MUTATION IN HUMAN POPULATION

A PRESENTATION FOR
DEPARTMENT OF MEDICAL BIOLOGY
CRIMEA STATE MEDICAL UNIVERSITY
SIMFEROPOL , CRIMEA .

STUDENTS NAME : SHEIKH SANA
GROUP : 191A
GUIDED BY :
ANNA ZHUKOVA ALEXANDROVNA MAM

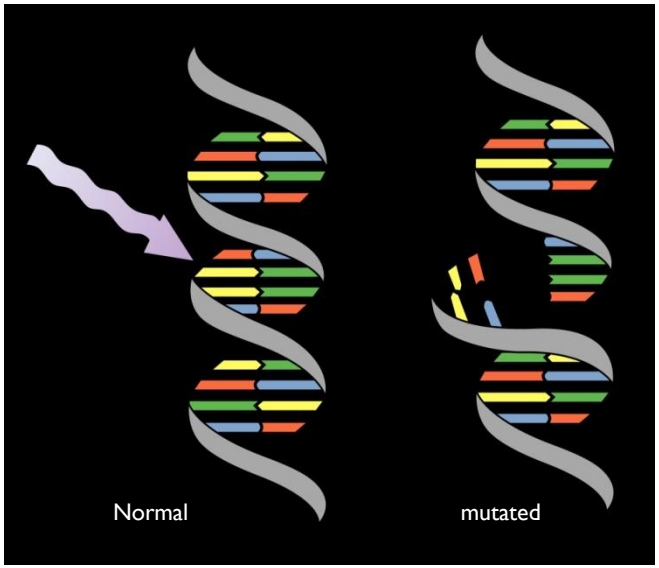
MUTATION

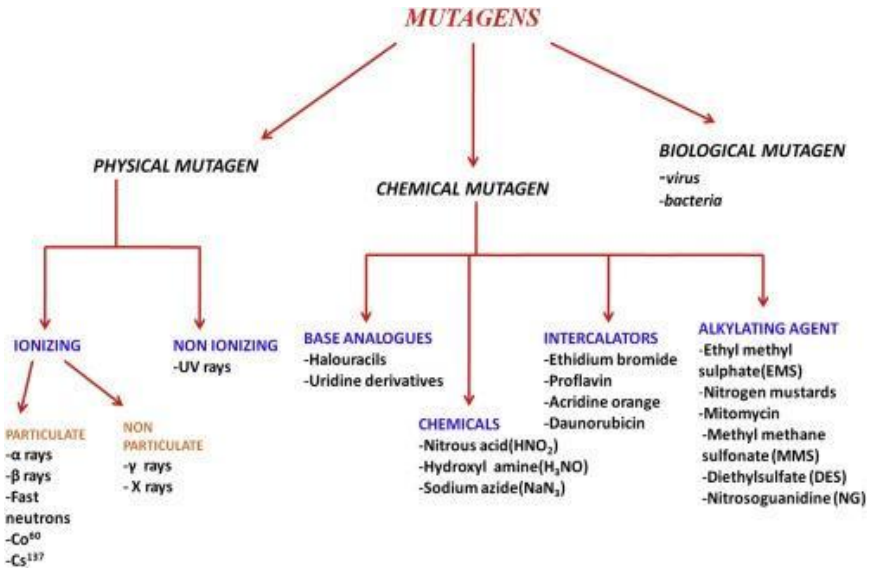
- It is a change that occurs in our DNA sequence , or abnormal changes in chromosomes .
- It causes by various factors known as MUTAGENIC FACTORS
- Mutagenic factors are of 2 types :

EXOGENIC factors : that are outside our body or environmental factors

1. physical factors : X-rays , UV light , particle radiation (alpha,beta ,etc)
2. chemical factors : carcinogens , benzo(a)pyrene , colchicine
3. biological factors : by bacteria and viruses

ENDOGENC factors : that are inside human body , it includes wrong DNA replication





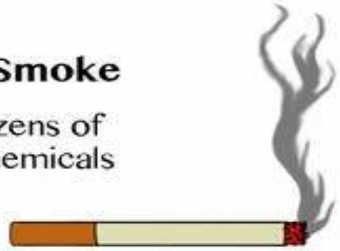
Radiation

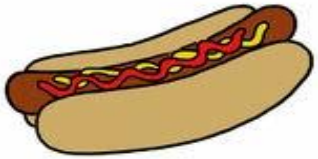
UV Radiation
Both natural sunlight and tanning beds

X-Rays
Medical, dental, airport security screening

Chemicals

Cigarette Smoke
Contains dozens of mutagenic chemicals





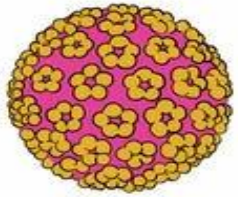
Benzoyl Peroxide
Common ingredient in acne products

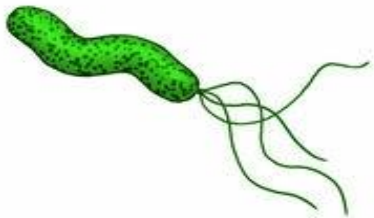
Nitrate & Nitrate Preservatives
In hot dogs and other processed meats

Barbecuing
Creates mutagenic chemicals in foods

Infectious Agents

Human Papillomavirus (HPV)
Sexually transmitted virus





Helicobacter pylori
Bacteria spread through contaminated food

HISTORY

INTRO-

The mutations in genes was discovered by Hugo De Vries.

He began this discovery quite by accident.

Following along the lines of Gregor Mendel, De Vries crossed primroses and saw that when they grew, the varieties were sometimes different. He called these differences, mutations.

