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Short Communication

Cytogenetics Approaches and Medicinal Applications

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DESCRIPTION

Cytogenetics is a field of genetics that also consists of elements of cell biology and cytology. It is concerned with how chromosomes affect cell behaviour, notably during mitosis and meiosis.

Karyotyping, examination of G-banded chromosomes, additional cytogenetic banding methodologies, and molecular cytogenetics techniques such as Fluorescence *In Situ* Hybridization (FISH) and comparative genomic hybridization are among the strategies utilized. Various techniques are involved in cytogenetics. They are:

Karyotyping

Routine genome analysis (karyotyping) is the examination of metaphase chromosomes that have been banded with trypsin and then stained with Giemsa, Leishmann's, or a combination of both. This results in distinct chromosomal banding patterns. Although the chemical mechanism and cause of these patterns are unknown, they are most likely connected to replication time and chromatin packing [1].

In cytogenetics laboratories, several chromosome-banding procedures are used. Quinacrine banding (Q-banding) was the first approach to creating precise banding patterns. This approach, which necessitates the use of a fluorescence microscope, is no longer as popular as Giemsa banding (G-banding). Heat treatment is required for reverse banding, which reverses the conventional black-and-white pattern seen in G-bands and Q-bands. This approach is very useful for staining chromosomal distal ends [2]. C-banding and nucleolar organising region stains are two further staining techniques (NOR stains). These new approaches dye specific chromosomal segments. NOR staining emphasizes the satellites and stalks of acrocentric chromosomes, while C-banding highlights constitutive heterochromatin, which is frequently seen at the centromere.

The staining of chromosomes during prophase or early metaphase (prometaphase) before they achieve maximum condensation is classified as top banding. Because prophase and prometaphase chromosomes are longer than metaphase chromosomes, the number of bands visible on all chromosomes(bands per haploid set, BPH; "band level") rises from 300 to 800.

Slide preparation

A salt solution, usually 2X SSC, is used to age the slide (salt, sodium citrate). After dehydrating the slides with ethanol, the probe combination is added. After that, the sample and probe DNA are co-denatured on a hot plate and re-annealed for at least 4 hours [3]. After washing to remove any unbound probe, the slides are counterstained with 4',6-Diamidino-2-phenylindole (DAPI) or propidium iodide.

Analysis

A clinical laboratory specialist in cytogenetics analyses FISH specimens using fluorescence microscopy. In oncology, a large number of interphase cells are counted and scored in order to rule out low-level residual illness, often between 200 and 1000 cells. Generally, 20 metaphase cells are evaluated for chromosomal defects.

Human abnormalities and medical applications

Despite the launch of procedures that made chromosome enumeration simple, findings about abnormal chromosomes and chromosome numbers were made swiftly. Cytogenetics revealed the nature of the chromosomal abnormality in some congenital illnesses, such as Down syndrome: a "simple" trisomy. Cells with aneuploidy (additions or deletions of whole chromosomes) in one or both of the parents or the fetus can result from nondisjunction occurrences. Sex chromosomal abnormalities were detected as well as other numerical abnormalities [4]. Turner syndrome is caused by a female having only one X chromosome, whereas Klinefelter syndrome is caused by a male having an additional X chromosome, resulting in 47 total chromosomes. Many more sex chromosomal combinations, such as XXX, XYY, and XXXX, are compatible with live birth. Mammals' ability to tolerate aneuploidies in the sex chromosomes is due to their ability to inactivate them, which is required in normal females to compensate for having two copies of the chromosome. Individuals with extra X chromosomes have a phenotypic effect because not all genes on the X chromosome are inactivated.

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CONCLUSION

It is concerned with how chromosomes affect cell behavior, notably during mitosis and meiosis. Techniques Karyotyping Routine genome analysis (karyotyping) is the examination of metaphase chromosomes that have been banded with trypsin and then stained with Giemsa, Leishmann's, or a combination of both. Cytogenetics are developed by various techniques.

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