Complete genomics

The Complete Genomics Reanalysis Service

The Complete Genomics Analysis Pipeline is continually being enhanced to provide customers with more valuable information about their sequenced genomes. Access to new versions of the Analysis Pipeline for previously sequenced genomes is available through the Complete Genomics Reanalysis Service. Through reanalysis, it is possible to gain all of the benefits of the latest analysis developments and advantages without the need to sequence the same DNA samples a second time.

The Complete Genomics Analysis Pipeline begins by mapping raw sequencing reads to the human reference genome. Analysis is then performed to identify, score, and annotate variations in the genome, including SNPs, indels, copy number variations, mobile element insertions, and structural variations. For detailed information on new features and enhancements, please refer to the Analysis Pipeline Release Notes. Reanalysis is available for Analysis Pipeline versions 1.5 and later.

A selection of recent improvements to the Complete Genomics Analysis Pipeline is shown here:

ANALYSIS VERSION	NEW FEATURES
2.4	• Added Lesser Allele Fraction (LAF) estimates for all samples to aid in identification of LOH or UPD • Added multiple hypotheses for variants assigned a no-call due to ambiguous evidence • Improved somatic detection with new, additional somatic score and bidirectional comparisons • Improved sensitivity in the mitochondrial genome with modifications that include treating the sequence as diploid rather than haploid
2.2	• Added a Variant Call Format (VCF) file as part of the standard data package
2.0	• Added cancer-specific functionality for tumor-normal pairs and tumor-tumor-normal trios, including identification of somatic events • Improved small variant calling algorithm for increased sensitivity to SNPs, indels, and block substitutions, along with scores indicating confidence of each call under diploid and non-diploid assumptions • Updated the copy number variation (CNV) and structural variation (SV) reference baseline set with new genomes • Added translation of identified SV junctions into structural variation events • Added summary of detected variants and other information into Circos Plot
1.12	• Added master variation file, aggregating variant calls and annotation information from various Complete Genomics export files • Added mobile element insertion detection results
1.11	• Added coverage distribution and coverage by GC content for coding region of the genome • Expanded reported summary statistics and metrics
1.10	• Added genomic CNV and SV results

Find out more at: www.completegenomics.com or contact your local Complete Genomics sales representative.

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