main

October 18, 2021

1 Examine overlaps with published data

```
[1]: import pandas as pd import numpy as np
```

1.1 Public si-eQTL analysis

```
[2]: shen = ["GDAP2", "AIM2", "SLAMF6", "RLF", "ATG4C", "FUT7",
             "TMEM218", "C11orf74", "RAB35", "TMEM5", "HNRNPK",
             "CDCA3", "ERCC5", "GJB6", "SNTB2", "SPNS3",
             "XAF1", "RBBP8", "RUFY4", "CA2", "RAPGEF1"]
     print("Shen et al.:")
     print(len(shen))
     kukurba = ["NOD2", "WDR36", "BSCL2", "MAP7D3", "RHOXF1", "DNAH1"]
     print("Kukurba et al.:")
     print(len(kukurba))
     yao = ["NOD2", "HLA-DRB5", "HLA-DRB5", "KIAAO586", "PPP2R5A",
            "TSNAXIP1", "MUT", "GRIK2", "C15orf37", "LIMA1", "IL6ST",
            "HCG8", "BLOC1S3", "NKX3-1", "CXorf23"]
     print("Yao et al.:")
     print(len(np.unique(yao)))
     print("Total of Yao + Kukurba:")
     len(set(yao) | set(kukurba))
    Shen et al.:
    21
    Kukurba et al.:
    Yao et al.:
    Total of Yao + Kukurba:
[2]: 19
```

1.2 Load BrainSeq si-eQTL results

1.2.1 Interacting variant-gene pairs

```
[3]: bs0 = pd.read_csv("../../summary_table/_m/
     →BrainSeq_sexGenotypes_4features_3regions.txt.gz", sep='\t')
     bs0["ensembl_gene_id"] = bs0.gene_id.str.replace("\\..*", "", regex=True)
     biomart = pd.read_csv("../../residualized_expression/final_plot_eqtls/_h/
     ⇔biomart.csv", index_col=0)
     bs = bs0.merge(biomart, on="ensembl_gene_id").drop_duplicates(subset="gene_id")
     print(bs.shape)
     bs.tail(2)
    (974, 11)
[3]:
                 Tissue
                                    gene_id
                                                      gencodeID \
                         ENSG00000185386.14 ENSG00000185386.14
     15826
           Hippocampus
     15827
           Hippocampus
                          ENSG00000267251.2
                                              ENSG00000267251.2
                    variant_id segnames
                                                   Type ensembl_gene_id \
                                             lfsr
           chr22:50563003:G:C
                                  chr22 0.049299 Gene
                                                         ENSG00000185386
     15826
           chr18:80244356:T:C
                                  chr18 0.049375 Gene
                                                         ENSG00000267251
     15827
           external_gene_name entrezgene \
                                   5600.0
     15826
                       MAPK11
     15827
                   AC139100.1
                                      NaN
                                                  description
           mitogen-activated protein kinase 11 [Source: HG...
     15826
     15827
[4]: bs[(bs['external_gene_name'].isin(shen))].to_csv("siEQTL_Shen_comparison.csv",_
      →index=False)
[5]: bs[(bs['external_gene_name'].isin(kukurba))]
[5]: Empty DataFrame
     Columns: [Tissue, gene_id, gencodeID, variant_id, seqnames, lfsr, Type,
     ensembl_gene_id, external_gene_name, entrezgene, description]
     Index: []
[6]: bs[(bs['external_gene_name'].isin(yao))]
[6]: Empty DataFrame
     Columns: [Tissue, gene_id, gencodeID, variant_id, seqnames, lfsr, Type,
     ensembl_gene_id, external_gene_name, entrezgene, description]
     Index: []
[7]: bs[(bs['external_gene_name'].isin(shen+kukurba+yao))]
```

```
[7]:
            Tissue
                               gene_id
                                                 gencodeID
                                                                   variant_id \
      4670 Caudate ENSG00000125703.14 ENSG00000125703.14 chr1:63060301:G:A
                        lfsr Type ensembl_gene_id external_gene_name
           segnames
                                                                        entrezgene \
              chr1 0.049896 Gene ENSG00000125703
                                                                 ATG4C
                                                                           84938.0
      4670
                                                 description
      4670 autophagy related 4C cysteine peptidase [Sourc...
     1.2.2 eigenMT corrected p-values
 [8]: df0 = pd.read_csv("../../../summary_table/_m/
      →Brainseq sex interacting 4features 3regions.eFeatures.txt.gz", sep='\t')
      df = df0[(df0["Type"] == "Gene")].drop_duplicates(subset="gene_id").

