Changes in Chromosome Structure

Changes in the physical structure of chromosomes can occur:

1. Spontaneously
2. As a result of irradiation
3. After exposure to certain chemicals

Structural Changes in Chromosomes

- Deletion
- Duplication
- Inversion
- Translocation

Deletion

- A deletion removes a chromosomal segment
- Loss of a piece of chromosome #5
  - Called Cri-du-chat
  - Affects the larynx making cat sounds

Duplication

- A duplication repeats a segment
- Duplication in the X chromosome
  - Called Fragile X syndrome

Inversion

- An inversion reverses a segment within a chromosome
- Some forms of autism
Translocation

- A translation moves a segment from one chromosome to another, non-homologous one
- Down Syndrome: chromosomes 14 and 21
- Leukemia: chromosomes 22 and 9

Nondisjunction

- Sometimes, chromosomes fail to separate from each other during meiosis. This produces gametes (eggs / sperm) which have either too many or too few chromosomes
- If a gamete which does not have the correct number of chromosomes is involved in fertilization, a zygote will be produced which has either too many or too few chromosomes
- This creates an embryo whose cells contain either more or less than 46 chromosomes. These embryos are usually aborted by the mother, but some survive and have genetic disorders

Nondisjunction

1. Meiosis I starts normally. Telophase II in middle of cell.
2. Then one set of homologs does not separate (a nondisjunction).
3. Meiosis II occurs normally.
4. All gametes have an abnormal number of chromosomes—either one too many or one too few.

Types of Nondisjunction

- Trisomy - When an individual inherits an extra chromosome.
- Monosomy - When an individual inherits one less chromosome.

Human Genetic Analysis

- Geneticists are able to analyze the patterns of human inheritance using two methods
  - Examination of karyotypes
  - Construction of pedigrees

Human Karyotype

- Within our body cells, humans normally possess 46 chromosomes.
  - 44 of these are autosomes (body chromosomes)
  - 2 are sex chromosomes.
- A karyotype is a photograph of the chromosomes which are located in the nucleus of a somatic cell.
- Once a photograph has been taken of the chromosomes in a cell's nucleus, they are cut out and arranged in pairs according to their size, shape, and appearance.
- By observing the karyotype, disorders may become apparent.
**Turner Syndrome**

- An individual inherits only a single X chromosome, as well the Y chromosome is missing.
- This results in a female with the genotype XO
  - O represents a missing chromosome

**Turner Syndrome Clinical features**

- Short stature
- Loe haemangi
- Small stature
- Narrow chest
- Small hands
- Brown spots (nevus)
- No menstruation

**Klinefelter Syndrome**

- A male who has an extra X chromosome.
- These individuals have the genotype XXY instead of XY

**Klinefelter Syndrome**

- Lower IQ than sibs
- Tall stature
- Poor muscle tone
- Reduced secondary sexual characteristics
- Gynaecomastia (male breasts)
- Small testes/infertility

**Down Syndrome (Trisomy 21)**

- This occurs when an individual receives three copies of chromosome 21 instead of the normal two.
Jacobs Syndrome

- Males with an extra Y chromosome, having the genotype XYY

Symptoms of Jacob’s syndrome:

- attention difficulties.
- delayed motor skill development, such as with writing.
- delayed or difficult speech.
- emotional or behavioral issues.
- hand trembling or involuntary muscle movements.
- hypotonia (weak muscle tone)