

# ClinVar Report

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*November 4, 2016*

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**Sourcing ClinVar input from:** clinvar\_2013-05-06.vcf

**Sending output to:** Report\_2013-05-06.pdf

# 1 Collect and Merge ClinVar Data

## 1.1 Import ClinVar VCF

```
## Processed ClinVar data frame 67470 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	67470
LP/P-ClinVar	13342
LP/P-ClinVar & ACMG	969
LP/P-ClinVar & ACMG & ExAC	212
LP/P-ClinVar & ACMG & 1000 Genomes	65

```
## Breakdown of ACMG-1000 Genomes Variants
```

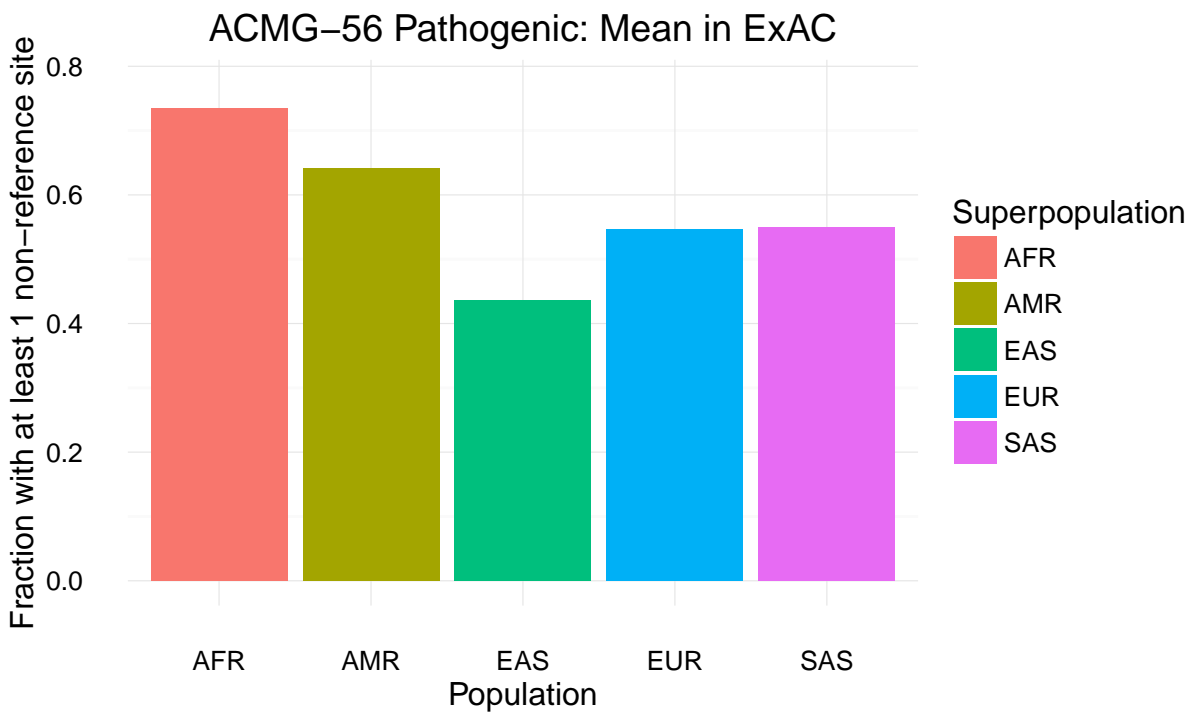
