

# ClinVar Report

*James Diao*

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**Sourcing ClinVar input from:** clinvar\_2014-07-02.vcf

**Sending output to:** Report\_2014-07-02.pdf

# 1 Collect and Merge ClinVar Data

## 1.1 Import ClinVar VCF

## Processed ClinVar data frame 78506 x 14 (selected rows/columns):

## 1.2 Merge ClinVar with 1000 Genomes and ExAC

## Breakdown of ClinVar Variants

Subset_ClinVar	Number_of_Variants
Total ClinVar	78506
LP/P-ClinVar	17873
LP/P-ClinVar & ACMG	3306
LP/P-ClinVar & ACMG & ExAC	535
LP/P-ClinVar & ACMG & 1000 Genomes	109

## Breakdown of ACMG-1000 Genomes Variants

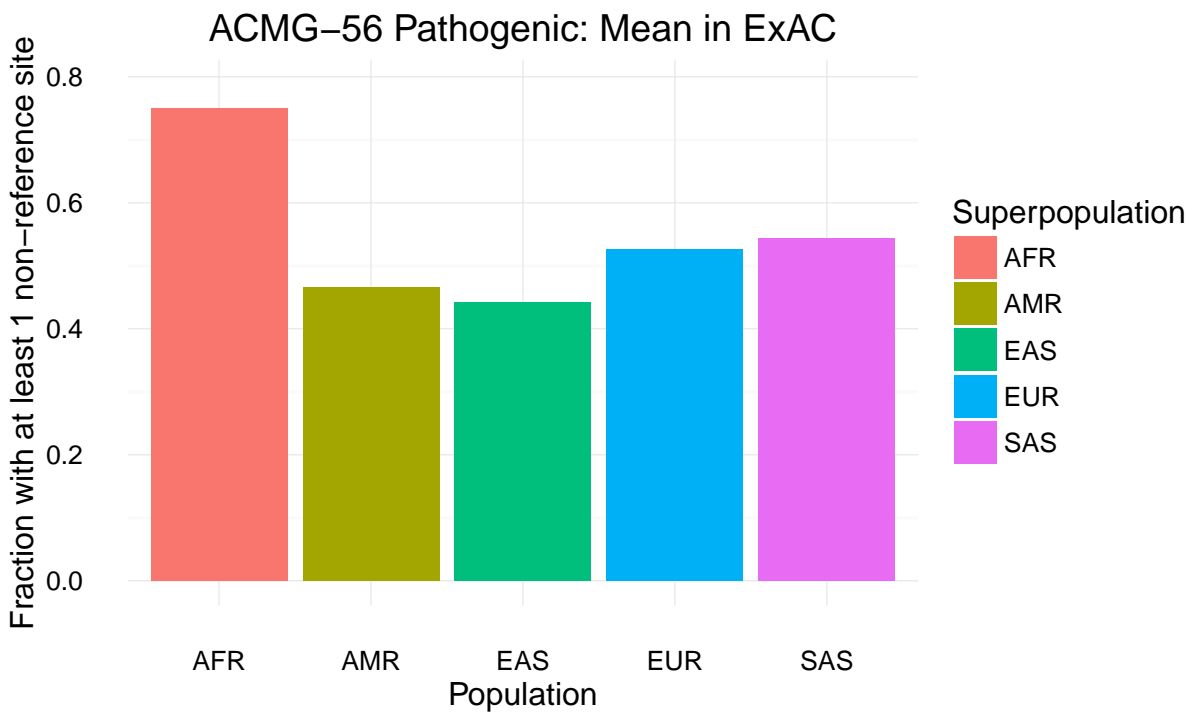
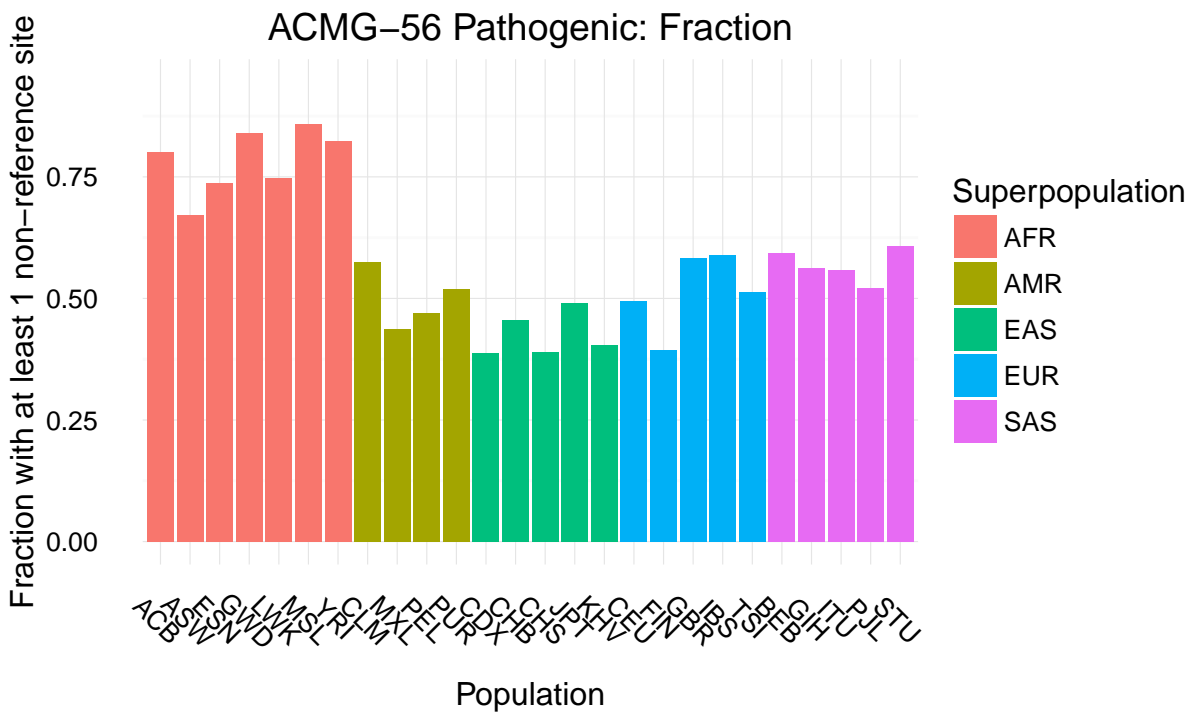
Subset_1000_Genomes	Number_of_Variants
Total 1000_Genomes & ACMG	139335
1000_Genomes & ACMG & ClinVar	1791
1000_Genomes & ACMG & LP/P-ClinVar	109