→drop("Tissue", axis=1)
      df["ensembl_gene_id"] = df.gene_id.str.replace("\\..*", "", regex=True)
      dft = df.merge(biomart, on="ensembl_gene_id")
      print(dft.shape)
      dft.head(2)
     (5177, 14)
 [8]:
                variant_id
                                       gene_id
                                                         gencodeID
                                                                       slope \
         chr7:42936690:A:C ENSG00000002746.14 ENSG00000002746.14 0.436502
      1 chr17:48075934:C:T ENSG00000002919.14 ENSG00000002919.14 -0.536903
        statistic pval_nominal
                                           eigenMT_BH
                                                       TESTS
                                                              Type
                                             0.445692
                                                         445
                                                              Gene
      0 7.820971
                       0.000023 0.010399
      1 -7.959488
                       0.000127 0.037424
                                             0.549367
                                                         294 Gene
        ensembl_gene_id external_gene_name entrezgene \
      0 ENSG0000002746
                                     HECW1
                                               23072.0
      1 ENSG00000002919
                                     SNX11
                                               29916.0
                                              description
      O HECT, C2 and WW domain containing E3 ubiquitin...
      1 sorting nexin 11 [Source: HGNC Symbol; Acc: HGNC:...
 [9]: ## Not all genes significant via mashr are also significant with eigenMT
      len(set(bs.gene_id) & set(dft.gene_id))
 [9]: 568
[10]: dft[(dft['external_gene_name'].isin(shen))]
[10]:
                                                             gencodeID
                                                                           slope \
                    variant_id
                                           gene_id
                                                                        0.210967
      486
             chr1:63052156:A:G ENSG00000125703.14 ENSG00000125703.14
            chr16:68729086:G:C ENSG00000168807.16 ENSG00000168807.16 0.378104
      1029
```

```
2804
             chr11:36347842:A:G
                                 ENSG00000166352.15
                                                     ENSG00000166352.15 0.518875
      3861
              chr8:84969351:T:C
                                  ENSG00000104267.9
                                                      ENSG00000104267.9 -0.230131
      4091
              chr17:6627584:T:C ENSG00000132530.16 ENSG00000132530.16 -0.515449
                                               eigenMT_BH
                                                           TESTS
                                                                  Type \
            statistic pval_nominal
                                           BF
            10.344946 7.923880e-05 0.037955
                                                 0.551098
                                                              479
                                                                  Gene
      486
      1029 11.451744 1.090640e-04 0.033483
                                                 0.541553
                                                             307
                                                                  Gene
      2399 10.551991 8.335770e-07 0.000542
                                                             650 Gene
                                                 0.317735
      2804
             8.165337 1.671190e-05 0.009492
                                                             568
                                                                  Gene
                                                 0.539284
      3861 -11.113888 3.085070e-04 0.040723
                                                              132
                                                                  Gene
                                                 0.693507
      4091 -14.299219 5.135140e-05 0.044214
                                                 0.698000
                                                             861 Gene
            ensembl_gene_id external_gene_name
                                                entrezgene
      486
            ENSG00000125703
                                         ATG4C
                                                   84938.0
      1029 ENSG00000168807
                                         SNTB2
                                                    6645.0
      2399 ENSG00000121742
                                          GJB6
                                                   10804.0
      2804 ENSG00000166352
                                      C11orf74
                                                  119710.0
      3861 ENSG00000104267
                                           CA2
                                                     760.0
      4091 ENSG00000132530
                                          XAF1
                                                   54739.0
                                                  description
      486
            autophagy related 4C cysteine peptidase [Sourc...
      1029 syntrophin beta 2 [Source: HGNC Symbol; Acc: HGNC...
            gap junction protein beta 6 [Source: HGNC Symbo...
      2804 chromosome 11 open reading frame 74 [Source: HG...
           carbonic anhydrase 2 [Source: HGNC Symbol; Acc: H...
      3861
      4091
           XIAP associated factor 1 [Source: HGNC Symbol; A...
