main

July 12, 2021

1 Quality control comparison between LIBD and public RNA-seq data

- CMC
- GTEx brain regions

```
[1]: library(tidyverse)
     library(synapser)
     library(ggpubr)
      Attaching packages
                                               tidyverse
    1.3.1
     ggplot2 3.3.5
                         purrr
                                 0.3.4
     tibble 3.1.2
                         dplyr
                                 1.0.7
     tidyr 1.1.3
                         stringr 1.4.0
     readr
             1.4.0
                         forcats 0.5.1
      Conflicts
    tidyverse_conflicts()
     dplyr::filter() masks stats::filter()
     dplyr::lag()
                     masks stats::lag()
    New synapser version detected:
        You are using synapser version 0.9.77.
        synapser version 0.10.101 is detected at http://ran.synapse.org.
        To upgrade to the latest version of synapser, please run the following
```

TERMS OF USE NOTICE:

When using Synapse, remember that the terms and conditions of use require that you:

1) Attribute data contributors when discussing these data or results from these data.

install.packages("synapser", repos="http://ran.synapse.org")

- 2) Not discriminate, identify, or recontact individuals or groups represented by the data.
 - 3) Use and contribute only data de-identified to HIPAA standards.
 - 4) Redistribute data only under these same terms of use.

1.1 Load metrics

1.1.1 Load Lieber RNA-seq metrics

A data.table: 2×14	SampleID	Sex	Race	Dx	Age	mitoRate	$rRNA_Rate$	totalAssignedO
	<chr $>$	<chr $>$	<chr $>$	<chr $>$	<dbl $>$	<dbl $>$	<dbl $>$	<dbl></dbl>
	R11135	Male	EA	CTL	18.77	0.2572796	0.0001690954	0.5231321
	R11137	Male	EA	CTL	41.44	0.3840272	0.0000884558	0.5933431

1.1.2 Load CMC RNA-seq metrics

```
[3]: # Process functions
    downloadFile_version <- function(id , version){</pre>
      data.table::fread(synGet(id, version = version)$path, data.table = F)
    synLogin()
     # Download clinical metadata
    CLINICAL ID = 'syn3354385'
    clinical = downloadFile_version(CLINICAL_ID, version = 4)
    # Download RNASeq metadata
    METADATA_QC_DLPFC_ID = 'syn18358379'
    metadata = downloadFile_version(METADATA_QC_DLPFC_ID, version = 3)
    # Join clinical and RNASeg metadata
    md = right_join(clinical, metadata, by = c("Individual ID" = "Individual_ID"))__
     ,>%
        mutate(Dx = fct_recode(Dx, AFF_BP = "BP", AFF_BP = "AFF", Other =__
     Control = "Control", SCZ = "SCZ"))
     # Compute read pair metrics and add Institution-Dx variable
```

Welcome, kj.benjamin!

NULL

[1] 991 13

		SampleID	Dx	$Reported_Gender$	Sex	Ethnicity
A data.frame: $2 \times 13 \frac{1}{1}$		<chr></chr>	<fct $>$	<chr $>$	<chr $>$	<chr $>$
	1	MSSM_RNA_PFC_155	Control	Female	XX	African-American
	2	MSSM_RNA_PFC_280	Control	Female	XX	Caucasian

1.1.3 Load GTEx metrics and

```
SampleID
                                                     RIN
                                                             SMTS
                                                                     Brain Region
                  <chr>
                                                     <dbl>
                                                             <chr>
                                                                     <chr>
                  GTEX-1117F-0011-R10a-SM-AHZ7F
                                                     NA
                                                             Brain
                                                                     Brain - Frontal Cortex (BA9)
                                                                     Brain - Frontal Cortex (BA9)
                  GTEX-1117F-0011-R10b-SM-CYKQ8
                                                     7.2
                                                             Brain
A data.table: 6 \times 6
                  GTEX-1117F-3226-SM-5N9CT
                                                     6.2
                                                             Brain
                                                                     Brain - Cortex
                  GTEX-111FC-0011-R10a-SM-AHZ7K
                                                     NA
                                                             Brain
                                                                     Brain - Frontal Cortex (BA9)
                                                                     Brain - Frontal Cortex (BA9)
                  GTEX-111FC-0011-R10a-SM-CYKQ9
                                                     8.5
                                                             Brain
                  GTEX-111FC-3126-SM-5GZZ2
                                                                     Brain - Cortex
                                                     6.1
                                                             Brain
```