TYPES OF MUTATION

- MAINLY 2 TYPES OF :
  - GENE MUTATION
  - CHROMOSOMAL MUTATION

GENE MUTATION VERSUS CHROMOSOMAL MUTATION

Gene mutation is an alteration of the nucleotide sequence of a gene	Chromosomal mutations are alterations in the chromosome structure or chromosome number
Caused by errors in DNA replication and mutagens such as UV and chemicals	Caused by errors in crossing over during meiosis
The alteration occurs in the nucleotide sequence of a gene	The alteration occurs in a segment of a chromosome
A single gene is affected	Several genes are affected
Influence is comparatively low	Can sometimes be lethal
Can cause sickle cell anemia, hemophilia, cystic fibrosis, Huntington syndrome, Tay-Sachs disease, and cancers	Can cause Klinefelter syndrome, Turner syndrome, and Down syndrome

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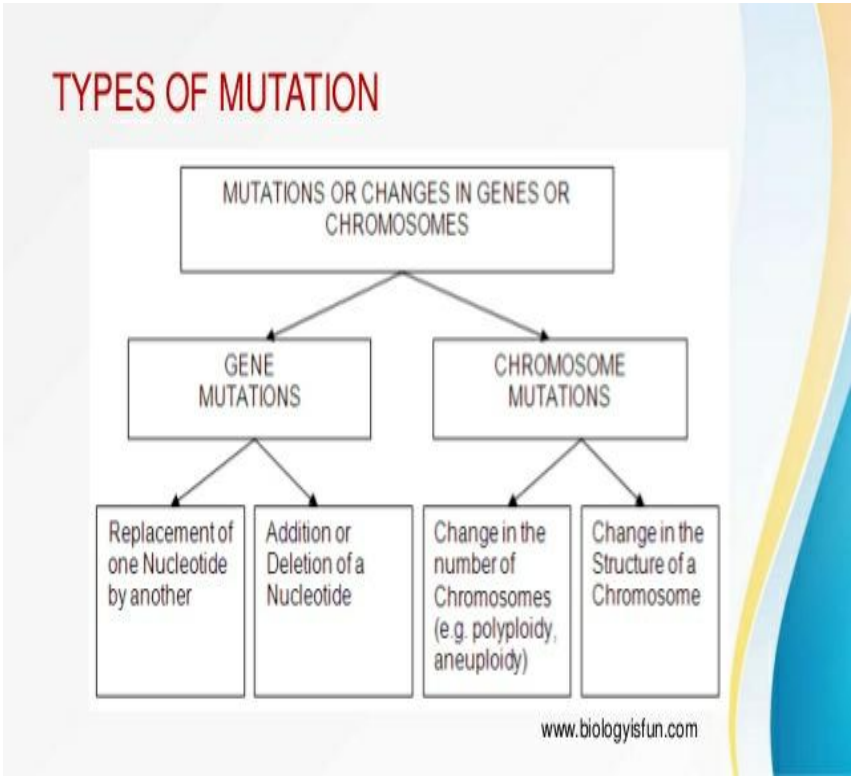
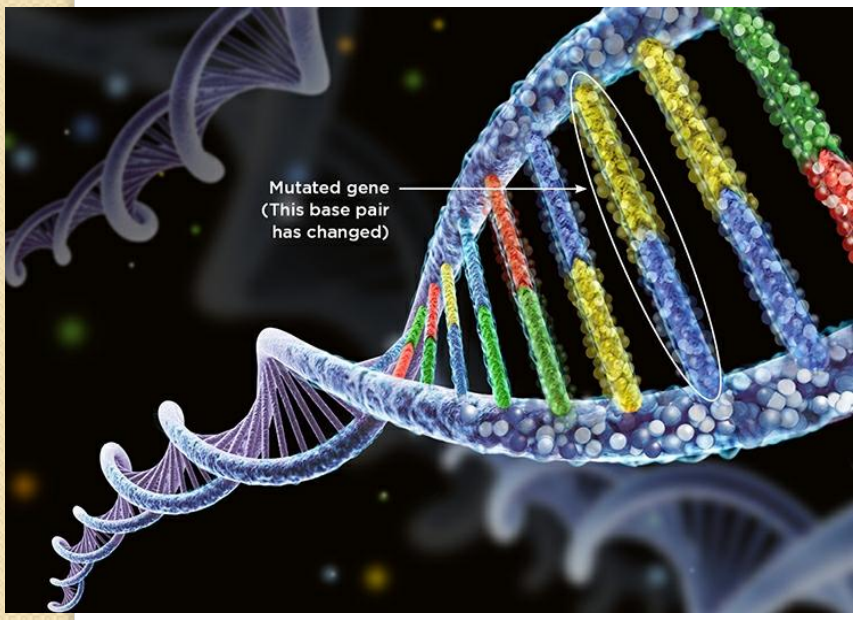
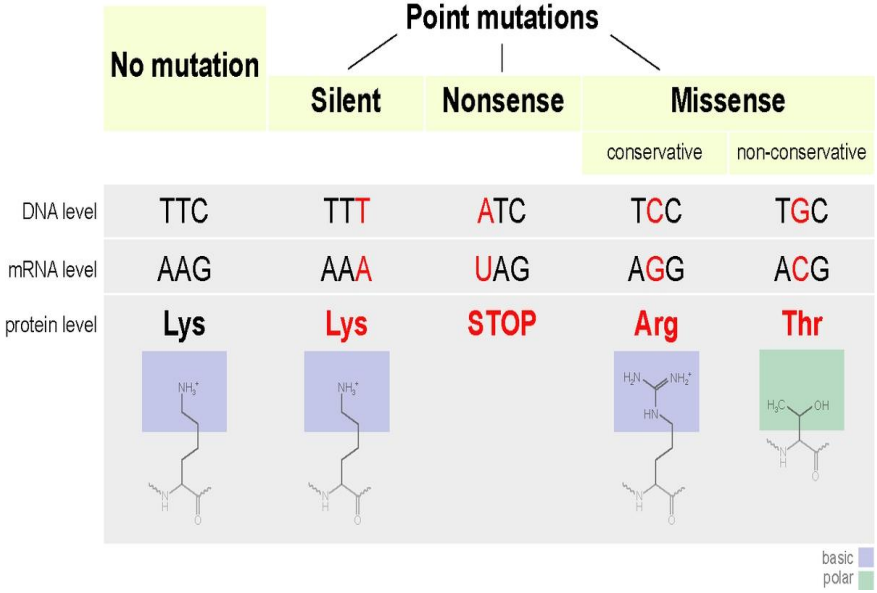


GENE MUTATION

- A **gene mutation** is a permanent alteration in the DNA sequence that makes up a **gene**, such that the sequence differs from what is found in most people. **Mutations** range in size; they can affect anywhere from a single DNA building block (base pair) to a large segment of a chromosome that includes multiple **genes**.

