

Changes from v1.2.2 to v2.0.4

Added and Updated the Following Features, Corrected the Following Bugs

Feature	Description	Risk
STR/YSTR analysis	Capacity to analyze STR/YSTR data using preloaded PowerSeq® panels or custom panels. Results include alleles called, sequences of alleles and isoalleles, depth of coverage, tables and histograms.	Major
Added a "template length" filter to the filter setting dialog	Filter by read length for non-paired data; Keep the paired read if TLEN >= length or TLEN <= - length.	Moderate
Added EMPOP variant lists to a file	Saving xx_variants.txt as the EMPOP variant report.	Minor
No template file to store bam file	Without template file, the space needed for storage is cut to half and the speed for reading bam remains the same.	Moderate
Additional requirement for assigning mtDNA amplicons and calculating coverage	When analyzing paired end data, forward and reverse reads must sort to the same amplicon.	Major
Check "Motifs" as default setting	Checking or unchecking "Motifs" does not change the instrument type. Enable "motifs" when an instrument is selected.	Minor
Coverage Report Customization	Added a "Settings" button next to the "Save As" button. "Total Coverage" only shows the black line. "Directional Coverage" only shows the red and blue lines. "Both" is checked by default and shows all the lines.	Minor
Move "adding variant at a given position" options to a submenu	Showing a submenu of "Add A at position X", "Add C at position X", "Add G at position X", "Add T at position X", "Add del at position X", "Add insC at position" for the pileup right-click.	Minor
Global Coverage image saving	Saving an image similar to the current global viewer with forward coverage stacked on top of reverse coverage.	Minor
Updated IUPAC calls in the table viewer	Updated the format for deletion and some other allele at the same position.	Minor
Added template length report	The 'Template Length' report shows a length distribution based on positive TLEN values in the BAM file for paired-end data.	Minor
Added read length report	Displaying two plots, "Read Length" which shows the length of the sequences in the BAM file and "Aligned Read Length" which shows the length of the sequences, ignoring any soft-clipping in the CIGAR string.	Minor
Added Amplicon GUI on viewer	Displaying the amplicon regions in the viewer when Amplicon Settings is used. Hovering on an amplicon will list the chromosome, start, end, and the number of reads assigned to it.	Minor

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Added "Jump to Position" right-click option in the pileup	The user can enter a position number to move the viewer to display the designated position is in the middle of the viewer.	Minor
Added "STR and SNP Amplicon tabs" in the filter setting dialog	STR and SNP Amplicon tabs are used for STR analysis.	Minor
Added a "Match Proportion" filter	Match Proportion means proportion of read that is aligned and not soft-clipped.	Moderate
Added an "Identity" by number filter	"Identity" by number means number of differences between the read and the reference.	Moderate
Fixed "Copy Consensus" bug	Fixed the bug which copies 'A' instead of 'N'.	Minor
Fixed "Amplicon Settings" bug	Amplicons spanning the origin can be input manually and loaded from the BED file now.	Minor
Fixed amplicon region trimming	Improved trimming of positions outside amplicon region boundaries	Minor
Fixed the "GUI zoom" bug	Fixed the GUI issue when changing the "Zoom" level in Windows	Minor

Description of risk categories:

Minor – cosmetic; such as changes to the graphic user interface and reporting options (no algorithm change).

Moderate – includes minor algorithm changes that may affect sample grouping and final reports.

Major – includes significant algorithm changes that may result in alignment and mutation call detection differences.