



Managing Data from Cytogenetic and Molecular Studies

Laboratory information systems must harness the power of microarrays

By Chris Callahan, Vice President of Product Management, Sunquest Information Systems

New technologies are revolutionizing the cytogenetic and molecular arenas. High-throughput microarray technology, for example array-based comparative genomic hybridization, can perform the equivalent of hundreds or even thousands of fluorescence in situ hybridization (FISH) experiments in one array. This technology has become the preferred method for molecular cytogenetic research, and recent advances in the technology are making the system appropriate for use by clinical pathology labs.

At present, most anatomic pathology laboratory information systems (AP LISs) are not capable of providing results that allow pathologists to perform the necessary queries and outcome analyses. To embrace the advancements in cytogenetic and molecular testing fully, the AP LIS functionality must be enhanced, so it can be the de facto manager of this critical information.

Laboratories looking for this enhanced functionality are turning to Sunquest CoPathPlus™. Users are empowered to define protocols, workflows, and reports tailored to each laboratory's standard operating procedures for cytogenetics. Further, integration between clinical and anatomic systems enables ongoing use of their existing systems, including instrument interfaces, to report out the discrete data. Pathologists can use these comprehensive data sets to provide detailed interpretations to the caregiver, improving patient treatment.

Genetic testing poised to assume a larger role in pathology

Cytogenetics is the study of the structure and function of the cell, particularly the chromosomes. The term encompasses routine analysis of G-Banded chromosomes and other cytogenetic banding techniques, as well as molecular cytogenetics, including fluorescent in situ hybridization (FISH), and comparative genomic hybridization (CGH) techniques. [1]

In recent years, scientists have moved towards the use of microarrays, a laboratory technology used to study if a large number of genes are switched on or off in a given tissue sample. A microarray is also called a DNA chip or a gene chip and is the technology used to obtain a genomic profile. A microarray consists of different nucleic acid probes that are chemically attached to a substrate, which can be a microchip, a glass slide, or a microsphere-sized bead.

New applications of technologies such as microarrays are extremely useful for diagnostic, prognostic, or therapeutic purposes in individual patients. DNA microarrays make it possible to analyze the mRNA expression of thousands of genes simultaneously. [2]

Molecular and cytogenetic testing has been important in clinical pathology for a number of years, since results from these procedures can greatly enhance the quality of a pathologist's rendered diagnosis. With the development of new testing technologies for cytogenetics, the pathologist has more information available to provide significant



“meaningful data” when rendering a diagnosis, consequently delivering detailed interpretations to the caregiver, which will assist in the treatment of the patient.

Automation, decrease in test kit costs, and advanced training for this type of testing has enabled many laboratories to add cytogenetics to their routine test catalogue. Automation and other advancements are beginning to drive down the cost of molecular and cytogenetic testing, which will allow medium to smaller laboratories to begin using these procedures more frequently.

According to a June 2009 article in Genetic Engineering News, “In 2008, the U.S. array-based molecular cytogenetic market was estimated at \$41.7 million; approximately one-third of the total U.S. molecular cytogenetic market.” Some industry analysts estimate that by 2015, use of microarrays will catch up to FISH-based technology and represent fully 50 percent of the market. It is clear that genetic testing of all kinds will become an ever-larger piece of the pathology laboratory business.

Laboratory software must keep pace with advances

One of the issues that arise from the increase in molecular and cytogenetic testing is the management of data received so that it can be manipulated, searched, and analyzed. In many systems, laboratories can share the complex information produced by anatomic pathology, cytology, and cytogenetic departments, but the system enters the data as unstructured text, making it difficult to manage and analyze the data.

Thus, any data manipulation must be carried out using the performing instrument’s native software. This problem is further exacerbated if the testing is handled through a third party; in this instance, the laboratory does not have any direct access to the discrete data.

Pathology groups that want to stay on the leading edge of laboratory medicine will need their AP LIS to capture and manage the data in a more meaningful way. Many Sunquest clients are establishing cytogenetic laboratories as part of their key business structure and using Sunquest CoPathPlus integrated with Sunquest Laboratory™ to process samples, generate results, and report these results.

With this solution, the user can define specific protocols, workflows, and reports tailored to each unique laboratory and its standard operating procedures for cytogenetics. Integration between the clinical and anatomic systems allows the laboratory to use its existing systems, including instrument interfaces, to report out the discrete data necessary when performing this testing.

This solution has been adopted by laboratories around the country. These include Nationwide Children’s Hospital in Columbus, OH; Aurora Consolidated Laboratories in West Allis, WI; and, Carolinas Health System in Charlotte, NC.

The user can add such information as volumes, mitosis, media, and slide qualities to the cytogenetic result and can define calculations, for example, averages and ratios. The user can also determine which data transmitted from the instruments will become part of the patient report. “Quick text” values can be defined to provide standardization of results. Images such as karyotype can be easily added to the case and patient report.



Whether cytogenetic testing is a component of a complex anatomic pathology case or a single cytogenetic or molecular test, the user orders these specific procedures. The test is performed and results generated. The user receives the report and inputs the test results into the CoPathPlus Procedure. The procedure is forwarded to the assigned pathologist who reviews the procedure results and renders an interpretation if needed. At this time, the pathologist can electronically sign out this testing.

If the testing is not performed in the anatomic pathology laboratory, the site can electronically transmit the order to a reference lab or another laboratory within their institution using the reference lab interface.

Innovative sample tracking capabilities from Sunquest streamline workflow technological advancements. These include the capability to define specific workflows for each of the testing methodologies and route the specimens to the appropriate laboratory or working bench. As the specimen is processed, it will be tracked so that the users can determine the status of the specimen and project turnaround times for completion.

As the testing volumes increase, new instrumentation will be developed and Sunquest will connect to these instruments and update the patient case or record with these discrete values. This will provide the end user the ability to perform searches, outcomes analysis, and correlations studies to provide trending information to the caregiver. Patient reports will be improved to allow the end user to highlight, selectively, the pertinent information so that the caregiver can better manage their patient's therapies.

One hospital that has significantly expanded its cytogenetic and molecular reference lab business in recent years is Nationwide Children's Hospital of Columbus, Ohio. The hospital reports an increase in cytogenetic cases to approximately 5,000 per year, with an additional 2,500 molecular cases per year.

According to Bhaskar AV, Sunquest Vice President of Product Development, "We are seeing a great increase in cytogenetic and molecular testing. Pathologists increasingly turn to this technology to provide meaningful data for diagnosis and treatment.

"Sunquest is committed to high performing laboratory systems for advanced cyto and molecular testing. Key to our success is a robust laboratory information system that allows us to integrate our clinical and anatomic systems."



[1] Stedman's Medical Dictionary (28th Ed.). (2006). Baltimore, MD: Lippincott Williams.

[2] High-throughput microarray technologies: from genomics to clinics. Bubendorf L., Institute for Pathology, University of Basel, Switzerland. lbubendo@bluewin.ch, Retrieved April 26, 2010, PMID: 11528203 [PubMed - indexed for MEDLINE]