

CYTOGENETIC METHOD

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CYTOGENETICS

- ▶ Cytogenetics = The study of chromosome number, structure, function, and behavior in relation to gene inheritance, organization and expression

CHROMOSOME

▶ Chromosome

Chromo = colored in
response to dye

Some = body

Chromosome of Eukaryotes have been the traditional subject for
cytogenetic

analysis because they are large enough to be examined with light
microscope

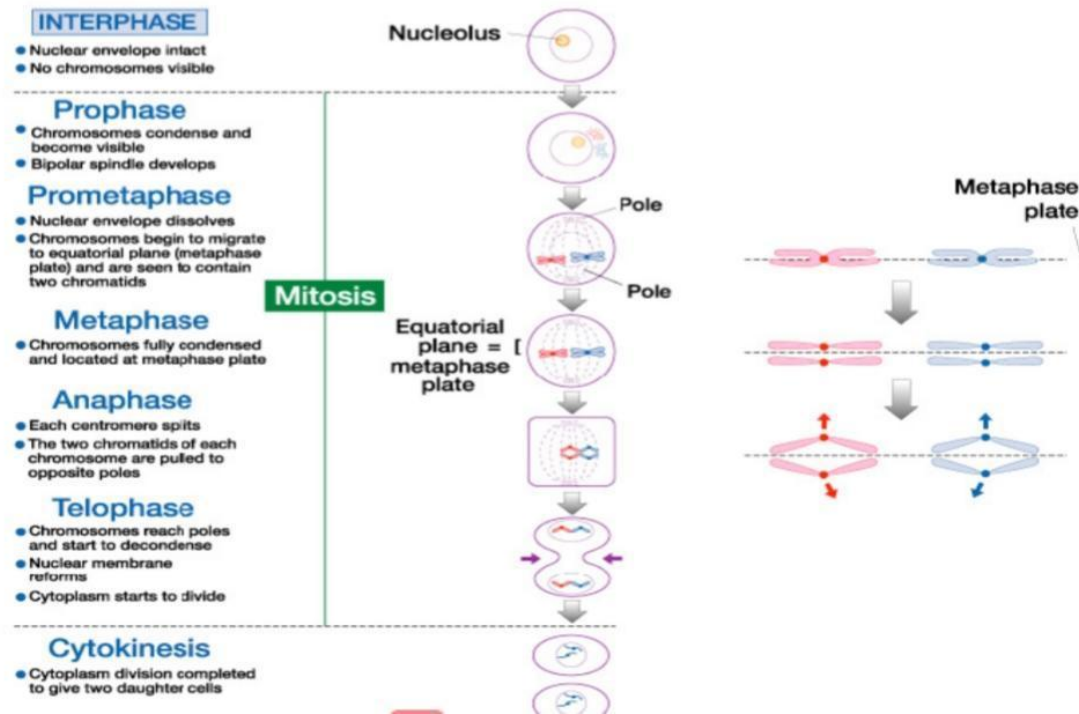


Why Analyse Chromosomes and Genes?

Genetic errors arise from deletions or insertions of genetic material, abnormal numbers of whole chromosomes or genes, and even from misplacement of a single base in the DNA sequence.

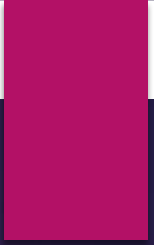
Genetic abnormalities can range from relatively harmless to severe: from vitamin deficiency and food allergies to cancer, birth defects and infant mortality.

Cytogenetic methods to detect chromosomal abnormalities underlying human birth defects usually involve analysis of mitotic chromosome



What tissues are appropriate for chromosome study?

- A tissue that can be stimulated to undergo cell division in-vitro
 - It is only during mitosis of the cell cycle that distinct chromosomes can be visualized with a light microscope
- After culturing, in-vitro, a proportion of cells are arrested in mitosis, and are then “harvested” for chromosome analysis. After harvesting, the cell preparations are dropped onto glass slides and stained. For most chromosome analyses, a G-banding technique is utilized for staining.
- Metaphase spread



The chromosomes are so named as they may be stained by certain dyes

Chromosomes are composed of chromatin, which is composed of protein and DNA

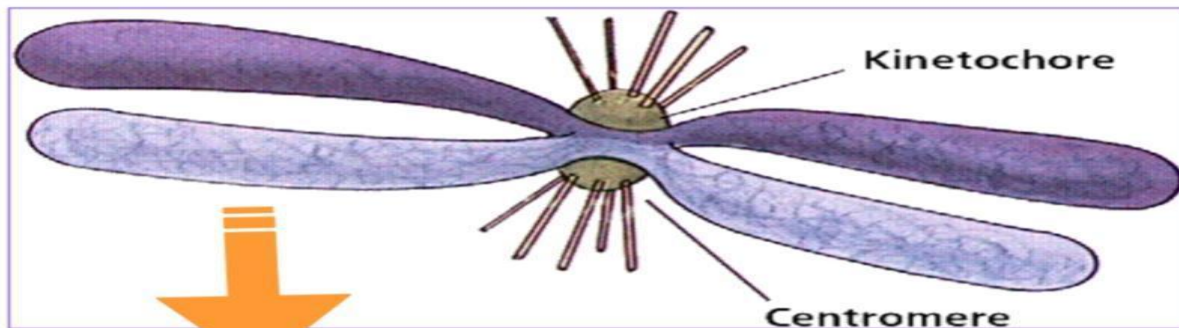
When cells are not dividing, the genetic material is decondensed