Ρ

N

0

1.2 Clean and organize data

```
gtex$Brain_Region = gsub(" $", "", str_replace(gtex$Brain_Region, "Frontal_\( \to \text{Cortex}", "DLPFC")) gtex %>% head(2)
```

```
RIN
                                                                                   rRNA Rate
                 SampleID
                                                    Dx
                                                             Brain Region
                 <chr>
                                                    <chr>
                                                             <chr>
                                                                            <dbl>
                                                                                   <dbl>
A data.table: 2 \times 7
                 GTEX-1117F-0011-R10a-SM-AHZ7F
                                                    Control
                                                             DLPFC
                                                                           NA
                                                                                   NA
                 GTEX-1117F-0011-R10b-SM-CYKQ8
                                                    Control
                                                             DLPFC
                                                                           7.2
                                                                                   NA
```

```
[6]: cmc = md %>% select("SampleID", "Dx", "Brain_Region", "RIN", "rRNA_Rate", □

→"Percent_Aligned") %>%

mutate("Dataset"="CMC")

cmc %>% head(2)
```

```
SampleID
                                              Dx
                                                       Brain Region
                                                                     RIN
                                                                             rRNA Rate
                                                                                          Percent .
                                              <fct>
                                                       < chr >
                                                                     <dbl>
                                                                             <dbl>
                                                                                           <dbl>
A data.frame: 2 \times 7
                     MSSM_RNA_PFC_155
                                                       DLPFC
                                                                     8.4
                                                                             0.000250992
                                              Control
                                                                                          0.9798915
                  2 MSSM RNA PFC 280
                                                      DLPFC
                                                                     8.4
                                                                             0.000373524
                                              Control
                                                                                          0.9688145
```

```
[7]: libd = libd %>% select("SampleID", "Dx", "Brain_Region", "RIN", "rRNA_Rate", □

→ "Percent_Aligned") %>%

mutate("Dataset"="LIBD", "Dx"=gsub("CTL", "Control", libd$Dx),

"Brain_Region"=gsub("HIPPO", "Hippocampus", libd$Brain_Region))

libd %>% head(2)
```

A data.table: 2×7	SampleID	Dx	Brain_Region	RIN	$rRNA_Rate$	Percent_Aligned	Dataset
	<chr $>$	<chr $>$	<chr $>$	<dbl $>$	<dbl $>$	<dbl></dbl>	<chr $>$
	R11135	Control	Hippocampus	5.9	0.0001690954	0.8746	LIBD
	R11137	Control	Hippocampus	9.2	0.0000884558	0.9149	LIBD

```
[8]: df = bind_rows(libd, gtex, cmc) #%>% mutate_if(is.character, as.factor) df %>% head(2)
```

pleID Dx	Brain_Region	RIN	$rRNA_Rate$	Percent_Aligned	Dataset
r> <chr></chr>	<chr $>$	<dbl $>$	<dbl $>$	<dbl></dbl>	<chr $>$
135 Control	Hippocampus	5.9	0.0001690954	0.8746	LIBD
137 Control	Hippocampus	9.2	0.0000884558	0.9149	LIBD
]	135 Control	r> <chr> <chr> <chr> Hippocampus</chr></chr></chr>	r>	r>	r>

[9]: df\$Brain_Region %>% unique

- 1. 'Hippocampus' 2. 'DLPFC' 3. 'Caudate' 4. 'Cortex' 5. 'Cerebellum' 6. 'Cerebellar Hemisphere' 7. 'Nucleus accumbens' 8. 'Putamen' 9. 'Hypothalamus' 10. 'Spinal cord' 11. 'Anterior cingulate cortex' 12. 'Substantia nigra' 13. 'Amygdala'
- 1.3 Descriptive analysis
- 1.3.1 Check if NA present

GTEx

RIN

```
[10]: gtex %>% group_by(Brain_Region) %>% count(is.na(RIN)) %>%
    pivot_wider(names_from=`is.na(RIN)`, values_from="n") %>%
    mutate("TRUE"=replace_na(`TRUE`,0))
```

	Brain_Region	FALSE	TRUE
	<chr></chr>	<int $>$	<dbl $>$
-	Amygdala	177	0
	Anterior cingulate cortex	213	0
	Caudate	291	0
	Cerebellar Hemisphere	263	0
	Cerebellum	297	1
A grouped_df: 13×3	Cortex	325	0
	DLPFC	303	122
	Hippocampus	232	11
	Hypothalamus	236	0
	Nucleus accumbens	277	0
	Putamen	232	0
	Spinal cord	182	0
	Substantia nigra	164	0

rRNA Rate

```
[11]: gtex %>% group_by(Brain_Region) %>% count(is.na(rRNA_Rate)) %>%
    pivot_wider(names_from=`is.na(rRNA_Rate)`, values_from="n") %>%
    mutate("TRUE"=replace_na(`TRUE`,0))
```

