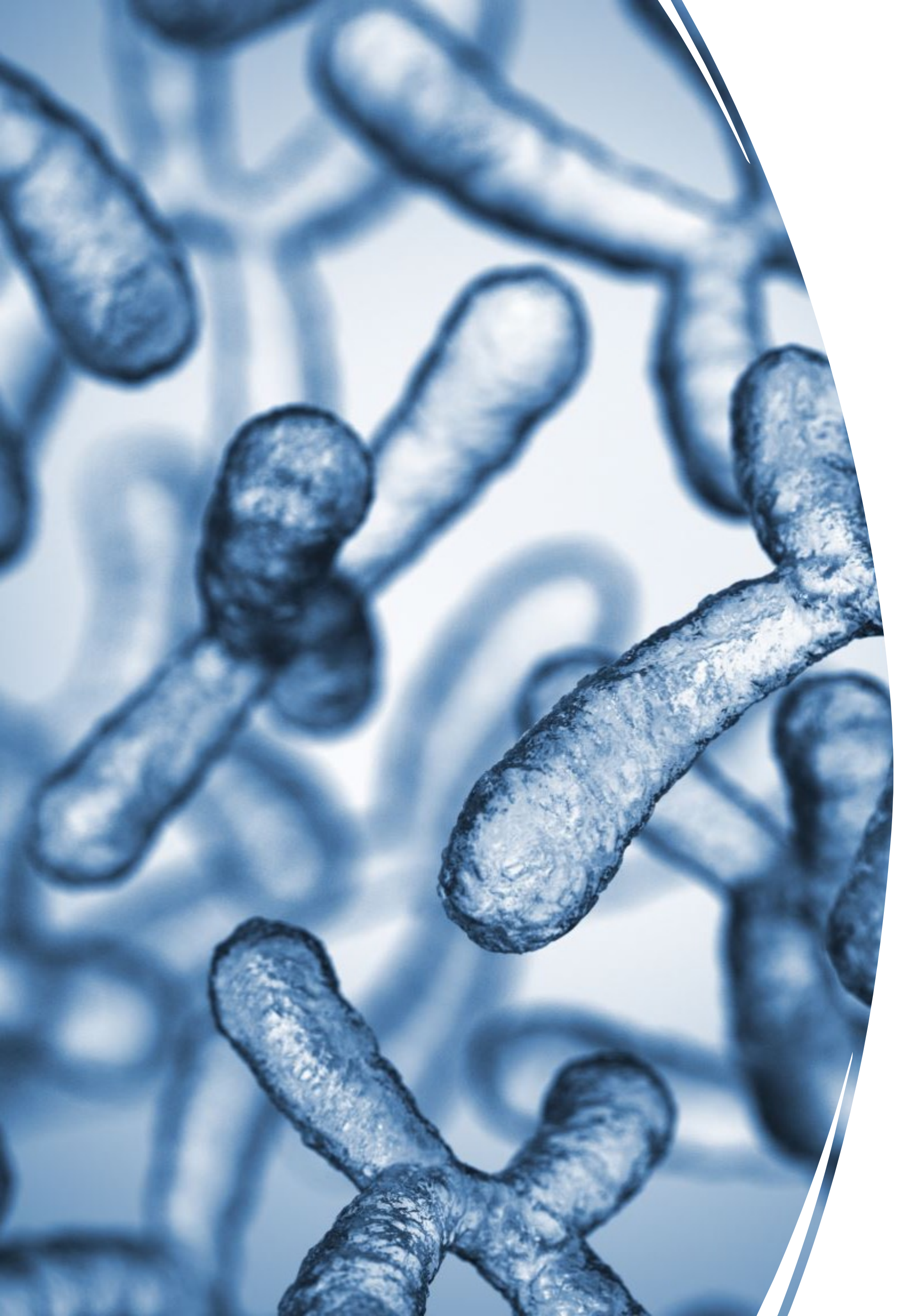


Chromosomes Structure and Organization II

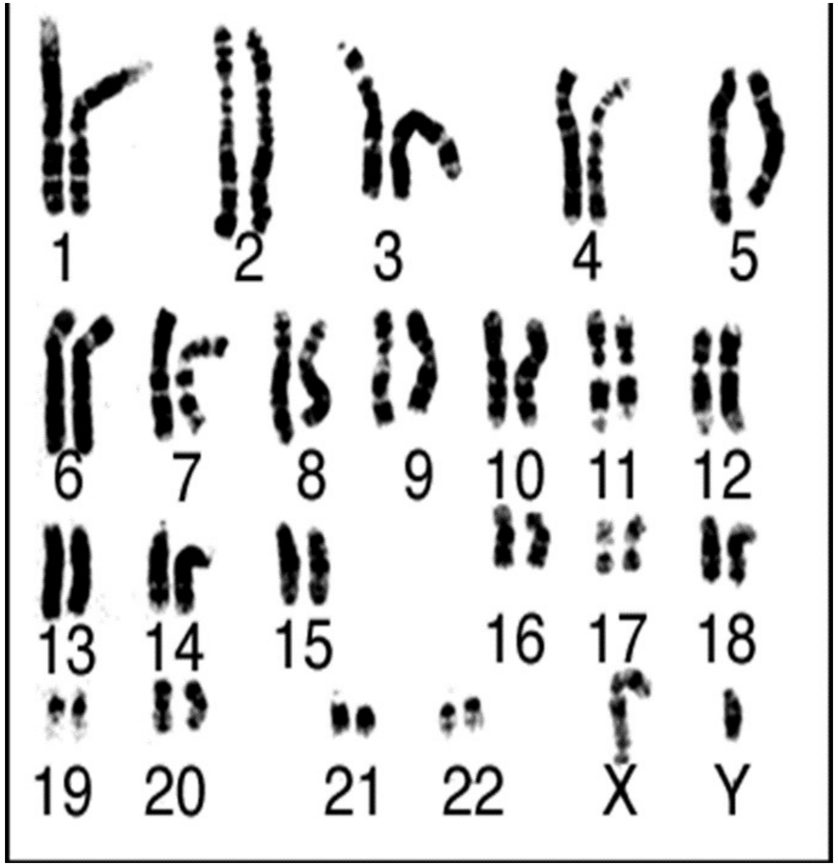
Lecture 5

Chromosome

- Chromosome made of DNA and protein
- Each human cell has 23 pairs of chromosome

  Except gametes cells (sperm, ovum)
- Autosome chromosomes from
- 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)
  - A normal sperm contain 22 **autosomes** (chromosome that does not have a gene that determines sex) and one (either X or Y) **sex chromosome.**
  - A normal egg contain 22 autosomes and one sex chromosome (always X).

