main

September 24, 2021

1 Examine sample make-up

1.1 Samples after quality control

```
[2]: save_ggplots <- function(p, fn, w, h){
    for(ext in c('.pdf', '.png', '.svg')){
        ggsave(pasteO(fn, ext), plot=p, width=w, height=h)
    }
}</pre>
```

1.1.1 Load Caudate data

1.1.2 Load DLPFC data

1.1.3 Load Hippocampus data

1.1.4 Load DG data

1.1.5 Merge data

1.2 STRUCTURE analysis

```
[8]: ancestry = data.table::fread("../../input/ancestry_structure/structure.

→out_ancestry_proportion_raceDemo_compare")

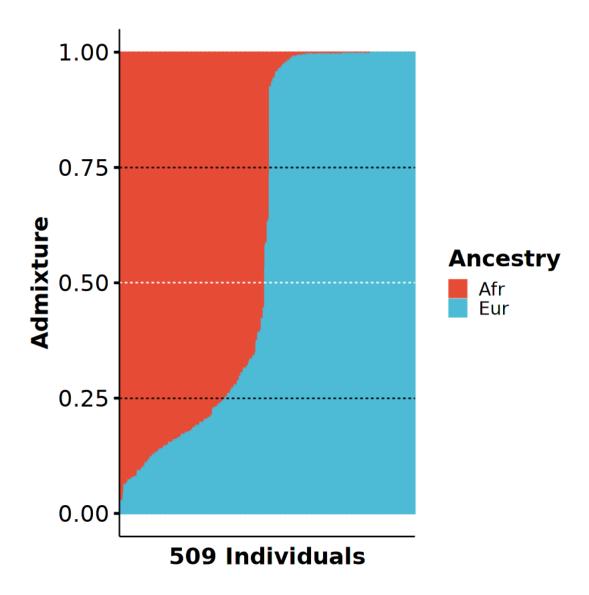
ancestry %>% head(2)
```

```
id
                             Afr
                                      Eur
                                              group
                    <chr>
                             <dbl>
                                      <dbl>
                                              < chr >
A data.table: 2 \times 4
                    Br2374
                             0.007
                                      0.993
                                              CAUC
                    Br1857
                             0.001
                                      0.999
                                              CAUC
```

