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LABORATORY DIAGNOSIS OF CHROMOSOMAL DISORDERS

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Abstract

Chromosomal disorders are genetic conditions caused by numerical or structural abnormalities in chromosomes. They represent a significant cause of congenital anomalies, developmental delay, intellectual disability, infertility, and pregnancy loss. Accurate laboratory diagnosis plays a crucial role in early detection, clinical management, genetic counseling, and prevention strategies. This article reviews the main laboratory methods used in the diagnosis of chromosomal disorders, including conventional cytogenetics, molecular cytogenetic techniques, and advanced genomic technologies.

Keywords

chromosomal disorders, karyotyping, FISH, CGH, prenatal diagnosis, genetic testing

Introduction

Chromosomes are structures within the cell nucleus that contain genetic material in the form of DNA. Humans normally possess 46 chromosomes, arranged in 23 pairs. Any deviation in chromosome number or structure can result in chromosomal disorders. Common examples include Down syndrome (trisomy 21), Turner syndrome (45,X), and Klinefelter syndrome (47,XXY).

Laboratory diagnostics is essential for identifying chromosomal abnormalities, confirming clinical suspicion, and guiding patient management. Advances in genetic technologies have significantly improved the accuracy and sensitivity of chromosomal analysis.[1,2,3]

Classification of Chromosomal Disorders

Chromosomal disorders are broadly classified into two categories:

1. Numerical Abnormalities

These occur due to gain or loss of entire chromosomes:

Aneuploidy (e.g., trisomy 21, trisomy 18, monosomy X)

Polyploidy (rare in live births)[4,5,6,7,8]

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2. Structural Abnormalities

These involve rearrangements within or between chromosomes:

Deletions

Duplications

Inversions

Translocations (reciprocal and Robertsonian)

Ring chromosomes[9,10,11,12,13,14]

Laboratory Diagnostic Methods

1. Conventional Karyotyping

Karyotyping is the classical cytogenetic method used to visualize chromosomes under a light microscope after Giemsa staining (G-banding).

Procedure:

Cell culture (usually lymphocytes, amniocytes, or chorionic villi)

Mitotic arrest

Chromosome staining and microscopic analysis[15,16,17,18,19]

Advantages:

Detects numerical and large structural abnormalities

Widely available and cost-effective

Limitations:

Low resolution (cannot detect microdeletions)

Time-consuming (cell culture required)

2. Fluorescence In Situ Hybridization (FISH)

FISH uses fluorescently labeled DNA probes that bind to specific chromosome regions.

Applications:

Detection of microdeletions (e.g., DiGeorge syndrome)

Rapid prenatal diagnosis of common aneuploidies

Identification of translocations

Advantages:

High specificity

Can be performed on interphase cells

Faster than karyotyping

Limitations:

Targeted technique (requires prior suspicion)

Cannot analyze the whole genome[20,21,22,23,24]

3. Cmparative Genomic Hybridization (CGH) and Array-CGH

Array-CGH compares patient DNA with normal reference DNA to detect copy number variations (CNVs).

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Advantages:

High resolution

Detects submicroscopic deletions and duplications

Genome-wide analysis[25,26,27,28,29]

Limitations:

Cannot detect balanced translocations or inversions

Higher cost

4. Quantitative Fluorescent PCR (QF-PCR)

QF-PCR is a molecular method used mainly for rapid prenatal screening.

Applications:

Detection of trisomy 21, 18, 13, and sex chromosome abnormalities

Advantages:

Rapid results (24–48 hours)

High sensitivity

Limitations:

Limited to common aneuploidies

Not suitable for structural abnormalities[30,31]

5. Next-Generation Sequencing (NGS)

NGS-based techniques are increasingly used in prenatal and postnatal diagnostics.

Applications:

Detection of copy number changes

Identification of complex chromosomal rearrangements

Advantages:

Very high resolution

Comprehensive genomic analysis

Limitations:

High cost

Complex data interpretation

Prenatal and Postnatal Diagnosis

Laboratory diagnosis can be performed at different stages:

Prenatal: amniocentesis, chorionic villus sampling, non-invasive prenatal testing (NIPT)

Postnatal: peripheral blood analysis in newborns or children with developmental anomalies

Early diagnosis allows informed decision-making, appropriate treatment planning, and genetic counseling for families.[33,34]

Clinical Significance and Genetic Counseling

Accurate laboratory diagnosis of chromosomal disorders is essential for:

Confirming clinical diagnosis

Assessing prognosis

Providing reproductive counseling

Preventing recurrence in future pregnancies

Genetic counseling should always accompany laboratory testing to explain results and their implications.[35,36,37]

Conclusion

Laboratory diagnostics of chromosomal disorders has evolved from traditional karyotyping to advanced molecular and genomic techniques. Each method has specific advantages and limitations, and often a combination of techniques is required for accurate diagnosis. Continuous technological advancements are improving early detection, diagnostic accuracy, and patient care in the field of medical genetics.

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