***Variant Workbench Examples***

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This document is informed by the general narrative:

The general use case: identify variants of interest > identify subjects with those variants > examine patient-level characteristics (HPO terms, diagnoses, etc.)

link to notebooks: <https://cavatica.sbgenomics.com/u/jared.rozowsky/ashg-variant-workbench/analysis/cruncher/variant-workbench-starter-kit#>

# Example 1

Scientific question: Identify probands in the CHD study with variants of the TNK1 gene that have a HIGH VEP value.

Approach:

1. load consequences, and occurrences for the PCGC study
2. join consquences and occurrences
3. filter by gene name (TNK1)
4. filter by VEP
5. filter by is\_proband
6. examine resulting data frame

# Example 2

Scientific question: What are the phenotypic characteristics (HPO terms) of subjects with variants in the BSN gene?

Approach:

1. load consequences, and occurrences for the PCGC study, and HPO table
2. join consquences and occurrences
3. filter by gene name (BSN)
4. join phenotypes table, mapping patient\_id to HPO terms
5. examine resulting data frame