ClinVar Report

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Contents

Sourcing ClinVar input from: clinvar_2013-12-30.vcf
Sending output to: Report_2013-12-30.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	42990
LP/P	16854
ACMG LP/P	2091
ACMG LP/P in gnomAD	495
ACMG LP/P in ExAC	343
ACMG LP/P in 1000 Genomes	90

```
## Breakdown of ACMG-gnomAD Variants
```

Subset_gnomAD	Number_of_Variants
ACMG in gnomAD	96742
ClinVar-ACMG in gnomAD	3129
LP/P-ACMG in gnomAD	495

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

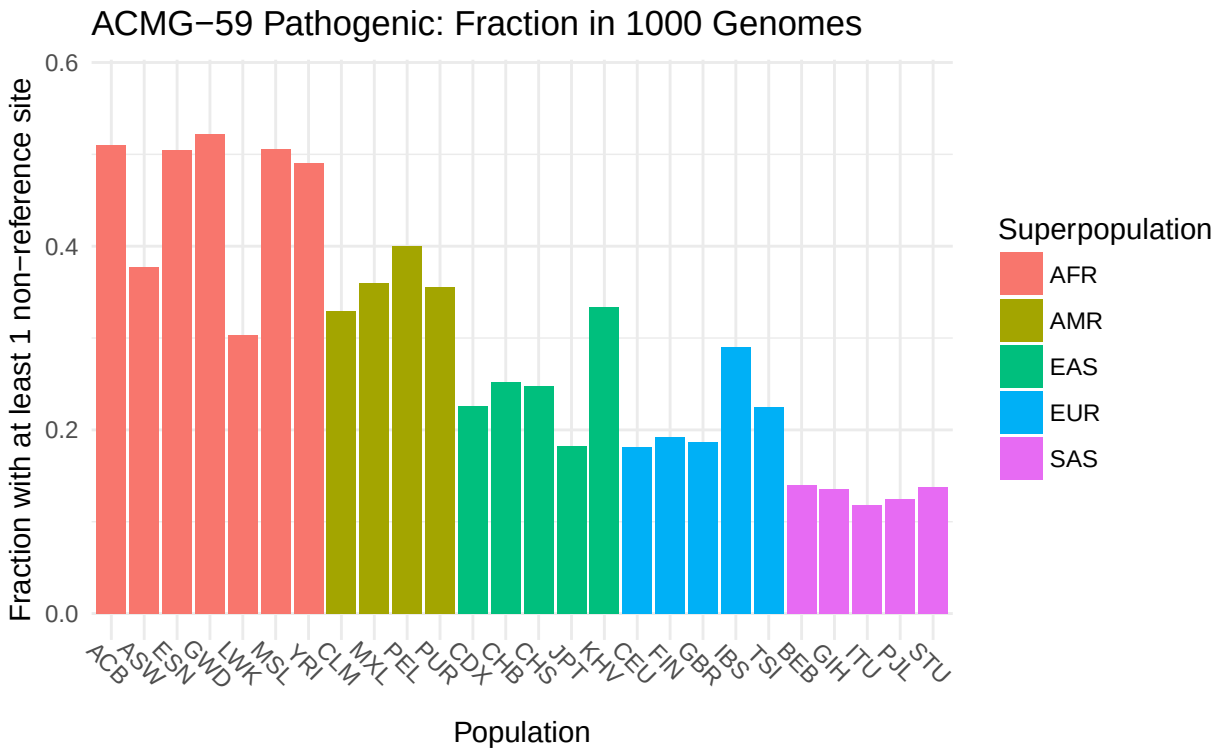
Subset_gnomAD	Number_of_Variants
ACMG in ExAC	59883
ClinVar-ACMG in ExAC	2600
LP/P-ACMG in ExAC	343

