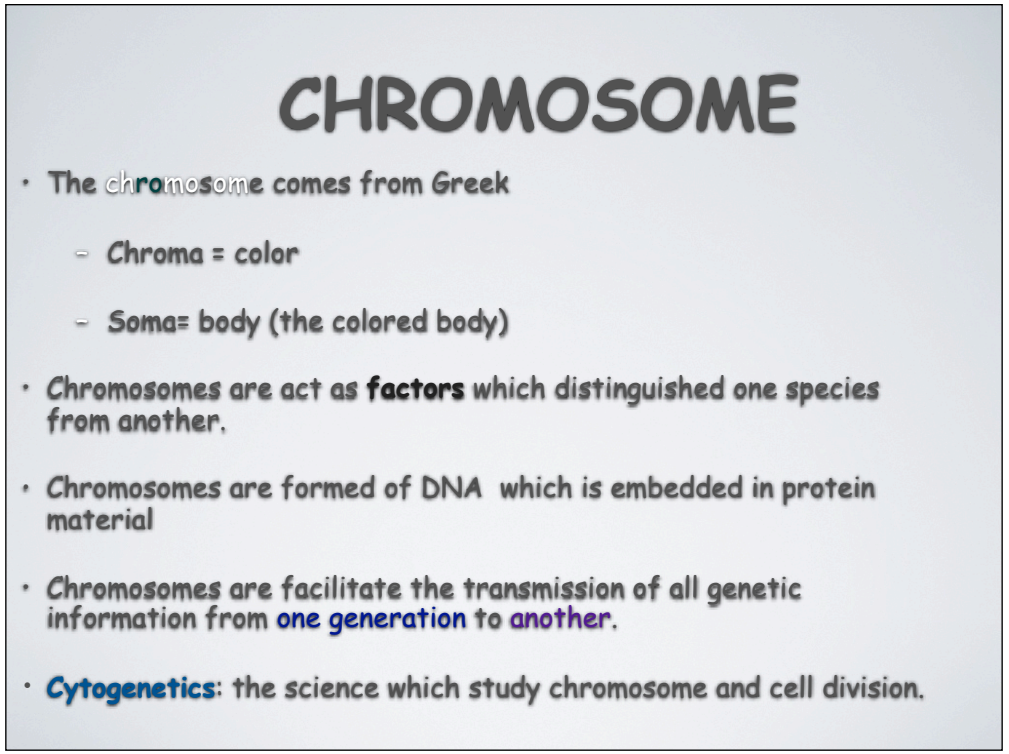
CHROMOSOMES



Chromosomes are the factors that distinguish one species from another and that enable transmission of genetic information from one generation to the next. Their behavior at somatic cell division in mitosis provides a means of ensuring that each daughter cell retains its own complete genetic complement. Similarly, their behavior during gamete formation in meiosis enables each mature ovum and sperm to contain a unique single set of parental genes. Chromosomes are quite literally the vehicles that facilitate reproduction and the maintenance of a species.

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

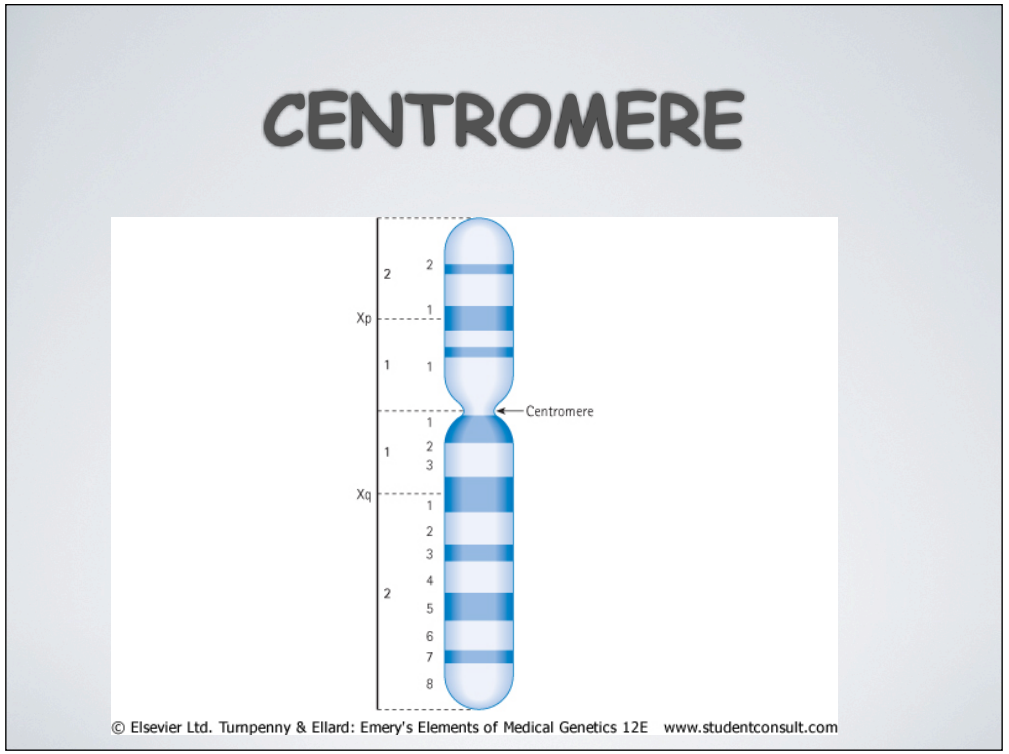
CHROMOSOME

These sister chromatids can be seen to be joined at a primary constriction known as the centromere.

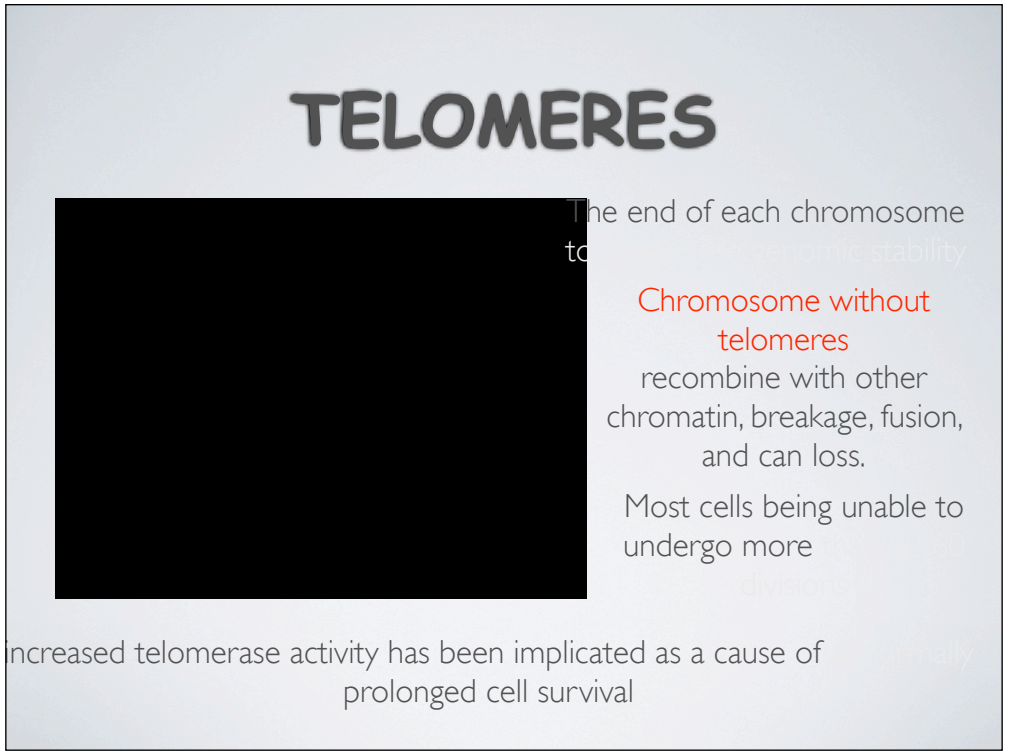
Centromeres are responsible for the movement of chromosomes at cell division.

