

# Basic genetics for cardiologists: cytogenetic approach

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Interest in cytogenetics may be traced to the development of the chromosomal theory of inheritance that emerged from efforts to provide the basis for Darwin's theory "On the origin of species by means of natural selection." Despite their fundamental place in biology, chromosomes and genetics had little impact on medical practice until the 1960s. The discovery that a chromosomal defect caused Down syndrome was the spark responsible for the emergence of medical genetics as a clinical discipline. Prenatal diagnosis of trisomies, biochemical disorders, and neural tube defects became possible and hence the proliferation of genetic counseling clinics. In a clinical setting, chromosome abnormalities account for a large proportion of cases involving individuals referred with congenital malformations, developmental delay, mental retardation, or infertility; women with gonadal dysgenesis; spontaneous abortuses, and couples with repeated spontaneous miscarriages. Additionally, the field of cytogenetics is important in the diagnosis and workup of patients with hematologic/oncologic disorders. Cytogenetic testing in this area is used for diagnosis, classification of disease, determining treatment regimens, and to monitor disease status and recovery.

Cytogenetic analysis is highly informative in the majority of cases,

although interpretation of the findings may be difficult if the karyotype is complex or multiple clones are present. In spite of continual technical developments in methodology, some samples fail to produce a successful result because of a low mitotic index or poor chromosome morphology. The growth of techniques in molecular cytogenetics, or fluorescence in situ hybridization (FISH), has overcome some of these limitations. FISH has produced major improvements in the sensitivity, accuracy and reliability of cytogenetic analysis for both research and routine applications. The introduction of interphase-FISH has made one of the most important contributions, allowing large numbers of non-dividing cells to be screened. Analysis is no longer restricted to cytogenetic cell suspensions. The increasing availability of commercial probes means that FISH has become accessible to most routine cytogenetics laboratories. Molecular cytogenetics has become an integral part of the discipline of cytogenetics and is indispensable in accurate chromosomal analysis of leukemias.

In conclusion, chromosome research has been behind the major advances in the field of genetics, and it will continue to be the key to future progress.