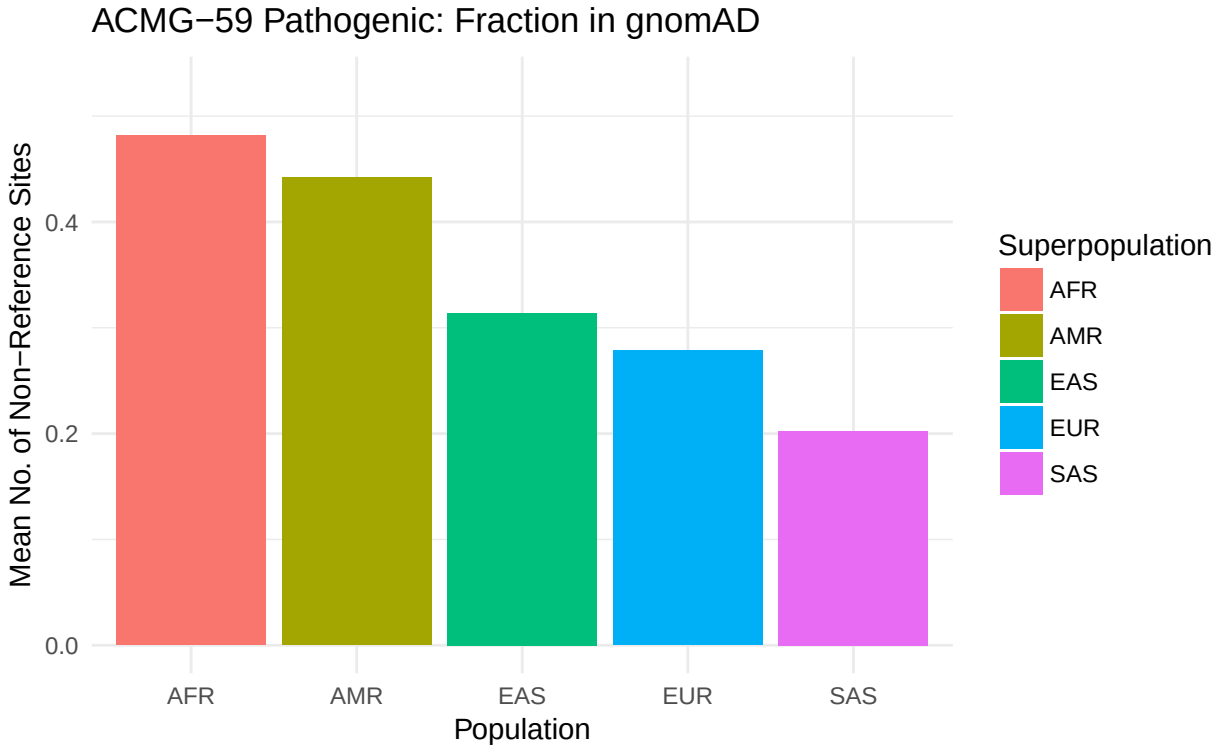
```
## Breakdown of ACMG-1000G Variants
```

Subset_gnomAD	Number_of_Variants
ACMG in 1000G	141466
ClinVar-ACMG in 1000G	1287
LP/P-ACMG in 1000G	90