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.

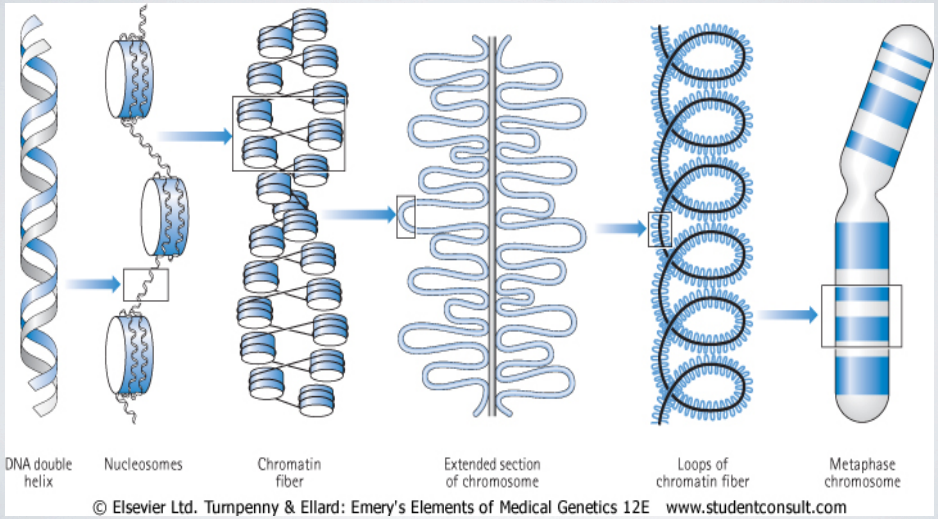


each chromosome can be seen to consist of two identical strands known as *chromatids*, or *sister chromatids*, which are the result of DNA replication having taken place during the S (synthesis) phase of the cell cycle (p. 43). These sister chromatids can be seen to be joined at a primary constriction known as the *centromere*Centromeres consist of several hundred kilobases of repetitive DNA and are responsible for the movement of chromosomes at cell division. Each centromere divides the chromosome into short and long arms, designated p (= petite) and q ('g' = grande), respectively.

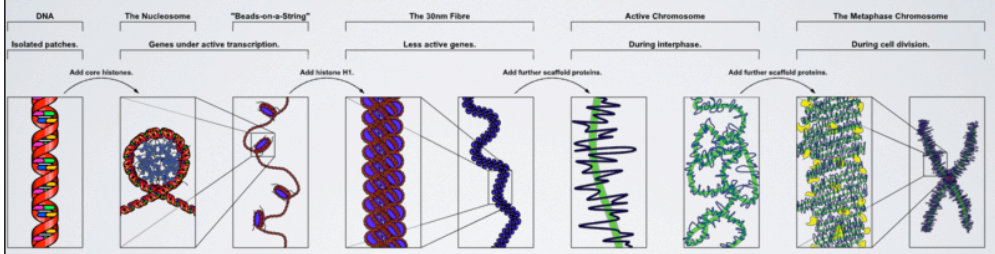


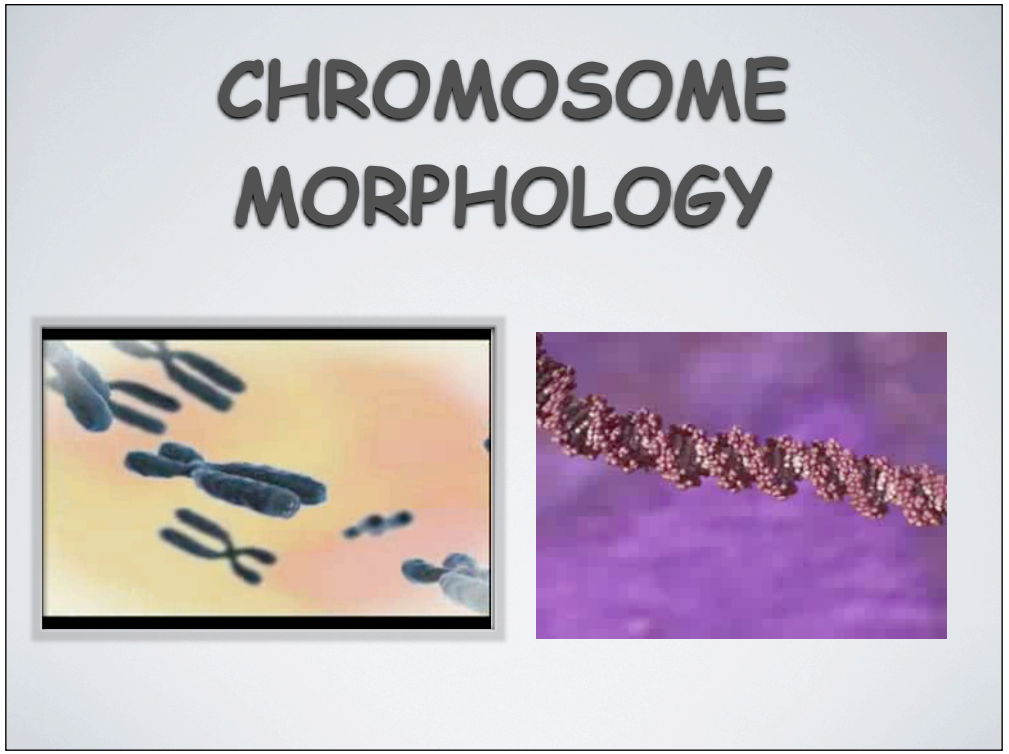
The tip of each chromosome arm is known as the *telomere*. Telomeres play a crucial role in sealing the ends of chromosomes and maintaining their structural integrity. Telomeres have been highly conserved throughout evolution and in humans they consist of many tandem repeats of a TTAGGG sequence. During DNA replication an enzyme known as *telomerase* replaces the 5' end of the long strand (p. 18), which would otherwise become progressively shorter until a critical length is reached when the cell can no longer divide and thus becomes senescent. This is in fact part of the normal cellular aging process, with most cells being unable to undergo more than 50-60 divisions. However, in some tumors increased telomerase activity has been implicated as a cause of abnormally prolonged cell survival