Chromosomes become visible as distinct structures when the cell divides

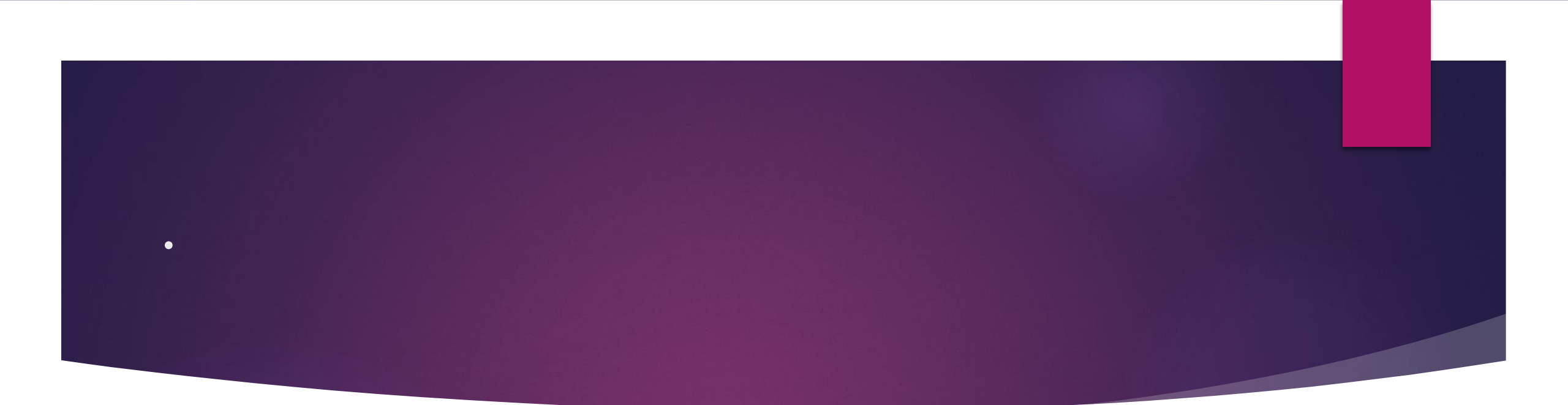
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Chromosome

Chromosome



Sister Chromatides



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Chromosomes of different species differ
in number and information content
Humans and several other species of
organisms have 46 chromosomes

Karyotyping

Karyotype

A pictorial display of metaphase

chromosomes from a mitotic cell

Homologous chromosomes-pairs

Karyotyping

Karyotyping is the analysis of chromosomes

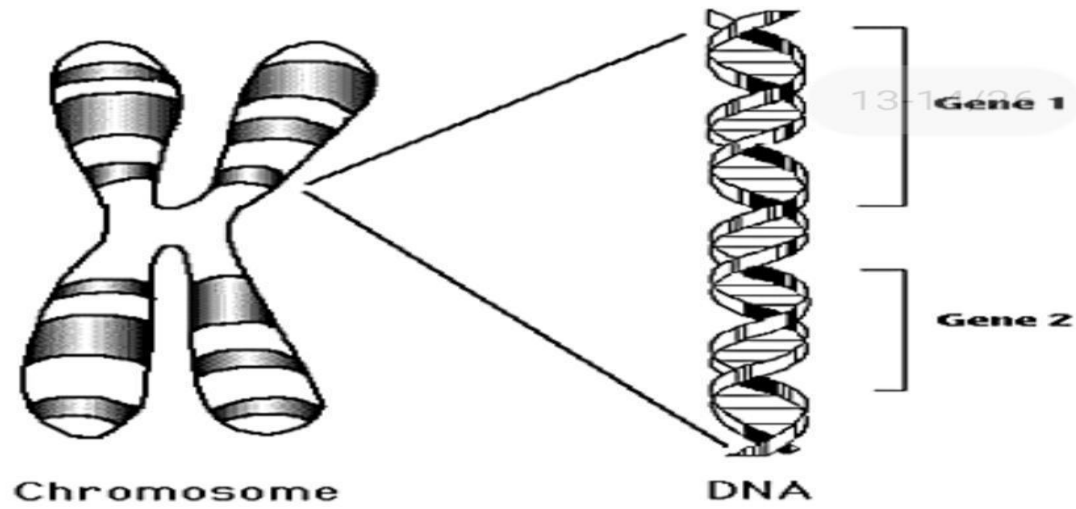
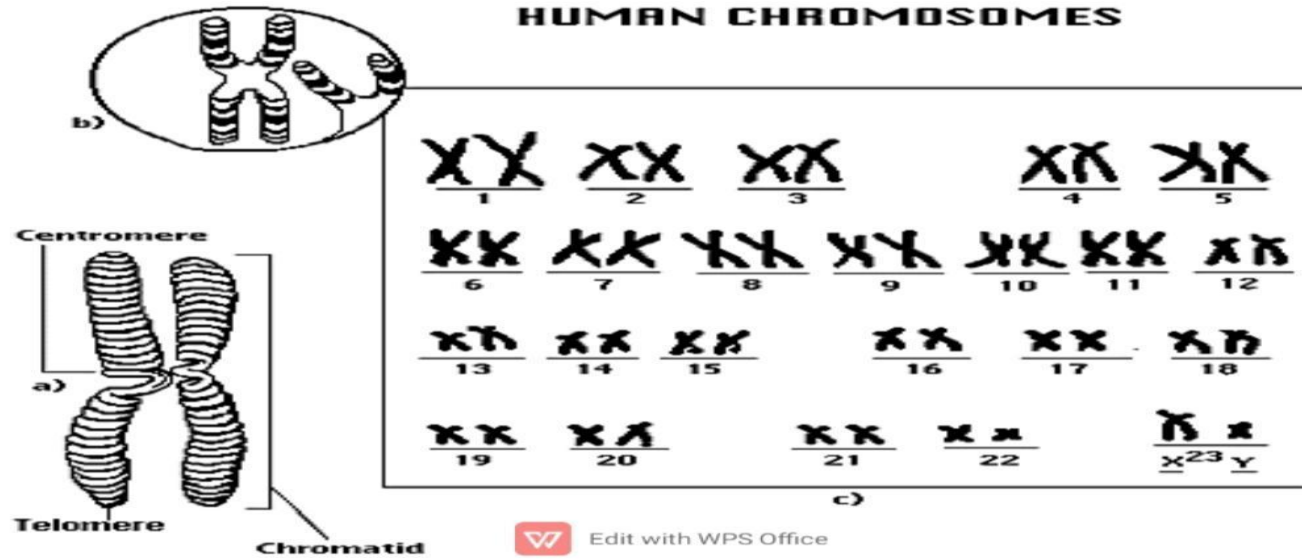
Cytogenetics is the study of chromosomes and inheritance