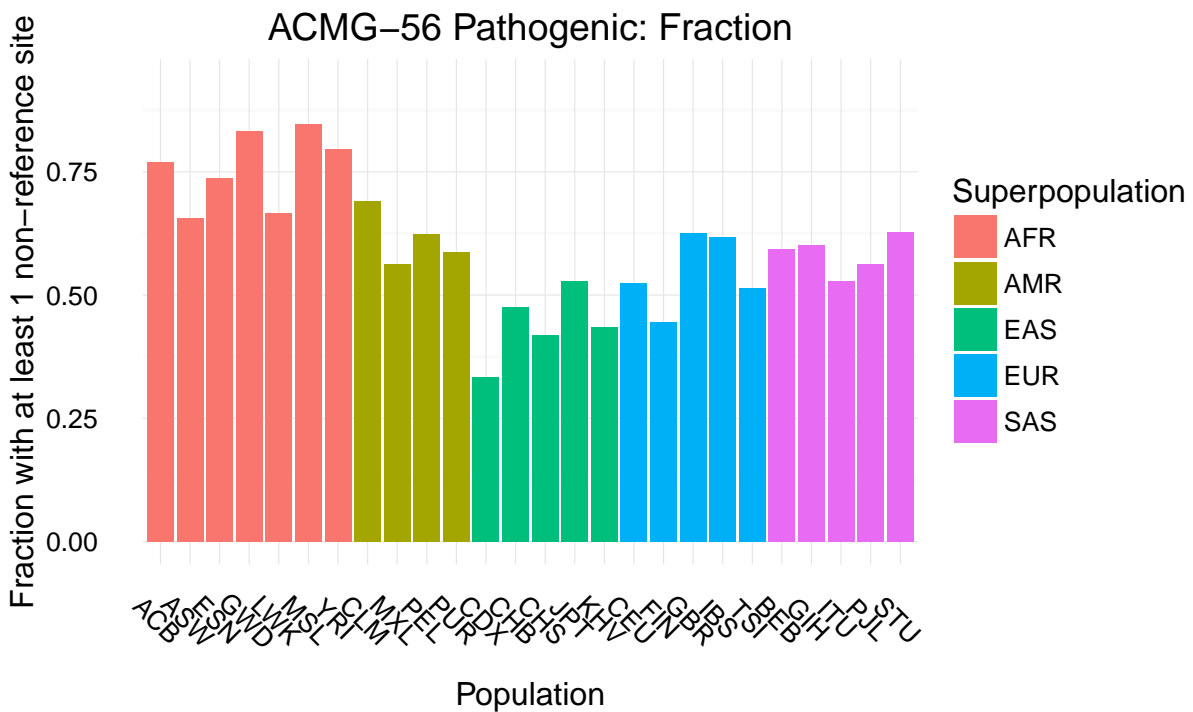
Subset_1000_Genomes	Number_of_Variants
Total 1000_Genomes & ACMG	139335
1000_Genomes & ACMG & ClinVar	1226
1000_Genomes & ACMG & LP/P-ClinVar	65

```
## Breakdown of ACMG-ExAC Variants
```

Subset_ExAC	Number_of_Variants
Total ExAC & ACMG	58873
ExAC & ACMG & ClinVar	2274