CHROMOSOME MORPHOLOGY



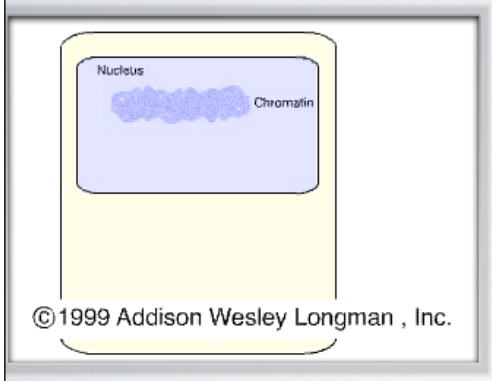
CHROMOSOME MORPHOLOGY

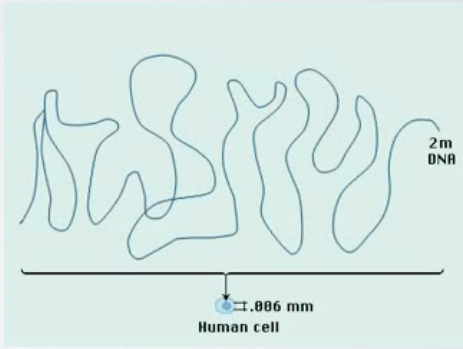




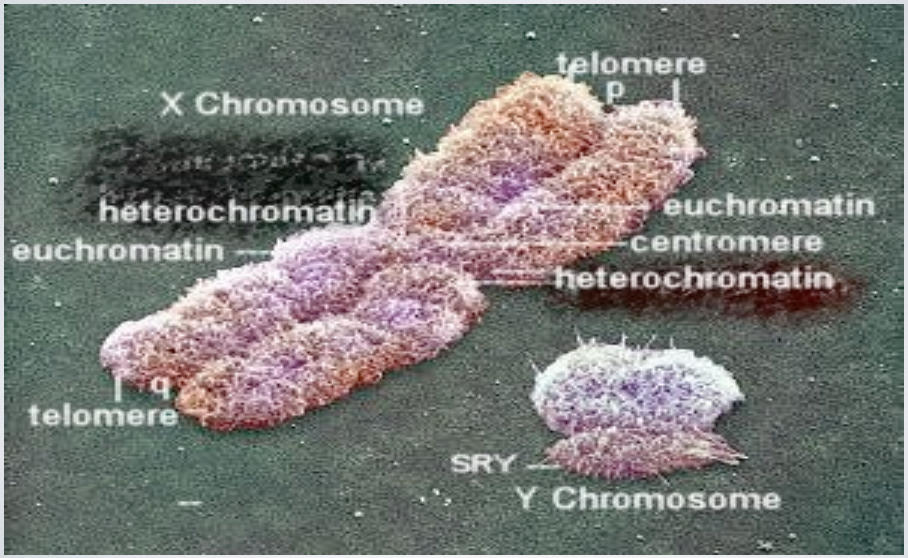
chromosome is mad of 2 sister chromatin histones and non histones protein
chromatin repeating unit Nucleosome 146 bp of DNA and 8 histones this give the shape and strutter