2 Summary Statistics

2.1 Fraction of Individuals with Pathogenic Non-Reference Sites

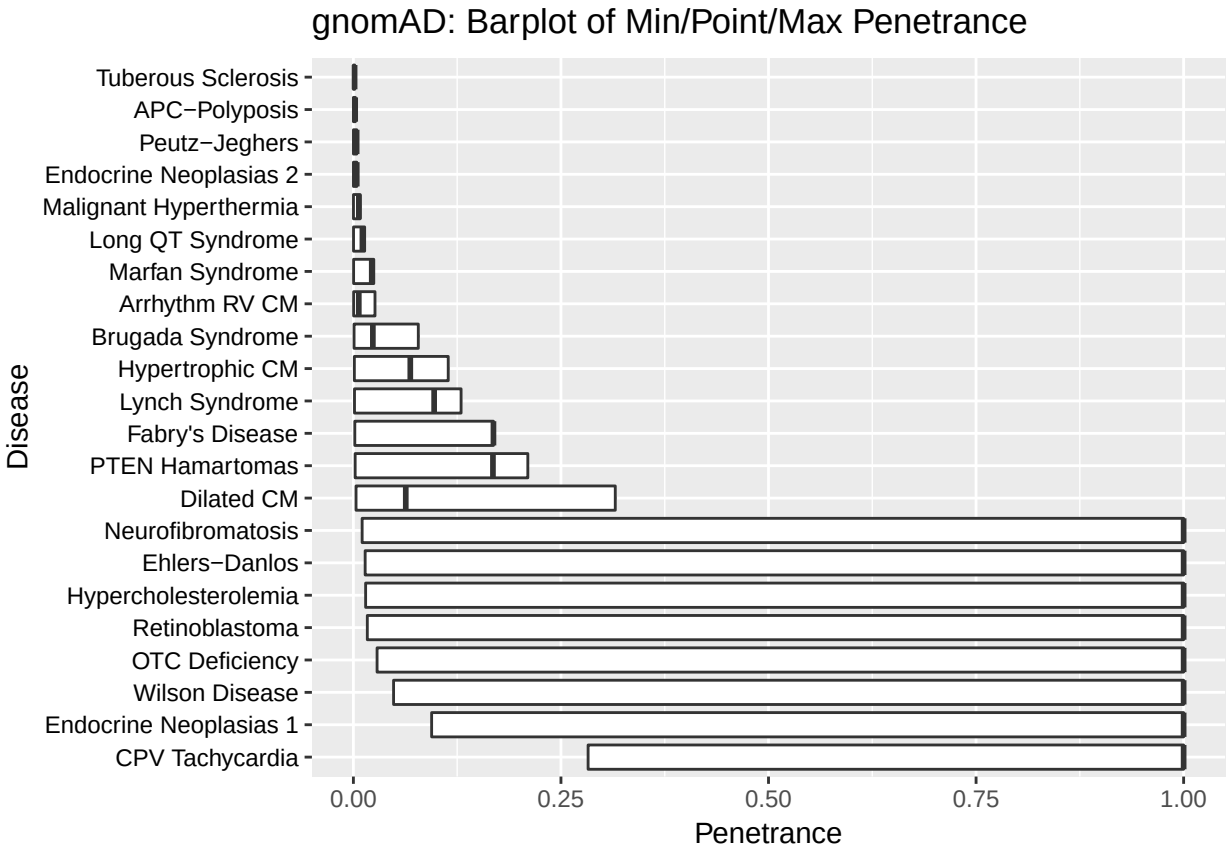




