ClinVar Report

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Sourcing ClinVar input from: clinvar_2014-02-11.vcf
Sending output to: Report_2014-02-11.pdf

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1.2 Merge ClinVar with 1000 Genomes and ExAC

2 Summary Statistics

2.1 Fraction of Individuals with Pathogenic Non-Reference Sites

3 Penetrance Estimates

3.1 Max/Min Penetrance as a Function of P(D) and P(V|D)

The left end of the boxplot indicates P(V|D) = 0.01,
the bold line in the middle indicates P(V|D) = point value,
the right end of the boxplot indicates P(V|D) = 1.

Note: Some diseases have mean theoretical penetrance = 1 because the assumed allelic heterogeneity is greater than is possible, given the observed prevalence and allele frequencies.

3.2 Penetrance Estimates by Ancestry