

COLOC visualisation

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2020-07-08

```
knitr::opts_chunk$set(warning = FALSE, echo = FALSE, message = FALSE)
library(tidyverse)
```

```
## -- Attaching packages -----
## v ggplot2 3.3.2      v purrr  0.3.4
## v tibble  3.0.1      v dplyr  1.0.0
## v tidyr   1.1.0      v stringr 1.4.0
## v readr   1.3.1      v forcats 0.5.0

## Warning: package 'ggplot2' was built under R version 3.6.2
## Warning: package 'tibble' was built under R version 3.6.2
## Warning: package 'tidyr' was built under R version 3.6.2
## Warning: package 'purrr' was built under R version 3.6.2
## Warning: package 'dplyr' was built under R version 3.6.2

## -- Conflicts -----
## x dplyr::filter() masks stats::filter()
## x dplyr::lag()     masks stats::lag()
```

```
library(ggrepel)
```

Read in COLOC eQTL results

create separate disease plots for Daner and Stahl GWAS results.

```
##
##      Daner_2020      IMSGC_2019      Jansen_2018      Kunkle_2019
##      116865          400284          73334            45503
##      Lambert_2013    Marionni_2018  Nalls23andMe_2019  Nicolas_2018
##      44487           197941          241487           20717
##      Ripke_2014      Stahl_2019      Wray_2018
##      193878          66964           46652

##
##      DLPFC_ROSMAP      Microglia_all_regions
##      22897              31073
##      Microglia_all_regions_sQTL  Microglia_all_regions_Young
##      172996              23682
##      Microglia_MFG      Microglia_sQTL_MFG
##      30293              209370
##      Microglia_sQTL_STG  Microglia_sQTL_SVZ
##      211254              226093
##      Microglia_sQTL_THA      Microglia_STG
```

```
##                210780                30293
##      Microglia_SVZ                Microglia_THA
##                30293                30293
##      Microglia_Young                Monocytes_Fairfax
##                41545                30799
##      Monocytes_MyND                Monocytes_sQTL_MyND
##                31625                114826
```

Fixing AD loci

4 different GWAS - all use different names for the same locus

```
## [1] 41
## [1] "ADAMTS4"    "ABCA7"      "SCIMP"      "ADAM10"     "KAT8"
## [6] "PILRA"      "CASS4"      "SORL1"      "APH1B"      "PICALM"
## [11] "TREM2"      "APOE"       "AC074212.3" "CD2AP"      "HLA/DRB"
## [16] "ABI3"       "EPA1"       "CD33"       "CR1"        "MS4A6A"
## [21] "HS3ST1"     "CLU"        "BIN1"       "BZRAP1-AS1" "SLC24A4"
## [26] "INPP5D"     "ECHDC3"     "CLNK"       "HESX1"      "SUZ12P1"
## [31] "ALPK2"      "CNTNAP2"    "SPI1"       "IQCK"       "ACE"
## [36] "FERMT2"     "WVOX"       "ADAMTS1"    "MEF2C"      "NME8"
## [41] "DSG2"
```

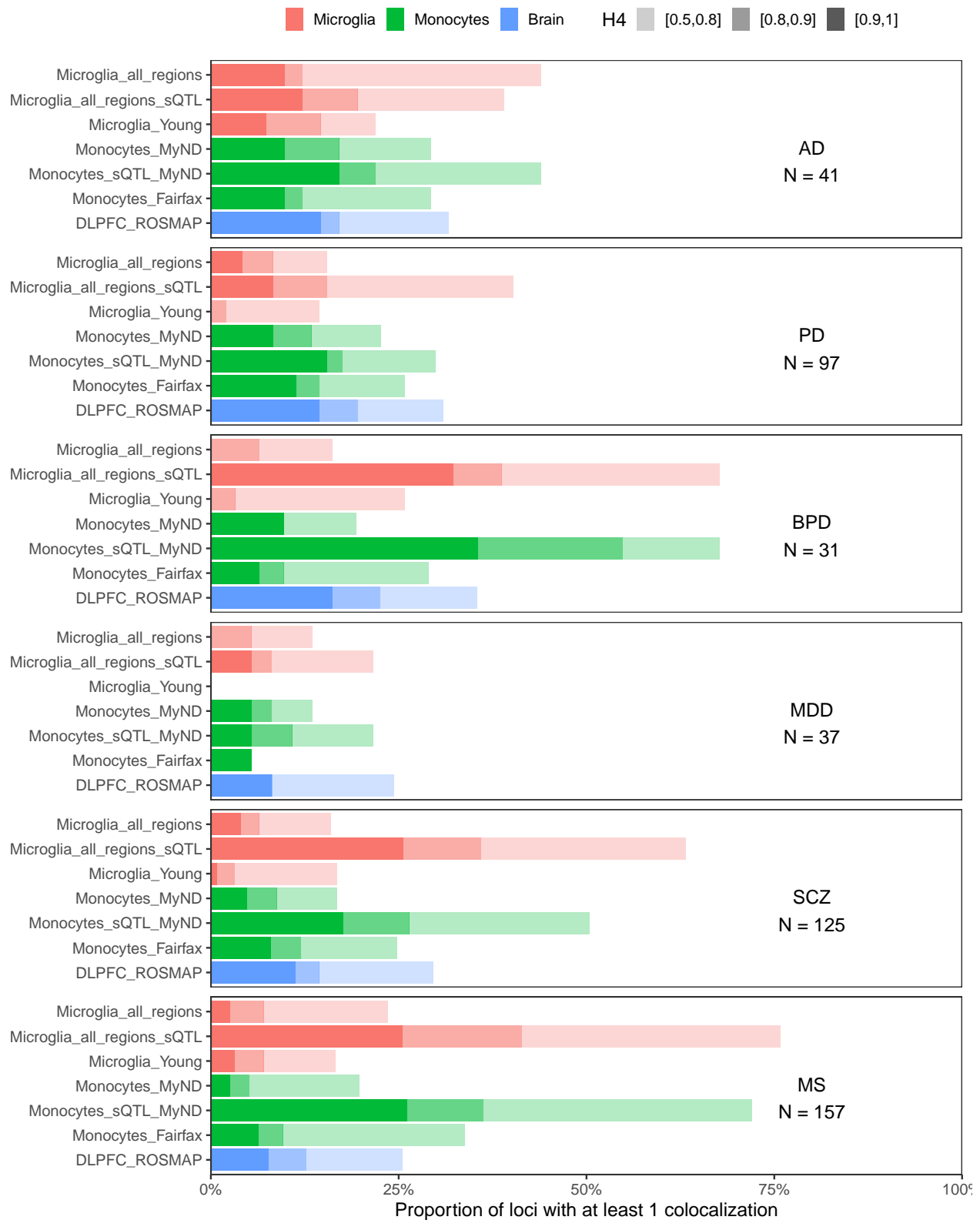
Summarising COLOC results

How many COLOC hits at 0.8 in each disease?

multiple GWAS for the same disease - same locus comes up multiple times sQTLs - multiple junctions for each QTL

Summary counts

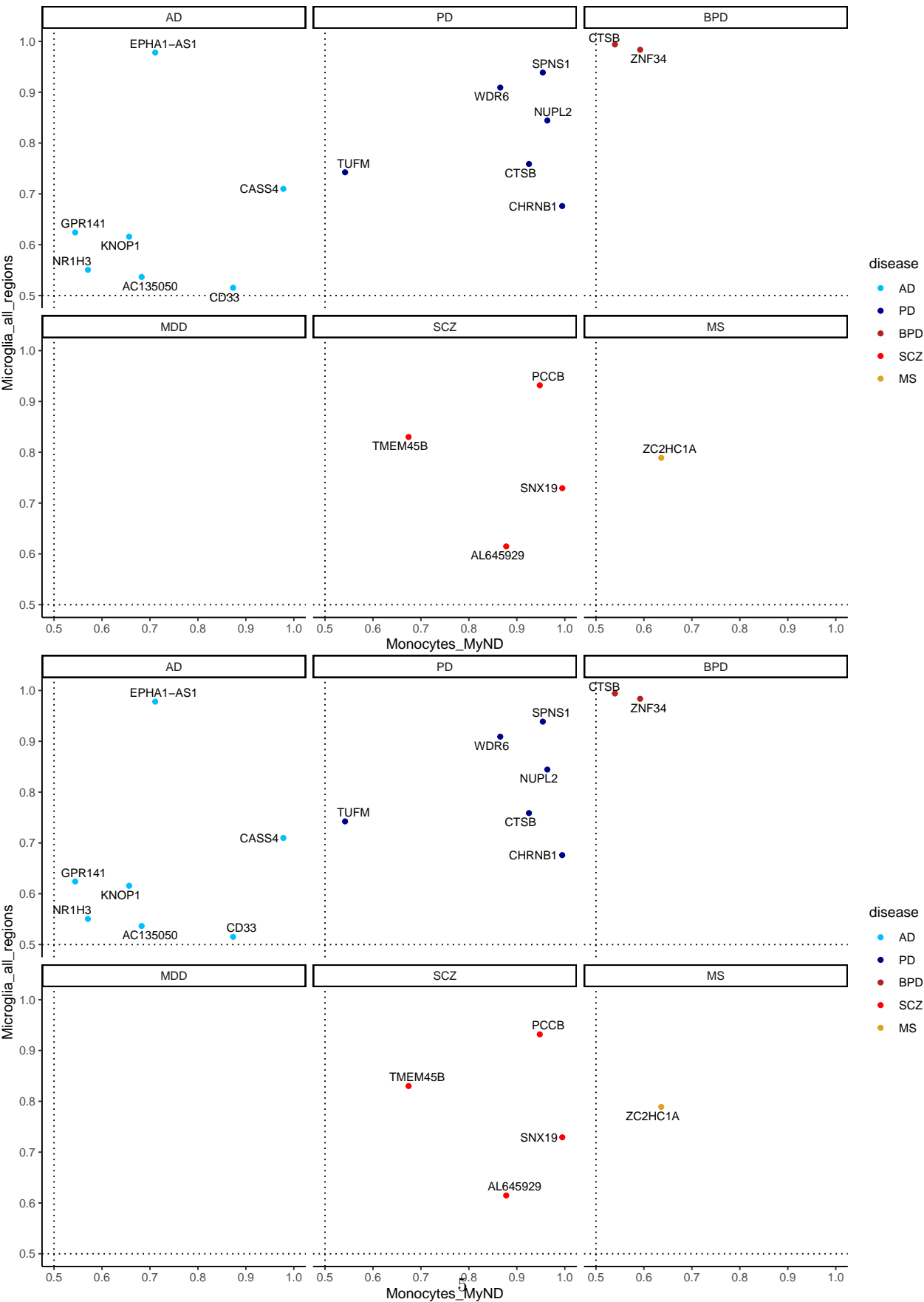
For each disease GWAS - or union of GWAS, of the total number of loci, how many have at least 1 COLOC $H4 > 0.8$?

