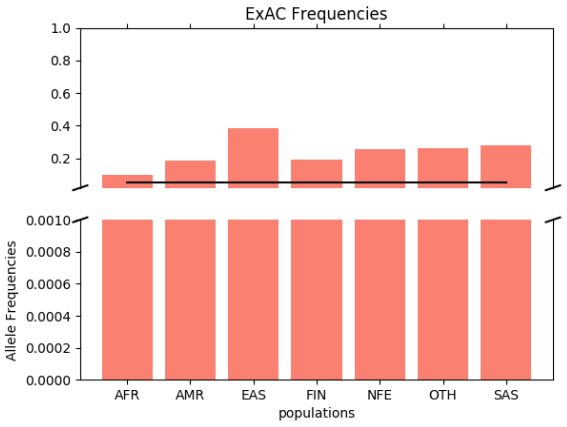
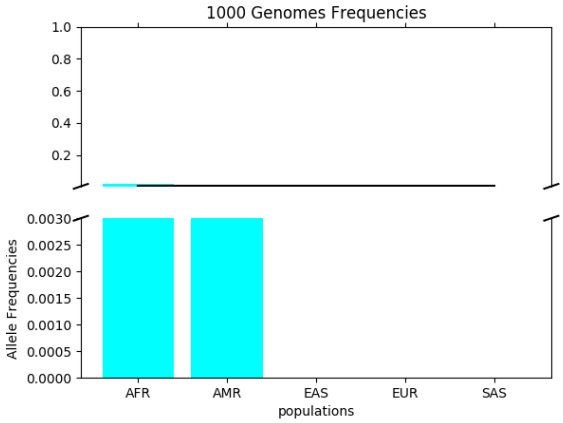
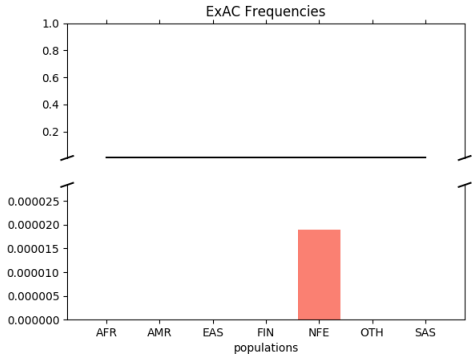
Allele Frequency Visualizations

These visualizations are protypes for the BRCA Exchange. The below graphs were made from ExAC and 1000 Genomes data pulled files in the BRCA Exchange output. Subpopulation data can be helpful to provide context for a variant's allele frequency. If a variant is rare overall, but prevalent in a subpopulation, than it might be lower risk for individuals who have it.

The subpopulations represented in each are automatically generated from the data. The values in the top portion of the bar charts are fixed, always going up to 1.0 for maximum allele frequencies. The truncation, represented by a break in the axis, autofits to a value less than 0.0004 for extremely small values, and otherwise uses 0.003 as the default truncation point. This feature of the program is in need of refinement to handle more test cases. There is a horizontal black line plotted at an allele frequency of 0.01 to represent ENIGMA's significance value cut off range for Allele Frequencies.







When implemented on the website, the truncation can be presented as a toggle feature for the user. Additionally, the cutoff values for the truncation might be further refined, or even an option for the user to modify. These visualizations are intended to responsibly and effectively allow a user to make assessments about significance based on subpopulation allele frequency data.