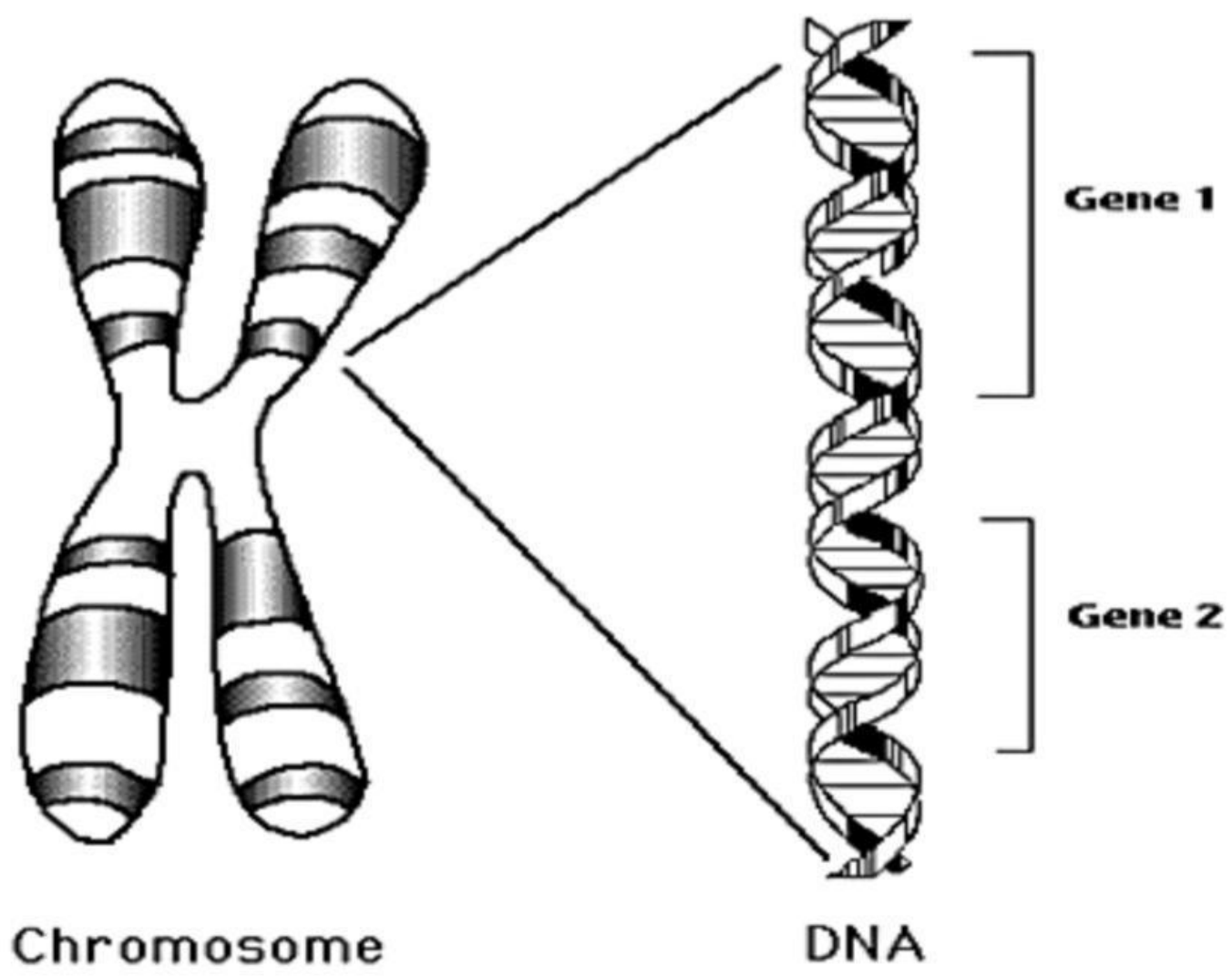
Cytogenetics is based on studies of humans as well as *Drosophila* and other organisms

Preparing a karyotype

- ▶ Harvested cells are first cultured
- ▶ The cells are then treated with colchicine which arrests the cells in metaphase, and then treated and stained to observe the chromosomes
- ▶ Chromosomes can be photographed or visualized using a computer, and then analyzed
- ▶ Chromosomes are identified by size, position of the centromere, and banding and staining regions