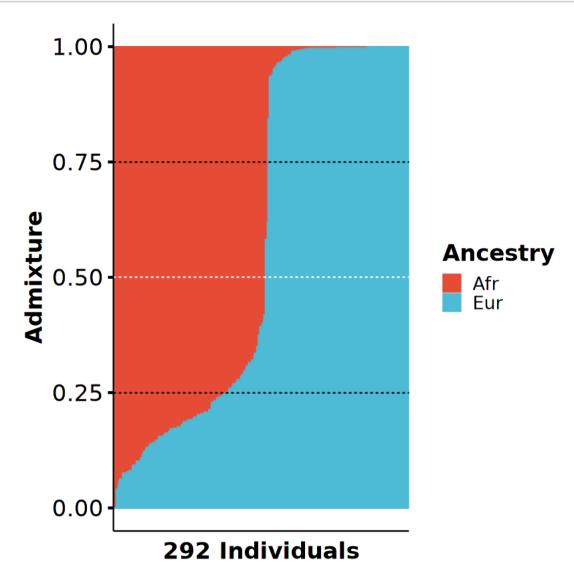
```
[9]: ancestry %>% mutate_if(is.character, as.factor) %>%
    group_by(group) %>% summarize(AA=mean(Afr), EA=mean(Eur))
```

```
AA_mean
                                  AA sd
                                              AA_{max}
                                                        AA_min EA_mean
                                                                               EA sd
                                                                                           EA max
              group
                                                         <dbl>
                                                                               <dbl>
                                                                                           <dbl>
              <fct>
                      <dbl>
                                  <dbl>
                                               < dbl >
                                                                   <dbl>
A tibble: 2 \times 9
              AA
                      0.78962609
                                  0.10611682
                                              0.999
                                                         0.381
                                                                   0.2103739
                                                                               0.10611682
                                                                                           0.619
              CAUC
                      0.03087879
                                  0.02997578
                                              0.156
                                                         0.001
                                                                   0.9691212
                                                                               0.02997578
                                                                                           0.999
```

```
[11]: brp = ancestry %>% inner_join(pheno, by=c("id"="BrNum")) %>%
          select(id, Race, Afr, Eur) %>%
          mutate if(is.character, as.factor) %>% distinct %>%
          pivot_longer(-c("Race", "id"), names_to="Ancestry", values_to="Proportion")
       →%>%
          mutate_if(is.character, as.factor) %>% group_by(Ancestry) %>%
          mutate(ID = fct reorder(id, desc(Proportion))) %>%
          ggbarplot(x="ID", y="Proportion", fill = "Ancestry", color="Ancestry",
                    palette="npg", ylab="Admixture", xlab="509 Individuals",
                    ggtheme=theme_pubr(base_size=20), legend="right") +
          geom_hline(yintercept=0.5, linetype="dashed", color="white") +
          geom_hline(yintercept=0.75, linetype="dashed", color="black") +
          geom_hline(yintercept=0.25, linetype="dashed", color="black") +
          font("xy.title", face="bold") + font("legend.title", face="bold") +
          rremove("x.text") + rremove("x.ticks")
      save_ggplots(brp, "ancestry_structure_barplot_eqtl", 12, 5)
      brp
```



```
font("xy.title", face="bold") + font("legend.title", face="bold") +
    rremove("x.text") + rremove("x.ticks")
save_ggplots(brp, "ancestry_structure_barplot", 12, 5)
brp
```



```
ncol=2, rug=TRUE, add="mean", palette="npg", ylab="Population

→Density",

xlab="Ancestry Proportion", panel.labs.font=list(face='bold'),

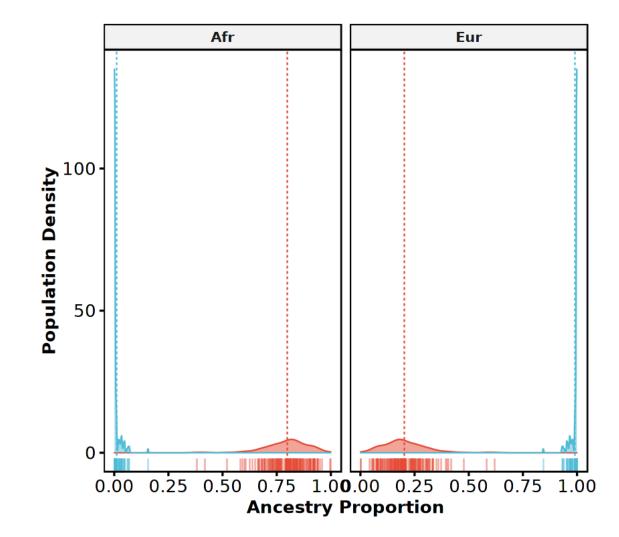
ggtheme=theme_pubr(base_size=15, border=TRUE)) +

font("xy.title", face="bold") + font("legend.title", face="bold")

save_ggplots(bxp, "ancestry_structure_distribution", 10, 5)

bxp
```





1.3 eQTL analysis

```
[14]: pheno %>% dim
```

1. 1334 2. 21

```
[15]: print(paste("There are", unique(pheno$BrNum) %>% length, "unique BrNum."))
     [1] "There are 509 unique BrNum."
[16]: pheno %>% select(BrNum, Region) %>% distinct %>%
          mutate_if(is.character, as.factor) %>%
          group_by(Region) %>% count()
                          Region
                                        n
                          <fct>
                                        <int>
                          Caudate
                                        400
     A grouped df: 4 \times 2
                         DentateGyrus
                                        161
                          DLPFC
                                        378
                          HIPPO
                                        395
[17]: pheno %>% select(BrNum, Race) %>% distinct %>%
          mutate_if(is.character, as.factor) %>%
          group_by(Race) %>% count()
                          Race
                          <fct>
                                 <int>
     A grouped df: 2 \times 2
                          AA
                                 256
                          EA
                                 253
[18]: pheno %>% select(BrNum, Race, Region) %>% distinct %>%
          mutate_if(is.character, as.factor) %>%
          group_by(Region, Race) %>% count()
                          Region
                                        Race
                                               n
                          <fct>
                                        <fct>
                                               <int>
                          Caudate
                                        AA
                                               206
                          Caudate
                                        EA
                                               194
                                               78
                         DentateGyrus
                                        AA
     A grouped_df: 8 \times 3
                          DentateGyrus
                                        EA
                                               83
                          DLPFC
                                        AA
                                               204
                          DLPFC
                                        EA
                                               174
                          HIPPO
                                        AA
                                               213
                          HIPPO
                                        EA
                                               182
[19]: pheno %>% select(BrNum, Sex, Region) %>% distinct %>%
```

mutate_if(is.character, as.factor) %>%

group_by(Region, Sex) %>% count()