[11]: dft[(dft['external_gene_name'].isin(shen)) & (dft["eigenMT_BH"] < 0.25)]
[11]: Empty DataFrame
      Columns: [variant_id, gene_id, gencodeID, slope, statistic, pval_nominal, BF,
      eigenMT_BH, TESTS, Type, ensembl_gene_id, external_gene_name, entrezgene,
      description]
      Index: []
[12]: dft[(dft['external_gene_name'].isin(kukurba)) & (dft["eigenMT_BH"] < 0.25)]
[12]: Empty DataFrame
      Columns: [variant_id, gene_id, gencodeID, slope, statistic, pval_nominal, BF,
      eigenMT_BH, TESTS, Type, ensembl_gene_id, external_gene_name, entrezgene,
      description]
      Index: []
[13]: dft[(dft['external_gene_name'].isin(yao)) & (dft["eigenMT_BH"] < 0.25)]
```

ENSG00000121742.16

ENSG00000121742.16 0.593176

2399

chr13:20258404:TC:T

```
[13]: Empty DataFrame
      Columns: [variant_id, gene_id, gencodeID, slope, statistic, pval_nominal, BF,
      eigenMT_BH, TESTS, Type, ensembl_gene_id, external_gene_name, entrezgene,
      description]
      Index: []
     1.3 GTEx comparison
[14]: gtex = pd.read_csv("/ceph/users/jbenja13/projects/sex_sz_ria/input/gtex_v8/"+\
                         "_m/GTEx_Analysis_v8_sbeQTLs/GTEx_Analysis_v8_sbeQTLs.txt",
                         sep='\t')
      gtex.iloc[0:2, 0:10]
「14]:
           ensembl_gene_id
                            hugo_gene_id
                                                        gene_type \
      0 ENSG00000241860.6 RP11-34P13.13
                                             processed_transcript
      1 ENSG00000227232.5
                                           unprocessed_pseudogene
                                   WASH7P
                 variant id
                                   rs_id
                                                        Tissue
                                                                     maf
      0 chr1_14677_G_A_b38 rs201327123
                                          Adipose_Subcutaneous
                                                                0.051635
      1 chr1_64764_C_T_b38 rs769952832
                                         Adipose_Subcutaneous
                                                                0.061102
        pval_nominal_sb slope_sb
                                    slope se sb
      0
                0.847114 0.055080
                                       0.285537
                0.316881 0.222928
                                       0.222511
      1
[15]: gtex.iloc[0:2, 10:14]
[15]:
        numtested pvals.corrected
                                               pval_nominal_f
                                         qval
                           0.847114 1.000000
                                                     0.022302
                 1
      1
                           0.316881
                                     0.981254
                                                     0.003978
[16]: ## qual threshold equal to number of published sb-eQTL
      gtex[(gtex['qval'] < 0.25) & (gtex["Tissue"].str.contains("Brain"))]\</pre>
          .loc[:, ["ensembl_gene_id", "hugo_gene_id", "Tissue", "pvals.corrected", __
       \rightarrow 'qval']].head(10)
Г16]:
                 ensembl_gene_id hugo_gene_id \
      62155
              ENSG00000026025.15
                                           MIV
      116842 ENSG00000160818.16
                                       GPATCH4
      121904 ENSG00000141562.17
                                          NARF
      122123
             ENSG00000267174.5 CTC-510F12.4
                                             Tissue pvals.corrected
                                                                          qval
      62155
                                     Brain_Amygdala
                                                                      0.012836
                                                            0.000004
      116842 Brain_Nucleus_accumbens_basal_ganglia
                                                            0.000088
                                                                      0.198445
      121904 Brain_Nucleus_accumbens_basal_ganglia
                                                            0.000056
                                                                      0.198445
              Brain_Nucleus_accumbens_basal_ganglia
      122123
                                                            0.000083
                                                                      0.198445
```

```
[17]: ## qual threshold equal to number of published sb-eQTL
      gtex[(gtex['qval'] < 0.25) & (gtex["Tissue"].str.contains("Whole"))]\</pre>
          .loc[:, ["ensembl_gene_id", "hugo_gene_id", "Tissue", "pvals.corrected", __
       \rightarrow 'qval']].head(10)
[17]:
                 ensembl_gene_id hugo_gene_id
                                                      Tissue
                                                             pvals.corrected \
               ENSG00000221571.3 RNU6ATAC35P
      362961
                                                 Whole Blood
                                                                      0.000039
               ENSG00000196743.8
                                                 Whole Blood
      365043
                                          GM2A
                                                                      0.000011
      367164
              ENSG00000148459.15
                                         PDSS1
                                                 Whole_Blood
                                                                      0.000027
                  qval
      362961
              0.139762
              0.116825
      365043
      367164
              0.139762
[18]: gtex_sig = gtex[(gtex['qval'] < 0.25)]
      gtex_sig.shape
[18]: (369, 22)
[19]:
      gtex_sig.head(10)
[19]:
                ensembl_gene_id hugo_gene_id
                                                                          gene_type \
      1096
              ENSG00000076356.6
                                       PLXNA2
                                                                    protein_coding
      5262
             ENSG00000170632.13
                                       ARMC10
                                                                    protein_coding
      5644
             ENSG00000120907.17
                                       ADRA1A
                                                                    protein_coding
      6414
             ENSG00000136830.11
                                      FAM129B
                                                                    protein_coding
      7220
              ENSG00000166787.3
                                                transcribed_unprocessed_pseudogene
                                        SAA3P
      8540
              ENSG00000183463.5
                                         URAD
                                                                    protein_coding
      9191
              ENSG00000282651.2
                                   IGHV5-10-1
                                                                          IG_V_gene
