

IN SILICO APPROACH TO THE IDENTIFICATION OF
REGULATORY POLYMORPHISMS WITHIN PROMOTER
REGIONS OF HOMO SAPIENS USING COMPARATIVE
GENOMICS, REGULATORY FEATURE AND SEQUENCE
COMPOSITION METRICS

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Advances in the computational identification of functional non-coding polymorphisms will aid in cataloging novel determinants of health. We have investigated the discriminatory properties of 68 literature-surveyed functional regulatory polymorphisms from our ORegAnno database (www.oreganno.org) compared to 346 polymorphisms of unknown function selected from the promoter regions of associated genes. We have utilize information derived from up to 23 different mammalian genomes and chicken combined with coexpression-selected library models and regulatory characteristics, like CpG island presence and distance to transcription start sites. Metrics are differentially supplied to a machine learning algorithm. Several disease-related genes have been selected and are being functionally tested at the time of writing. Software is being made available under the name Chum: Cis-acting human mutation modules from <http://www.bcgsc.ca>