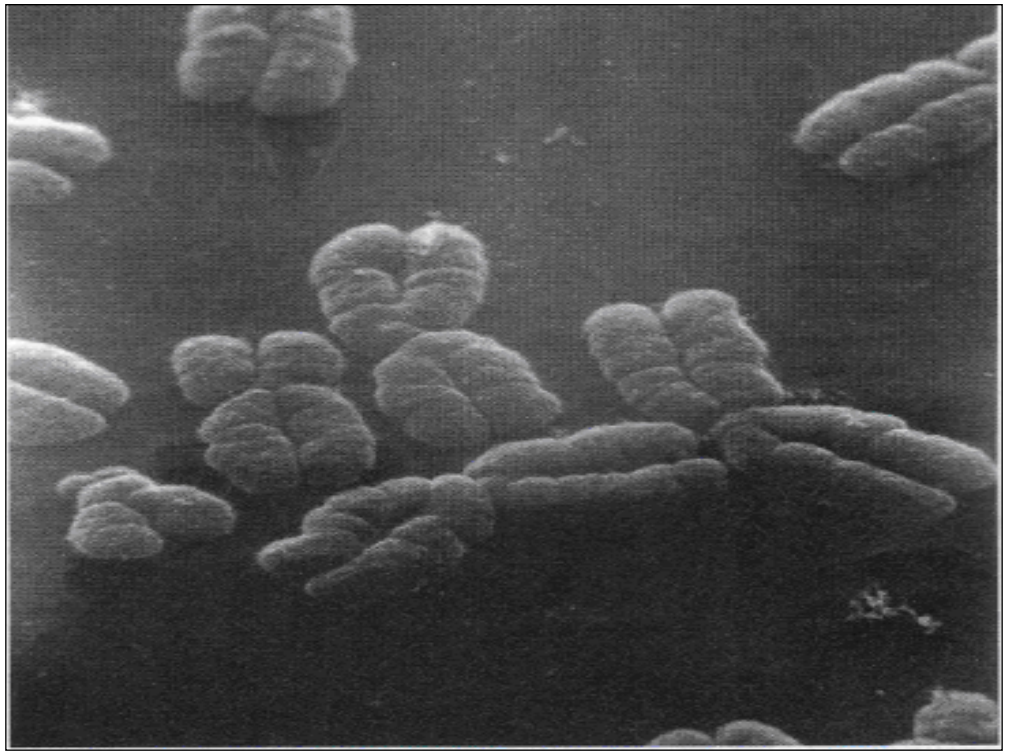
CHROMOSOME MORPHOLOGY

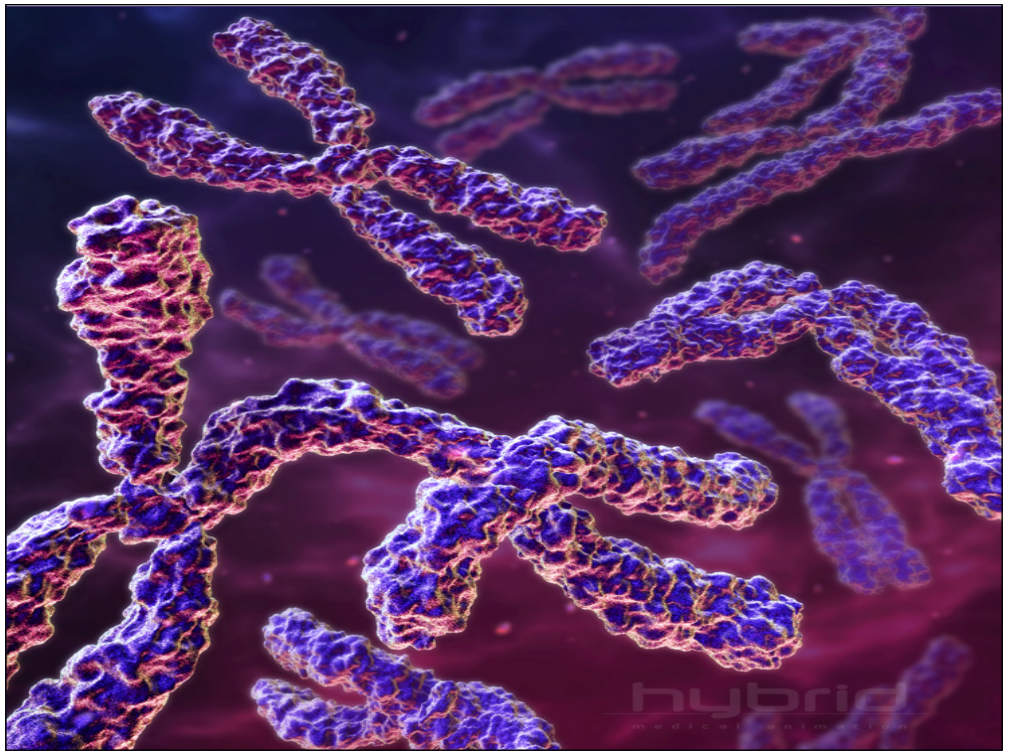


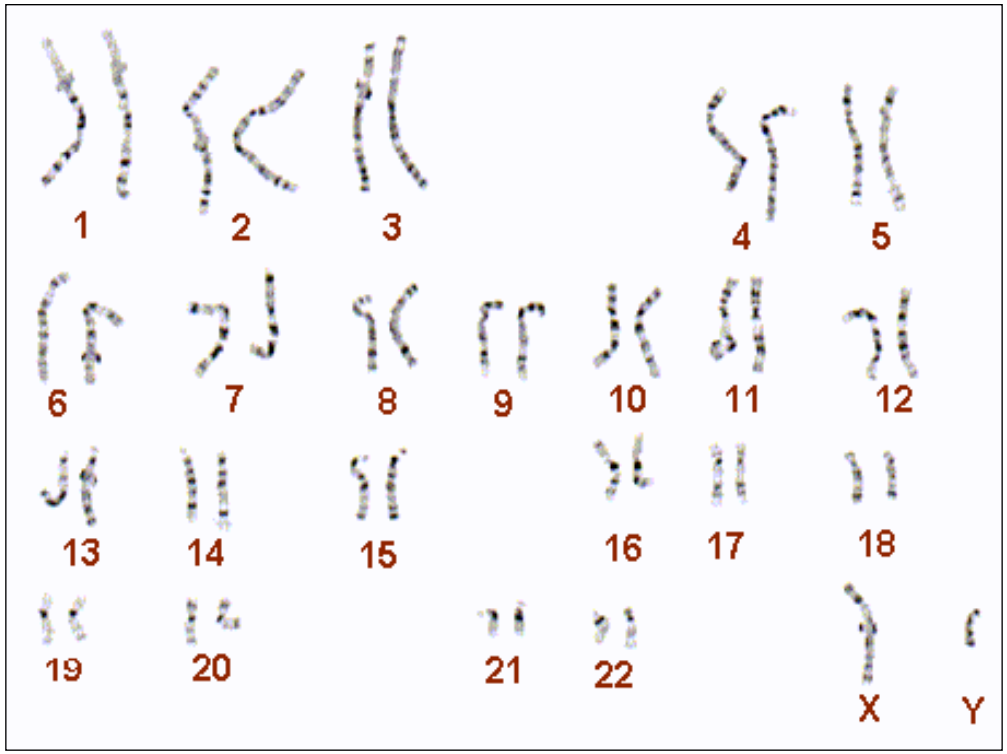


CHROMOSOME MORPHOLOGY





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CHROMOSOMAL CLASSIFICATION

The classification of chromosome is made according to three important parameters

Length of chromosome

Position of centromere

- Metacentric
- Submetacentric
- Acrocentric

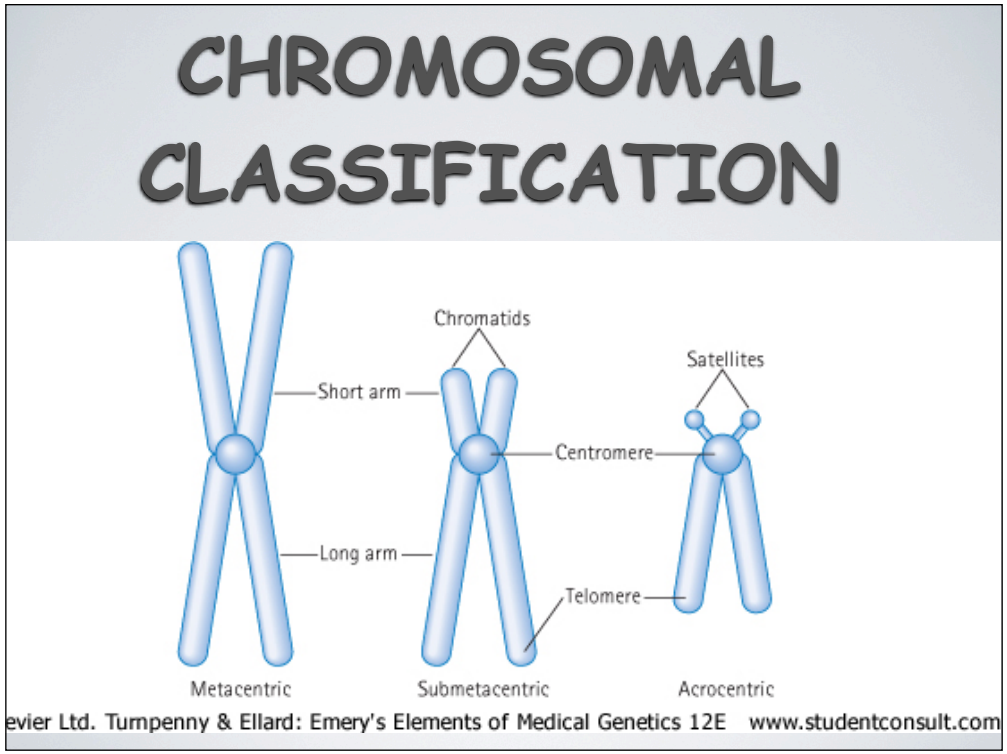
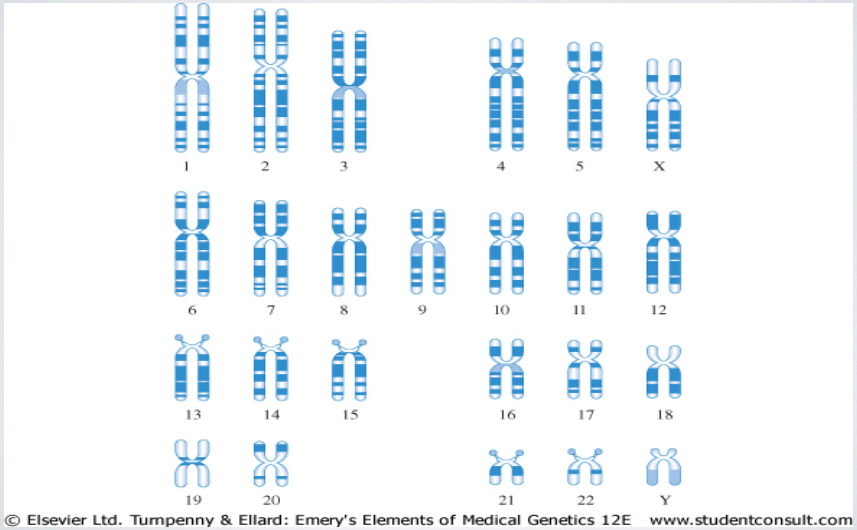