      14611
             ENSG00000143933.16
                                        CALM2
                                                                    protein_coding
      15082
              ENSG00000144410.4
                                          CPO
                                                                    protein_coding
      17452
                                        TRGV4
              ENSG00000211698.2
                                                                          TR_V_gene
                                                                        Tissue
                           variant_id
                                             rs_id
      1096
              chr1_208030492_G_A_b38
                                                         Adipose_Subcutaneous
                                         rs3811383
      5262
              chr7_103076937_C_T_b38
                                         rs6958836
                                                         Adipose_Subcutaneous
      5644
               chr8_26839198_G_A_b38
                                                         Adipose_Subcutaneous
                                       rs117380715
      6414
              chr9_127584339_G_A_b38
                                        rs10739693
                                                         Adipose_Subcutaneous
      7220
              chr11_18269355_T_C_b38
                                                         Adipose Subcutaneous
                                        rs34068567
      8540
              chr13_27990205_T_A_b38
                                         rs7335293
                                                         Adipose_Subcutaneous
      9191
             chr14_106114510_A_G_b38
                                         rs4573838
                                                         Adipose Subcutaneous
               chr2_46225349_C_T_b38
                                                     Adipose_Visceral_Omentum
      14611
                                        rs12477148
              chr2 206822186 C T b38
                                                     Adipose Visceral Omentum
      15082
                                        rs12470278
      17452
               chr7_38361995_A_C_b38
                                                     Adipose_Visceral_Omentum
                                        rs10233345
                  maf pval_nominal_sb slope_sb slope_se_sb ...
                                                                         qval \
```

```
1096
       0.123924
                    5.391600e-05 0.338278
                                               0.083064
                                                         ... 0.121068
5262
       0.169535
                    5.011130e-05 0.357403
                                               0.087384
                                                            0.192900
5644
       0.216867
                    1.045890e-05 -0.323552
                                               0.072676
                                                            0.084548
6414
       0.304647
                    7.387010e-07 -0.283660
                                               0.056579
                                                            0.004976
7220
       0.278830
                    2.207290e-05 0.323030
                                               0.075427 ... 0.074347
8540
       0.500000
                    9.078700e-09 -0.444892
                                               0.076123 ... 0.000122
9191
                                               0.094541 ...
       0.419105
                    2.025150e-05 -0.406760
                                                            0.074347
14611
      0.072495
                    4.497930e-05 -0.480557
                                               0.116471 ... 0.161955
15082
      0.097015
                    3.204120e-05 0.682291
                                               0.162191 ...
                                                            0.115370
                                               0.105837 ...
17452
      0.335821
                    6.438100e-05 0.427491
                                                            0.139089
      pval_nominal_f
                                 slope_se_f pval_nominal_m
                        slope_f
                                                              slope_m
1096
         1.718880e-08
                      0.456729
                                   0.075705
                                               9.155700e-01 0.009739
5262
         4.933240e-01 -0.054539
                                   0.079379
                                               3.219220e-07 -0.429800
5644
         4.637410e-18 -0.779707
                                   0.076596
                                               3.976660e-10 -0.469672
6414
         1.978000e-06 -0.333315
                                   0.066772
                                               1.653380e-01 -0.082625
7220
         6.409400e-08 0.453034
                                   0.078725
                                               3.138000e-01 0.063002
8540
         9.982650e-21 -0.887723
                                   0.078738
                                               1.892290e-09 -0.457733
9191
         5.248710e-12 -0.682629
                                   0.089412
                                               2.805060e-03 -0.289091
                                               9.165740e-01 0.013043
         4.747150e-04 -0.491287
14611
                                   0.134732
15082
         1.165430e-01 0.280837
                                   0.176978
                                               4.431060e-06 -0.558002
17452
         7.011230e-06 -0.481758
                                   0.100091
                                               8.857670e-15 -1.068840
       slope se m pval nominal
                                    slope
                                           slope se
1096
         0.091682
                  2.747400e-05 0.171830
                                           0.040604
5262
         0.079545 8.797530e-08 -0.216374
                                           0.039857
5644
         0.069091 5.637370e-52 -0.568916
                                           0.033334
         0.059205 1.393160e-08 -0.168762 0.029260
6414
7220
         0.062292 2.433600e-08 0.211910
                                           0.037395
8540
         0.070571 3.077310e-53 -0.640604 0.036976
         0.094806 3.458420e-21 -0.445408 0.045073
9191
         0.124116 2.197750e-05 -0.246023
14611
                                           0.057281
15082
         0.113158 7.896400e-06 -0.320288
                                           0.070745
17452
         0.112111 1.630390e-49 -0.838766
                                           0.049090
```

[10 rows x 22 columns]

1.3.1 mashr

```
[20]: gtex_overlap = bs[(bs['gene_id'].isin(gtex_sig.ensembl_gene_id))].

