

# ClinVar Report

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**Sourcing ClinVar input from:** clinvar\_2013-08-08.vcf

**Sending output to:** Report\_2013-08-08.pdf

# 1 Collect and Merge ClinVar Data

## 1.1 Import ClinVar VCF

## 1.2 Merge ClinVar with 1000 Genomes and ExAC

## Breakdown of ClinVar Variants

Subset_ClinVar	Number_of_Variants
Total ClinVar	68182
LP/P	14125
ACMG LP/P	1614
ACMG LP/P in gnomAD	436
ACMG LP/P in ExAC	314
ACMG LP/P in 1000 Genomes	82

## Breakdown of ACMG-gnomAD Variants

Subset_gnomAD	Number_of_Variants
ACMG in gnomAD	96742
ClinVar-ACMG in gnomAD	2963
LP/P-ACMG in gnomAD	436

## Breakdown of ACMG-ExAC Variants

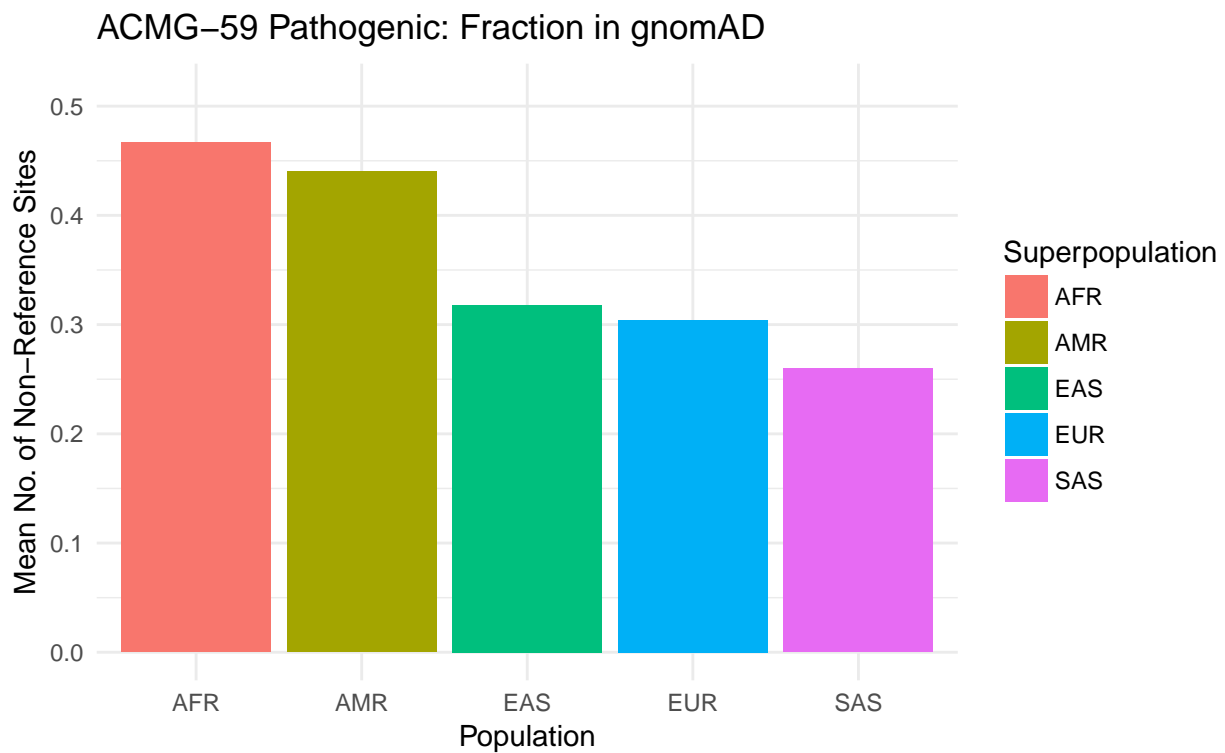
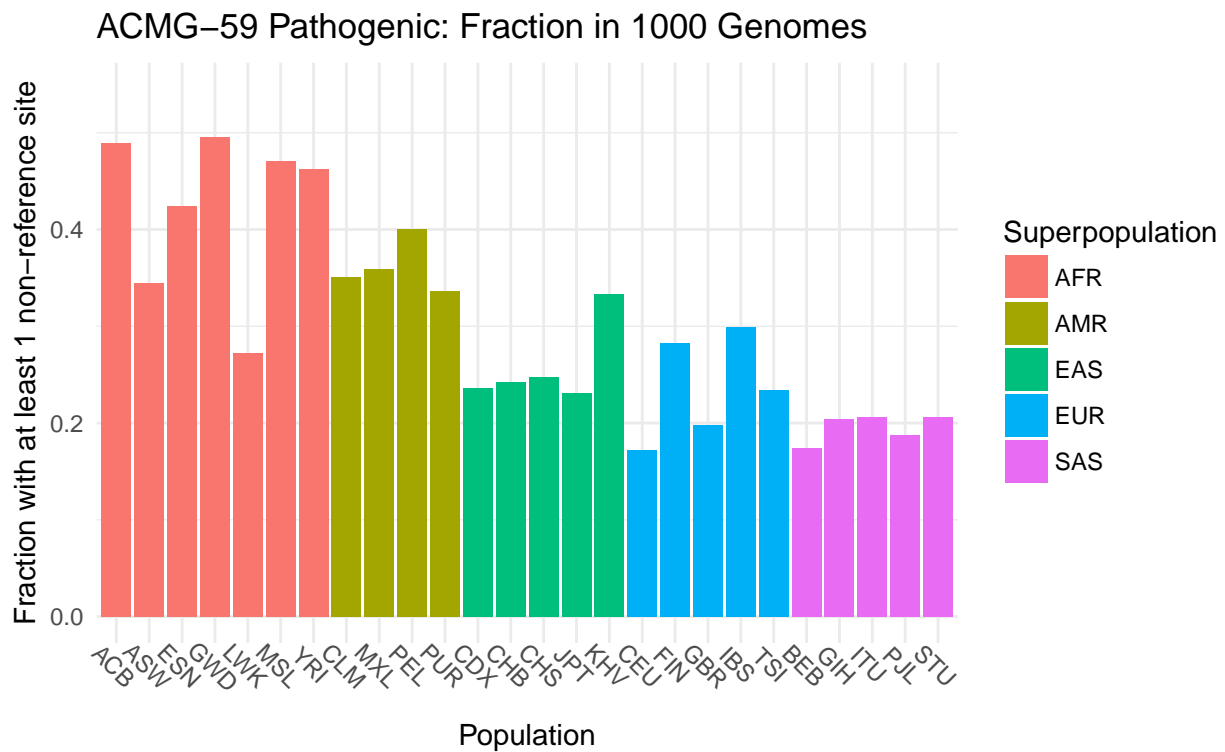
Subset_gnomAD	Number_of_Variants
ACMG in ExAC	59883
ClinVar-ACMG in ExAC	2419
LP/P-ACMG in ExAC	314

## Breakdown of ACMG-1000G Variants

Subset_gnomAD	Number_of_Variants
ACMG in 1000G	141466
ClinVar-ACMG in 1000G	1300
LP/P-ACMG in 1000G	82

