What molecular diagnostics laboratory systems offer

December 2016—As personalized and predictive medicine have progressed from not-in-my-lifetime to now available, health care information technology vendors have faced the challenge of how to manage a mass of molecular data and direct molecular testing processes. CAP TODAY asked vendors of molecular diagnostics lab information systems to explain what their products contribute to the flourishing field of molecular testing. Here are their responses.

Callahan

Chris Callahan, general manager of GeneInsight, a Sunquest company: UniConnect LC, which was recently acquired by Sunquest Information Systems (see "Newsbytes," page 60), designed the Precision Molecular Diagnostics, or pMDx, laboratory information management system platform to meet exacting molecular diagnostics laboratory requirements for complex data handling and deep process management.

The recent integration of UniConnect's pMDx solution with Sunquest's GeneInsight genetics information system provides a molecular and genetics platform that manages workflow from instrument to electronic medical record. PMDx offers the following.

- Integrated molecular wet lab and dry lab functionality via pMDx and GeneInsight.
- Elimination of spreadsheets and paper with workflow automation from accessioning through extraction, quantitation, polymerase chain reaction, analysis, and final report.
- Installation as a standalone LIMS platform or as software that augments existing clinical lab information systems with rigorous molecular functionality. It is also available via HIPAA-compliant cloud hosting, which provides secure Web portal access for external users, including ordering physicians, partner laboratories, sales teams, and billing partners.
- Billing interface with multiple third-party systems, including Quadax, Premier Source, and Xifin.
- Scalability to meet business growth.

- Graphical dashboards that present real-time information display of desired data sets, tracking every aspect of the science and business, and that are accessible only through adherence to stringent user-access guidelines.
- Custom applications for next-generation sequencing, pharmacogenomics, companion diagnostics, molecular toxicology, oncology, virology, infectious disease, and cytogenetics—commercial or laboratory-developed tests.
- Biobank sample tracking with precise sample location and dynamic visual search.
- Compliance support for industry standards, guidance, and regulations.
- Functionality to track and qualify reagents (including new lot validation, as required per the CAP), instruments, other consumables, and personnel.

Fehrs-Battey

Amber Fehrs-Battey, senior product specialist, Sunquest Information Systems: Sunquest Information Systems offers solutions to help clients maximize the efficiency and quality of their esoteric laboratory testing, in part through complex workflow management, data analysis, inventory management, and data sharing. The solutions also manage polymerase chain reactions, sequencing, karyotyping, DNA and RNA extraction, chromosomal microarrays, and next-generation sequencing.

Sunquest's molecular and genetics solution, which includes the GeneInsight, UniConnect, and Sunquest Molecular suite of products, addresses many types of workflows, including those for molecular genetics, molecular microbiology, cytogenetics, human leukocyte antigens, and anatomic pathology. The products can track samples and containers. They also manage inventory and interface with instruments and robotics, as well as manage the millions of data points associated with complex NGS and microarray variants.

For health care entities that want to bring testing in-house, Sunquest's bioinformatics staff provide expert services, including complex secondary and tertiary validations.

Furthermore, Sunquest provides several out-of-the-box tests and system features, such as single and multiplexed PCR, generic Sanger sequencing, NGS, and microarray protocols, which speed implementation and support customization.

GeneInsight, a Sunquest company, manages the analysis of complex genetic data and has been doing so for more

than a decade within CLIA-accredited laboratories. GeneInsight has supported the interpretation and reporting workflow for more than 50,000 clinical reports. The platform integrates with Sunquest's molecular laboratory solutions to pull data from next-generation sequencers and chromosomal microarray instruments, their accompanying software, or bioinformatics systems. From there, complex genomes are processed using filtration, annotation, analysis, reporting, and clinician tools.

In addition, GeneInsight participates in variant data-sharing working groups to establish new standards for genetic data throughout the medical community. Among the large national data-sharing initiatives that use the company's systems are the Canadian Open Genetics Repository and eMERGE (Electronic Medical Records and Genomics) Network.

Hakim

Gilbert Hakim, founder and CEO, SCC Soft Computer: SCC's Genetics Information Systems Suite, a single platform utilizing a single database, allows labs to fully incorporate and automate the molecular/genetics/NGS testing process.