→drop_duplicates()

print(gtex_overlap.shape)
gtex_overlap
```

(14, 11)

```
[20]:
              Tissue
                                   gene_id
                                                      gencodeID
                                                                            variant_id
      2638
              Caudate
                       ENSG00000105695.14
                                            ENSG00000105695.14
                                                                   chr19:35065453:C:T
      3163
              Caudate
                       ENSG00000109775.10
                                             ENSG00000109775.10
                                                                   chr4:185462033:A:G
      4518
             Caudate
                       ENSG00000124406.16
                                             ENSG00000124406.16
                                                                    chr4:42387723:C:A
              Caudate
      5118
                       ENSG00000132199.18
                                             ENSG00000132199.18
                                                                     chr18:381997:C:G
      5635
             Caudate
                       ENSG00000136830.11
                                            ENSG00000136830.11
                                                                   chr9:127456523:G:A
      6605
              Caudate
                       ENSG00000143933.16
                                             ENSG00000143933.16
                                                                    chr2:47607174:T:C
      6687
              Caudate
                       ENSG00000146530.11
                                            ENSG00000146530.11
                                                                    chr7:12170840:G:A
      10405
             Caudate
                        ENSG00000181264.8
                                              ENSG00000181264.8
                                                                  chr11:120171373:G:A
      11623
             Caudate
                       ENSG00000189067.12
                                             ENSG00000189067.12
                                                                   chr16:11300271:A:G
      12455
             Caudate
                       ENSG00000204248.10
                                             ENSG00000204248.10
                                                                    chr6:33170900:G:A
      14958
             Caudate
                        ENSG00000235098.8
                                              ENSG00000235098.8
                                                                     chr1:1425750:T:C
      15443
             Caudate
                        ENSG00000267056.2
                                              ENSG00000267056.2
                                                                   chr19:16385981:G:A
      15478
             Caudate
                        ENSG00000268362.5
                                              ENSG00000268362.5
                                                                   chr19:23910181:A:G
      15565
             Caudate
                        ENSG00000272977.1
                                              ENSG00000272977.1
                                                                   chr22:25901440:A:C
                                        ensembl_gene_id external_gene_name
            seqnames
                           lfsr
                                  Type
      2638
                       0.044690
                                  Gene
                                        ENSG00000105695
               chr19
                                                                         MAG
      3163
                 chr4
                       0.034580
                                  Gene
                                        ENSG0000109775
                                                                       UFSP2
      4518
                 chr4
                       0.013706
                                  Gene
                                        ENSG00000124406
                                                                      ATP8A1
      5118
                chr18
                       0.044197
                                  Gene
                                        ENSG00000132199
                                                                      ENOSF1
                                  Gene
      5635
                 chr9
                       0.044628
                                        ENSG00000136830
                                                                     FAM129B
      6605
                 chr2
                       0.022399
                                  Gene
                                        ENSG00000143933
                                                                       CALM2
                       0.023059
      6687
                 chr7
                                  Gene
                                        ENSG00000146530
                                                                        VWDE
               chr11
                       0.002548
                                  Gene
      10405
                                        ENSG00000181264
                                                                     TMEM136
      11623
                chr16
                       0.007562
                                  Gene
                                        ENSG00000189067
                                                                       LITAF
                       0.045996
      12455
                 chr6
                                  Gene
                                        ENSG00000204248
                                                                     COL11A2
      14958
                 chr1
                       0.006797
                                  Gene
                                        ENSG00000235098
                                                                     ANKRD65
      15443
                chr19
                       0.040125
                                  Gene
                                        ENSG00000267056
                                                                  AC005336.1
      15478
                chr19
                       0.042637
                                  Gene
                                        ENSG00000268362
                                                                  AC092279.1
      15565
               chr22
                       0.004803
                                        ENSG00000272977
                                                                  AL008721.2
                                  Gene
              entrezgene
                                                                   description
      2638
                  4099.0
                          myelin associated glycoprotein [Source: HGNC Sy...
                          UFM1 specific peptidase 2 [Source: HGNC Symbol; ...
      3163
                 55325.0
                          ATPase phospholipid transporting 8A1 [Source: H...
      4518
                 10396.0
                          enolase superfamily member 1 [Source: HGNC Symb...
