ClinVar Report

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Contents

Sourcing ClinVar input from: clinvar_2014-06-04.vcf
Sending output to: Report_2014-06-04.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	77997
LP/P	17835
ACMG LP/P	3660
ACMG LP/P in gnomAD	803
ACMG LP/P in ExAC	556
ACMG LP/P in 1000 Genomes	118

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

Subset_gnomAD	Number_of_Variants
ACMG in gnomAD	96742
ClinVar-ACMG in gnomAD	4264
LP/P-ACMG in gnomAD	803

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

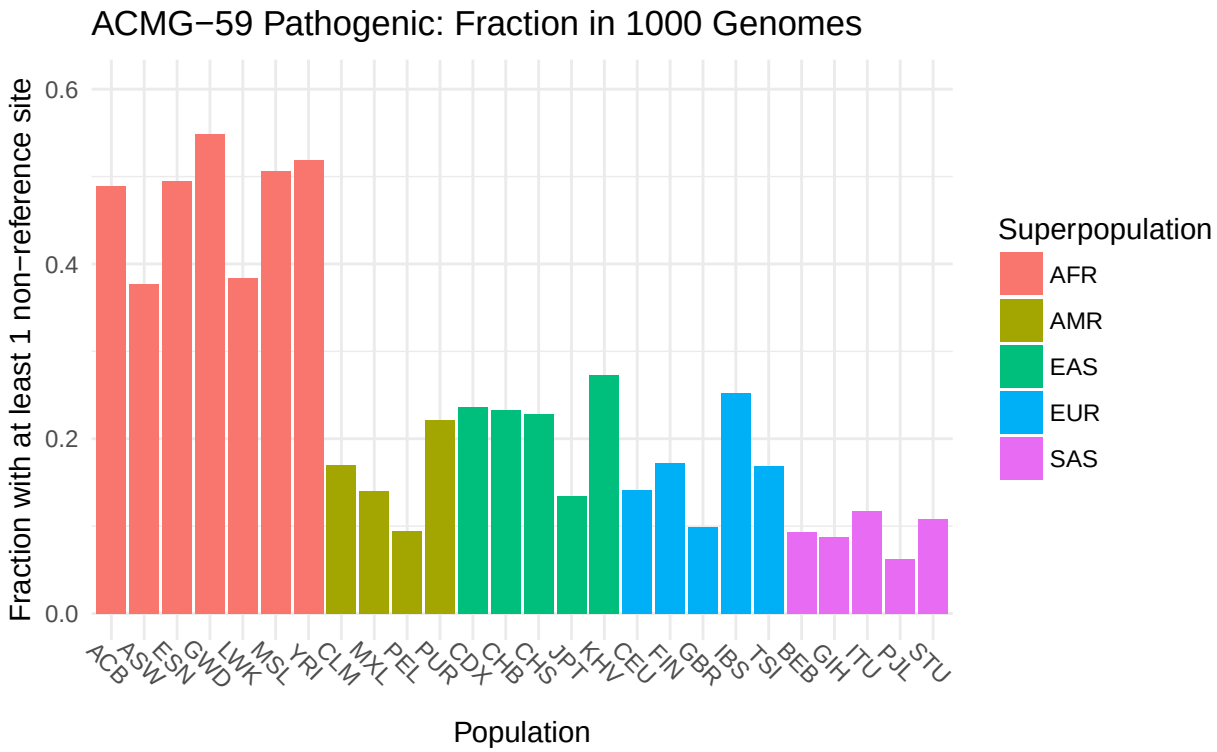
Subset_gnomAD	Number_of_Variants
ACMG in ExAC	59883
ClinVar-ACMG in ExAC	3519
LP/P-ACMG in ExAC	556

