

# ClinVar Report

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**Sourcing ClinVar input from:** clinvar\_2012-06-16.vcf

**Sending output to:** Report\_2012-06-16.pdf

# 1 Collect and Merge ClinVar Data

## 1.1 Import ClinVar VCF

## Processed ClinVar data frame 33292 x 13 (selected rows/columns):

## 1.2 Merge ClinVar with 1000 Genomes and ExAC

## Breakdown of ClinVar Variants

Subset_ClinVar	Number_of_Variants
Total ClinVar	33292
LP/P-ClinVar	12040
LP/P-ClinVar & ACMG	779
LP/P-ClinVar & ACMG & ExAC	191
LP/P-ClinVar & ACMG & 1000 Genomes	64

## Breakdown of ACMG-1000 Genomes Variants

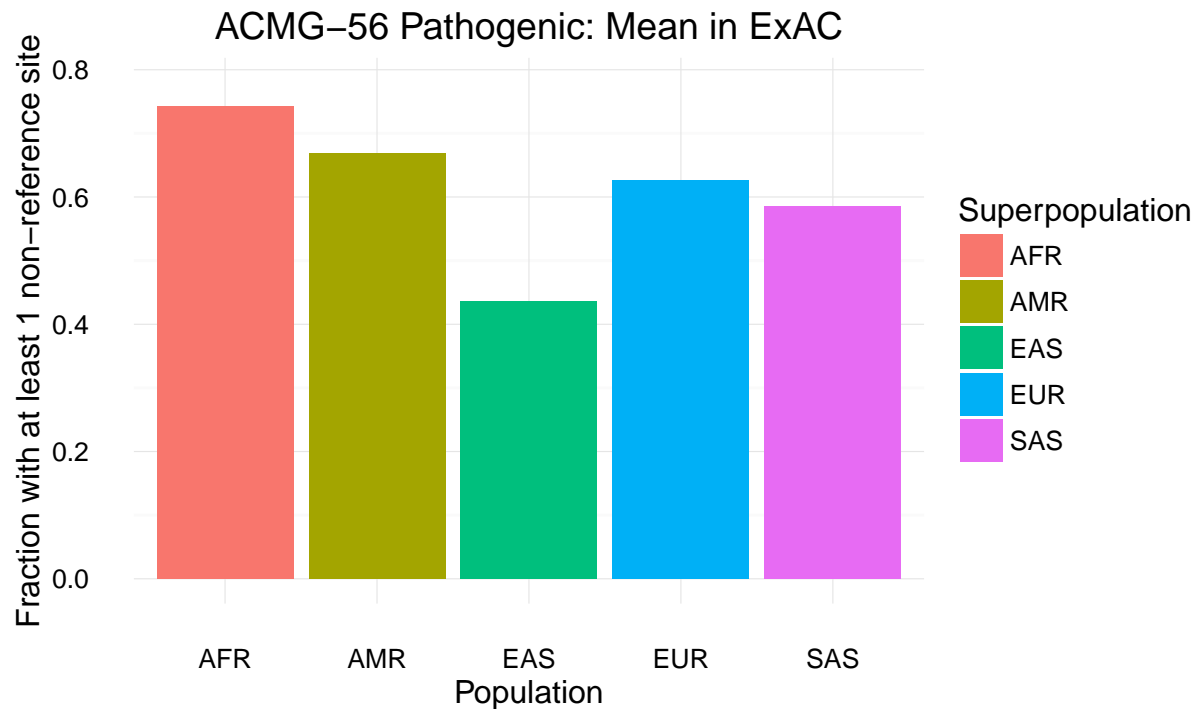
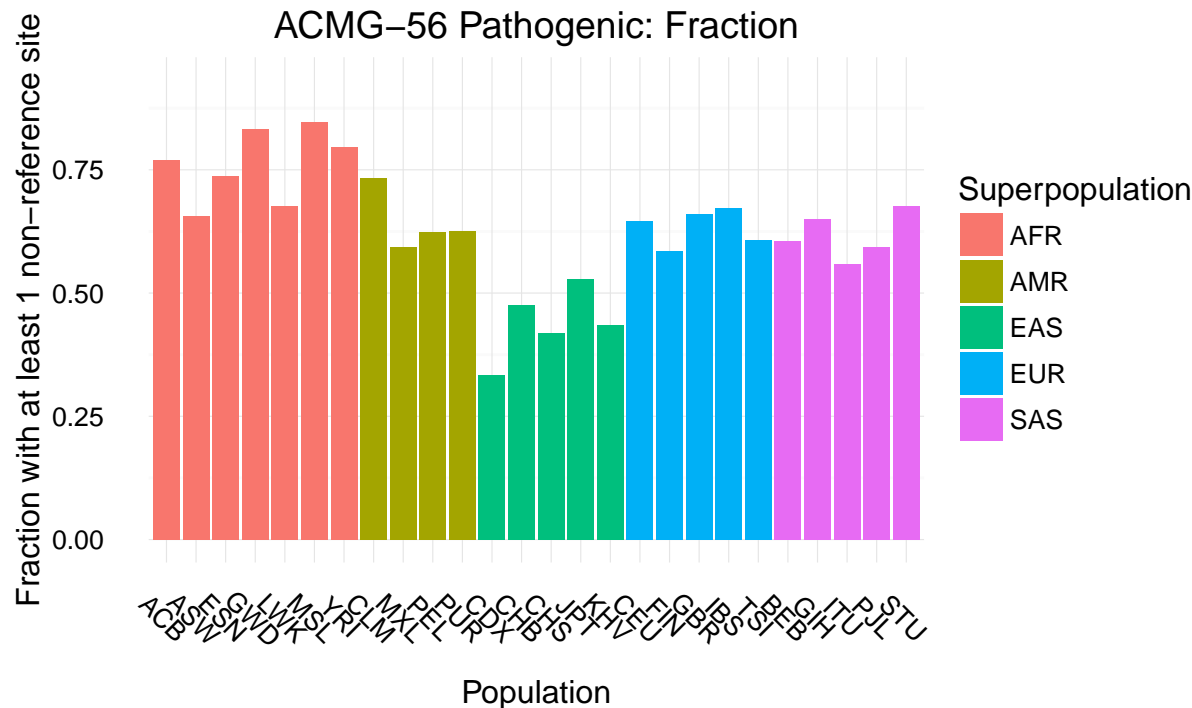
Subset_1000_Genomes	Number_of_Variants
Total 1000_Genomes & ACMG	139335
1000_Genomes & ACMG & ClinVar	1091
1000_Genomes & ACMG & LP/P-ClinVar	64