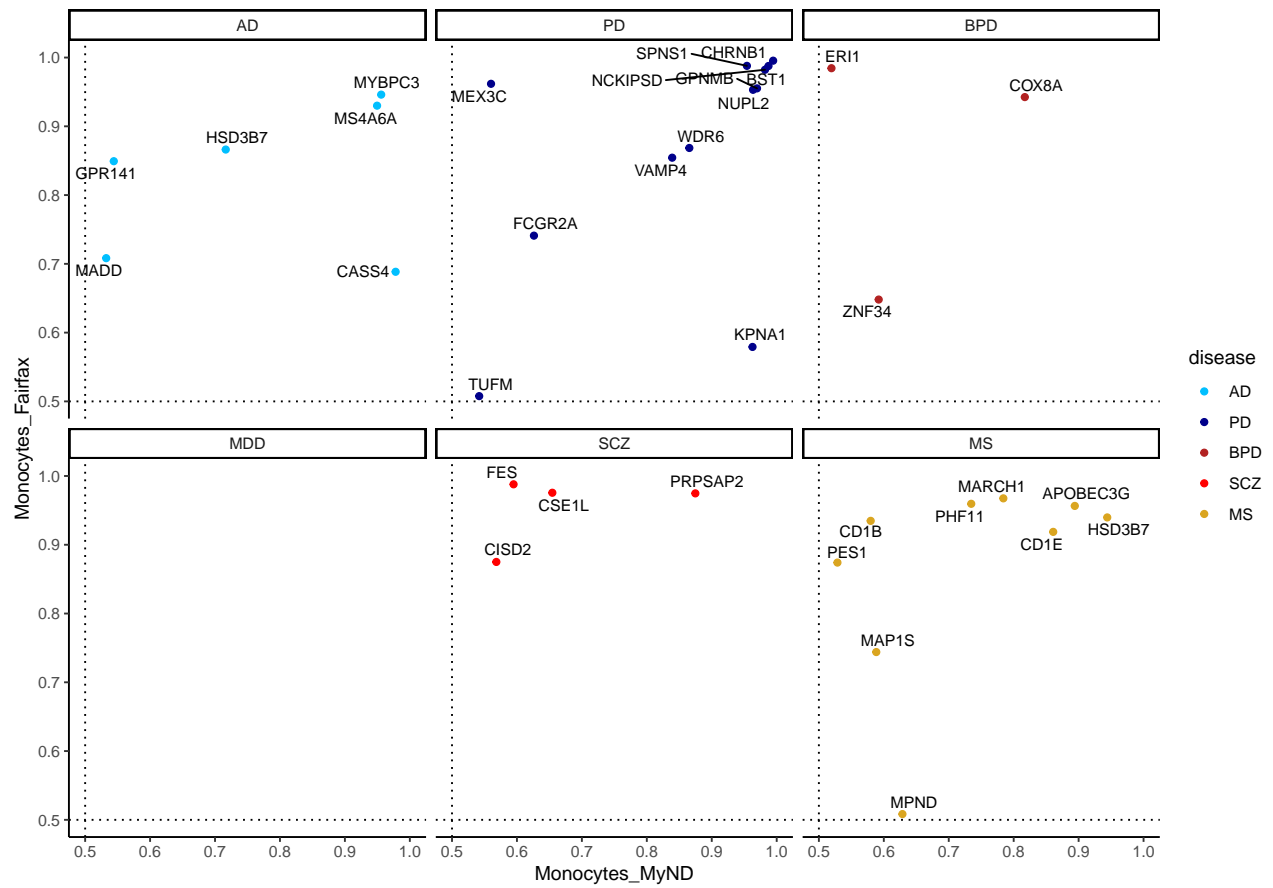


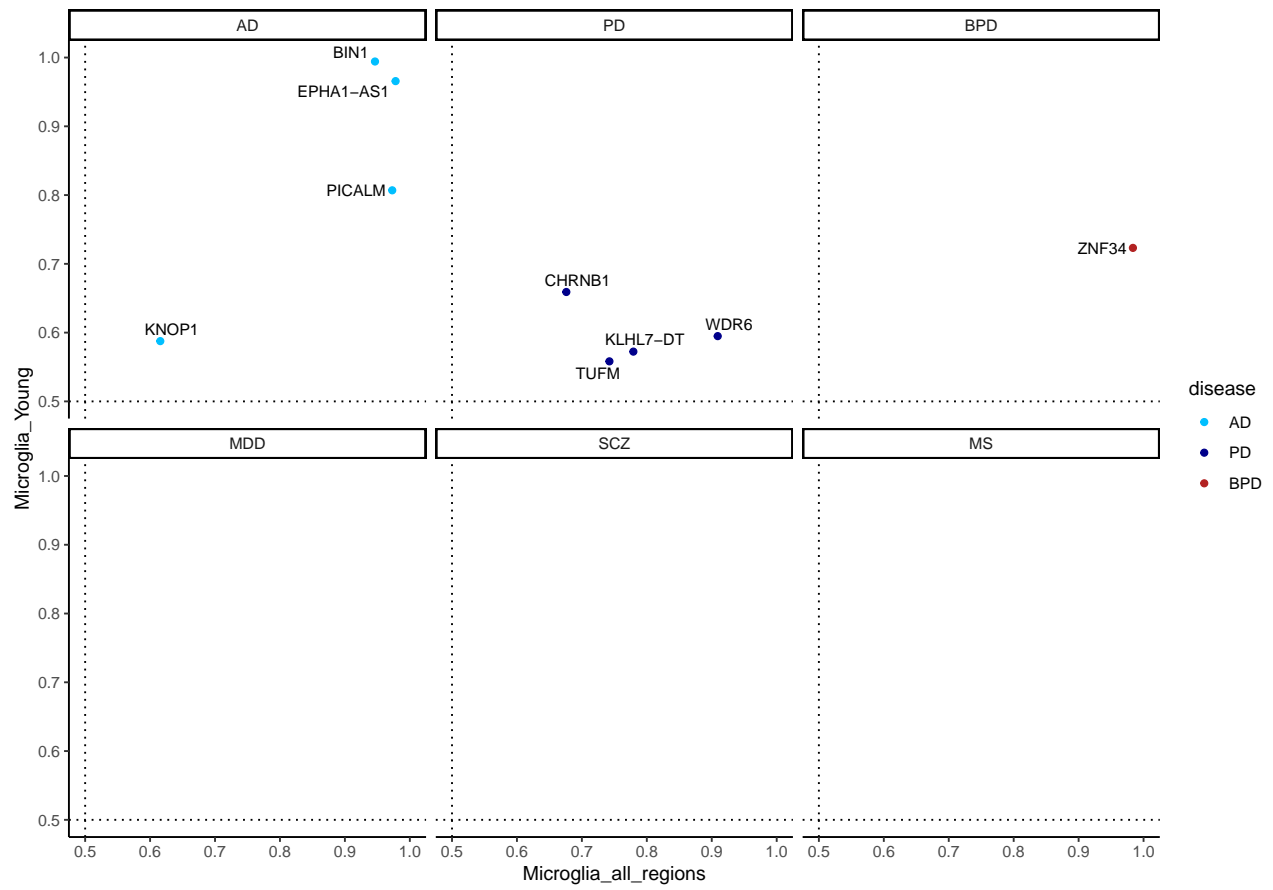
disease	QTL	n_loci	h4_0.5	h4_0.8	h4_0.9
AD	DLPFC_ROSMAP	41	6	1	6
AD	Monocytes_Fairfax	41	7	1	4

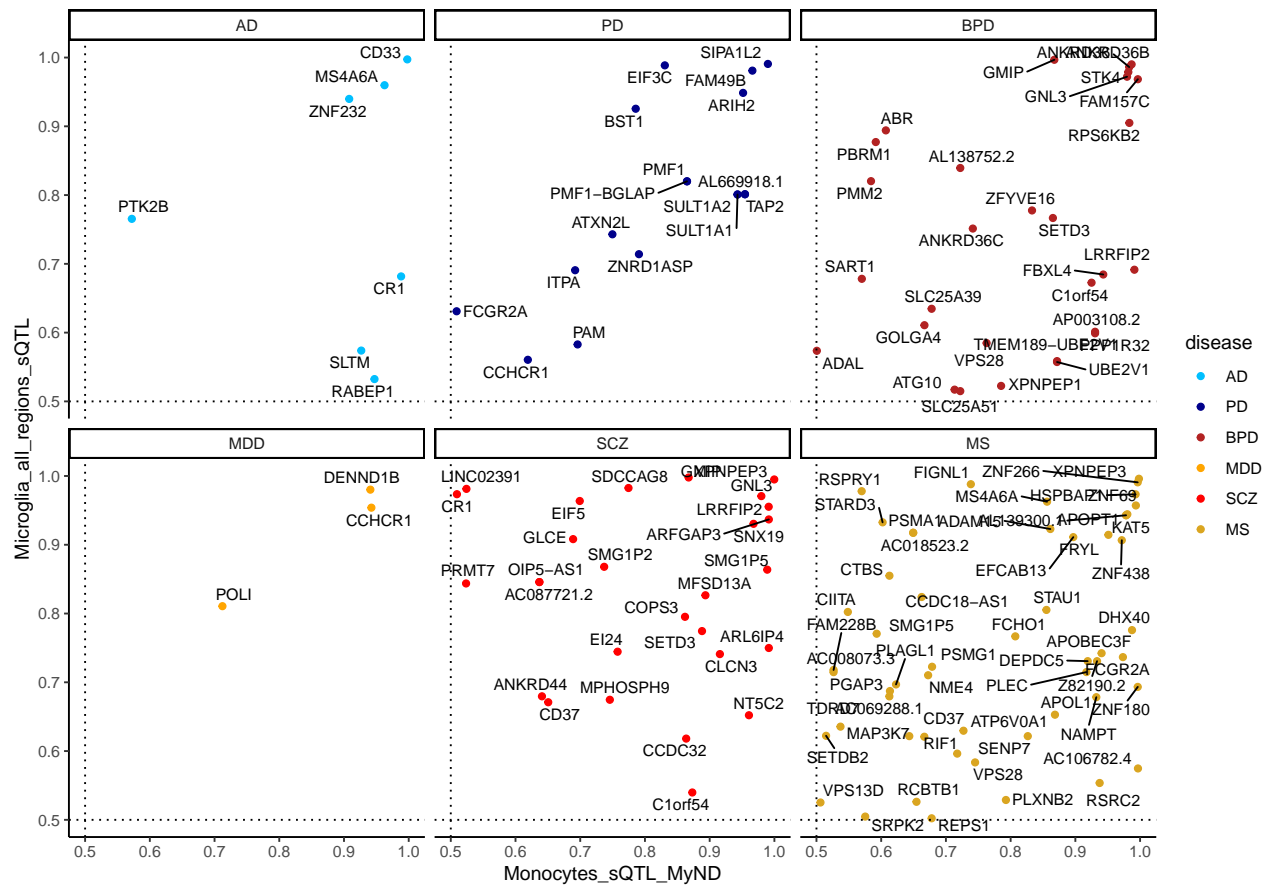
disease	QTL	n_loci	h4_0.5	h4_0.8	h4_0.9
AD	Monocytes_sQTL_MyND	41	9	2	7
AD	Monocytes_MyND	41	5	3	4
AD	Microglia_Young	41	3	3	3
AD	Microglia_all_regions_sQTL	41	8	3	5
AD	Microglia_all_regions	41	13	1	4
PD	DLPFC_ROSMAP	97	11	5	14
PD	Monocytes_Fairfax	97	11	3	11
PD	Monocytes_sQTL_MyND	97	12	2	15
PD	Monocytes_MyND	97	9	5	8
PD	Microglia_Young	97	12	2	0
PD	Microglia_all_regions_sQTL	97	24	7	8
PD	Microglia_all_regions	97	7	4	4
BPD	DLPFC_ROSMAP	31	4	2	5
BPD	Monocytes_Fairfax	31	6	1	2
BPD	Monocytes_sQTL_MyND	31	4	6	11
BPD	Monocytes_MyND	31	3	0	3
BPD	Microglia_Young	31	7	1	0
BPD	Microglia_all_regions_sQTL	31	9	2	10
BPD	Microglia_all_regions	31	3	2	0
MDD	DLPFC_ROSMAP	37	6	0	3
MDD	Monocytes_Fairfax	37	0	0	2
MDD	Monocytes_sQTL_MyND	37	4	2	2
MDD	Monocytes_MyND	37	2	1	2
MDD	Microglia_Young	37	0	0	0
MDD	Microglia_all_regions_sQTL	37	5	1	2
MDD	Microglia_all_regions	37	3	2	0
SCZ	DLPFC_ROSMAP	125	19	4	14
SCZ	Monocytes_Fairfax	125	16	5	10
SCZ	Monocytes_sQTL_MyND	125	30	11	22
SCZ	Monocytes_MyND	125	10	5	6
SCZ	Microglia_Young	125	17	3	1
SCZ	Microglia_all_regions_sQTL	125	34	13	32
SCZ	Microglia_all_regions	125	12	3	5
MS	DLPFC_ROSMAP	157	20	8	12
MS	Monocytes_Fairfax	157	38	5	10
MS	Monocytes_sQTL_MyND	157	56	16	41
MS	Monocytes_MyND	157	23	4	4
MS	Microglia_Young	157	15	6	5
MS	Microglia_all_regions_sQTL	157	54	25	40
MS	Microglia_all_regions	157	26	7	4

