

# ClinVar Report

*James Diao*

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**Sourcing ClinVar input from:** clinvar\_2015-06-03.vcf

**Sending output to:** Report\_2015-06-03.pdf

# 1 Collect and Merge ClinVar Data

## 1.1 Import ClinVar VCF

## Processed ClinVar data frame 109899 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                      | 109899             |
| LP/P-ClinVar                       | 25124              |
| LP/P-ClinVar & ACMG                | 5096               |
| LP/P-ClinVar & ACMG & ExAC         | 822                |
| LP/P-ClinVar & ACMG & 1000 Genomes | 151                |

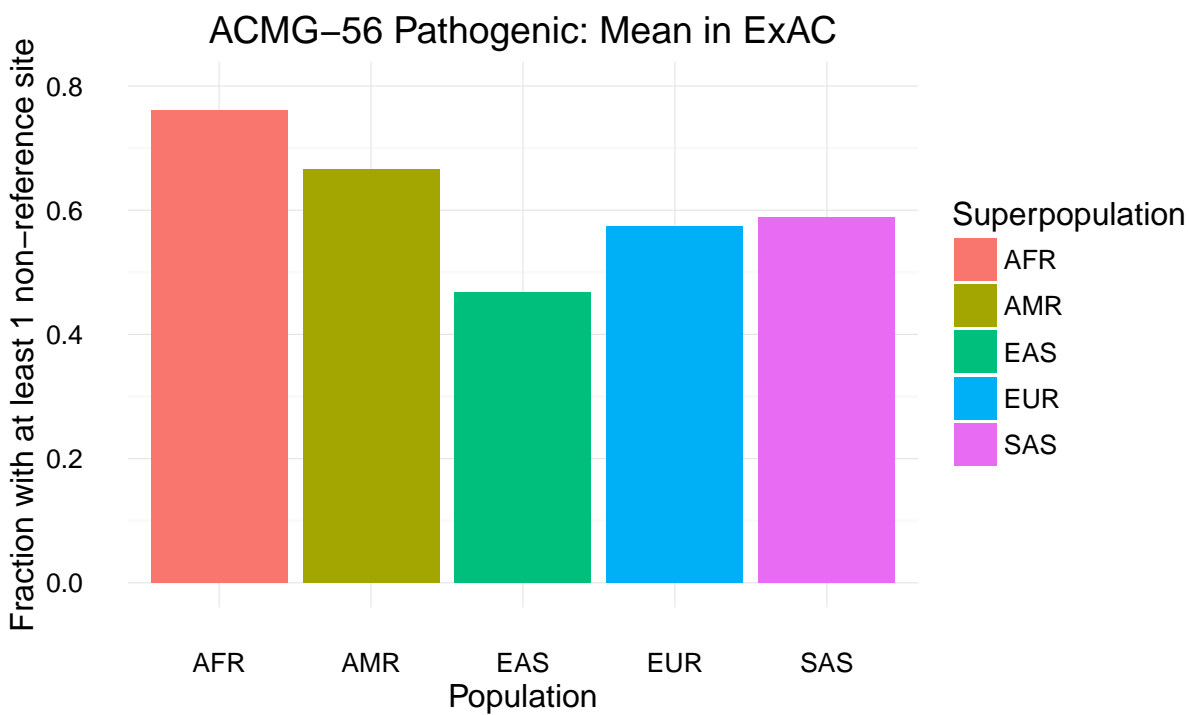
## Breakdown of ACMG-1000 Genomes Variants

| Subset_1000_Genomes                | Number_of_Variants |
|------------------------------------|--------------------|
| Total 1000_Genomes & ACMG          | 139335             |
| 1000_Genomes & ACMG & ClinVar      | 2824               |
| 1000_Genomes & ACMG & LP/P-ClinVar | 151                |

## Breakdown of ACMG-ExAC Variants

| Subset_ExAC                | Number_of_Variants |
|----------------------------|--------------------|
| Total ExAC & ACMG          | 58873              |
| ExAC & ACMG & ClinVar      | 6622               |
| ExAC & ACMG & LP/P-ClinVar | 822                |

