Ingenuity® Variant Analysis™

For identification of causal variants from resequencing data

Ingenuity Variant Analysis combines powerful analytical tools and unparalleled content from the Ingenuity Knowledge Base to rapidly identify the most compelling disease variants in human sequencing data. Hundreds of samples can be analyzed in just hours, using selection criteria based on published evidence and knowledge of disease biology.

Ingenuity Variant Analysis enables:

- Faster analysis in just a few hours
- Prioritization of variants along biologically relevant filter criteria
- Focus on variants demonstrated to be implicated in the phenotype of interest
- Filtering variants based on robust statistics for cancer, kindred, proband, and cohort studies

Powerful insights from the Interactive Filter Cascade

Variant Analysis compiles all gene variants within a dataset and enables this list to be quickly narrowed down through an interactive series of filters. This Interactive Filter Cascade can be adapted to reflect selection criteria of interest and their importance to the research question at hand (Figure 1).

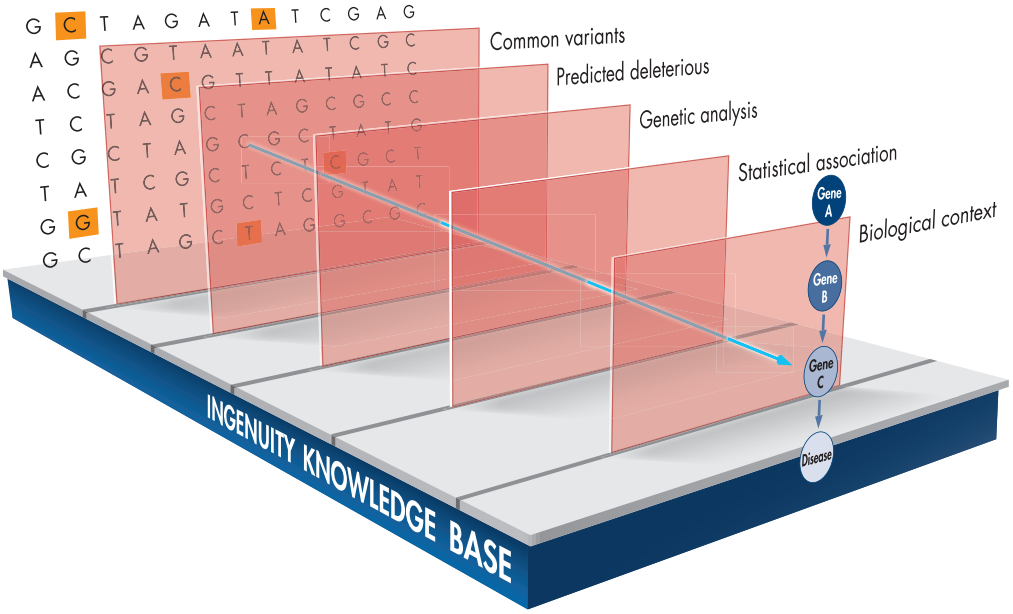

Figure 1. Rapid prioritization of variants. A list of gene variants relevant to an analyzed dataset can be drilled down to those most relevant to the research question by defining a series of filters that reflect the most important selection criteria.

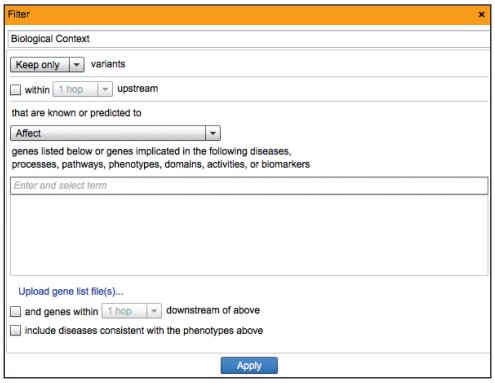

Figure 2. Interactive filters to zero in on biologically interesting variants. Flexible and interactive filters allow you to rapidly narrow down variant lists to those that are relevant to your research question.

Interactive filtering to deliver impactful results

Filters of the Interactive Filter Cascade are based on information in the Ingenuity Knowledge Base, including manually curated primary literature on human mutations in patients with particular diseases or abnormal phenotypes. These filters offer a broad portfolio of selection criteria that go above and beyond identifying variants that impact symptoms, pathways, processes and genes implicated in drug response or disease progression. Additional biologically relevant filters (Figure 2) allow, for example, identification of diseases consistent with clinical features and deleterious variants in medical genomes, or use of single and bidirectional statistical burden tests to find genes or pathways with significantly more deleterious variants in one study group compared to another. Variants can also be filtered according to quality considerations such as call confidence and PASS status from upstream variant calling pipeline and read depth, and can be selected for consistency with Mendelian inheritance patterns.

Integrated content for fast and accurate insights

Variant Analysis leverages an industry-leading knowledge base that has been actively augmented and curated for over 15 years. This knowledge base includes nearly 4 million manually curated findings from the literature and information on mutations from databases such as COSMIC, OMIM, and TCGA. Downstream information includes known and inferred effects on protein function resulting from sequence variants in coding and regulatory regions, copy number variation, and fusions. Additionally, our novel integrated cancer disease models enable rapid identification of driver variants.

Ingenuity Variant Analysis is intended for molecular biology applications. This product is not intended for the diagnosis, prevention or treatment of a disease.

For up-to-date licensing information and product-specific disclaimers, see the respective Ingenuity product site. Further information can be requested from support@ingenuity.com or by contacting your local account manager.

Learn more and streamline your analysis of variants! Visit www.ingenuity.com/variants.

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