

Man (OMIM),⁷ via the ClinVar database. Resources such as dbSNP,⁸ the Ensembl 1,000 Genomes Project,⁹ and Exome Variant Server¹⁰ provide information about the occurrence and frequencies of variants within a population. By delivering rapid and thorough annotation processes, VariantStudio software empowers researchers to identify biological significance in variant data.

Flexible Filtering Options

Exome and whole-genome sequencing often detect large numbers of variants per sample, requiring further analysis to identify the few variants associated with a given phenotype. Virtual subpanels from these broad sequencing assays can be generated within VariantStudio software, enabling the push-button creation of a broad sequencing portfolio. Using a cascade of filtering options, researchers can rapidly isolate the key variants that are consequential to the phenotype under study. For ease of use, VariantStudio software offers commonly applied filters, including variant quality, frequency, functional impact, and known disease association, which are easily accessible through the user interface (Figure 3). To maximize flexibility, the VariantStudio tool also enables users to filter variants based on any available associated information, including user-imported custom annotations. For further convenience, this application offers the option to construct workflows by saving concatenated filters, enabling researchers to standardize workflows and streamline analysis.

In addition to single-sample filtering, VariantStudio software enables multisample comparisons that accelerate identification of causative variants. Somatic mutations can be quickly identified in tumor-normal pairs by filtering for variants that are present in a tumor sample but absent in its normal counterpart. To support family-based analyses of inherited disease, this software application provides a collection of filters that identifies variants consistent with specified inheritance modes and patterns of disease progression. Using the VariantStudio tool, researchers can confidently isolate causative variants underlying inherited diseases and traits.

Streamlined Variant Classification

A critical component of translating genetic information into meaningful biological insight is determining the impact of identified variants within the context of observed phenotypes. The classification feature in VariantStudio software facilitates this process, enabling clinical researchers to combine their expertise with provided annotations to categorize variants. Classified variants and associated information entered by the user are saved to a local database. The classifications can then be applied easily to the same variants when they are observed in other samples (Figure 4). Researchers can enrich this database further by importing classified variants from external sources. By enabling automatic recording, tracking, and management of classified variants, VariantStudio software simplifies and accelerates data interpretation.

Customizable Reporting

Concise and effective reporting is critical for isolating actionable information from complex data. The VariantStudio application provides powerful and flexible report generation capabilities to facilitate variant reporting. Researchers can create and store multiple templates to optimize reporting for different disease areas. When a template

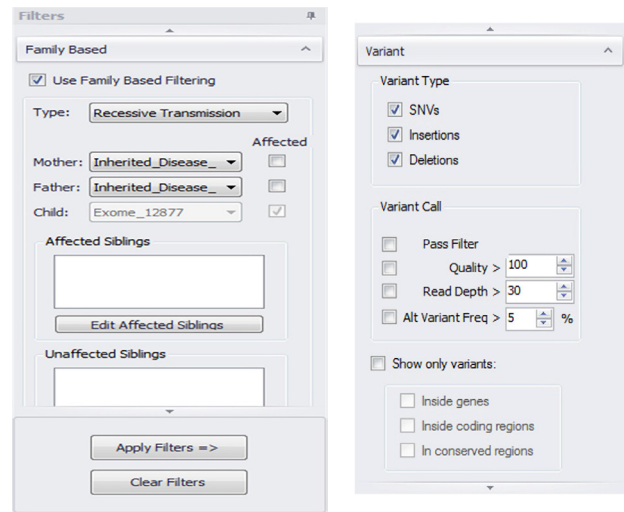


Figure 3: Comprehensive and Customizable Filters—Flexible filtering options enable users to filter variants based on inheritance mode, quality, frequency, functional impact, or custom annotations.

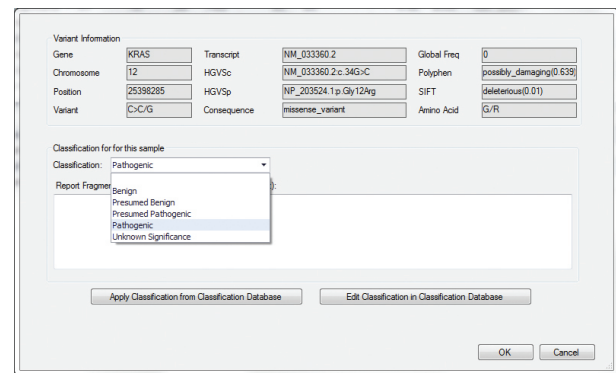


Figure 4: Information Storage Streamlines Interpretation—The VariantStudio tool stores classified variants and associated notes in a local database, making the information easily accessible for future analysis.

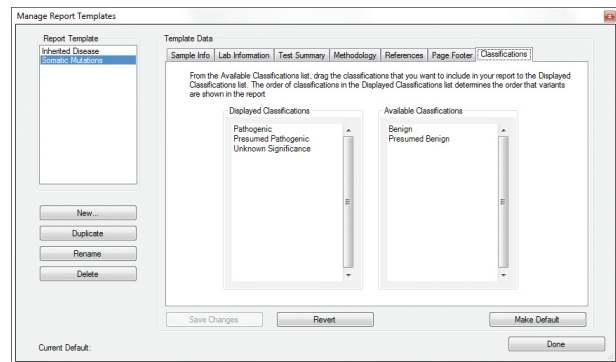


Figure 5: Templates Enable Customizable Reporting—Guided template generation and addition of sample-specific information enable customized reporting for different clinical research areas.