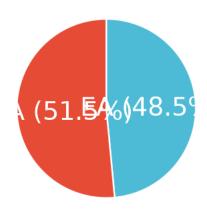
```
Region
                                            Sex
                                                    n
                            <fct>
                                            < fct >
                                                    <int>
                            Caudate
                                            \overline{\mathrm{F}}
                                                    126
                            Caudate
                                            Μ
                                                    274
                            DentateGyrus
                                           F
                                                    48
      A grouped df: 8 \times 3
                            DentateGyrus
                                           Μ
                                                    113
                            DLPFC
                                            \mathbf{F}
                                                    121
                            DLPFC
                                            Μ
                                                    257
                            HIPPO
                                            \mathbf{F}
                                                    126
                            HIPPO
                                            Μ
                                                    269
[20]: pheno %>% group_by(Region) %>%
         summarise_at(vars(c("Age")), list(mean = mean, sd = sd))
                      Region
                                      mean
                                                \operatorname{sd}
                      <chr>
                                      < dbl >
                                                 <dbl>
                      Caudate
                                      49.12390
                                                16.05379
      A tibble: 4 \times 3
                      DentateGyrus
                                      50.06770
                                                15.43849
                      DLPFC
                                      45.83574
                                                16.49445
                      HIPPO
                                      45.49527 \quad 16.41527
[21]: pheno %>% group_by(Region, Race) %>%
         summarise_at(vars(c("Age")), list(mean = mean, sd = sd))
                            Region
                                            Race
                                                    mean
                                                               \operatorname{sd}
                            <chr>
                                            <chr>
                                                     <dbl>
                                                               <dbl>
                            Caudate
                                                               14.49676
                                            AA
                                                    48.81325
                            Caudate
                                            EA
                                                    49.45376
                                                               17.58900
                            DentateGyrus
                                            AA
                                                    50.18423
                                                               15.53374
      A grouped df: 8 \times 4
                            DentateGyrus
                                           EA
                                                               15.44210
                                                    49.95819
                            DLPFC
                                            AA
                                                    46.97896
                                                               15.34261
                            DLPFC
                                            EA
                                                    44.49542
                                                               17.70090
                            HIPPO
                                            AA
                                                    46.34080
                                                               15.61922
                            HIPPO
                                            EA
                                                    44.50571
                                                               17.29140
[22]: pheno %>% filter(RIN != "NA") %>% mutate("RIN"=as.numeric(unlist(RIN))) %>%
           group_by(Region) %>% summarise_at(vars(c("RIN")), list(mean = mean, sd =__
        sd))
                      Region
                                      mean
                                                \operatorname{sd}
                      <chr>
                                      <dbl>
                                                 <dbl>
                      Caudate
                                      7.861000
                                                0.8648983
      A tibble: 4 \times 3
                      DentateGyrus
                                      5.208403
                                                1.1871187
                      DLPFC
                                      7.699471
                                                0.9340876
                      HIPPO
                                      7.616962
                                                1.0311104
[23]: pheno %>% filter(RIN != "NA") %>% mutate("RIN"=as.numeric(unlist(RIN))) %>%
           group_by(Region, Race) %>% summarise_at(vars(c("RIN")), list(mean = mean,__
        \rightarrowsd = sd))
```