	Brain_Region <chr></chr>	FALSE <int></int>	TRUE <dbl></dbl>
	Amygdala	161	16
	Anterior cingulate cortex	199	14
	Caudate	269	22
	Cerebellar Hemisphere	245	18
	Cerebellum	275	23
A grouped_df: 13×3	Cortex	280	45
	DLPFC	221	204
	Hippocampus	214	29
	Hypothalamus	219	17
	Nucleus accumbens	258	19
	Putamen	219	13
	Spinal cord	171	11
	Substantia nigra	150	14

Percent Alignment

```
[12]: gtex %>% group_by(Brain_Region) %>% count(is.na(Percent_Aligned)) %>%
    pivot_wider(names_from=`is.na(Percent_Aligned)`, values_from="n") %>%
    mutate("TRUE"=replace_na(`TRUE`,0))
```

	Brain_Region	FALSE	TRUE
	<chr></chr>	<int $>$	<d b l $>$
	Amygdala	161	16
	Anterior cingulate cortex	199	14
	Caudate	269	22
	Cerebellar Hemisphere	245	18
	Cerebellum	275	23
A grouped_df: 13×3	Cortex	280	45
	DLPFC	221	204
	Hippocampus	214	29
	Hypothalamus	219	17
	Nucleus accumbens	258	19
	Putamen	219	13
	Spinal cord	171	11
	Substantia nigra	150	14

CMC

RIN

[13]: cmc %>% group_by(Brain_Region) %>% count(is.na(RIN))

rRNA Rate

[14]: cmc %>% group_by(Brain_Region) %>% count(is.na(rRNA_Rate))

Percent Alignment

[15]: cmc %>% group_by(Brain_Region) %>% count(is.na(Percent_Aligned))

	Brain_Region	is.na(Percent_Aligned)	\mathbf{n}
A grouped_df: 1×3	<chr $>$	<lgl $>$	<int $>$
	DLPFC	FALSE	991

LIBD

RIN

[16]: libd %>% group_by(Brain_Region) %>% count(is.na(RIN))

rRNA Rate

[17]: libd %>% group_by(Brain_Region) %>% count(is.na(rRNA_Rate))

	Brain_Region	$is.na(rRNA_Rate)$	\mathbf{n}
	<chr $>$	<lgl $>$	<int $>$
A grouped_df: 3×3	Caudate	FALSE	464
	DLPFC	FALSE	453
	Hippocampus	FALSE	447

Percent Alignment

[18]: libd %>% group_by(Brain_Region) %>% count(is.na(Percent_Aligned))

	Brain_Region	$is.na(Percent_Aligned)$	n
	<chr $>$	<lgl $>$	<int $>$
A grouped_df: 3×3	Caudate	FALSE	464
	DLPFC	FALSE	453
	Hippocampus	FALSE	447

1.3.2 Summarize data

[`]summarise()` has grouped output by 'Dataset'. You can override using the `.groups` argument.

	Dataset	Brain_Region	RIN_mean	RIN_sd	$rRNA_mean$	${\rm rRNA_sd}$
	<chr $>$	<chr $>$	<dbl $>$	<dbl $>$	<dbl></dbl>	<dbl $>$
•	CMC	DLPFC	7.595863	0.9002318	2.825332e-04	1.016402e-
	GTEx	Amygdala	6.594915	0.6356163	8.679170 e-02	3.724921e
	GTEx	Anterior cingulate cortex	6.683568	0.7626681	7.020330e-02	3.407544e
	GTEx	Caudate	7.390034	0.8786767	6.669918e-02	2.858002e-
	GTEx	Cerebellar Hemisphere	7.457034	1.0033617	2.524327e-02	2.178709e-
	GTEx	Cerebellum	6.851852	0.7893323	2.060865 e-02	1.212267e-
	GTEx	Cortex	6.588308	0.6811292	4.852958e-02	2.031282e-
A grouped_df: 17×8	GTEx	DLPFC	7.168647	0.8580205	5.299829 e-02	2.901991e-
	GTEx	Hippocampus	6.765517	0.7559981	7.530126e-02	3.385451e
	GTEx	Hypothalamus	6.855085	0.7763085	6.479683e-02	3.499553e-
	GTEx	Nucleus accumbens	7.140433	0.8426733	7.279581e-02	3.489604e-
	GTEx	Putamen	7.085776	0.9074560	7.632170e-02	2.873567e-
	GTEx	Spinal cord	7.063187	0.7335050	4.625480 e - 02	2.817331e
	GTEx	Substantia nigra	6.578659	0.7163100	7.752818e-02	3.039102e
	LIBD	Caudate	7.834483	0.8673440	5.759414e-05	3.599073e-
	LIBD	DLPFC	7.779691	0.9663740	5.022716e-05	3.060435e
	LIBD	Hippocampus	7.723490	1.1208014	2.929714e-05	2.628273e-

