

NextGENe Change Log

Version 2.4.2.3

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Mutation Report	Add Order Options for Variation Track Settings	No	Low
Reports	AMP/CAP pipeline validation guidelines - phase 1	No	Low
STR	STR Application Improvement	No	Low
Track Manager	Support dbNSFP3.5	No	Low
Track Manager	Support ClinVar updates	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Alignment	Fix GRCh38 Projects Failing to open in Viewer for certain cases	No	Low
Alignment	Fix issue with Hide Unmatched Ends and Amplicon BED File	Yes	Medium
Autorun	Correct AutoRun Format Conversion Settings Display	No	Low
BAM	Fix incomplete BAM export due to special case paired end read name format	No	Low
CNV	Fix issues with CNV Tool	Yes	Medium
CNV	Fix CNV Tool Floating Point Overflow Error with Certain Samples	No	Low
Coverage Curve	Fix Coverage Curve Report's 3'UTR HGVS nomenclature	No	Low
Mutation Report	Fix reference allele complement issue with Mutation Call Relative to mRNA nomenclature	Yes	Low
Mutation Report	Homopolymer Indel Calling Issues	Yes	Medium
Mutation Report	Fix Mutation Report Limit Region Filters Not Applied for Splice Sites When Selected Under Type	No	Low
Mutation Report	Update Mutation Report ROI BED filter to show variants that partially overlap when phasing is turned on	Yes	Medium
Reports	Fix SV Detection Bugs	No	Low
Sequence Operations	Fix issue with Remove Duplicates by UMI regarding targets sharing UMIs with different target	Yes	Medium
Statistics	Fix Special Case Statistics and Mutation Detection Crash	No	Low
	Other minor bug fixes		

Version 2.4.2.2

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
CNV	Enable CNV Tool to process a batch of samples (for Dispersion/HMM)	No	Low
Variant Comparison	Add "Show all transcripts" feature to Variant Comparison Tool	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Coverage Curve	Fix Coverage Curve Report using Hotspot BED file missing 1bp regions	Yes	Low
Mutation Call	Fix incorrectly calling of homopolymer indels that meet "indel" calling threshold but not "homopolymer indel" threshold	Yes	High
Mutation Report	Fix Show All Transcripts Incorrectly Filtering Splice Variants	Yes	Medium
Mutation Report	Fix Filtering Multiple Base Deletions by Bias	Yes	Medium
	Other minor bug fixes		

Version 2.4.2.1

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Autorun	Add loaded sample file count to Job File Editor	No	Low
Autorun	Load Multiple PJTs for Report-only Jobs in AutoRun	No	Low
CNV	Increase the number of control projects for CNV	Yes	Medium
Format Conversion	Remove uncalled bases from end of read when "trim or reject read" option is selected	No	Low
Mutation Call	Update forensic mutation call nomenclature to include reference allele	No	Low
Mutation Report	Add Mutation Call to VCF	No	Low
Mutation Report	Enable Mutation Report column ordering	No	Low
Project Files	List BED File in StatInfo for Mito Amplicon application	No	Low
RNA-Seq	Include amino acids and nucleotides in Zoom Display of Detected Transcripts	No	Low
SAD Tool	Make SAD Tool compatible with using NextGENe license	No	Low

Track Manager	Import Reference tool update to support local disk (not just DVD)	No	Low
Variant Comparison	Add copy sequence for comparison tools	No	Low
Viewer	Update mRNA to Exon	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Alignment	Fix Crash with Alignment to reads across contigs of a BED-based Preloaded Reference	No	Low
Alignment	Fix large deletion/insertion alignment issue	Yes	Medium
Autorun	Fix issue with AutoRun rewriting Project Settings configuration file	No	Low
Autorun	Fix Viewing All File Types in AutoRun Job Editor	No	Low
Autorun	Intermittent AutoRun Failed Job error	No	Low
CNV	Fix Batch CNV tool's Viewer crash issues	No	Low
CNV	Update Graphics When Loading New Sample or Control in CNV Tool - SNP Method	No	Low
CNV	Fix Search Function in CNV Tool	No	Low
Coverage Curve	Fix HGVS columns in Coverage Curve Report	No	Low
Mutation Call	Fix issue with mutation calls in repeat at beginning of ROI	Yes	Medium
Mutation Call	Fix issue with incorrectly filtering Indel by SNP mutation percentage when Indel mutation percentage is lower	Yes	High
Mutation Report	Fix Amino Acid Change and Function issues when Merge phased variants option is selected	No	Low
Mutation Report	Fix issue with Mutation Report Settings ignoring Filter by Annotation Regions in previous versions' settings files	No	Low
Mutation Report	Fix balance ratio calculation issue for homopolymer indels	Yes	High
Mutation Report	Fix unfiltered VCF missing some filtered indel mutation calls	No	Medium
Mutation Report	Fix blank Mutation Call values of non-preferred transcripts for All Transcripts option	No	Low
Mutation Scores	Calculate Homopolymer Scores for Ion Torrent	Yes	Medium
Mutation Scores	Add scores for Phased variants	No	Low
Project Files	Fix file write speed	No	Low
RNA-Seq	Fix Access Violation Error for Post-processing Transcript Report with No Settings File	No	Low
Sequence Operations	Fix FASTA input issue with Remove Duplicates	No	Low

Somatic Mutation Comparison	Fix Annotation Missing from Somatic Mutation Comparison Tool	Yes	Medium
Track Manager	Fix dbNSFP Population Frequency filtering issue	Yes	Medium
Track Manager	Fix issue with Track Manager recognizing "chr" in Contig	No	Low
Variant Calling	Improve In-read Phasing for multiple alleles	Yes	High
Viewer	Remember height of mutation report	No	Low
Viewer	Fix issue with Geneticist Assistant viewing position in NextGENe Viewer when path has space	No	Low
	Other minor bug fixes		

