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PERCH: ENHANCED GENETIC VARIANT PRIORITIZATION

DIAGNOSTICS

Software that integrates Bayesian analyses for enhanced prediction of genetic variants' pathogenicity.

TECHNOLOGY TYPE

Bioinformatics
Software

STAGE OF DEVELOPMENT

Commercially ready and
available for deployment.

LEARN MORE

Reference Number: U-6647

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TECHNOLOGY SUMMARY

There are multiple tools available to identify, score, and annotate genetic variants within a genome. However, the majority of these tools are restricted to high-penetrance genes for Mendelian diseases and can only analyze certain pedigree structures.

A University of Utah researcher has developed a framework for prioritizing genetic disease variants. This framework, Polymorphism Evaluation, Ranking, and Classification for Heritable traits (PERCH), predicts the pathogenicity of genetic variants better than competing methods. PERCH uses BayesDel, BayesSeg, BayesHLR, and BayesGBA to prioritize variants or gene sets. PERCH measures the biological relevance of each gene to the disease of interest, searching for disease susceptibility genes through whole-exome, whole-genome, or gene-panel sequencing data.

FEATURES AND BENEFITS

- Provides a more accurate disease variant pathogenicity score than competing tools.
- Can be used for disease gene discovery research and new rare variant association.
- Can be implemented in IARC guidelines or can be integrated into ACMG guidelines.
- Automates analysis by calculating every possible variant in the human exome.
- Enables a variety of study designs including case-control samples, extended pedigrees, nuclear pedigrees, or admixtures of the above.

RECENT PUBLICATIONS

Feng, B. (2017). PERCH: A Unified Framework for Disease Gene Prioritization. *Human Mutation*, 38(3), 243-251. doi: [10.1002/humu.23158](https://doi.org/10.1002/humu.23158)

INVENTOR PROFILE

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