ExAC & ACMG & LP/P-ClinVar	212

## 2 Summary Statistics

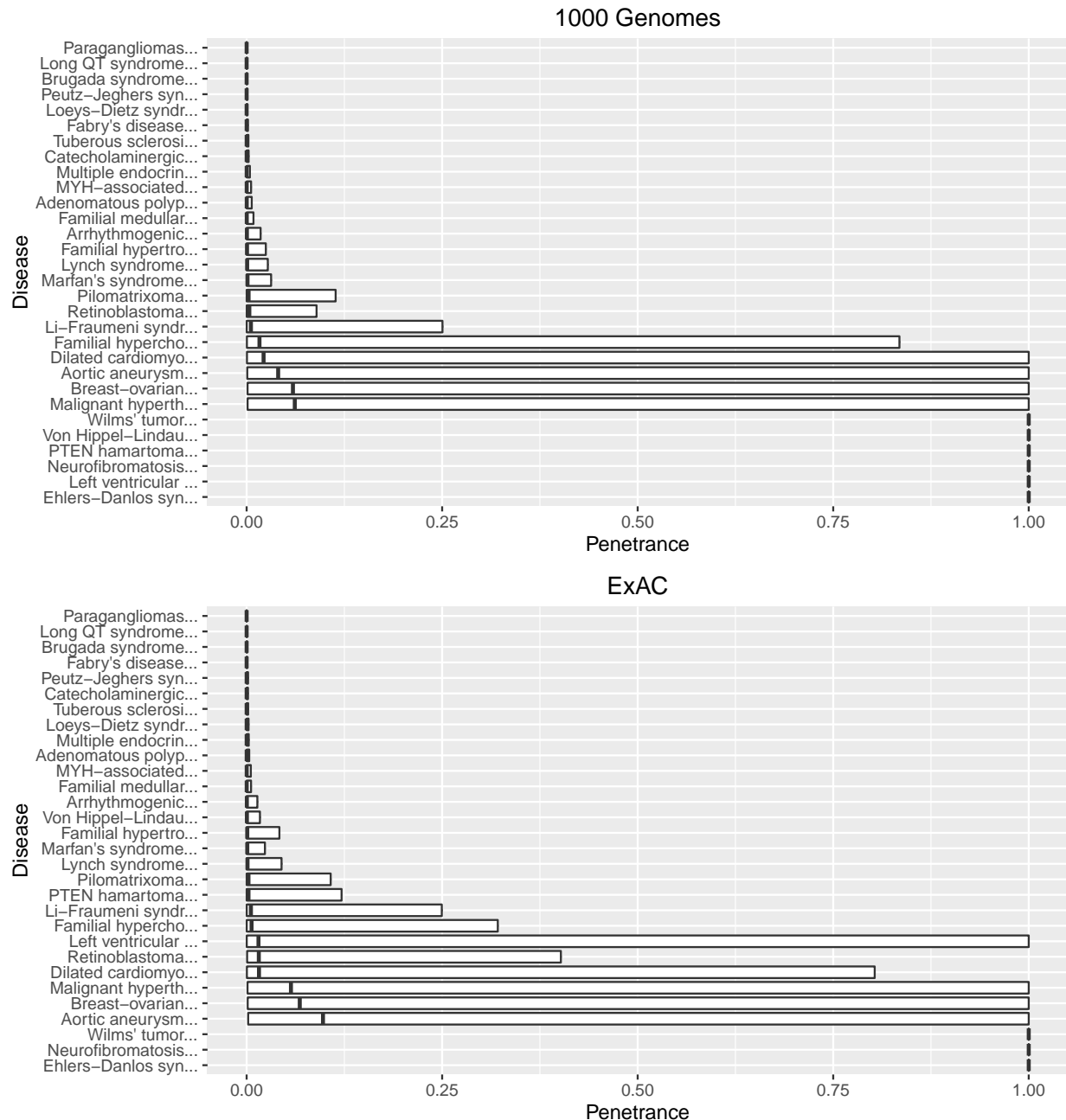
### 2.1 Fraction of Individuals with Pathogenic Non-Reference Sites



### 3 Penetrance Estimates

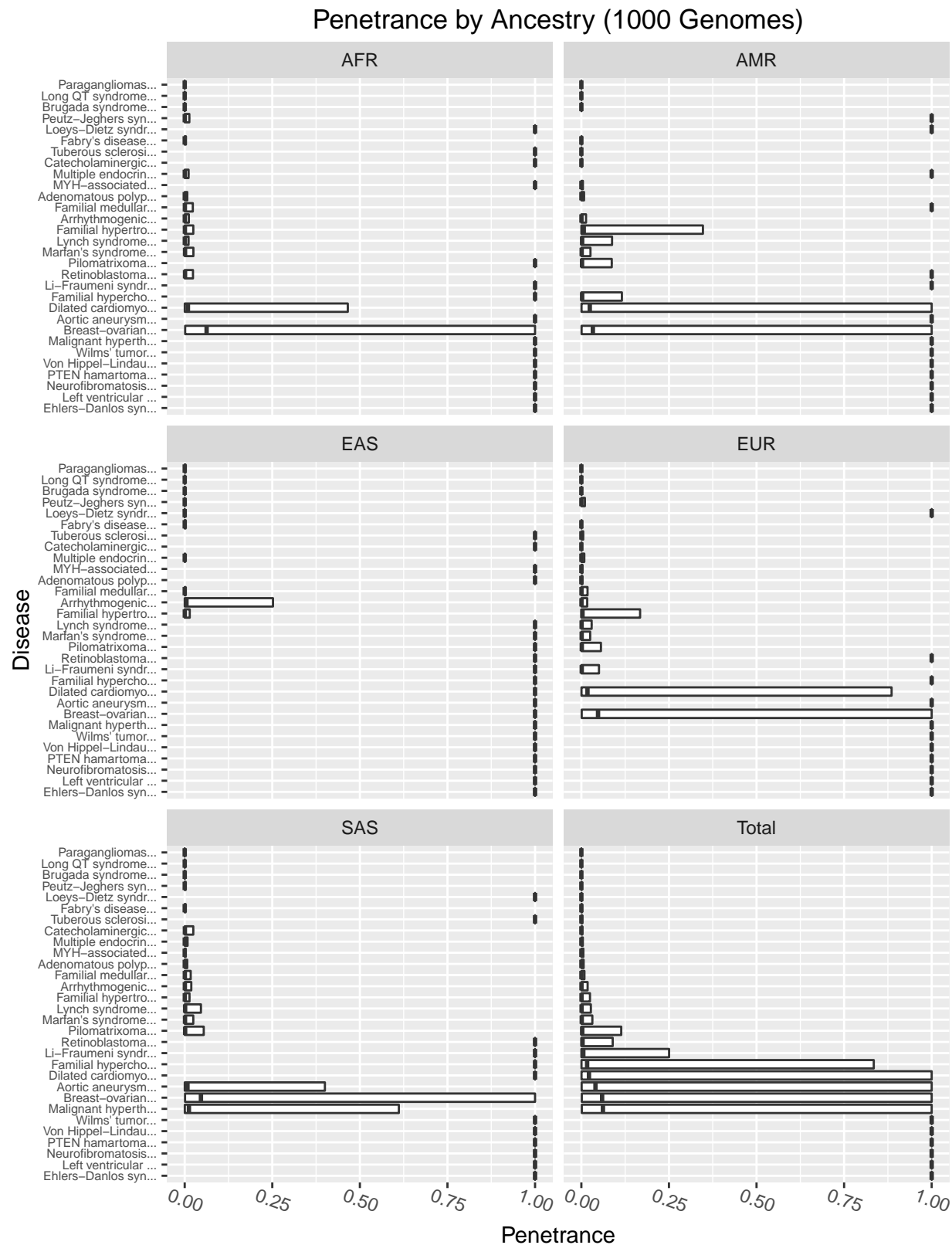
#### 3.1 Max/Min Penetrance as a Function of $P(D)$ and $P(V|D)$

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.