## Breakdown of ACMG-ExAC Variants

Subset_ExAC	Number_of_Variants
Total ExAC & ACMG	58873
ExAC & ACMG & ClinVar	2035
ExAC & ACMG & LP/P-ClinVar	191

2 Summary Statistics

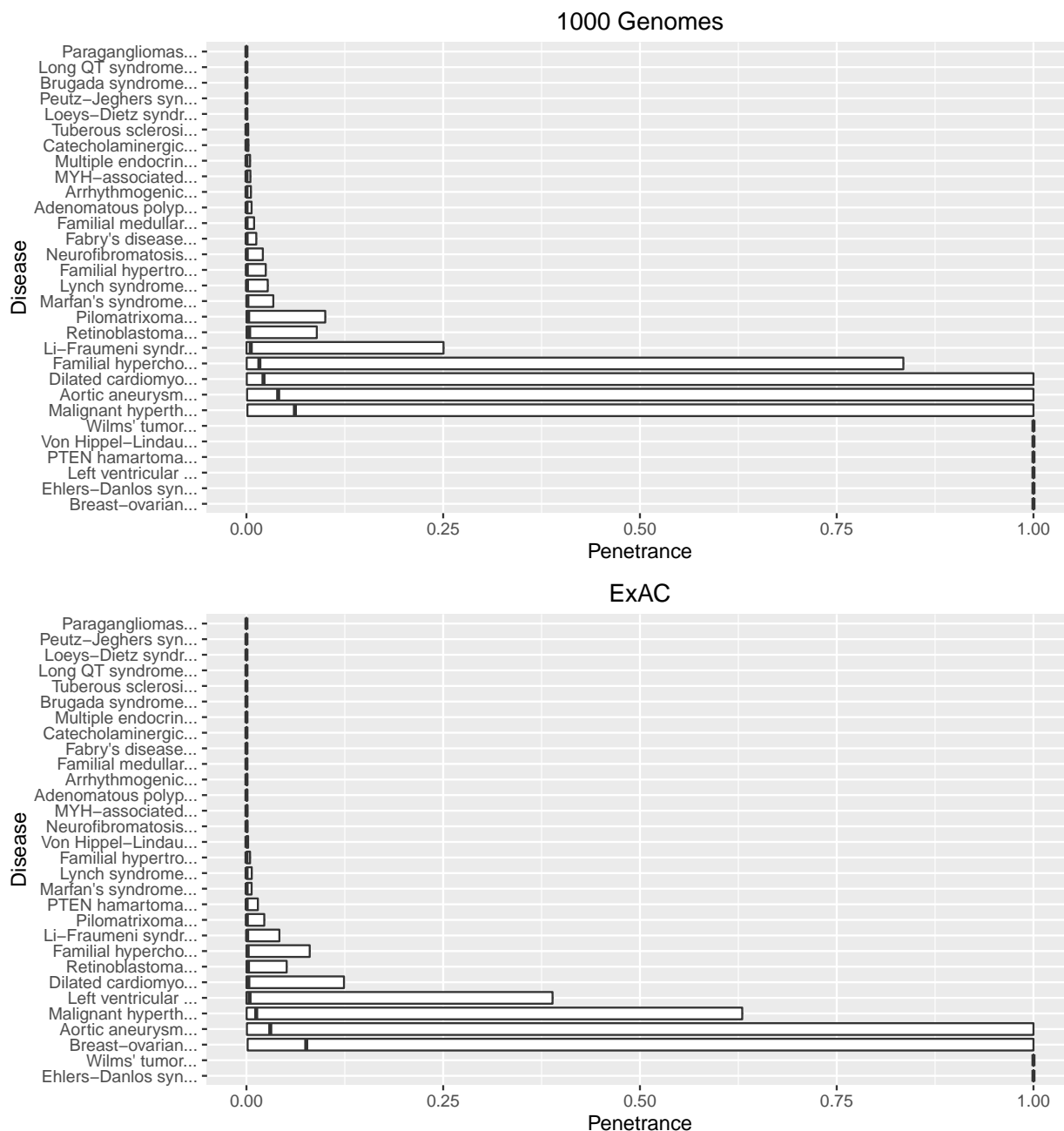
2.1 Fraction of Individuals with Pathogenic Non-Reference Sites



