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The medical genomics community is assembling vast, population-based and ailment-specific datasets in order to identify causative genetic variants and disease-associated genes. At present, scientists studying their own patient cohorts with such diseases must rely on indirect avenues to utilize the results from larger studies, e.g. use the ExAC/gnomAD database to look up variant population frequencies, and look up ClinVar for known pathogenic variants. Moreover, there are currently no tools that allow researchers and clinicians to answer subtle yet critical questions about patient variants in the context of the relevant, large research cohorts increasingly available to the community, e.g. whether a specific variant in the patient is enriched in a phenotypically similar subset of the large cohort.

To address this issue, we set out to create web-accessible, visually-driven software with a two-fold goal: to elucidate genetic variants enriched within a filtered dataset, and compare analyst-provided data to that of the enriched cohort. The cohort-gene.iobio application (http://cohort-gene.iobio.io) launches after the analyst selects a sample database, a gene or region of interest, and phenotypic filters for analysis. Within seconds of starting, cohort-gene.iobio compares sample variants to the selected dataset - user supplied or publicly available catalogs such as 1000G and ExAC - and generates annotations sourced from VEP, SIFT, PolyPhen, ClinVar, and others. Each group of samples is displayed within a single, cumulative variant track; variants are visualized to facilitate quantitative comparison between tracks, clearly and recognizably indicating enrichment in one cohort versus another. Additional annotation filters can be applied to variants while in the cohort-gene iobio application, and multiple genes or regions can be further selected for simultaneous analysis. To give the analyst an indication of the guality of called variants, coverage tracks are displayed when possible. cohort-gene.iobio also provides convenient reporting functionality, saving visualization information in PDF format, and variant information in VCF format. In conjunction with our other iobio applications, cohort-gene.iobio offers powerfully complex variant processing in a streamlined, intuitive interface, and stands to help close the gap between intricate genomics data and directed patient care.