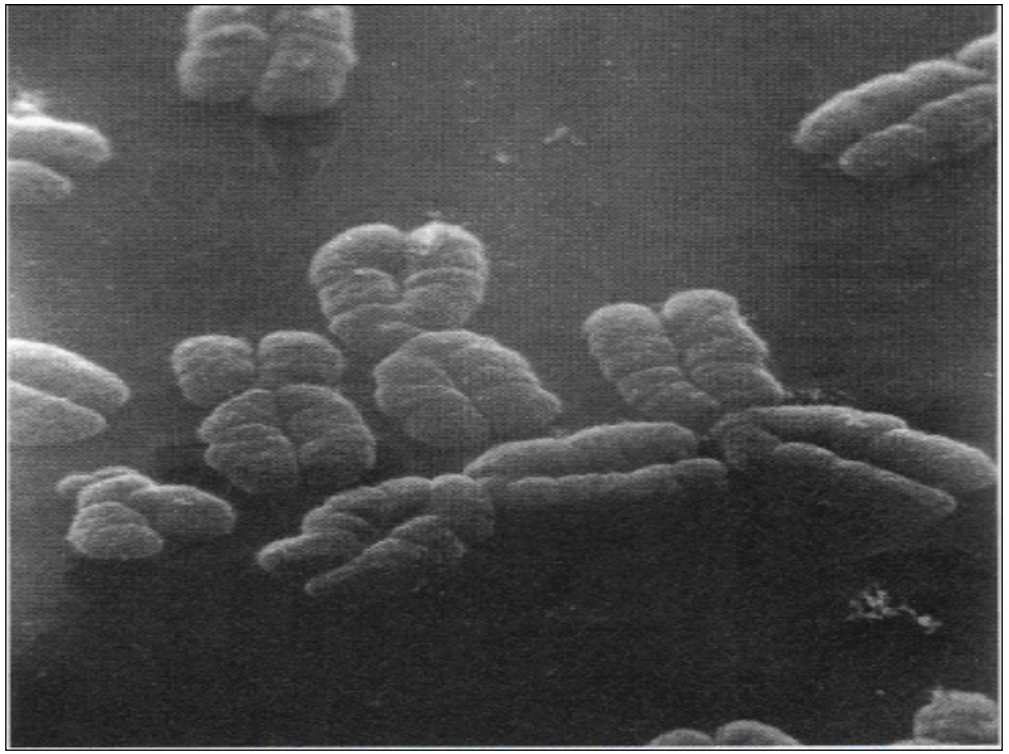
Figure 3.2 Morphologically chromosomes are divided into metacentric, submetacentric and acrocentric depending on the position of the centromere.

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

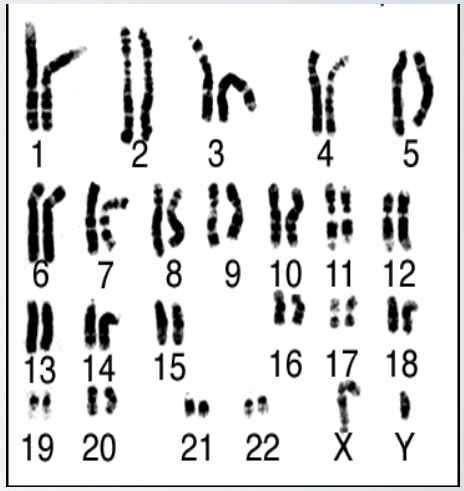
CHROMOSOMAL CLASSIFICATION

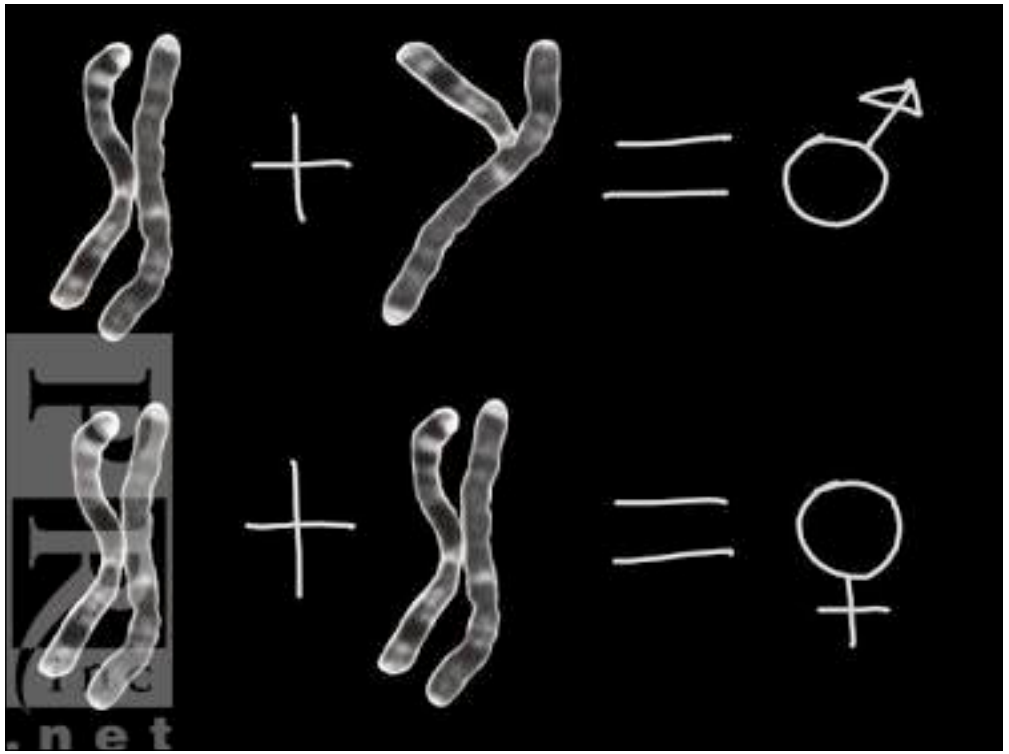




CHROMOSOME

- 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)