### 3 Penetrance Estimates

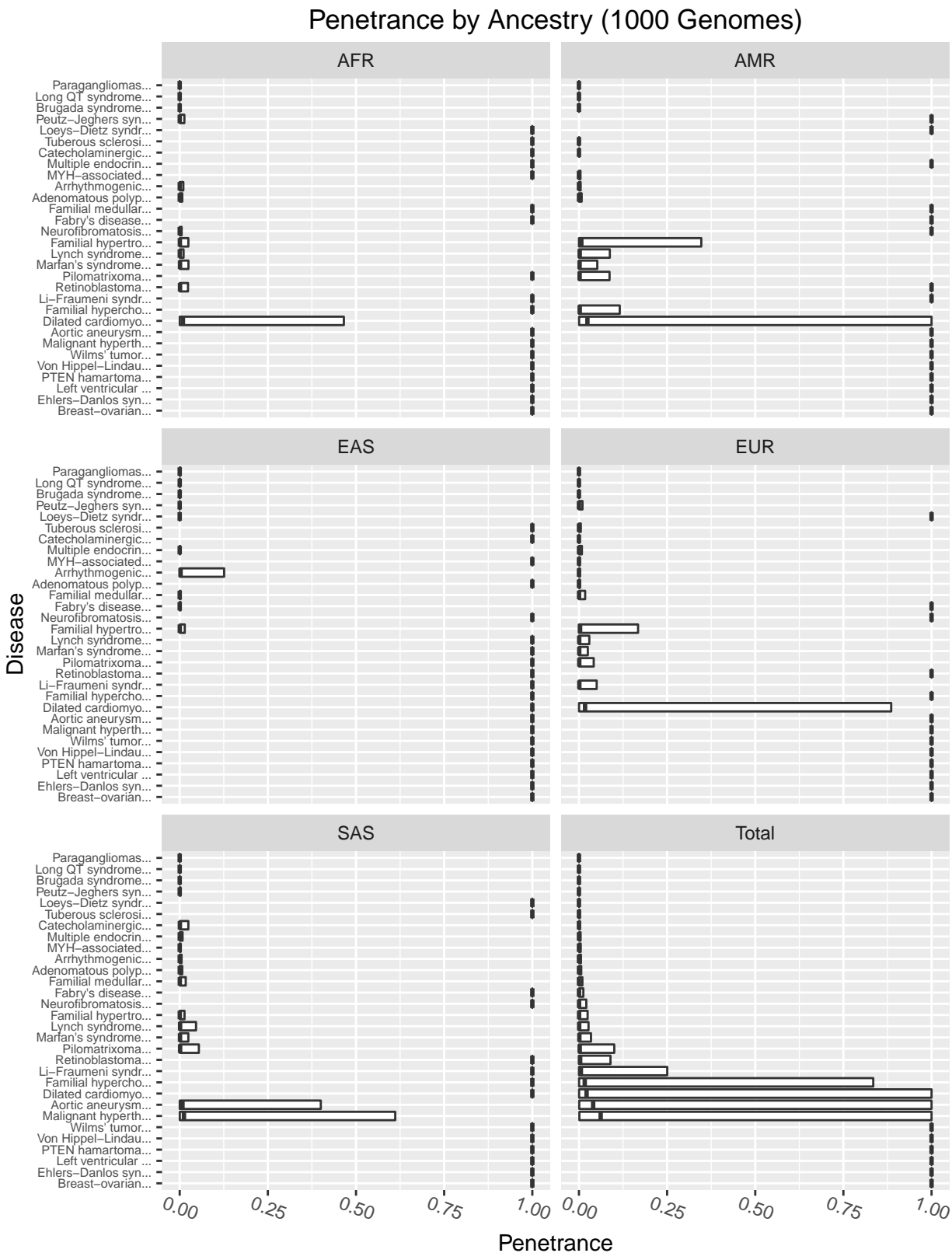
#### 3.1 Max/Min Penetrance as a Function of $P(D)$ and $P(V|D)$

The left end of the boxplot indicates  $P(D)$  AND  $P(V|D)$  = lower value,  
the bold line in the middle indicates  $P(D)$  AND  $P(V|D)$  = geometric\_mean(values),  
the right end of the boxplot indicates  $P(D)$  AND  $P(V|D)$  = upper value.