## Breakdown of ACMG-ExAC Variants

Subset_ExAC	Number_of_Variants
Total ExAC & ACMG	58873
ExAC & ACMG & ClinVar	3555
ExAC & ACMG & LP/P-ClinVar	535

## 2 Summary Statistics

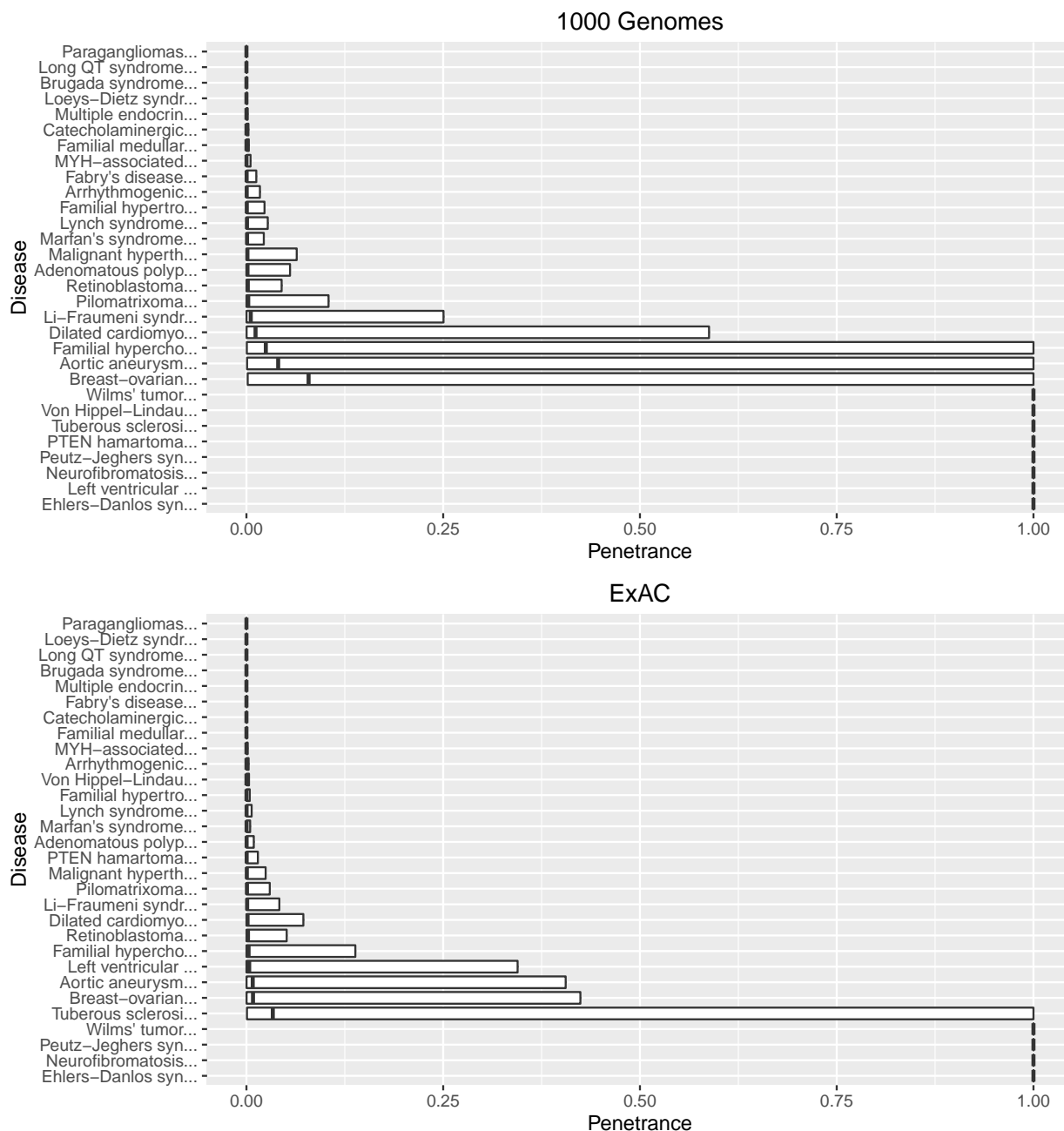
### 2.1 Fraction of Individuals with Pathogenic Non-Reference Sites