HUMAN CHROMOSOMES



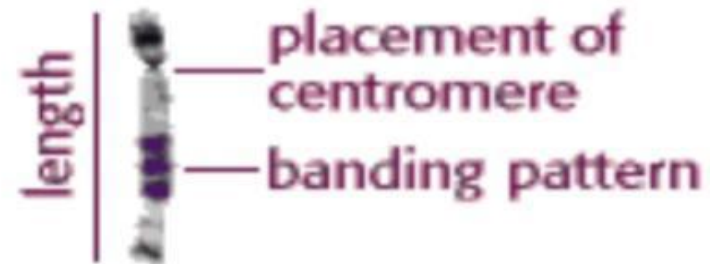


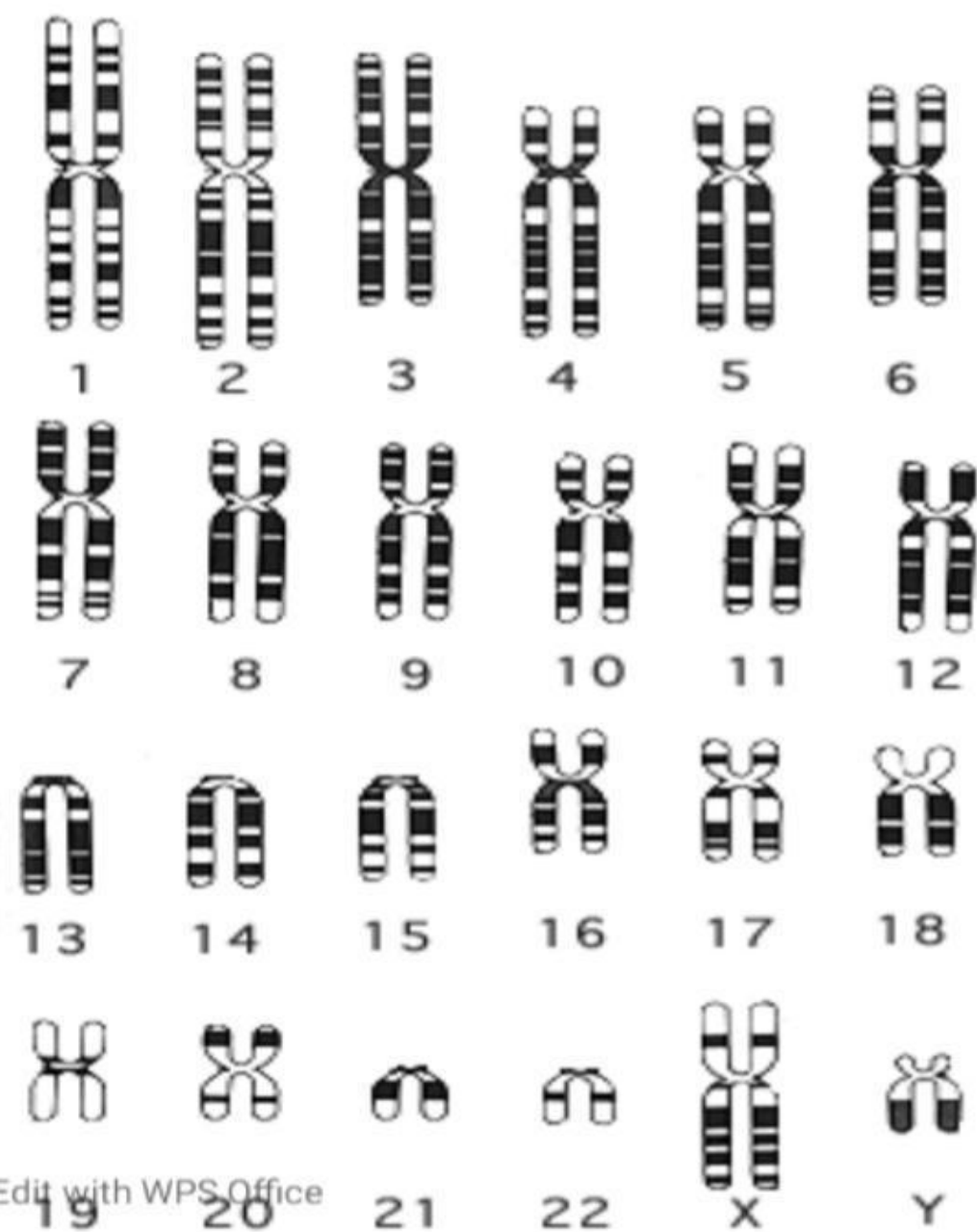
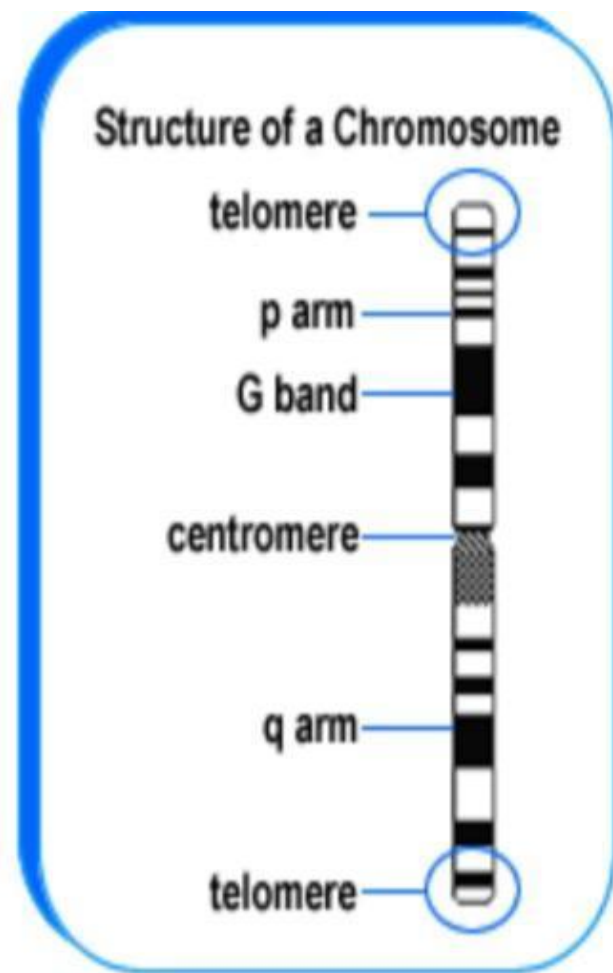
The analysis involves comparing chromosomes for their length, the placement of centromeres (areas where the two chromatids are joined), and the location and sizes of G-bands.