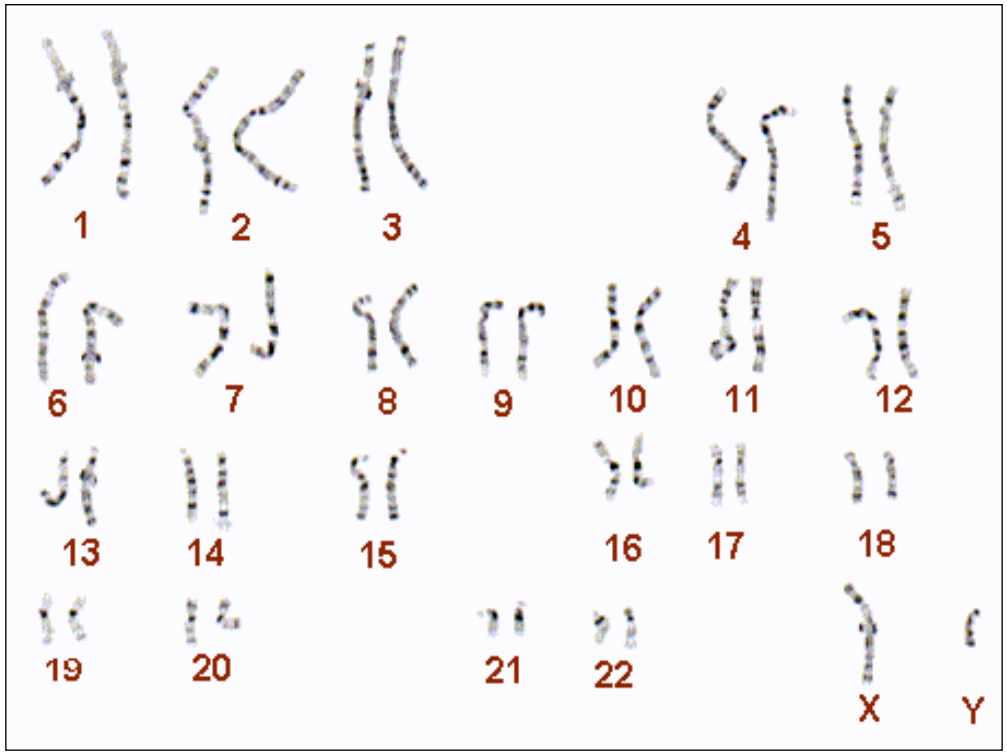
HOMOLOGOUS PAIRS OF CHROMOSOMES

- 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.

Several different staining methods can be utilized to identify individual chromosomes.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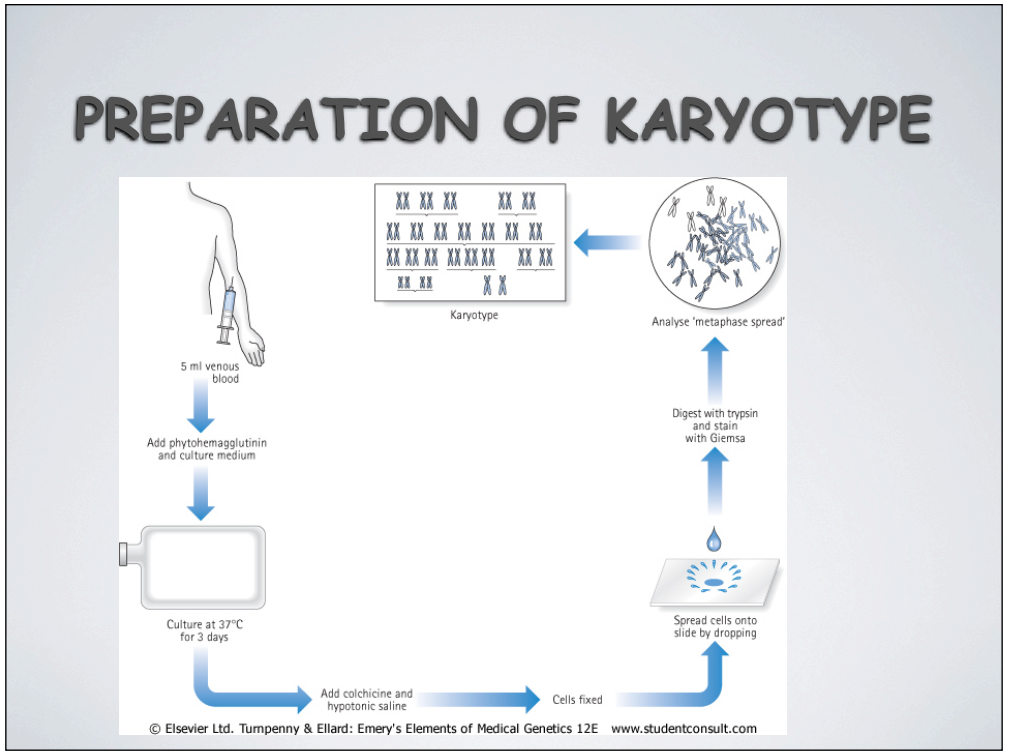
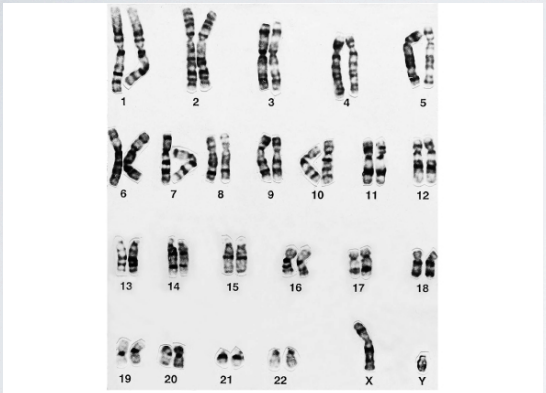


Figure 3.4 Preparation of a karyotype

G BANDING



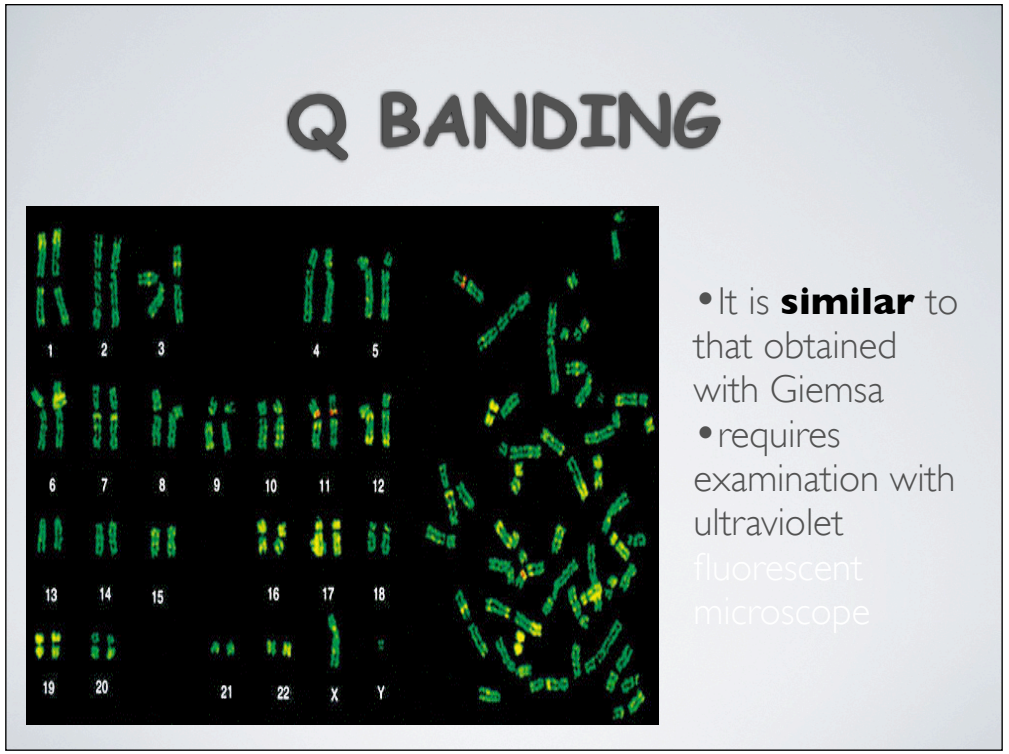
- Most common use
- In the **Metaphase**
  - chromosome treated with **trypsin**
  - Then stained with **Giemsa**

