variantyx

Case Study

Launching a Hereditary Cancer Panel Test

Customer needs

Premier Medical Laboratory Services (PMLS) is an established CLIA/COLA reference lab with an extensive test menu based on gPCR and other non-NGS technologies. When we first met, they were exploring the possibility of adding NGS capabilities to their lab to support the demand they were seeing from their customers for cancer predisposition testing.

Why cancer predisposition testing? Approximately 5-10% of cancer cases are hereditary in nature. With approximately 1 in 300 individuals within the general population carrying a genetic mutation that confers increased risk of developing cancer, genetic testing for cancer predisposition mutations can help identify susceptible individuals.

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Knowledge of increased risk can set the stage for lifestyle change, early screening and intervention programs that contribute to increased life expectancy. With increasing awareness of the benefits of genetic testing, the demand for hereditary cancer screening has grown significantly.

Experienced in running a clinical testing lab, PMLS's staff had the know how to set up the wet bench operations but realized it would take time to acquire new domain expertise in analysis, interpretation and reporting of hereditary cancer data. They partnered with Variantyx to expand their test menu and bring their new NGS test to market sooner.

Project scope

The partnership focused on pairing PMLS's established wet lab expertise with Variantyx's bioinformatic, automation and clinical interpretation and reporting expertise. The project scope included:

- Assay validation
- LIMS and data plane integration
- Automated data processing
- Data analysis and interpretation
- Report template generation
- Clinical report sign off

Streamlined assay validation

Developing a new clinical test requires a combination of secondary and tertiary validation exercises. As a clinical lab that has processed thousands of samples, Variantyx has extensive experience working with known positive and negative variants that we use to calculate sensitivity, specificity and positive predictive value for a wide variety of testing applications, including hereditary cancer testing. Utilizing our expertise significantly streamlined the validation process.

Leveraging the Genomic Intelligence® platform for seamless automation

Variantyx's proprietary Genomic Intelligence® platform provides an automated end-to-end system that simplifies NGS data analysis, interpretation and clinical reporting. Because

PMLS performs its own sequencing in their COLA-certified South Carolina facility, custom LIMS and data plane integration were required to ensure secure and high fidelity transfer of patient information and variant data from their systems to our cloud-based servers. Once received, the data is analyzed and interpreted by our clinical geneticists who return a signed clinical report in PDF format accompanied by meta information. The deliverables are picked up by PMLS's LIMS system which generates a header page containing additional patient information, attaches the transmitted PDF report and delivers the completed package to the ordering physician.

Each case is logged and trackable via the online Genomic Intelligence® console. Custom implemented standard operating procedures (SOPs) and exception management protocols ensure that from the time PMLS's staff begins the sequencing run for a patient sample to the time the report is delivered to the ordering physician, no manual input is required on their side.

Leveraging Variantyx's clinical interpretation and report sign off capabilities

As a clinical lab, our staff includes board-certified clinical and molecular geneticists trained in data interpretation who have the authority to sign off on clinical reports. Interpretation protocols designed to follow standards set by the American College of Medical Geneticists (ACMG) employ our up-to-date database of aggregated variant information to ensure that identified variants are annotated with relevant, curated clinical observations and algorithmically-generated severity scores. Preset filters customized for PMLS's 30 gene hereditary cancer assay were constructed to consistently and automatically select the most relevant variants for review and reporting by our geneticists. Selected variants are imported into a custom report template featuring PMLS's own branding, test description, data fields and medicolegal language. After final review and sign off by our geneticists, the completed report is delivered.

The cloud-based Genomic Intelligence® interpretation platform makes it possible for PMLS's staff to take over interpretation and clinical report sign off activities in the future.

Summary

Working in partnership with Variantyx, leveraging the powerful automation and simplified NGS data analysis, interpretation and clinical reporting components of our Genomic Intelligence® platform, PMLS was able to expand their test menu and launch their new hereditary cancer panel test within a short period of time. Since taking the assay live, patient samples have been moving through the system as fast as they can be sequenced.

For more information about Variantyx or Genomic Intelligence®, please visit www.variantyx.com.