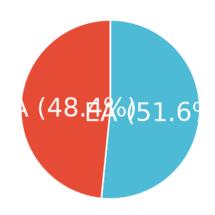
```
Region
                                      Race
                                               mean
                                                         \operatorname{sd}
                      <chr>
                                      <chr>
                                                         <dbl>
                                               <dbl>
                      Caudate
                                      \overline{AA}
                                               7.859709
                                                         0.8416464
                      Caudate
                                      EA
                                               7.862371
                                                         0.8911055
                      DentateGyrus
                                      AA
                                                         1.2062837
                                               5.206349
A grouped df: 8 \times 4
                      DentateGyrus
                                     EA
                                              5.210714
                                                         1.1760765
                      DLPFC
                                      AA
                                              7.678922
                                                         0.9445184
                      DLPFC
                                      EA
                                               7.723563
                                                         0.9238440
                      HIPPO
                                      AA
                                               7.604225
                                                         1.0509344
                                      EA
                      HIPPO
                                               7.631868
                                                         1.0101014
```

1.3.1 Pie chart

Caudate

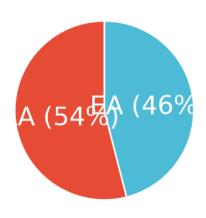
Dentate Gyrus

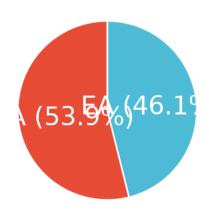




DLPFC

Hippocampus





1.4 Adult individuals for expression related analysis

```
[26]: pheno = pheno %>% filter(Age > 17, Dx == "Control")
pheno %>% dim
```

1. 785 2. 21

[27]: print(paste("There are", unique(pheno\$BrNum) %>% length, "unique BrNum."))

[1] "There are 292 unique BrNum."

```
Region
                                           n
                            <fct>
                                            <int>
                            Caudate
                                           240
      A grouped df: 4 \times 2
                            DentateGyrus
                                           90
                            DLPFC
                                           212
                            HIPPO
                                           243
[29]: pheno %>% select(BrNum, Race) %>% distinct %>%
           mutate_if(is.character, as.factor) %>%
           group_by(Race) %>% count()
                            Race
                                    \mathbf{n}
                            <fct>
                                    <int>
      A grouped_df: 2 \times 2
                            \overline{AA}
                                    151
                            EA
                                    141
[30]: pheno %>% select(BrNum, Race, Region) %>% distinct %>%
           mutate if(is.character, as.factor) %>%
           group_by(Region, Race) %>% count()
                            Region
                                           Race
                                                   n
                            <fct>
                                            < tct >
                                                    <int>
                            Caudate
                                           \overline{AA}
                                                    122
                            Caudate
                                           EA
                                                   118
                            DentateGyrus
                                           AA
                                                   47
      A grouped df: 8 \times 3
                            DentateGyrus
                                           EA
                                                   43
                            DLPFC
                                           AA
                                                   123
                            DLPFC
                                           EA
                                                   89
                            HIPPO
                                           AA
                                                   133
                            HIPPO
                                           EA
                                                   110
[31]: pheno %>% select(BrNum, Sex, Region) %>% distinct %>%
           mutate_if(is.character, as.factor) %>%
           group_by(Region, Sex) %>% count()
                            Region
                                           Sex
                                                   \mathbf{n}
                            <fct>
                                            < fct >
                                                   <int>
                            Caudate
                                           \overline{\mathbf{F}}
                                                    71
                            Caudate
                                           Μ
                                                    169
                            DentateGyrus
                                           F
                                                    26
      A grouped df: 8 \times 3
                            DentateGyrus
                                           Μ
                                                    64
                            DLPFC
                                           F
                                                    66
                            DLPFC
                                           Μ
                                                   146
                            HIPPO
                                           F
                                                    74
                            HIPPO
                                           Μ
                                                   169
[32]: pheno %>% group_by(Region) %>%
```