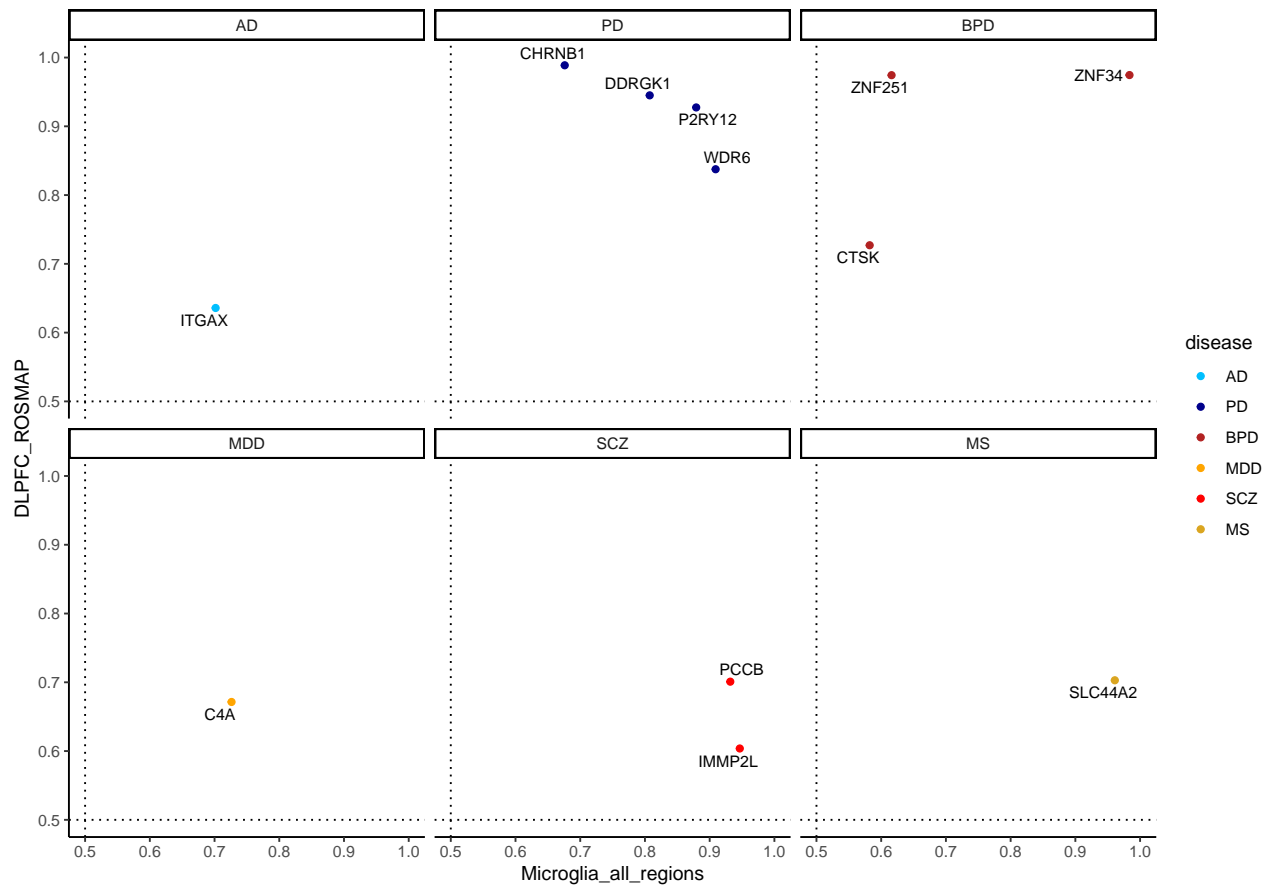
Sharing of colocalised genes

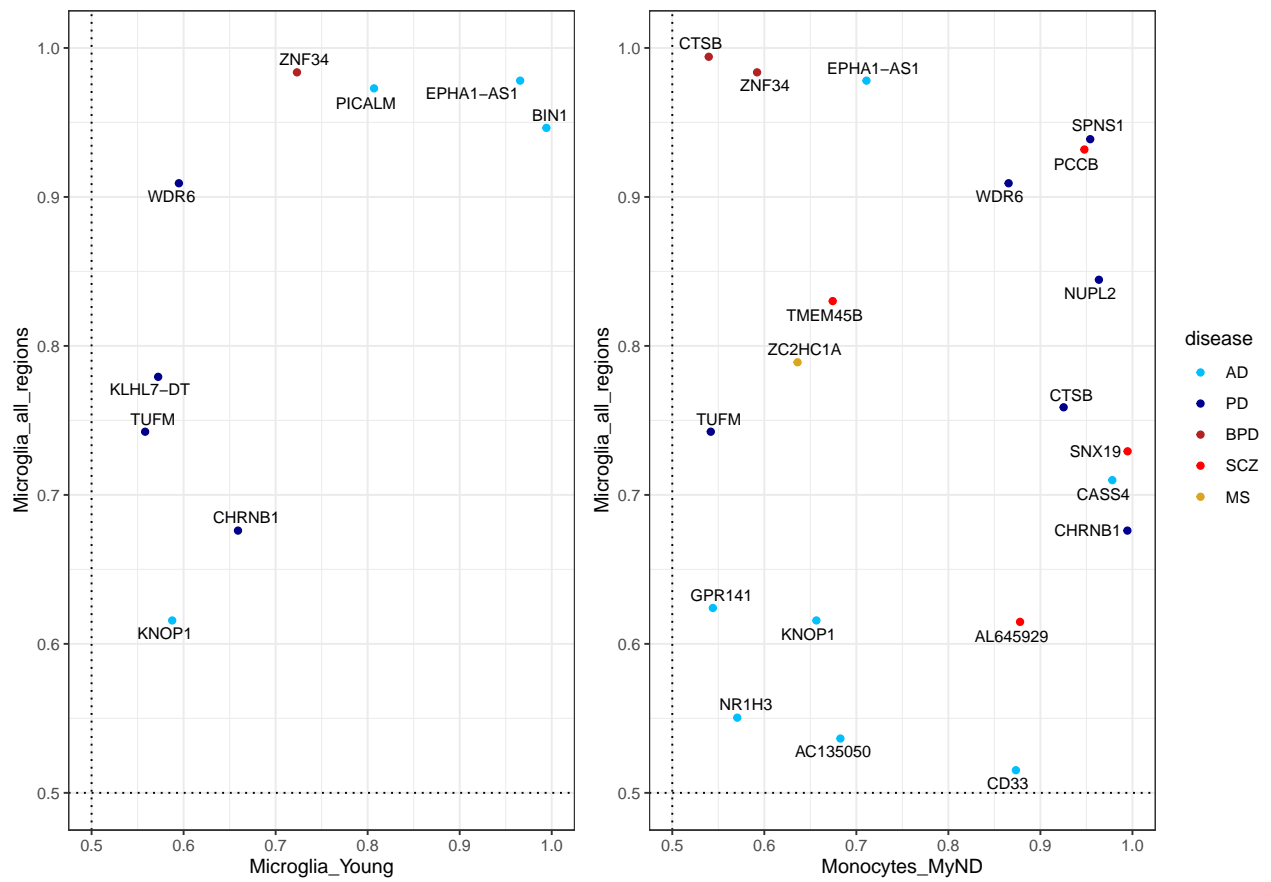




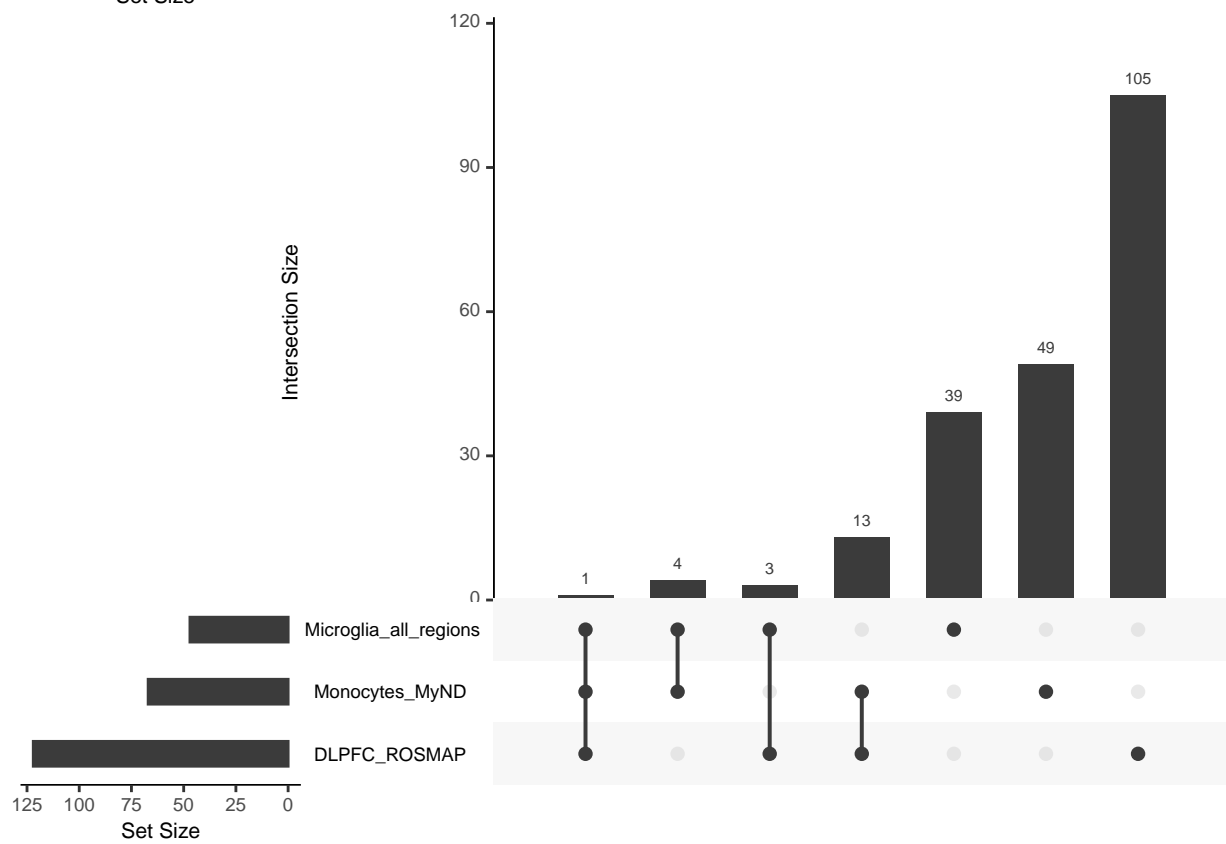
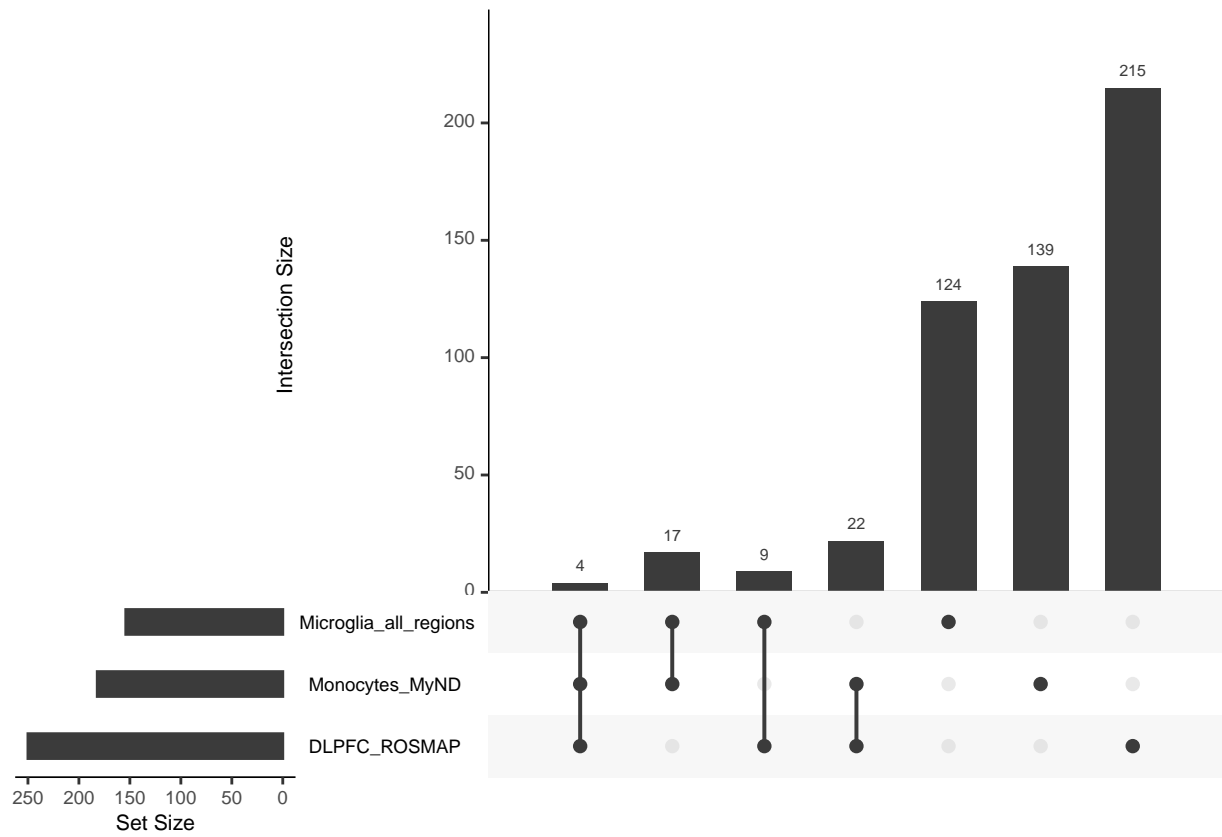


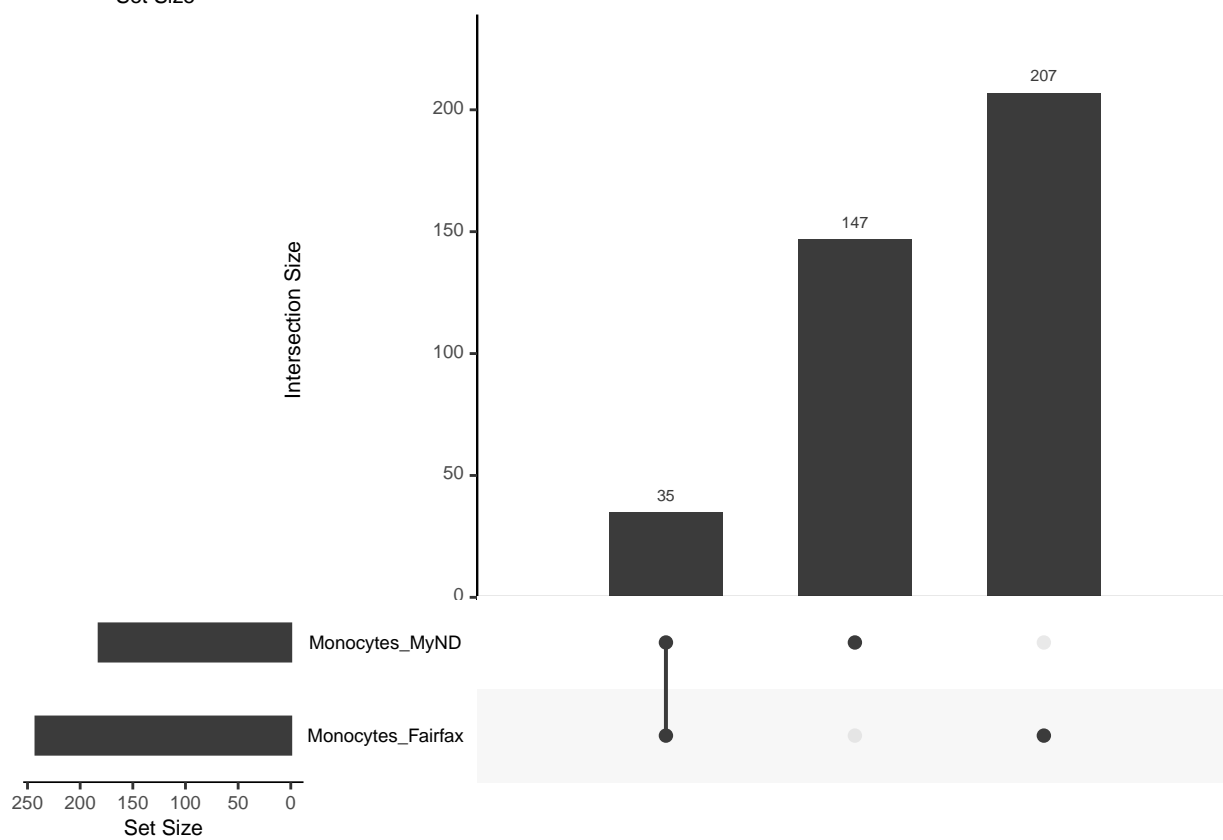
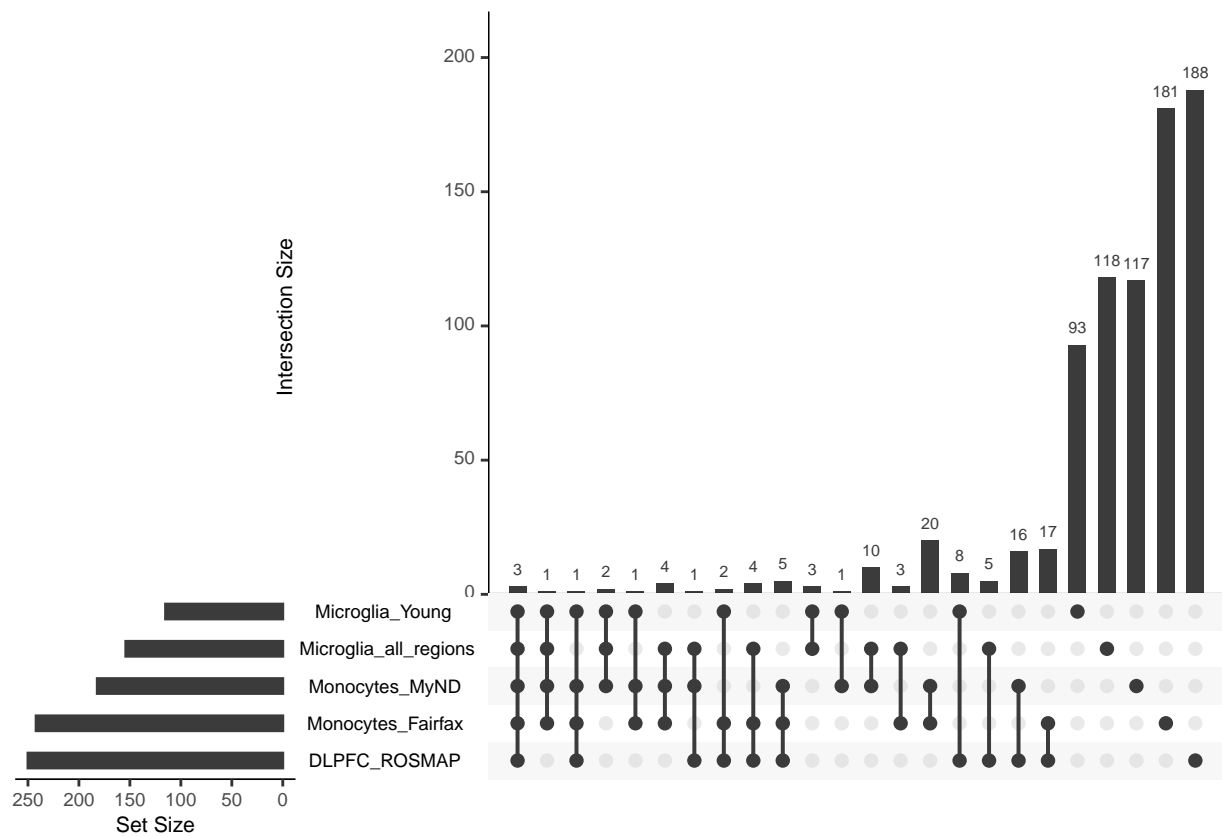