      5118
                 55556.0
                          family with sequence similarity 129 member B [...
      5635
                 64855.0
      6605
                   805.0
                             calmodulin 2 [Source: HGNC Symbol; Acc: HGNC: 1445]
      6687
               221806.0
                          von Willebrand factor D and EGF domains [Sourc...
               219902.0
                          transmembrane protein 136 [Source: HGNC Symbol; ...
      10405
                          lipopolysaccharide induced TNF factor [Source:...
      11623
                  9516.0
      12455
                  1302.0
                          collagen type XI alpha 2 chain [Source: HGNC Sy...
               441869.0
                          ankyrin repeat domain 65 [Source: HGNC Symbol; A...
      14958
      15443
                     NaN
                                                                            NaN
      15478
                     NaN
                                                                            NaN
      15565
                     NaN
                                                                            NaN
```

```
gtex_overlap.shape[0]/bs.shape[0] * 100
[21]: 1.4373716632443532
      gtex_sig[(gtex_sig['ensembl_gene_id'].isin(bs.gene_id))]
[22]:
                 ensembl_gene_id
                                    hugo_gene_id
                                                              gene_type \
      6414
              ENSG00000136830.11
                                                         protein coding
                                         FAM129B
                                                         protein_coding
      14611
              ENSG00000143933.16
                                            CALM2
      45230
               ENSG00000181264.8
                                         TMEM136
                                                         protein_coding
      135656
               ENSG00000235098.8
                                         ANKRD65
                                                         protein_coding
      137724
              ENSG00000124406.16
                                           ATP8A1
                                                         protein_coding
      137946
              ENSG00000109775.10
                                            UFSP2
                                                         protein_coding
                                         COL11A2
      138621
              ENSG00000204248.10
                                                         protein_coding
      138995
              ENSG00000146530.11
                                            VWDE
                                                         protein_coding
      142471
              ENSG00000189067.12
                                            LITAF
                                                         protein_coding
      143690
               ENSG00000267056.2
                                      AC005336.4
                                                   processed_pseudogene
      143783
               ENSG00000268362.5
                                   CTD-2017D11.1
                                                                 lincRNA
      143823
              ENSG00000105695.14
                                              MAG
                                                         protein_coding
      263405
              ENSG00000132199.18
                                          ENOSF1
                                                         protein_coding
               ENSG00000272977.1
                                   CTA-390C10.10
                                                         sense intronic
      338770
                            variant id
                                               rs_id
                                                                         Tissue
               chr9_127584339_G_A_b38
      6414
                                         rs10739693
                                                          Adipose_Subcutaneous
      14611
                chr2_46225349_C_T_b38
                                         rs12477148
                                                      Adipose_Visceral_Omentum
      45230
              chr11_120493228_T_C_b38
                                                               Artery_Coronary
                                          rs4938809
      135656
                 chr1_1419214_A_G_b38
                                          rs3766165
                                                         Breast_Mammary_Tissue
      137724
                chr4_42653682_A_T_b38
                                         rs17448575
                                                         Breast_Mammary_Tissue
               chr4_185425919_G_C_b38
                                                         Breast_Mammary_Tissue
      137946
                                         rs11132303
                                                         Breast_Mammary_Tissue
               chr6_33081200_G_GA_b38
      138621
                                        rs113353922
                                                         Breast_Mammary_Tissue
      138995
                chr7_12312811_T_C_b38
                                          rs4721084
      142471
               chr16_11551157_A_T_b38
                                         rs11644920
                                                         Breast_Mammary_Tissue
      143690
               chr19_15934939_G_A_b38
                                         rs12985091
                                                         Breast_Mammary_Tissue
      143783
               chr19_24059854_C_T_b38
                                         rs17206087
                                                         Breast_Mammary_Tissue
                                                         Breast_Mammary_Tissue
      143823
               chr19_35309759_G_T_b38
                                         rs10411704
                                                               Muscle Skeletal
      263405
                 chr18 712037 G A b38
                                          rs2612103
               chr22_25459662_G_A_b38
      338770
                                           rs6004655
                                                                         Spleen
                   maf
                         pval_nominal_sb
                                          slope_sb
                                                     slope_se_sb
                                                                          qval
      6414
              0.304647
                            7.387010e-07 -0.283660
                                                        0.056579
                                                                      0.004976
      14611
              0.072495
                            4.497930e-05 -0.480557
                                                        0.116471
                                                                      0.161955
      45230
              0.403756
                            1.832690e-05
                                         0.332900
                                                        0.075523
                                                                      0.098269
      135656
              0.131313
                            7.063370e-08
                                          0.499559
                                                        0.090555