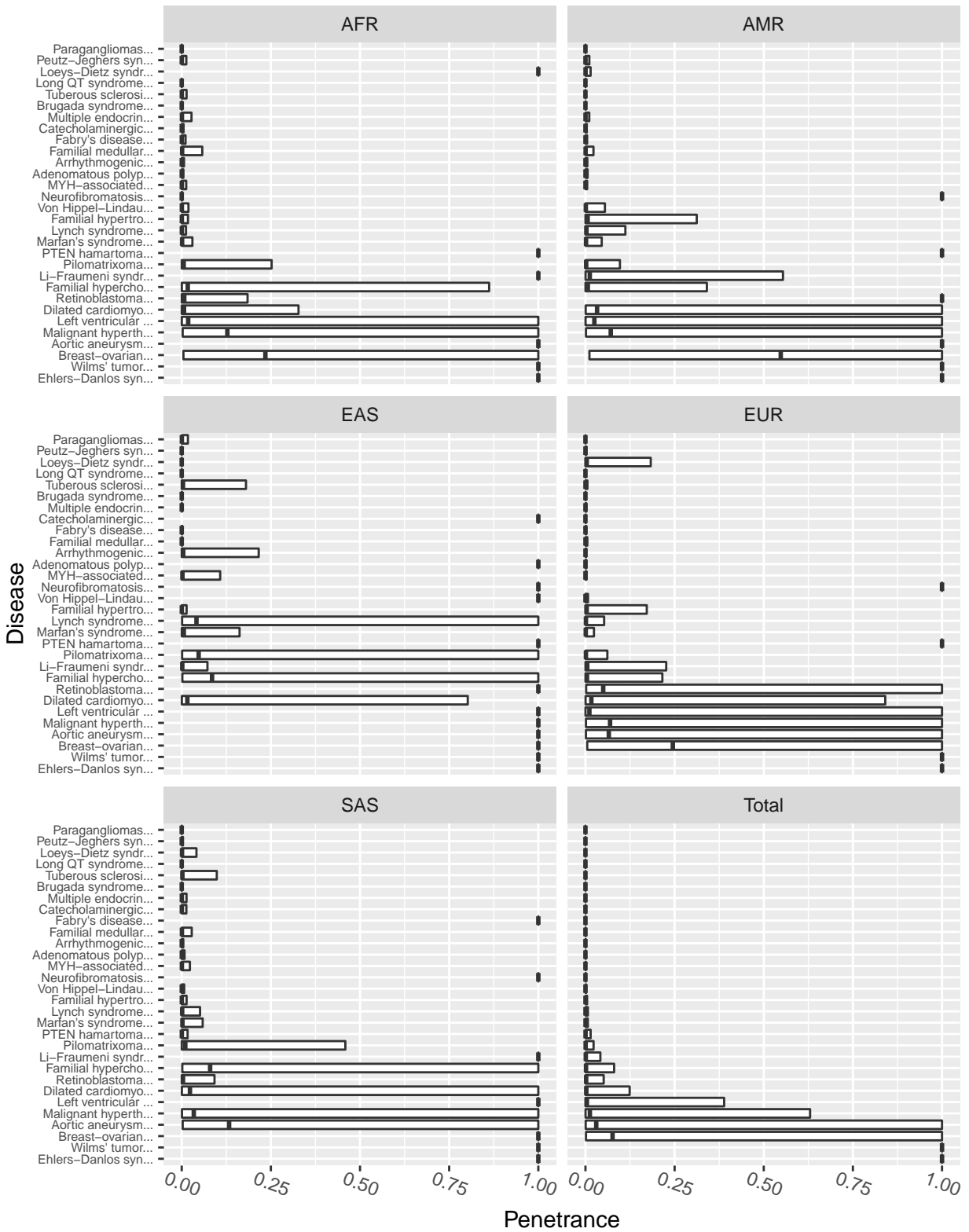


Note: Prevalence ranges of 5x were assumed for all point estimates of prevalence.  
For example: a point estimate of 0.022 would be given the range 0.01-0.05.

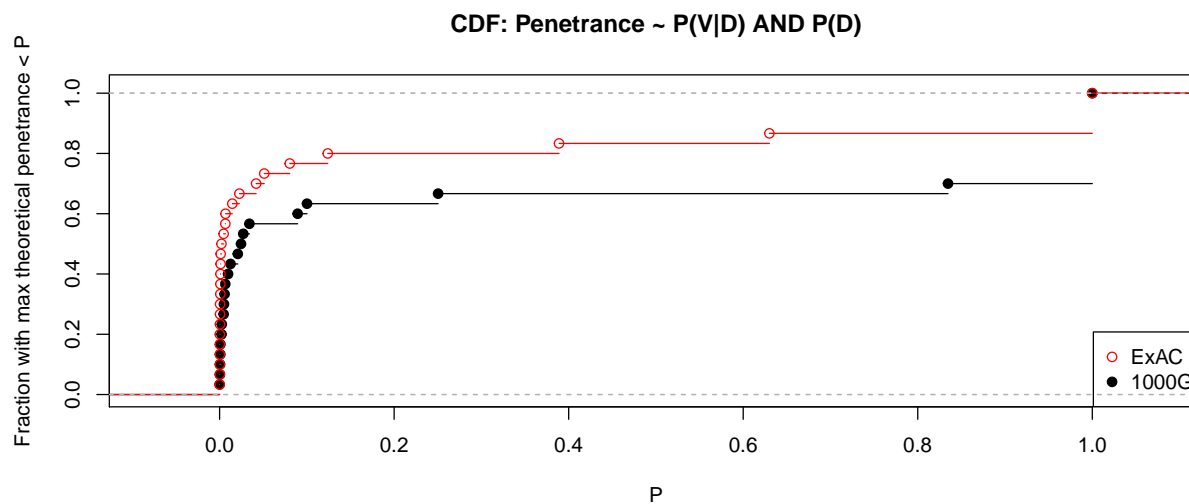
### 3.2 Penetrance Estimates by Ancestry



