## main

## September 7, 2021

## 1 Generate supplemental data of all significant integration analysis

```
[1]:
    import pandas as pd
[2]: genes = pd.read_csv('../../m/genes/integration_by_symbol.txt', sep='\t').
      →rename(columns={'gene': 'Feature_ID'})
     genes['Feature_Type'] = 'Gene'
     genes.head()
[2]:
                             Symbol
                                                    variant_id
                                                                        rsid A1
                   gene_id
                             ZSCAN9
     0 ENSG00000137185.11
                                             chr6:28651576:A:G
                                                                 rs34724414
                                             chr6:28662914:G:A
       ENSG00000137185.11
                             ZSCAN9
                                                                 rs13194504
     2 ENSG00000137185.11
                             ZSCAN9
                                     chr6:28468237:TAAACATA:T
                                                                rs138024639
                                                                              Τ
     3 ENSG00000137185.11
                                            chr6:28594470:TG:T
                                                                              Τ
                             ZSCAN9
                                                                  rs34119086
     4 ENSG00000137185.11
                                             chr6:28608366:T:A
                             ZSCAN9
                                                                  rs4947336
              A2
                         OR
                                           pval_nominal
                                                          adj.P.Val
                                                                        logFC
     0
               G
                  1.275700
                             1.120000e-43
                                                0.001482
                                                           0.022627 -0.07713
     1
                  1.275700
                             1.180000e-43
                                                0.001482
                                                           0.022627 -0.07713
     2
                  0.770435
                             2.595000e-43
                                                0.000917
       TAAACATA
                                                           0.022627 -0.07713
     3
              TG
                  0.787730
                             5.296000e-42
                                                0.001482
                                                           0.022627 -0.07713
     4
                  1.268900
                             7.480000e-42
                                                0.001482
               Α
                                                           0.022627 -0.07713
                  eqtl_slope
                               de_dir
                                       eqtl_gwas_dir agree_direction
     0 -3.126621
                           -1
                                   -1
                                                   -1
                                                                   Yes
     1 -3.126621
                           -1
                                   -1
                                                   -1
                                                                  Yes
     2 -3.126621
                           -1
                                   -1
                                                   -1
                                                                  Yes
     3 -3.126621
                           -1
                                   -1
                                                   -1
                                                                  Yes
     4 -3.126621
                           -1
                                   -1
                                                   -1
                                                                  Yes
        pgc2_a1_same_as_our_counted Feature_Type
     0
                               False
     1
                               False
                                             Gene
     2
                                True
                                             Gene
     3
                                True
                                             Gene
     4
                               False
                                             Gene
```

```
[3]: trans = pd.read_csv('../../_m/transcripts/integration_by_symbol.txt', sep='\t').
     →rename(columns={'gene': 'Feature_ID', 'gene_name': 'Symbol'})
    trans['Feature Type'] = 'Transcript'
    trans.head()
[3]:
                 gene_id Symbol
                                         variant_id
                                                          rsid A1
                                                                   A2
                                                                             OR
    0 ENST00000418983.1
                           HCG4
                                  chr6:29854872:C:T rs2517857 C
                                                                       1.166700
    1 ENST00000418983.1
                           HCG4
                                  chr6:29638984:G:A rs3095268 G
                                                                       1.146100
                           HCG4 chr6:29956359:AG:A rs5875221 A
    2 ENST00000418983.1
                                                                   AG
                                                                       0.859074
    3 ENST00000418983.1
                           HCG4
                                  chr6:29852420:A:G rs9258609 A
                                                                    G
                                                                       1.159100
                           HCG4
    4 ENST00000418983.1
                                  chr6:29850949:A:G rs2523761 A
                                                                    G
                                                                      1.156400
                  P pval_nominal adj.P.Val
                                                 logFC
                                                                  eqtl slope
      1.170000e-22 7.560360e-05
                                    0.028722 0.251395
                                                        3.568189
    1 4.090000e-22 1.498240e-04
                                    0.028722 0.251395
                                                        3.568189
                                                                           1
    2 1.624000e-21 3.937150e-05
                                    0.028722 0.251395 3.568189
                                                                           1
    3 1.860000e-21 1.103540e-04
                                    0.028722 0.251395 3.568189
                                                                           1
    4 2.000000e-21 1.462340e-09
                                    0.028722 0.251395 3.568189
                                                                           1
               eqtl_gwas_dir agree_direction pgc2_a1_same_as_our_counted
       de dir
    0
                                         Yes
    1
            1
                           1
                                         Yes
                                                                    False
    2
            1
                           1
                                         Yes
                                                                     True
    3
            1
                           1
                                         Yes
                                                                    False
            1
                           1
                                         Yes
                                                                    False
      Feature_Type
        Transcript
    1
        Transcript
    2
        Transcript
        Transcript
    3
        Transcript
[4]: exons = pd.read_csv('../../_m/exons/integration_by_symbol.txt', sep='\t').