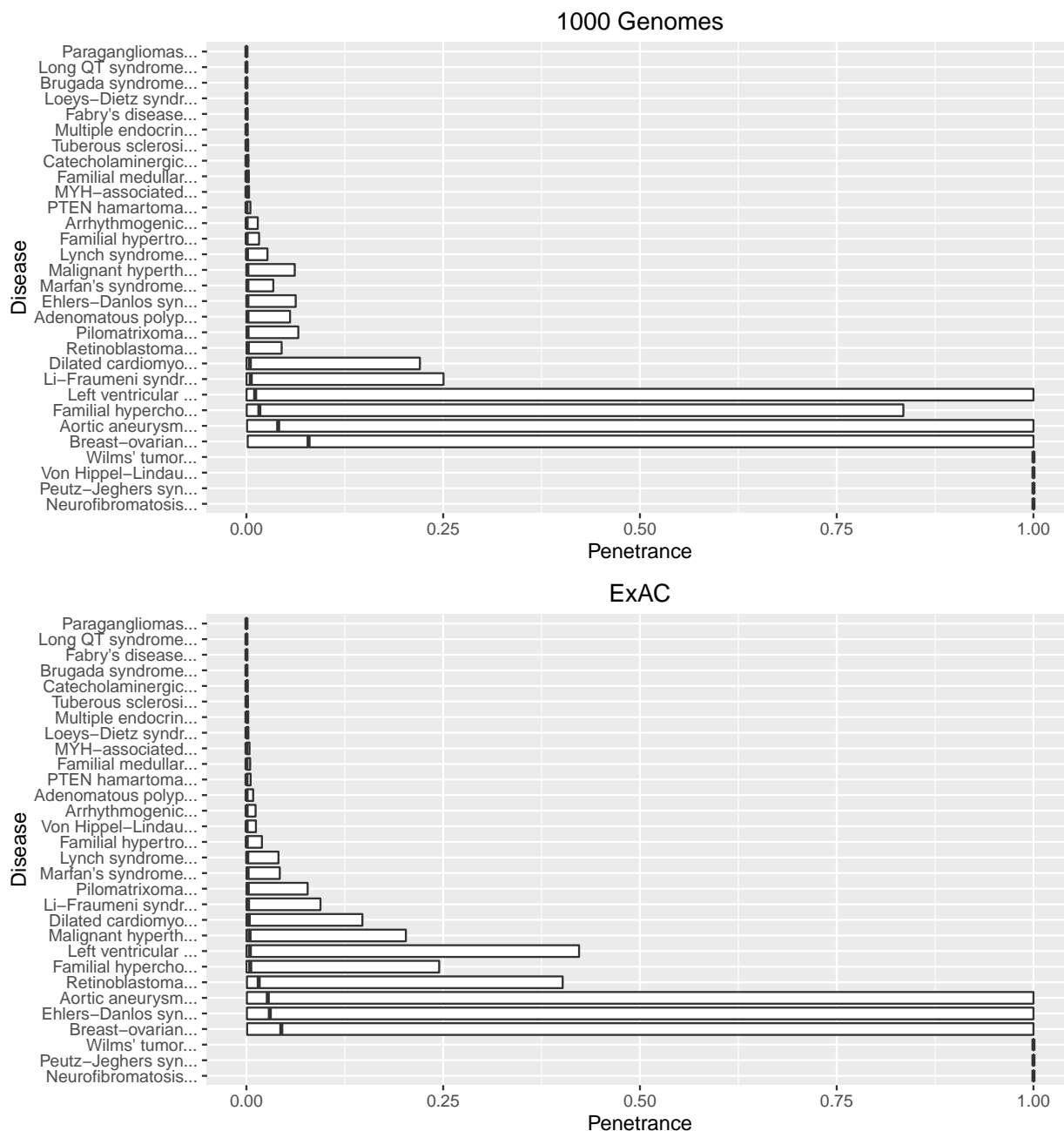
## 2.1 Fraction of Individuals with Pathogenic Non-Reference Sites



