## main

## September 4, 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]:
                   gene_id Symbol
                                             variant_id A1 A2 risk_allele
                                                                               OR.
     0 ENSG00000148842.17
                            CNNM2
                                   chr10:102825368:C:A C
                                                            Α
                                                                        С
                                                                           1.0604
     1 ENSG00000148842.17
                            CNNM2
                                   chr10:102825648:A:T
                                                            Τ
                                                                           1.0635
                                                                        Α
     2 ENSG00000148842.17
                            CNNM2
                                   chr10:102832918:G:C
                                                            C
                                                                        G 1.0644
     3 ENSG00000148842.17
                            CNNM2
                                   chr10:102835663:T:G
                                                                        Т
                                                                           1.0617
     4 ENSG00000148842.17 CNNM2
                                                                           1.0633
                                   chr10:102835962:C:T C
                                                   logFC
                      pval_nominal
                                    adj.P.Val
                                                                 t
                                                                    eqtl_slope
     0 1.120000e-09
                      2.413790e-11
                                     0.003899
                                                0.053701
                                                          3.799656
                                                                     -0.169331
     1 2.300000e-10 1.944530e-05
                                     0.003899
                                                0.053701
                                                          3.799656
                                                                     -0.119926
     2 1.330000e-10 3.352800e-06
                                                                     -0.127135
                                     0.003899
                                               0.053701
                                                          3.799656
     3 4.970000e-10 2.310390e-10
                                     0.003899
                                                0.053701
                                                          3.799656
                                                                     -0.163240
     4 2.590000e-10 3.624190e-09
                                     0.003899
                                                0.053701
                                                         3.799656
                                                                     -0.152542
       de_dir eqtl_gwas_dir agree_direction Feature_Type
     0
           Uр
                       Down
                                                     Gene
                                         No
     1
           Uр
                       Down
                                         No
                                                     Gene
     2
                       Down
                                                     Gene
           Uр
                                         No
     3
           Uр
                       Down
                                         No
                                                     Gene
     4
                                                     Gene
           Uр
                       Down
                                         No
[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()
```

```
gene_id Symbol
       ENST00000369878.8
                          CNNM2 chr10:102825368:C:A
     0
                                                       C
                                                          Α
                                                                         1.0604
     1 ENST00000369878.8
                          CNNM2
                                  chr10:102825648:A:T
                                                       Α
                                                          Т
                                                                      Α
                                                                         1.0635
     2 ENST00000369878.8
                          CNNM2
                                  chr10:102832918:G:C
                                                       G
                                                          C
                                                                      G
                                                                         1.0644
     3 ENST00000369878.8
                          CNNM2
                                  chr10:102835663:T:G
                                                       Т
                                                          G
                                                                      Τ
                                                                         1.0617
                                                                         1.0633
     4 ENST00000369878.8 CNNM2
                                  chr10:102835962:C:T
                                                       С
                                                                      C
                     pval_nominal
                                    adj.P.Val
                                                  logFC
                                                                   eqtl_slope \
      1.120000e-09 4.928250e-11
                                     0.000852 0.084962
                                                                    -0.179745
                                                         4.753266
     1 2.300000e-10 7.617130e-05
                                     0.000852
                                               0.084962
                                                         4.753266
                                                                    -0.119358
     2 1.330000e-10 1.354810e-05
                                     0.000852
                                               0.084962
                                                         4.753266
                                                                    -0.128029
     3 4.970000e-10 9.108410e-11
                                     0.000852
                                               0.084962
                                                                    -0.179316
                                                         4.753266
     4 2.590000e-10 7.879660e-09
                                     0.000852
                                               0.084962
                                                         4.753266
                                                                    -0.160431
       de_dir eqtl_gwas_dir agree_direction Feature_Type
     0
          Up
                       Down
                                              Transcript
     1
          Uр
                       Down
                                         No
                                              Transcript
     2
                       Down
                                         No
                                              Transcript
          Uр
     3
                       Down
                                         No
                                              Transcript
          Uр
     4
          Uр
                       Down
                                         No
                                              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]:
       gene_id Symbol
                                 variant_id A1 A2 risk_allele
                                                                   OR
     0 e598887
                CNNM2
                        chr10:102825368:C:A
                                                Α
                                                            C
                                                               1.0604
     1 e598887
                 CNNM2
                        chr10:102825648:A:T
                                             Α
                                                Τ
                                                            Α
                                                               1.0635
     2 e598887
                CNNM2
                        chr10:102832918:G:C
                                             G
                                                C
                                                            G
                                                               1.0644
                                                            Т
     3 e598887
                CNNM2
                        chr10:102835663:T:G T
                                                G
                                                               1.0617
     4 e598887
                CNNM2
                        chr10:102835962:C:T C
                                                            C
                                                               1.0633
                     pval nominal
                                    adj.P.Val
                                                 logFC
                                                                  eqtl_slope
                     1.275560e-10
       1.120000e-09
                                     0.001824
                                               0.08217 4.275115
                                                                   -0.172984
     1 2.300000e-10 8.685790e-05
                                     0.001824
                                               0.08217
                                                        4.275115
                                                                   -0.117049
     2 1.330000e-10 5.547170e-05
                                     0.001824
                                               0.08217
                                                        4.275115
                                                                   -0.117347
     3 4.970000e-10 5.187010e-10
                                     0.001824 0.08217
                                                        4.275115
                                                                   -0.169565
     4 2.590000e-10 9.561270e-09
                                     0.001824 0.08217
                                                       4.275115
                                                                   -0.157295
       de_dir eqtl_gwas_dir agree_direction Feature_Type
     0
          Uр
                       Down
                                         No
                                                    Exon
                       Down
                                         No
                                                    Exon
     1
          Uр
     2
          Uр
                       Down
                                         No
                                                    Exon
     3
          Uр
                       Down
                                         No
                                                    Exon
                       Down
                                         No
                                                    Exon
          Uр
```

variant\_id A1 A2 risk\_allele

OR. \

[3]:

```
[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]:
                                       Symbol
                             gene_id
                                                          variant_id A1
                                                                            A2
          chr11:47180691-47182402(-)
                                      PACSIN3
                                               chr11:46729945:T:TAGG
                                                                          TAGG
                                                  chr11:46945082:T:C
     1
          chr11:47180691-47182402(-)
                                      PACSIN3
                                                                             C
     2 chr11:113412884-113415420(-)
                                         DRD2
                                                 chr11:113447023:G:C G
                                                                             C
     3 chr11:113412884-113415420(-)
                                         DRD2
                                                 chr11:113457959:A:C
                                                                             C
     4 chr11:113412884-113415420(-)
                                         DRD2
                                                 chr11:113463378:C:T C
                                            pval nominal adj.P.Val
       risk_allele
                          OR
                                                                         logFC
     0
              TAGG
                    0.925705 3.875000e-09
                                                0.000221
                                                           0.022512 -0.186281
     1
                 С
                    0.896840
                              3.720000e-08
                                                0.000115
                                                           0.022512 -0.186281
                 G 1.066000 4.300000e-10
                                                0.000011
                                                           0.004208 0.302443
     2
     3
                 A 1.065800 3.690000e-10
                                                0.000024
                                                           0.004208
                                                                     0.302443
                  1.064600 8.050000e-10
                                                0.000005
                                                           0.004208
                                                                     0.302443
                  eqtl_slope de_dir eqtl_gwas_dir agree_direction Feature_Type
     0 - 3.419199
                    0.208793
                               Down
                                               Uр
                                                               No
                                                                       Junction
     1 - 3.419199
                    0.421745
                               Down
                                               Up
                                                               No
                                                                       Junction
     2 4.079084
                   -0.260710
                                 Up
                                             Down
                                                               No
                                                                       Junction
     3 4.079084
                   -0.252745
                                 Uр
                                             Down
                                                               No
                                                                       Junction
     4 4.079084
                   -0.272205
                                 Uр
                                                               No
                                                                       Junction
                                             Down
[6]: df = pd.concat([genes, trans, exons, juncs], axis=0, sort=False)
     print(df.shape)
     df.head(2)
    (13169, 17)
[6]:
                   gene_id Symbol
                                            variant_id A1 A2 risk_allele
     0 ENSG00000148842.17
                            CNNM2
                                   chr10:102825368:C:A C A
                                                                          1.0604
     1 ENSG00000148842.17 CNNM2
                                   chr10:102825648:A:T A T
                                                                          1.0635
                                                                       Α
                   P pval_nominal adj.P.Val
                                                                t eqtl slope
                                                  logFC
      1.120000e-09 2.413790e-11
                                     0.003899
                                               0.053701
                                                                     -0.169331
                                                         3.799656
     1 2.300000e-10 1.944530e-05
                                     0.003899 0.053701
                                                        3.799656
                                                                     -0.119926
       de_dir eqtl_gwas_dir agree_direction Feature_Type
     0
           Uр
                       Down
                                         No
                                                    Gene
     1
           Uр
                       Down
                                         No
                                                    Gene
[7]: df.to_csv('BrainSeq_Phase3_Caudate_GWAS_DE_eQTL_Integration.txt.gz',_
      →index=False, header=True, sep='\t')
```