# Why Your Laboratory Should Offer These 3 Cytogenic Tests

*By Austin McGarrett*

Over the past several years, the study of genetics, as well as the application of genetic tools, has evolved beyond academia and research facilities. Cytogenetic tests are increasingly being offered by medical laboratories as medical lab directors and technicians become more aware and more comfortable with performing and analyzing cytogenetic tests. The market for cytogenetic testing has grown steadily over the past several years; at $8.5 billion in 2017, the market is expected to increase to $19.1 billion by 2024, at a Compound Annual Growth Rate (CAGR) of 12.2%.
Since the inception of cytogenetic testing, there has been an increase in the number and types of genetic tests available. As a result, medical laboratories trying to branch into the field of cytogenetic testing may be at a loss of how and where to begin. This post discusses the three primary cytogenetic tests that should be offered by any laboratory wishing to offer cytogenetic testing; it also addresses how medical laboratories can benefit from offering cytogenetic tests.

# 1) CHROMOSOME ANALYSIS

Also known as karyotype analysis or simply karyotyping, chromosome analysis is the study of the chromosome for any abnormalities. The typical person has twenty-three pairs of chromosomes or forty-six in all; karyotyping analyzes these chromosomes for their structural integrity as well as their number. Traditionally, blood or amniotic fluid is the specimen type used for karyotyping. Some examples of genetic disorders that can be detected by chromosome analysis are Down's syndrome (Trisomy 21), Edward's syndrome (Trisomy 18), and Patau syndrome (Trisomy 13). In addition, chromosome analysis can also be used to screen for some types of cancers that have a genetic component; some examples of these cancerous genes are BCR-ABL found in individuals with chronic myeloid leukemia (CML) and the BRCA gene which is used to screen for breast cancer.

# 2) FLUORESCENCE IN SITU HYBRIDIZATION (FISH)

FISH is a cytogenetic test that is used to analyze in detail the individual genes within chromosomes. It has a shorter turnaround time compared to karyotyping because it has a shorter processing time. The initial phase of FISH is the development of hybridized probes, either directly or indirectly; once developed, these hybridized probes are used to look for genetic chromosomal mutations. Some examples of disorders that can be detected by FISH are Prader-Willi syndrome, Angelman syndrome, Huntington's disease, and cancer-related genes.

# 3) MICROARRAY COMPARATIVE GENOMIC HYBRIDIZATION (aCGH)

Microarray comparative genomic hybridization is another type of cytogenetic test that is vastly superior to karyotyping or FISH. aCGH is used to detect very minor genetic imbalances such as chromosomal gain or loss which is not readily detected by other traditional cytogenetic tests. Usually, two DNA sources are compared, a test DNA and a source DNA, for any variations in their chromosomal compositions. This test has been used to detect analyze genetic abnormalities in different types of cancer, and other genetic disorders such as Cri du Chat.

# BENEFITS OF CYTOGENETIC TESTING

There are several reasons why medical laboratories should expand their testing capabilities and offer cytogenetic tests:

- **Profitability:** Cytogenetic testing is a highly lucrative field; as stated earlier, the market share for cytogenetic testing is expected to grow at a CAGR of 12.2%. Due to an increased focus in preventative healthcare, an increased incidence of chronic diseases, and an increasingly

elderly population, the demand for cytogenetic testing has markedly risen. As such, medical laboratories offering these tests are highly profitable.

- **Enhanced diagnostic capabilities:** Cytogenetic tests enable laboratories to offer more in-depth analyses into various medical conditions. This allows for more accurate diagnoses and the results enable providers to offer more relevant and precise treatment to their patients. In some instances, enhanced diagnostic capabilities can also be used to head off some medical conditions before the symptoms appear.
- **Future growth:** Cytogenetic testing is the future of laboratory medicine. As more genetic advances are made and the costs of cytogenetic tests become cheaper, providers will increasing request cytogenetic tests as part of the management of their patients. Laboratories that do not offer cytogenetic testing will eventually experience a decreased demand for their services which may lead to their ultimate closure if there are no remedial attempts made. Therefore, to prevent the risk of obsolescence, laboratories should expand their services to include cytogenetic testing sooner rather than later.

At Psychē, we know all about cytogenetic testing. Our Molecular Information Management System (MIMS), nucleoLIS Ē.finity, is specially designed to manage and support the workflow of molecular and genetic testing. For more information about our services, contact us.