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Editorial

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Cytogenetics is Essentially a Branch of Genetics that is concerned with the Chromosomes

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Editorial Note

Techniques used include karyotyping, analysis of G-banded chromosomes, other cytogenetic banding techniques, as well as molecular cytogenetics such as fluorescent in situ hybridization (FISH) and comparative genomic hybridization (CGH). Chromosomes were first observed in plant cells by Karl Wilhelm von Nägeli in 1842. Their behavior in animal (salamander) cells was described by Walther Flemming, the discoverer of mitosis, in 1882. The name was coined by another German anatomist, von Waldeyer in 1888. The next stage took place after the development of genetics in the early 20th century, when it was appreciated that the set of chromosomes (the karyotype) was the carrier of the genes. Levitsky seems to have been the first to define the karyotype as the phenotypic appearance of the somatic chromosomes, in contrast to their genic contents. Investigation into the human karyotype took many years to settle the most basic question: how many chromosomes does a normal diploid human cell contain? In 1912, Hans von Winiwarter reported 47 chromosomes in spermatogonia and 48 in oogonia, concluding an XX/XO sex determination mechanism. Painter in 1922 was not certain whether the diploid number of humans was 46 or 48, at first favoring 46. He revised his opinion later from 46 to 48, and he correctly insisted on humans having an XX/XY system of sex-determination. Considering their techniques, these results were quite remarkable. In science books, the number of human chromosomes remained at 48 for over thirty years.

In the 1930s, Dobzhansky and his coworkers collected Drosophila pseudoobscura and D. persimilis from wild populations in California and neighboring states. Using Painter's techniquethey studied the polytene chromosomes and discovered that the wild populations were polymorphic for chromosomal inversions. All the flies look alike whatever inversions they carry: this is an example of a cryptic polymorphism.

Evidence rapidly accumulated to show that natural selection was responsible. Using a method invented by L'Héritier and Teissier, Dobzhansky bred populations in population cages, which enabled feeding, breeding and sampling whilst preventing escape. This had the benefit of eliminating migration as a possible explanation of the results. Stocks containing inversions at a known initial frequency can be maintained in controlled conditions. It was found that the various chromosome types do not fluctuate at random, as they would if selectively neutral, but adjust to certain frequencies at which they become stabilised. By the time Dobzhansky published the third edition of his book in 1951he was persuaded that the chromosome morphs were being maintained in the population by the selective advantage of the heterozygotes, as with most polymorphisms.

Advent of Banding Techniques

In the late 1960s, Torbjörn Caspersson developed a quinacrine fluorescent staining technique (Q-banding) which revealed unique banding patterns for each chromosome pair. This allowed chromosome pairs of otherwise equal size to be differentiated by distinct horizontal banding patterns. Banding patterns are now used to elucidate the breakpoints and constituent chromosomes involved in chromosome translocations. Deletions and inversions within an individual chromosome can also be identified and described more precisely using standardized banding nomenclature. G-banding (utilizing trypsin and Giemsa/ Wright stain) was concurrently developed in the early 1970s and allows visualization of banding patterns using a bright field microscope. Diagrams identifying the chromosomes based on the banding patterns are known as idiograms. These maps became the basis for both prenatal and oncological fields to quickly move cytogenetics into the clinical lab where karyotyping allowed scientists to look for chromosomal alterations. Techniques were expanded to allow for culture of free amniocytes recovered from amniotic fluid, and elongation techniques for all culture types that allow for higher-resolution banding.

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