- Basically the gene mutation is the abnormal changes in nitrogenous bases (purines and pyrimidines) of DNA .





Types of Gene Mutations

- Include:
  - –Point Mutations
  - –Substitutions
  - –Insertions
  - –Deletions
  - –Frameshift

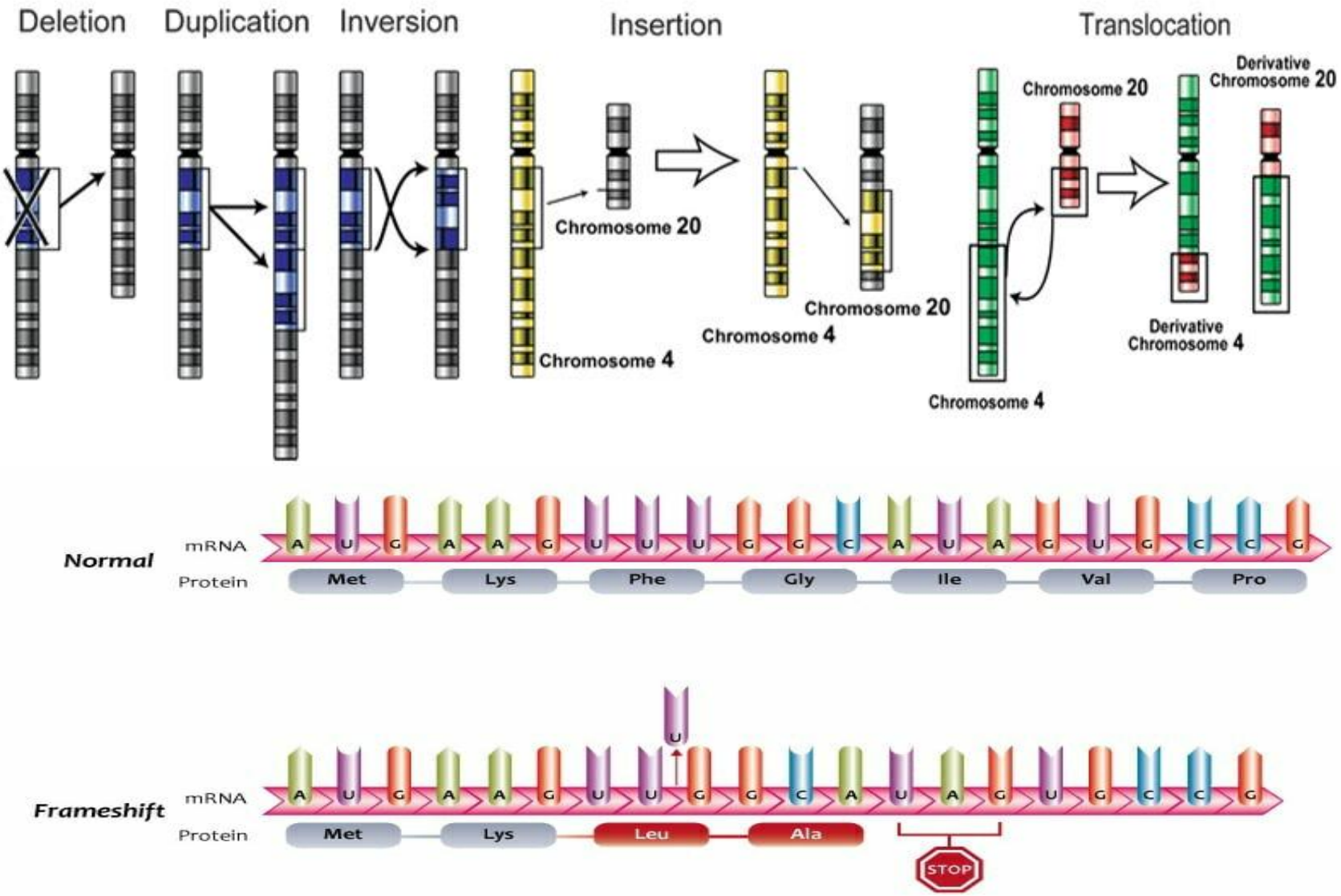
A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation.**

insertion mutation is the addition of one or more nucleotide base pairs into a DNA sequence. **Insertions** can be anywhere in size from one base pair incorrectly inserted into a DNA sequence to a section of one chromosome inserted into another.

A **deletion mutation** occurs when part of a DNA molecule is not copied during DNA replication. This uncopied part can be as small as a single nucleotide or as much as an entire chromosome. The loss of this DNA during replication can lead to a genetic disease.

A **frameshift mutation** is a type of **mutation** involving the insertion or deletion of a nucleotide in which the number of deleted base pairs is not divisible by three. "Divisible by three" is important because the cell reads a gene in groups of three bases.

Types of Mutations



Adapted from Campbell NA (ed). Biology, 2nd ed, 1990.

