## main

## August 3, 2021

## 1 Extract male bias genes on the X chromosome

```
[1]: import pandas as pd
[2]: def get_deg(tissue):
         fn = "../../../differential_expression/%s/" % tissue +\
         "metrics_summary/_m/chrom_annotation_genes.txt"
         return pd.read_csv(fn, sep='\t').loc[:, ["gene_id", "seqname", "Symbol",
                                                              "t", "adj.P.Val"]]
[3]: df = pd.DataFrame()
     for tissue in ["caudate", "dlpfc", "hippocampus"]:
         dt = get_deg(tissue)
         dt["Tissue"] = tissue
         df = pd.concat([df, dt], axis=0)
     df["ensemblID"] = df.gene_id.str.replace("\\..*", "", regex=True)
     df.shape
[3]: (1058, 7)
[4]: xci = pd.read_csv("../../_h/xci_status_hg19.txt", sep='\t')
     xci["ensemblID"] = xci["Gene ID"].str.replace("\\..*", "", regex=True)
     xci.head(2)
[4]:
      Gene name
                            Gene ID Chr Start position End position \
                                                 192989
                                                               220023
     0
         PLCXD1 ENSG00000182378.8
                                      Х
         GTPBP6 ENSG00000178605.8
                                                 220025
                                                               230886
      Transcript type Combined XCI status
                                                  ensemblID
     0 protein_coding
                                    escape ENSG00000182378
     1 protein_coding
                                    escape
                                            ENSG00000178605
[5]: xci.groupby("Combined XCI status").size()
[5]: Combined XCI status
     escape
                  99
     inactive
                 431
     variable
                 101
```

