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Cytogenetics and chromosomes are two terms that are used interchangeably.

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Perspective

Chromosomes

A chromosome is a well-organized DNA package located in the cell nucleus. The number of chromosomes in each organism varies. Humans have 23 pairs of chromosomes: 22 pairs of autosomes (numbered chromosomes) and one pair of X and Y chromosomes. Each parent contributes one chromosome to each pair, giving offspring half of their mother's chromosomes and half of their father's. In a cell, a chromosome is the structure that houses DNA. Chromosomes have a complex structure that contains elements required for processes like replication and segregation. In terms of number and arrangement, each species has its own set of chromosomes. Humans, for example, have 23 pairs of chromosomes: 22 pairs of autosomes (numbered 1 through 22) and one pair of sex chromosomes (X and Y). Each pair of parents provides one chromosome to their offspring.

There are 22 pairs of autosomes and one pair of sex chromosomes in a normal human karyotype. On karyotypes, aneuploidies, or variations in chromosomal number, are easily observed. Because of the resulting imbalance in gene expression, most aneuploidies in humans are fatal. Trisomy 21, or Down syndrome, is a notable exception, as it is routinely found during prenatal screening of older moms. In humans, X inactivation maintains near-normal expression levels for X-linked genes, thus sex chromosomal aneuploidies are tolerated. Karyotypes can reflect more subtle changes in chromosomal shape in addition to variations in chromosome number. In fact, a chromosome's natural banding pattern acts as a "bar code" that may be translated into a chromosome map. The positions of structural abnormalities, such as deletions, duplications, and translocations, can then be identified to within a few megabases of DNA using coordinates on these approximate chromosome maps, or idiograms.

Cytogenetics

The field of genetics that investigates the structure of DNA within the cell nucleus is known as cytogenetics. During cell division, this DNA is condensed and forms chromosomes. The quantity and form of chromosomes are studied in cytogenetics. Chromosomal banding procedures (traditional cytogenetics) or fluorescently labelled hybridization probes (molecular cytogenetics). In most cells of the body, the quantity and shape of chromosomes in a cell of a certain species remain always constant (with the exception of reproductive cells and others such as the liver). This is a trait unique to each species; for example, humans have 46 chromosomes.

When scientists learned that chromosomes are the physical bearers of genes, the field of cytogenetics arose in the early twentieth century. Researchers built on the data of their colleagues to synthesise the chromosomal hypothesis of inheritance, as is customary in science. This revolutionary theory was based on cytologists' comprehensive observations of chromosome motions during mitosis and meiosis, which revealed that chromosomal behaviour may explain Mendel's inheritance laws.

The term "cytogenetics" refers to the study of genetic material under a microscope. Previously, this was done with a light microscope and chromosome analysis. Even before we understood they were formed of DNA, chromosomes were visible to those who used microscopes. We can now utilise improved cytogenetics techniques like fluorescence in situ hybridization (FISH) to look at the genetic material in a cell through a light microscope with considerably higher resolution than before.

Scientists struggled to discern individual chromosomes in the early days of cytogenetics, but over time, they refined the conditions for maintaining and staining chromosomes to the reproducible standard that is now anticipated in clinical cytogenetics. Metaphase chromosomes are stained to produce different banding patterns, and chromosomal pairs are then grouped into a standardised structure known as a karyotype in today's techniques. Cytogeneticists have been able to detect numerous abnormalities in chromosomal number and shape that are related with disease states and developmental disorders because karyotypes are surprisingly similar among individuals of a species.

Fluorescence in situ hybridization

Flexible approaches based on fluorescence in situ hybridization (FISH) have transformed cytogenetics into a molecular science and given cytogeneticists powerful new tools over the last few decades. Labeled DNA or RNA probes are hybridised with their complementary target DNA sequences on chromosomes in FISH methods. Because numerous probes, each tagged with a spectrally unique fluorescent dye, can be utilised in the same experiment, FISH investigations often produce colourful findings. A single gene or a cluster of genes spaced out throughout the length of a chromosome can be used as the target DNA sequence. In clinical cytogenetics, FISH methods are now commonly used. Spectral karyotyping gives a picture of any major chromosome rearrangements or variations in the number of chromosomes in a patient's cells. Cytogeneticists can also positively identify the genes impacted by chromosomal alterations using gene-specific probes. Recently, researchers have started to use comparative genomic hybridization to look at subtle quantitative differences in people's DNA, such as copy number variants (CNVs).

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