## 2 Summary Statistics

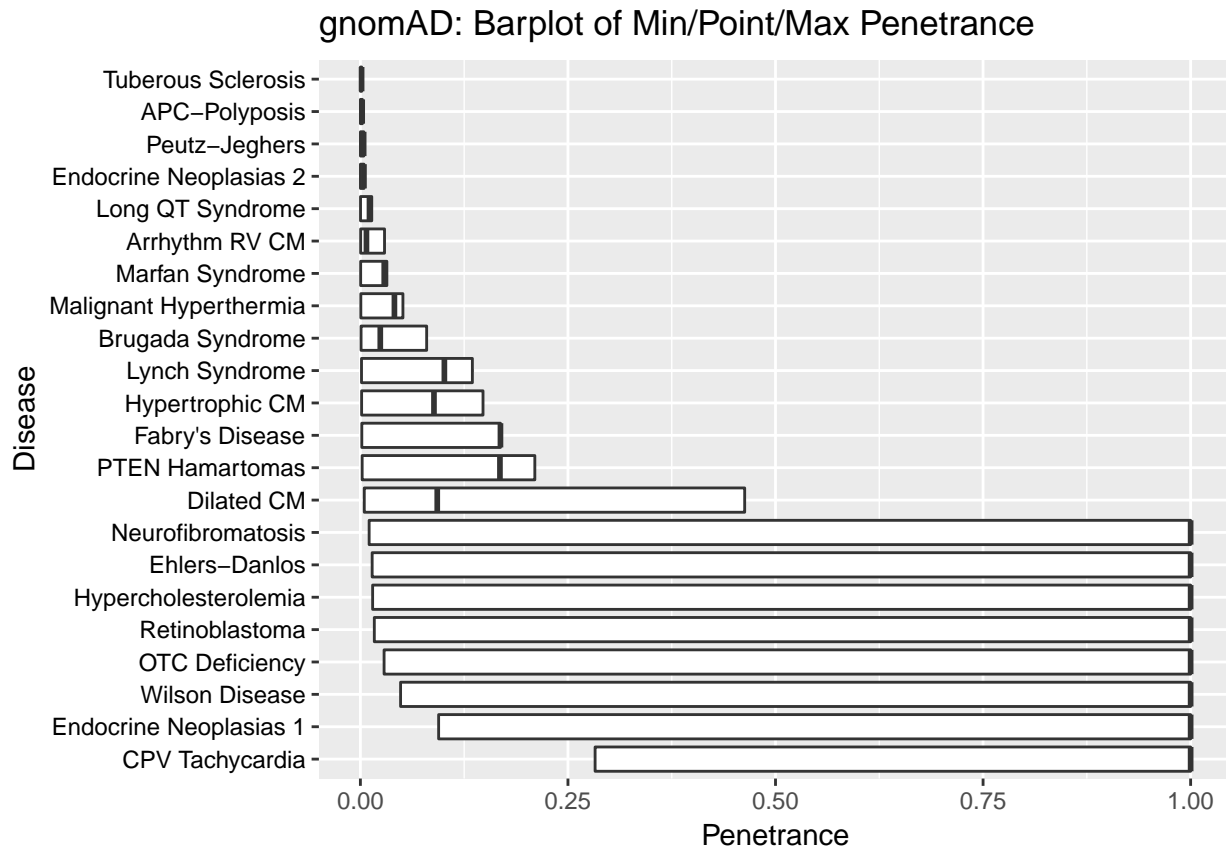
### 2.1 Fraction of Individuals with Pathogenic Non-Reference Sites