dtype: int64

```
[6]: tt = df.merge(xci[(xci["Combined XCI status"] == "escape")], on="ensemblID")
     tt[(tt['t'] > 0)]
[6]:
                                            Symbol
                                                                 adj.P.Val
                          gene_id seqname
                                                           t
     55
         ENSG00000182378.13_PAR_Y
                                            PLCXD1
                                                    6.988151
                                                              4.239261e-09
                                      chrY
     56
         ENSG00000182378.13_PAR_Y
                                            PLCXD1
                                                              9.826634e-07
                                      chrY
                                                    6.107645
     57
         ENSG00000182378.13_PAR_Y
                                      chrY
                                           PLCXD1
                                                    4.772588 7.966110e-04
        ENSG00000002586.18 PAR Y
                                              CD99
                                      chrY
                                                    4.264277
                                                              3.645847e-03
     73
        ENSG00000002586.18 PAR Y
                                      chrY
                                              CD99
                                                    3.901722 1.477514e-02
     74 ENSG00000002586.18 PAR Y
                                              CD99
                                                    4.073952 1.481474e-02
                                      chrY
     80 ENSG00000169093.15_PAR_Y
                                      chrY
                                             ASMTL
                                                    3.831986 1.701108e-02
     83
        ENSG00000178605.13 PAR Y
                                      chrY
                                           GTPBP6 3.599197 2.847852e-02
                                                             Gene ID Chr
              Tissue
                            ensemblID Gene name
                                                                          \
                      ENSG00000182378
                                                   ENSG00000182378.8
     55
             caudate
                                          PLCXD1
     56
               dlpfc
                      ENSG00000182378
                                          PLCXD1
                                                   ENSG00000182378.8
                                                                        X
     57
         hippocampus
                      ENSG00000182378
                                          PLCXD1
                                                   ENSG00000182378.8
                                                                        Х
     72
             caudate
                     ENSG00000002586
                                            CD99
                                                  ENSG0000002586.13
                                                                        Х
     73
               dlpfc
                     ENSG00000002586
                                            CD99
                                                  ENSG00000002586.13
                                                                        Х
     74
                                                                        X
         hippocampus
                     ENSG00000002586
                                            CD99
                                                  ENSG0000002586.13
     80
               dlpfc
                      ENSG00000169093
                                           ASMTL
                                                  ENSG00000169093.10
                                                                        Х
     83
               dlpfc
                      ENSG00000178605
                                          GTPBP6
                                                   ENSG00000178605.8
                                                                        Х
         Start position
                         End position Transcript type Combined XCI status
     55
                 192989
                               220023
                                       protein coding
                                                                     escape
     56
                 192989
                               220023
                                       protein_coding
                                                                     escape
     57
                 192989
                               220023
                                       protein coding
                                                                     escape
                                       protein_coding
     72
                2609220
                              2659350
                                                                     escape
     73
                                       protein coding
                2609220
                              2659350
                                                                     escape
     74
                2609220
                              2659350
                                       protein coding
                                                                     escape
                                       protein_coding
     80
                1522032
                              1572655
                                                                     escape
     83
                 220025
                               230886
                                       protein_coding
                                                                     escape
    Escaped genes are also located on the PAR regions of the Y chromosome.
[7]: xlinked = df[(df['seqname'] == 'chrX')].copy()
     xlinked_male = xlinked[(xlinked["t"] > 0)].copy()
     xlinked_female = xlinked[(xlinked["t"] < 0)].copy()</pre>
[8]: xlinked.groupby("Tissue").size()
[8]: Tissue
     caudate
                    45
     dlpfc
                    60
    hippocampus
                    31
     dtype: int64
```

```
[9]: xlinked_male.groupby("Tissue").size()
 [9]: Tissue
      caudate
                       3
      dlpfc
                      18
      hippocampus
                       1
      dtype: int64
[10]: xlinked_female.groupby("Tissue").size()
[10]: Tissue
                      42
      caudate
                      42
      dlpfc
      hippocampus
                      30
      dtype: int64
[11]: xlinked_male
[11]:
                       gene_id seqname
                                            Symbol
                                                                adj.P.Val
                                                                                 Tissue
      126
            ENSG00000213468.4
                                                                0.000705
                                             FIRRE
                                                     4.689886
                                                                                caudate
                                   chrX
      132
            ENSG00000186675.6
                                   chrX
                                            MAGEE2
                                                     4.613375
                                                                 0.000956
                                                                                caudate
      133
           ENSG00000102001.12
                                   chrX
                                           CACNA1F
                                                     4.608831
                                                                                caudate
                                                                 0.000963
      97
           ENSG00000172465.13
                                   chrX
                                            TCEAL1
                                                     4.402338
                                                                 0.003347
                                                                                  dlpfc
      99
            ENSG00000236064.1
                                   chrX
                                               NaN
                                                     4.364580
                                                                0.003866
                                                                                  dlpfc
      125
            ENSG00000277883.1
                                   chrX
                                           NLRP3P1
                                                     4.155232
                                                                0.007473
                                                                                  dlpfc
      130
           ENSG00000184515.10
                                   chrX
                                              BEX5
                                                     4.126387
                                                                0.008105
                                                                                  dlpfc
      199
                                   chrX
                                                                                  dlpfc
           ENSG00000204071.10
                                            TCEAL6
                                                     3.836015
                                                                0.017011
      232
           ENSG00000147155.10
                                   chrX
                                                     3.751150
                                                                                  dlpfc
                                               EBP
                                                                0.020360
      277
            ENSG00000232119.7
                                   chrX
                                             MCTS1
                                                     3.632172
                                                                 0.026693
                                                                                  dlpfc
      291
                                                                                  dlpfc
           ENSG00000198932.12
                                   chrX
                                           GPRASP1
                                                     3.611593
                                                                 0.027517
      314
            ENSG00000186675.6
                                   chrX
                                            MAGEE2
                                                     3.555431
                                                                0.031388
                                                                                  dlpfc
      330
                                                     3.530376
                                                                                  dlpfc
           ENSG00000102054.17
                                   chrX
                                             RBBP7
                                                                0.032470
      361
            ENSG00000278530.4
                                   chrX
                                          CHMP1B2P
                                                     3.485690
                                                                0.035151
                                                                                  dlpfc
      384
            ENSG00000133169.5
                                   chrX
                                              BEX1
                                                     3.454265
                                                                0.037068
                                                                                  dlpfc
      399
            ENSG00000184905.8
                                   chrX
                                            TCEAL2
                                                     3.428241
                                                                                  dlpfc
                                                                0.039058
                                            ARMCX2
      408
           ENSG00000184867.13
                                   chrX
                                                     3.417176
                                                                0.039832
                                                                                  dlpfc
      483
           ENSG00000102401.19
                                   chrX
                                            ARMCX3
                                                     3.328593
                                                                 0.045811
                                                                                  dlpfc
      486
                                                                                  dlpfc
           ENSG00000133134.11
                                   chrX
                                              BEX2
                                                     3.325992
                                                                 0.045811
      505
            ENSG00000224204.1
                                   chrX
                                          PHEX-AS1
                                                     3.311985
                                                                 0.046573
                                                                                  dlpfc
      528
                                                                                  dlpfc
            ENSG00000178947.8
                                   chrX
                                         SMIM10L2A
                                                     3.297549
                                                                 0.047002
      85
           ENSG00000147124.12
                                             ZNF41
                                                                 0.013322
                                                                           hippocampus
                                   chrX
                                                     4.105295
                  ensemblID
      126
           ENSG00000213468
      132
           ENSG0000186675
      133
           ENSG0000102001
```