The software suite, which includes SoftMolecular, is a workflow solution for the diagnostic molecular genetics laboratory. It allows geneticists and pathologists to create customized protocols for managing testing workflows, derive data and images from automated instruments via interfaces, auto-populate report text based on result-driven reporting rules, and easily access patient and family medical and testing histories.

The software provides customizable next-generation sequencing functionality. Customizable requisitions can capture test-specific information, allowing users to build and store family pedigrees in the system. The product also easily handles aliquoting and extraction processes, whether automated or performed manually.

Additional functionality provided via SoftMolecular includes:

- support for clinical and research applications of NGS, including targeted panels, whole exome sequencing, and whole genome sequencing, as well as for other common molecular technologies.
- easy categorization of the reportable variants and versioning into relevant databases to facilitate variant entry, management, and interpretation.
- direct interfaces to molecular instrumentation for uploading or downloading data.
- a Web-based orders and results reporting portal.
- functionality to manage patient and family cases, track sample storage, and control inventory.
- the ability to combine different types of result values on one graph for comparison purposes or provide multiple graphs for a single test.
- features to combine patient results with the information in the software's

gene master table and variant/mutation table.

- a task list that supports PCR, qPCR, arrays, multiplex ligation-dependent probe amplification, fragment analysis, and various sequencing tests.
- Sanger sequencing for reflex testing.
- support for lab interfacing and communication with other laboratories and health agencies.
- the ability to record all levels of results review and provide customizable reports and labels.
- automated billing.
- automatic linkage to external databases to perform inquiries regarding variant significance and to track variant, gene-classification, and disease-association information in the system.

Clifford

Lisa-Jean Clifford, CEO, Psyche Systems: NucleoLIS is a molecular information management system designed to streamline the complexities of molecular testing. The software, which was built in collaboration with Psyche's molecular and cytogenetic customers, is a fully automated solution for DNA sequencing, FISH, immunology, karyotyping, PCR, and other molecular techniques.

NucleoLIS features consolidated results entry and viewing and can generate report previews via on-demand management reporting tools. Users can quickly enter results for all tests or assay types on one screen. Interpretation of test results, notes, and images can be added and stored as part of the case information. The system also applies automatic logic for range checking and reflex testing and enables report distribution via outreach or secure email.

Other key features of NucleoLIS allow the software to:

- handle all molecular disciplines and workflows.
- integrate with laboratory instrumentation.
- support results delivery directly to the result field and automatically attach images.
- generate specimen tracking and case status steps that provide the location of specimens or cases.
- distribute reports based on case types and physician requests.

Ioan Cucoranu, MD, chief medical officer, Advanced Health Informatics: Molecular Insights is a HIPAA-compliant, cloud-based, cross-platform clinical LIS slated for commercial availability in mid-2017. It is designed to streamline the molecular laboratory workflow by managing every preanalytical, analytical, and postanalytical aspect of the

molecular lab. And because of the system’s intuitive, straightforward user interface, it requires minimal end-user training.

Molecular Insights will be offered as a standalone molecular lab system or as middleware that can be integrated with a clinical or anatomic pathology lab information system or an enterprisewide health information system in a variety of laboratory settings. The system allows significant customization using minimal information technology resources.

Molecular Insights provides workbench access, extensive configurability, integration with automated molecular instruments, longitudinal integration of test results, document management, discrete data collection, and storage and processing automation. It also provides specimen and reagent inventory management with just-in-time ordering, test control data management, support for barcoding, and radio-frequency identification tracking technology.

End-user dashboards encompass test-status notifications and allow communication between users. The system generates a paperless environment with user-defined workflows or reporting templates and provides on-demand, print-out capabilities. A key feature is integrated support for the complex workflow of next-generation sequencing and integration with NGS bioinformatics pipelines.

Support for quality assurance and quality control activities provides real-time identification of issues that need immediate attention. The system also automatically generates clinical reports, including results and interpretations, that can be linked with other areas of the pathology lab, such as hematopathology, surgical pathology, cytology, and microbiology, integrating the results for molecular, cytogenetics, and FISH, as well as flow cytometry data.

Molecular Insights provides support for personnel management, proficiency testing, and training, as well as test validation documentation.

Cloud-based architecture allows for fast implementation of product features using minimal in-house IT resources. Furthermore, the system’s modular design supports a rapid software development cycle.[hr]