Version 2.4.2

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Alignment Settings	Hide Unmatched Ends Update	Yes	High
Autorun	Add "Automatically Start" option to AutoRun Settings	No	Low
Autorun	Update AutoRun settings to replace "Start Time" with "Delay Detect" option	No	Low
BAM	Add mapping quality to BAM output	No	Low
BAM	Add ROI BAM as Export type for Post-processing	No	Low
Barcode Sorting	Remove unnecessary functions and improve FASTQ support for "Barcode in Separate Files"	No	Low
CNV	Add Export option for CNV Tool graphics	No	Low
CNV	Update CNV SNP comparison method: using both SNP and Read Count	No	Low
CNV	Update Batch CNV Tool calculations, filters and graphics	Yes	Low
CNV	Add Reporting Filter to CNV Tool	No	Low
CNV	CNV Dispersion HMM - Change Manual Dispersion Value to Fixed Dispersion Value, add Auto-detect	No	Low
Filtering Settings	Update Mutation Filter's balance ratio filter (move to Viewer and include ratio relative to percentage)	Yes	Medium
Geneticist Assistant	Geneticist Assistant input optimization	No	Low
Mutation Call	HGVS Nomenclature Improvements	No	Low
Mutation Call	Support "In-Read Phasing" of simple Deletions/Insertions in Mutation Report	Yes	High
Mutation Report	Save negative positions (hotspots) to VCF	No	Low
Mutation Report	Mutation report setting columns and dialog reorganization	No	Low
Mutation Report	Update HGVS Nomenclature	No	Low

Mutation Scores	Add "Allele Score" to Mutation Report's Display and Filter settings	No	Low
Post-Processing Reports	Add Transcript Report as one of the available Post-processing Reports	No	Low
Project Files	Check file write	No	Low
Project Files	Update project output folder structure - Reports and Files	No	Low
Sequence Operations	Add a "Random Subsample" tool to Sequence Operations	No	Low
Sequence Operations	Support UMIs using Illumina I2 files	No	Low
Settings	Enable the use of local temp directory for any input/output locations	No	Low
Templates	Add NEBNext templates	No	Low
Track Manager	Support Genome Build Information in References	No	Low
Variant Calling	Improve Allele Calling for Determining Zygosity	Yes	High
Viewer	Make NextGENeViewer 64-bit compatible	Yes	Medium
Viewer	"Add Mutation" is permitted for calls with inadequate coverage	No	Low
Viewer	Remember Mutation Report location and column widths	No	Low
Viewer	Add ROI track to Viewer	No	Low
Viewer	Remove Paired Reads Viewer	No	Low
Viewer	Improve status bar and zooming	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Alignment	Fix issue with paired alignment and no allowable ambiguous alignments	Yes	High
Alignment	Fix IUPAC issue for "N" bases being shown as "A" with Detect large indels	No	Low
Alignment	Check 5p positions prior to hiding unmatched ends for Paired Alignment with Amplicon BED file	Yes	Medium
Alignment	Fix soft clipping issue due to selecting wrong amplicon region for some reads	Yes	Medium
Alignment	Fix issue for Paired End Reads mapping on circular reference appropriately	Yes	Medium
Autorun	Fix AutoRun Template Saving Issue with Sequence Trim Conversion Settings File	No	Low
Autorun	Fix Secondary Analysis Input issue	No	Low
Autorun	Fix error when some special language characters are used in AutoRun	No	Low
BAM	Transcriptome Project Export BAM MD tag fix	No	Low
BAM	Fix BAM output related to circular alignments	No	Low
BAM	Fix SAM/BAM "Reference Sequence Name" when	No	Low

	Reference input is ChrM GBK		
BAM	Fix SAM/BAM export issue with soft-clipping of ROI	No	Low
BAM	Fix Issue with Large Deletion Detection from BAM Files	No	Low
Condensation	Fix error with Condensation only projects	No	Low
Mutation Call	mtDNA mutation call nomenclature fixes	Yes	Medium
Mutation Report	Save Consensus Sequence relative to mutation report filters bug	Yes	Medium
Mutation Report	Fix some VCF Export information for Indels	No	Low
Mutation Report	Fix issue with reporting Ambiguous Gain/Loss Penalty values in VCF	No	Low
Mutation Report	Fix incorrect Mutation Report Summary values	No	Low
Project Files	Fix long path issue	No	Low
RNA-Seq	Fix exon link issue	Yes	Medium
RNA-Seq	Fix Transcriptome Error issue with Ion BAM Files	No	Low
SAD Tool	Fix X/Y cutoffs in SAD Tool	No	Low
Track Manager	Fix Issue with CAF/GMAF Fields from dbSNP with overlapping indels	No	Low
Variant Comparison	Variant Comparison Tool Score Filter Fix	No	Low
	Other minor bug fixes		

Version 2.4.1.2

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Annotation	Integrate with Elsevier's Pathway Studio	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
	Other minor bug fixes		

Version 2.4.1.1

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
RNA-Seq	Add Contig Column to Transcript Report	No	Low
Track Manager	Separate CAF values for dbSNP custom track	No	Low

	import		
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Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Alignment	Fix for "Hide Unmatched Ends" and Detect Large Indels	Yes	Medium
Alignment	Update for paired read handling improvements	Yes	Medium
Alignment	Detect Large Indels memory management crash issue	No	Low
Alignment	"Set Amplicon BED file" changes to handling Paired-End data	Yes	Medium
Alignment Settings	Fix missed low covered hmz mutation calls due to issue with "Except for Homozygous" settings and tracks	Yes	High
Autorun	Load NGJOB issue	No	Low
BAM	Fix issue with some BAM files only being partially loaded	No	Low
Distribution Report	Fix issue with Distribution Coverage Report zero coverage positions	No	Low
Format Conversion	Fix wrong ASCII table selected in special cases	No	Low
Mutation Report	Fix Access Violation when VCF is blank	No	Low
Mutation Report	Fix ROI filter ignored when Keep all splice variants selected	No	Low
Mutation Report	Fix formatting of Track Info for VCF files	No	Low
Reports	Fix Gene Distance bug in Peak Identification report	No	Low
Track Manager	dbNSFP v2.5 Import Incomplete	No	Low
Track Manager	Fix dbSNP VCF import in Track Manager	No	Low
Track Manager	Gene Tracks fix	No	Low
Track Manager	Fix incorrect dbSNP annotation for some insertion calls	No	Low
Track Manager	Fix issue displaying track information in Mutation Report (with long file path)	No	Low
Variant Comparison	Access Violation Error Saving VCF from Variant Comparison Tool	No	Low
Viewer	Viewer performance issue for Mitochondrial Amplicon with large numbers of reads	No	Low
Viewer	Fix Viewer crash issue with preloaded reference built from BED file and a non-preferred transcript difference at splice site	No	Low
	Other minor bug fixes		

