

Release notes

New Features in Alamut® Visual version 2.8

• Alamut® Visual version 2.8.1 (13 July 2016)

- This minor release fixes a bug in 2.8.0 whereby stop loss variations could cause software crashes.

• Alamut® Visual version 2.8.0 (15 June 2016)

- **New feature:** An integrated access to the Ranomics functional datasets. **Ranomics** is a startup company that provides comprehensive functional data from biologically relevant high-throughput functional assays for variant classification. Ranomics' first comprehensive dataset includes 2000 genetic variants within the BRCA1 N-terminal RING domain (2-100). This dataset was built by experimentally testing the E3 ligase and BARD1 RING domain binding functionalities of each variant. Each variant is assigned a functional score that provides >99% sensitivity and specificity for predicting the functional outcome. Ranomics functional variant assessments are available in Alamut Visual 2.8.0. To activate data access you will need to acquire a separate subscription from Ranomics (see <https://app.ranomics.com/signup>) and then to check the Ranomics option in Alamut Visual (Tools > Options > Misc > External Functional Data).
- **New feature:** Terms "variant" and "variation" are now used throughout the software, instead of "mutation" or "polymorphism"
- **New feature:** New 'VariantValidator' button in the variant window allows to easily check variant descriptions with VariantValidator
- **New feature:** Improved color-coding for HGMD variants (disease causing: red - likely disease causing, not disease-associated frameshift or truncating: yellow - polymorphisms: green)
- **New feature:** Improved HGVS protein-level variant descriptions
- Minor bugs are fixed.