### 3 Penetrance Estimates

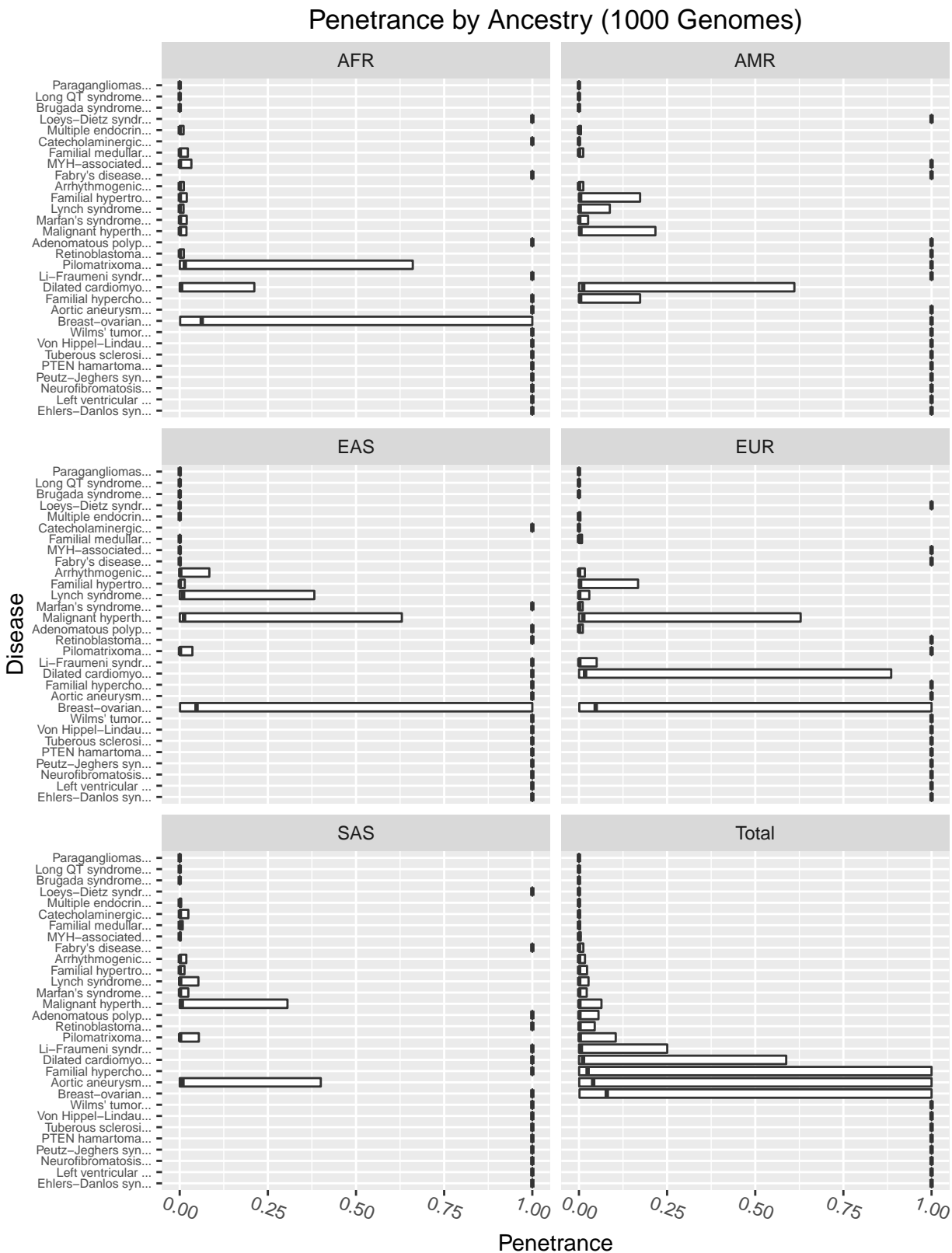
#### 3.1 Max/Min Penetrance as a Function of $P(D)$ and $P(V|D)$

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.