     →rename(columns={'gene': 'Feature_ID'})
    exons['Feature Type'] = 'Exon'
    exons.head()
[4]:
                 Symbol
                                                 rsid A1 A2
                                                                  OR
       gene_id
                                variant_id
    0 e378566 ZSCAN26
                         chr6:28744470:A:G rs1233578 A
                                                             1.22280
    1 e378558 ZSCAN26
                         chr6:28744470:A:G rs1233578 A
                                                             1.22280
    2 e378566 ZSCAN26
                         chr6:28716406:T:G rs9295778
                                                      Т
                                                             0.82449
    3 e378558 ZSCAN26
                         chr6:28716406:T:G rs9295778 T
                                                          G
                                                             0.82449
    4 e378566 ZSCAN26
                         chr6:28721895:C:A rs9468413 C
                  P pval_nominal adj.P.Val
                                                 logFC
                                                                  eqtl slope \
       4.320000e-44 7.294750e-10
                                    0.018675 -0.084735 -3.408383
```

```
1 4.320000e-44 1.080300e-09
                                     0.019079 -0.084425 -3.399939
                                                                           -1
     2 4.520000e-42 6.790160e-10
                                     0.018675 -0.084735 -3.408383
                                                                            -1
     3 4.520000e-42 8.899240e-10
                                     0.019079 -0.084425 -3.399939
                                                                           -1
     4 6.860000e-42 6.790160e-10
                                     0.018675 -0.084735 -3.408383
                                                                           -1
       de_dir
                eqtl_gwas_dir agree_direction pgc2_a1_same_as_our_counted \
     0
           -1
                           -1
                                          Yes
                                                                     False
     1
            -1
                           -1
                                          Yes
                                                                     False
     2
           -1
                           -1
                                          Yes
                                                                     False
     3
           -1
                           -1
                                          Yes
                                                                     False
     4
            -1
                           -1
                                          Yes
                                                                     False
      Feature_Type
     0
               Exon
     1
               Exon
     2
               Exon
     3
               Exon
     4
               Exon
[5]: juncs = pd.read_csv('../../_m/junctions/integration_by_symbol.txt', sep='\t').
     →rename(columns={'gene': 'Feature_ID'})
     juncs['Feature_Type'] = 'Junction'
     juncs.head()
[5]:
                          gene_id
                                    Symbol
                                                          variant_id
                                                                             rsid \
                                   ZSCAN26
     0 chr6:28271935-28272021(+)
                                                   chr6:28744470:A:G
                                                                        rs1233578
     1 chr6:28271935-28272021(+)
                                   ZSCAN26 chr6:28468237:TAAACATA:T rs138024639
     2 chr6:28271935-28272021(+)
                                   ZSCAN26
                                                   chr6:28716406:T:G
                                                                        rs9295778
     3 chr6:28271935-28272021(+)
                                   ZSCAN26
                                                   chr6:28721895:C:A
                                                                        rs9468413
     4 chr6:28271935-28272021(+)
                                   ZSCAN26
                                                   chr6:28426903:C:T
                                                                       rs13201681
                                            pval_nominal adj.P.Val
       A1
                A2
                           OR.
                                                                        logFC \
                    1.222800 4.320000e-44 9.439080e-11
     0
       Α
                                                            0.018932 -0.26177
     1
       Τ
          TAAACATA 0.770435 2.595000e-43 2.249520e-04
                                                            0.018932 -0.26177
     2
      Т
                  G 0.824490 4.520000e-42 1.737900e-10
                                                            0.018932 -0.26177
                  A 0.825110 6.860000e-42 1.737900e-10
     3 C
                                                            0.018932 -0.26177
     4 C
                  T 1.265200 1.320000e-41 2.249520e-04
                                                            0.018932 -0.26177
                eqtl_slope de_dir eqtl_gwas_dir agree_direction \
     0 - 3.493012
                          -1
                                                 -1
                                  -1
                                                                Yes
     1 -3.493012
                          -1
                                  -1
                                                 -1
                                                                Yes
     2 -3.493012
                          -1
                                  -1
                                                 -1
                                                                Yes
     3 -3.493012
                          -1
                                  -1
                                                 -1
                                                                Yes
     4 -3.493012
                                                                Yes
                          -1
                                  -1
                                                 -1
       pgc2_a1_same_as_our_counted Feature_Type
     0
                              False
                                        Junction
```

```
1
                              True
                                       Junction
    2
                             False
                                       Junction
    3
                             False
                                       Junction
    4
                             False
                                       Junction
[6]: df = pd.concat([genes, trans, exons, juncs], axis=0, sort=False)
    print(df.shape)
    df.head(2)
    (13169, 18)
[6]:
                  gene_id Symbol
                                         variant id
                                                           rsid A1 A2
                                                                           OR \
    O ENSGO0000137185.11 ZSCAN9 chr6:28651576:A:G rs34724414 A G 1.2757
    1 ENSG00000137185.11 ZSCAN9 chr6:28662914:G:A rs13194504 G A 1.2757
                  P pval_nominal adj.P.Val
                                                             t eqtl_slope \
                                               logFC
                         0.001482
    0 1.120000e-43
                                    0.022627 -0.07713 -3.126621
    1 1.180000e-43
                         0.001482
                                    0.022627 -0.07713 -3.126621
                                                                        -1
       de_dir eqtl_gwas_dir agree_direction pgc2_a1_same_as_our_counted \
    0
           -1
                                         Yes
                                                                   False
                          -1
    1
           -1
                          -1
                                         Yes
                                                                   False
      Feature_Type
    0
              Gene
    1
              Gene
[7]: df.to_csv('BrainSeq_Phase3_Caudate_GWAS_DE_eQTL_Integration.txt.gz',
     →index=False, header=True, sep='\t')
```