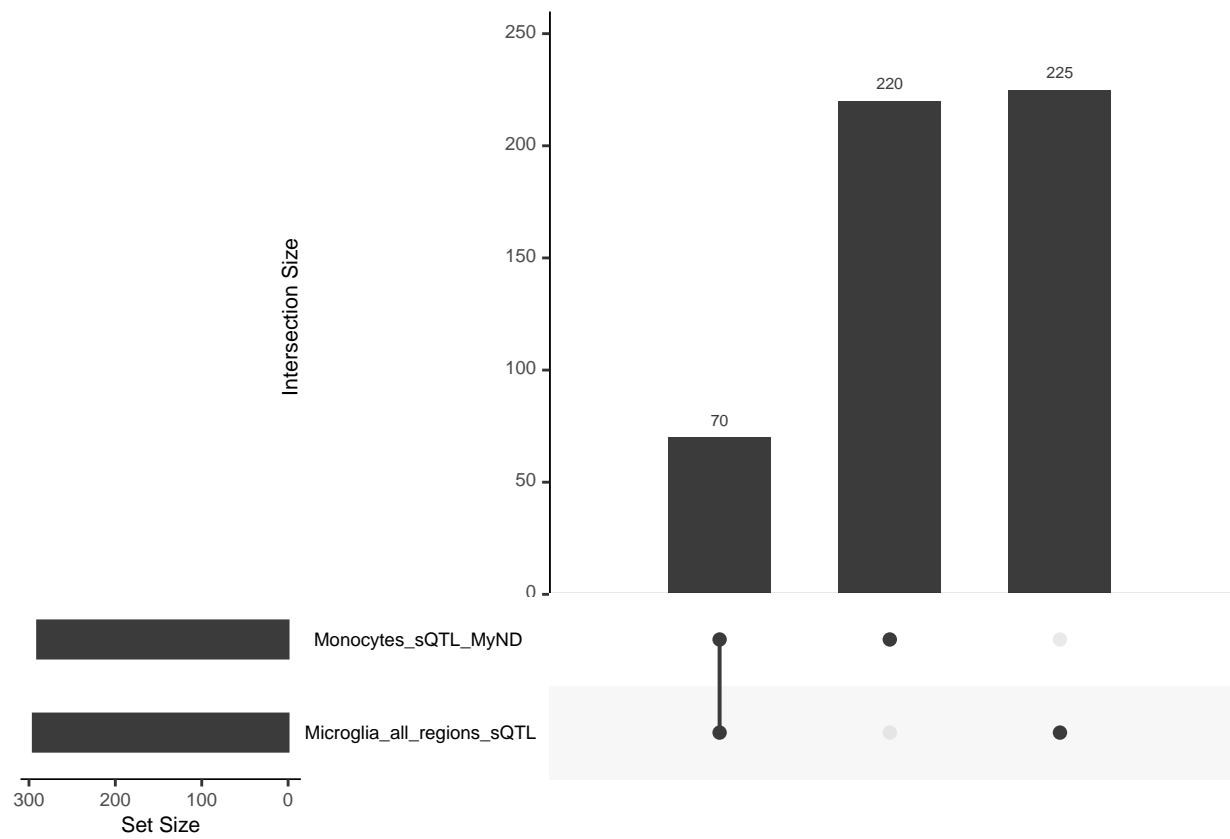




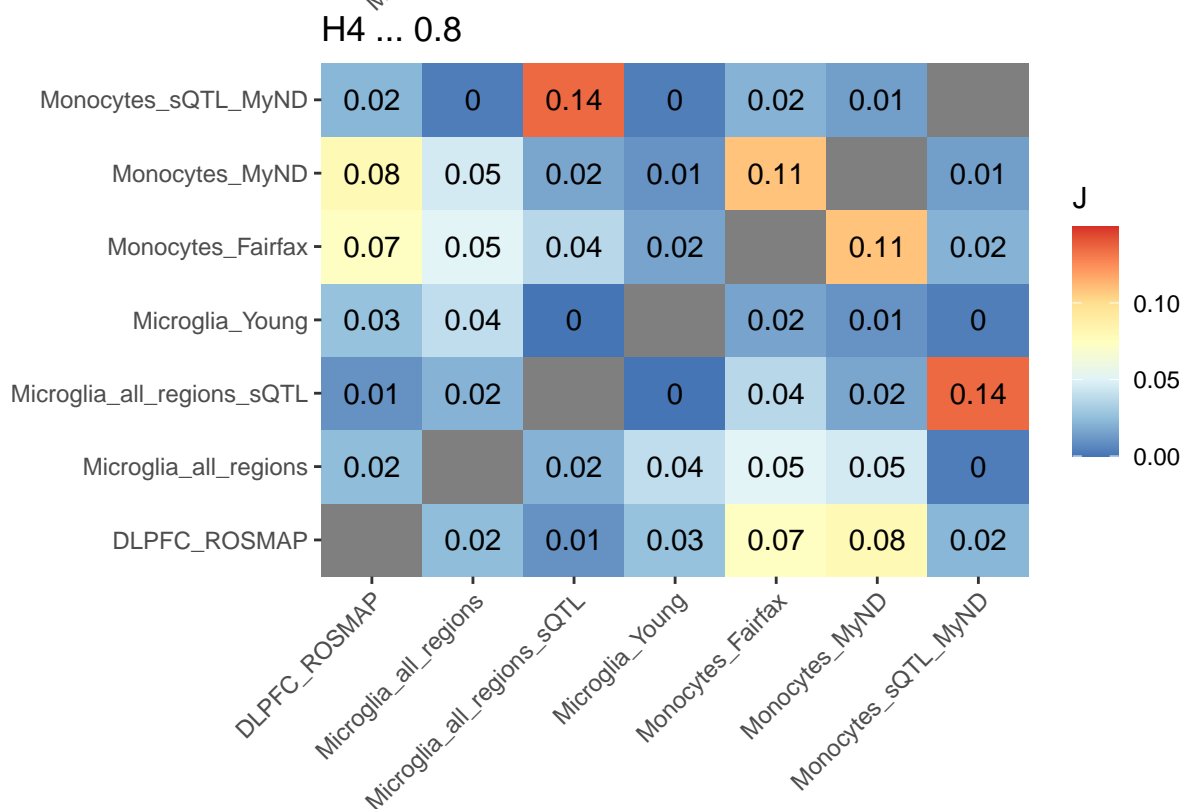
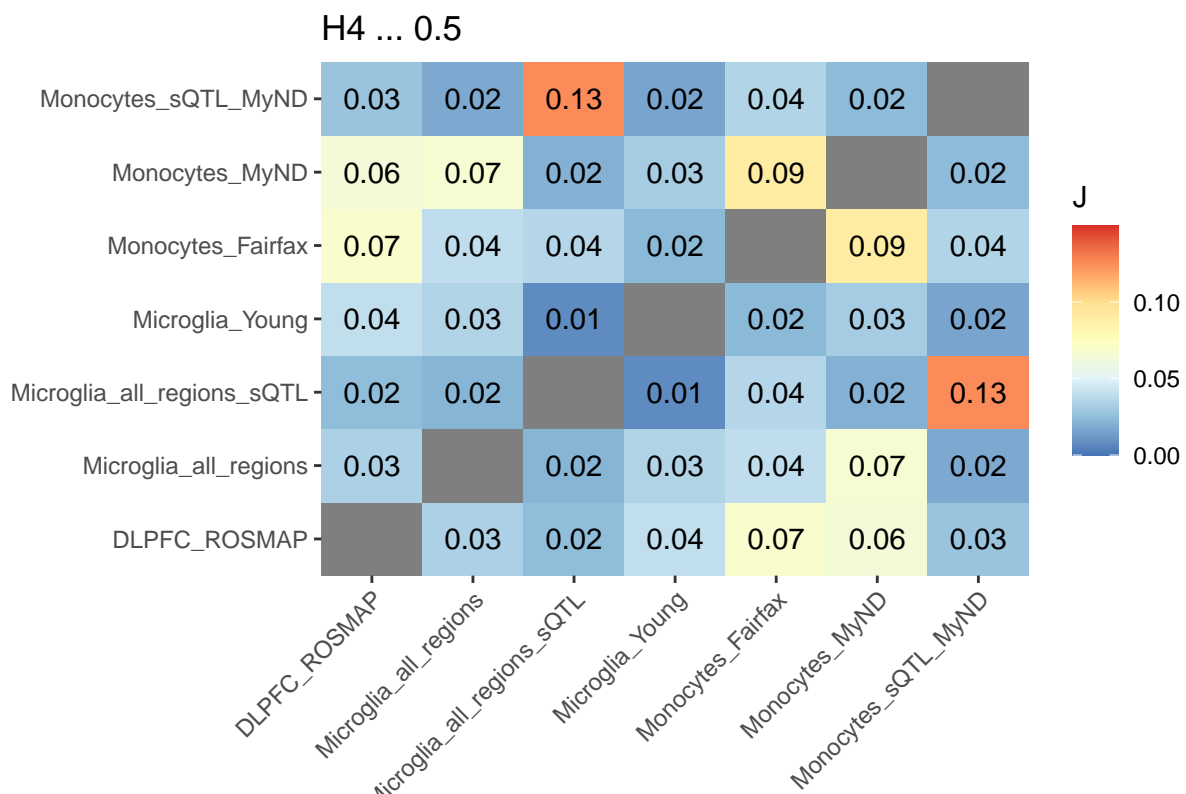
Upset plots of Colocalised QTLs (PP4 > 0.5) between Monocytes, Microglia and DLPFC





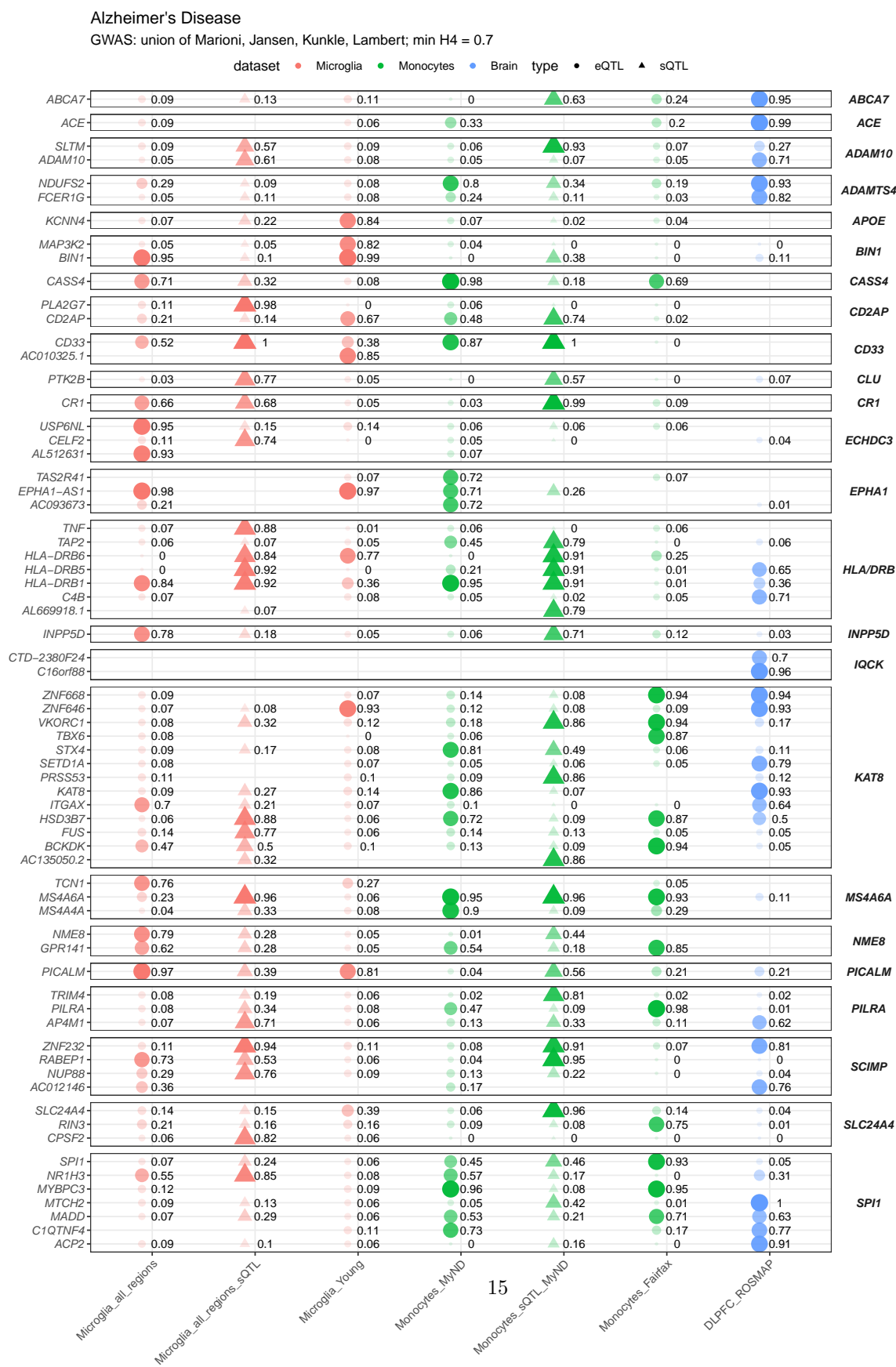


Jaccard distances between each QTL set - take the intersection of loci at a threshold divided by the union.



