Brief Report
Volume 15:04, 2024

Journal of Cytology & Histology

ISSN: 2157-7099 Open Access

Molecular Cytogenetics: Bridging the Gap Between G-banding and Next-generation Sequencing

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Introduction

G-banding, or Giemsa banding, has been a fundamental technique in cytogenetics since its introduction in the 1970s. This method involves staining chromosomes with Giemsa dye, producing a distinct banding pattern that is unique to each chromosome. The bands correspond to regions of DNA that are rich or poor in Adenine-Thymine (AT) base pairs, allowing for the identification of chromosomal abnormalities such as aneuploidies, translocations, deletions and duplications [1].

Description

Advantages of g-banding:

- **Resolution:** G-banding can detect large-scale chromosomal changes (greater than 5-10 Mb).
- **Visualization:** It provides a clear visual representation of the entire karyotype.
- **Accessibility:** The technique is relatively simple and cost-effective, making it widely accessible in clinical laboratories.

Limitations of g-banding:

- **Resolution:** It lacks the resolution to detect small genetic changes, such as microdeletions or point mutations.
- **Interpretation:** The interpretation of G-banded karyotypes requires significant expertise and experience.
- **Preparation:** High-quality metaphase spreads are necessary, which can be challenging to obtain from some cell types.

The advent of molecular cytogenetics

The limitations of G-banding led to the development of molecular cytogenetic techniques that offer higher resolution and specificity. Fluorescence *In Situ* Hybridization (FISH) was one of the first molecular methods to enhance traditional cytogenetics [2].

Fluorescence *In Situ* Hybridization (FISH): FISH involves the use of fluorescently labeled DNA probes that bind to specific DNA sequences on chromosomes. This allows for the detection of specific genetic abnormalities at a higher resolution than G-banding [3].

Advantages of FISH:

- **Targeted Analysis:** FISH can identify specific genetic changes, such as translocations, amplifications and deletions, with high sensitivity.
- **Resolution:** It offers better resolution than G-banding, detecting changes as small as a few kilobases.
- **Versatility:** FISH can be applied to interphase nuclei, making it useful for analyzing non-dividing cells.

Limitations of FISH:

- **Target specificity:** FISH requires prior knowledge of the genetic region of interest, limiting its use in genome-wide screening.
- **Probe design:** The design and validation of FISH probes can be time-consuming and costly.
- **Quantification:** While FISH is qualitative, it is not inherently quantitative.

Next-Generation Sequencing (NGS) in cytogenetics

Next-Generation Sequencing (NGS) represents a paradigm shift in molecular cytogenetics, offering comprehensive and high-throughput analysis of the genome. NGS technologies enable the sequencing of millions of DNA fragments simultaneously, providing detailed insights into genetic variations, including Single Nucleotide Polymorphisms (SNPs), insertions, deletions, Copy Number Variations (CNVs) and structural rearrangements [4].

Advantages of NGS

Resolution: NGS can detect genetic changes at single-nucleotide resolution.

Comprehensive analysis: It allows for genome-wide analysis without prior knowledge of specific regions.

Quantitative: NGS provides quantitative data, enabling precise measurement of genetic variations.

Scalability: High-throughput sequencing facilitates large-scale studies and population genetics.

Limitations of NGS

Cost: Despite decreasing costs, NGS remains more expensive than traditional cytogenetic methods.

Data interpretation: The vast amount of data generated requires sophisticated bioinformatics tools and expertise for analysis and interpretation.

Turnaround time: While improving, the turnaround time for NGS can be longer than for FISH or G-banding, particularly for whole-genome sequencing.

Bridging the Gap: Integrating G-Banding, FISH and NGS

Modern cytogenetic laboratories increasingly adopt an integrative approach, combining G-banding, FISH and NGS to leverage the strengths of each method while compensating for their limitations. This multi-modal strategy provides a comprehensive assessment of genetic abnormalities [5].

Clinical applications:

Cancer diagnostics: Integrative cytogenetics is crucial in oncology for identifying chromosomal aberrations associated with specific cancers, guiding treatment decisions and monitoring disease progression.

Prenatal testing: Combining G-banding and NGS enhances the detection

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Received: 01 July, 2024, Manuscript No. jch-24-143567; **Editor Assigned:** 03 July, 2024, PreQC No. P-143567; **Reviewed:** 15 July, 2024, QC No. Q-143567; **Revised:** 22 July, 2024, Manuscript No. R-143567; **Published:** 29 July, 2024, DOI: 10.37421/2157-7099.2024.15.755

of chromosomal abnormalities in prenatal samples, providing more accurate risk assessments for genetic disorders.

Genetic research: The integration of these techniques accelerates the discovery of novel genetic variants and their associations with diseases, contributing to personalized medicine.

Case studies

Chronic Myeloid Leukemia (CML): In CML, the Philadelphia chromosome (t(9;22)(q34;q11)) is a hallmark genetic abnormality. G-banding first identified this translocation, but FISH and NGS have since provided more detailed characterizations. FISH confirms the presence of the BCR-ABL1 fusion gene, while NGS detects additional mutations that may influence treatment response.

Prenatal diagnosis of down syndrome: Traditional G-banding detects the extra copy of chromosome 21 in Down syndrome. FISH can rapidly confirm trisomy 21 in interphase cells and NGS-based Non-Invasive Prenatal Testing (NIPT) offers highly accurate screening using maternal blood samples, detecting trisomies with high sensitivity and specificity.

The field of molecular cytogenetics is continually evolving, with advancements in technology and bioinformatics driving new applications. Emerging techniques such as single-cell sequencing and CRISPR-based diagnostics hold promise for further enhancing the resolution and specificity of cytogenetic analyses. Additionally, the integration of Artificial Intelligence (AI) and Machine Learning (ML) in data interpretation will streamline the analysis of complex genetic data, making advanced cytogenetic diagnostics more accessible and accurate.

Conclusion

The transition from G-banding to next-generation sequencing represents a significant advancement in cytogenetics, offering unparalleled resolution and comprehensive analysis of genetic variations. By integrating traditional and molecular techniques, modern cytogenetics provides a powerful toolkit for diagnosing genetic disorders, guiding treatment decisions and advancing genetic research. As technology continues to evolve, the future of molecular cytogenetics promises even greater precision and insights into the complexities of the human genome.

Acknowledgement

None.

Conflict of Interest

There are no conflicts of interest by author.

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How to cite this article: Caleb, Ezekiel. "Molecular Cytogenetics: Bridging the Gap Between G-banding and Next-generation Sequencing." *J Cytol Histol* 15 (2024): 755.