

Gene Expression and Mutation

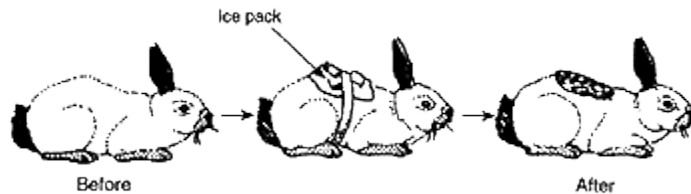
GENE EXPRESSION:

There are hormonal and environmental factors that may cause the expression of some genetic information. Some examples are:

1. The two- colour pattern of some Siamese cats involves one hair-colour gene producing a temperature sensitive enzyme that produces a darker pigments on cooler areas of the body - tips of ears, tail, feet, snout.



2. Environmental differences in identical twins - one could be a different size due to eating habits
3. Temperature affects the coloration of some animals, like Arctic fox and snowshoe hare, which turn white in lower temperatures. The Himalayan rabbit turns dark in response to cold.



4. Hormones can affect balding.
5. The sex of reptile offspring is affected by temperature.
6. The absence of females in a limpet colony will cause some males to turn into females

MUTATIONS

A mutation is a permanent change in the genetic material of an organism.

Most mutations are harmful, some are beneficial, and some have no effect on the organism. They are, however, an important source of species variation.

All mutations can be passed on to other cells in the body but only mutations that happen in the sex cells (**germ cell mutations**) can be passed on to offspring.

Mutations that happen in regular body cells and only affect that organism are called **somatic cell mutations**.

Causes of Mutations:

Many mutations are simple mistakes in copying - **spontaneous mutations**. Every cell undergoes spontaneous mutations.

An **induced mutation** is caused by agents outside the cell called **mutagens**. Mutagens fall into two general categories:

1. **Physical Mutagens** - high energy radiation, like X-rays and gamma rays can cause the worst mutations.

Exposure to UV light (sunbathing, tanning salons), while not as damaging as X-rays or gamma rays can lead to skin cancer.

2. **Chemical Mutagens** - molecules that can enter the cell and react chemically with the DNA. Many of these mutagens are cancer causing (carcinogenic) Many types of drugs, alcohol, bacteria or viruses that may be present in a mother's blood would act as chemical mutagens on the developing fetus. (Remember these are called **teratogens**)

TYPES OF MUTATIONS

1. **Point mutations** - a chemical change that only affects just one or a few nucleotides. They often affect only one gene.

There are two types of point mutations:

- A. **Substitution** - one nucleotide is substituted for another. There are several types:
 - i. **Silent mutation** - has no affect on the final structure of the protein
 - ii. **Mis-sense mutation** - a substitution leads to an altered protein that may be harmful- serious consequences
 - iii. **Nonsense mutation** - the substitution renders the gene unable to produce the protein at all - severe consequences.

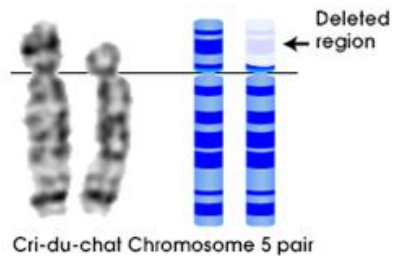
B. **Frameshift mutations** - these are the most severe of the point mutations, because they may cause more than one gene to be useless. Frameshift mutations can be due to:

- i. Insertion
- ii. Deletion

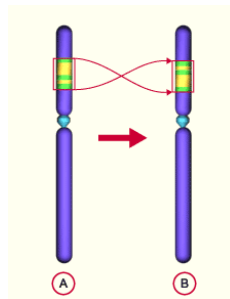
2. Chromosome Mutations

There are five basic types of chromosome mutation. **These would be more serious than point mutations because they would affect so many genes.**

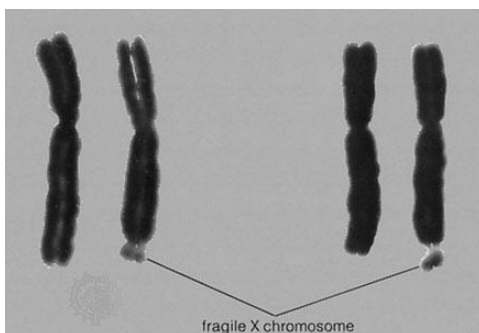
A. **Deletion** - A portion of the chromosome is lost. (Example: cri-du-chat disorder)



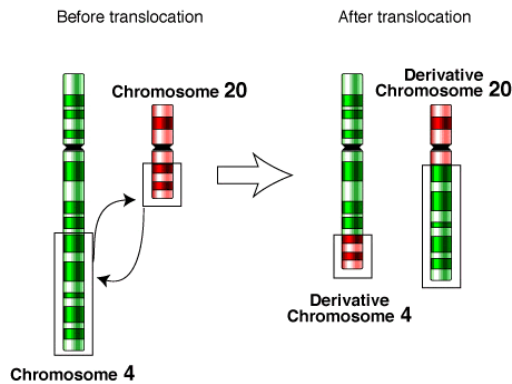
B. **Inversion** - a segment becomes free from the chromosome and is reinserted upside down. (Example: some forms of autism)