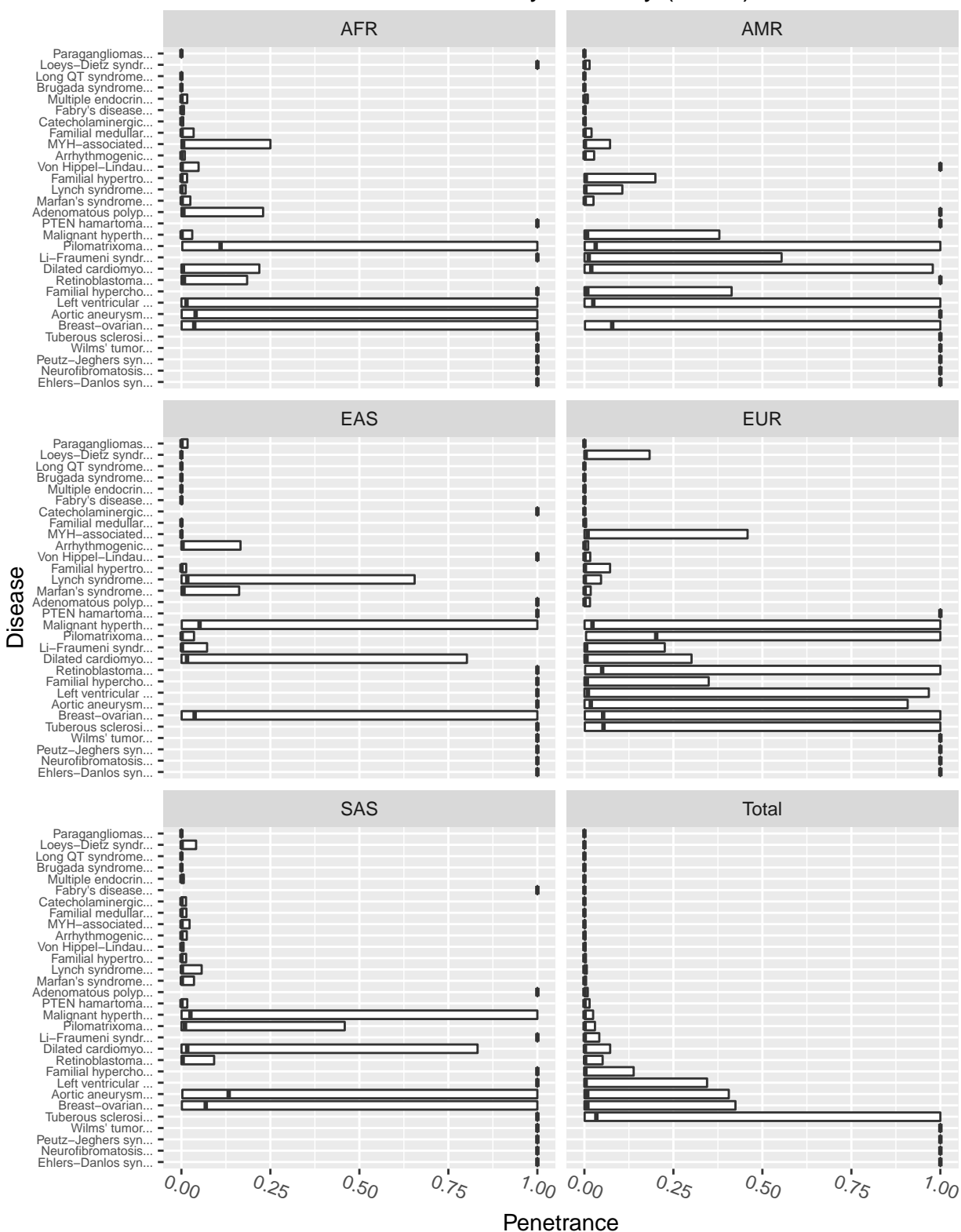


Note: Prevalence ranges of 5x were assumed for all point estimates of prevalence.  
For example: a point estimate of 0.022 would be given the range 0.01-0.05.

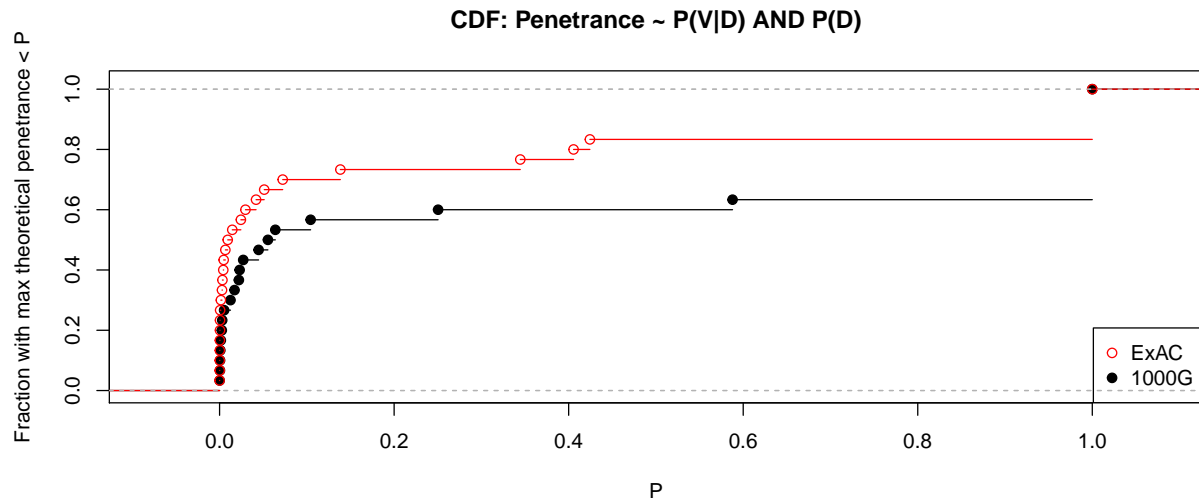
### 3.2 Penetrance Estimates by Ancestry



