CHROMOSOMES
CHROMOSOME

• The chromosome comes from Greek
  - Chroma = color
  - Soma = body (the colored body)

• Chromosomes are act as factors which distinguished one species from another.

• Chromosomes are formed of DNA which is embedded in protein material

• Chromosomes are facilitate the transmission of all genetic information from one generation to another.

• Cytogenetics: the science which study chromosome and cell division.
CHROMOSOME MORPHOLOGY

• Submicroscopic
  - Chromosome made up of supercoils of DNA.

• Microscopic
  - Chromosome consists of two chromatids or sister chromatids
  - Join together by centromere
  - Centromere divided the chromosome into short (p=petite) and long (q=grande).
  - The tip of each chromosome arms in named telomere
These sister chromatids can be seen to be joined at a primary constriction known as the centromere.

Centromeres are responsible for the movement of chromosomes at cell division.

The tip of each chromosome arm is known as the telomere.

Telomeres play a crucial role in sealing the ends of chromosomes and maintaining their structural integrity.
CENTROMERE
TELOMERS

The end of each chromosome to ensure the genomic stability

Chromosome without telomeres recombine with other chromatin, breakage, fusion, and can loss.

Most cells being unable to undergo more than 50-60 divisions.

Increased telomerase activity has been implicated as a cause of abnormally prolonged cell survival.
CHROMOSOME MORPHOLOGY

DNA double helix → Nucleosomes → Chromatin fiber → Extended section of chromosome → Loops of chromatin fiber → Metaphase chromosome

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CHROMOSOME MORPHOLOGY
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Image of a chromosome with various labeled parts:
- X Chromosome
- heterochromatin
- euchromatin
- centromere
- euchromatin
- heterochromatin
- telomere
- telomere
- SRY
- Y Chromosome
The classification of chromosome is made according to three important parameters:

- Length of chromosome
- Position of centromere
  - Metacentric
  - Submetacentric
  - Acrocentric
CHROMOSOMAL CLASSIFICATION
CHROMOSOMAL CLASSIFICATION

• Presence or absence of satellite

• According to the above parameters: chromosomes are divided into 7 groups

  • A= 1-3   B= 4-5   C=6-12+X   D=13-15
  • E=16-18   F=19-20   G=21-22+Y
CHROMOSOMAL CLASSIFICATION
CHROMOSOME

- Made of DNA and protein
- 23 pair
- Autosomes
  - Pair 1-22
- Sex chromosomes
  - X and Y
CHROMOSOME

• In humans the normal cell nucleus contains 46 chromosomes, made up of 22 pairs of autosomes and a single pair of sex chromosomes - XX in the female and XY in the male.

• Each human cell (somatic cells) contains 46 chromosomes (diploid number of chromosomes) except mature gametes (sperms and ova) each cell contains 23 chromosomes (haploid number of chromosomes) i.e. 22 autosomes + one sex chromosome)
HOMOLOGOUS PAIRS OF CHROMOSOMES

- Members of a pair of chromosome are known as **homologues** (carrying the same gene).

- Each chromosome has a certain gene on it.

- A **homologous pair** is a pair with the same gene one from mother, **one from** father.
**BANDING (STAINING) TECHNIQUES**

- The development of chromosome banding (staining) enable very precise recognition of individual chromosomes and the detection of chromosome abnormalities.

- The technique (staining) also revealed that chromatin, exists in two main forms.
  - **Euchromatin** stains lightly and consists of genes which are actively expressed.
  - **Heterochromatin** stains darkly and is made up largely of inactive unexpressed repetitive DNA.
PREPARATION OF KARYOTYPE

1. 5 ml venous blood
2. Add phytohemagglutinin and culture medium
3. Culture at 37°C for 3 days
4. Digest with trypsin and stain with Giemsa
5. Spread cells onto slide by cropping
6. Add colchicine and hypotonic saline
7. Cells fixed
8. Analyse "metaphase spread"
G BANDING

• Most common use
• In the **Metaphase**
  • chromosome treated with **trypsin**
  • Then stained with **Giemsa**

R band
• Stain light
• GC rich
• have the highest gene density

G bands
• Stain dark
• AT rich
• have relatively **fewer** genes than R bands
Q BANDING

- It is similar to that obtained with Giemsa
- requires examination with ultraviolet fluorescent microscope
R-bandung

The chromosomes are **heat-denatured** then staining with **Giemsa**.
C-banding

- Chromosomes are pretreated with acid
- followed by alkali
- then Giemsa

Human female C-bands
CHROMOSOME
THE SEX CHROMOSOMES

In humans both the male and the female have two sex chromosomes

- XX in the female and
- XY in the male.

The Y chromosome is much smaller than the X chromosome.

Genes on the Y chromosome include testis determining factor known as SRY gene.

Other genes on the Y chromosome are known to be important in maintaining spermatogenesis.
THE SEX CHROMOSOMES

In the female each ovum carries an X chromosome.

As there is a roughly equal chance of either an X-bearing sperm or a Y-bearing sperm fertilizing an ovum, the numbers of male and female conceptions are approximately equal.

In fact slightly more male babies are born than females, although during childhood and adult life the sex ratio evens out at 1:1.
PUNNETT'S SQUARE SHOWING SEX CHROMOSOME COMBINATIONS FOR MALE AND FEMALE GAMETES