Version 2.4.1

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Alignment	Improve "Set Amplicon BED file" option in overlapping amplicons	Yes	Medium
BAM	BAM Output update for Soft Clipping and BED Target	Yes	Low
Coverage Curve	Add "Use Original Coverage" setting in Coverage Curve	No	Low
Export	Load BED file for BAM/SAM export	No	Low
Expression Report	Expression Report to include read counts relative to amplicons	No	Low
Expression Report	Add Minimum Forward Read Coverage and Minimum Reverse Read Coverage to Expression Report	No	Low
Mutation Report	Add Setting to Keep All Splice Variants for Mutation Report	Yes	Medium
SAD Tool	Improve SAD Tool (Beta)	Yes	Medium
Settings	Create a template for RainDance Technologies' panels	No	Low
Summary Report	Add Settings File Names to Summary Report	No	Low
Track Manager	Separate MAF values of EspVCF database to individual columns	No	Low
Track Manager	dbNSFP 2.9 support	No	Low
Track Manager	dbscSNV Support	No	Low
User Management/Tracking	Add Audit Trail	No	Low
Variant Comparison	Add a Somatic Mutation Comparison Tool to NextGENe Viewer	No	Low
Variant Comparison	Improve speed for saving the variant comparison report	No	Low
Viewer	Improve speed of Expression Report and CNV tool for large projects	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Alignment	"Unmatched_Paired" files are incomplete with "Detect Large Indels"	Yes	Medium
Alignment	"Hide unmatched ends" fix	Yes	Medium
Autorun	Fix issues with Create More Projects not sending Run Name and Date/Time to Geneticist Assistant	No	Low
Build	Build Preloaded Reference bug when limiting to region	No	Low

Reference	defined in BED file		
CNV	CNV Bug fixes	Yes	Medium
Export	Fix BED export 0-based offset issue	Yes	Medium
Mutation Report	Variation Tracks Settings Issues with Postprocessing and User Defaults	No	Low
Mutation Report	EEFACE Error Saving VCF	No	Low
Track Manager	Microsoft Windows Display text size issue	No	Low
Variant Comparison	Fix for some variants missing from VCT	Yes	High
Viewer	UNC Reference paths not recognized correctly on remote computer	No	Low
	Other minor bug fixes		

Version 2.4.0.2

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Alignment	Update "Hide Unmatched Ends" to hide a mismatched first or last base	Yes	Medium
Mutation Report	Revert VCF output for indels in repeat regions where multiple occur in the same repeat	No	Medium
StatInfo	Add "Aligned Reads" Counts to run statistics and update expression report calculations	Yes	Medium
Variant Comparison	Improve speed of Variant Comparison Tool over network	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Alignment	Fix an issue with some insertions detected as string of substitutions	Yes	Medium
Alignment	Alignment issue with reads that have several mismatches to paralogous locations	Yes	Medium
Alignment	Issue with Set ROI regions from BED file and ambiguous reads	Yes	Medium
Alignment	Detect Large Indel and Hide Unmatched Ends Alignment Bug	Yes	Medium
Autorun	Fix Load Processed Projects in AutoRun issue	No	Low
BAM	Fix BAM output of paired reads for aligned flag and unmapped reads mapping score	Yes	Medium
BAM	Convert to BAM issue with a special case of paired reads and targeted BED regions	Yes	Medium

Coverage Curve	Coverage Report's "Aligned Reads (Including Ambiguous Locations)" number is smaller than "Aligned Reads" number fixed	No	Low
Export	BED Export Bug fix for large projects	No	Low
Mutation Call	Fix Viewer confidence score filters issue, mutation calls hidden	No	Low
Mutation Call	HGVS Amino Acid nomenclature indel offset fix	Yes	Medium
Mutation Report	Fix hiding of reported variants issue when project has no tracks due to some deletions	Yes	Medium
Mutation Report	Fix ROI files on network issue with filtered mutation calls	Yes	Medium
Mutation Report	Fix a bug preventing post-processing output of mutation reports with some older mutation report settings files	No	Low
Mutation Report	Mixed Mutation call VCF issue	Yes	Medium
Mutation Report	Fix bug with Include Negative Positions within ROI	Yes	Medium
Variant Comparison	Fix a Show All issue in Variant Comparison Report regarding filtered and deleted mutation calls in some (but not all) projects	Yes	Medium

Version 2.4.0.1

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Statistics	Read count fix in Coverage Curve and StatInfo files	No	Low
Variant Comparison	Fix crash issue in Variant Comparison Tool with projects having no tracks	No	Low

Version 2.4.0

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Alignment	Paired Reads handling improvements	Yes	Medium
Alignment	hide unmatched end improvement	Yes	Medium
Alignment	Improvements to handling of reads that align ambiguously	Yes	Medium
Alignment Settings	Soft Clipping Support - Off-target/primer sequences and unmatched ends	Yes	Medium

Assembly	Include Original Read Names in AssembledSequences.fasta file when using the Save Original Sequences setting	No	Low
Autorun	Enable the generation of a secondary run that is dependent on output of primary run	No	Low
Autorun	Load AutoRun Template	No	Low
Autorun	Add Sample Grouping Tool to AutoRun Job File Editor	No	Low
Autorun	Remove AutoRun "Local work folder" option	No	Low
Barcode Sorting	454 Sorting, add "Orient Reads Before Sorting" option	No	Low
Build Reference	Circular References support	Yes	Medium
CNV	CNV- Simple Multiple-Control Methods	Yes	Medium
CNV	Add Normalized Reads Counts method, an alternative to RPKM (based on DESeq Normalization)	Yes	Medium
CNV	Link CNV reports to DGV for human data	No	Low
CNV	Add a new "Batch CNV Tool" to Beta tools of NextGENe Viewer Tools menu	No	Low
Coverage Curve	Add HGVS nomenclature to Coverage Curve	No	Low
Export	Add Export Sequences option to Post-Processing	No	Low
Export	Option to export SAM/BAM for selected chromosome(s)	No	Low
Expression Report	Add Fragment Count (FPKM) to Expression Report	No	Low
Forensics	NextGENe Forensics Application Improvements	Yes	Medium
Format Conversion	Enable Format Conversion of Multiple-Sample BAM files.	No	Low
Format Conversion	Update Format Conversion's Interface and Output options	No	Low
Geneticist Assistant	AutoRun can move to next job while NG is submitting previous job to GA	No	Low
HLA	HLA Interface Improvements	Yes	Medium
HLA	Identify Low Coverage Regions	Yes	Medium
HLA	Display HLA consensus nucleotide sequence for all alleles	No	Low
HLA	Add option to remove short reads from alignment	Yes	Medium
Index Creation	FASTA Reference Display Improvements	No	Low
Mutation Call	Display additional Mutation Call information (assist with ACMG NGS Guidelines)	Yes	High
Mutation Call	No-stop Mutation clarification	No	Low
Mutation Call	Support mitochondrial forensic nomenclature	No	Low
Mutation Report	dbSNP Track Import Including Clinical Relevance Filter	No	Low
Mutation Report	Amino Acid Formatting update	No	Low