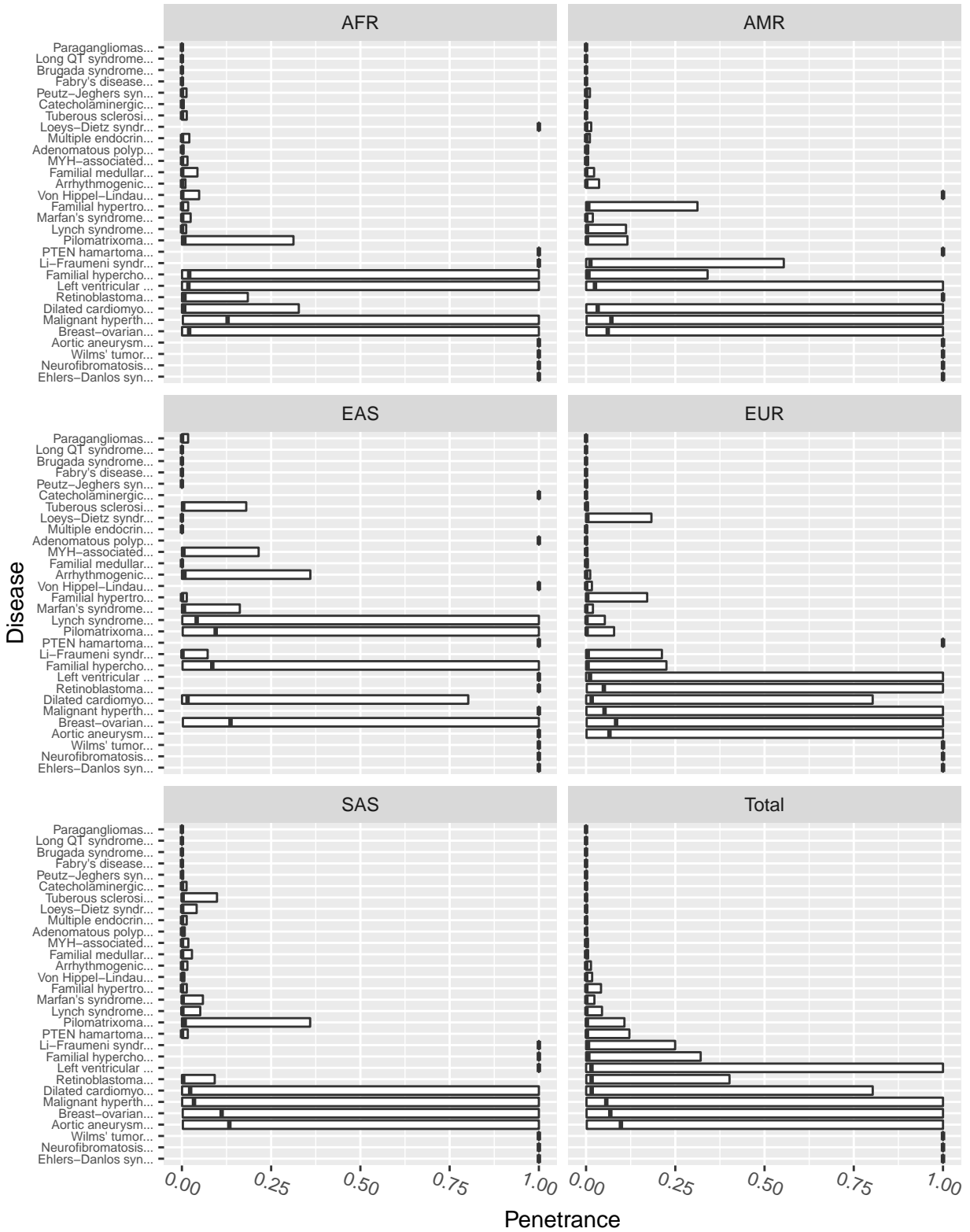


Note: Prevalence ranges of 5x were assumed for all point estimates of prevalence.  
For example: a point estimate of 0.022 would be given the range 0.01-0.05.

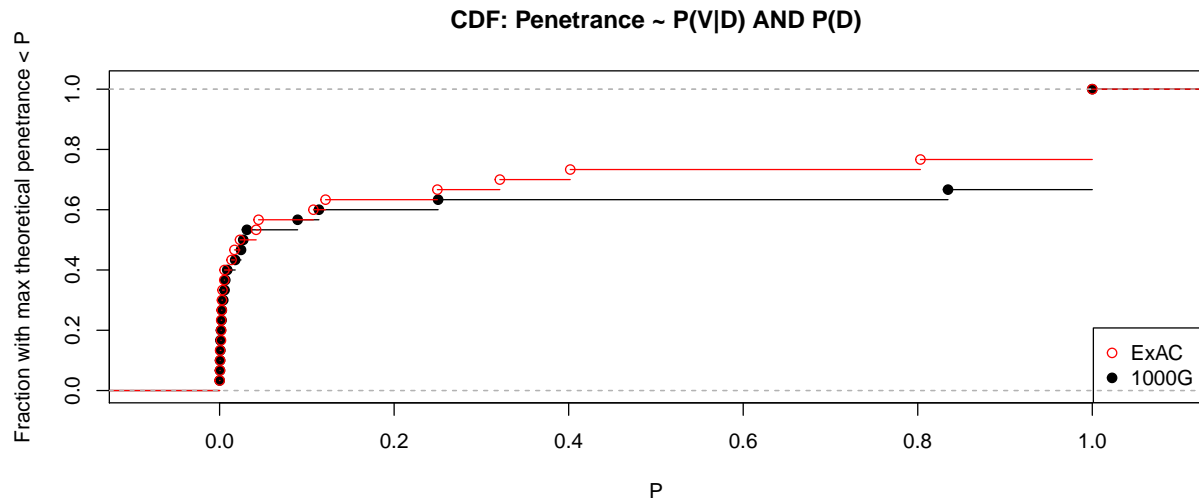
3.2 Penetrance Estimates by Ancestry