                                                                      0.000111
                                                                      0.090512
      137724
              0.354430
                            2.577750e-04 0.246240
                                                        0.066638
              0.309343
                            4.674540e-07 -0.348749
                                                        0.067806
                                                                      0.000399
      137946
      138621
              0.118687
                            6.199050e-04 0.418594
                                                        0.121102
                                                                      0.102197
      138995
              0.452020
                            1.741340e-06 -0.301523
                                                        0.061908
                                                                      0.000744
```

```
142471 0.339646
                           1.196880e-04 0.247997
                                                       0.063687
                                                                 ... 0.035705
      143690
              0.478535
                           7.228630e-05 -0.373013
                                                       0.092781
                                                                    0.015797
      143783
              0.170455
                           2.481440e-03 -0.260946
                                                       0.085573
                                                                    0.155454
      143823
              0.179293
                           3.726460e-06 -0.621945
                                                       0.132132
                                                                     0.001446
              0.443343
                           2.579330e-05 -0.266917
                                                       0.062968
                                                                    0.123976
      263405
      338770
              0.167401
                           3.787520e-05 0.413128
                                                       0.097856 ... 0.212883
              pval_nominal_f
                               slope f
                                         slope_se_f
                                                     pval_nominal_m
                                                                       slope m
                1.978000e-06 -0.333315
                                           0.066772
                                                       1.653380e-01 -0.082625
      6414
      14611
                4.747150e-04 -0.491287
                                                       9.165740e-01 0.013043
                                           0.134732
                6.159840e-08 0.485968
      45230
                                           0.074741
                                                       1.990100e-01 0.113737
      135656
                7.568940e-06 -0.439928
                                           0.091937
                                                       1.443540e-11 -0.879777
      137724
                2.989190e-07 -0.323359
                                           0.057862
                                                       1.095640e-13 -0.613169
      137946
                9.942900e-10 0.432381
                                           0.062550
                                                       7.750080e-18 0.758610
                2.304030e-01 -0.125417
                                                       2.720980e-04 -0.553034
      138621
                                           0.103786
      138995
                5.408710e-12 -0.446355
                                           0.055294
                                                       1.501580e-03 -0.237816
      142471
                3.307190e-04 -0.171901
                                           0.045841
                                                       2.925070e-07 -0.385154
                1.961980e-07 -0.587505
      143690
                                           0.103261
                                                       1.935340e-03 -0.245596
      143783
                1.501750e-17 0.858155
                                           0.078738
                                                       8.474450e-17 0.981955
      143823
                8.536830e-14 -1.030720
                                           0.114726
                                                       1.809540e-04 -0.520457
                1.058820e-29 -0.787918
      263405
                                           0.056652
                                                       4.149110e-16 -0.508209
                7.931210e-14 -1.129030
                                           0.107327
                                                       5.155260e-16 -1.276590
      338770
              slope se m pval nominal
                                            slope
                                                   slope se
                0.059205
                          1.393160e-08 -0.168762
                                                   0.029260
      6414
      14611
                0.124116
                          2.197750e-05 -0.246023
                                                   0.057281
      45230
                0.087221 8.491350e-08 0.231096
                                                   0.041304
                0.111990 3.281960e-30 -0.726153
      135656
                                                   0.057266
      137724
                0.068668
                          1.462140e-26 -0.370315
                                                   0.031672
                0.068668 2.014910e-57 0.706155
      137946
                                                   0.035862
                0.145261
                          4.482050e-06 -0.293079
      138621
                                                   0.062803
      138995
                0.072364
                          2.765010e-20 -0.319694
                                                   0.032375
                0.068855
                          9.495520e-22 -0.323697
      142471
                                                   0.031398
      143690
                0.076638
                          1.176010e-12 -0.331468
                                                   0.044801
      143783
                0.093415
                          1.509120e-67 0.972900
                                                   0.043631
      143823
                0.132593
                          8.545960e-22 -0.693095
                                                   0.067157
                0.056303
                          5.746020e-65 -0.613437
      263405
                                                   0.031989
      338770
                0.104690
                          4.896640e-51 -1.193670
                                                   0.056659
      [14 rows x 22 columns]
[23]: gtex_sig[(gtex_sig['ensembl_gene_id'].isin(bs.gene_id))]\
          .to_csv("siEQTL_gtex_comparison.csv", index=False)
```

1.3.2 eigenMT corrected

```
[24]: gtex_overlap = dft[(dft['gene_id'].isin(gtex_sig.ensembl_gene_id)) & (dft["BF"]__
      print(gtex_overlap.shape)
      gtex_overlap.sort_values("BF").head()
     (57, 14)
[24]:
                    variant_id
                                           gene_id