3 Penetrance Estimates

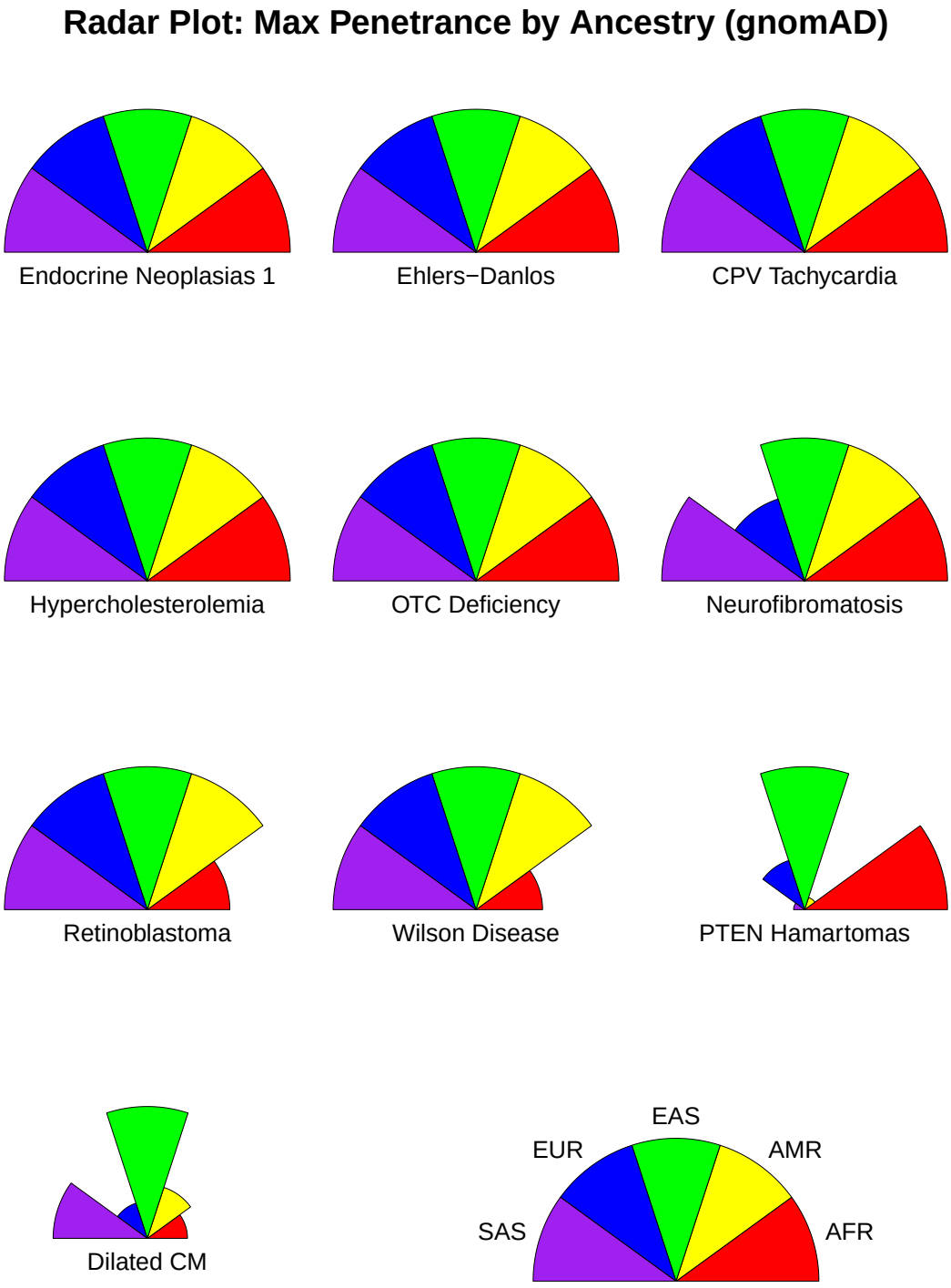
3.1 Max/Min Penetrance as a Function of P(D) and P(V|D)

