

Chapter

Cytogenetics and Microdeletion Syndromes

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Abstract

The emergence of molecular cytogenetic technologies has transformed the process of identifying new microdeletion syndromes. Traditional methods of identifying these syndromes relied on the chance discovery of patients exhibiting specific clinical features and visible chromosomal rearrangements through techniques like G-banding. However, with the introduction of comparative genomic hybridization using microarrays, it has become possible to detect previously unknown and recurring imbalances in individuals presenting with conditions such as mental retardation and nonspecific features. In contrast to the conventional "phenotype-first" approach in cytogenetics, array-based comparative genomic hybridization allows for a "genotype-first" approach. This means that the focus is on identifying genetic variations or imbalances in the genome before associating them with specific clinical phenotypes. By facilitating the identification of novel genomic disorders and recurrent imbalances, this technology has revolutionized the field of cytogenetics and deepened our understanding of the genetic underpinning of microdeletion syndromes. The present chapter emphasizes the role of cytogenetics technologies in delineating the aetiology of microdeletion syndromes.

Keywords: cytogenetics, microdeletion syndromes, fluorescent *in situ* hybridization, comparative genomic hybridization, chromosomal microarray

1. Introduction

Cytogenetic analysis plays an important role in delineating the aetiology of numerous suspected chromosomal disorders. Cytogenetics examines the connection between chromosomal abnormalities and genetic disorders in humans [1]. Microdeletion syndromes are a group of disorders caused by the deletion of small chromosomal segments (< 5 Mb in size) that contain multiple genes, each potentially contributing independently to the disease phenotype. These disorders usually arise from the haploinsufficiency of critical genes in the affected region [2].

Microdeletion syndromes often occur when a chromosomal segment is lost, resulting in a deficiency of a small number of crucial genes or, in some instances, a

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