MutationForecaster®

A comprehensive genomic variant interpretation suite to interpret and validate mutations affecting mRNA splicing and protein coding. It finds and validates mutations that other software cannot.

Are you finding all the mutations in your genomic sequence data?

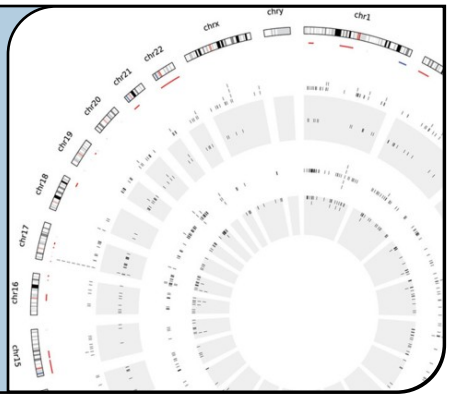
Mutations that affect splicing are among the most common causes of Mendelian disorders.

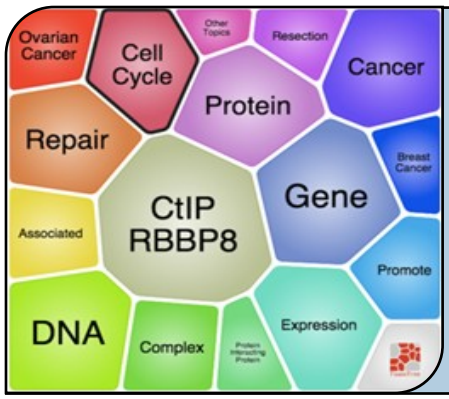
Locate novel splicing variants on a Genome-scale

The popular Automated Splice Site Analysis and Exon Definition (ASSEDA) server has set the standard for splicing mutation analyses worldwide over the last decade. It utilizes information theory-based variant interpretation methods and been cited in hundreds of studies. Potential splice isoforms are selected using a combination of donor/acceptor binding site strength and exon length. Constitutive, cryptic, and regulatory mutations can be interpreted using ASSEDA.

The Shannon Human Splicing pipeline accelerates ASSEDA's core functionality to a genome-scale. A VCF file containing 15 million variants can be examined in approximately 30 minutes. Variants are filtered, resulting in a tractable number of potentially deleterious mutations.

In 2014, Dorman et al. used the Shannon pipeline in combination with the validation software described below to discover and validate 988 splicing variants in 442 breast cancer tumour and matched normal exomes.



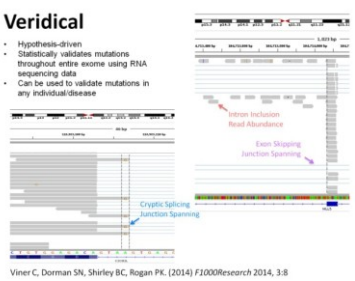


Start to finish variant interpretation

MutationForecaster® is a "one stop shop" for variant interpretation. While we specialize in splicing mutations, we have incorporated other tools which work together to help provide a full picture of each variant. Ensembl's Variant Effect Predictor (VEP) reports the effect of variants on protein coding sequences. The Cytognomix User Variation Database is based on the well-known database software LOVD and allows results generated by MutationForecaster® tools to be stored and compared with other sources of genomic variation (dbSNP, ClinVar, Exome Variant Server, Pubmed, public locus specific LOVDs). CytoVA is a cytogenetic decision support tool which can locate peer reviewed literature assotiated with user specified genes or phenotypes. It can also process results of other MutationForecaster® tools to discover variants within the results known to be associated with a user specified HPO phenotype.

Validation

Integrated software Veridical examines RNASeq BAM files to validate mutations predicted to be potentially deleterious by the Shannon pipeline. Veridical compares normalized counts of sequence reads from mutant and control samples, eliminating the need to examine sequences manually.



Reporting

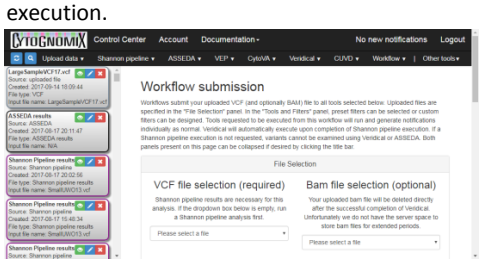
Results from any tool within the MutationForecaster® suite can be combined and examined in a single, interactive report interface. Variants are placed into overlapping subsets based on the results of all tools which have examined the variants. Clicking a subset causes the report to zoom in, revealing additional subsets.

Each variant subset can be included/excluded from a final report set. Selected variants can be exported to PDF format.

Interconnectivity

The "control-center" page is a hub within which you can upload genomic variants, specify settings for and execute any MutationForecaster® tool, and access results from the sidebar. Most tools run in the background without user oversight and notify you when they are complete.

Automated "Workflow" functionality allows multiple tools to run in sequence – or simultaneously where possible – with no user intervention required after the workflow begins execution.



MutationForecaster® homepage: mutationforecaster.com
Company site: CytoGnomix.com
Company e-mail: info@CytoGnomix.com

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