Mutation Report	Use m. nomenclature for mtDNA mutation calls	No	Low
Mutation Report	Update "Relative to CDS" Nomenclature to use "c." in intronic regions	No	Low
Mutation Report	Add Functional Consequence to Mutation Report and VCF	No	Low
Mutation Report	Output read balance statistics in VCF reports	No	Low
Mutation Report	HGVS Nomenclature relative to genomic sequences	No	Low
Mutation Report	Add Zygosity Column to Mutation Report and Variant Comparison Tool Report	No	Low
NIPT	Sensitive Aneuploidy Detection Tool (SAD) Alpha	Yes	Low
Performance	Optimize AutoRun for running jobs in parallel	No	Low
Post-Processing Reports	Add option to save a copy of post-processing reports to a single location	No	Low
Reports	Add information to Paired Read reports	No	Low
Settings	Reorganize Settings Dialogs and Configuration Files	No	Low
Track Manager	Support annotation in GFF3 format through Track Manager	No	Low
Track Manager	Track Manager VCF Support improvements	No	Low
Track Manager	Read (nonstandard) COSMIC VCF format	No	Low
Track Manager	dbNSFP 2.5 Support	No	Low
User Management/Tracking	Add permissions for users/groups	No	Low
User Management/Tracking	Generate User Login History Report	No	Low
Variant Comparison	Navigate from variant in viewer to variant in report	No	Low
Variant Comparison	Add a Somatic Mutation Comparison Tool	No	Low
Viewer	Increase Zoom Out capabilities	No	Low
Viewer	Increase Whole Genome View resolution	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Advanced GBK Editor	Fix Add SNPs function in Advanced GBK Editor	No	Low
Annotation	Fix Chromosome Position Bug related to error in Annotation.gbk	No	Low
Assembly	Fix error with Assembled Sequences Count When "Save the Original Sequences with Assembled Ones" is selected	No	Low
Assembly	Update Parameters.txt file for Floton Assembly	No	Low
Autorun	AutoRun Issue when All Licenses are in use	No	Low
Autorun	Project truncated when AutoRun stops due to network	No	Low

	instability		
BAM	Fix display error of BAM import settings	No	Low
Expression Report	Fix bug with Expression Report not recognizing a single ROI in genbank file correctly	No	Low
License Server Manager	Project does not complete when connection with server is interrupted	No	Low
Mutation Call	Mutation Report Statistics for Negative Mutations is wrong	No	Low
Mutation Call	c.DNA numbering sometimes wrong for one of the alleles when position has a mixture	Yes	Medium
Mutation Call	Splice Site UTR issue	Yes	Medium
Mutation Report	VCF Allele Frequency is reported lower than expected for some low frequency variants	Yes	Medium
Reports	Use Post-Processing Reports settings in NextGENe Viewer	No	Low
Sequence Operations	Remove Duplicate bug fix	No	Low
Track Manager	Fix issue with incorrect values in columns for 1000 genomes custom track import	No	Low
Variant Comparison	Filtering out Silent Variants in Variant Comparison Tool Not Working	Yes	Medium

Version 2.3.4.6

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Geneticist Assistant	Geneticist Assistant Speed Optimization	No	Low

Version 2.3.4.5

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Mutation Report	Add Read Balance Column to Mutation Report	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Assembly	Assembled sequences count in statinfo file incorrect when using the Save Original Sequences setting	No	Low
Assembly	Update Parameters.txt file for Floton Assembly	No	Low

ChIP-Seq	Peak Identification report "Gene Distance" bug	No	Low
Mutation Call	Balance Ratios - short homopolymer length issues.	Yes	Medium
Sequence Operations	Remove Duplicate Reads Summary Statistics Fix	No	Low
Variant Comparison	Variant Comparison Tool Access Violation Error	No	Low

Version 2.3.4.4

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Reports	Expression Comparison Report Visualization Improvements	No	Low
Variant Comparison	Add Project Name to Sequence View Panel in Variant Comparison Tool	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Mutation Call	Missed Insertion due to imbalanced deletion at same position	Yes	Medium
Mutation Report	Saving unfiltered VCF through Post-processing not working	No	Low
Post-Processing Reports	COSMIC field not included in mutation report through post-processing	No	Low
Variant Comparison	Variant Comparison reported dbSNP filtering issue	Yes	Medium
Viewer	Coverage Calculated Incorrectly in Add Mutation Dialog	No	Low

Version 2.3.4.3

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Autorun	Load Multiple PJT files in AutoRun Job File Editor and Split Job	No	Low
Sequence Operations	Add statistics to Remove Duplicates Log	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm	Risk
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		Change	
CNV	EEFFACE (Memory Allocation) Error in the CNV Tool	No	Low
Database	NextGENe fails to retrieve the correct CDS start information from the database	Yes	Low
Export	SOLiD BAM Export Bug	No	Low
Expression Report	Expression Report - Keep duplicate regions for report generated by BED file	No	Low
Geneticist Assistant	Geneticist Assistant Fields blank in Project Wizard	No	Low
Mutation Report	Save Consensus Bug	No	Low
Settings	Language Settings conflict	No	Low
Settings	Indel Balance Mutation Filter update	Yes	Medium