1.4 Plot metrics

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

Warning message:

Warning message:

Warning message:

Warning message:

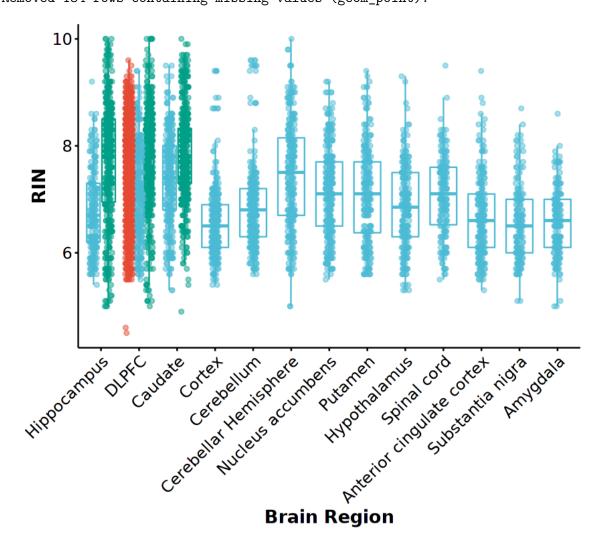
"Removed 134 rows containing missing values (geom_point)." Warning message:

[&]quot;Removed 134 rows containing non-finite values (stat_boxplot)."

[&]quot;Removed 134 rows containing missing values (geom_point)."

[&]quot;Removed 134 rows containing non-finite values (stat_boxplot)."

```
"Removed 134 rows containing non-finite values (stat_boxplot)."
Warning message:
"Removed 134 rows containing missing values (geom_point)."
Warning message:
"Removed 134 rows containing non-finite values (stat_boxplot)."
Warning message:
"Removed 134 rows containing missing values (geom_point)."
```



Dataset

CMC

GTEx

LIBD

```
outlier.shape=NA, ylab='rRNA Rate', add.

→params=list(alpha=0.5),

legend="bottom", xlab="Brain Region") +

font("xy.text", size=14) + font("xy.title", size=16, face="bold") +

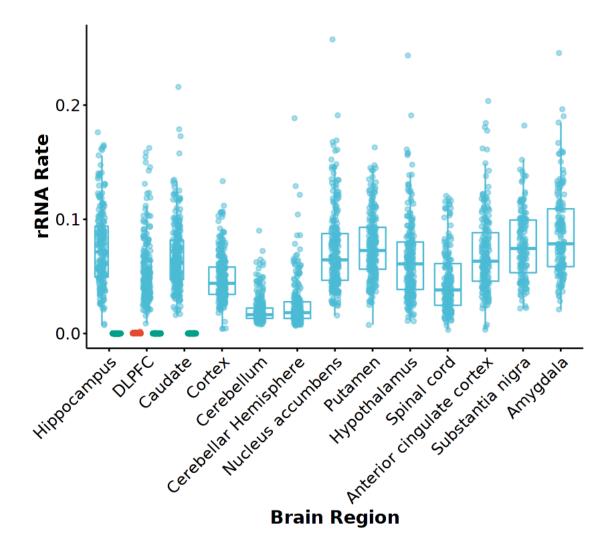
font("legend.title", size=18, face="bold") + font("legend.text", size=18) +

rotate_x_text(45)

save_ggplots("boxplot_brain_region_comparison_rRNA_rate", bxp, 12, 8)

bxp
```

```
Warning message:
"Removed 445 rows containing non-finite values (stat_boxplot)."
Warning message:
"Removed 445 rows containing missing values (geom_point)."
Warning message:
"Removed 445 rows containing non-finite values (stat_boxplot)."
Warning message:
"Removed 445 rows containing missing values (geom_point)."
Warning message:
"Removed 445 rows containing non-finite values (stat_boxplot)."
Warning message:
"Removed 445 rows containing missing values (geom_point)."
Warning message:
"Removed 445 rows containing non-finite values (stat_boxplot)."
Warning message:
"Removed 445 rows containing missing values (geom_point)."
```



Dataset

CMC

GTEx

LIBD

Warning message:

"Removed 445 rows containing non-finite values (stat_boxplot)."