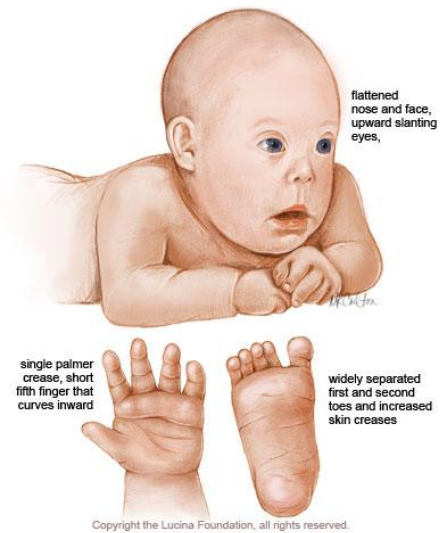
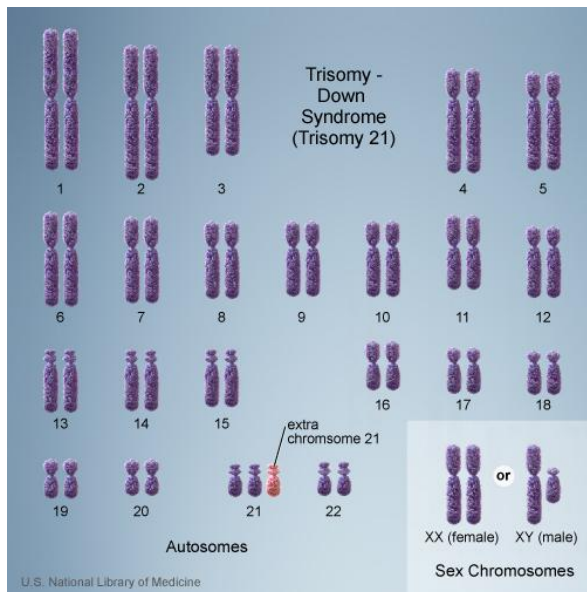
C. **Duplication** - a gene sequence is repeated one or more times within chromosomes (Example: Fragile X syndrome)



- D. **Translocation** - a part of a chromosome changes places with another part of the same chromosome or with a non-homologous chromosome (Example: certain cancers, especially leukemia)



- E. **Nondisjunction** - chromosomes or chromatids do not separate during meiosis. An extra chromosome is called a **trisomy** like with Down's syndrome, Jacobs Syndrome, Triple X, and Klinefelter Syndrome. One fewer is called **monosomy**, like with Turner syndrome. Often a human embryo with too few or too many chromosomes will not survive and will be miscarried (about 30% of all pregnancies)



HUMAN GENETIC DISEASES

Two basic divisions :

1. **Sex-linked** - carried on the Sex chromosomes (sex specific)
2. **Autosomal** - carried on any of the other 22 pairs of chromosomes (not sex specific)

1. **Sex - linked** - these are carried on the X- chromosome so they are recessive in females and dominant in males. This is called **X-linked recessive inheritance** . Diseases and disorders include:

- **Colourblindness**
- **Hemophilia** - blood lacks the ability to clot.
- **Duchenne Muscular Dystrophy** - muscle tissue wastes away and death usually comes by the late teens and early twenties

2. **Autosomal** - we can sub-divide these into other categories:

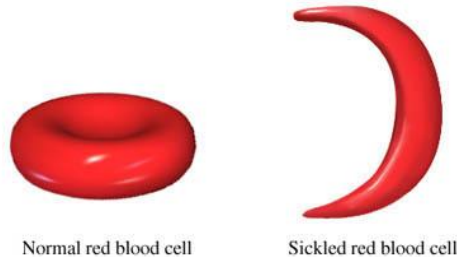
A. **Autosomal Recessive Inheritance**

- **Tay-Sachs Disease** Children born with disease are lacking an important enzyme. They appear normal at birth but by eight months their brains and spinal cord begin to deteriorate. By their first birthday they are blind, mentally handicapped and have little movement. Death usually comes by the fifth birthday. No treatment, but a test is available to see if adults are heterozygous (in other words a carrier)
- **PKU- phenylketonuria** - A missing or defective enzyme responsible for breaking down phenylalanine causes damage to developing nervous systems in young children. Appear normal at birth but will become mentally handicapped within a few months if not diagnosed and put on a special diet. Presently all babies are tested at birth to make sure. Proper diet ensures there will be no mental deficiencies.
- **Albinism** - individuals lack the ability to produce melanin - a skin pigment. At risk for sun damage to skin and eyes.



B. Co-dominant Inheritance

- **Sickle Cell Anemia** - affects the hemoglobin in red blood cells - leads to clots, reduced blood flow and constant pain. The trait has not died out because heterozygous individuals are more likely to survive malaria.



C. Autosomal Dominant Inheritance

An autosomal dominant allele will appear in every generation, if the affected individual lives long enough to reproduce. They are rare. Some are the result of rare, chance mutations.

- **Progeria** - a rare disorder that causes the individual to age very rapidly. Affects about one in 8 million newborns.



- **Huntington Disease** - a lethal disease that does begin until about mid-thirties, and is usually fatal within 15 years. The brain deteriorates over this time. There is no cure or treatment and many people die before knowing if they have passed on the disease to their children.

D. Incomplete Dominance

- **Familial Hypercholesterolemia (FH)** Excess cholesterol can lead to heart attacks and strokes. Individuals that are heterozygous (1 in 500) have double the normal cholesterol level, and may have heart attacks by the age of 35. Homozygous individuals (1 in 1 million) have six times the normal level, and may have heart attacks by the age of 2!