Homologous pairs of chromosomes

- Members of a pair of chromosome are known as **homologs**

  (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.

Chromosome banding

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

This technique (staining) also revealed that **chromatin**, the combination of DNA and histone proteins that comprise chromosome, exists in two main forms.

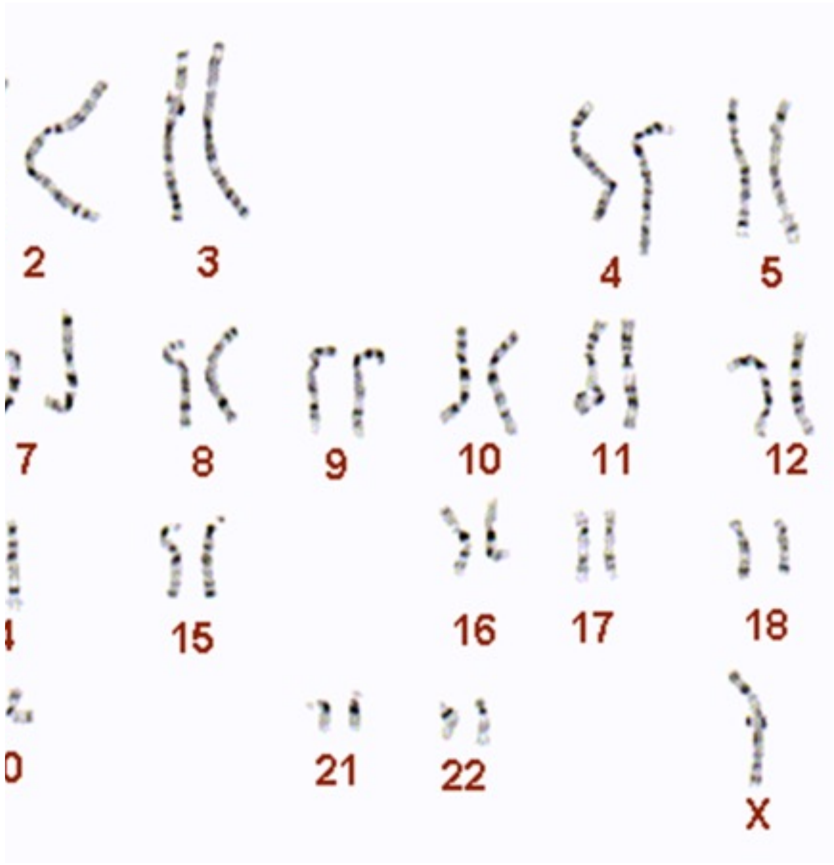
Euchromatin stains lightly and consists of genes which are actively expressed.