Chromosome smear



Identifying features of a chromosome



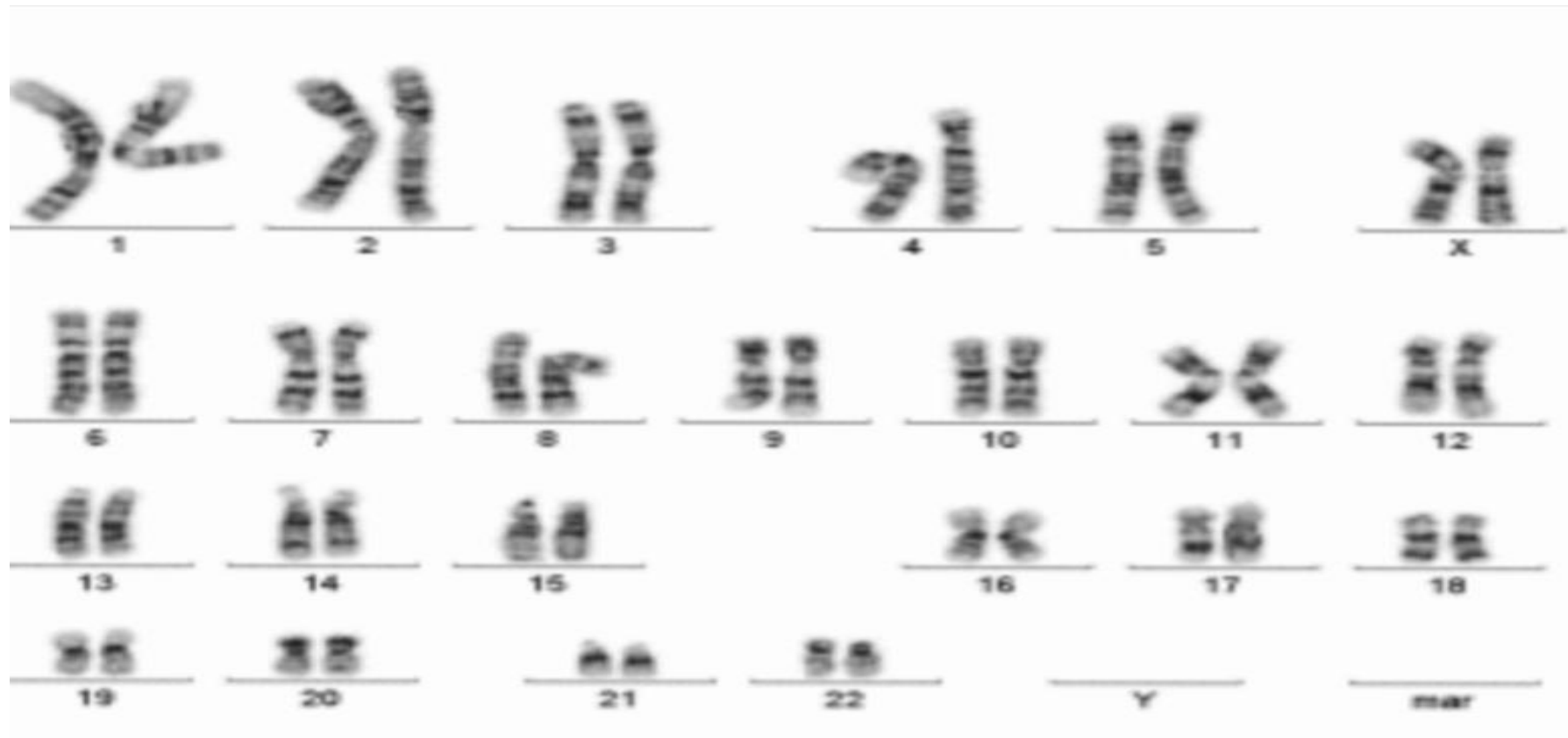


[A cartoon of a chromosome. The ends of the chromosome are called telomeres. The centromeres is the narrowing of the chromosome. The chromosome has 2 arms – the smaller is called the p arm, the larger, the q arm. The black bands are called G bands.]