### 3 Penetrance Estimates

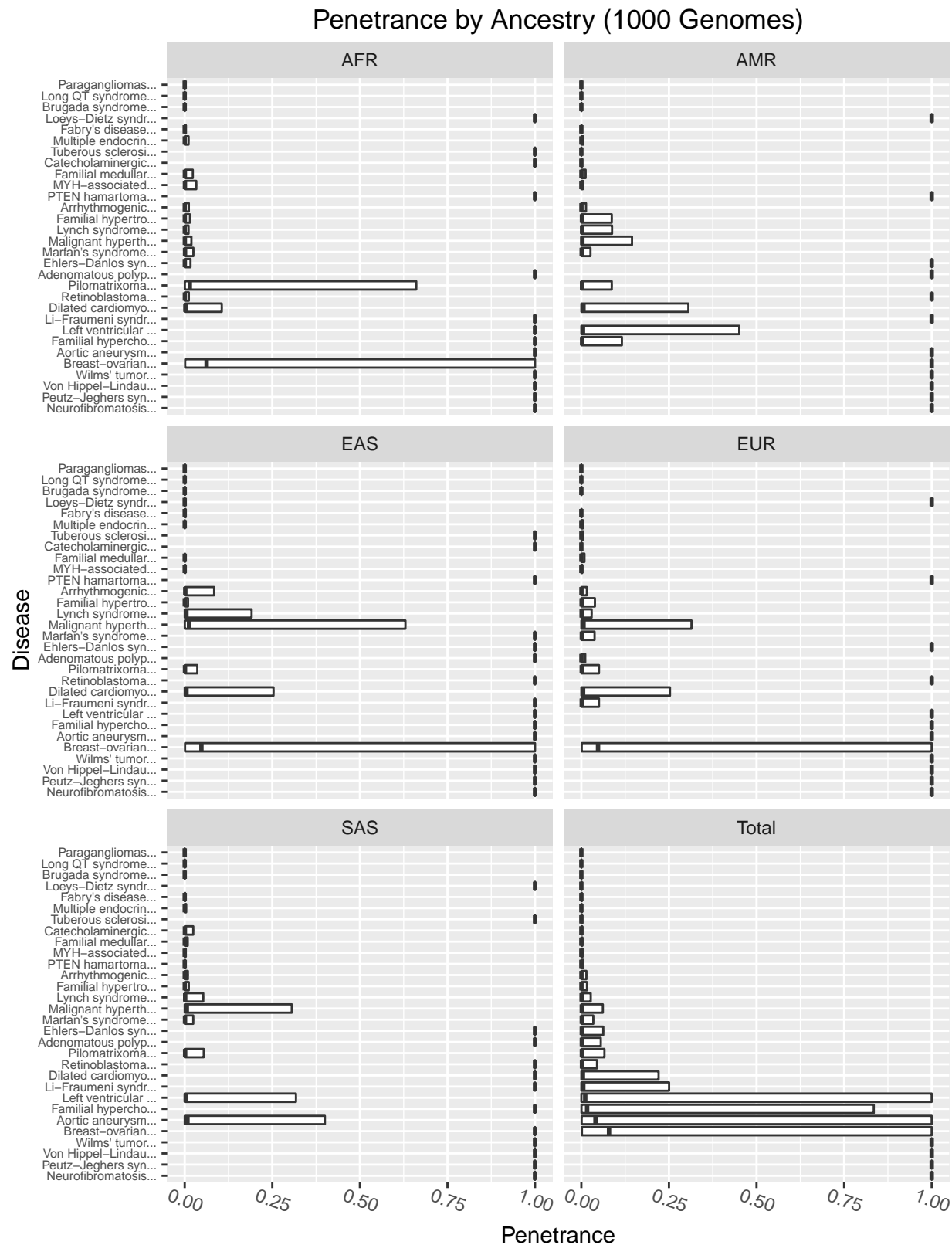
#### 3.1 Max/Min Penetrance as a Function of $P(D)$ and $P(V|D)$

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.