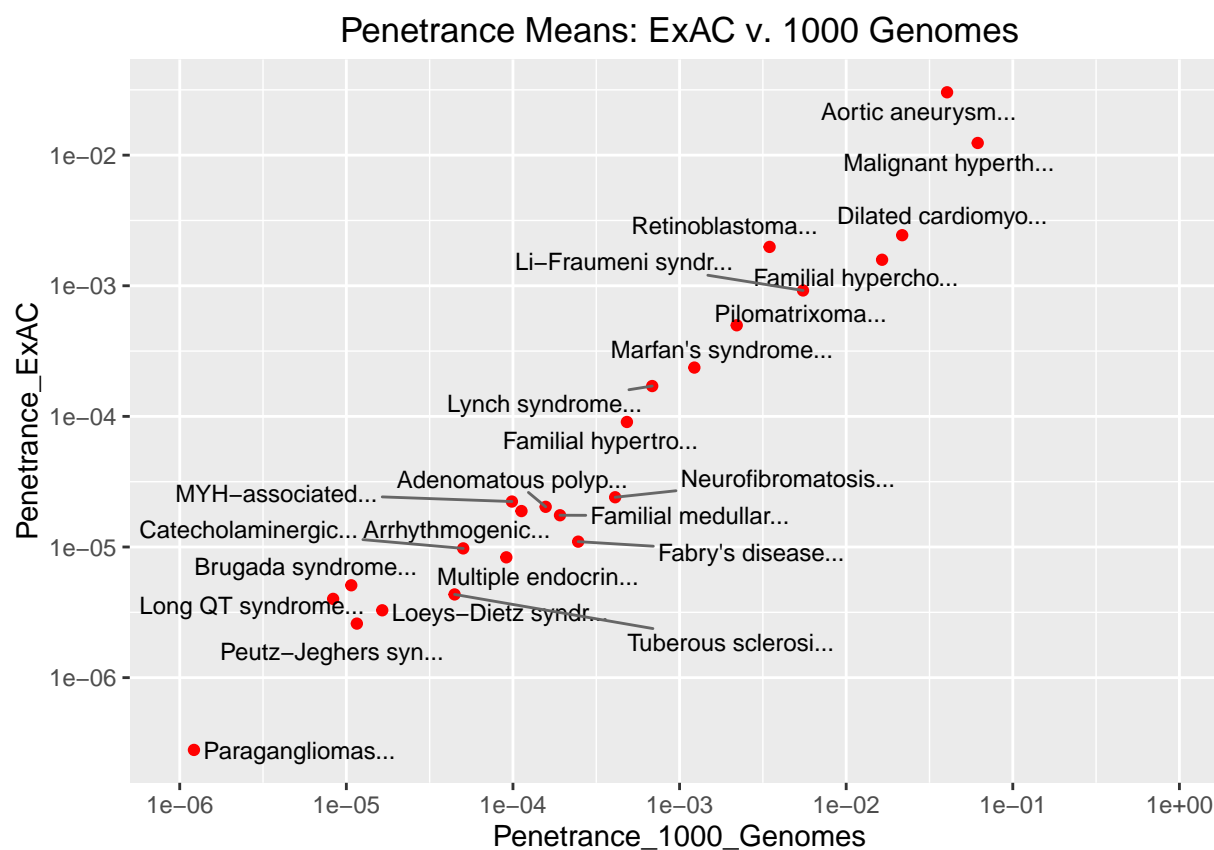
## Penetrance by Ancestry (ExAC)



### 3.3 Empirical CDFs for All Penetrance Plots



### 3.4 Comparing Mean Penetrance between ExAC and 1000 Genomes



The Pearson correlation is 0.78.

Max penetrance values computed using 1000 Genomes are 1.8-fold larger than those computed using ExAC.