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.

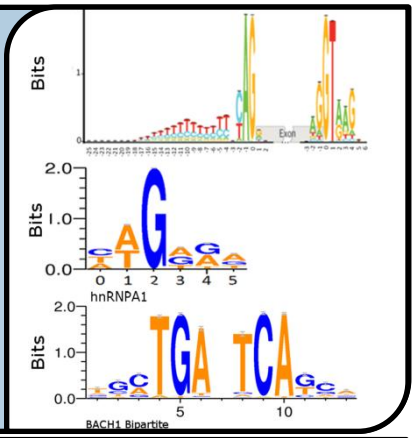
Are you finding all the non-coding mutations in your genomic sequence data?

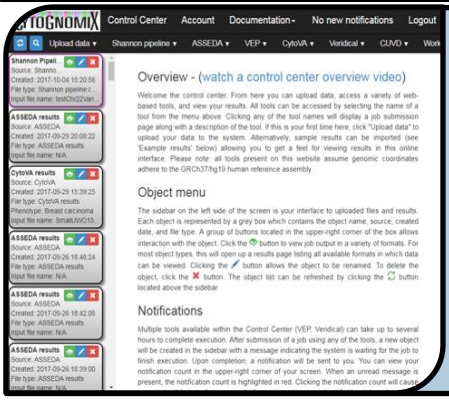
Mutations that affect splicing are among the most common causes of Mendelian disorders.

Interpret novel non-coding 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. New functionality has since been incorporated which examines splicing regulatory elements. ***ASSEDA*** utilizes information theory-based variant interpretation methods and has been cited in hundreds of published 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. Researchers 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 (Dorman et al., 2014, *Sci Rep*., 4:7063).



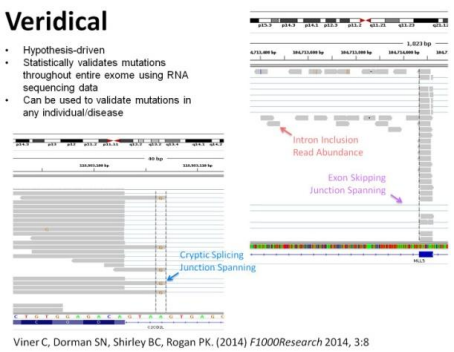


Start to finish variant interpretation

MutationForecaster® is a "one stop shop" for variant interpretation. While we specialize in non-coding 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 (CUVD)*** 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 associated with user specified genes or phenotypes. It can also process results of other ***MutationForecaster®*** tools to flag variants known to be associated with a user specified HPO phenotype.

Validation

Integrated software ***Veridical*** examines RNA-Seq 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.



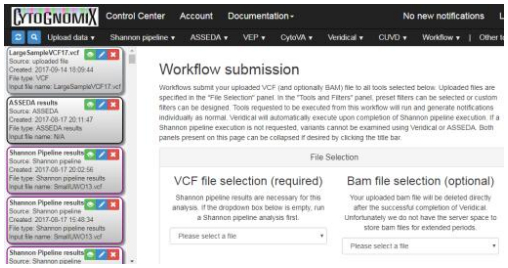
Viner C, Dorman SN, Shirley BC, Rogan PK. (2014) *F1000Research* 2014, 3:8

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 dynamically zoom in, revealing additional subsets. Each variant subset can be included or 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 and execute any ***MutationForecaster®*** tool, and access results from the sidebar. Most tools run in the background and notify you when completed. Automated "Workflow" functionality allows multiple tools to run in sequence – or simultaneously – with no user intervention required after the workflow begins execution.



MutationForecaster® homepage: mutationforecaster.com

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