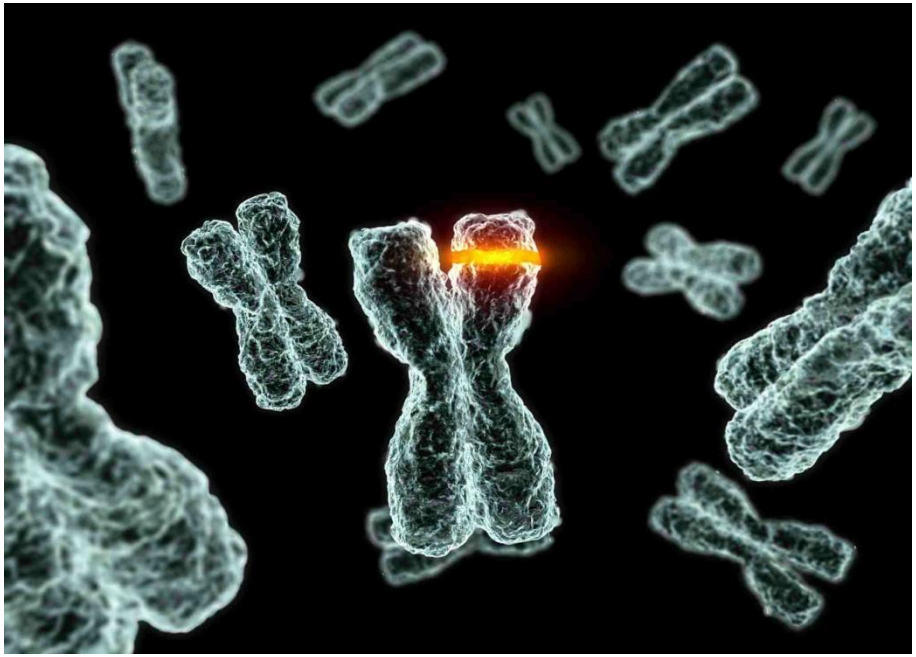
CHROMOSOMAL MUTATION

- A **chromosomal mutation** is any change or error that occurs within the chromosome. Unlike gene **mutations** that involve the alteration of a gene or a segment of DNA in the chromosome, **chromosomal mutations** occur and change the entirety of the chromosome itself**.**

Chromosome Mutations

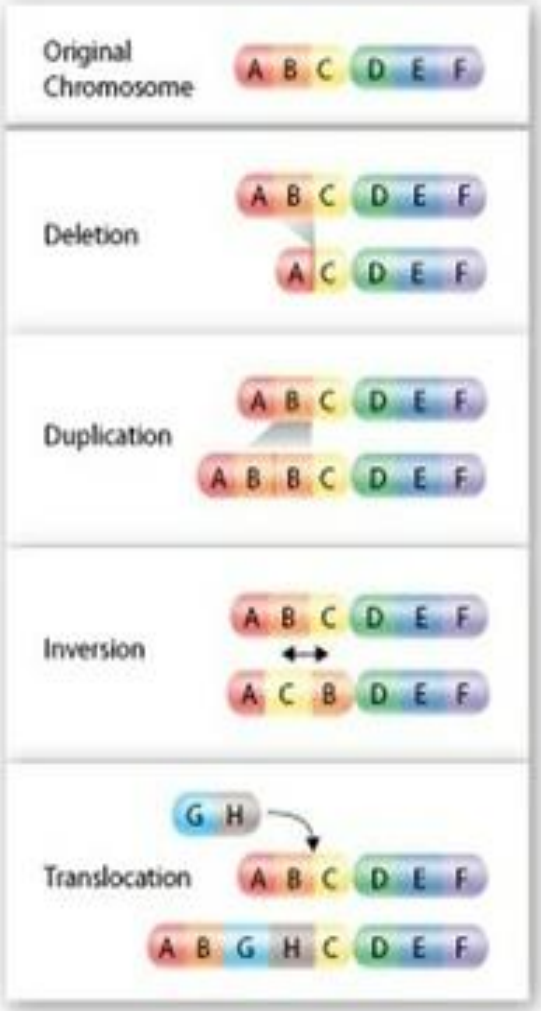
- May Involve:
  - Changing the structure

loss or gain



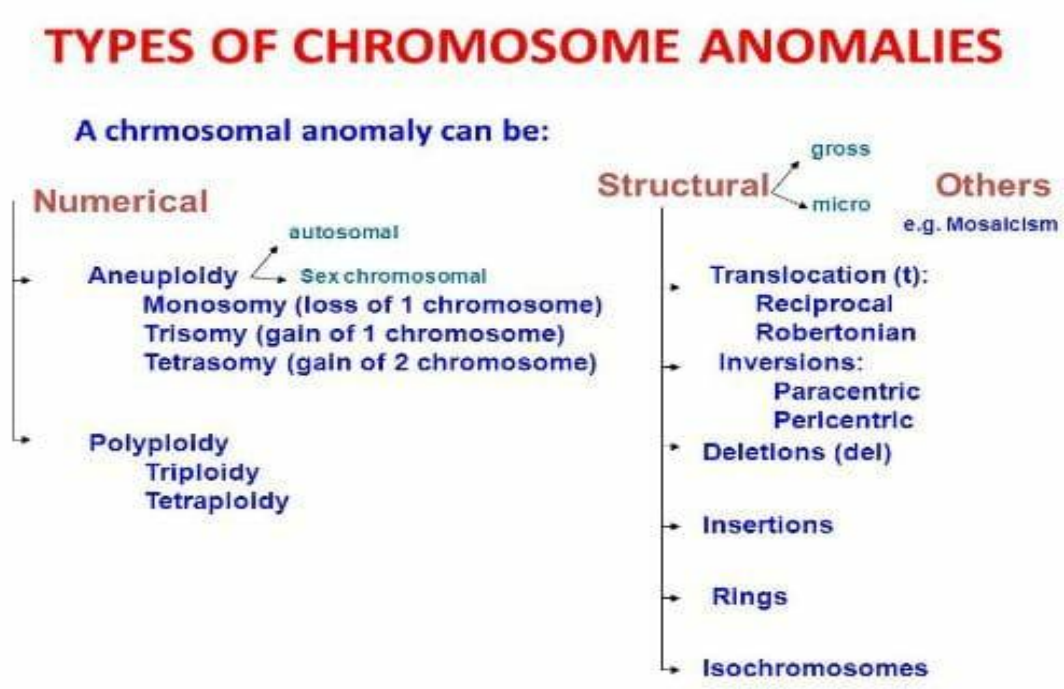
4 Types of Chromosomal Mutations

- Deletion- loss of part of chromosome
- Duplication- extra copy made of part of chromosome
- Inversion- reverses direction of part of chromosome
- Translocation- part of one attaches to another chromosome