Version 2.3.4.2

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
CNV	Dispersion&HMM - Tumor-Normal Adjustment Setting	Yes	Medium
CNV	Add CNV Settings for Minimum Region Length, Custom Fitting Point Number and other Advanced Settings	No	Low
CNV	Update to CNV scores	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Autorun	Remove "Please do alignment" message when processing unmapped BAM files in AutoRun	No	Low
BAM	BAM Conversion crashes or fails when sorting large BAM files, and is too slow	No	Low
BAM	BAM Input Problems	No	Low
Mutation Report	Chr reported as N/A for mtDNA genbank file	No	Low
Other Reports	Expression Comparison Report Issues	No	Low
Other Reports	Summary Report's Coverage Curve Sections mis-labeled	No	Low
Tools	"Reads Simulator" and "Pseudo Paired Read Constructor" Filename Problems	No	Low
Viewer	Coverage Calculated Incorrectly for Manually Adding Mutations for WGA	No	Low

Version 2.3.4.1

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Mutation Call	Mutation Filter additions for indels	Yes	High
Track Manager	dbNSFP 2.1 support	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Autorun	AutoRun/GA issue with hyphens	No	Low
BAM	BAM Conversion crashes or fails when sorting large BAM files, and is too slow	No	Low
Barcode Sorting	Barcode Sorting Improvement - for similar tags	No	Low
Expression Report	Continuous mRNA/CDS display Gene issue	No	Low
Format Conversion	XSQ Format Conversion bug	No	Low
Mutation Report	Ambiguous Gain/Loss Export Fix	No	Low
Mutation Report	Mutation Report filter issue with a Gene Text File - only one instance of gene that appears in multiple locations is kept.	No	Low
Mutation Report	Mutation Report Filter by Annotation Issue with Track memory	No	Low
Post-Processing	Summary Report issue with many post-processing reports	No	Low
Reports	"Set Incremental Segment Length" issue with contigs and chromosome coordinates	No	Low
Sequence Operations	Complex Trimming issue fix	Yes	Medium
Sequence Operations	Remove Duplicates for Paired End Requires name recognition issue	No	Low
Variant Comparison	Variant Comparison Issue	No	Low
Variant Comparison	Filtering by Reported vs. Unreported in dbSNP in the Variant Comparison Tool Doesn't Work	No	Low
Variant Comparison	Variant Comparison - Filtering by dbNSFP not working	No	Low

Version 2.3.4

Added and Updated the Following Features

Feature	Description	Algorithm Change	Risk
Alignment	Detect Large Indels Memory Management	Yes	High
Autorun	Load Projects in AutoRun to generate Post-Processing Reports	No	Low
BAM	Output Project in BAM for Paired Reads	Yes	Medium
BAM	Load Projects in AutoRun to generate BAM file	No	Low
Build Reference	Add option to Build Preloaded Reference to prevent merge of overlapping amplicons	Yes	Medium
CNV	Add graphics to CNV Tool	No	Low
CNV	New Dispersion-HMM CNV Method	Yes	Medium
CNV	Add CNV 'Block Report'	No	Low
Expression Report	Add "FPKM" to the Expression Report for Paired-end Data	No	Low
File Formats	Support GZIP Input	Yes	Medium
Filtering Settings	Improvements to the "Annotation" Filter tab, including noncoding filter	Yes	Medium
Geneticist Assistant	Interface with Geneticist Assistant	No	Low
HLA	HLA Allele Call Improvements	Yes	High
Mutation Report	Calculate Allele Frequencies for Add Mutation Dialog	No	Low
Performance	Increase Speed of Alignment and Mutation Detection	Yes	Medium
Post-Processing Reports	Add Distribution Report Save Coverage, Save Consensus, Save SNP Consensus	No	Low
Post-Processing Reports	Save PDF/TXT of sub-reports	No	Low
Post-Processing Reports	Automatically create Summary Report PDF with Project	No	Low
Post-Processing Reports	Save Target Region Statistics with Coverage Curve	No	Low
Post-Processing Reports	Post-Processing Reports no longer require Reference/Chromosome position	No	Low
Project Files	Decrease project size with an Option to Link annotation files to project folders	No	Low
Project Wizard	Project Output location automatically updated	No	Low
Sequence Operations	Sequence Trimming - Advanced Settings - Option to opt out of Primer dimers/trimers check	Yes	Medium
Sequence Operations	Add stringency flexibility for Remove Duplicate Reads	Yes	Medium
Track Manager	Support additional SNP Tracks	No	Low
Track Manager	Add VCF support to Track Manager	No	Low

Track Manager	Support dbNSFP 2.0	No	Low
Track Manager	Show all Tracks in Main Display	No	Low
Track Manager	Support Exome Variant Server (ESP)	No	Low
Track Manager	Support COSMIC in VCF Format	No	Low
User Management / Tracking	Add User Management to NextGENe	No	Low
Variant Comparison	Add mutant allele frequency column	No	Low
Viewer	Hovering over variants in aligned reads shows statistics, hold Ctrl to highlight current read	No	Low

Corrected the Following Bugs

Feature	Description	Algorithm Change	Risk
Assembly	Floton assembly issue with sorted data	Yes	Medium
Autorun	Loading .fq files in AutoRun	No	Low
BAM	Mutation Report missing some insertions and deletions with BAM input	Yes	High
BAM	BAM error with NextGENe Viewer's Export BAM for Reverse Complement GBK	Yes	Medium
Coverage Curve	Coverage Curve Report missing single base regions that fall below threshold	Yes	Medium
Export	Gap Fasta Export fix	Yes	Medium
Mutation Call	Proper export of Insertions/Deletions in VCF	Yes	High
Mutation Report	Mutation calls for deletions across exon boundaries	Yes	High
Mutation Report	Allele Frequency Missing from VCF Output	No	Low
Mutation Report	Using Reference Re-created by BED File - Includes False Variants at Ends of Segments	Yes	High
Sequence Operations	Sequence Operations Configuration File discrepancy fix	No	Low
Sequence Operations	Remove Duplicates removes too few reads with PE Fastq files	Yes	Medium
Settings	NextGENe Viewer error when opened for first time on computer without NextGENe	No	Low
Setups	NextGENe Viewer "BamApi.dll not found" error on Windows Server2008	No	Low
Variant Comparison	Variant Comparison Settings File saving bug	Yes	Medium

Version 2.3.3

Added and Updated the Following Features:

- Enable LSM to display Client User Information

Corrected the Following Bugs:

- GBK Reference fetching wrong dbSNPs
- Format Conversion Trim Bug
- Transcriptome Alignment- Unmatched Reads for Paired-End Data

Version 2.3.2

Added and Updated the Following Features:

- Format Conversion
 - Format Conversion compatible with Default output of Torrent Suite v3.4
 - Make BAM format conversion work with Paired Read data
- Accept BAM files as input for alignment
- Sample File Tools
 - Support FASTQ files in more Sequence Operation tools
 - Create Log File for Remove Duplicates of Sequence Operations
 - Remove Duplicates checks Reverse Complement between Paired Reads in Sequence Operations
- Automatically query tracks for GBK references
- Hide Unmatched Ends checks BWT Allowable Mismatched Bases
- Viewer Changes
 - Reports
 - Mutation Report
 - Mutation Call made to all transcripts
 - Save Filtered VCF (only include variants that passed all filters)
 - Add mutant allele frequency column
 - Link Summary Report positions to Main Graphical Displays

- Peak Identification Report Improvements
- Coverage Curve Report's Target Region Statistics - add "Aligned Reads(Including Ambiguous Locations)"
- Add hotkey shortcuts for Variant Comparison Tool
- Add Option to Export SV Paired Reads

Corrected the Following Bugs:

- Alignment of a read at a segment junction should should only be to one segment
- VCF Formatting Bug for Insertions and Deletions
- Unknown Chromosome Bug
- CNV Report "Allele" numbers fix
- AutoRun Format Conversion/Network Output Issue
- BED Format zero-based indexing
- Negative/Deleted Mutations Shown in Mutation Report
- Preferred transcript not always used
- AutoRun with Sequence Operation for Paired Data only aligns 1 file
- Paired Reads Detect Large Indels Memory Leak fix
- Some COSMIC IDs not retrieved with v37_3 reference
- Ability to save the Transcript report was lost
- Some COSMIC IDs not retrieved with v37_3 reference
- Other minor bug fixes

Version 2.3.1

Added and Updated the Following Features:

- Add CNV-Seq Application
- Summary Report to NextGENe Viewer
- HLA best match and Display Improvements
- Sample File Tools

- Update Sequence Operations format inputs and Trimming capabilities
- Add Dual-Barcode Sorting option
- Alignment Improvements
 - Add a frequency setting for balance filters
- AutoRun Changes
 - Add Sequence Operations to AutoRun
 - Add a "Duplicate" option to AutoRun's Job File Editor
 - Add "Split Job" option in AutoRun to create multiple projects when input is multiple sample files
- Change Floton Settings in the Project Wizard
- Viewer Changes
 - Add Drop-Down list for the Next and Previous Options
- Mutation Report
 - Add COSMIC ID descriptions
 - Add option to Save Consensus Sequence Relative to Mutation Report Filtering
- Expression Report
 - Display Amplicon ID in Expression Report
 - Display Accession Numbers in Expression Report
- Variant Comparison Tool
 - Check allele counts for negative mutations improvements
- Coverage Curve Target Region relative to ROI in GenBank
- Output BAI with BAM
- Export Sequences using a list of positions

Corrected the Following Bugs:

- NextGENe AutoRun with Format Conversion Creates *_Copy1 Output Folders
- "miRNA Trimming" doesn't trim partial adapters.
- Report coverage for individual segments/contigs in Coverage Curve

- Advanced GBK Editor can't see CDS for some GBK files
- Sequence Operations Trim By File not trimming close matches
- Recreated BED Index bug in Viewer
- Fix for ACE format in assembly
- Peak Identification Report Save Bug
- Some RNA Accessions wrong for Mutation Reports saved through AutoRun
- Viewer unable to load projects with many (~1000) reference files
- Expression Comparison Report - Region settings not applied
- Sequence Operation - many trim options were unavailable
- SOLiD data not pairing correctly
- Coverage Curve not displaying BED file description
- Variant Comparison "Default Settings" bug with Mutation Report Settings Bugs
- "Detect Large Indels" crashes NextGENe when used with Paired-end data
- Saved Mutation Report Missing Positions From BED File
- AutoRun Settings Bug
- Recreated BED Index Alignment Errors
- Alignment Crash bug due to file share violation
- COSMIC option unavailable in Variant Comparison
- Bam Reader - Crash due to User Account Controls
- Transcriptome Alignment Crashes with seed size 17 or lower
- Roche Homopolymer Trimming issue in Format Conversion
- Other minor bug fixes

Version 2.3.0

Added and Updated the Following Features:

- Sample File Tools

- Ion Torrent Format Conversion Improvements, keep more reads
 - Expand on FASTQ support in Format Conversion Tool and AutoRun
 - Support SOLiD XSQ Format
 - Additional Filtering feature, keep bases x to y
 - Add function to Import a list of sequences to trim to assist with Removing primers/adapters
 - Add miRNA trimming option, better handling of variable adaptor lengths
 - Remove Duplicates Improvement, input FASTQ, rather than only FASTA
 - Improvements to Overlap Merger Tool for Ion PGM data
 - Add HLA Tool
- Flotom Assembly
 - Cutoff option updated
 - Trim function added
 - Coverage Normalization option added, Decreases processing time by ignoring reads, or low quality ends of reads, where coverage is above the set threshold
 - Improve Batch Processing's ease of use with Inspect Input Files and NGJOB file formats
 - Add Post-Processing setup to Project Wizard and AutoRun – to allow users to specify reports to have generated automatically
 - Add function to create reference subset of indexed genome from BED file
- Project Overwrite Check
- Alignment Settings' Balance Ratio Optimization
 - Increase alignment speed
- Viewer Changes
 - Mutation Report
 - Add Mutation Report Summary
 - Import COSMIC track to Mutation Report
 - Filter by VCF file

- Show all positions in ROI
- Variant Comparison Tool
- Add VarMD export option to Variant Comparison Tool
- Add Gene Association Comparison method to Variant Comparison Tool
- Add Gene Drop-down list for quick filtering of variants
- Increase speed
- Improve output by including project name in each header column and saving a settings file
- BAM/SAM improvements
- Select Chromosomes
- Save header
- Expression Report
- Expand the Expression Report grouping options
- Limit improvement when coverage is greater than 65K
- Improved Viewer navigation
- Display Primer information in Viewer
- Improved Viewer display for fasta references
- RNA-Seq Display improvements
- Filtered mutation calls tick marks now gray
- Distribution Report display region relative to BED file