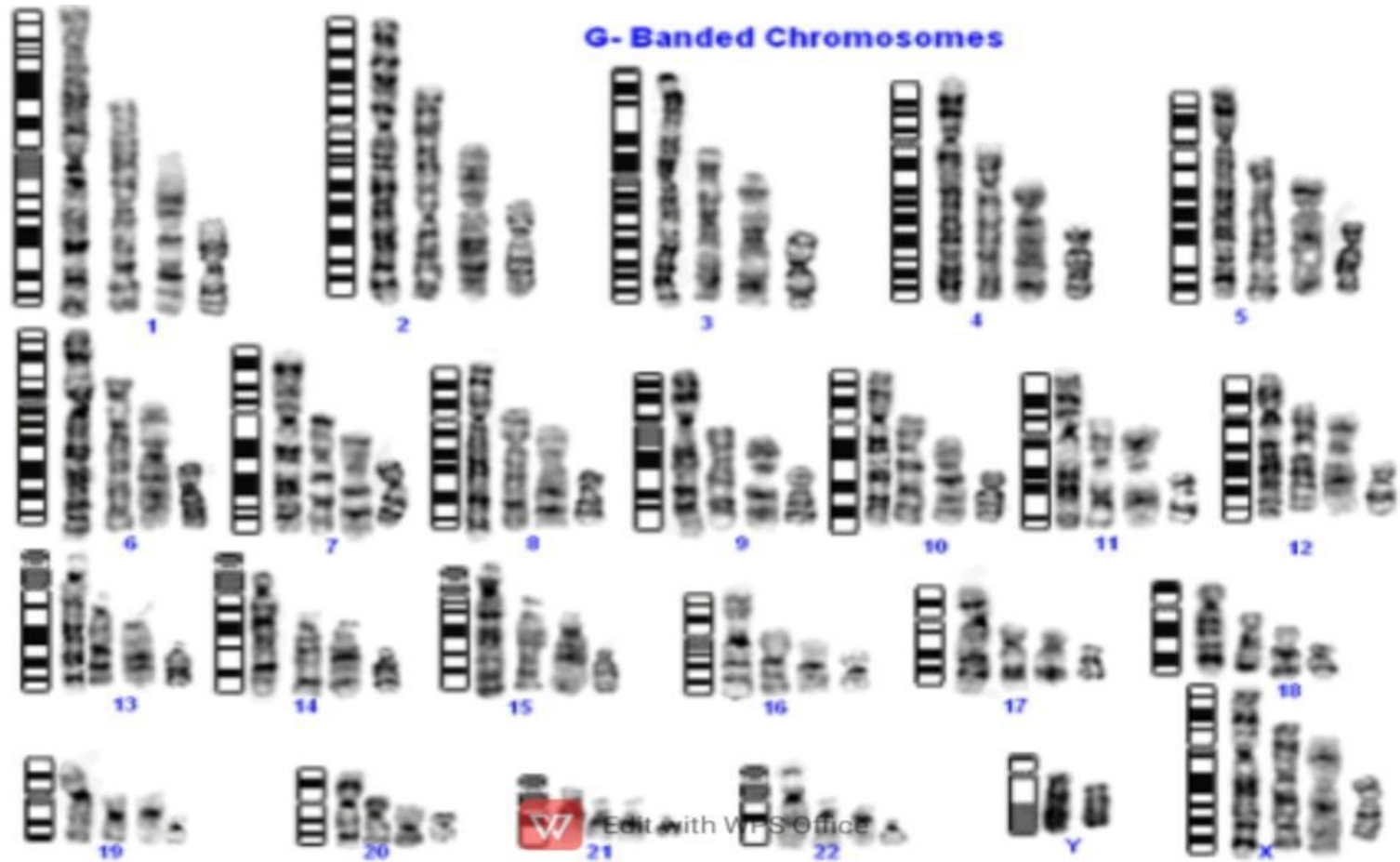
METAPHASE CHROMOSOMES



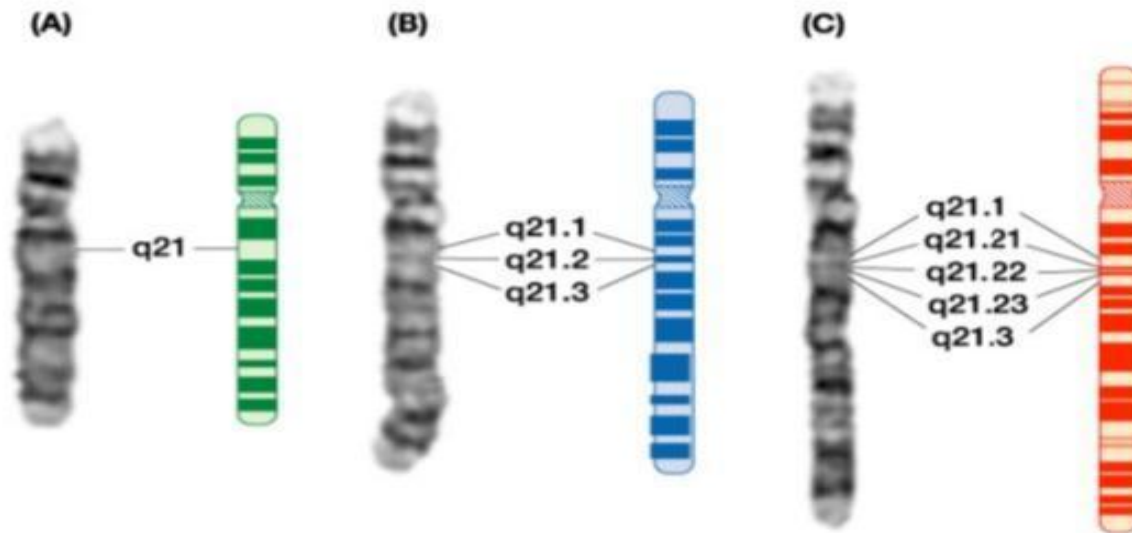
KARYOTYPED CHROMOSOMES



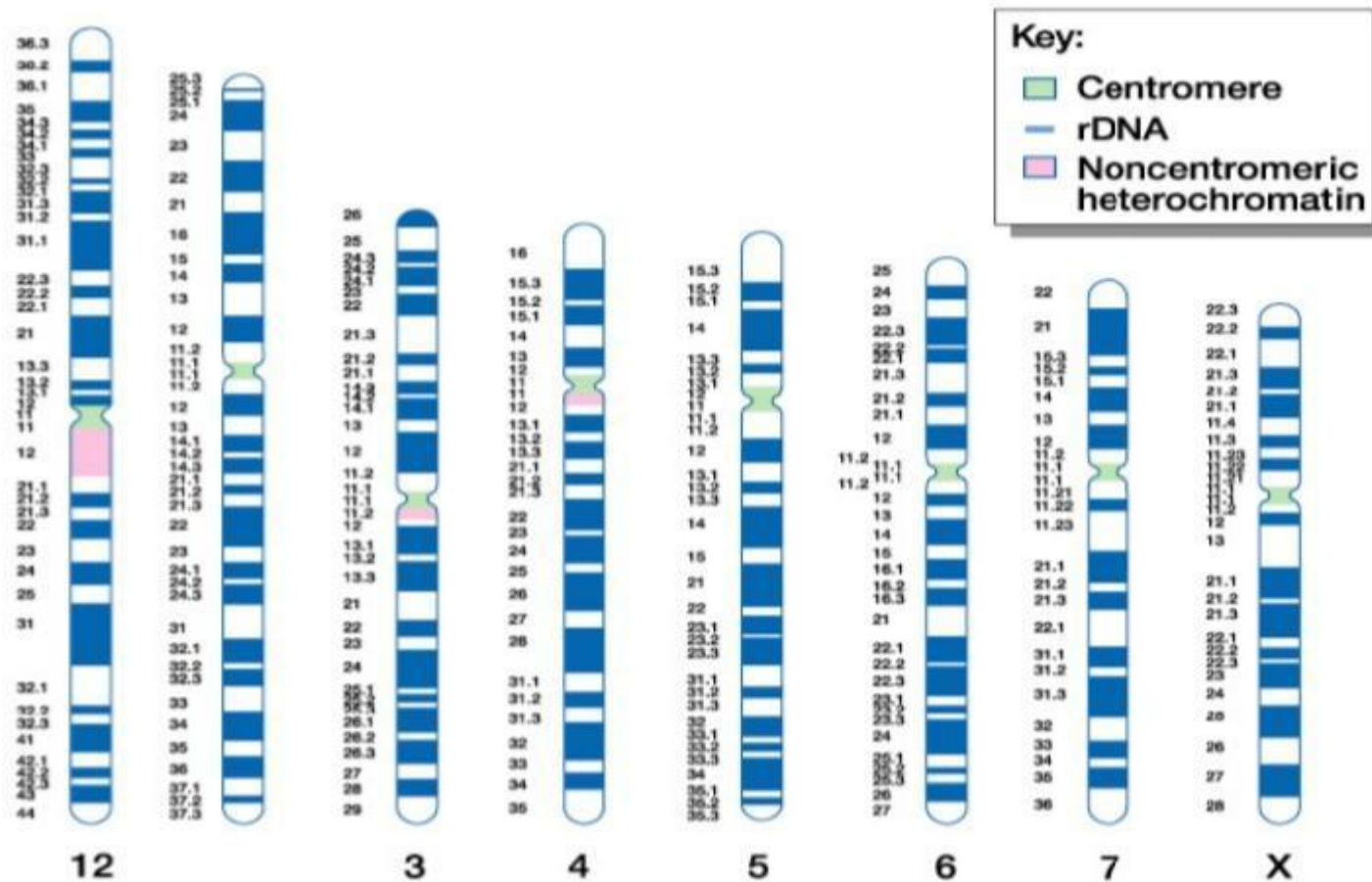
G – BANDED CHROMOSOMES

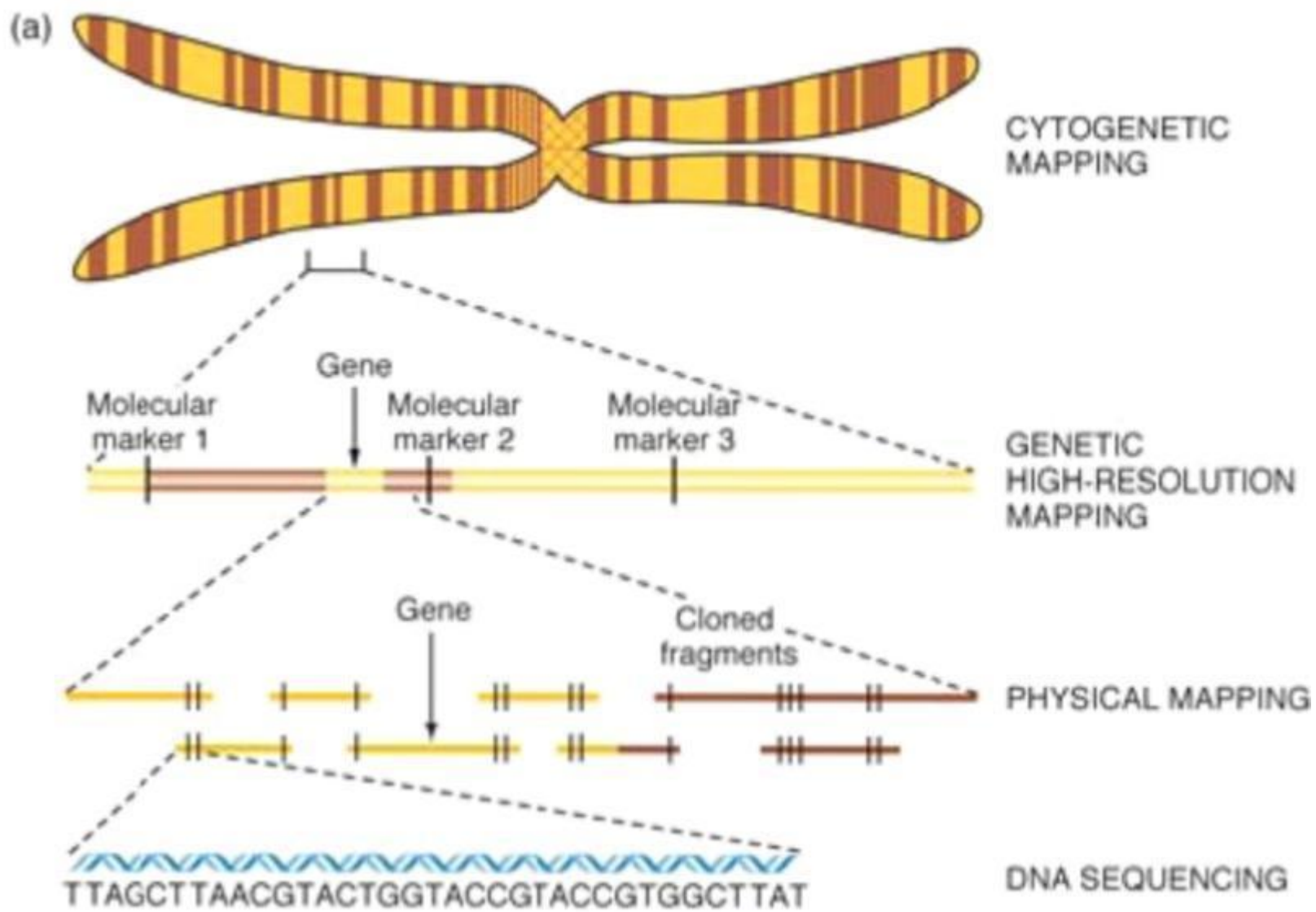


Banding patterns on human mitotic chromosomes due to regions of condensed chromatin (darker - G bands) and less condensed chromatin (lighter - R bands)



Human chromosome number is determined by their length in "mitotic figures"





International System for Cytogenetic Nomenclature, (ISCN,1995)