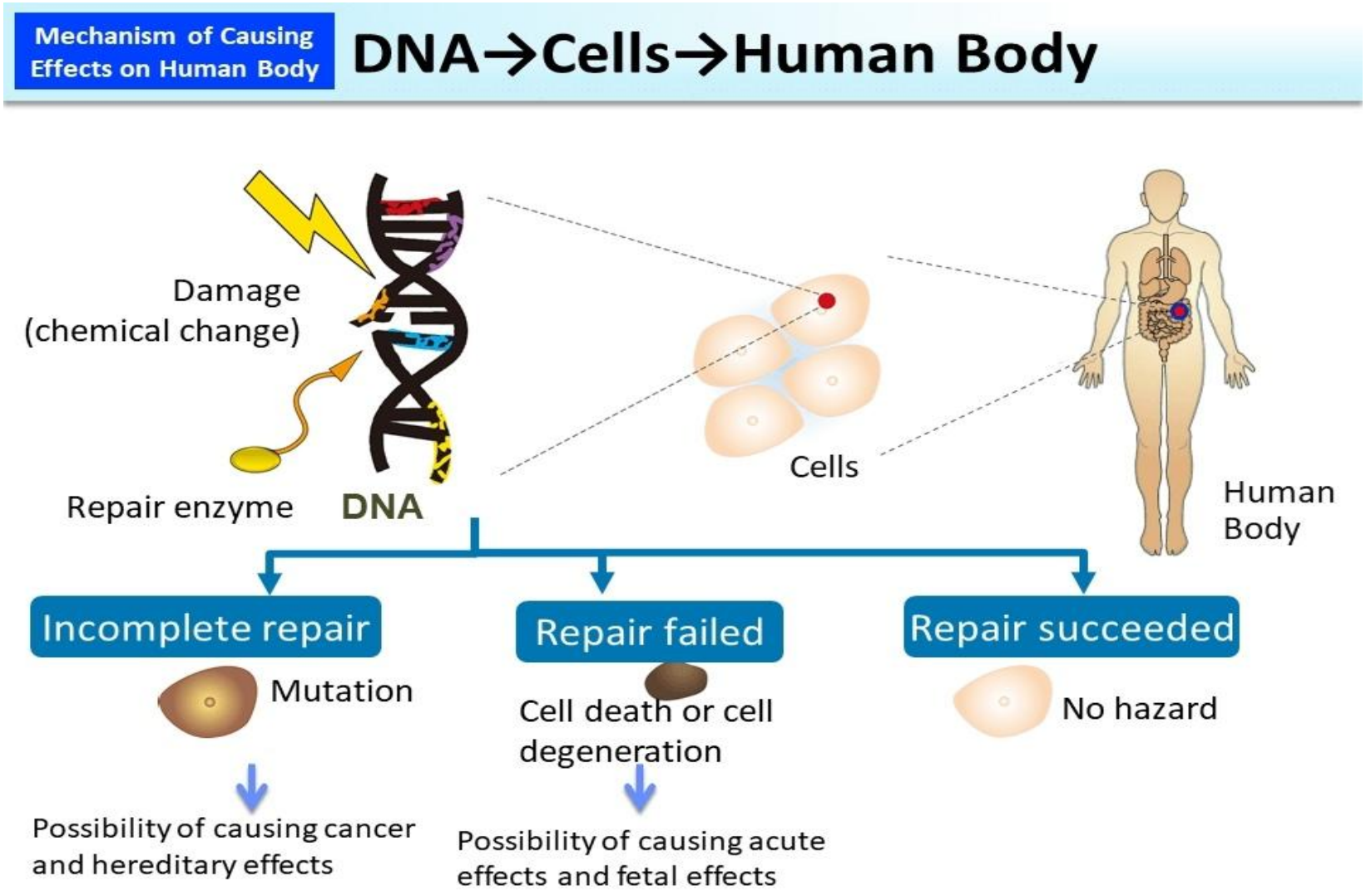


Chromosomal Aberrations

- The somatic (2n) and gametic (n) chromosome numbers of a species ordinarily remain constant.
- This is due to the extremely precise mitotic and meiotic cell division.
- Somatic cells of a diploid species contain two copies of each chromosome, which are called homologous chromosome.
- Their gametes, therefore contain only one copy of each chromosome, that is they contain one chromosome complement or genome.
- Each chromosome of a genome contains a definite numbers and kinds of genes, which are arranged in a definite sequence.

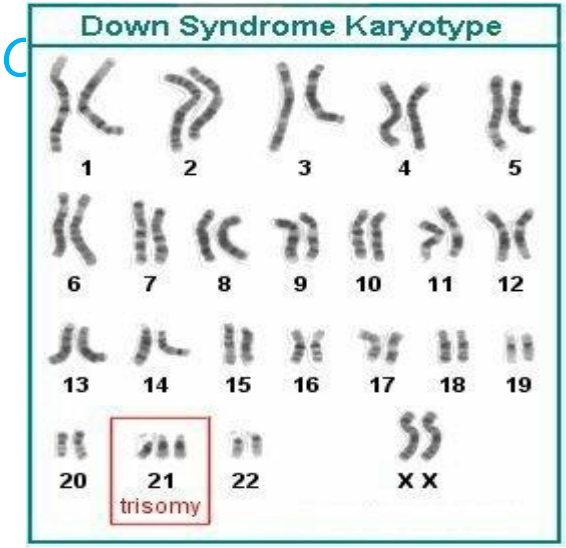


EFFECTS OF MUTATION IN HUMAN BODY



DOWN SYNDROME

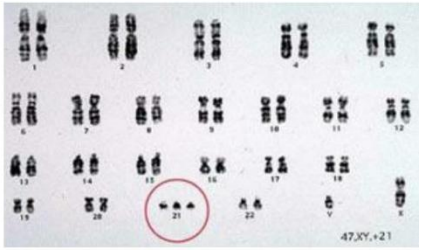
- *CAUSED BY TRISOMY OF 21st CHRO*



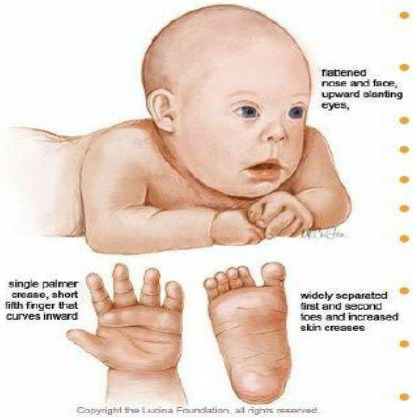
Autosomal Disorders
Chromosomal Mutations