97

ENSG00000172465

```
125
          ENSG00000277883
      130
           ENSG00000184515
      199
           ENSG00000204071
      232 ENSG00000147155
      277
           ENSG00000232119
      291
          ENSG00000198932
      314
          ENSG00000186675
      330
          ENSG00000102054
      361
          ENSG00000278530
      384 ENSG00000133169
          ENSG00000184905
      408
          ENSG00000184867
      483 ENSG00000102401
      486
          ENSG00000133134
      505
          ENSG00000224204
      528
           ENSG00000178947
      85
           ENSG00000147124
[12]: xci["ensemblID"] = xci["Gene ID"].str.replace("\\..*", "", regex=True)
      xlinked_male["ensemblID"] = xlinked_male.gene_id.str.replace("\\..*", "",__
       →regex=True)
      xlinked_male.merge(xci[["ensemblID", "Combined XCI status"]], on="ensemblID")
[12]:
                     gene_id seqname
                                         Symbol
                                                           adj.P.Val
                                                                            Tissue \
      0
                                                             0.000705
           ENSG00000213468.4
                                 chrX
                                          FIRRE 4.689886
                                                                           caudate
      1
          ENSG00000102001.12
                                        CACNA1F
                                                 4.608831
                                                             0.000963
                                                                           caudate
                                 chrX
      2
                                         TCEAL1
                                                 4.402338
                                                             0.003347
          ENSG00000172465.13
                                 chrX
                                                                             dlpfc
      3
           ENSG00000236064.1
                                 chrX
                                                 4.364580
                                                             0.003866
                                                                             dlpfc
                                            NaN
      4
           ENSG00000232119.7
                                 chrX
                                          MCTS1
                                                 3.632172
                                                             0.026693
                                                                             dlpfc
      5
          ENSG00000198932.12
                                 chrX
                                        GPRASP1
                                                 3.611593
                                                             0.027517
                                                                             dlpfc
      6
          ENSG00000102054.17
                                 chrX
                                          RBBP7
                                                 3.530376
                                                             0.032470
                                                                             dlpfc
      7
           ENSG00000184905.8
                                 chrX
                                         TCEAL2
                                                 3.428241
                                                             0.039058
                                                                             dlpfc
      8
          ENSG00000184867.13
                                 chrX
                                         ARMCX2 3.417176
                                                             0.039832
                                                                             dlpfc
      9
          ENSG00000102401.19
                                 chrX
                                         ARMCX3
                                                 3.328593
                                                             0.045811
                                                                             dlpfc
                                                                             dlpfc
      10
          ENSG00000133134.11
                                 chrX
                                           BEX2
                                                 3.325992
                                                             0.045811
                                                                             dlpfc
      11
           ENSG00000224204.1
                                 chrX PHEX-AS1
                                                 3.311985
                                                             0.046573
      12
         ENSG00000147124.12
                                 chrX
                                          ZNF41
                                                 4.105295
                                                             0.013322 hippocampus
                ensemblID Combined XCI status
          ENSG00000213468
      0
                                      variable
      1
          ENSG00000102001
                                      inactive
      2
          ENSG00000172465
                                      inactive
      3
          ENSG00000236064
                                      inactive
      4
          ENSG00000232119
                                      inactive
      5
          ENSG00000198932
                                      inactive
          ENSG00000102054
                                      variable
```

99

ENSG00000236064

7	ENSG00000184905	inactive
8	ENSG00000184867	inactive
9	ENSG00000102401	inactive
10	ENSG00000133134	inactive
11	ENSG00000224204	inactive
12	ENSG00000147124	inactive

## []:[