SOFTGENETICS®
Software PowerTools for Genetic Analysis

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

**FOR IMMEDIATE RELEASE**

# No Cost CNV Analysis offered by SoftGenetics

**September 20, 2013 State College PA**, SoftGenetics announced today that it will provide interested NGS users with up to 10 Copy Number Variation (CNV) analyses at no cost. The analyses will be performed with the company's latest Hidden Markov Model based technology found in NextGENe software CNV tool. To participate in this limited time offer, interested NGS users need to electronically provide up to 10 individual WES or targeted panels BAM files as well as one control BAM file. SoftGenetics will quickly perform the CNV analysis utilizing NextGENe's proprietary CNV technology and furnish both graphical and annotated report of the results. Furnished BAM files must be sanitized of identifying information.

*"This is a great way to experience the kind of results that can be expected from this new analysis tool without having to invest time in analyzing the data."* stated John McGuigan developer of the technology.

Kevin LeVan, NextGENe software Product Manager *added "our collaborators have demonstrated that NextGENe software's sensitivity in NGS CNV-Seq samples parallels that of MLPA analyses in capillary sequencing. "*

NextGENe software's CNV detection tool 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 as 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.

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).

Interested parties in the free CNV analyses may request the free CNV by visiting http://www.softgenetics.com/NextGENe.html or by email: info@softgenetics.com .

*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.*