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 insertionsof 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 deficiencie 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 invitro

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

Chromosome

Chromosome





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 chromosomespairs

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 cltured
- 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 visualizedd using a computer, and then analyzed
- Chromosomes are identified by size, position of the centromere, and banding and staining regionss





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.





METAPHASE CHROMOSOMES



KARYOTYPED CHROMOSOMES



G – BANDED CHROMOSOMES



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



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



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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

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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.

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?