Corrected the Following Bugs:

- AutoRun stalls when license is about to expire
- RNA-Seq crash with PE data
- Transcriptome - Remove from Parameters File Settings that aren't in Project Wizard
- "Number of Unmatched Bases" doesn't include insertions
- Alt Splicing VS Alt Transcript Start/Stop fix
- Inaccurate background highlighting in SOLiD data

- Sequence trimming bugs (Barcode Sorting, Format Conversion, and Sequence Operation)
- Remove Duplicates for Fastq problematic with new Illumina format
- HGVS Nomenclature bug at 3'UTR
- Format Conversion Instrument type gets inadvertently changed
- Multiple base reported deletions are recorded as a single position, but need to be recorded at each position
- VCF Contig Header Bug
- Other minor bug fixes

Version 2.2.2

Corrected the Following Bugs:

- NextGENe AutoRun Not Using Format Conversion Settings File
- Coverage Curve not including regions at Highlight Coverage Threshold
- Conversion of CSFASTA > FASTA Removed Reads bug
- Increase speed of MiSeq barcode sorting
- Overlap Merger not Using Overlap Min Bases Setting
- Variant Comparison Shows Incorrect Annotation for Complement Genes
- Saved Mutation Report with ROI text file differs from report in viewer
- SFF files are read incorrectly during format conversion
- Mutation Calls for Some Coding Variants Shown as IVS
- Mutation Calls include Reference Positions
- Other minor bug fixes

Version 2.2.1

Added and Updated the Following Features:

- Add Capture Quality Metrics and Plots to the Coverage Curve Report
- Add VCF Export to Mutation Report

- Barcode Support for Single-Indexed MiSeq sample files
- With 2 Mutation Calls Show both Amino Acid Changes

Corrected the Following Bugs:

- Opening Old Projects - gbk refs, reverse genes mis-annotated
- BAM Reader Automatching Fix
- Barcode Sorting Bugs
- SAM Export Access Violation Error
- Convert dbNSFP Tool errors - language settings
- Arrange Paired Reads bug for large files
- Paired Read Alignment Causing some extra Unmatched reads
- BED File Format Bug, ChromEND should not be included in region
- Merge Files fails with large files
- Mutation Calls for Some Coding Variants Shown as IVS
- Mutation Excluded for Large Datasets
- LSM NextGENe Viewer Licensing Issue
- Exp Rpt - Min & Max Counts Columns are inaccurate
- Reads not shown in the viewer
- NextGENe AutoRun Not Using Format Conversion Settings File
- Querying dbNSFP Issue for Complement Genes GBK Files
- AutoRun fails to load barcode settings file
- Indel Alignment Bug
- Mutation Calls Sometimes Incorrect for Mutation in 1 gene, closest CDS is in another gene
- Other minor bug fixes

Version 2.2.0

Added and Updated the Following Features:

- Distribution Report - Add Save Coverage for Entire Reference Range option
- Barcode Sorting Tool - Paired Read Support
- Add Mutation Call Nucleotides Relative to Strand or Gene Direction
- Display SAGE Report as Main Viewer report for SAGE projects
- RNA-Seq Improvements
- Add Advanced Settings for Barcode Sorting
- AutoRun Interface Improvements
- Query Annotation of Preloaded Reference When Using GenBank References
- Add Flon Assembler for Ion PGM data assembly
- Add Ion Paired End Support

Corrected the Following Bugs:

- Coverage Curve Report is Incomplete
- Mutation report - selecting mutation doesn't move top view
- Runtime Error Due to Allowable Mismatched Bases
- Paired Read Statistics Fix for SOLiD Paired End Data
- Transcriptome Project Setup fix for fasta reference files vs Alternate Splicing method
- A reverse complemented GBK should not be adjusted to Plus orientation
- Barcode Sorting Fix for tags identical in reverse complement
- Viewer Registration Issue
- Original Allele settings with Elongated Data could show Wrong Amino Acid Change for heterozygotes
- Variant Comparison- Minimum Coverage Setting conflict with Mutation Type Comparison Method
- Viewer has difficulty loading and navigating fasta projects
- Wrong Allele Percentages in Mutation Report for Consolidation's Original Sequences
- SIFT Output for Het. Mutations

- Remove Duplicate Reads removing some unique reads
- SIFT Output for Indels broken into multiple lines
- Show GB file types in Advanced GBK File Editor
- Format Conversion Crash
- Expression Report - using BED file, discrepant # of lines
- AutoRun does not recognize Version 1 NGSettings
- SOLiD WrongAllele Score 0.0 after Position 2GB
- Unicode Support - Barcode Sorting - all reads sent to Other Tags File
- Amino Acid Change does not agree with Mutation Call
- Sort References alpha numerically
- Mutation Report's Splice Sites selection bug with filtering calls after chr11
- PE Crash Fix
- Incomplete BAM file generated
- BAM File Export - EEFACE error for large projects
- Other minor bug fixes

Version 2.17

Added and Updated the Following Features:

- StatInfo.txt file written at each new step, not just at end of project
- Update Database Settings Dialog
- Variant Comparison Tool Sort Significant Variations to Top of List
- When "Load" Reference is selected for one of the Indexed Genomes, Alignment Algorithm updates
- NextGENe Autorun Format Conversion- Ion PGM added
- Specify number of cores for Processing in Project Wizard
- Barcode Sorting - Barcodes at 5' End Only Option
- Variant Comparison Interface Updates

Corrected the Following Bugs:

- Mut Rpt using BED file with overlapping regions causes negative mutations
- Scrolling through large genes (TTN) takes a long time
- Barcode Sorting is Overtrimming
- Elongated Paired End Files Not Recognized as Paired (SOLiD)
- CSFASTA > FASTA Conversion not working
- Barcode Sort issue when one is reverse complement of another
- Negative Values in Format Conversion Log
- SIFT Report Saves Homozygous Mutations Incorrectly
- Other minor bug fixes

Version 2.16

Added and Updated the Following Features:

- BAM Reader Improvements - Add an interface that allows users to see which chromosomes and contigs are able to match perfectly between the BAM file's references and NextGENe's genome references
- Ion PGM Version - NextGENe PGM version has defaults ideal for PGM users
- Mutation and Variant Comparison Reports - Inclusion/Exclusion Regions of Interest Lists
- Add Ambiguous Gain and Loss Scores to Mutation and Variant Comparison Report to assist with call confidence
- Include dbNSFP information to assist with assigning functional predictions
- Add "Check Allele Counts" to show this information in Variant Comparison Report for negative mutations
- Transcriptome Analyzer Improvements - Setup Transcriptome Projects from the Project Wizard, Single Strand Sequencing Improvements
- New Illumina Fastq- PE Read Names and Quality File Format Change to be supported
- Add Rare Disease Module to Variant Comparison Tool - This tool allows users to filter mutations relative to Family Relationships and Phenotypes

Corrected the Following Bugs:

- Batch Processing does not read Version 1 Settings Files
- Chr Positions Displayed wrong when multiple NC_ GBK files are used
- Issue reading Barcode identifier in SOLiD PE Data Files
- dbSNP number not shown in Mutation Report for some GBK reference files
- Other minor bug fixes

Version 2.14

Added and Updated the Following Features:

- Ion Torrent enhancements
- Read alignment improvements
- Delete Small Homopolymer Indel improvements
- Transcriptome
 - Alignment Algorithm Improvements
 - Detection Improvements
 - Alternative Splice Site Detection Improvements
 - Multiple Transcript Detection Improvements
 - Fusion Gene Detection Improvements
 - Single Project saved for Transcriptome Projects, merging the ability to create reports for Mutation Calls, Expression Results and Transcript Identification into a single project
- BAM Format
 - Improvements to reading of BAM files
 - Improvements to exporting of BAM files
- NextGENe AutoRun
 - Added the "NextGENe AutoRun" tool to launch NextGENe jobs automatically
- Load Alternate Gene information for Whole Genome Alignments
- Updated Manual

- Updated Help
- Registration and Security Validation Improvements related to virtual servers
- License Server Manager improvements
 - Registration and security validation improvements
 - Additional Products
- Reference Genome Improvements
 - Annotation Database can now contain multiple transcripts (such as human build 37.2)
 - Preferred transcript is assigned as one with the longest CDS. Annotations in the Viewer and Mutation Report will be displayed relative to the Preferred transcript.

Corrected the Following Bugs:

- Variant Comparison Report Updates
 - Variant Comparison Gene and Chr field was previously blank when first project was negative for variant.
 - Annotation information now shows in the variant comparison report even if the mutation is absent in the first project
 - The "display as normal" option for the variant comparison report now sets the reference allele to 100% rather than 100 counts for absent mutations
- Mutation Report Updates
 - Viewer Mutation Report's Negative SNP reference nucleotide was previously wrong.
 - dbSNP number previously not shown in projects with some GBK reference files
 - Substitution/Indel filter prioritization improvements
 - Other minor bug fixes

Version 2.10

Added and Updated the Following Features:

- Forensic Data Analysis Support of STR – Homopolymers and repeats are handled differently. When identifying the number of short tandem repeats, reads that contain tags at both the 5' and 3' end of read are required. Alignment of these reads is improved, which aids in accurate interpretation of expression levels with NextGENe Viewer's Expression Report.

- Transcriptome Analyzer – When aligning to SoftGenetics’ human build 37 database that contains all transcripts for each gene, NextGENe now has improved alignment to each exon. NextGENe maps reads to exon-exon junctions as well as novel exons. Reporting is available that identifies the expressed transcripts, as well as novel transcripts and alternate splice variants.

- Output Improvements:

- Paired Read Reports – Additional columns of information, like gene names and chromosome numbers, enabling more sorting and filtering options.

- Distribution Report – can load Bed File to view statistics

- Coverage Report – can load a Bed File of Regions of Interest, and can adjust the Coverage Threshold after alignment has already been completed.

- Expression Report – In addition to choosing to report expression levels relative to Gene Start/End positions, you can now report relative to each CDS Start/End or Region of Interest Start/End. Also, there are additional columns in the report, including Chromosome, Contig, CDS – improving sorting options.

- Structural Variation Report -there are additional columns in the report, including Chromosome and Gene – improving sorting options.

- Added “Score Distribution Report”

- Mutation Report

- Mutation Score improvements, especially with projects where Condensation was used

- Additional columns available, including Gene Direction, RNA Accession number

- Additional filtering available – substitutions vs. Indels

- Save Consensus (Nucleotide) Sequence – can specify range relative to text or Bed files or relative to start/end position.

Corrected the Following Bugs:

- WGA Memory allocation

- Format Conversion

- Other minor bug fixes

Version 2.01

Added and Updated the Following Features:

- Assembly

- For 454 Assembly "Save Original Sequences with Assembled Ones" - change output file name to AssembledContigsWithOrg.fasta and add read names
- Long PE Assembler Mapping tool was ignoring minus signs for reverse contigs
- Mutation Report
 - Mutation Scoring - when elongation is used, allow users to deselect "ignore F/R balance" and "ignore allele balance"
 - Output Points of Interest - filter by mutation report settings applied to scoring filter settings
- Remove Duplicate Reads Tool added to Sequence Operations to remove duplicate reads.

Corrected the Following Bugs:

- Expression Report
 - Original coverage values can now be saved when consolidation was used.
 - Fix N/A in fields upon first opening the report
 - Fix resort/save bug with description column
- Mutation Report
 - Output Points of Interest BED file fix.
 - Increase speed for saving mutation report for All chromosomes, especially an issue with build 37 of human genome.
 - Fix Save Consensus
- Assembly crash fix when PE Assembly is used with "save original sequences with assembled ones" selected
- Modify Register.exe
- Other minor bug fixes

Version 2.00

Added and Updated the Following Features:

- Support for Human Genome Build 37
- Mutation Report – filter by score
- SNP Comparison – now called Variant Comparison

- mutation score shown
- filter by score

Description of Risk Categories

- Low – cosmetic; such as changes to the graphic user interface and reporting options (no algorithm change).
- Medium – includes minor algorithm changes that may affect sample grouping and final reports.
- High – includes significant algorithm changes that may result in alignment and mutation call detection differences.