main

July 13, 2021

1 Feature summary analysis

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

1.1 Summary plots

1.1.1 MSSM Penn Pitt

```
[2]: mpp = pd.read_csv('../../_m/mssm_penn_pitt_maleVfemale.tsv', sep='\t')
mpp = mpp[(mpp['adj.P.Val'] < 0.05)].sort_values('adj.P.Val')
mpp.head()</pre>
```

```
[2]:
                              logFC
                                                                    adj.P.Val \
                    Geneid
                                      AveExpr
                                                           P. Value
        ENSG00000241859.7 7.426322 0.211930
                                               106.905258
                                                                0.0
                                                                          0.0
    1 ENSG00000206159.11 6.829826 -0.454650
                                               101.687435
                                                               0.0
                                                                          0.0
                                                               0.0
                                                                          0.0
    2 ENSG00000183878.15 8.566599
                                     2.821199
                                                96.624132
    3 ENSG00000099725.14 7.234079
                                     0.094070
                                                96.796064
                                                               0.0
                                                                          0.0
    4 ENSG00000215580.11 6.602361 -0.667862
                                                               0.0
                                                                          0.0
                                                95.449115
```

```
Coef
                                   Symbol
                                           Entrez Chrom
0 794.024282 Reported_GenderMale
                                   ANOS2P
                                              NaN
                                                      Υ
1 769.944857
              Reported GenderMale
                                   GYG2P1
                                               NaN
                                                      Υ
2 747.659203
              Reported_GenderMale
                                      UTY
                                           7404.0
                                                      Y
              Reported GenderMale
                                     PRKY
                                              NaN
                                                      Y
3 747.089270
4 739.995713 Reported_GenderMale
                                   BCORP1
                                              NaN
                                                      Y
```

1.1.2 NIMH HBCC

```
[3]: hbcc = pd.read_csv('../../_m/nimh_hbcc_maleVfemale.tsv', sep='\t')
hbcc = hbcc[(hbcc['adj.P.Val'] < 0.05)].sort_values('adj.P.Val')
hbcc.head()
```

```
[3]:
                    Geneid
                                logFC
                                       AveExpr
                                                                  P. Value \
    O ENSG00000229807.11 -11.504527
                                      1.451875 -158.907479 9.677013e-244
        ENSG00000241859.7
                            8.165803
                                      0.195418
                                                 98.530405 4.690014e-195
    1
    2 ENSG00000206159.11
                            7.594584 -0.185954
                                                 92.012611 3.727481e-188
    4 ENSG00000215580.11
                            6.980341 -0.607569
                                                 89.120159 6.068671e-185
```

```
3 ENSG00000067646.12 9.872471 2.014148 88.829183 1.293023e-184
```

```
adi.P.Val
                          В
                                            Coef
                                                 Symbol
                                                         Entrez Chrom
0 1.849374e-239
                 541.117577
                             Reported_GenderMale
                                                    XIST
                                                             NaN
1 4.481543e-191 425.604156
                             Reported_GenderMale ANOS2P
                                                             NaN
                                                                    Υ
                             Reported_GenderMale GYG2P1
2 2.374530e-184 411.071479
                                                            NaN
                                                                    Y
4 2.899459e-181 404.120608
                             Reported_GenderMale
                                                                    Y
                                                 BCORP1
                                                            NaN
                             Reported_GenderMale
3 4.942193e-181 404.158301
                                                     ZFY 7544.0
                                                                    Y
```

1.2 DE summary

1.2.1 DE (feature)

```
[4]: gg1 = len(set(mpp['Geneid']))
gg2 = len(set(hbcc['Geneid']))
print("Gene MPP:\t%d\nGene HBCC:\t%d" % (gg1, gg2))
```