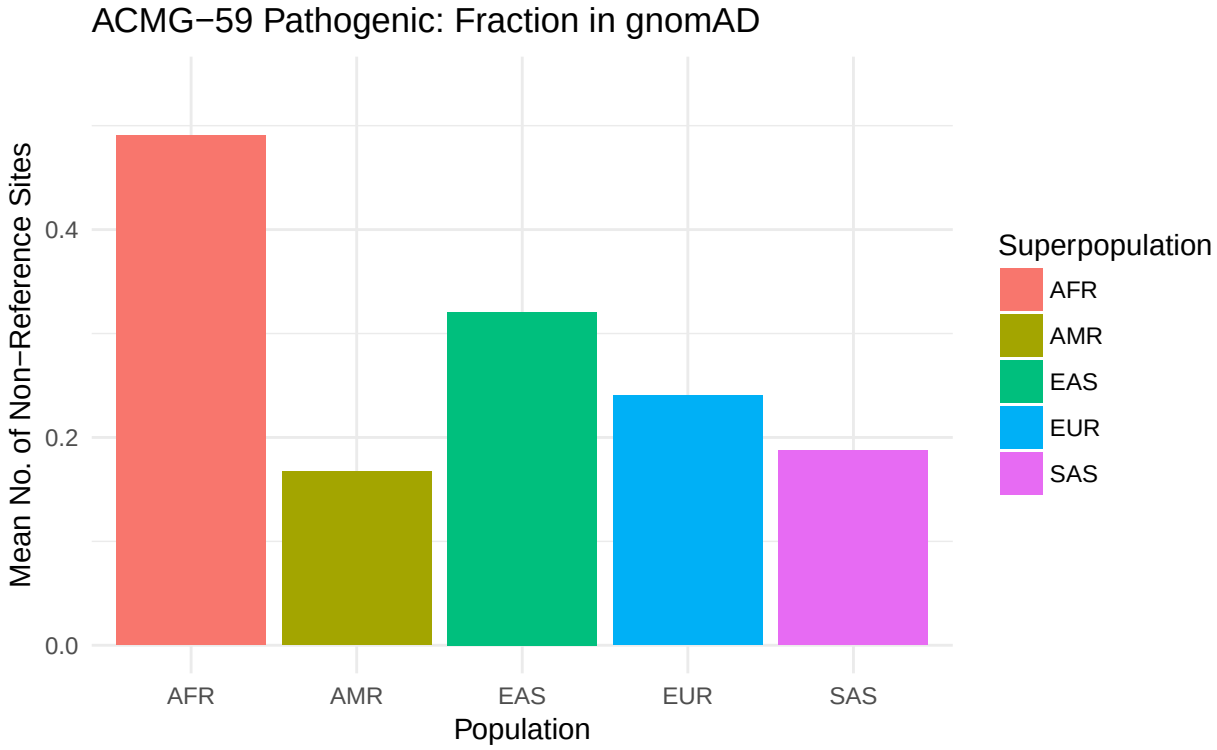
```
## Breakdown of ACMG-1000G Variants
```

Subset_gnomAD	Number_of_Variants
ACMG in 1000G	141466
ClinVar-ACMG in 1000G	1748
LP/P-ACMG in 1000G	118

