



Chromosomal Abnormalities and Cytogenetics in Human Cancer

Noah Bennett*

Department of Cytogenetics, Institute of Biomedical Sciences, University of Copenhagen, Copenhagen, Denmark

DESCRIPTION

Cytogenetics is the branch of genetics that focuses on the study of chromosomes, their structure, function, behavior, and abnormalities within cells. It combines principles of cytology and genetics to investigate chromosomal alterations associated with inherited disorders, developmental abnormalities, and various human diseases, particularly cancer. Chromosomes contain the genetic information required for normal cellular function, and their integrity is essential for maintaining genomic stability. Structural or numerical abnormalities in chromosomes can disrupt gene expression, activate oncogenes, inactivate tumor suppressor genes, and contribute to malignant transformation. Consequently, cytogenetics has become an indispensable discipline in cancer biology, clinical diagnostics, and precision medicine by providing valuable insights into the genetic basis of disease and guiding therapeutic decision-making.

Human cells normally contain forty-six chromosomes organized into twenty-three pairs, including twenty-two pairs of autosomes and one pair of sex chromosomes. During normal cell division, chromosomes are accurately replicated and equally distributed between daughter cells, and errors during Deoxyribonucleic Acid (DNA) replication, chromosome segregation, or DNA repair may result in chromosomal abnormalities. These abnormalities are broadly classified as numerical or structural changes. Numerical abnormalities include aneuploidy and polyploidy, while structural abnormalities encompass deletions, duplications, inversions, translocations, ring chromosomes, and isochromosomes. Such alterations may influence gene dosage, disrupt regulatory regions, or generate fusion genes that contribute to disease development.

Chromosomal abnormalities play a fundamental role in carcinogenesis. Many cancers exhibit characteristic cytogenetic alterations that contribute directly to tumor initiation and progression. Chromosomal translocations may create fusion genes encoding abnormal proteins with oncogenic activity, whereas deletions frequently result in the loss of tumor

suppressor genes. Gene amplification increases the expression of growth-promoting genes, while aneuploidy contributes to genomic instability by altering chromosome number. These abnormalities accumulate during tumor evolution and promote uncontrolled proliferation, resistance to apoptosis, angiogenesis, invasion, and metastasis. The identification of specific chromosomal abnormalities has therefore become an important component of cancer classification and prognostic evaluation.

Several laboratory techniques are employed in cytogenetic analysis. Conventional karyotyping remains a valuable method for examining chromosome number and large structural abnormalities during metaphase. Fluorescence in situ hybridization enables the detection of specific DNA sequences using fluorescent probes and provides greater sensitivity for identifying chromosomal rearrangements, deletions, and gene amplifications. Comparative genomic hybridization and chromosomal microarray analysis allow genome-wide evaluation of copy number variations with higher resolution than conventional cytogenetics. More recently, next-generation sequencing has complemented cytogenetic approaches by detecting chromosomal breakpoints and complex genomic rearrangements with exceptional precision. Together, these technologies provide comprehensive information regarding chromosomal abnormalities in both inherited and acquired diseases.

In conclusion, cytogenetics provides essential insights into chromosome structure and function while serving as a cornerstone of cancer diagnosis, genetic research, and precision medicine. The identification of chromosomal abnormalities has significantly improved the understanding of carcinogenesis and other genetic disorders by revealing the molecular alterations responsible for disease development. Continued advances in cytogenetic technologies, combined with genomic and molecular approaches, will further enhance diagnostic accuracy, therapeutic decision-making, and personalized patient care, ultimately contributing to improved clinical outcomes and a deeper understanding of human disease.

Correspondence to: Noah Bennett, Department of Cytogenetics, Institute of Biomedical Sciences, University of Copenhagen, Copenhagen, Denmark, E-mail: noah.bennett@university.edu

Received: 22-Apr-2026, Manuscript No. JCM-26-32083; **Editor Assigned:** 24-Apr-2026, Pre QC No. JCM-26-32083 (PQ); **Reviewed:** 08-May-2026, QC No. JCM-26-32083; **Revised:** 15-May-2026, Manuscript No. JCM-26-32083 (R); **Published:** 22-May-2026, DOI: 10.35248/2157-2518.26. S59.005

Citation: Bennett N (2026). Chromosomal Abnormalities and Cytogenetics in Human Cancer. S59:005.

Copyright: ©2026 Bennett N. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.