

Longitudinal data exploratory analysis

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## ##	The following object is masked _byGlobalEnv:	
##	root_dir	

Project: Comprehensive Omics Catalogue for Hartwell

St. Jude Children's Research Hospital Bio
Hackathon Team ${\bf 1}$

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1 Information about this notebook

This is an exploratory analysis of the data availability in terms of assays in the Comprehensive Omics Catalogue for Hartwell. This is critical for mitigating duplicate sequencing requests and efforts through Hartwell. This notebook aims to showcase: (1) which samples have already been sequenced by Hartwell, and (2) what omics data are available per sample.

For demo purposes, we use dummy data cohort and subset by human brain tumor samples. Finally, we investigate the number of samples per cancer_type_brain, Assay and longitudinal.

2 Set up

```
suppressPackageStartupMessages({
  library(tidyverse)
})
```

3 Directories and paths to file Inputs/Outputs

```
attach(params)
## The following object is masked _by_ .GlobalEnv:
##
       root dir
analysis dir <- file.path(root dir, "analyses", "data-exploratory-analysis")
input_dir <- file.path(analysis_dir, "input")</pre>
# We will first read in metadata file as we need to define sample_name
metadata_file <- file.path(input_dir, input_file) # metadata input file</pre>
palette_file <- file.path(root_dir, "figures", "palettes", "assay_color_palette.tsv")</pre>
tumor_palette_file <- file.path(root_dir, "figures", "palettes", "tumor_color_palette.tsv")</pre>
longitudinal_palette_file <- file.path(root_dir, "figures", "palettes", "longitudinal_color_palette.tsv</pre>
# File path to `plots` directory
plots_dir <- file.path(analysis_dir, "plots")</pre>
figures_plots_dir <- file.path(plots_dir, "figures-longitudinal")</pre>
if (!dir.exists(figures_plots_dir)) {
  dir.create(figures_plots_dir)}
source(paste0(analysis_dir, "/util/function-create-barplot.R"))
source(paste0(root dir, "/figures/scripts/theme plot.R"))
```

4 Read metadata file

We will subset by human brain tumor samples.

```
# Read metadata
df <- read.csv(metadata_file, stringsAsFactors=FALSE)

# Number of samples per cancer_type_brain
assays_number <- length(df$SJUID)
samples_number <- length(unique(df$SJUID))</pre>
```

4.1 Color palette for plotting

```
# Read color palette
palette_df <- readr::read_tsv(palette_file, guess_max = 100000, show_col_types = FALSE)

# Define and order palette
palette <- palette_df$hex_codes
names(palette) <- palette_df$color_names

# Read color palette for tumor type
tumor_palette_df <- readr::read_tsv(tumor_palette_file, guess_max = 100000, show_col_types = FALSE)

# Define and order palette
tumor_palette <- tumor_palette_df$hex_codes
names(tumor_palette) <- tumor_palette_df$color_names

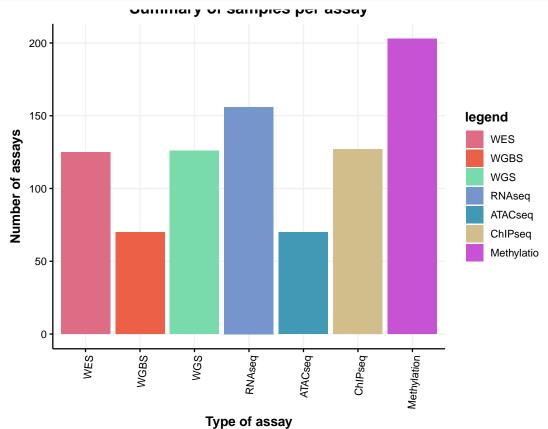
# Read color palette for longitudinal
longitudinal_palette_df <- readr::read_tsv(longitudinal_palette_file, guess_max = 100000, show_col_type)

# Define and order palette
longitudinal_palette <- longitudinal_palette_df$hex_codes
names(longitudinal_palette) <- longitudinal_palette_df$color_names</pre>
```

5 Number of samples with assay information

Table 1: Summary of samples per assay

Assay	n
WES 1	25
WGBS	70
WGS 1	26
RNAseq 1	56
ATACseq	70
ChIPseq 1	27
Methylation 2	203



```
pdf(file = fname, width = 6, height = 5)
print(p)
dev.off()
```

pdf ## 2

6 Number of samples per brain cancer type and assay

6.1 Overall assays

There are 59 brain tumor samples with 877 assays in total.