Down Syndrome

- 1 in 1,000 live births.
- Extra Chromosome 21
- Risk increases with mom's age
- Pregnancies of women over age 35 accounts for 20% of Down syndrome births



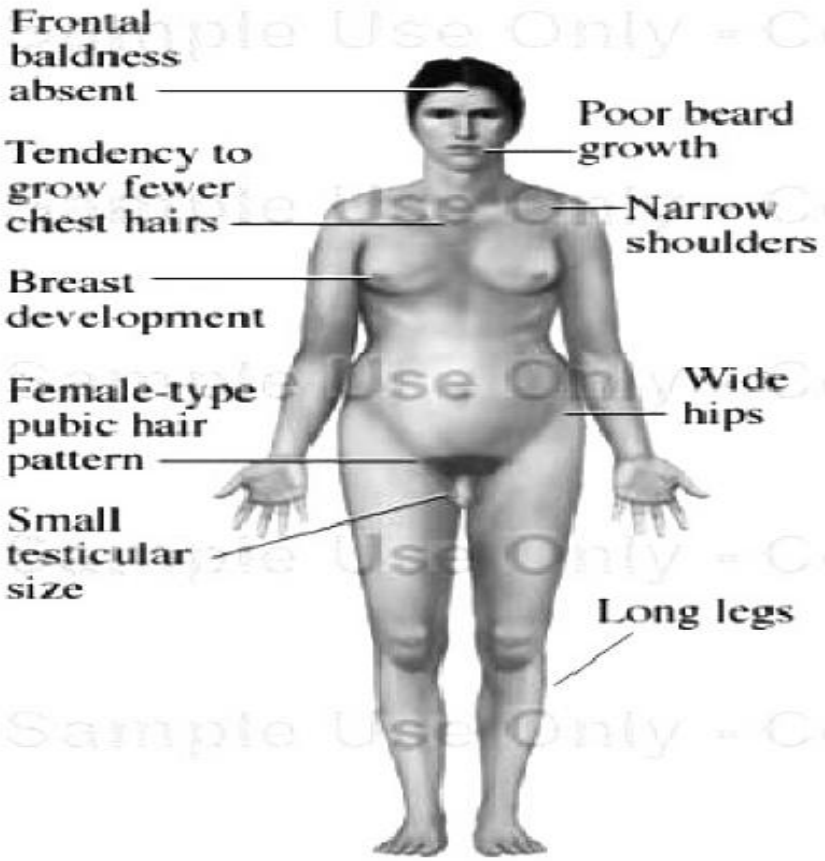
Common physical signs include:

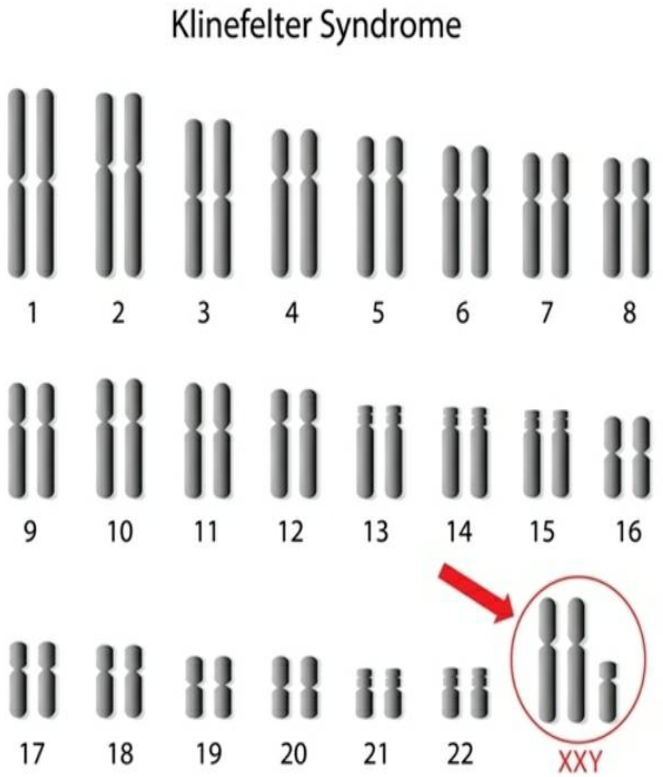


- Decreased muscle tone at birth
- Excess skin at the nape of the neck
- Flattened nose
- Upward slanting eyes
- Small ears
- Small mouth
- Wide, short hands with short fingers
- Separated joints between the bones of the skull
- Single crease in the palm of the hand
- White spots on the colored part of the eye

