Alamut™ Visual Plus version 1.12
December 16, 2024

Don't miss out! Click on the blue "UPDATE" button today to unlock exciting new features and enhancements designed to elevate your variant interpretation experience.

What's NEW?

New Mitochondrial Variant Annotations: MITOMAP and MitoTIP

- MITOMAP - This human mitochondrial genome database uses the mtDNA sequence as the unifying element for bringing together information on mitochondrial genome structure and function, pathogenic mutations and their clinical characteristics, population associated variation, and gene-gene interactions.
  It is now available as a track and in the mitochondrial variant annotation panel.
- MitoTIP – MitoTIP scores predict the impact of novel mitochondrial tRNA mutations.
  These are now available in the variant annotation panel and as a tooltip when hovering over the track.

New Splicing Predictor: SpliceAI Lookup

- The SpliceAI Lookup tool developed by Broad Institute is now accessible from the Annotations tab under the protein level variant features, where clicking on the button opens a pop-up window that displays scores for the given variant.

UPDATES

ACMG Improvements

- The ACMG criteria strength suffix has been added in the ACMG dialog suggested/selected labels.
- The ACMG rule strength used by each user is now saved in the local variant database (LVD).
- All categories of user-defined rule strengths are now displayed in the variant classification.

Occurrence Management Updates

- A new “Created By” column has been added to the occurrence tab.
- There is a new occurrence management system, whereby the unique identifiers for a variant are the occurrence ID + Family ID. Remaining columns (Phenotype, HPO IDs, RNA Analysis, Comment, Created/Created By and Updated/Updated By) are independent of the variant

occurrence and any changes in those fields will not impact other variants linked to the same occurrence.

Database Version Updates

Database updates include:

- ClinVar updated to 2024-10-09
- Mastermind updated to 2024-10-02
- HGNC updated to November 2024
- RefSeq transcripts updated to November 2024

Small Improvements

- The Variant Panel layout has been improved for large insertion variants, by wrapping the long string of nucleotides in one box. In addition, long cDNA strings are now displayed on multiple lines in the Report tab.
- You can now view the total number of introns or exons in the Variant Panel, when applicable.
- Changes to splicing prediction options and flanking region size are now saved when the flanking region size is modified at the bottom of the "Splicing Prediction Options" dialog and the "Apply" button is clicked when generating the splicing report (accessed via the "Report" button in the splicing view).
- Rs-number searches that were not properly returning gene annotations have now been fixed.
- The Variant nomenclature algorithm version has been updated for variants within the -8 or +8 regions of the codon, which are now assigned "p.?", while other intronic variants remain assigned "p.(=)".
- When closing Alamut™ Visual Plus, if any Variant Panels are open with unsaved changes, a warning will be displayed. If several Variant Panel tabs are open, the warning will be displayed only once.
- It is now possible to close all tabs at once by right clicking on any tab.
- The automatic pop-up indicating the successful opening of a BAM file has now been removed.
- The GeneSplicer value has been added to the splicing variant Report as a percentage, similar to MaxEntScan, SSF, and NN splice.

For additional details on these updates, please refer to the extended release notes.

Please consult our regularly updated User Manual to discover more about the new and updated features mentioned in these Release Notes.

To enjoy all the benefits of Alamut™ Visual Plus and stay supported, we recommend upgrading to the latest version. It's quick and easy—just click the blue "Upgrade" button in your software!

Keep in mind, starting January 1st, 2025, versions below 1.11 will no longer receive bug fixes, technical support, or updates. But don't worry—upgrading now means you'll be ready to take full advantage of everything we've improved.

Please contact our support team with any questions via the Customer Support Portal.