### 3 Penetrance Estimates

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

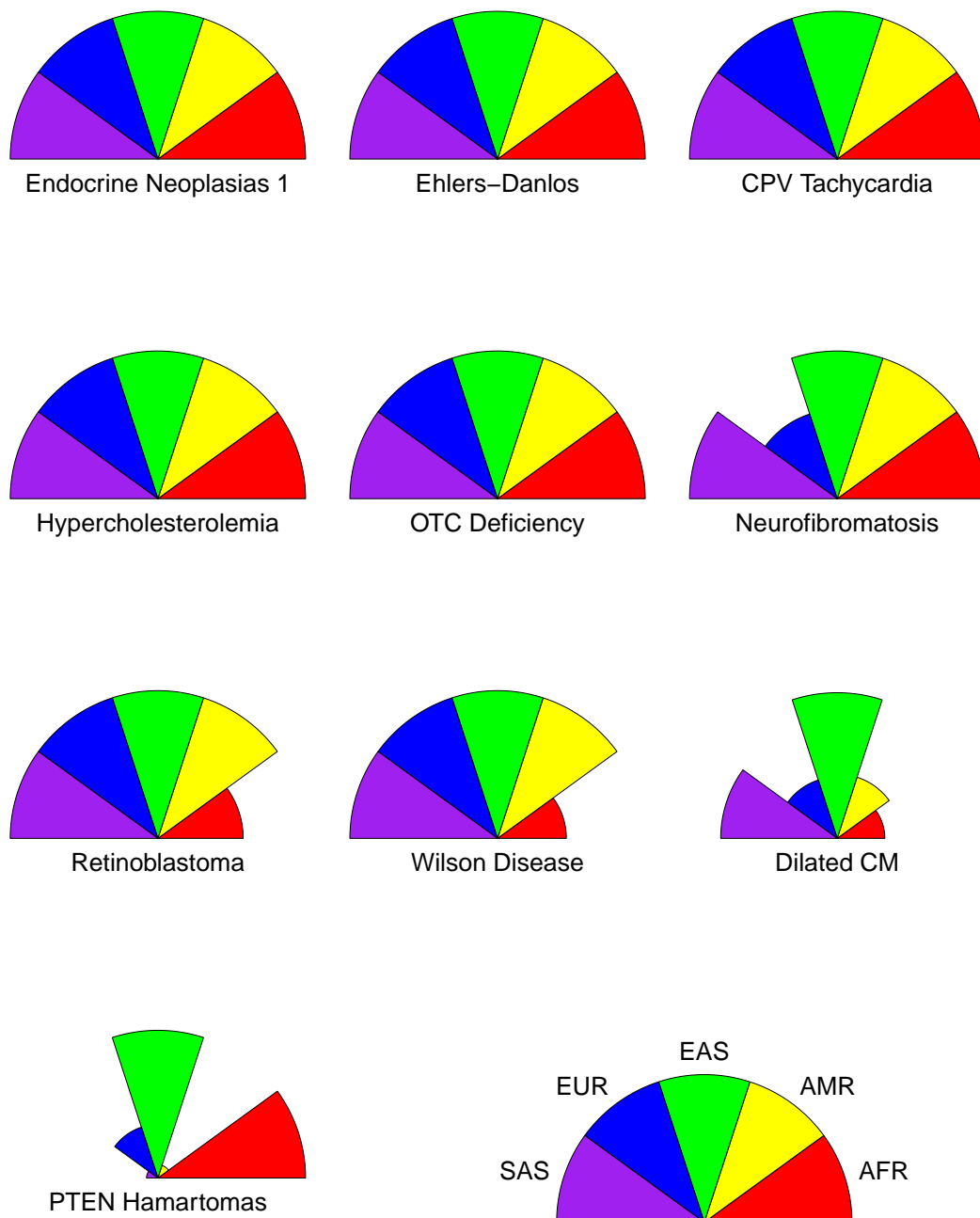
The left end of the boxplot indicates  $P(V|D) = 0.01$ ,  
the bold line in the middle indicates  $P(V|D) = \text{point value}$ ,  
the right end of the boxplot indicates  $P(V|D) = 1$ .