Heterochromatin stains darkly and is made up largely of inactive unexpressed repetitive DNA.

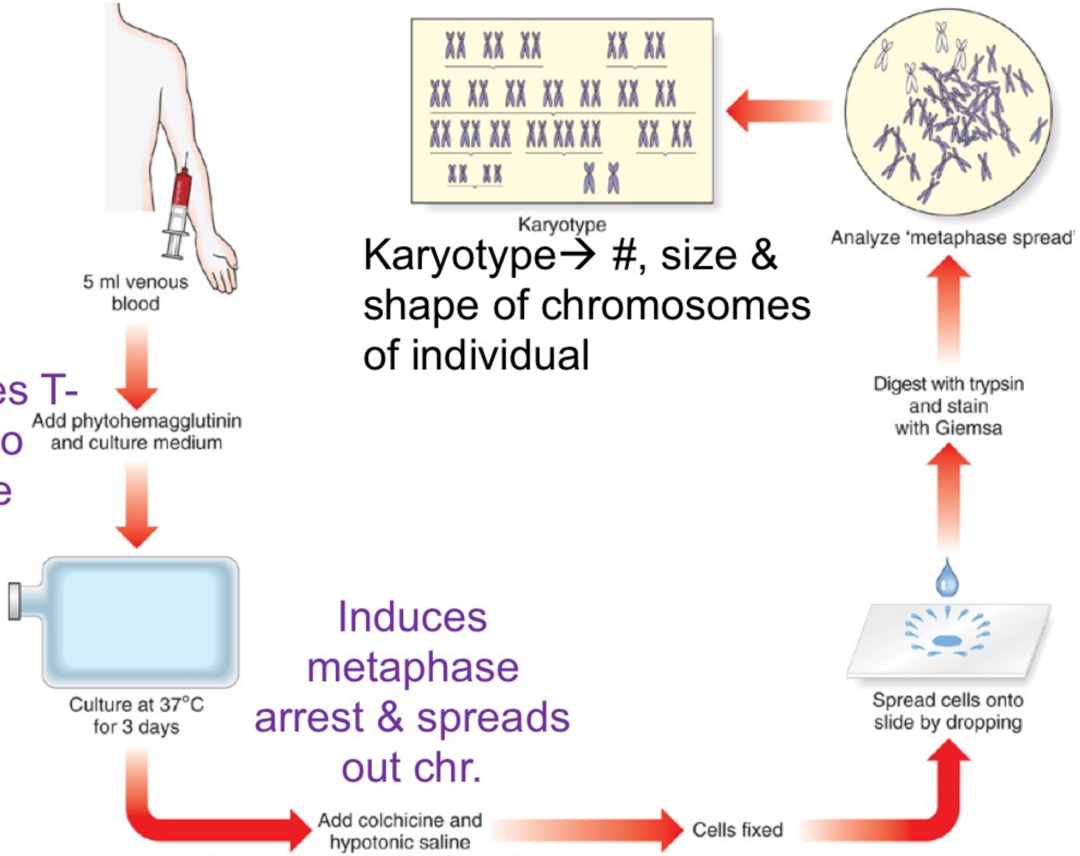
Chromosome and karyotyping

Karyotype

- Picture of chromosome
- Karyotyping is the determination of the number, size and shape of the chromosomes of an individual.
- This term is also used to describe a photomicrograph of an individual's chromosomes arranged in a standard manner.
- Special stains selectively taken up by DNA have enabled each individual chromosome to be identified.



Preparing a Karyotype



HA stimulates T-cells to divide

Karyotype→ #, size & shape of chromosomes of individual

Induces metaphase arrest & spreads out chr.

