# main

July 23, 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 0.11.7 is detected at http://ran.synapse.org.
        To upgrade to the latest version of synapser, please run the following
        install.packages("synapser", repos="http://ran.synapse.org")
```

#### 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.

- 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

	SampleID	Sex	Race	Dx	Age	mitoRate	$rRNA\_Rate$	totalAssignedO
A data.table: $2 \times 14$	<chr $>$	<chr $>$	<chr $>$	<chr $>$	<dbl $>$	<dbl $>$	<dbl $>$	<dbl></dbl>
	R11135	Male	EA	CTL	18.77	0.2572796	0.0001690954	0.5231321
	R11137	Male	$\mathrm{EA}$	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
                                                                                          0.9798915
                                              Control
                  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)
```

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

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

D Dx	Brain_Region	RIN	$rRNA\_Rate$	Percent_Aligned	Dataset
<chr $>$	<chr $>$	<dbl $>$	<dbl $>$	<dbl></dbl>	<chr $>$
Control	Hippocampus	5.9	0.0001690954	0.8746	LIBD
Control	Hippocampus	9.2	0.0000884558	0.9149	LIBD
[.	ID Dx <pre><chr></chr></pre> Control Control	<pre><chr></chr></pre>	<chr><chr>ControlHippocampus5.9</chr></chr>	<chr><chr><dbl>          Control         Hippocampus         5.9         0.0001690954</dbl></chr></chr>	<chr> <chr> <dbl> <dbl>         Control       Hippocampus       5.9       0.0001690954       0.8746</dbl></dbl></chr></chr>

# [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

RIN

```
[10]: rin = df %>% group_by(Dataset, Brain_Region) %>% count(is.na(RIN)) %>%
    pivot_wider(names_from=`is.na(RIN)`, values_from="n") %>%
    mutate("TRUE"=replace_na(`TRUE`,0), `RIN (%na)`=`TRUE`/(`FALSE`+`TRUE`))
    rin %>% head(2)
```

```
Dataset
                             Brain Region
                                            FALSE
                                                     TRUE
                                                             RIN (%na)
                    <chr>
                              <chr>
                                            <int>
                                                     < dbl >
                                                             <dbl>
A grouped df: 2 \times 5
                    CMC
                             DLPFC
                                            991
                                                     0
                                                             0
                    GTEx
                                            177
                                                     0
                                                             0
                             Amygdala
```

#### rRNA Rate

#### Percent Alignment

#### Combined summary

	Dataset	Brain_Region	RIN (%na)	rRNA Rate (%na)	Percent Aligned
	<chr $>$	<chr></chr>	<dbl></dbl>	<dbl $>$	<dbl></dbl>
-	CMC	DLPFC	0.000000000	0.00000000	0.00000000
	GTEx	Amygdala	0.000000000	0.09039548	0.09039548
	GTEx	Anterior cingulate cortex	0.000000000	0.06572770	0.06572770
	GTEx	Caudate	0.000000000	0.07560137	0.07560137
	GTEx	Cerebellar Hemisphere	0.000000000	0.06844106	0.06844106
	GTEx	Cerebellum	0.003355705	0.07718121	0.07718121
	GTEx	Cortex	0.000000000	0.13846154	0.13846154
A grouped_df: $17 \times 5$	GTEx	DLPFC	0.287058824	0.48000000	0.48000000
	GTEx	Hippocampus	0.045267490	0.11934156	0.11934156
	GTEx	Hypothalamus	0.000000000	0.07203390	0.07203390
	GTEx	Nucleus accumbens	0.000000000	0.06859206	0.06859206
	GTEx	Putamen	0.000000000	0.05603448	0.05603448
	GTEx	Spinal cord	0.000000000	0.06043956	0.06043956
	GTEx	Substantia nigra	0.000000000	0.08536585	0.08536585
	LIBD	Caudate	0.000000000	0.00000000	0.00000000
	LIBD	DLPFC	0.000000000	0.00000000	0.00000000
	LIBD	Hippocampus	0.000000000	0.00000000	0.00000000

# 1.3.2 Summarize data

<sup>`</sup>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></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	6.669918 e-02	2.858002e-
	GTEx	Cerebellar Hemisphere	7.457034	1.0033617	2.524327e-02	2.178709e-
A grouped_df: $17 \times 8$	GTEx	Cerebellum	6.851852	0.7893323	2.060865 e-02	1.212267e-
	GTEx	Cortex	6.588308	0.6811292	4.852958 e-02	2.031282e-
	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	5.022716 e-05	3.060435e-
	LIBD	Hippocampus	7.723490	1.1208014	2.929714e-05	2.628273e