KLINFELTER`S SYNDROME

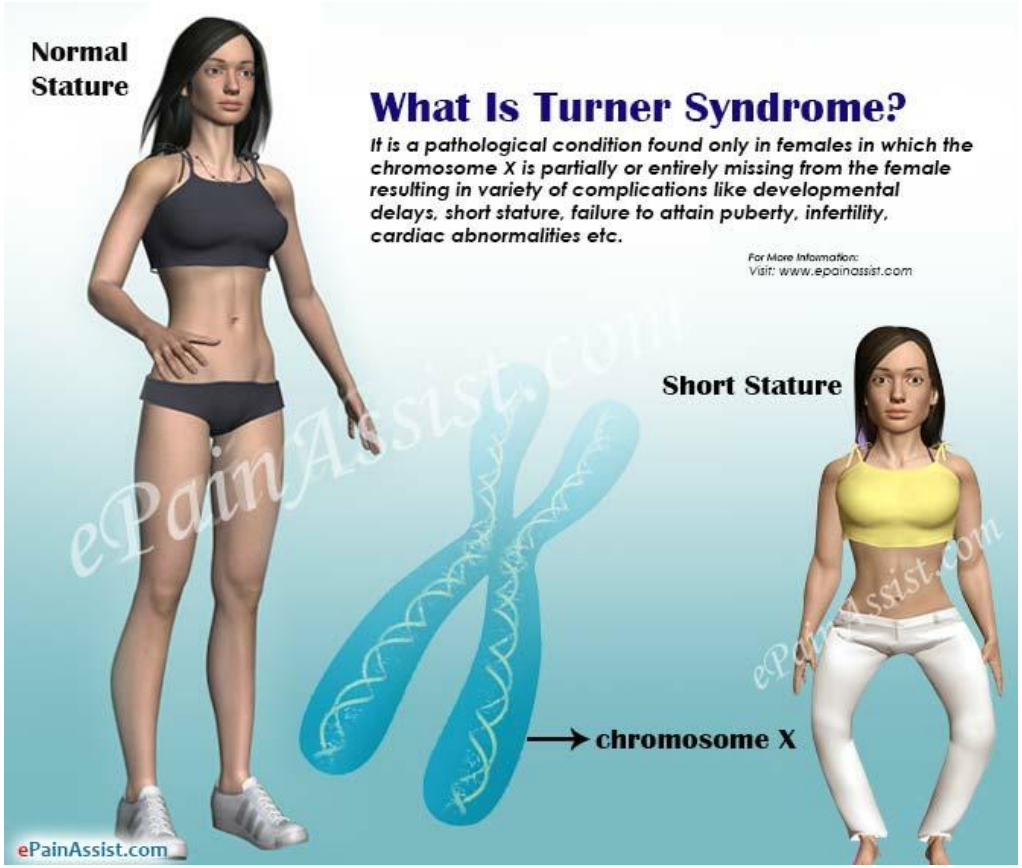
- *CAUSED BY TRISOMY OF SEX CHROMOSOME*

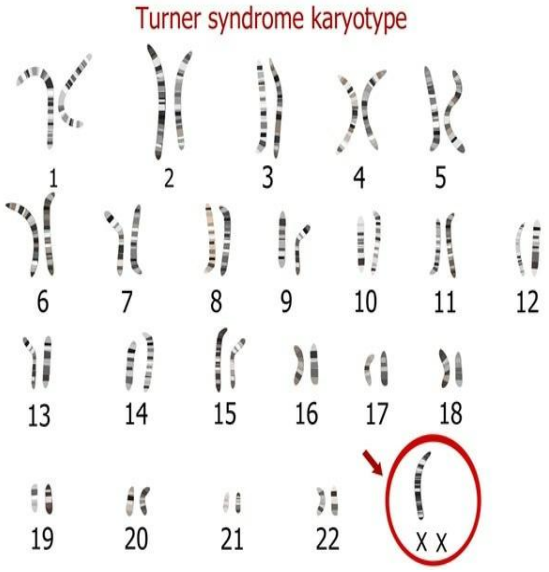




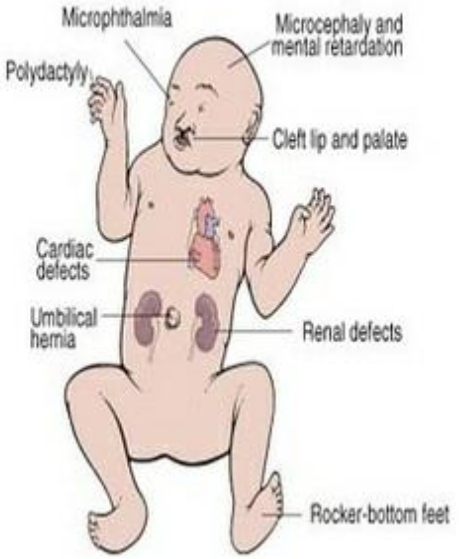
TURNER`S SYNDROME

- ***CAUSED BY MONOSOMY OF SEX CHROMOSOME***





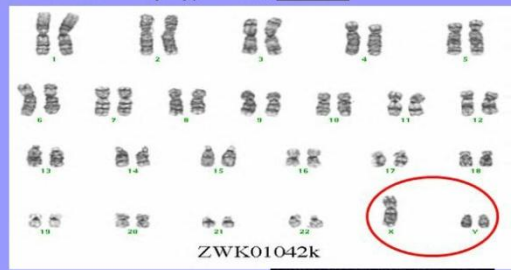
Trisomy 13



- Incidence :1 in 15,0000
- Karyotypes:
  - Trisomy13 type:47xx+13
  - Translocation type:46,xx,+13,der(13;14)(q10;q10)
  - Mosaic type:46,xx/47,xx,+13

Jacob's syndrome

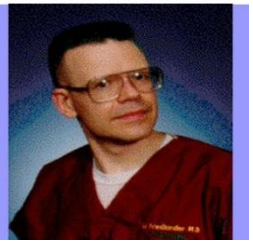
Jacob's syndrome: An extra Y chromosome. Karyotype label, **47, XYY**.



Frequency: 1 in every 1000 male births.

Symptoms (problems): The **extra Y** chromosome may have some affects on the male phenotype:

1. They tend to be somewhat taller than the average male.
2. They tend to have slightly lower mental ability.
3. They tend to have excess acne on their body.
4. Studies of the prison system show that 85% of inmates in prison for violent crimes have this syndrome.



