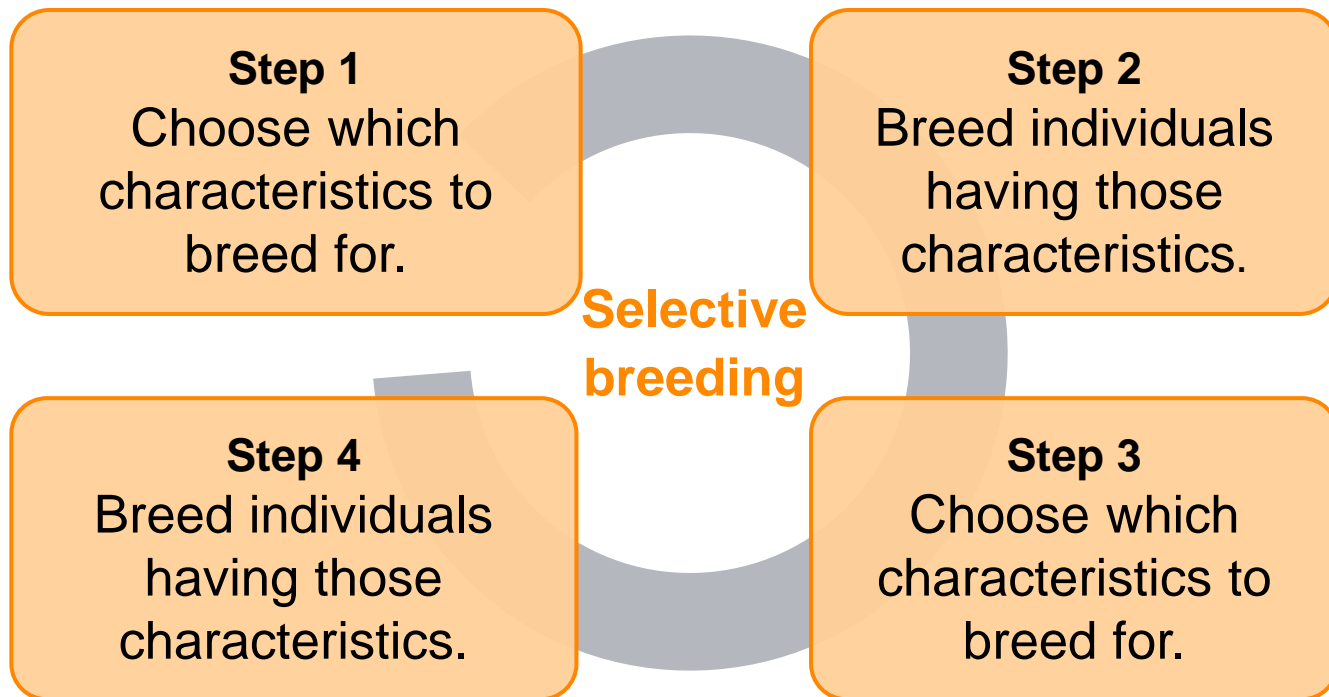
Animals:

- Faster growth
- Greater docility (less aggression)
- Higher milk, egg, honey, or meat production

Genetic engineering makes breeding specific – they target by adding specific genes.