#### 1.4 Plot metrics

```
[15]: 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:

<sup>&</sup>quot;Removed 134 rows containing non-finite values (stat\_boxplot)."

<sup>&</sup>quot;Removed 134 rows containing missing values (geom\_point)."

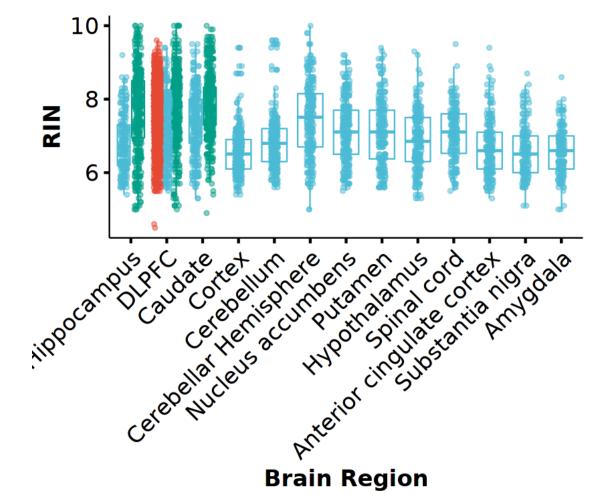
<sup>&</sup>quot;Removed 134 rows containing non-finite values (stat\_boxplot)." Warning message:

<sup>&</sup>quot;Removed 134 rows containing missing values (geom\_point)."

Warning message:

Warning message:

<sup>&</sup>quot;Removed 134 rows containing missing values (geom\_point)."



```
[17]: set.seed(20210723)
bxp = df %>% ggboxplot(x="Brain_Region", y="rRNA_Rate", color="Dataset", ⊔

→add='jitter',
```

<sup>&</sup>quot;Removed 134 rows containing non-finite values (stat\_boxplot)."

<sup>&</sup>quot;Removed 134 rows containing missing values (geom\_point)." Warning message:

<sup>&</sup>quot;Removed 134 rows containing non-finite values (stat\_boxplot)." Warning message:

```
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:

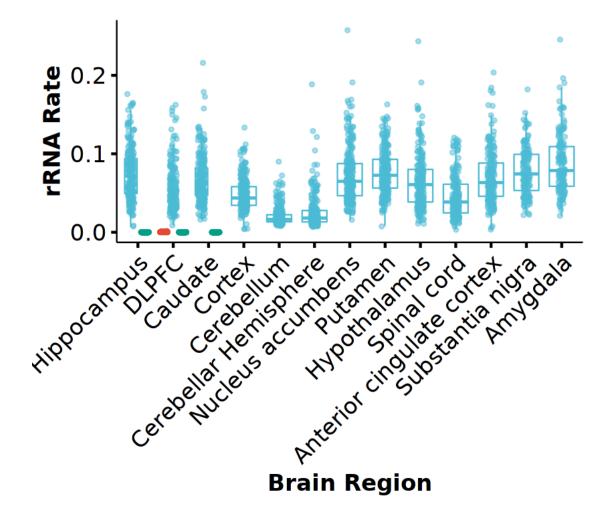
"Removed 445 rows containing missing values (geom\_point)." Warning message:

"Removed 445 rows containing non-finite values (stat\_boxplot)." Warning message:

"Removed 445 rows containing missing values (geom\_point)." Warning message:

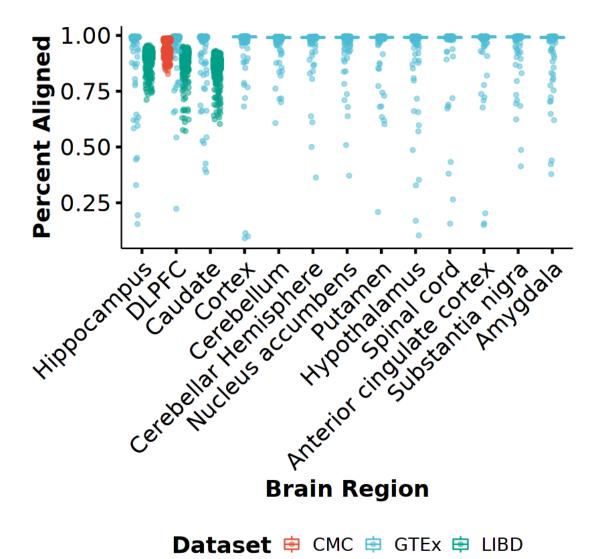
"Removed 445 rows containing non-finite values (stat\_boxplot)." Warning message:

"Removed 445 rows containing missing values (geom\_point)."