Compare eQTL effect sizes between Young and our Microglia

Alzheimer's Disease - all loci (max H4 per QTL across 4 GWAS)

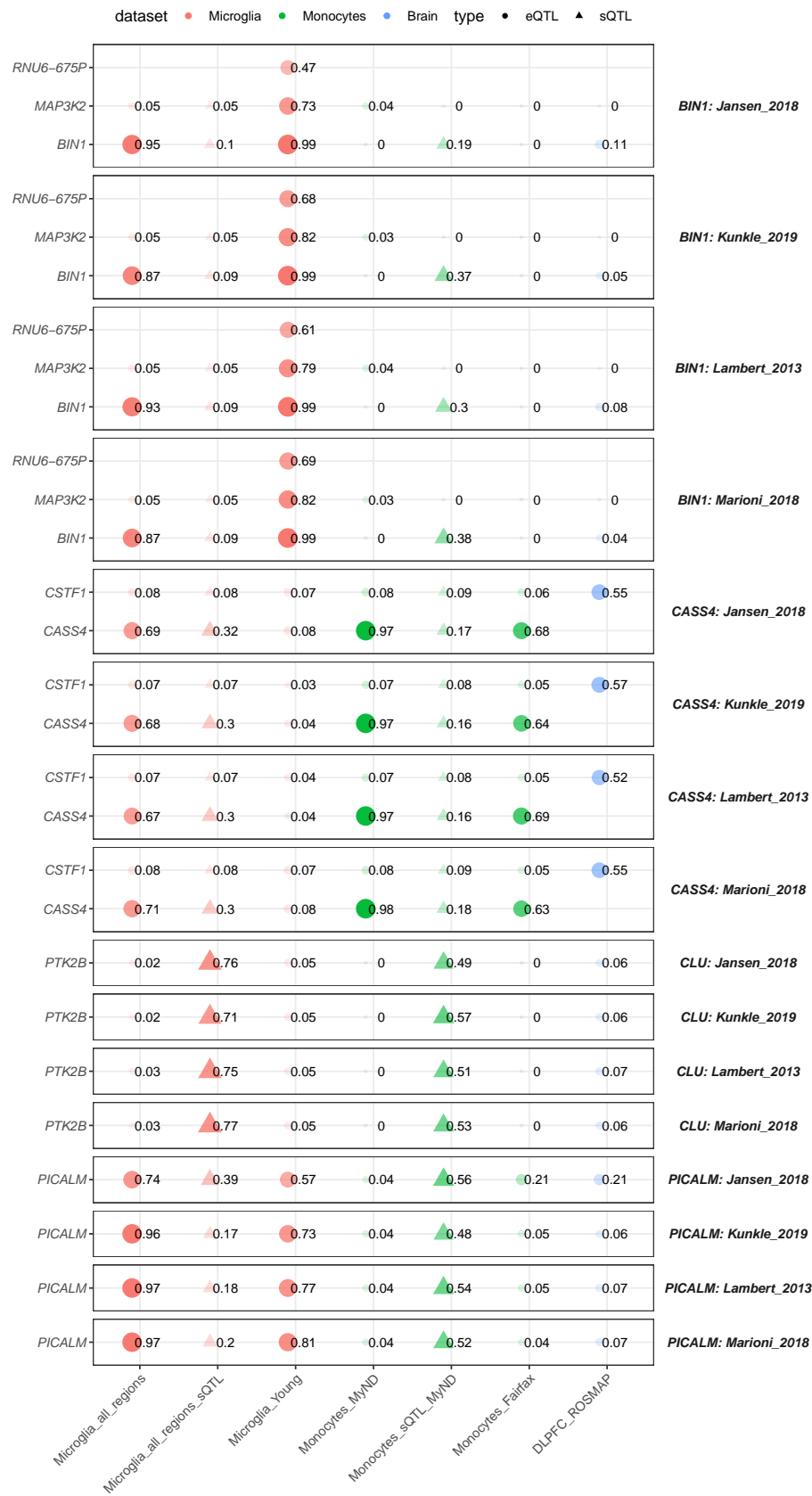


AD - Compare loci between the 4 GWAS

Plot some well known AD loci shared between GWAS - do the same Colocalisations occur?

Alzheimer's Disease

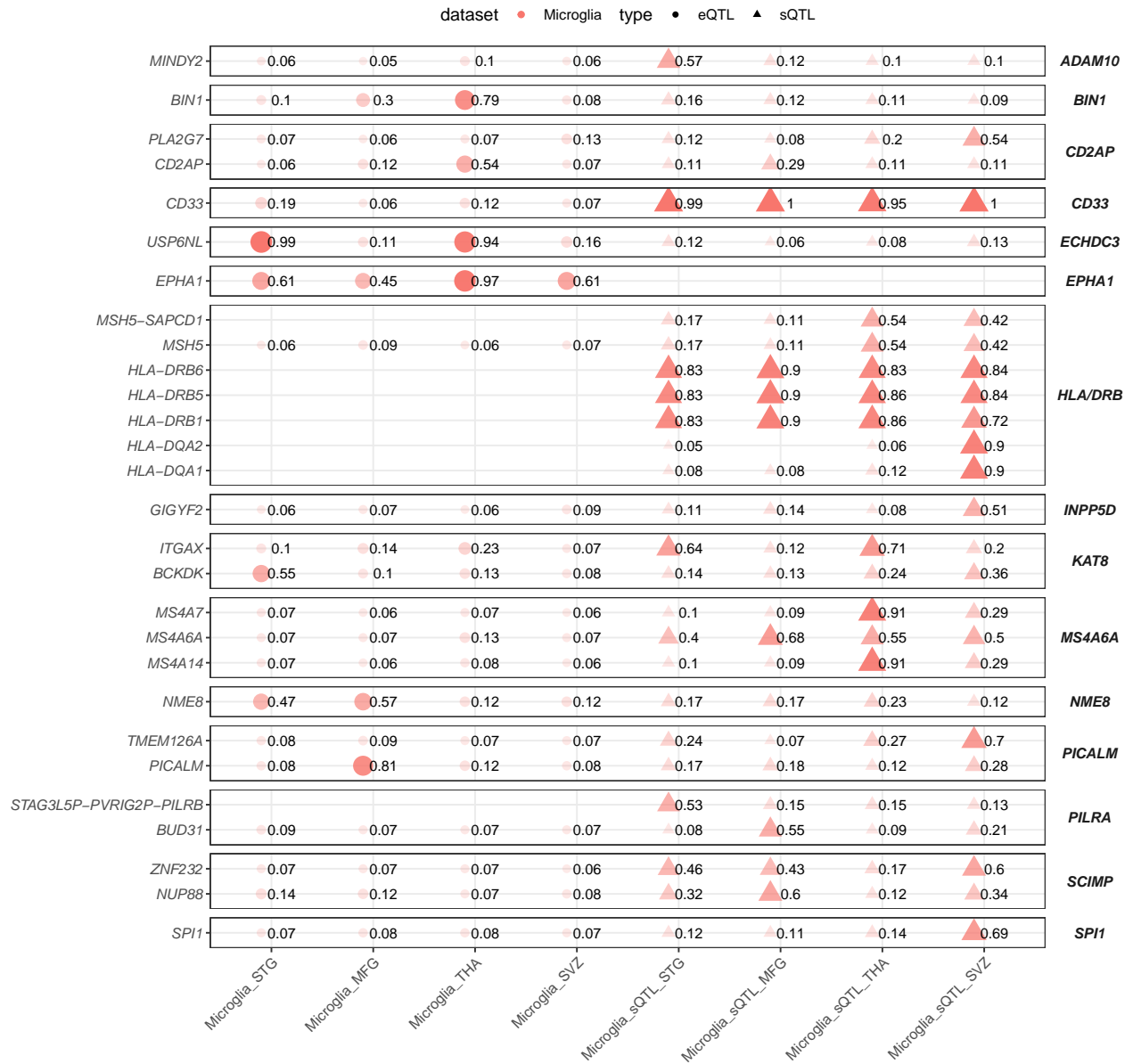
GWAS: union of Marioni, Jansen, Kunkle, Lambert; min H4 = 0.5