summarise_at(vars(c("Age")), list(mean = mean, sd = sd))

```
<chr>
                                     < dbl >
                                                <dbl>
                      Caudate
                                     48.31150
                                               \overline{15.84692}
      A tibble: 4 \times 3
                      DentateGyrus
                                     47.88311
                                               15.02380
                      DLPFC
                                     45.16991
                                               14.76717
                      HIPPO
                                     44.56724
                                               14.73045
[33]: pheno %>% group_by(Region, Race) %>%
         summarise_at(vars(c("Age")), list(mean = mean, sd = sd))
                           Region
                                           Race
                                                   mean
                                                              \operatorname{sd}
                           <chr>
                                                              <dbl>
                                           <chr>
                                                   <dbl>
                           Caudate
                                           AA
                                                   45.63770
                                                              14.72979
                           Caudate
                                           EA
                                                   51.07593
                                                              16.53588
                           DentateGyrus
                                           AA
                                                   45.85043
                                                              16.32827
      A grouped df: 8 \times 4
                           DentateGyrus
                                           EA
                                                   50.10488
                                                              13.28980
                           DLPFC
                                           AA
                                                   44.12511
                                                              14.97092
                           DLPFC
                                           EA
                                                   46.61386
                                                              14.43996
                           HIPPO
                                           AA
                                                   43.30015
                                                              14.73609
                           HIPPO
                                           EA
                                                   46.09927
                                                              14.64404
[34]: pheno %>% filter(RIN != "NA") %>% mutate("RIN"=as.numeric(unlist(RIN))) %>%
           group_by(Region) %>% summarise_at(vars(c("RIN")), list(mean = mean, sd =__
        ⇒sd))
                      Region
                                     mean
                                               \operatorname{sd}
                      <chr>
                                     <dbl>
                                                <dbl>
                      Caudate
                                     7.850000
                                               0.7956997
      A tibble: 4 \times 3
                      DentateGyrus
                                     5.315152
                                               1.2186048
                      DLPFC
                                     7.699057
                                               0.8803807
                      HIPPO
                                     7.735391
                                               0.9668378
[35]: pheno %>% filter(RIN != "NA") %>% mutate("RIN"=as.numeric(unlist(RIN))) %>%
           group_by(Region, Race) %>% summarise_at(vars(c("RIN")), list(mean = mean,__
       \rightarrowsd = sd))
                           Region
                                           Race
                                                              \operatorname{sd}
                                                   mean
                                                              <dbl>
                            <chr>
                                           <chr>
                                                   <dbl>
                           Caudate
                                           AA
                                                              0.7993477
                                                   7.829508
                           Caudate
                                           EA
                                                   7.871186
                                                              0.7947587
                           DentateGyrus
                                           AA
                                                   5.447368
                                                              1.2173824
      A grouped df: 8 \times 4
                           DentateGyrus
                                           EA
                                                   5.135714
                                                              1.2190507
                           DLPFC
                                           AA
                                                   7.696748
                                                              0.8851169
                           DLPFC
                                           EA
                                                   7.702247
                                                              0.8787876
                           HIPPO
                                           AA
                                                   7.715038
                                                              0.9754173
                                           EA
                           HIPPO
                                                   7.760000
                                                              0.9602370
[36]: ## Get and annotate plot
      cc_pie = annotate_figure(plot_pie("Caudate"),
```