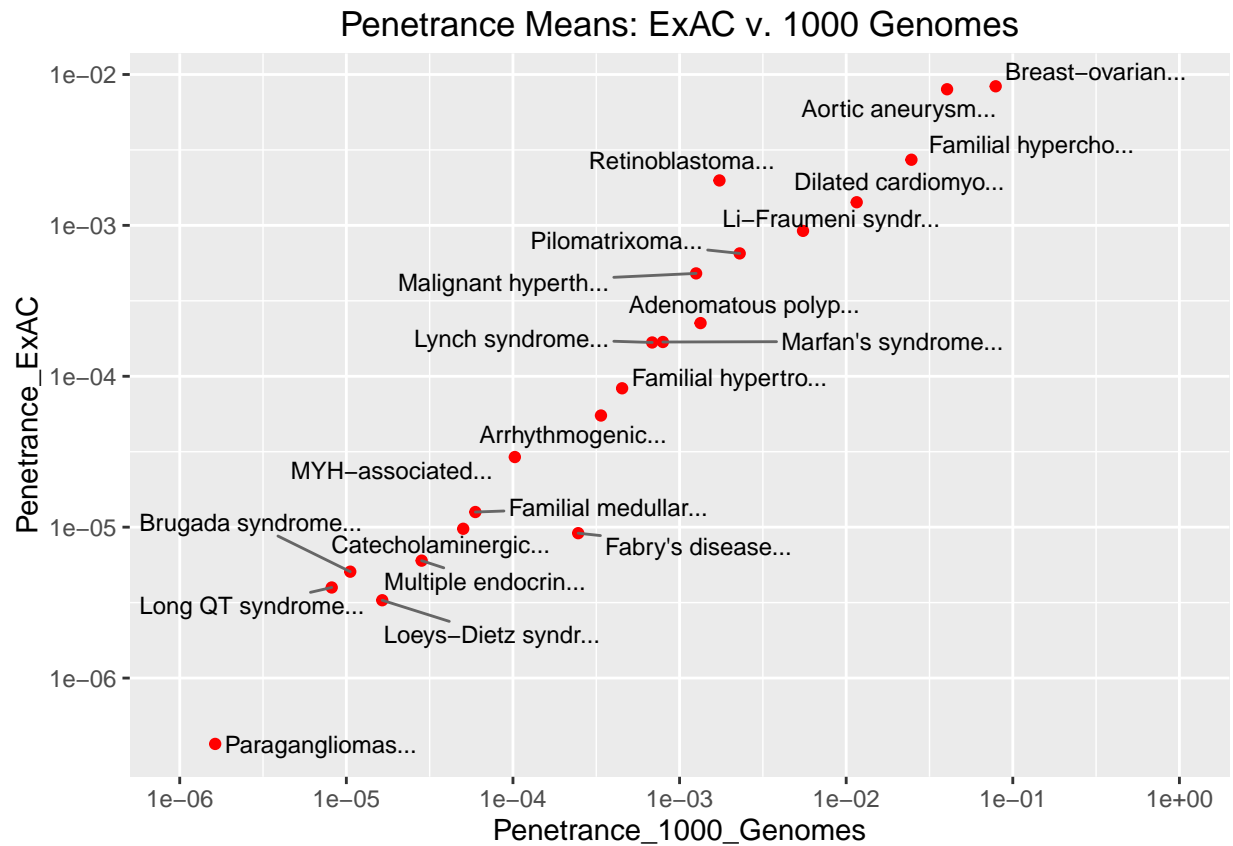
## Penetrance by Ancestry (ExAC)



### 3.3 Empirical CDFs for All Penetrance Plots



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



The Pearson correlation is 0.94.

Max penetrance values computed using 1000 Genomes are 7.4-fold larger than those computed using ExAC.