2 Summary Statistics

2.1 Fraction of Individuals with Pathogenic Non-Reference Sites

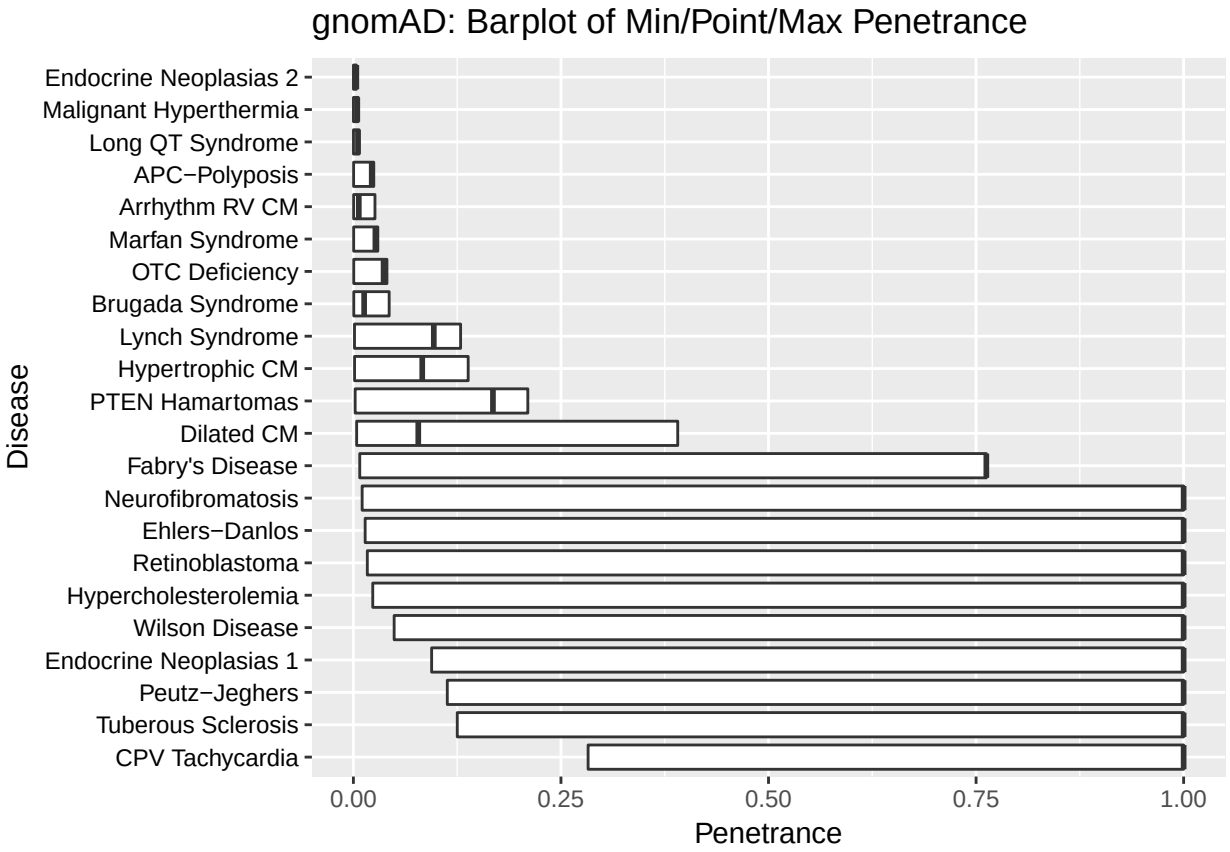




3 Penetrance Estimates

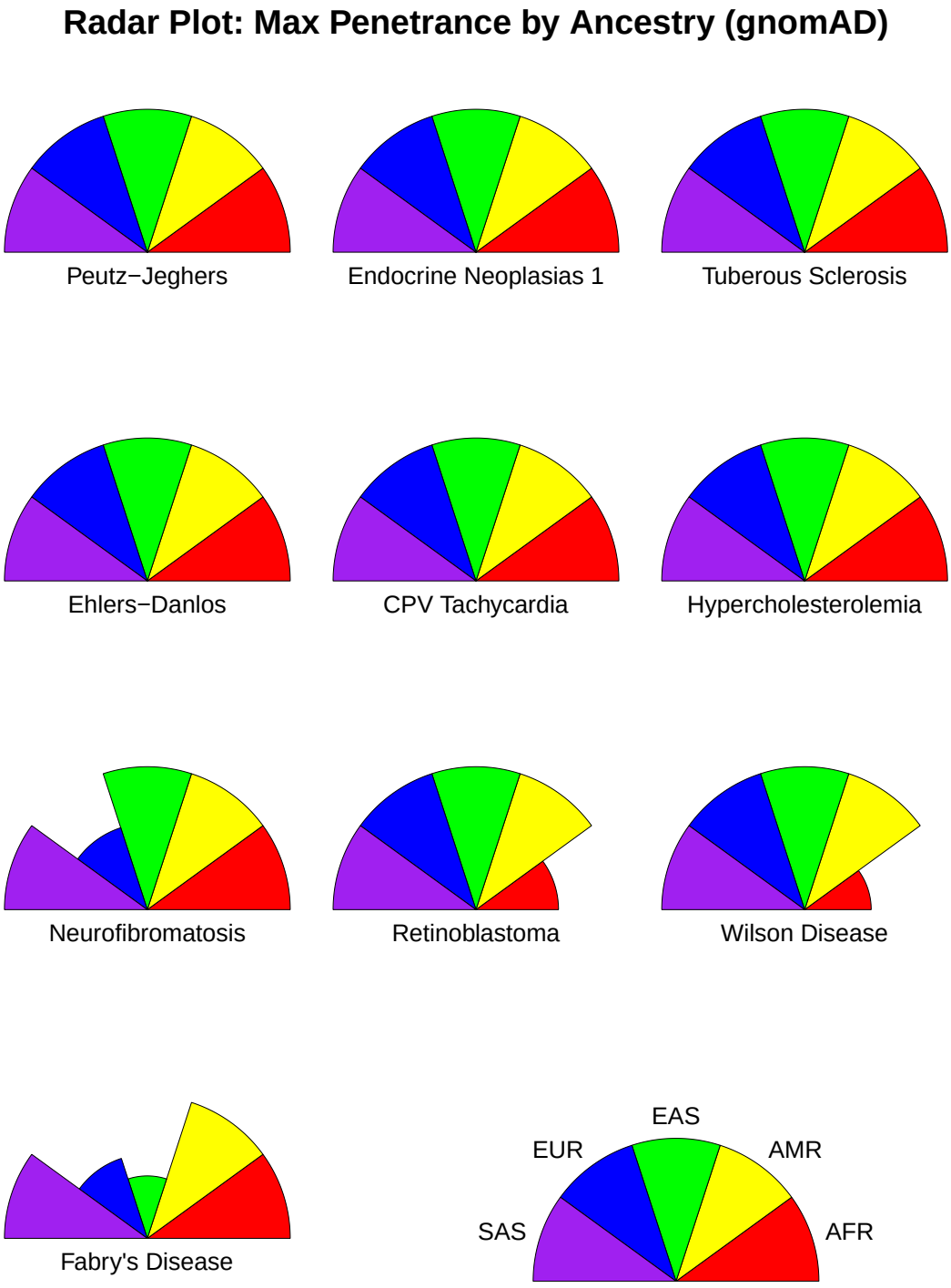
3.1 Max/Min Penetrance as a Function of P(D) and P(V|D)

