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**Report:****Clinical cytogenetics and molecular cytogenetics***LI Marilyn¹, PINKEL Daniel²*(¹Medical School, Tulane University, New Orleans, LA 70112-2699, USA)**(²Laboratory of Medicine, University of California, San Francisco, California 94143, USA)*E-mail: mli2@tulane.edu; pinkel@cc.ucsf.edu

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Abstract: The short report will be focused on helping our students to understand commonly used conventional and cutting edge cytogenetic techniques and their clinical applications, the advances and drawbacks of each technique, and how to pick the right test(s) for a specific patient in order to achieve a proper diagnosis efficiently and economically.

Key words: Cytogenetics, Chromosome, FISH, Microarray, Comparative genomic hybridization (CGH)

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Chromosomes are the vehicles of inheritance that reside in the cell nucleus. Clinical cytogenetics is the study of the relationship of chromosomal alterations and genetic diseases in human beings. Molecular cytogenetics is a study of genetic disorders using new technologies that combine cytogenetic and molecular techniques (Speicher and Carter, 2005). This lecture covers basic cytogenetic concepts and techniques, mechanism of chromosomal alterations, chromosomal nomenclature, common cytogenetic disorders, diagnosis of chromosomal disorders, genetic counseling, introduction of molecular cytogenetics, and commonly used FISH techniques and their clinical significance (Li, 2005). The goal of this lecture is to help our students to understand commonly used conventional and cutting edge cytogenetic techniques and their clinical applications, advances and drawbacks of each technique, and how to pick the right test(s) for a specific patient in order to achieve a proper diagnosis efficiently and economically (Li *et al.*, 2000; Pinkel and Albertson, 2005).

Conventional cytogenetics has a very powerful ability to scan the genome for aberrations that involve

both gains and losses of portions of the genome, as well as rearrangements within and among chromosomes. However, the staining patterns produced on the chromosomes by banding procedures are sometimes ambiguous, and the resolution is limited by the optical characteristics of microscopes and the complex manner in which DNA is packaged into chromosomes. In some cases analysis can be improved by using FISH with probes for specific genetic loci, such as telomeres or sites of known gain or loss of sequence. However the complexity of the staining pattern that can be produced with FISH is limited by the number of FISH probes that can be distinguished, and the same optical and chromosome structure considerations that affect chromosome banding. Aberrations that involve gain or loss of chromosome segments are particularly important in medical cytogenetics. Comparative genomic hybridization (CGH) allows screening the entire genome for these aberrations. Using microarrays, the resolution of CGH can be much greater than with standard cytogenetics. Microarrays with dense coverage of the entire genome allow screening for aberrations at any location without the need to have prior knowledge about where to test. Thus array CGH offers the ability to screen genomes of affected individuals to discover new aberrations, determine if apparently balanced transloca-

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tions also involve gains or losses of DNA, establish phenotype-genotype relationships for conditions with variable aberrations, and provide comprehensive clinical diagnostic information. In combination with techniques such as chromosome microdissection or chromosome sorting, array CGH can map translocation breakpoints with high precision.

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