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

G bands
- Stain dark
- AT rich
- have relatively fewer genes than R bands

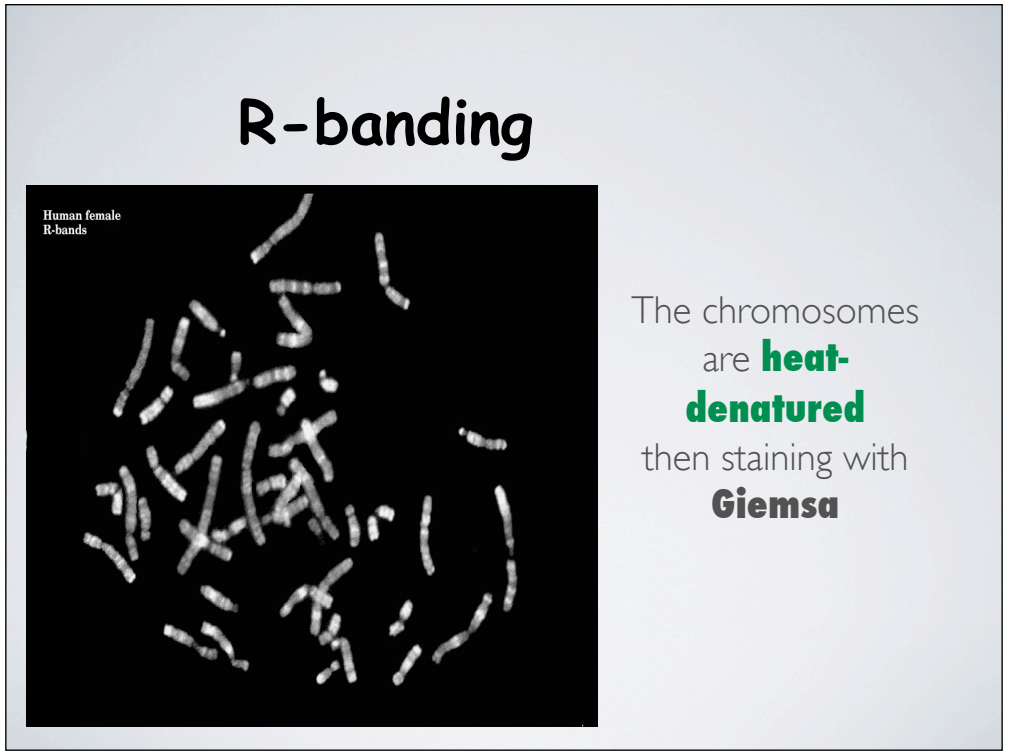
G (giemsa) banding

This is the method most commonly used. The chromosomes are treated with trypsin, which denatures their protein content, and then stained with a DNA-binding dye known as Giemsa, which gives each chromosome a characteristic and reproducible pattern of light and dark bands (Fig. 3.5).



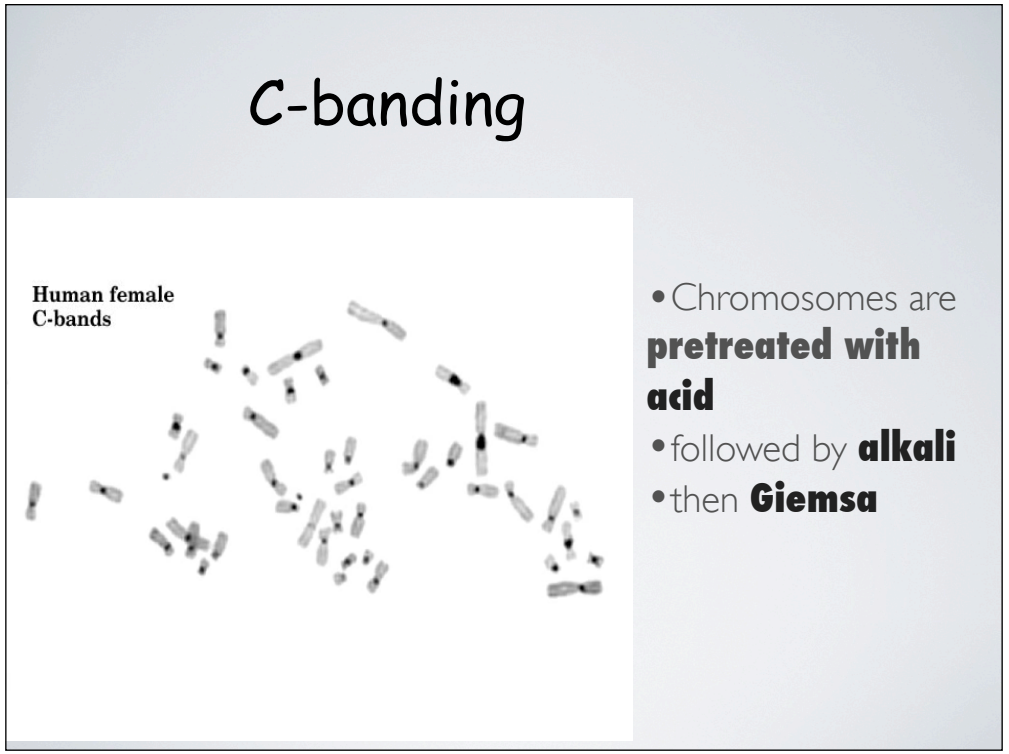
Q (quinacrine) banding

This gives a banding pattern similar to that obtained with Giemsa, and requires examination of the chromosomes with an ultraviolet fluorescent microscope.



