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## FOR IMMEDIATE RELEASE

## DNA Mutation & Methylation Quantification added to Mutation Surveyor<sup>®</sup> software

**March 5, 2009, State College PA** SoftGenetics announced a quantification feature has been added to its Mutation Surveyor DNA Variant analysis software that provides status quantification of specific nucleotides of Sanger Sequencing traces. This new feature is valuable for a variety of applications including disease study and diagnosis, quantification of drug resistance in a population, such as, antibiotic resistance in bacteria, and heteroplasmy studies of mitochondrial DNA.

Dr. David Hulce, SoftGenetics Technical Support indicates:"Quantification of allele ratios in gene expression and genomic analyses is useful for disease diagnosis and developing treatment protocols for patients in individualized medicine. Understanding the early stages of disease is invaluable since early detection is vital for efficacious treatment. Quantification of the presence of disease-causing markers can be used as a tool for early screening for diseases, such as, methylation status of genes associated with various cancer and heteroplasmy in mitochondrial DNA. Quantification of variant alleles can be used to monitor specific genes for drug-resistant mutations: BRC-ABL gene in CML patients and antiviral-target genes of hepatitis C virus (HCV) and human immunodeficiency virus (HIV). Mutation quantification can be used to measure antibiotic-resistant mutations in bacterial strains within a sample, useful for monitoring infections and microbial source tracking."

However, Dr. Hulce continued, "Determining the concentration of different alleles in DNA can be problematic. Two alleles of one gene may differ at only one position with sequence identity for all other positions of the gene. Mutation detection from cancer cells isolated from tumors may contain normal genes from co-isolated normal cells, resulting in a low frequency mutation allele. Promoter regions may be differentially or hyper-methylated and mtDNA may contain regions of heteroplasmy. Quantification of regions containing a high degree of variability may result in some positions not analyzed. Quantification of alleles that occur at low frequency may be below the detection limit of the technique used. The "Mutation Quantifier" function of Mutation Surveyor software solves the above problems. Mutation Surveyor software will align reference and sample sequences, detect mutations and quantify the wild type and variant alleles.

The company offers 30-day trials and no cost web-based training on Mutation Surveyor as well as all of its genetic analysis software. Interested parties may request the software on the company website: <u>www.softgenetics.com</u> or via email: info@softgenetics.com.

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SoftGenetics, LLC specializes in the development of genetic analysis tools for both research and diagnostic applications. Hallmarks of SoftGenetics software tools are advanced technologies, providing exceptional accuracy, and sensitivity in an easy-to use Windows® user interface.

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