SOFTGENETICS®
Software PowerTools for Genetic Analysis

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FOR IMMEDIATE RELEASE

New dispersion based Copy Number Variation (CNV) Tool added to NextGENe® Software

The CNV detection tool of NextGENe version 2.3.4 forward includes a sophisticated new coverage-based algorithm developed for NGS sequencing data from instruments such as Illumina, Roche, and Ion Torrent sequencing platforms. Applications include both whole exome as well targeted sequencing panels such as Ion Torrent AmpliSeq panels or the HaloPlex Target Enrichment System from Agilent Technologies. Copy number variations can now be detected in NGS sequencing data using dispersion measurements and a novel Hidden Markov Model (HMM) not found in other NGS analysis programs.

With the new technology specified regions of a "sample" and a "control" are used to determine a coverage ratio measured in RPKM for every region (sample divided by sample plus control). CNV calls are made on the basis of changes in coverage, utilizing automatic measurement of noise (dispersion) and a novel Hidden Markov Model. Additionally, each region receives a phred-scale score for insertions and deletions, with results available in a table and unique graphical view.

Copy number variations are detected by comparing the coverage of regions in the sample and control projects. Regions are defined by annotation or incremental length for whole exome sequencing projects, and a BED file for targeted sequencing projects. A beta-binomial model is fit to the coverage ratio (similar to ExomeDepth software) in order to model the amount of dispersion (noise). Simple classifications are generated for each region, such as "Insertion" (increased copy number), "Normal" (little evidence of CNV), "Deletion", or "Uncalled" (due to low coverage).

John McGuigan, Biologist at SoftGenetics indicates that "Like many other CNV tools, this approach is based on the assumption that the copy number affects the level of coverage. This new algorithm takes it a step further and tests that assumption to provide an appropriate level of confidence. It then uses a Hidden Markov Model (HMM) to translate this confidence into CNV calls. The entire process is implemented in an easy-to-use interface that allows for simple fully-automated analysis or for manual tweaking of parameters to get the best results. The new graphical view makes it easy to visualize the raw data and the final results in an integrated way."

Interested parties may request a beta version of version 2.3.4 by email info@softgenetics.com, or review further information and download an Application Note: http://www.softgenetics.com/NextGENe_013.html or view a webinar on CNV detection: http://www.softgenetics.com/webinar/NG_CNV_algorthm_2013.html.

NextGENe is a comprehensive, free-standing Windows® based analysis program compatible with all NGS sequencing platforms. The easy-to-use user interface requires no scripting, provides highly graphical results and reporting, with multiple filtering options, multiple analysis comparison capabilities as well as causative prediction from multiple databases including COSMIC, dbNSFP which includes 1000 genome frequency, PolyPhen and other functional prediction databases. The company offers 30-day trials and no cost web-based training on its genetic analysis software packages.