Warning message:

"Removed 445 rows containing missing values (geom_point)."

Warning message:

"Removed 445 rows containing non-finite values (stat_boxplot)." Warning message:

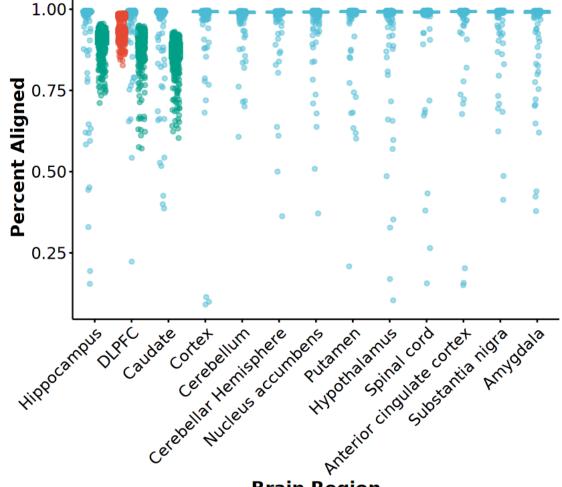
"Removed 445 rows containing missing values (geom_point)." Warning message:

"Removed 445 rows containing non-finite values (stat_boxplot)." Warning message:

"Removed 445 rows containing missing values (geom_point)." Warning message:

"Removed 445 rows containing non-finite values (stat_boxplot)." Warning message:

"Removed 445 rows containing missing values (geom_point)."



Brain Region

Dataset

CMC

GTEx

LIBD

1.5 Session Info

[24]: Sys.time()

```
proc.time()
options(width = 120)
sessioninfo::session_info()
[1] "2021-07-12 15:50:21 EDT"
         system elapsed
  user
17.106
          0.769
                15.800
 Session info
setting value
version
          R version 4.0.3 (2020-10-10)
          Arch Linux
os
system
          x86_64, linux-gnu
ui
language (EN)
          en_US.UTF-8
collate
ctype
          en_US.UTF-8
tz
          America/New_York
          2021-07-12
date
 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)
broom
                  0.7.8
                            2021-06-24 [1] CRAN (R 4.0.3)
                  1.5-12.2 2020-07-07 [1] CRAN (R 4.0.2)
Cairo
car
                  3.0-11
                            2021-06-27 [1] CRAN (R 4.0.3)
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
                  3.0.0
                            2021-06-30 [1] CRAN (R 4.0.3)
                  0.2-16
                            2018-12-24 [2] CRAN (R 4.0.3)
codetools
                  2.0 - 2
                            2021-06-24 [1] CRAN (R 4.0.3)
colorspace
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)
DBI
                  1.1.1
                            2021-01-15 [1] CRAN (R 4.0.2)
                            2021-04-06 [1] CRAN (R 4.0.3)
dbplyr
                  2.1.1
digest
                  0.6.27
                            2020-10-24 [1] CRAN (R 4.0.2)
                * 1.0.7
                            2021-06-18 [1] CRAN (R 4.0.3)
dplyr
                  0.3.2
                            2021-04-29 [1] CRAN (R 4.0.3)
ellipsis
```