Microglia regions

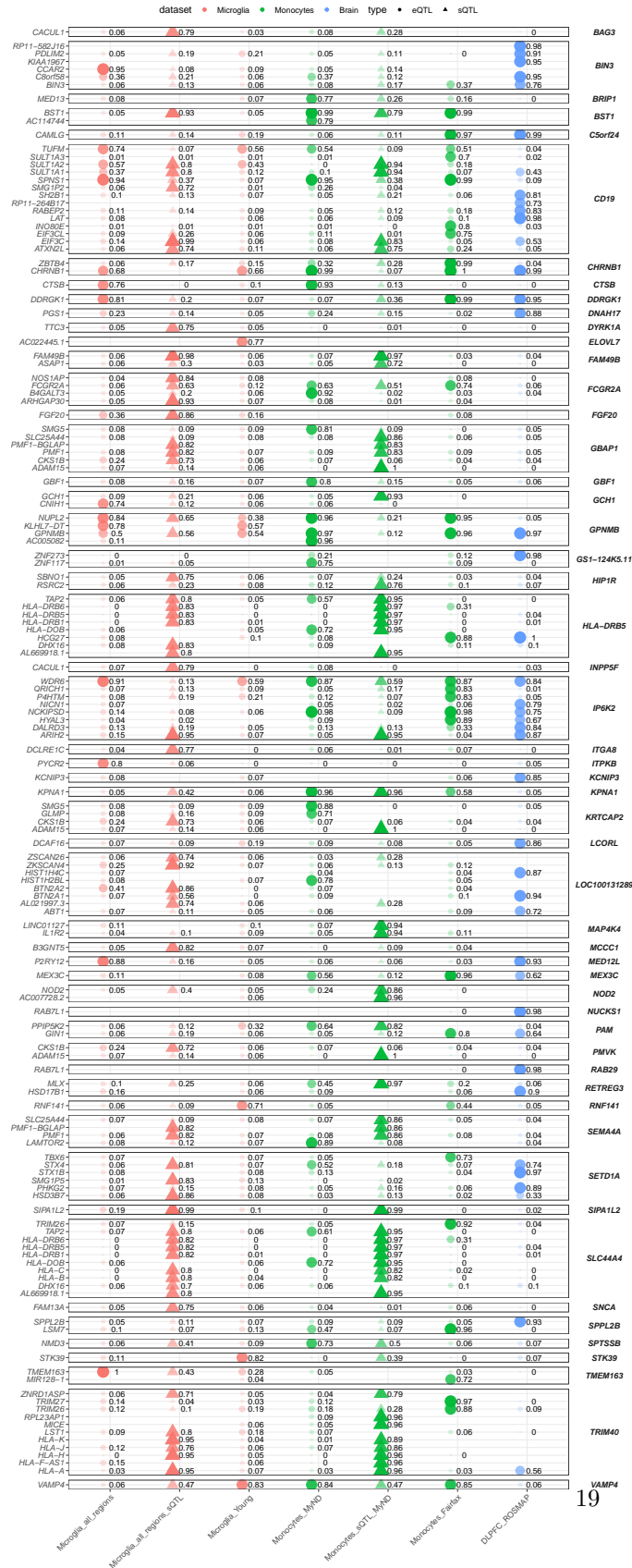
Alzheimer's Disease

GWAS: union of Marioni, Jansen, Kunkle, Lambert; min H4 = 0.5



Microglia, Monocytes and DLPFC

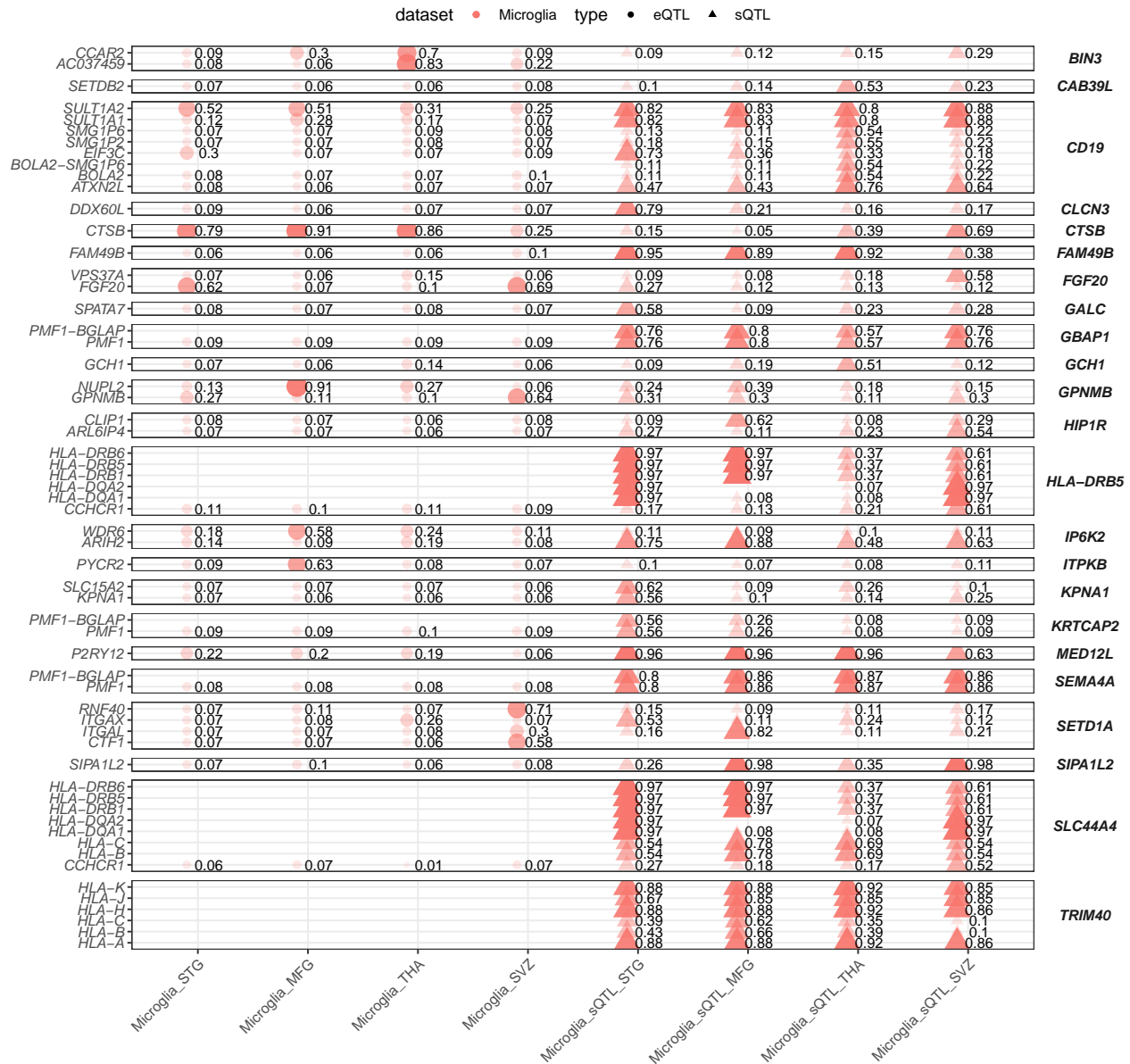
Parkinson's Disease
GWAS: Nalls et al 2019; min H4 = 0.7