Gene MPP: 482 Gene HBCC: 148

1.2.2 Feature effect size summary

```
[5]: feature_list = ['Genes: MPP', 'Genes: HBCC', 'Exons', 'Junctions']
  feature_df = [mpp, hbcc]
  ii = 0

for ii in [0,1]:
    ff = feature_df[ii]
    half = len(set(ff[(np.abs(ff['logFC']) >= 0.5)].Geneid))
    one = len(set(ff[(np.abs(ff['logFC']) >= 1)].Geneid))
    print("\nThere are %d unique %s with abs(log2FC) >= 0.5" % (half, u)
    ofeature_list[ii]))
    print("There are %d unique %s with abs(log2FC) >= 1" % (one, u)
    ofeature_list[ii]))
```

```
There are 36 unique Genes: MPP with abs(log2FC) >= 0.5
There are 25 unique Genes: MPP with abs(log2FC) >= 1

There are 40 unique Genes: HBCC with abs(log2FC) >= 0.5
There are 27 unique Genes: HBCC with abs(log2FC) >= 1
```

1.3 Autosomal only

```
[6]: mpp.Chrom.fillna('?', inplace=True)
      auto1 = mpp[(mpp.Chrom.str.contains('\d+'))].copy()\
              .rename(columns={'Geneid': 'gene_id'})
      auto1 = auto1[['gene_id', 'Chrom', 'Symbol', 'logFC', 'adj.P.Val']]
      print(auto1.shape)
      auto1.head()
     (418, 5)
 [6]:
                     gene_id Chrom
                                        Symbol
                                                   logFC
                                                             adj.P.Val
          ENSG00000205611.4
                                    LINC01597 0.977532 3.625963e-25
      33
                                20
      35 ENSG00000255346.10
                                15
                                          NOX5 0.964489 8.867594e-22
      38
          ENSG00000283443.1
                                20
                                   AC018688.1 0.879612 8.234590e-18
      39
          ENSG00000258484.4
                                15
                                        SPESP1 0.701390 3.003091e-17
      41 ENSG00000149531.15
                                20
                                        FRG1BP 0.528819 6.561622e-13
 [7]: auto1.sort_values('adj.P.Val').to_csv('autosomal_DEG_mpp.csv', index=False,_
       →header=True)
 [8]: hbcc.Chrom.fillna('?', inplace=True)
      auto2 = hbcc[(hbcc.Chrom.str.contains('\d+'))].copy()\
              .rename(columns={'Geneid': 'gene_id'})
      auto2 = auto2[['gene_id', 'Chrom', 'Symbol', 'logFC', 'adj.P.Val']]
      print(auto2.shape)
      auto2.head()
     (98, 5)
 [8]:
                     gene_id Chrom
                                        Symbol
                                                   logFC
                                                             adj.P.Val
           ENSG00000095932.6
                                        SMIM24 -0.813149 9.014157e-15
      35
                                19
      37 ENSG00000149531.15
                                20
                                        FRG1BP 0.739522 6.859297e-12
      41
          ENSG00000283443.1
                                20 AC018688.1 1.149979 2.357916e-09
      42
          ENSG00000205611.4
                                20
                                    LINC01597 1.015293 8.239587e-09
      43
          ENSG00000258484.4
                                15
                                        SPESP1 0.776058 1.221874e-08
 [9]: auto2.sort_values('adj.P.Val').to_csv('autosomal_DEG_hbcc.csv', index=False,__
       →header=True)
     1.4 DE summary
     1.4.1 DE (feature)
[10]: | gg1 = len(set(auto1['gene_id']))
      gg2 = len(set(auto2['gene_id']))
      print("Gene MPP:\t%d\nGene HBCC:\t%d" % (gg1, gg2))
```

Gene MPP: 418
Gene HBCC: 98

1.4.2 Feature effect size summary

```
There are 8 unique Genes: MPP with abs(log2FC) >= 0.5
There are 0 unique Genes: MPP with abs(log2FC) >= 1

There are 12 unique Genes: HBCC with abs(log2FC) >= 0.5
There are 2 unique Genes: HBCC with abs(log2FC) >= 1
```