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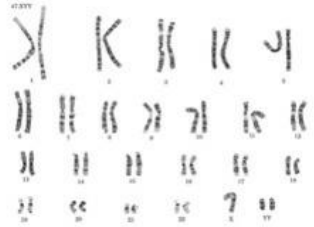
Trisomy 18 (Edward's Syndrome)

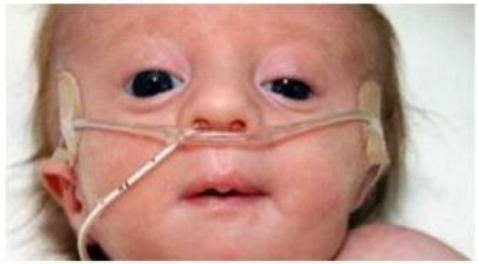
Edward's syndrome also known as trisomy 18 is a genetic disorder. This is a condition where the child is born with three copies of chromosome number 18. The condition leads to death caused due to the malfunctioning of heart. The chances of getting this syndrome are higher in late pregnancy.

Many effects the child might suffer are growth failure, mental retardation, open scull sutures at birth, high arched eyebrows, low set deformed ears, short sternum, ventricular spectrum defect, flextime deformities of fingers, abnormal kidneys, persistent ductus arteriosis, deformity of hips, muscular hypertonus, prominent heel, and dorsal flexing of big toes.



Edwards' syndrome occurs in approximately one in every 5,000 live births and one in every 5,000 stillborn births; it affects girls more often than boys. Women older than their early thirties have a greater risk of conceiving a child with Edwards' syndrome, but it can also occur with younger mothers.

Most children born with Edwards' syndrome appear weak and fragile, and they are often underweight. A child with Edwards' syndrome is likely to have many medical and development needs. Parents should develop good working relationships with their doctor, other specialists, and therapists, and should consult them as needed.



If a woman gives birth to a child with Edwards' syndrome and plans to have another child, a doctor as well as a genetic counselor should be consulted so that prenatal screening and genetic counseling can be conducted.

There is no cure for Edwards' syndrome. Since babies with Edwards' syndrome frequently have major physical abnormalities, doctors and parents face difficult choices regarding treatment. Abnormalities can be treated to a certain degree with surgery, but extreme procedures may not be in the best interests of an infant whose lifespan is measured in days or weeks. Medical therapy often consists of supportive care with the goal of making the infant comfortable, rather than having the child live longer. Children with Edwards' syndrome appear to have increased risk of developing a Wilms' tumor, a cancer of the kidney that primarily affects children.

Classification of mutation

- **Based on the survival of an individual**

1. **Lethal mutation** – when mutation causes death of all individuals undergoing mutation are known as lethal

2. **Sub lethal mutation** - causes death of 90% individuals

3. **Sub vital mutation**– such mutation kills less than 90% individuals

4. **Vital mutation** -when mutation don't affect the survival of an individual are known as vital

5. **Supervital mutation** – This kind of mutation enhances the survival of individual

- **Based on causes of mutation**

1. **Spontaneous mutation-**

Spontaneous mutation occurs naturally without any cause. The rate of spontaneous mutation is very slow eg- Methylation followed by deamination of cytosine.

Rate of spontaneous mutation is higher in eukaryotes than prokaryotes.

Eg. UV light of sunlight causing mutation in bacteria

2. **Induced Mutation-**

Mutations produced due to treatment with either a chemical or physical agent are called induced mutation .

The agents capable of inducing such mutations are known as mutagen.

use of induced mutation for crop improvement program is known as **mutation breeding.**

Eg. X- rays causing mutation in cereals

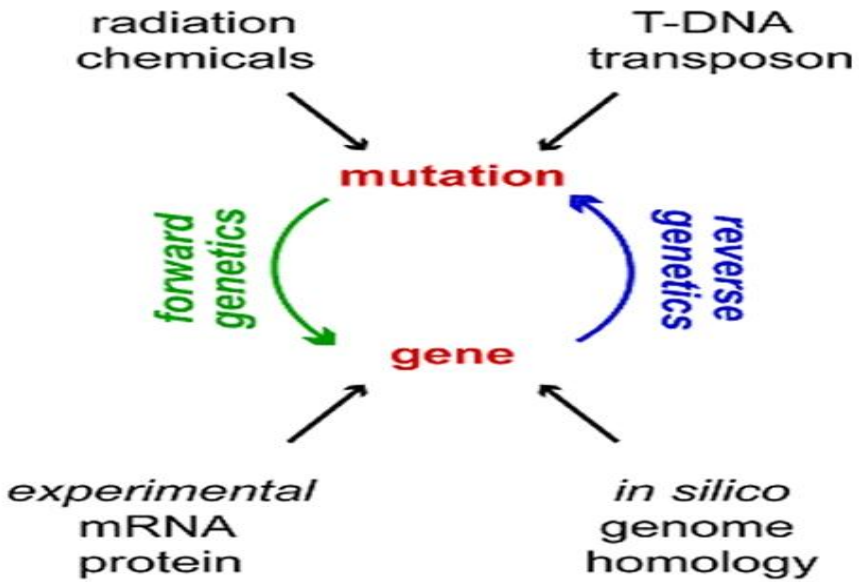
INDUCED MUTATION
VERSUS
SPONTANEOUS MUTATION

INDUCED MUTATION	SPONTANEOUS MUTATION
A mutation that is produced by treatment with a physical or chemical agent that affects the deoxyribonucleic acid molecules of a living organism	A mutation that arises naturally and not as a result of exposure to mutagens
Occur due to mutagens from the environment	Occur due to natural causes
Occur due to the incorporation of base analogs, base mispairing, and base damage	Occur due to errors in DNA replication, spontaneous lesions like depurination and deamination, and transposable genetic elements
Important in reverse genetics	Important in forward genetics

- **Based on direction of mutation**

1. **Forward mutation**- When mutation occurs from the normal/wild type allele to mutant allele are known as forward mutation

2. **Reverse mutation-** When mutation occurs in reverse direction that is from mutant allele to the normal/wild type allele are known as reverse mutation




THANK YOU