# 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)."
```



#### 1.5 Session Info

```
[19]: Sys.time()
      proc.time()
      options(width = 120)
      sessioninfo::session_info()
     [1] "2021-07-23 14:07:33 EDT"
        user system elapsed
               1.062 18.182
      19.432
      Session info
      setting value
```

version R version 4.0.3 (2020-10-10)

os Arch Linux

system x86\_64, linux-gnu

ui X11 language (EN)

collate en\_US.UTF-8
ctype en\_US.UTF-8
tz America/New\_York

date 2021-07-23

# Packages

1 ackages						
package	*	version	date	lib	sourc	ce
abind		1.4-5	2016-07-21	[1]	CRAN	(R 4.0.2)
assertthat		0.2.1	2019-03-21	[1]	CRAN	(R 4.0.2)
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)
Cairo		1.5-12.2	2020-07-07	[1]	CRAN	(R 4.0.2)
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)
cellranger		1.1.0	2016-07-27	[1]	CRAN	(R 4.0.2)
cli		3.0.0	2021-06-30	[1]	CRAN	(R 4.0.3)
codetools		0.2-16	2018-12-24	[2]	CRAN	(R 4.0.3)
colorspace		2.0-2	2021-06-24	[1]	CRAN	(R 4.0.3)
crayon		1.4.1	2021-02-08	[1]	CRAN	(R 4.0.3)
curl		4.3.2	2021-06-23	[1]	CRAN	(R 4.0.3)
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)
dbplyr		2.1.1	2021-04-06	[1]	CRAN	(R 4.0.3)
digest		0.6.27	2020-10-24	[1]	CRAN	(R 4.0.2)
dplyr	*	1.0.7	2021-06-18	[1]	CRAN	(R 4.0.3)
ellipsis		0.3.2	2021-04-29	[1]	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)
forcats	*	0.5.1	2021-01-27	[1]	CRAN	(R 4.0.2)
foreign		0.8-80	2020-05-24	[2]	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)
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)
gtable		0.3.0	2019-03-25	[1]	CRAN	(R 4.0.2)
haven		2.4.1	2021-04-23	[1]	CRAN	(R 4.0.3)
hms		1.1.0	2021-05-17	[1]	CRAN	(R 4.0.3)
htmltools		0.5.1.1	2021-01-22	[1]	CRAN	(R 4.0.2)

httr		1.4.2	2020-07-20	Γ <b>1</b> ]	CRAN (R 4.0.2)
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]	
labeling		0.4.2	2020-10-20	[1]	CRAN (R 4.0.2)
lifecycle		1.0.0	2021-02-15	[1]	CRAN (R 4.0.3)
lubridate		1.7.10	2021 02 13	[1]	CRAN (R 4.0.3)
magrittr		2.0.1	2021 02 20	[1]	
modelr		0.1.8	2020-05-19	[1]	CRAN (R 4.0.2)
munsell		0.5.0	2018-06-12	[1]	CRAN (R 4.0.2)
		4.2.4	2010-00-12	[1]	CRAN (R 4.0.2)
openxlsx		0.1-1	2021-00-10	[1]	local
pack		0.1-1	2021-02-23	[1]	CRAN (R 4.0.3)
pbdZMQ				[1]	CRAN (R 4.0.3)
pillar		1.6.1 2.0.3	2021-05-16		
pkgconfig	J.		2019-09-22	[1]	
purrr		0.3.4	2020-04-17	[1]	CRAN (R 4.0.2)
PythonEmbedInR		0.6.76	2021-02-23	[1]	local
R6		2.5.0	2020-10-28	[1]	CRAN (R 4.0.2)
Rcpp		1.0.7	2021-07-07	[1]	CRAN (R 4.0.3)
readr	*		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		1.7.3	2021-07-16	[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		
synapser	*	0.9.77	2021-02-23		
systemfonts		1.0.2	2021-05-11		
tibble	*	3.1.2	2021-05-16	[1]	CRAN (R 4.0.3)
tidyr	*	1.1.3	2021-03-03	[1]	CRAN (R 4.0.3)
tidyselect		1.1.1	2021-04-30	[1]	CRAN (R 4.0.3)
tidyverse	*	1.3.1	2021-04-15	[1]	CRAN (R 4.0.3)
utf8		1.2.1	2021-03-12	[1]	CRAN (R 4.0.3)
uuid		0.1-4	2020-02-26	[1]	CRAN (R 4.0.2)
vctrs		0.3.8	2021-04-29	[1]	CRAN (R 4.0.3)
withr		2.4.2	2021-04-18	[1]	CRAN (R 4.0.3)
xml2		1.3.2	2020-04-23	[1]	CRAN (R 4.0.2)
zip		2.2.0	2021-05-31	[1]	CRAN (R 4.0.3)

<sup>[1] /</sup>home/jbenja13/R/x86\_64-pc-linux-gnu-library/4.0
[2] /usr/lib/R/library