Genetic testing lab sees 25-fold scale-up with new informatics system

AdvaGenix increases test-throughput with QIAGEN Clinical Insight (QCI®) Interpret

About AdvaGenix

AdvaGenix, based in Rockville, Maryland, is the first all-encompassing preimplantation genetics laboratory that employs next-generation sequencing (NGS) for all testing. The lab provides one of the most advanced bioinformatics platforms to detect, annotate, and classify genomic variants associated with multiple disorders, including heart conditions, metabolic and pediatric disorders, and hereditary cancers.

The Challenge

The rapid pace of development in genomics is revolutionizing scientific research, causing a seismic shift in precision medicine. The increase in speed and decrease in cost of NGS technologies has outpaced Moore's law. The $1,000 genome is now a reality. The bottlenecks are now genome informatics and data portability; researchers don't have the computer power nor the infrastructure to manage the volume of raw sequence data they are creating.

"We looked at 15 or so vendors and we did an in-depth analysis comparing all of [them]," says William Kearns, co-founder and chief scientific officer of AdvaGenix. "We wanted a bioinformatics platform that could provide highly accurate analytics with 100% reproducibility and deterministic results." To accomplish this, AdvaGenix partnered with QIAGEN Clinical Informatics Services.

> "Now with automation, one lab director can do about 1,000 samples per month and it is not overwhelming. We have been able to increase our productivity by using the QCI Interpret bioinformatics platform by 35-40%."
>
> Dr. William Kearns
> Co-founder

QIAGEN

Why QIAGEN Clinical Informatics Services

AdvaGenix determined that moving to an automated bioinformatics platform was the best way to address its challenges. "We really needed intelligent bioinformatics. We wanted a fully automated, turn-key analytic that gave us a real end-to-end solution," says Kearns.

The company considered building its own bioinformatics pipeline and hiring an in-house variant curation team. After examining the cost of developing, validating, and maintaining the workflow's infrastructure, AdvaGenix researched the available clinical service options. The company chose QIAGEN Clinical Informatics Services because QIAGEN could deliver complete end-to-end testing solutions, fully-customizable and capable of supporting a broad range of indications.

Also instrumental in AdvaGenix's decision was the inclusion of QIAGEN Clinical Insight (QCI®) Interpret—the clinical decision support software found at the core of QIAGEN's Clinical Informatics Service offerings.

The Benefits

With the help of QCI Interpret and QIAGEN Clinical Informatics Services, AdvaGenix has scaled productivity by 35 to 40%.

"Prior to the automation of [our workflow], we were spending potentially 15 minutes—which is beyond-slow—per report," says Kearns. "Now with automation, one lab director can do about 1,000 samples per month and it is not overwhelming."

QCI Interpret reproducibly translates highly complex NGS data into concise, clinician-ready reports, using current clinical evidence and all 28 criteria of the ACMG/AMP variant interpretation guidelines. Unlike any other bioinformatics platform on the market, QCI Interpret is powered by manual curation.

The software leverages QIAGEN's Ingenuity Knowledge Base, a repository of expertly curated clinical and scientific content that is structured into an ontology containing more than 2.8 million classes. QCI Interpret captures both confirmatory and contradictory findings and includes the source publications and context, ensuring users have all the supporting evidence required to assess the pathogenicity of a specific variant.

AdvaGenix selected QIAGEN because the bioinformatics platform provided unsurpassed traceability and transparency, giving Dr. Kearns peace of mind and full control over his lab's results. "We had to come up with some type of solution that [would make] me comfortable to sign out clinical reports. We needed to see complete reproducibility in the samples analyzed," says Kearns.

AdvaGenix has also reduced IT management time with the support of QIAGEN Clinical Informatics Services. "We've been using QIAGEN for about a year," Kearns says. "We are very pleased with how the system works."

Go to www.qiagenbioinformatics.com/qci-interpret to learn more.