

Software PowerTools for Genetic Analysis

Contact Information: John Fosnacht (<u>john@softgenetics.com</u>) SoftGenetics LLC The Oakwood Centre State College PA 16803 USA

FOR IMMEDIATE RELEASE

Operational Management function incorporated into Geneticist Assistant NGS Interpretative Workbench

State College PA, SoftGenetics announced the incorporation of an Operational Management function into its newly enhanced Geneticist Assistant NGS Interpretative Workbench. The Operational Management function includes a unique workflow builder that permits users to mimic their physical workflow in "silico", providing real-time status tracking of patient samples throughout the workflow; operational statistics by department or function and automated email notification of status changes.

Developed in collaboration with Emory Genetics Laboratory, the new management system streamlines control and management of patient samples, provides "time-based" production information by department, identifies operational bottlenecks, and provides a sound base for future systemic improvements.

Additionally, the new release incorporates pathogenicity calling information from 17 public and custom databases; single click linkage to comprehensive databases such as Alamut and LOVD (Leiden Open Variation Database) and others, providing pathologists and geneticists with a *comprehensive overview of information regarding variant pathogenicity in a single detailed view*.

Significantly, the company has added many useful tools to the workbench including: CAP Validation Assistance that captures quality trending statistics over time; positive control verification capabilities; process control stats; an automated BED file builder, that indicates regions of clinical significance and "bioinformatics pipeline automation" all while building a historical knowledge base of Next Generation Sequencing Whole Exome or Targeted Disease Panels data. The Workbench is compatible with outputs from all major NGS sequencing platforms including those from Ion Torrent, Illumina and Roche.

Jason Park MD, PhD, Assistant Professor, University of Texas Southwestern Medical Center, Director, Advanced Diagnostics Laboratory, Children's Medical Center Dallas indicates that "Geneticist Assistant integrates all of the necessary tools for the comprehensive analysis of next-generation sequencing data, while providing us with a utilitarian platform for on-going process and quality improvement of our NGS workflow."

SoftGenetics offers no cost web review of Geneticist Assistant NGS Interpretative Workbench to interested parties. To arrange for the informative web meeting, please email: <u>info@softgenetics.com</u> or visit <u>www.softgenetics.com</u> for further information.

-30-