

Functional consequences of genetics variants



Search our repository

Or [browse](#) the 5,848,740 variants on 1,048 proteins that have been analyzed and are predicted to be either pathological or neutral.

e.g. BRCA2, 6pax,
ENSG00000133110, Q04917

Search



Analyze your mutations

Enter a list of mutations on any protein or sequence, and find out their pathology score using PMut2015 predictor.

New analysis

Train your model

Train a specific pre-trained model with your own variants and get new predictions.

The understanding of the functional consequences of genetic variants is key to evaluate their influence in diseases, and their possible implication for diagnosis or the design of treatment.

Summary

Genomic variants are one of the objects of study of large biomedical projects. Initiatives like TCGA, or ICGC catalogues a large number of somatic variants found in cancer development that add to the known variants related to genetic diseases. The increasing availability of cheap DNA sequencing allow to envision the possibility to obtain personal genomes, allowing the adaptation of health care to individual features. The understanding of the functional consequences of such genetic variants is key to evaluate their influence in diseases, and their possible implication for diagnosis or the design of treatment. Tools available in this field include PMut, a well-known predictor for pathological effects of protein sequence variants. PMut is being updated and extended to include new patterns of conservation, and introducing protein flexibility and dynamics features.

Objectives

Evaluate the usability of flexibility and dynamics properties of proteins as a tool for predicting functional

consequences of sequence variants in proteins.

Barcelona Supercomputing Center - Centro Nacional de Supercomputación

Source URL (retrieved on 1 jun 2025 - 14:44): <https://www.bsc.es/ca/research-development/research-areas/bioinformatics/functional-consequences-genetics-variants>