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

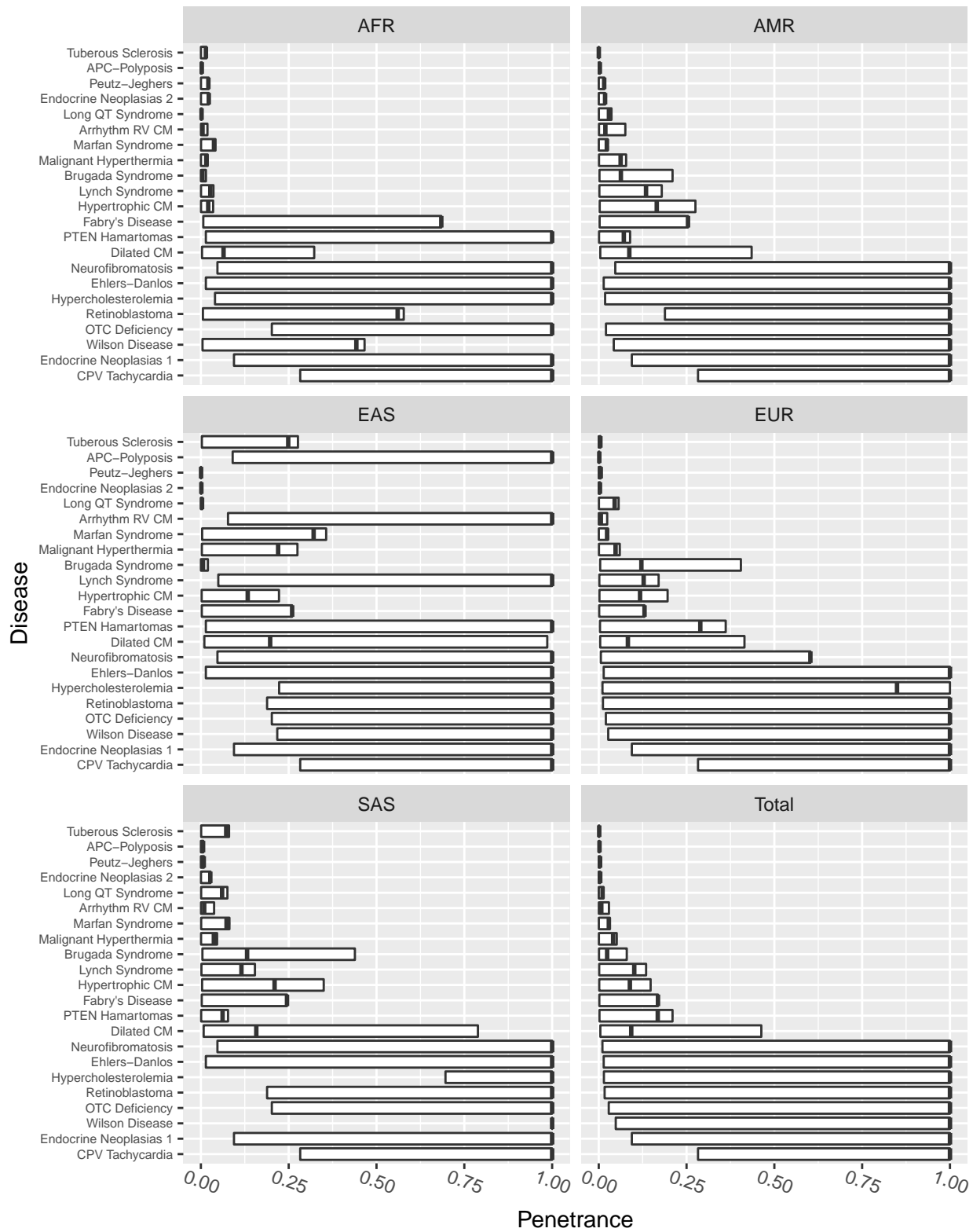
#### Radar Plot: Max Penetrance by Ancestry (gnomAD)