Process of Selective Breeding

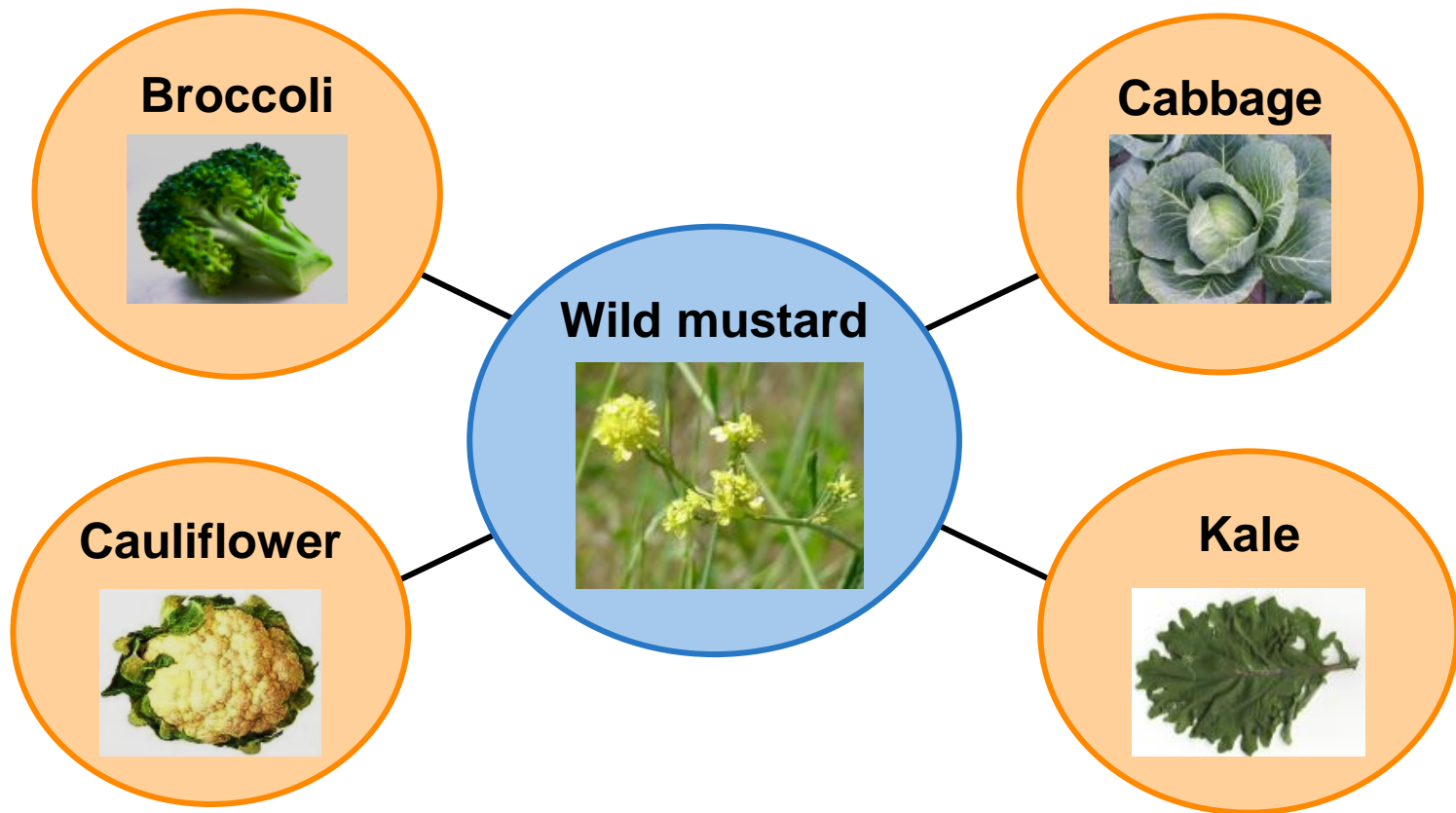
Each generation, desired characteristics increase in population.



Selective Breeding

- Selective breeding is a form of artificial selection to produce plants and animals with more desirable traits.
- Selective breeding is goal-directed, not random like natural selection.
- The process involves choosing the best parents to breed, then breeding offspring to build up traits in a population.
- Some problems include inbreeding—buildup of negative genes—and decreased population diversity.
- Wild mustard and wild cattle have been extensively selected.

From the Wild to the Garden





Selectively Bred Cow Traits

Cows that are domesticated:

- are herbivores.
- are large and relatively inactive.
- can be herded.
- provide milk.

Cows used for beef:

- are short and stocky
- have lots of muscle.

Cows used for dairy production:

- have long limbs & body build.
- have large udders.
- produce high volumes of milk.





Luther Burbank (1849–1926)

- Was an American plant breeder.
- Was famous for “Burbank potato”.
- Developed more than 800 new strains and varieties of plants.
- Increased world food supply.





Origins: Flowers and Pets

Today's flowers and pets came from ancient wild ancestors.



Wild rose –
ancestor of today's
hothouse roses



Grey wolf –
ancestor of today's
dogs



African wild cat –
likely ancestor of
today's house cats

Hybridization

Hybridization

Breeding between individual organisms with different parentage; reproduction between organisms that are distantly related

- Mendel is the best-known early plant hybridizer.
- Many crops are hybrids, combining good traits from different sources to restore “hybrid vigor.”



Which of the following are potential benefits of selective breeding?

- Crops with higher yield.
- Crops with better nutritional value.
- More aggressive farm animals.
- Higher milk, egg, or meat production.
- Decreased diversity of wild populations.



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Crops with higher yield.