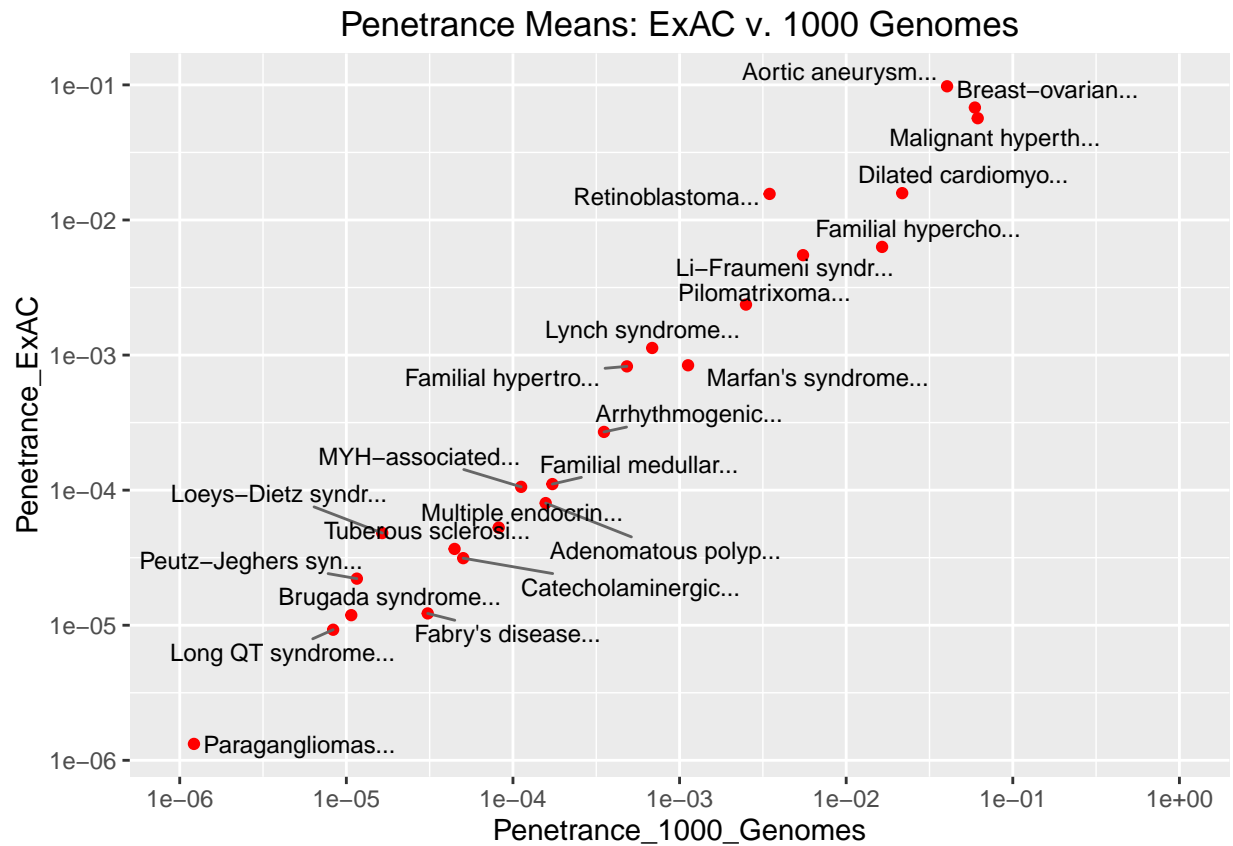
## Penetrance by Ancestry (ExAC)



### 3.3 Empirical CDFs for All Penetrance Plots



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



The Pearson correlation is 0.89.

Max penetrance values computed using 1000 Genomes are 0.64-fold larger than those computed using ExAC.