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.

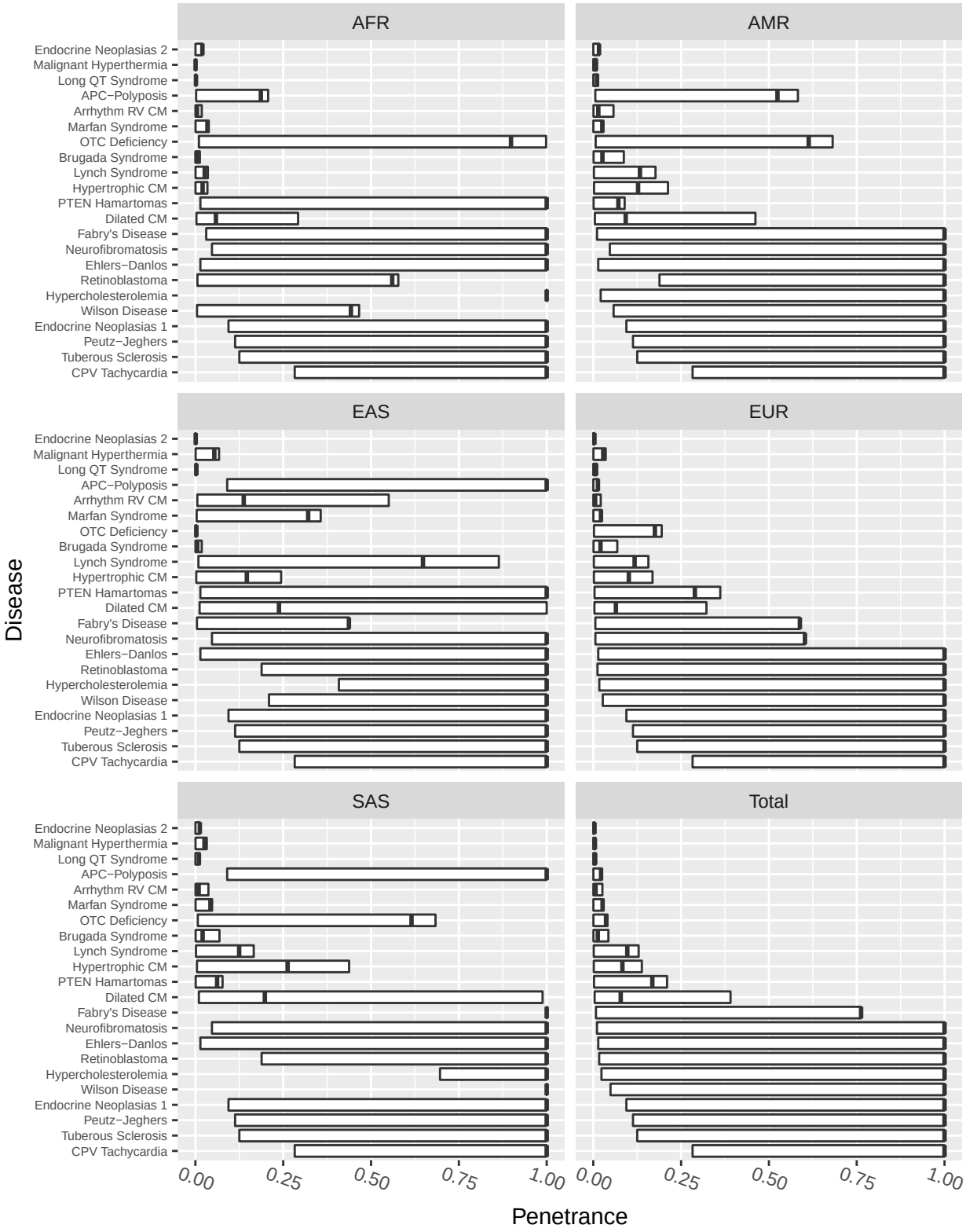


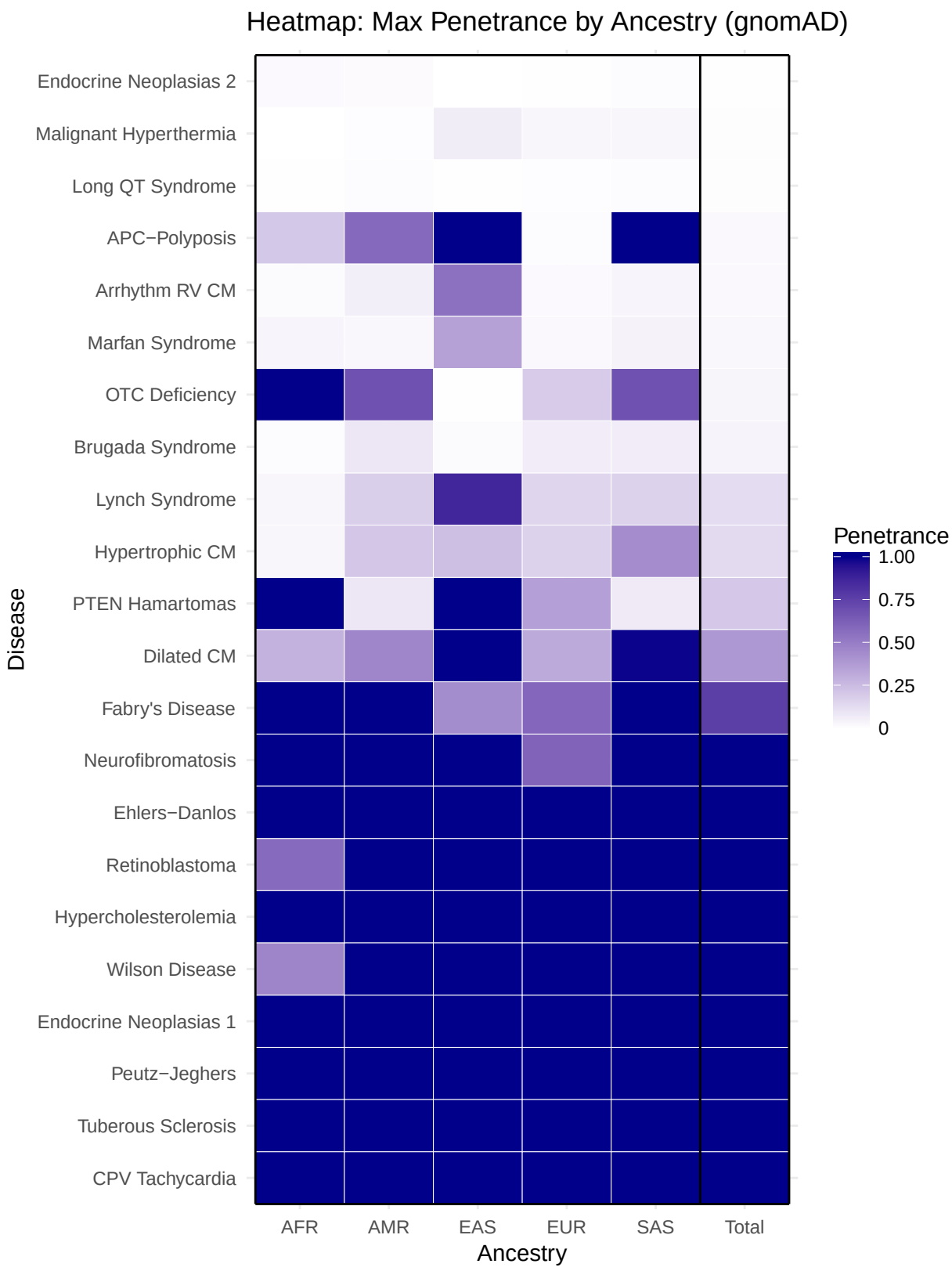
Note: Some diseases have mean theoretical penetrance = 1 because the assumed allelic heterogeneity is greater than is possible, given the observed prevalence and allele frequencies.

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



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