Appistry Expands Cancer Genome Analysis Suite

Appistry Inc.

Appistry, Inc. announced the expansion of its Cancer Genome Analysis (CGA) Suite to include additional tools for next-generation sequencing (NGS) analysis from the Broad Institute. The first addition to the suite is a method for estimating contamination levels in cancer genome data. The expanded partnership—coming just over one year after the Broad Institute named Appistry as the authorized distributor of the Genome Analysis Toolkit (GATK)— demonstrates the organizations' commitment to developing and delivering high quality genetic analysis algorithms and tools suited to the rigors of regulated clinical sequencing workflows.

"As more and more labs look to adopt NGS-based clinical tests, the need for robust, well-tested analysis pipelines has become acute," said Trevor Heritage, vice president of corporate development and strategy at Appistry. "We're thrilled to be delivering the innovative, easy-to-use-and-deploy genetic analysis products based on the highest quality science and algorithms from the Broad Institute."

Appistry and the Broad Institute premiered their partnership at the ASHG annual meeting in October 2012. The partnership was extended in July 2013, when Appistry unveiled the CGA Suite, a complete, integrated workflow for somatic mutation studies based on three Broad tools: the GATK; MuTect, which is engineered to provide the high sensitivity required for identifying low-frequency point mutations; and Somatic Indel Detector for identifying insertions and deletions in cancer genome data. Appistry's CGA Suite includes all of these tools along with a test dataset, results summary, and script to accurately connect and configure the workflow.

This newly expanded collaboration adds ContEst, a tool for estimating cross-contamination in NGS tumor/normal samples, to the CGA Suite. As part of the CGA Suite, ContEst will provide enhanced specificity by eliminating false positives called due to sample contamination.

Earlier this year, Appistry announced a significant upgrade to the GATK software available to for-profit users with commercial-grade support, featuring robust improvements in the Broad Institute's innovative variant caller, the Haplotype Caller. The Haplotype Caller now works at speeds on par with the GATK's legacy variant caller, the Unified Genotyper, while providing much more accurate calling of SNPs and indels.

"By selecting Appistry as a distribution partner for the Genome Analysis Toolkit last year, we enabled our software development team to focus on introducing innovative functionality and extending the utility of tools like the Haplotype Caller,"

said Issi Rozen, director of strategic alliances for the Broad Institute. “Broadening our distribution relationship with Appistry beyond the GATK highlights the depth of our genetic analysis offerings at the Broad, and Appistry’s choice to deliver these tools in the context of specific, high-value workflows such as cancer analysis will make it easier for organizations to implement proven, validated, best-in-class pipelines.”