Crops with better nutritional value.

More aggressive farm animals.

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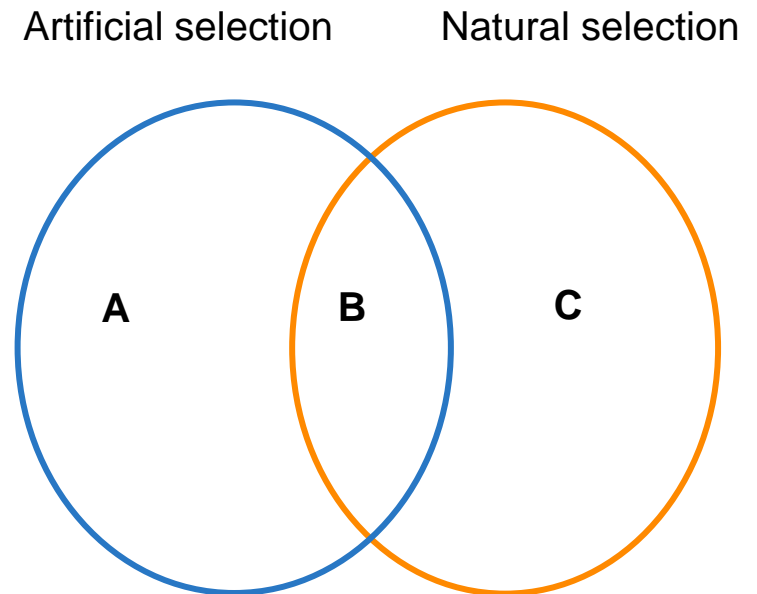
Comparing Artificial and Natural Selection

Decide if the following statements apply only to artificial selection (A), only to natural selection (C), or to both (B).

Results in genetic change in next generation.

Is goal-directed.

Is a result of individuals that best survive in their environment and reproduce.





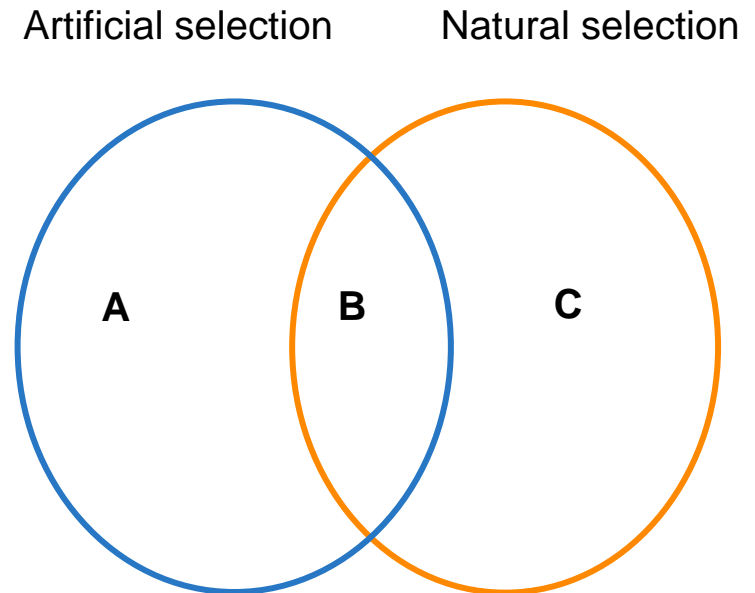
Comparing Artificial and Natural Selection

Decide if the following statements apply only to artificial selection (A), only to natural selection (C), or to both (B).

Results in genetic change in next generation. **B**

Is goal-directed. **A**

Is a result of individuals that best survive in their environment and reproduce. **C**



Potential Problems with Artificial Selection - **Inbreeding**

Inbreeding

Breeding between individual organisms who share similar genetic makeup or parentage; reproduction between organisms that are closely related.

Inbreeding causes unintended consequences:

- Negative, recessive traits build up in the population.
- The population becomes weaker and has less resilience.
- Diversity decreases in populations.