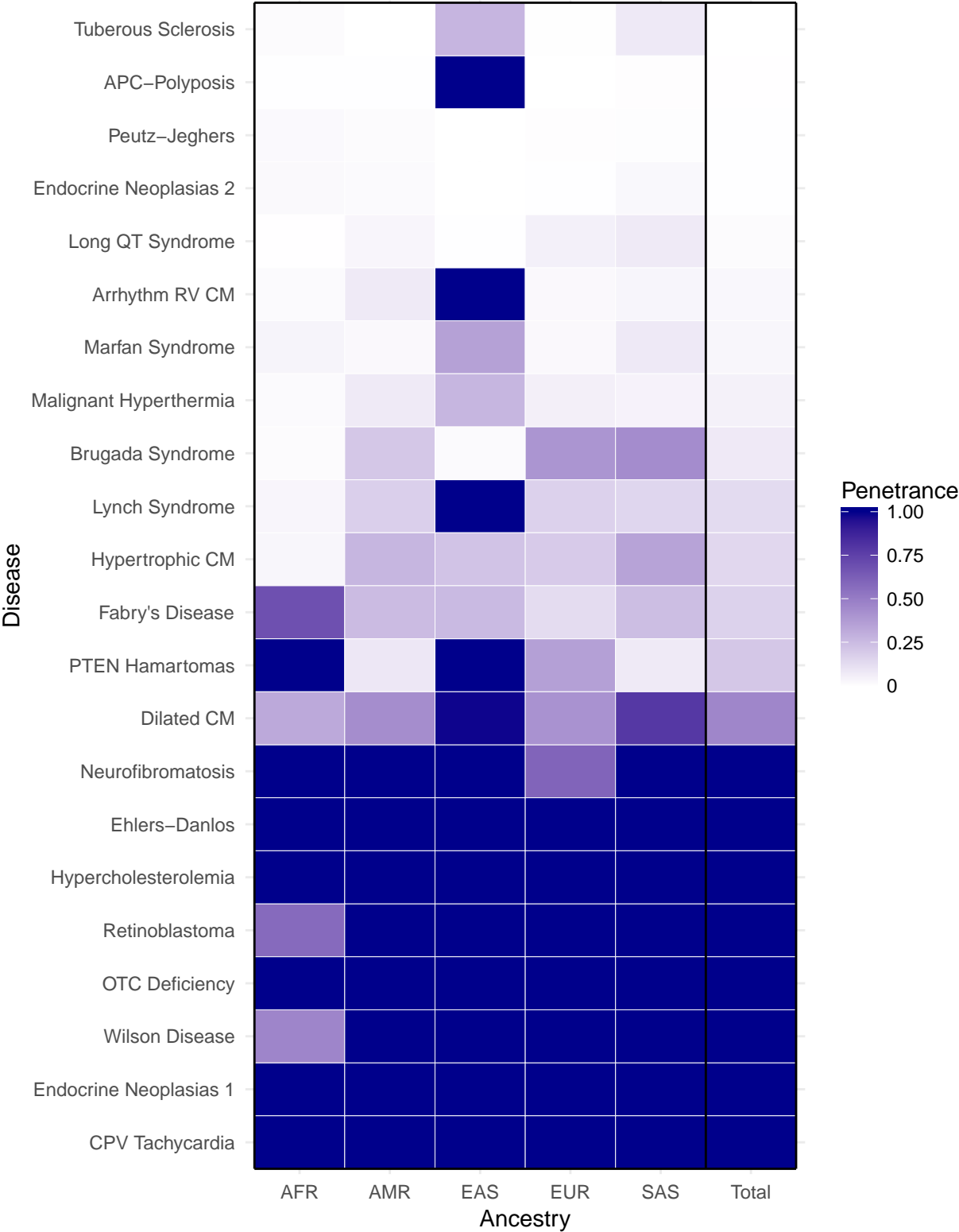
## [1] These are the top 10 diseases by summed allele frequencies. NULL values are not plotted.

## [1] Each radius is proportional to the penetrance of the disease in the given population.

Barplot: Penetrance by Ancestry (gnomAD)



Heatmap: Max Penetrance by Ancestry (gnomAD)



## Dark gray boxes are NA: no associated variants discovered in that ancestral population.