Microglia regions

Parkinson's Disease

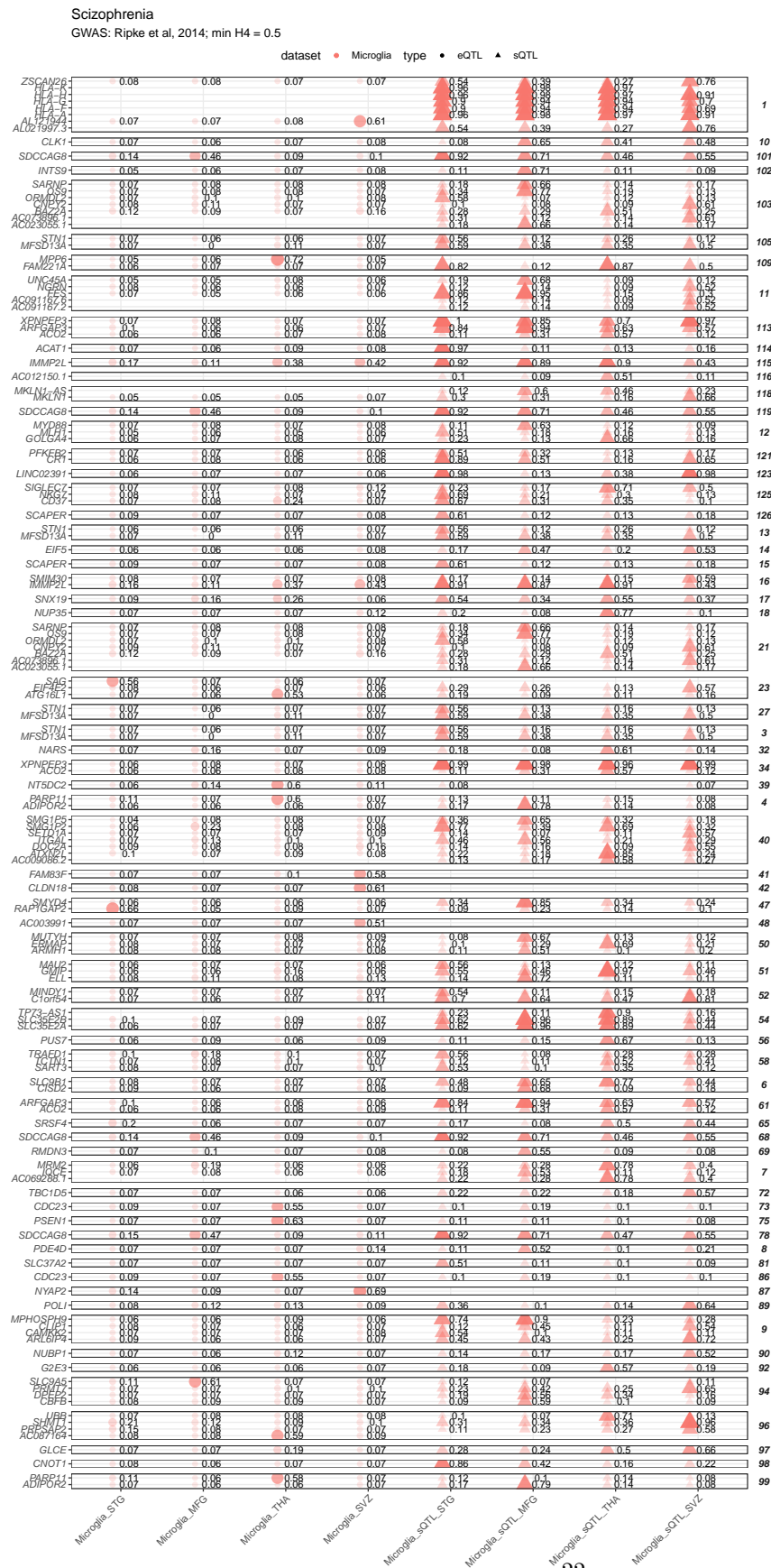
GWAS: Nalls et al 2019; min H4 = 0.5



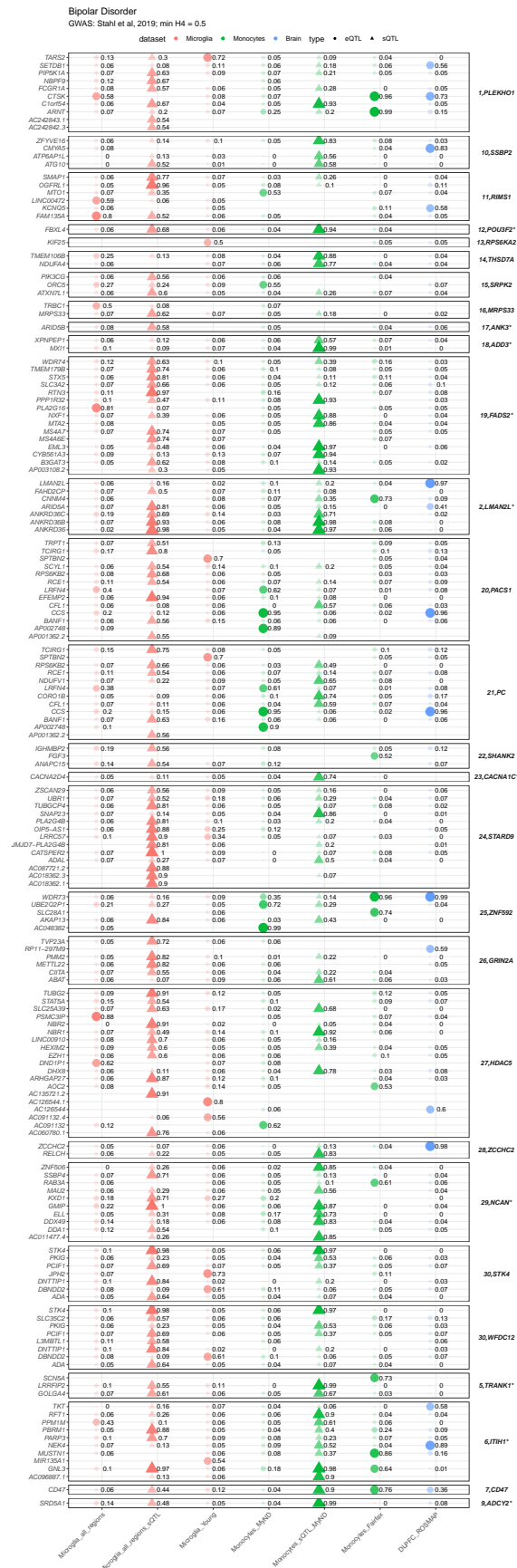
Microglia, Monocytes and DLPFC



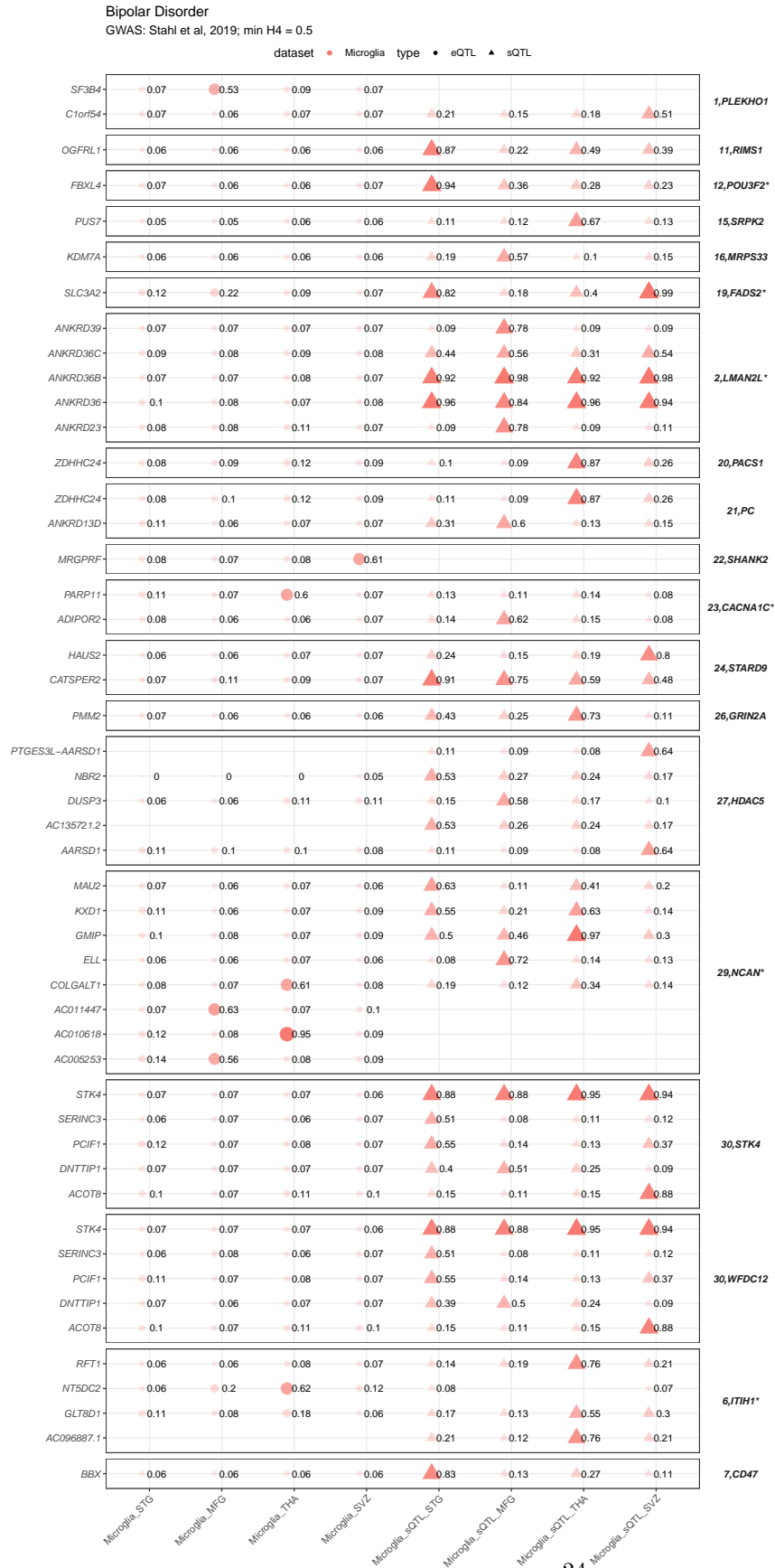
Microglia regions



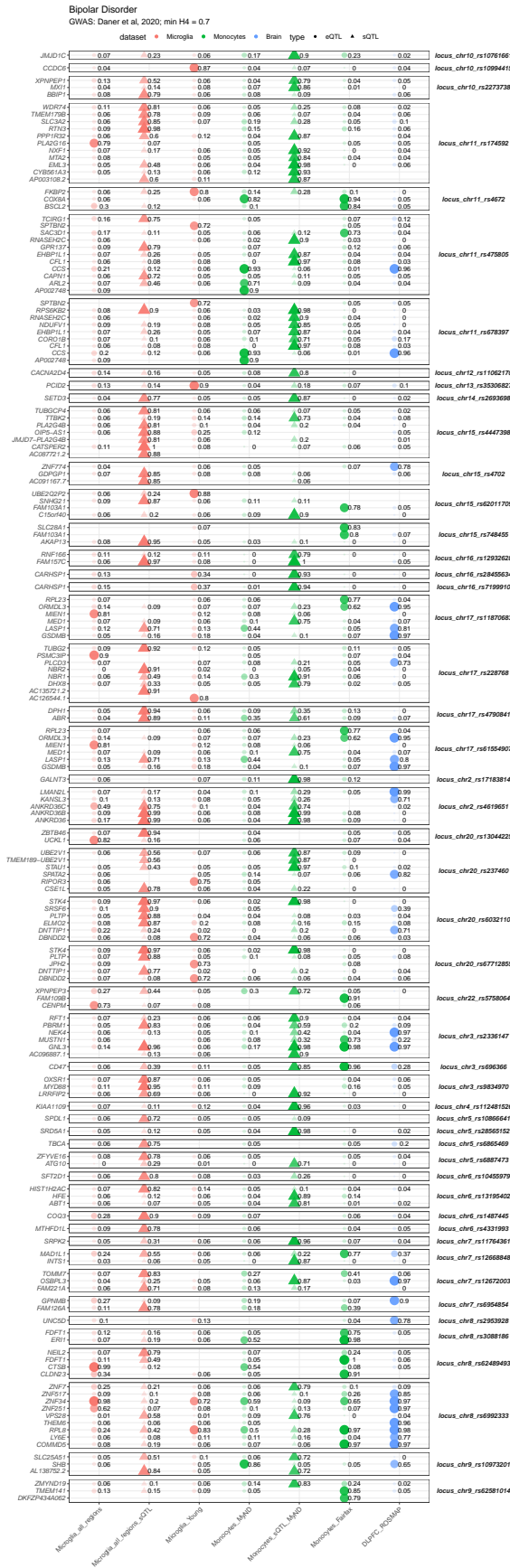
Microglia, Monocytes and DLPFC



Microglia regions



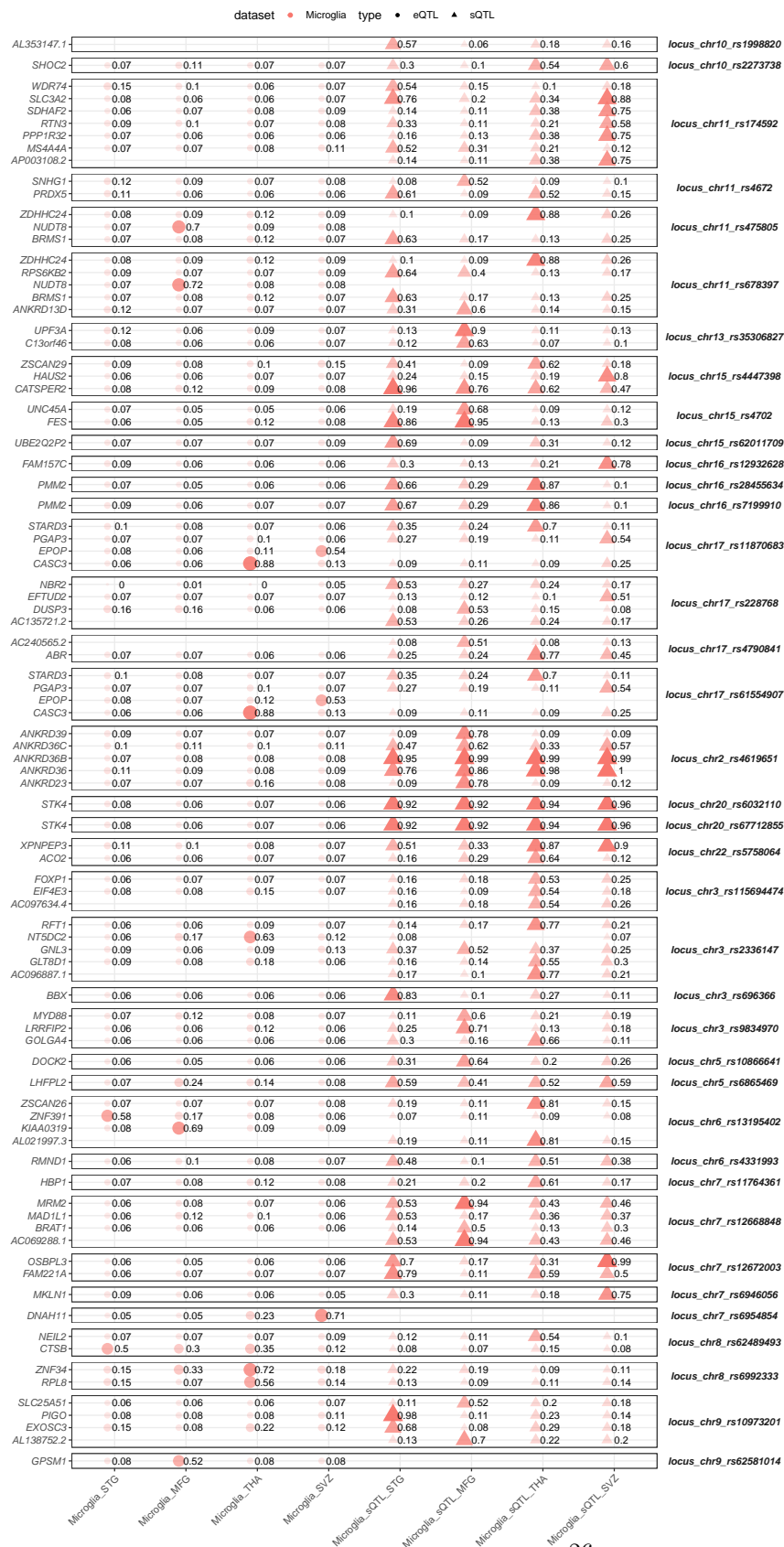
Bipolar 2 - Daner 2020



Microglia regions

Bipolar Disorder

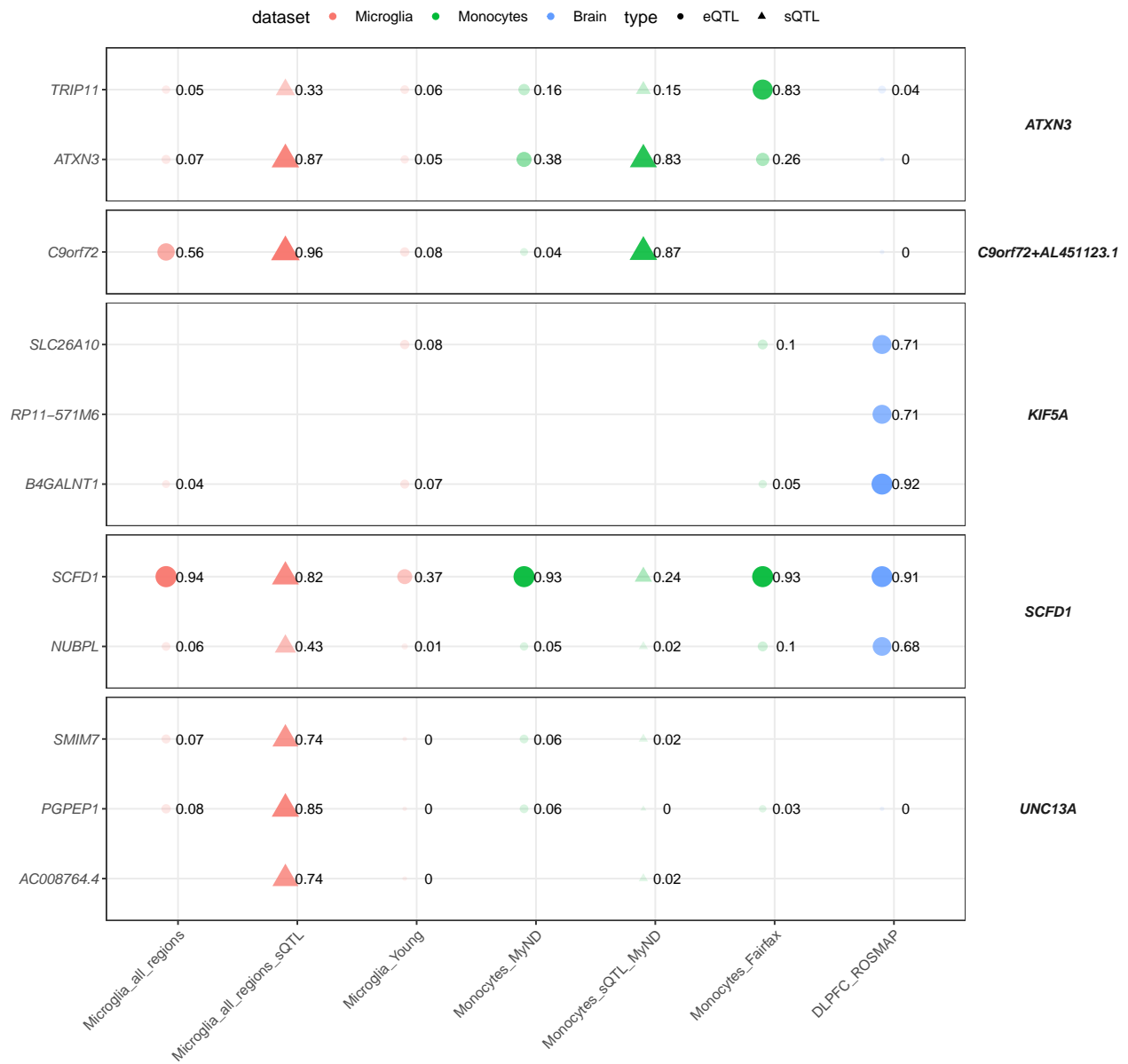
GWAS: Daner et al, 2020; min H4 = 0.5



ALS

Amyotrophic Lateral Sclerosis

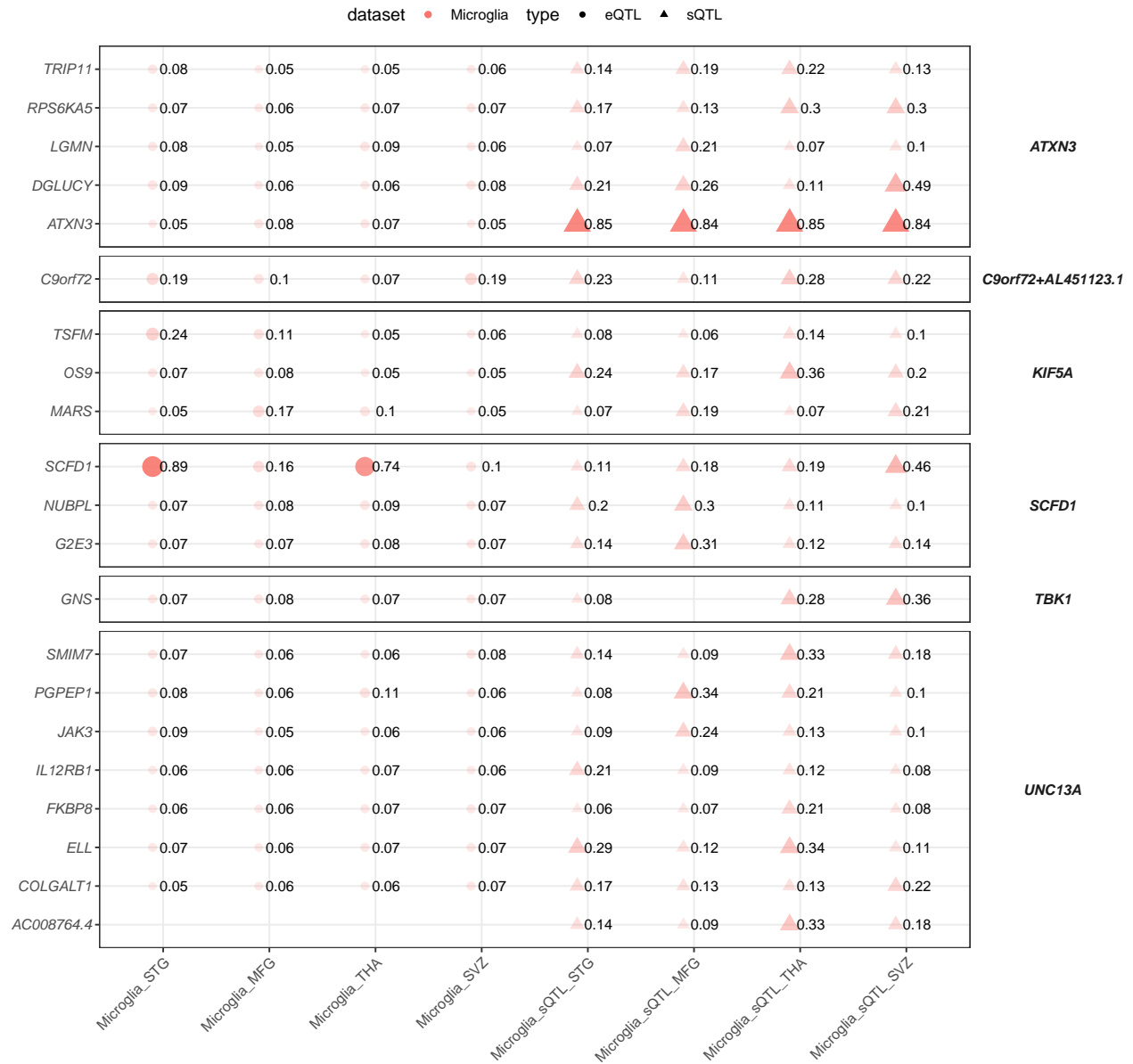
GWAS: Nicolas et al 2019; min H4 = 0.5



Microglia regions

Amyotrophic Lateral Sclerosis

GWAS: Nicolas et al 2019; min H4 = 0.2



Multiple Sclerosis

Multiple Sclerosis (MS)

Global MS Data - 2015-2016

Region

Country

Prevalence (per 100,000)

Incidence (per 100,000)

Age-standardized Prevalence (per 100,000)

Age-standardized Incidence (per 100,000)

Gender Ratio (M:F)

Age Ratio (M:F)

Health Status (0-100)

