

# Karyotyping to Genomic Profiling: The Evolution of Cytogenetics

Carrillo Pratt\*

Department of Human Genetics, University of Saimaa, South Karelia, Finland

## DESCRIPTION

Cytogenetics is a branch of genetics that delves into the study of chromosomes, their structure, function, and abnormalities. It plays a pivotal role in understanding the genetic basis of various diseases, aiding in diagnosis, prognosis, and therapeutic decisions. This study explores the fascinating world of cytogenetics, its principles, applications, and its contributions to unraveling the complexities of genetics and human health.

Chromosomes are the cellular structures where genetic information is stored. Humans typically have 46 chromosomes, comprising 23 pairs, with one set inherited from each parent. Cytogenetics focuses on studying these chromosomes and their variations.

### Role of karyotyping

Karyotyping is a fundamental cytogenetic technique that involves arranging and analyzing chromosomes from a cell in numerical and size order. It provides a visual snapshot of an individual's genome, helping to detect numerical and structural chromosome abnormalities.

### Techniques in cytogenetics

**Fluorescence *In Situ* Hybridization (FISH):** FISH is a powerful cytogenetic technique that uses fluorescent probes to bind specific DNA sequences on chromosomes. It is widely used to detect chromosomal abnormalities, study gene amplifications, and identify translocations in cancer cells.

**Comparative Genomic Hybridization (CGH):** CGH is a technique that enables the comparison of DNA from two different sources, often normal and tumor tissues. It helps identify DNA copy number changes associated with genetic disorders and cancer.

**Array Comparative Genomic Hybridization (ACGH):** ACGH is an advanced version of CGH that uses microarrays to analyze DNA copy number variations across the entire genome with high resolution.

**Fluorescence-Activated Cell Sorting (FACS):** FACS is a technique used to sort and isolate cells based on their fluorescence. It is

useful in studying specific cell populations and isolating chromosomes for further analysis.

### Applications of cytogenetics

**Cancer cytogenetics:** Cytogenetics has revolutionized cancer research and treatment. By identifying chromosomal alterations in cancer cells, it helps classify tumors, predict outcomes, and develop targeted therapies. For example, the Philadelphia chromosome in Chronic Myeloid Leukemia (CML) led to the development of the drug imatinib.

**Prenatal diagnosis:** Cytogenetics plays a crucial role in prenatal diagnosis, identifying chromosomal abnormalities like down syndrome, Turner syndrome, and trisomy 18 through techniques like amniocentesis and Chorionic Villus Sampling (CVS).

**Genetic counseling:** Cytogenetic analysis provides essential information for genetic counseling, helping individuals and families understand their genetic risks and make informed decisions about family planning.

**Developmental disorders:** Studying chromosomal abnormalities can elucidate the genetic basis of developmental disorders, such as autism and intellectual disabilities, enabling early intervention and support.

### Future directions in cytogenetics

**Precision medicine:** Cytogenetics is poised to play a pivotal role in the era of precision medicine by identifying specific genetic alterations that can inform targeted therapies for individuals with genetic disorders and cancer.

**Non-Invasive Prenatal Testing (NIPT):** NIPT, based on the analysis of fetal DNA in maternal blood, is becoming increasingly important in prenatal diagnosis, offering a safer and less invasive alternative to traditional methods.

**Functional cytogenetics:** Functional cytogenetics aims to understand the functional consequences of chromosomal alterations, going beyond identification to elucidate the mechanisms underlying diseases.

Cytogenetics is an indispensable field that continues to unlock the mysteries of the genetic code. From its role in diagnosing

**Correspondence to:** Carrillo Pratt, Department of Human Genetics, University of Saimaa, South Karelia, Finland, E-mail:carrillo@gmail.com

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diseases to guiding treatment decisions and advancing our understanding of genetics, cytogenetics stands as a beacon of progress in medicine. As technology evolves and our knowledge

deepens, cytogenetics promises to play an increasingly vital role in personalized medicine, ensuring that healthcare is tailored to the unique genetic makeup of each individual.