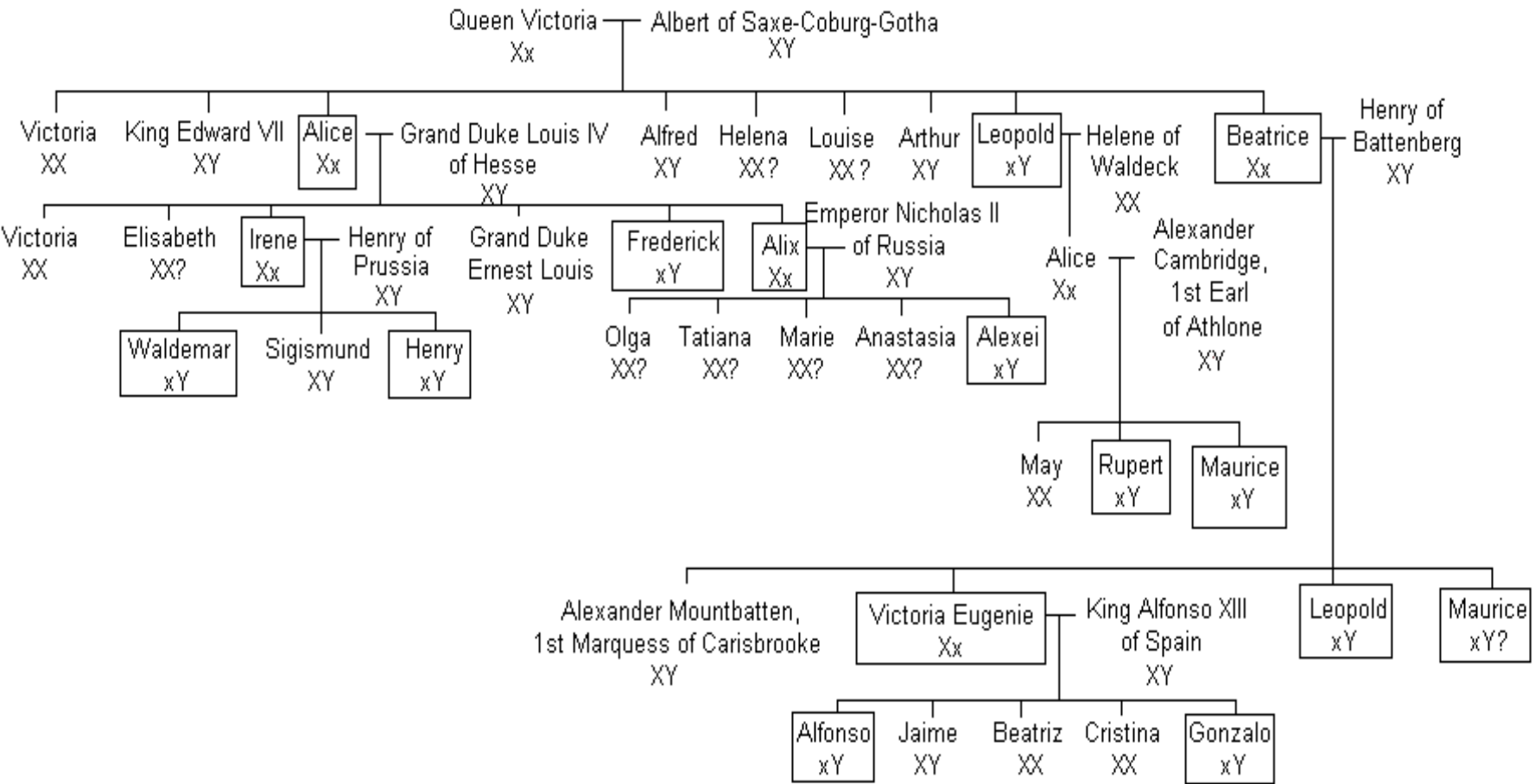


Human Inbreeding: Hemophilia

The British Haemophilia Line

KEY

- X: Unaffected X-chromosome
- Y: Y-chromosome
- x: Affected X-chromosome



Human Inbreeding: Hemophilia





Why can inbreeding be potentially harmful to populations? Check all that apply.

- Negative traits, such as diseases, may build up in the population.
- Useful traits may be “bred out” of the population.
- Genetic diversity of the population is increased.
- The population becomes weaker and less resilient.



Why can inbreeding be potentially harmful to populations? Check all that apply.

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Identifying Future Traits For Breeding

Agricultural geneticists research and brainstorming future plant foods that could be developed using selective breeding either by general methods or genetic engineering.

- resistance to pesticides
- addition of genes to make animal protein
- resistance to insects, fungi, and other pests
- addition of genes to improve color and taste
- modifications for specific climates (heat, cold, drought)
- addition of genes to increase concentrations of essential vitamins
- modification of food's appearance to make it more attractive to consumers