R (reverse) banding

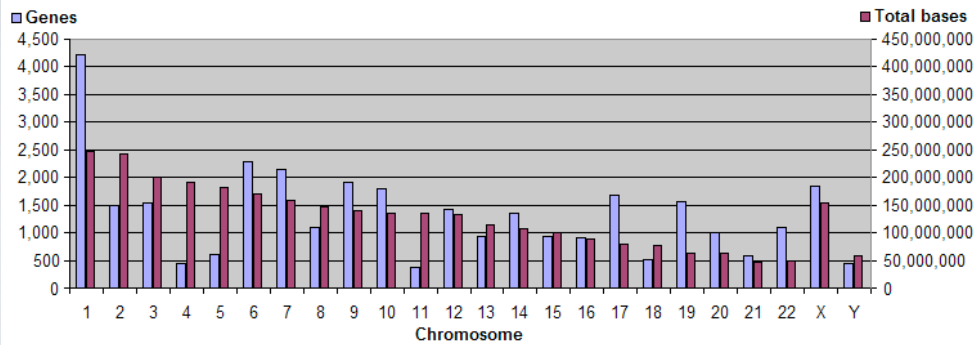
The chromosomes are heat-denatured before staining with Giemsa, yielding light and dark bands which are the reverse of those obtained using conventional G banding (Fig. 3.6).



C (centromeric heterochromatin) banding

If the chromosomes are pretreated with acid followed by alkali prior to G banding, the centromeres and other heterochromatic regions containing highly repetitive DNA sequences are preferentially stained.

CHROMOSOME


Genes
Total bases
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450,000,000
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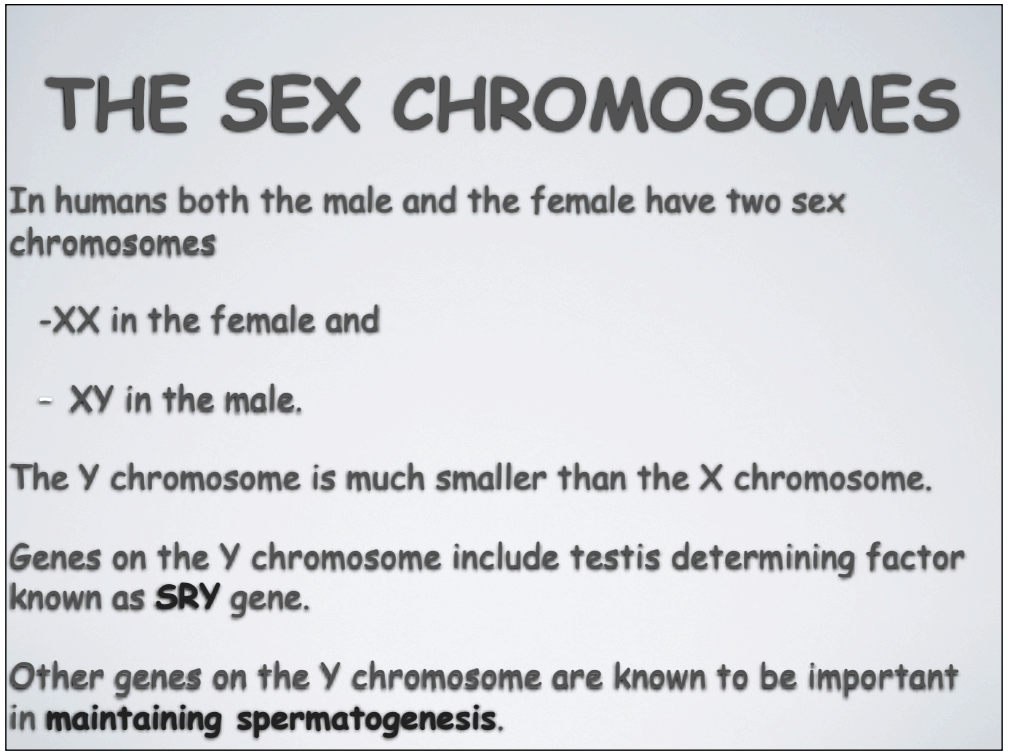
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y
Chromosome



The X and Y chromosomes are known as the sex chromosomes because of their crucial role in sex determination. The X chromosome was originally labeled as such because of uncertainty as to its function when it was realized that in some insects this chromosome is present in some gametes but not in others. In these insects the male has only one sex chromosome (X), whereas the female has two (XX). In humans, and in most mammals, 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 and carries only a few genes of functional importance, most notably the testis-determining factor, known as *SRY* (p. 97). Other genes on the Y chromosome are known to be important in maintaining spermatogenesis.

In the female each ovum carries an X chromosome, whereas in the male each sperm carries either an X or a Y 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 (Fig. 3.3). In fact, slightly more male babies are born than females, although during childhood and adult life the sex ratio evens out at 1:1.

THE SEX CHROMOSOMES

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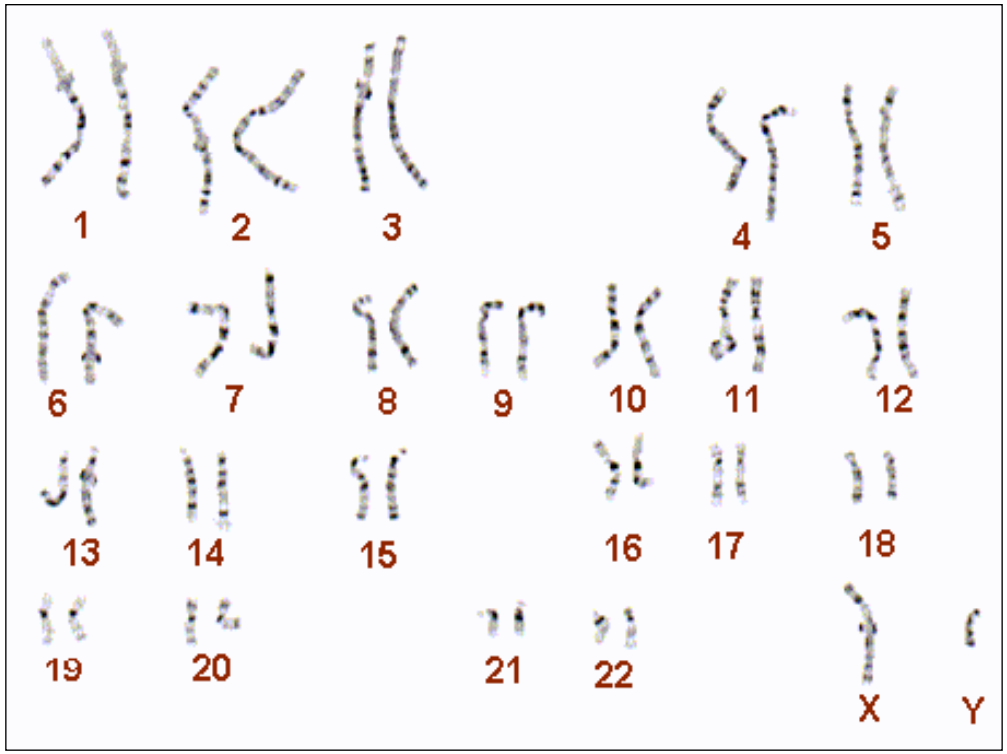
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PUNNETT`S SQUARE SHOWING SEX CHROMOSOME COMBINATIONS FOR MALE AND FEMALE GAMETES