G banding technique

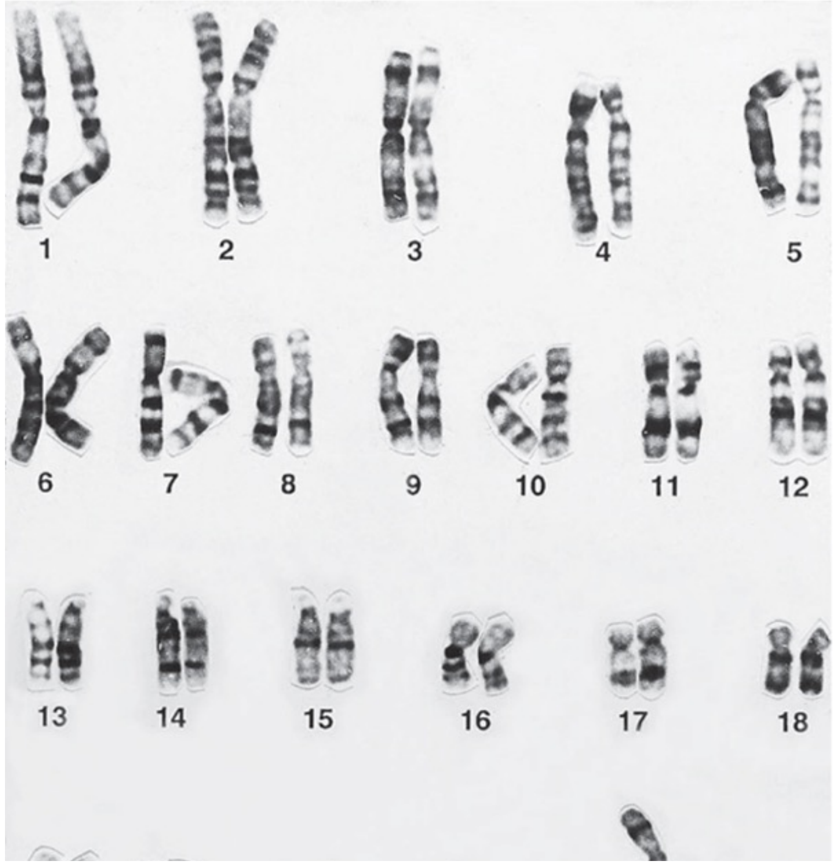
- G banding is the most common procedure, uses G (**Giemsa)** stain, chromosomes are treated with trypsin of metaphase.

G bands are areas that

- Stain dark with G banding
- Replicate DNA late in the S phase
- AT rich
- Have relatively **fewer genes** than R bands

R bands are areas that

- Stain light with G banding
- Replicate DNA early in the S phase
- GC rich
- Have the highest gene density

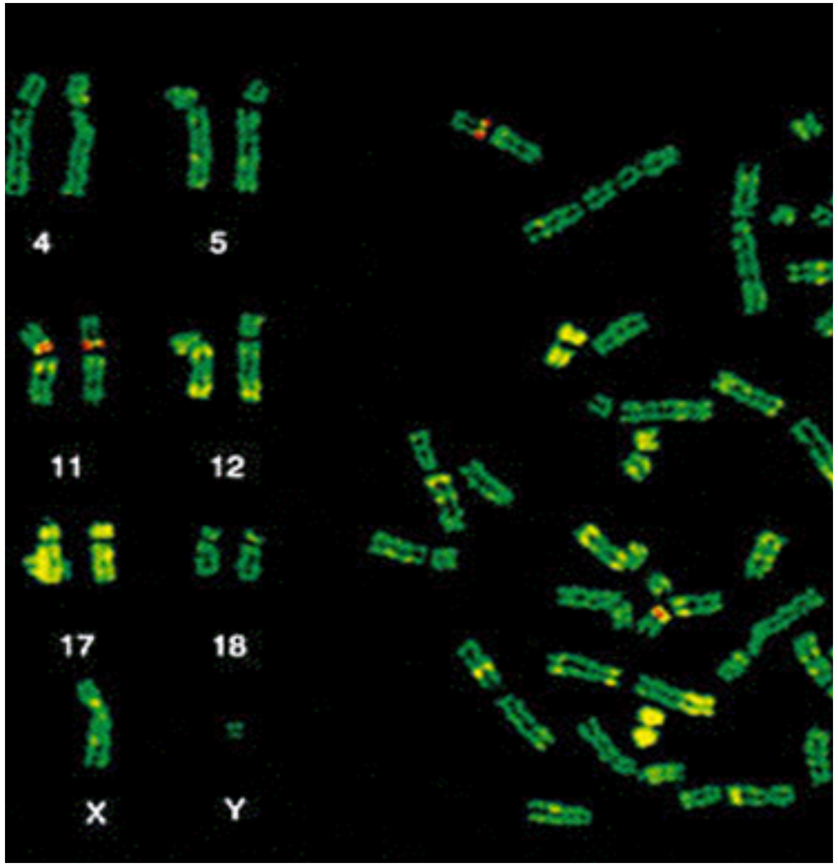


R banding

- R banding or **reverse** banding, uses Giemsa dyes under elevated temperatures to produce the reverse of the pattern seen in G banding or Q banding
- Fluorescent dyes with a high-binding affinity to GC-rich DNA can also produce R banding