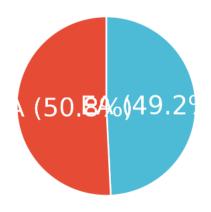
Region

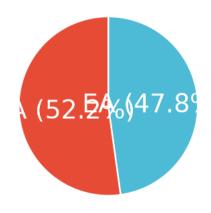
mean

 sd

Caudate

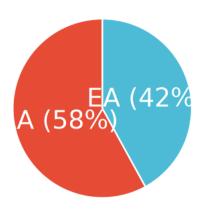
Dentate Gyrus

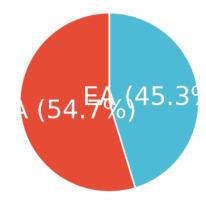




DLPFC

Hippocampus





1.5 Reproducibility Information

```
[37]: Sys.time()
      proc.time()
      options(width = 120)
      sessioninfo::session_info()
     [1] "2021-09-24 13:23:00 EDT"
        user system elapsed
      26.731
                1.054 28.316
       Session info
      setting value
      version R version 4.0.3 (2020-10-10)
      os
               Arch Linux
               x86_64, linux-gnu
      system
      ui
               X11
      language (EN)
               en_US.UTF-8
      collate
      ctype
               en_US.UTF-8
               America/New_York
      tz
      date
               2021-09-24
       Packages
      package
                            * version date
                                                   lib source
      abind
                              1.4-5
                                       2016-07-21 [1] CRAN (R 4.0.2)
                              0.2.1
                                       2019-03-21 [1] CRAN (R 4.0.2)
      assertthat
      backports
                              1.2.1
                                       2020-12-09 [1] CRAN (R 4.0.2)
      base64enc
                              0.1-3
                                       2015-07-28 [1] CRAN (R 4.0.2)
      Biobase
                            * 2.50.0
                                       2020-10-27 [1] Bioconductor
      BiocGenerics
                            * 0.36.1
                                       2021-04-16 [1] Bioconductor
                              1.0 - 7
                                       2021-04-24 [1] CRAN (R 4.0.3)
      bitops
      broom
                              0.7.9
                                       2021-07-27 [1] CRAN (R 4.0.3)
                              1.5-12.2 2020-07-07 [1] CRAN (R 4.0.2)
      Cairo
                                       2021-06-27 [1] CRAN (R 4.0.3)
      car
                              3.0-11
      carData
                              3.0-4
                                       2020-05-22 [1] CRAN (R 4.0.2)
                                       2016-07-27 [1] CRAN (R 4.0.2)
      cellranger
                              1.1.0
      cli
                              3.0.1
                                       2021-07-17 [1] CRAN (R 4.0.3)
                              2.0-2
                                       2021-06-24 [1] CRAN (R 4.0.3)
      colorspace
                                       2020-12-30 [1] CRAN (R 4.0.2)
      cowplot
                              1.1.1
      crayon
                              1.4.1
                                       2021-02-08 [1] CRAN (R 4.0.3)
                              4.3.2
                                       2021-06-23 [1] CRAN (R 4.0.3)
      curl
      data.table
                              1.14.0
                                       2021-02-21 [1] CRAN (R 4.0.3)
                              1.1.1
      DBI
                                       2021-01-15 [1] CRAN (R 4.0.2)
      dbplyr
                              2.1.1
                                       2021-04-06 [1] CRAN (R 4.0.3)
      DelayedArray
                              0.16.3
                                       2021-03-24 [1] Bioconductor
                              0.6.27
                                       2020-10-24 [1] CRAN (R 4.0.2)
      digest
                                       2021-06-18 [1] CRAN (R 4.0.3)
      dplyr
                            * 1.0.7
```