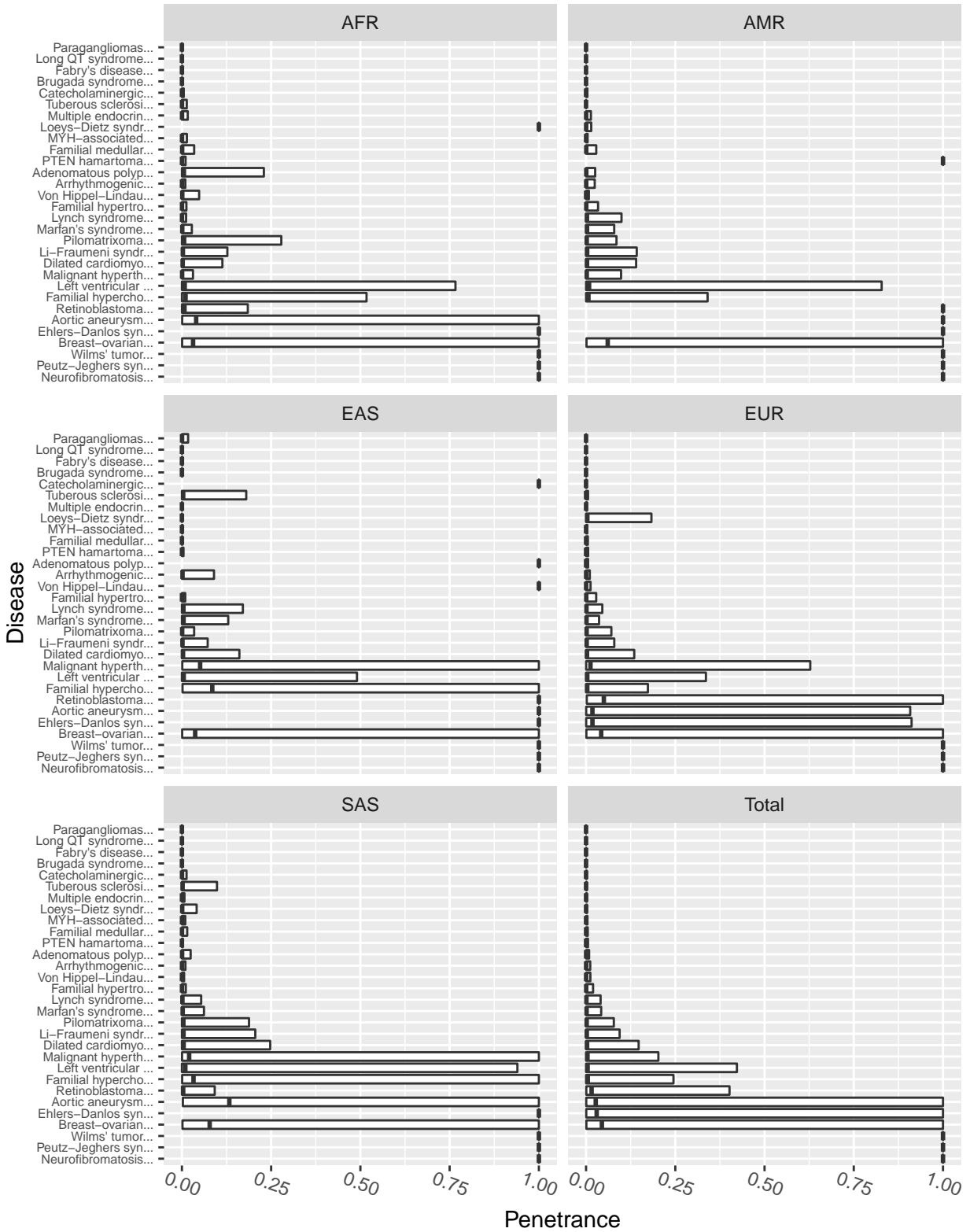


Note: Prevalence ranges of 5x were assumed for all point estimates of prevalence.  
For example: a point estimate of 0.022 would be given the range 0.01-0.05.

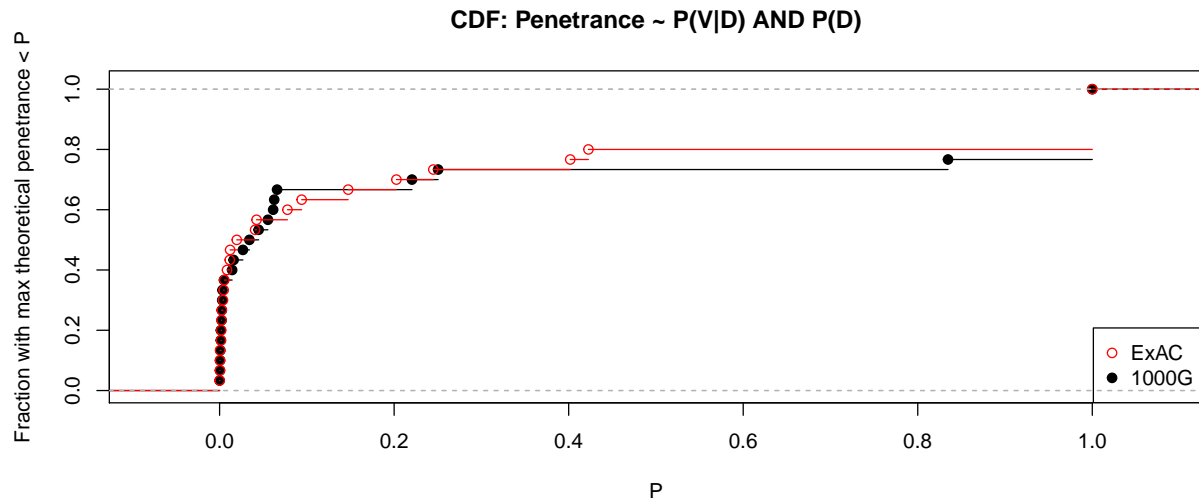
### 3.2 Penetrance Estimates by Ancestry



