CADD Scores

CADD is a tool for scoring the deleteriousness of single nucleotide variants as well as insertion/deletions variants in the human genome.

While many variant annotation and scoring tools are around, most annotations tend to exploit a single information type (e.g. conservation) and/or are restricted in scope (e.g. to missense changes). Thus, a broadly applicable metric that objectively weights and integrates diverse information is needed. Combined Annotation Dependent Depletion (CADD) is a framework that integrates multiple annotations into one metric by contrasting variants that survived natural selection with simulated mutations.

C-scores strongly correlate with allelic diversity, pathogenicity of both coding and non-coding variants, and experimentally measured regulatory effects, and also highly rank causal variants within individual genome sequences. Finally, C-scores of complex trait-associated variants from genome-wide association studies (GWAS) are significantly higher than matched controls and correlate with study sample size, likely reflecting the increased accuracy of larger GWAS.

CADD can quantitatively prioritize functional, deleterious, and disease causal variants across a wide range of functional categories, effect sizes and genetic architectures and can be used prioritize causal variation in both research and clinical settings.

For more information: http://cadd.gs.washington.edu/home

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References

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