| ellipsis | | 0.3.2 | 2021-04-29 | | CRAN (R 4.0.3) |
|------------------|---|---------|------------|-------|--------------------|
| evaluate | | 0.14 | 2019-05-28 | [1] | CRAN (R 4.0.2) |
| fansi | | 0.5.0 | 2021-05-25 | [1] | CRAN (R 4.0.3) |
| farver | | 2.1.0 | 2021-02-28 | [1] | CRAN (R 4.0.3) |
| fastmap | | 1.1.0 | 2021-01-25 | [1] | CRAN (R 4.0.2) |
| forcats | * | 0.5.1 | 2021-01-27 | [1] | CRAN (R 4.0.2) |
| foreign | | 0.8-80 | 2020-05-24 | | CRAN (R 4.0.3) |
| fs | | 1.5.0 | 2020-07-31 | [1] | CRAN (R 4.0.2) |
| generics | | 0.1.0 | 2020-10-31 | [1] | CRAN (R 4.0.2) |
| GenomeInfoDb | * | 1.26.7 | 2021-04-08 | [1] | Bioconductor |
| GenomeInfoDbData | | 1.2.4 | 2021-02-02 | [1] | Bioconductor |
| GenomicRanges | * | 1.42.0 | 2020-10-27 | [1] | Bioconductor |
| ggplot2 | * | 3.3.5 | 2021-06-25 | [1] | CRAN (R 4.0.3) |
| ggpubr | * | 0.4.0 | 2020-06-27 | [1] | CRAN (R 4.0.2) |
| ggsci | | 2.9 | 2018-05-14 | [1] | CRAN (R 4.0.2) |
| ggsignif | | 0.6.2 | 2021-06-14 | [1] | CRAN (R 4.0.3) |
| glue | | 1.4.2 | 2020-08-27 | [1] | CRAN (R 4.0.2) |
| gridExtra | | 2.3 | 2017-09-09 | [1] | CRAN (R 4.0.2) |
| gtable | | 0.3.0 | 2019-03-25 | [1] | CRAN (R 4.0.2) |
| haven | | 2.4.3 | 2021-08-04 | [1] | CRAN (R 4.0.3) |
| hms | | 1.1.0 | 2021-05-17 | [1] | CRAN (R 4.0.3) |
| htmltools | | 0.5.2 | 2021-08-25 | [1] | CRAN (R 4.0.3) |
| httr | | 1.4.2 | 2020-07-20 | [1] | CRAN (R 4.0.2) |
| IRanges | * | 2.24.1 | 2020-12-12 | [1] | Bioconductor |
| IRdisplay | | 1.0 | 2021-01-20 | [1] | CRAN (R 4.0.2) |
| IRkernel | | 1.2 | 2021-05-11 | [1] | CRAN (R 4.0.3) |
| jsonlite | | 1.7.2 | 2020-12-09 | [1] | CRAN (R 4.0.2) |
| labeling | | 0.4.2 | 2020-10-20 | [1] | CRAN (R 4.0.2) |
| lattice | | 0.20-41 | 2020-04-02 | [2] | CRAN (R 4.0.3) |
| lifecycle | | 1.0.0 | 2021-02-15 | [1] | CRAN (R 4.0.3) |
| lubridate | | 1.7.10 | 2021-02-26 | | CRAN (R 4.0.3) |
| magrittr | | 2.0.1 | 2020-11-17 | | CRAN (R 4.0.2) |
| Matrix | | 1.3-4 | 2021-06-01 | [1] | CRAN (R 4.0.3) |
| MatrixGenerics | * | 1.2.1 | | | Bioconductor |
| matrixStats | | 0.60.1 | | | CRAN (R 4.0.3) |
| modelr | | 0.1.8 | | | CRAN (R 4.0.2) |
| munsell | | 0.5.0 | 2018-06-12 | | CRAN (R 4.0.2) |
| openxlsx | | 4.2.4 | 2021-06-16 | [1] | CRAN (R 4.0.3) |
| pbdZMQ | | 0.3-5 | 2021-02-10 | | CRAN (R 4.0.3) |
| pillar | | 1.6.2 | 2021-07-29 | | |
| pkgconfig | | 2.0.3 | 2019-09-22 | | CRAN (R 4.0.2) |
| purrr | * | 0.3.4 | 2020-04-17 | | CRAN (R 4.0.2) |
| R6 | • | 2.5.1 | 2021-08-19 | | |
| Rcpp | | 1.0.7 | 2021-07-07 | | |
| RCurl | | | 2021-08-17 | | CRAN (R 4.0.3) |
| readr | * | 2.0.1 | 2021-08-10 | [1] | CRAN (R 4.0.3) |
| readxl | • | 1.3.1 | 2019-03-13 | | |
| repr | | 1.1.3 | | | CRAN (R 4.0.2) |
| - °F- | | 1.1.0 | 2021 01 21 | L + J | 0101111 (10 1.0.2) |

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2.0.1
                                 2021-08-05 [1] CRAN (R 4.0.3)
reprex
                       0.5.27
                                 2021-06-21 [1] CRAN (R 4.0.3)
rio
                                 2021-04-30 [1] CRAN (R 4.0.3)
                       0.4.11
rlang
                       0.7.0
                                 2021-02-13 [1] CRAN (R 4.0.3)
rstatix
                       0.13
                                 2020-11-12 [1] CRAN (R 4.0.2)
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S4Vectors
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                                 2020-05-11 [1] CRAN (R 4.0.2)
scales
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sessioninfo
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                                 2021-08-25 [1] CRAN (R 4.0.3)
stringi
                       1.7.4
                     * 1.4.0
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SummarizedExperiment * 1.20.0
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svglite
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                                 2021-05-11 [1] CRAN (R 4.0.3)
                       1.0.2
                     * 3.1.4
                                 2021-08-25 [1] CRAN (R 4.0.3)
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                                 2021-04-30 [1] CRAN (R 4.0.3)
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tidyverse
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utf8
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                                 2020-02-26 [1] CRAN (R 4.0.2)
uuid
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xml2
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XVector
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zip
                                 2020-10-27 [1] Bioconductor
zlibbioc
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^{[1] /}home/jbenja13/R/x86_64-pc-linux-gnu-library/4.0

^{[2] /}usr/lib/R/library