evaluate		0.14	2019-05-28	[1]	CRAN ((R 4.0.2)
fansi		0.5.0	2021-05-25	[1]		(R 4.0.3)
farver		2.1.0	2021-02-28	[1]		(R 4.0.3)
forcats	*	0.5.1	2021-01-27	[1]		(R 4.0.2)
foreign	•	0.8-80	2020-05-24	[2]		(R 4.0.3)
fs		1.5.0	2020-07-31	[1]		(R 4.0.2)
generics		0.1.0	2020-10-31	[1]		(R 4.0.2)
ggplot2	*	3.3.5	2021-06-25	[1]		(R 4.0.2)
ggpubr		0.4.0	2020-06-27	[1]		(R 4.0.2)
ggsci	•	2.9	2018-05-14	[1]		(R 4.0.2)
ggsignif		0.6.2	2010 05 14	[1]		(R 4.0.2)
glue		1.4.2	2020-08-27	[1]		(R 4.0.3)
gtable		0.3.0	2019-03-25	[1]		(R 4.0.2)
haven		2.4.1	2019-03-23	[1]		(R 4.0.2)
_		1.1.0	2021-04-23	[1]		(R 4.0.3)
hms				[1]		(R 4.0.2)
htmltools		0.5.1.1	2021-01-22			
httr		1.4.2	2020-07-20	[1]		(R 4.0.2)
IRdisplay		1.0	2021-01-20	[1]		(R 4.0.2)
IRkernel		1.2	2021-05-11	[1]		(R 4.0.3)
jsonlite		1.7.2	2020-12-09	[1]		(R 4.0.2)
labeling		0.4.2	2020-10-20	[1]		(R 4.0.2)
lifecycle		1.0.0	2021-02-15	[1]		(R 4.0.3)
lubridate		1.7.10	2021-02-26	[1]		(R 4.0.3)
magrittr		2.0.1	2020-11-17	[1]		(R 4.0.2)
modelr		0.1.8	2020-05-19	[1]		(R 4.0.2)
munsell		0.5.0	2018-06-12	[1]		(R 4.0.2)
openxlsx		4.2.4	2021-06-16	[1]		(R 4.0.3)
pack		0.1-1	2021-02-23	[1]	local	
pbdZMQ		0.3-5	2021-02-10	[1]		(R 4.0.3)
pillar		1.6.1	2021-05-16	[1]		(R 4.0.3)
pkgconfig		2.0.3	2019-09-22	[1]		(R 4.0.2)
purrr	*	0.3.4	2020-04-17	[1]		(R 4.0.2)
${\tt PythonEmbedInR}$		0.6.76	2021-02-23	[1]	local	
R6		2.5.0	2020-10-28	[1]		(R 4.0.2)
Rcpp		1.0.7	2021-07-07	[1]	CRAN	(R 4.0.3)
readr	*	1.4.0	2020-10-05	[1]	CRAN	(R 4.0.2)
readxl		1.3.1	2019-03-13	[1]	CRAN	(R 4.0.2)
repr		1.1.3	2021-01-21	[1]	CRAN	(R 4.0.2)
reprex		2.0.0	2021-04-02	[1]	CRAN ((R 4.0.3)
rio		0.5.27	2021-06-21	[1]	CRAN ((R 4.0.3)
rlang		0.4.11	2021-04-30	[1]	CRAN ((R 4.0.3)
rstatix		0.7.0	2021-02-13	[1]	CRAN	(R 4.0.3)
rstudioapi		0.13	2020-11-12	[1]	CRAN	(R 4.0.2)
rvest		1.0.0	2021-03-09	[1]	CRAN	(R 4.0.3)
scales		1.1.1	2020-05-11	[1]	CRAN	(R 4.0.2)
sessioninfo		1.1.1	2018-11-05	[1]	CRAN	(R 4.0.2)
stringi		1.6.2	2021-05-17	[1]	CRAN	(R 4.0.3)
stringr	*	1.4.0	2019-02-10	[1]	CRAN	(R 4.0.2)
=						

```
svglite
                 2.0.0
                          2021-02-20 [1] CRAN (R 4.0.3)
synapser
               * 0.9.77
                          2021-02-23 [1] local
systemfonts
                 1.0.2
                          2021-05-11 [1] CRAN (R 4.0.3)
tibble
               * 3.1.2
                          2021-05-16 [1] CRAN (R 4.0.3)
               * 1.1.3
                          2021-03-03 [1] CRAN (R 4.0.3)
tidyr
                          2021-04-30 [1] CRAN (R 4.0.3)
tidyselect
                 1.1.1
tidyverse
                          2021-04-15 [1] CRAN (R 4.0.3)
               * 1.3.1
                          2021-03-12 [1] CRAN (R 4.0.3)
utf8
                 1.2.1
                          2020-02-26 [1] CRAN (R 4.0.2)
uuid
                 0.1-4
vctrs
                 0.3.8
                          2021-04-29 [1] CRAN (R 4.0.3)
                 2.4.2
                          2021-04-18 [1] CRAN (R 4.0.3)
withr
xm12
                 1.3.2
                          2020-04-23 [1] CRAN (R 4.0.2)
                 2.2.0
                          2021-05-31 [1] CRAN (R 4.0.3)
zip
```

- [1] /home/jbenja13/R/x86_64-pc-linux-gnu-library/4.0
- [2] /usr/lib/R/library