Q-banding

- Q banding uses **quinacrine** to stain chromosomes, which are viewed with ultraviolet fluorescent microscopy.
- Q banding gives a banding pattern similar to that obtained with Giemsa



C-banding

- C banding requires **heating** in an **alkali solution** and staining with Giemsa.
- Or pretreated with acid followed by alkali then Giemsa.
- C bands are area of constitutive heterochromatin located adjacent to the **<u>centromeres</u>** of all chromosomes.

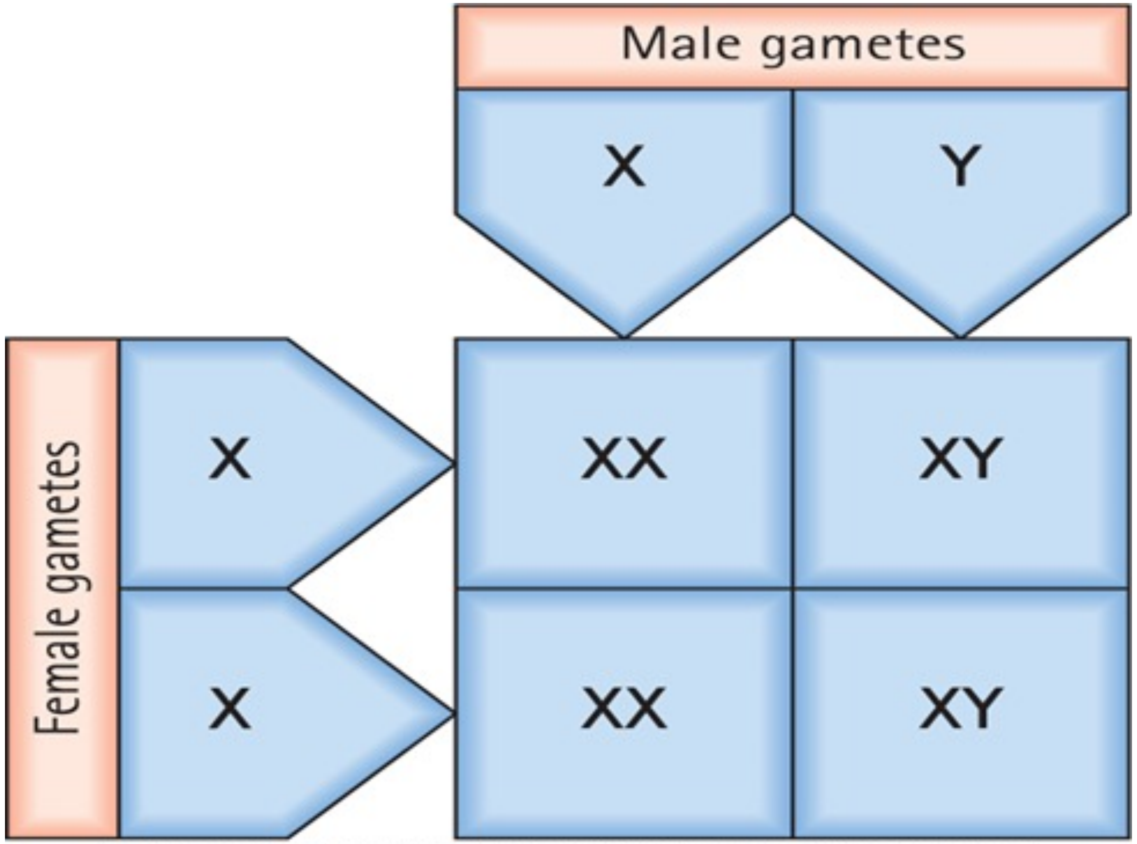
The sex chromosome

- 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 *<u>smaller</u>* than the X chromosome.
- A few genes are 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.

Determination the sex of the baby?

- **Sperm** cells can carry either an **X or a Y chromosome**.
- 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



Thank you