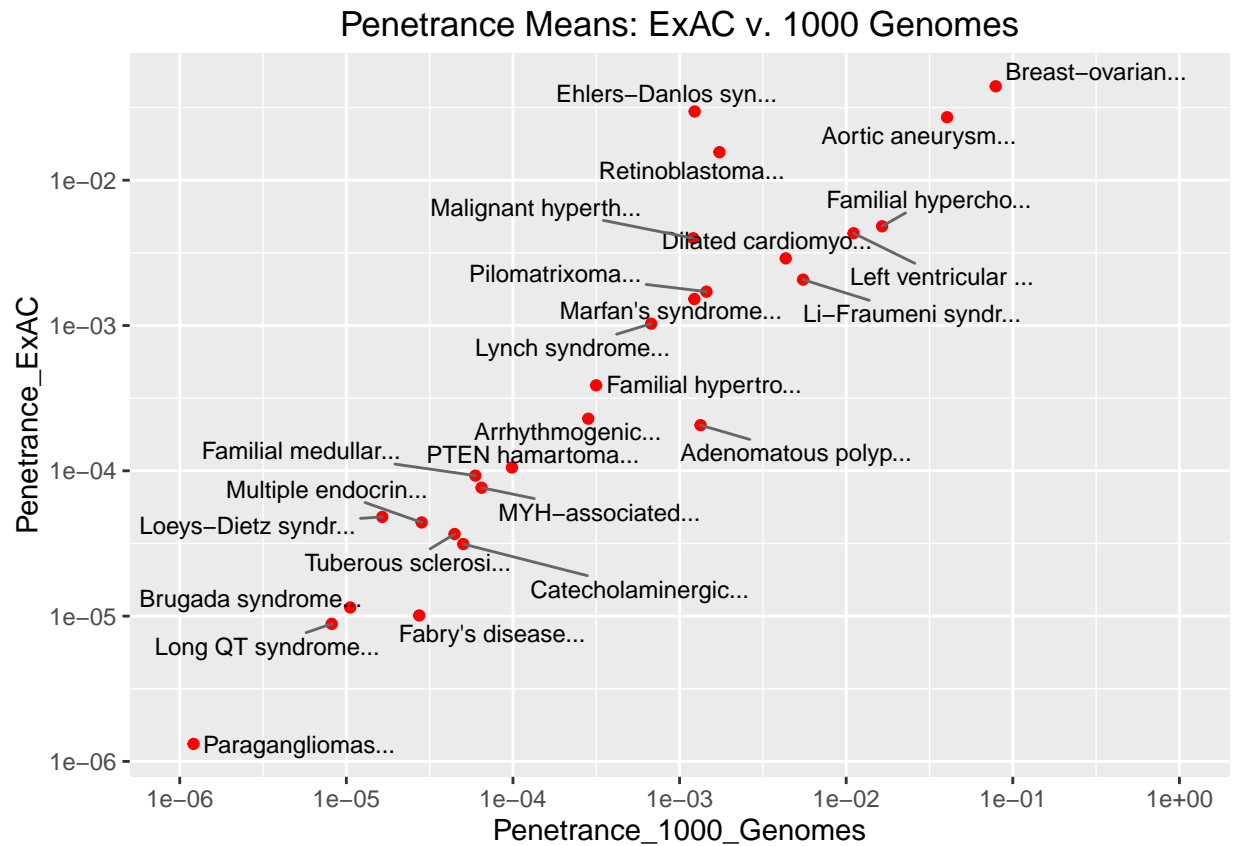
## Penetrance by Ancestry (ExAC)



### 3.3 Empirical CDFs for All Penetrance Plots



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



The Pearson correlation is 0.82.

Max penetrance values computed using 1000 Genomes are 1.2-fold larger than those computed using ExAC.