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

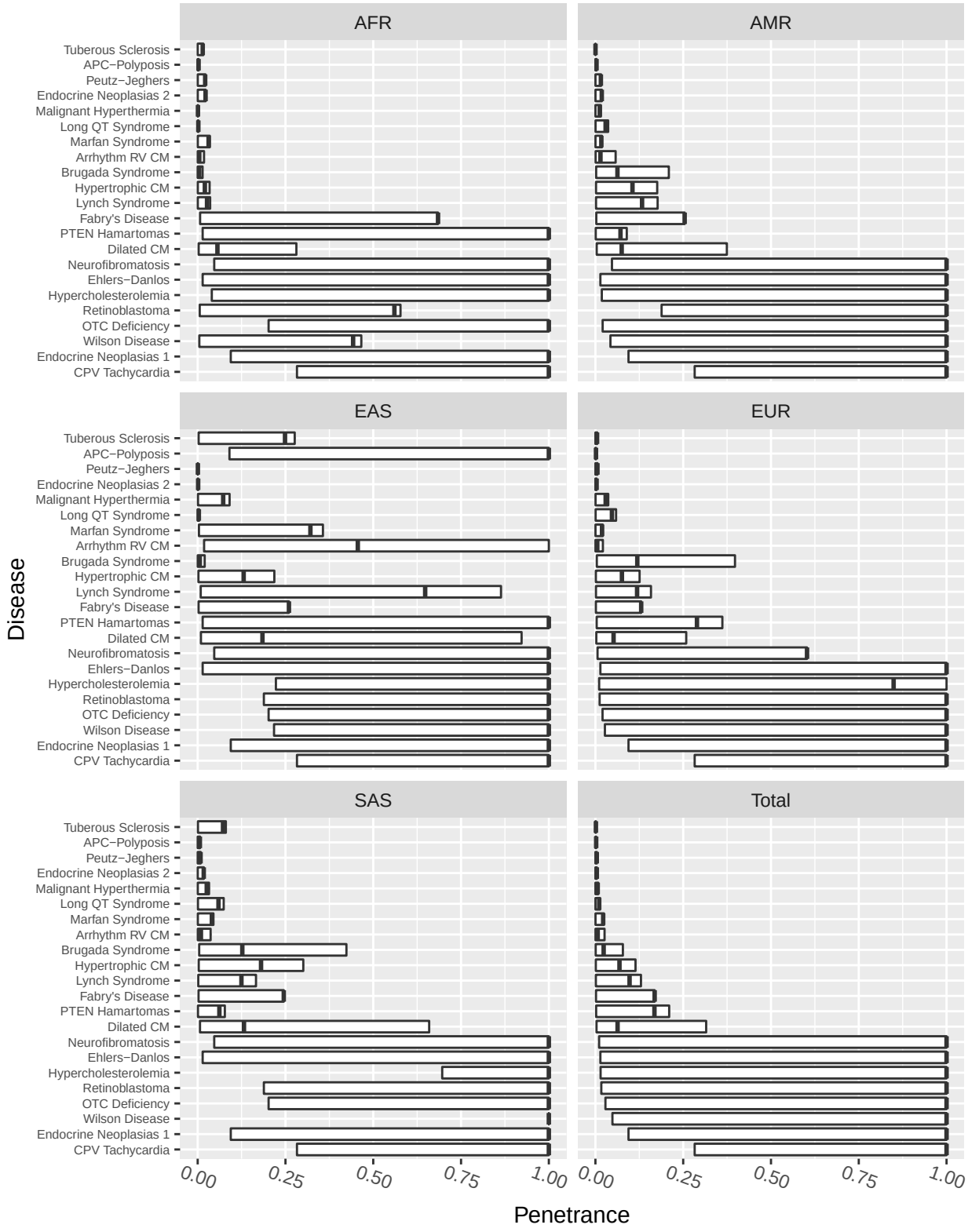


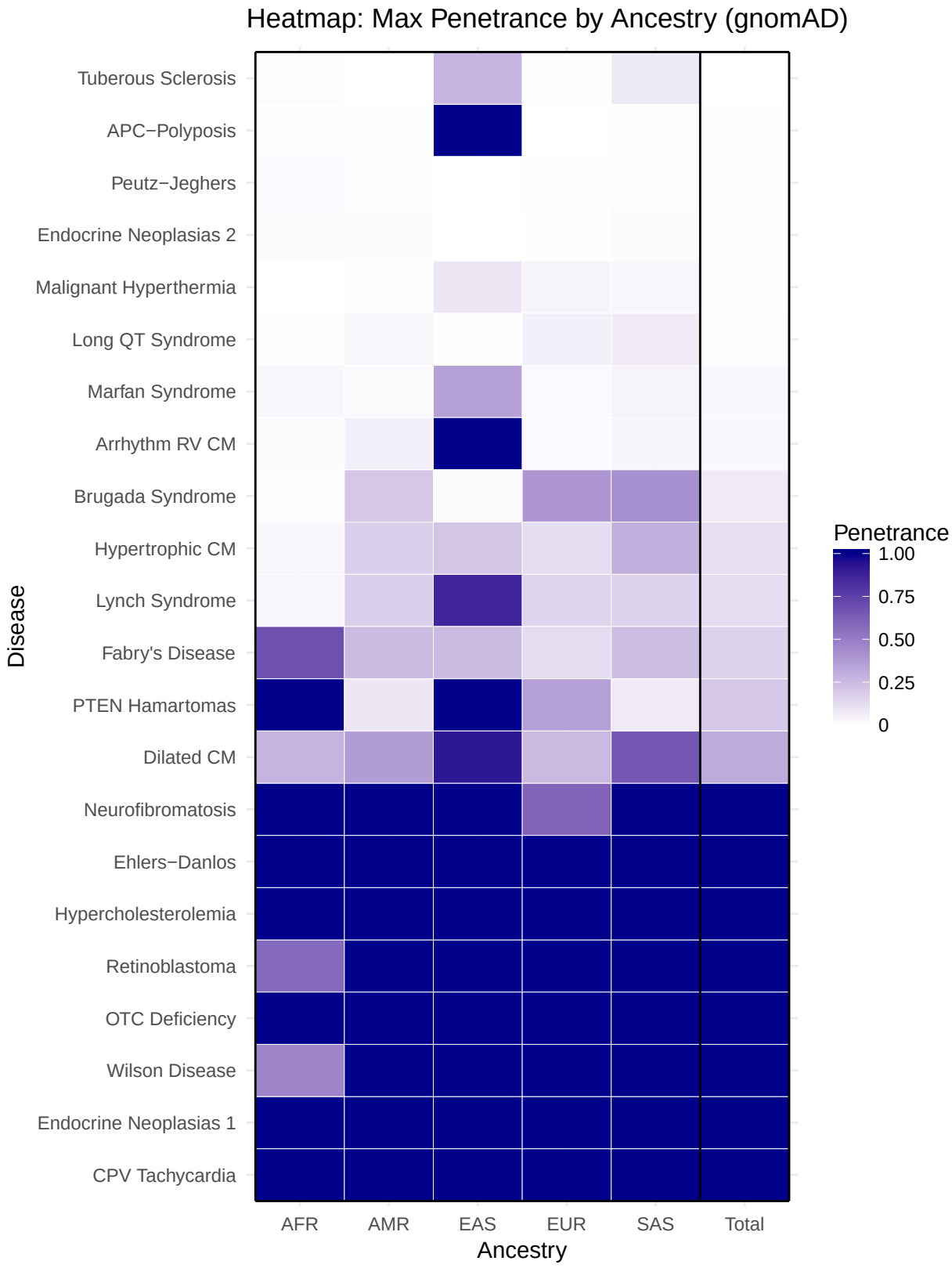
Note: Some diseases have mean theoretical penetrance = 1 because the assumed allelic heterogeneity is greater than is possible, given the observed prevalence and allele frequencies.

3.2 Penetrance Estimates by Ancestry




```
## [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)
AFR
AMR
EAS
EUR
SAS
Total
Disease
Tuberous Sclerosis
APC−Polyposis
Peutz−Jeghers
Endocrine Neoplasias 2
Malignant Hyperthermia
Long QT Syndrome
Marfan Syndrome
Arrhythm RV CM
Brugada Syndrome
Hypertrophic CM
Lynch Syndrome
Fabry's Disease
PTEN Hamartomas
Dilated CM
Neurofibromatosis
Ehlers−Danlos
Hypercholesterolemia
Retinoblastoma
OTC Deficiency
Wilson Disease
Endocrine Neoplasias 1
CPV Tachycardia
0.00
0.25
0.50
0.75
1.00
Penetrance



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