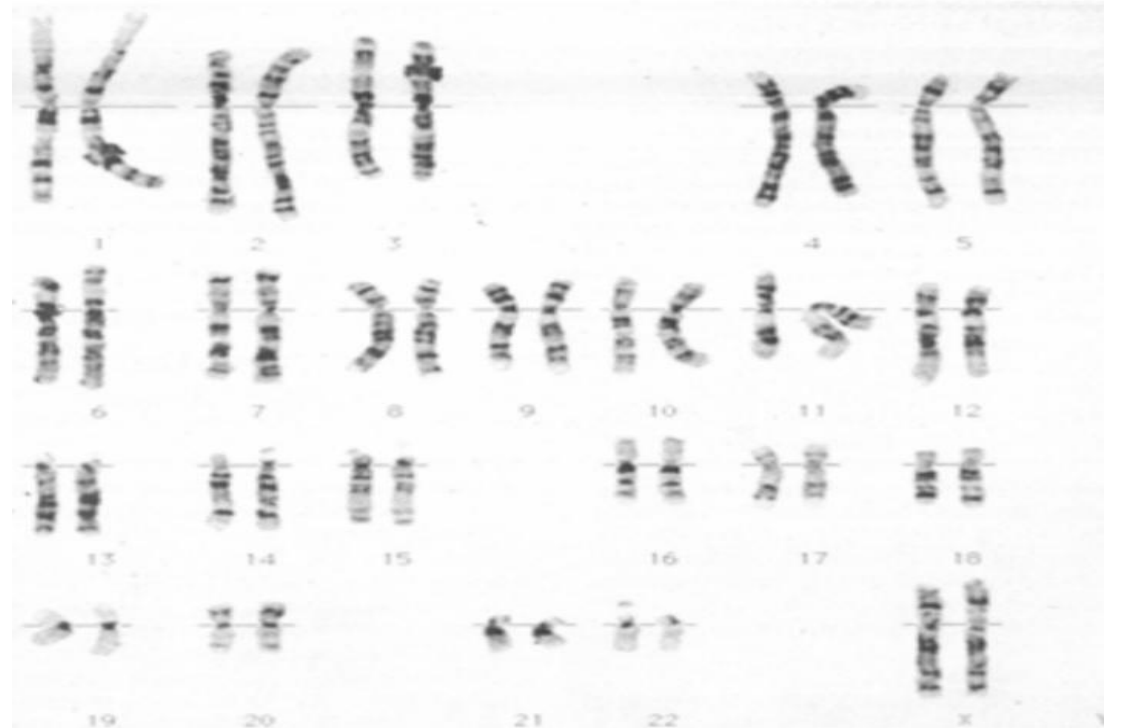
Short arm of the chromosome = p

Long arm of the chromosome = q

Bands are numbered independently on the short and long arms

Centromeres = p10,q10

Band numbers increase as move from the centromere to the telomere





Hundreds of genes are encompassed within a single G-band.

Therefore, most constitutional chromosome abnormalities are associated with multiple congenital anomalies.

Therefore, deletion of a single gene cannot be

detected by G-banding.

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Conclusion

The evolution of cytogenetic techniques and the mapping of the human genome have provided scientists with a great deal of insight into the causes of numerous genetic disorders. Though rooted in early chromosome staining and gene mapping techniques, modern FISH, SKY, and CGH methods have far outshone their predecessors by providing an unprecedented view of human chromosomes.

QUESTIONS

- 1) WHAT IS KARYOTYPING?
- 2) Why Analyse Chromosomes and Genes?



THANK YOU