SOFTGENETICS®
Software *PowerTools* for Genetic Analysis

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FOR IMMEDIATE RELEASE

New Paired Read Mapper designed to reduce false positives added to NextGENe® 2nd generation sequence analysis software

September 2, 2009, State College PA SoftGenetics announced the addition of an enhanced alignment algorithm which significantly reduces false positive rate of short read paired end and mate-pair sequences from the AB SOLiD System, Illumina Genome Analyzer and Roche FLX systems.

The NextGENe paired-read Mapper, unlike current technologies, which typically aligns both pairs to the reference prior to mating (which often duplicates alignment), pairs the reads prior to mapping in order to increase sequence uniqueness. The pairs are then mapped to the reference. This technique has been found to positively reduce the false positive rate of variants easing and speeding analysis of the data.

Reads in a pair are expected to align a given distance from each other with an expected relative orientation; pairs where the distance between reads or their orientation differs from the expected can indicate positions of structural rearrangements. Paired end reads, such as those from Illumina GA paired end sequencing, are generated by sequencing two ends of a molecule of DNA. Generally, the length of fragments used, known as the insert or library size is between 50-500 bp. Mate-paired reads, such as those from the SOLiD System, are generated following circularization of larger DNA fragments using an internal adaptor between ends of the fragment, then breaking of the circular DNA using restriction enzymes to produce smaller fragments with the internal adaptor between the two ends of the original fragment. Typical mate-paired library sizes are 0.6, 1, 2.5 and 4 kb. It is possible for the use of larger fragment sizes to result in some bias in the selectivity of fragments since breakage of DNA into larger fragments (0.6-4 kbp) can be less random than breaking into smaller fragment (50-500bp).

Pairs outside the expected gap size, indicating presence of possible structural variations, are reported and captured for further examination. Two Paired Read reports are available that can identify the locations where reads are mapping at abnormal distances and orientation, in addition to identifying reads where only one of the pair match.

“NextGENe’s paired end mapping technology, is a further extension of our philosophy to enlist novel technologies into NextGENe”, indicates Jonathan Liu, VP Development, which provide the highest accuracy while maintaining a simple, biologist-friendly user interface. The increased reporting capabilites is one to the keys to having accurate reliable results and we feel NextGENe is a unique tool in that respect.”

The company offers 30-day trials and no cost web-based training on its genetic analysis software packages Interested parties may request the software on the company website: www.softgenetics.com 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.