                                                            gencodeID
                                                                          slope \
      473
             chr4:42392391:T:G ENSG00000124406.16
                                                   ENSG00000124406.16 -0.291917
      4271
             chr8:11603602:A:G
                               ENSG00000154328.15
                                                   ENSG00000154328.15 0.380078
      406
             chr1:89159687:G:A ENSG00000117226.11
                                                   ENSG00000117226.11 0.431197
      2556
             chr8:63463308:T:C
                               ENSG00000137563.11
                                                   ENSG00000137563.11 0.527946
      3627 chr22:25059120:A:C
                                ENSG00000272977.1
                                                     ENSG00000272977.1 0.626299
            statistic pval_nominal
                                              eigenMT_BH TESTS
                                                                 Type
      473 -13.197929
                          0.000004
                                    0.002091
                                                0.362373
                                                            502
                                                                 Gene
      4271 14.183350
                          0.000002
                                    0.002130
                                                0.458802
                                                            888
                                                                 Gene
      406
            15.848961
                          0.000009 0.002374
                                                0.362373
                                                            266 Gene
      2556 10.192544
                                                            335
                                                                 Gene
                          0.000007 0.002460
                                                0.438136
      3627 16.616479
                          0.000003 0.002482
                                                0.438136
                                                            744 Gene
            ensembl_gene_id external_gene_name entrezgene
      473
           ENSG00000124406
                                       ATP8A1
                                                   10396.0
      4271 ENSG00000154328
                                        NEIL2
                                                 252969.0
      406
           ENSG00000117226
                                          GBP3
                                                   2635.0
      2556 ENSG00000137563
                                           GGH
                                                   8836.0
                                   AL008721.2
      3627 ENSG00000272977
                                                      NaN
                                                 description
      473
            ATPase phospholipid transporting 8A1 [Source: H...
           nei like DNA glycosylase 2 [Source: HGNC Symbol...
      4271
            guanylate binding protein 3 [Source: HGNC Symbo...
      406
      2556
           gamma-glutamyl hydrolase [Source: HGNC Symbol; A...
      3627
                                                         NaN
[25]: gtex_overlap = dft[(dft['gene_id'].isin(gtex_sig.ensembl_gene_id))].
      →drop_duplicates()
      print(gtex overlap.shape)
      gtex_overlap.sort_values("BF").head()
     (57, 14)
[25]:
                    variant_id
                                                            gencodeID
                                                                          slope \
                                           gene_id
      473
             chr4:42392391:T:G
                               ENSG00000124406.16
                                                   ENSG00000124406.16 -0.291917
      4271
             chr8:11603602:A:G
                               ENSG00000154328.15
                                                   ENSG00000154328.15
                                                                       0.380078
      406
             chr1:89159687:G:A ENSG00000117226.11
                                                   ENSG00000117226.11 0.431197
```

```
2556
             chr8:63463308:T:C ENSG00000137563.11 ENSG00000137563.11 0.527946
      3627 chr22:25059120:A:C
                                 ENSG00000272977.1
                                                      ENSG00000272977.1
                                                                         0.626299
            statistic pval_nominal
                                            BF
                                                eigenMT_BH TESTS
                                                                   Type
      473 -13.197929
                           0.00004
                                     0.002091
                                                  0.362373
                                                              502
                                                                   Gene
      4271 14.183350
                           0.000002
                                     0.002130
                                                  0.458802
                                                              888
                                                                   Gene
      406
            15.848961
                           0.000009
                                     0.002374
                                                  0.362373
                                                              266
                                                                   Gene
                                                              335
                                                                   Gene
      2556 10.192544
                           0.000007
                                     0.002460
                                                  0.438136
      3627 16.616479
                           0.000003 0.002482
                                                  0.438136
                                                              744 Gene
            ensembl_gene_id external_gene_name
                                                 entrezgene
      473
            ENSG00000124406
                                        ATP8A1
                                                    10396.0
      4271 ENSG00000154328
                                         NEIL2
                                                   252969.0
      406
            ENSG00000117226
                                           GBP3
                                                     2635.0
      2556 ENSG00000137563
                                            GGH
                                                     8836.0
      3627 ENSG00000272977
                                    AL008721.2
                                                        NaN
                                                   description
      473
            ATPase phospholipid transporting 8A1 [Source: H...
            nei like DNA glycosylase 2 [Source: HGNC Symbol...
      4271
      406
            guanylate binding protein 3 [Source: HGNC Symbo...
            gamma-glutamyl hydrolase [Source: HGNC Symbol; A...
      2556
      3627
                                                           NaN
[26]: gtex_overlap.shape[0]/dft.shape[0] * 100
[26]: 1.1010237589337455
 []:
```