Notes

North America

USA

12.5

0.8

12.5

0.8

1.0

1.0

95

Canada

Mexico

10.2

0.7

10.2

0.7

1.0

1.0

92

Brazil

Argentina

8.9

0.6

8.9

0.6

1.0

1.0

88

Europe

Germany

11.8

0.7

11.8

0.7

1.0

1.0

93

France

UK

10.5

0.6

10.5

0.6

1.0

1.0

90

Italy

Spain

9.2

0.5

9.2

0.5

1.0

1.0

87

Asia

China

13.1

0.9

13.1

0.9

1.0

1.0

96

India

Japan

11.4

0.8

11.4

0.8

1.0

1.0

91

South Korea

Australia

9.5

0.6

9.5

0.6

1.0

1.0

89

South America

Brazil

7.1

0.5

7.1

0.5

1.0

1.0

85

Colombia

Venezuela

6.8

0.4

6.8

0.4

1.0

1.0

82

Peru

Chile

6.2

0.3

6.2

0.3

1.0

1.0

79

Africa

Egypt

10.3

0.7

10.3

0.7

1.0

1.0

90

Sudan

Ethiopia

9.1

0.6

9.1

0.6

1.0

1.0

87

Kenya

Tanzania

8.5

0.5

8.5

0.5

1.0

1.0

84

Oceania

Australia

7.8

0.4

7.8

0.4

1.0

1.0

81

New Zealand

South Africa

7.2

0.3

7.2

0.3

1.0

1.0

78

India

China

6.4

0.2

6.4

0.2

1.0

1.0

74

Middle East

Israel

8.8

0.5

8.8

0.5

1.0

1.0

83

Iran

Turkey

8.2

0.4

8.2

0.4

1.0

1.0

80

Saudi Arabia

UAE

7.6

0.3

7.6

0.3

1.0

1.0

77

Central America

Costa Rica

7.0

0.2

7.0

0.2

1.0

1.0

74

Panama

Nicaragua

6.5

0.4

6.5

0.4

1.0

1.0

79

Honduras

Guatemala

6.1

0.3

6.1

0.3

1.0

1.0

76

Caribbean

Jamaica

5.8

0.2

5.8

0.2

1.0

1.0

73

Trinidad and Tobago

Barbados

5.4

0.2

5.4

0.2

1.0

1.0

70

Suriname

Guyana

5.1

0.1

5.1

0.1

1.0

1.0

67

Eastern Europe

Poland

11.5

0.8

11.5

0.8

1.0

1.0

94

Iran

Turkey

10.8

0.7

10.8

0.7

1.0

1.0

91

Saudi Arabia

UAE

10.1

0.6

10.1

0.6

1.0

1.0

88

Southeast Asia

Thailand

9.4

0.5

9.4

0.5

1.0

1.0

85

Philippines

Malaysia

8.7

0.4

8.7

0.4

1.0

1.0

82

Central Asia

Uzbekistan

8.0

0.3

8.0

0.3

1.0

1.0

75

Kazakhstan

Georgia

7.3

0.2

7.3

0.2

1.0

1.0

68

South Asia

India

6.6

0.1

6.6

0.1

1.0

1.0

61

East Asia

China

5.9

0.1

5.9

0.1

1.0

1.0

54

South America

Brazil

5.2

0.1

5.2

0.1

1.0

1.0

47

Colombia

Venezuela

4.5

0.1

4.5

0.1

1.0

1.0

40

Peru

Chile

3.8

0.1

3.8

0.1

1.0

1.0

33

Africa

Egypt

3.1

0.1

3.1

0.1

1.0

1.0

26

Sudan

Ethiopia

2.4

0.1

2.4

0.1

1.0

1.0

19

Kenya

Tanzania

1.7

0.1

1.7

0.1

1.0

1.0

12

Oceania

Australia

1.0

0.1

1.0

0.1

1.0

1.0

5

New Zealand

South Africa

0.3

0.1

0.3

0.1

1.0

1.0

-2

India

China

-0.9

0.1

-0.9

0.1

1.0

1.0

-9

Middle East

Israel

-1.6

0.1

-1.6

0.1

1.0

1.0

-16

Iran

Turkey

-2.3

0.1

-2.3

0.1

1.0

1.0

-23

Saudi Arabia

UAE

-3.0

0.1

-3.0

0.1

1.0

1.0

-30

Central America

Costa Rica

-3.7

0.1

-3.7

0.1

1.0

1.0

-37

Panama

Nicaragua

-4.4

0.1

-4.4

0.1

1.0

1.0

-44

Honduras

Guatemala

-5.1

0.1

-5.1

0.1

1.0

1.0

-51

Caribbean

Jamaica

-5.8

0.1

-5.8

0.1

1.0

1.0

-58

Trinidad and Tobago

Barbados

-6.5

0.1

-6.5

0.1

1.0

1.0

-65

Suriname

Guyana

-7.2

0.1

-7.2

0.1

1.0

1.0

-72

Eastern Europe

Poland

-7.9

0.1

-7.9

0.1

1.0

1.0

-79

Iran

Turkey

-8.6

0.1

-8.6

0.1

1.0

1.0

-86

Saudi Arabia

UAE

-9.3

0.1

-9.3

0.1

1.0

1.0

-93

Southeast Asia

Thailand

-10.0

0.1

-10.0

0.1

1.0

1.0

-100

Philippines

Malaysia

-10.7

0.1

-10.7

0.1

1.0

1.0

-107

Central Asia

Uzbekistan

-11.4

0.1

-11.4

0.1

1.0

1.0

-114

Kazakhstan

Georgia

-12.1

0.1

-12.1

0.1

1.0

1.0

-121

South Asia

India

-12.8

0.1

-12.8

0.1

1.0

1.0

-128

East Asia

China

-13.5

0.1

-13.5

0.1

1.0

1.0

-135

South America

Brazil

-14.2

0.1

-14.2

0.1

1.0

1.0

-142

Colombia

Venezuela

-14.9

0.1

-14.9

0.1

1.0

1.0

-149

Peru

Chile

-15.6

0.1

-15.6

0.1

1.0

1.0

-156

Africa

Egypt

-16.3

0.1

-16.3

0.1

1.0

1.0

-163

Sudan

Ethiopia

-17.0

0.1

-17.0

0.1

1.0

1.0

-170

Kenya

Tanzania

-17.7

0.1

-17.7

0.1

1.0

1.0

-177

Oceania

Australia

-18.4

0.1

-18.4

0.1

1.0

1.0

-184

New Zealand

South Africa

-19.1

0.1

-19.1

0.1

1.0

1.0

-191

India

China

-19.8

0.1

-19.8

0.1

1.0

1.0

-198

Middle East

Israel

-20.5

0.1

-20.5

0.1

1.0

1.0

-205

Iran

Turkey

-21.2

0.1

-21.2

0.1

1.0

1.0

-212

Saudi Arabia

UAE

-21.9

0.1

-21.9

0.1

1.0

1.0

-219

Central America

Costa Rica

-22.6

0.1

-22.6

0.1

1.0

1.0

-226

Panama

Nicaragua

-23.3

0.1

-23.3

0.1

1.0

1.0

-233

Honduras

Guatemala

-24.0

0.1

-24.0

0.1

1.0

1.0

-240

Caribbean

Jamaica

-24.7

0.1

-24.7

0.1

1.0

1.0

-247

Trinidad and Tobago

Barbados

-25.4

0.1

-25.4

0.1

1.0

1.0

-254

Suriname

Guyana

-26.1

0.1

-26.1

0.1

1.0

1.0

-261

Eastern Europe

Poland

-26.8

0.1

-26.8

0.1

1.0

1.0

-268

Iran

Turkey

-27.5

0.1

-27.5

0.1

1.0

1.0

-275

Saudi Arabia

UAE

-28.2

0.1

-28.2

0.1

1.0

1.0

-282

Southeast Asia

Thailand

-28.9

0.1

-28.9

0.1

1.0

1.0

-289

Philippines

Malaysia

-29.6

0.1

-29.6

0.1

1.0

1.0

-296

Central Asia

Uzbekistan

-30.3

0.1

-30.3

0.1

1.0

1.0

-303

Kazakhstan

Georgia

-31.0

0.1

-31.0

0.1

1.0

1.0

-310

South Asia

India

-31.7

0.1

-31.7

0.1

1.0

1.0

-317

East Asia

China

-32.4

0.1

-32.4

0.1

1.0

1.0

-324

South America

Brazil

-33.1

0.1

-33.1

0.1

1.0

1.0

-331

Colombia

Venezuela

-33.8

0.1

-33.8

0.1

1.0

1.0

-338

Peru

Chile

-34.5

0.1

-34.5

0.1

1.0

1.0

-345

Africa

Egypt

-35.2

0.1

-35.2

0.1

1.0

1.0

-352

Sudan

Ethiopia

-35.9

0.1

-35.9

0.1

1.0

1.0

-359

Kenya

Tanzania

-36.6

0.1

-36.6

0.1

1.0

1.0

-366

Oceania

Australia

-37.3

0.1

-37.3

0.1

1.0

1.0

-373

New Zealand

South Africa

-38.0

0.1

-38.0

0.1

1.0

1.0

-380

India

China

-38.7

0.1

-38.7

0.1

1.0

1.0

-387

Middle East

Israel

-39.4

0.1

-39.4

0.1

1.0

1.0

-394

Iran

Turkey

-40.1

0.1

-40.1

0.1

1.0

1.0

-401

Saudi Arabia

UAE

-40.8

0.1

-40.8

0.1

1.0

1.0

-408

Central America

Costa Rica

-41.5

0.1

-41.5

0.1

1.0

1.0

-415

Panama

Nicaragua

-42.2

0.1

-42.2

0.1

1.0

1.0

-422

Honduras

Guatemala

-42.9

0.1

-42.9

0.1

1.0

1.0

-429

Caribbean

Jamaica

-43.6

0.1

-43.6

0.1

1.0

1.0

-436

Trinidad and Tobago

Barbados

-44.3

0.1

-44.3

0.1

1.0

1.0

-443

Suriname

Guyana

-45.0

0.1

-45.0

0.1

1.0

1.0

-450

Eastern Europe

Poland

-45.7

0.1

-45.7

0.1

1.0

1.0

-457

Iran

Turkey

-46.4

0.1

-46.4

0.1

1.0

1.0

-464

Saudi Arabia

UAE

-47.1

0.1

-47.1

0.1

1.0

1.0

-471

Southeast Asia

Thailand

-47.8

0.1

-47.8

0.1

1.0

1.0

-478

Philippines

Malaysia

-48.5

0.1

-48.5

0.1

1.0

1.0

-485

Central Asia

Uzbekistan

-49.2

0.1

-49.2

0.1

1.0

1.0

-492

Kazakhstan

Georgia

-49.9

0.1

-49.9

0.1

1.0

1.0

-499

South Asia

India

-50.6

0.1

-50.6

0.1

1.0

1.0

-506

East Asia

China

-51.3

0.1

-51.3

0.1

1.0

1.0

-513

South America

Brazil

-52.0

0.1

-52.0

0.1

1.0

1.0

-520

Colombia

Venezuela

-52.7

0.1

-52.7

0.1

1.0

1.0

-527

Peru

Chile

-53.4

0.1

-53.4

0.1

1.0

1.0

-534

Africa

Egypt

-54.1

0.1

-54.1

0.1

1.0

1.0

-541

Sudan

Ethiopia

-54.8

0.1

-54.8

0.1

1.0

1.0

-548

Kenya

Tanzania

-55.5

0.1

-55.5

0.1

1.0

1.0

-555

Oceania

Australia

-56.2

0.1

-56.2

0.1

1.0

1.0

-562

New Zealand

South Africa

-56.9

0.1

-56.9

0.1

1.0

1.0

-569

India

China

-57.6

0.1

-57.6

0.1

1.0

1.0

-576

Middle East

Israel

-58.3

0.1

-58.3

0.1

1.0

1.0

-583

Iran

Turkey

-59.0

0.1

-59.0

0.1

1.0

1.0

-590

Saudi Arabia

UAE

-59.7

0.1

-59.7

0.1

1.0

1.0

-597

Central America

Costa Rica

-60.4

0.1

-60.4

0.1

1.0

1.0

-604

Panama

Nicaragua

-61.1

0.1

-61.1

0.1

1.0

1.0

-611

Honduras

Guatemala

-61.8

0.1

-61.8

0.1

1.0

1.0

-618

Caribbean

Jamaica

-62.5

0.1

-62.5

0.1

1.0

1.0

-625

Trinidad and Tobago

Barbados

-63.2

0.1

-63.2

0.1

1.0

1.0

-632

Suriname

Guyana

-63.9

0.1

-63.9

0.1

1.0

1.0

-639

Eastern Europe

Poland

-64.6

0.1

-64.6

0.1

1.0

1.0

-646

Iran

Turkey

-65.3

0.1

-65.3

0.1

1.0

1.0

-653

Saudi Arabia

UAE

-66.0

0.1

-66.0

0.1

1.0

1.0

-660

Southeast Asia

Thailand

-66.7

0.1

-66.7

0.1

1.0

1.0

-667

Philippines

Malaysia

-67.4

0.1

-67.4

0.1

1.0

1.0

-674

Central Asia

Uzbekistan

-68.1

0.1

-68.1

0.1

1.0

1.0

-681

Kazakhstan

Georgia

-68.8

0.1

-68.8

0.1

1.0

1.0

-688

South Asia

India

-69.5

0.1

-69.5

0.1

1.0

1.0

-695

East Asia

China

-70.2

0.1

-70.2

0.1

1.0

1.0

-702

South America

Brazil

-70.9

0.1

-70.9

0.1

1.0

1.0

-709

Colombia

Venezuela

-71.6

0.1

-71.6

0.1

1.0

1.0

-716

Peru

Chile

-72.3

0.1

-72.3

0.1

1.0

1.0

-723

Africa

Egypt

-73.0

0.1

-73.0

0.1

1.0

1.0

-730

Sudan

Ethiopia

-73.7

0.1

-73.7

0.1

1.0

1.0

-737

Kenya

Tanzania

-74.4

0.1

-74.4

0.1

1.0

1.0

-744

Oceania

Australia

-75.1

0.1

-75.1

0.1

1.0

1.0

-751

New Zealand

South Africa

-75.8

0.1

-75.8

0.1

1.0

1.0

-758

India

China

-76.5

0.1

-76.5

0.1

1.0

1.0

-765

Middle East

Israel

-77.2

0.1

-77.2

0.1

1.0

1.0

-772

Iran

Turkey

-77.9

0.1

-77.9

0.1

1.0

1.0

-779

Saudi Arabia

UAE</