Table 2: Summary of samples and assays per brain cancer type

cancer_type_brain	n
Ependymoma	400
HGG	93
LGG	346
Medulloblastoma	38

Brain tumor type

```
pdf(file = fname, width = 6, height = 5)
print(p)
dev.off()
```

pdf ## 2

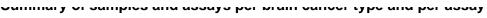
6.2 Per assay

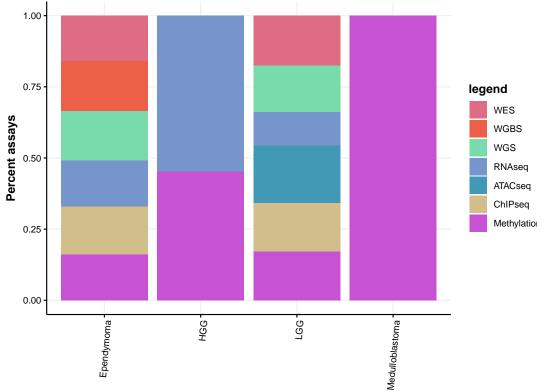
Table 3: Summary of samples and assays per brain cancer type and per assay

cancer_type_brain	WES	WGBS	WGS	RNAseq	ChIPseq	Methylation	ATACseq
Ependymoma	64	70	70	64	68	64	0

cancer_type_brain	WES	WGBS	WGS	RNAseq	ChIPseq	Methylation	ATACseq
HGG	0	0	0	51	0	42	0
LGG	61	0	56	41	59	59	70
Medulloblastoma	0	0	0	0	0	38	0

```
tables1 <- df %>% count(cancer_type_brain, Assay) %>%
  as.data.frame() %>%
  mutate_all(funs(replace_na(.,0)))
## Warning: `funs()` was deprecated in dplyr 0.8.0.
## i Please use a list of either functions or lambdas:
##
## # Simple named list: list(mean = mean, median = median)
##
## # Auto named with `tibble::lst()`: tibble::lst(mean, median)
##
## # Using lambdas list(~ mean(., trim = .2), ~ median(., na.rm = TRUE))
## Call `lifecycle::last_lifecycle_warnings()` to see where this warning was generated.
# Plot stacked barplot
fname <- paste0(figures_plots_dir, "/", "cancer-type-brain-per-assay-longitudinal.pdf")</pre>
p <- create_stacked_barplot(plot_df = tables1,</pre>
                             x_value = tables1$cancer_type_brain,
                             use_palette = palette,
                            xtitle ="Brain tumor type",
                             legend = tables1$Assay,
                             title_value = caption_value)
```





Brain tumor type

```
pdf(file = fname, width = 6, height = 5)
print(p)
dev.off()
```

6.3 Per disease stage

Table 4: Summary of samples and assays per brain cancer type, per disease stage and per assay $\,$

can-								ATAC-
cer_type_brain	longitudinal	WES	WGBS	WGS	RNAseq	ChIPseq	Methylation	seq
Ependymoma	Diagnosis	15	15	15	15	15	13	0
Ependymoma	Remission	15	17	17	15	17	17	0
Ependymoma	Relapse	16	18	18	16	17	16	0
Ependymoma	Deceased	18	20	20	18	19	18	0
HGG	Diagnosis	0	0	0	13	0	11	0
HGG	Remission	0	0	0	13	0	11	0
HGG	Relapse	0	0	0	14	0	12	0
HGG	Deceased	0	0	0	11	0	8	0
LGG	Diagnosis	16	0	14	11	15	15	17
LGG	Remission	14	0	13	10	14	14	17
LGG	Relapse	17	0	15	11	16	16	19
LGG	Deceased	14	0	14	9	14	14	17
Medulloblastoma	Diagnosis	0	0	0	0	0	9	0
Medulloblastoma	Remission	0	0	0	0	0	10	0
Medulloblastoma	Relapse	0	0	0	0	0	10	0
Medulloblastoma	Deceased	0	0	0	0	0	9	0

7 Number of samples per brain cancer type, longitudinal, assay, and SJUID

Table 5: Summary of cohort per brain cancer type, SJUID, disease stage, and assay $\,$

can-		longitudi-					Methyla-		ATAC-
cer_type_brain	SJUID	nal	WES	WGBS	WGS	ChIPseq	tion	RNAseq	seq
Ependymoma	SJH0H5WYF	RHPemission	1	1	1	1	1	0	0
Ependymoma	SJH0H5WYF	RE R elapse	1	1	1	1	1	0	0
Ependymoma	SJH0H5WYF	REDeceased	1	1	1	1	1	0	0
Ependymoma	SJH2HPPEE	KMiagnosis	1	1	1	1	1	1	0
Ependymoma	SJH2HPPEE	KRemission	1	1	1	1	1	1	0
Ependymoma	SJH2HPPEE	KMRelapse	1	1	1	1	1	1	0
Ependymoma	SJH2HPPEE	K M eceased	1	1	1	1	1	1	0
Ependymoma	SJH51B396IV	V Diagnosis	1	1	1	1	1	1	0
Ependymoma	SJH51B396IV	VRemission	1	1	1	1	1	1	0
Ependymoma	SJH51B396IV	V Relapse	1	1	1	1	1	1	0
Ependymoma	SJH51B396IV	V Deceased	1	1	1	1	1	1	0
Ependymoma	SJH5HCKPC	9Remission	0	1	1	1	1	1	0
Ependymoma	SJH5HCKPC	97Relapse	0	1	1	1	1	1	0
Ependymoma	SJH5HCKPC	97Deceased	0	1	1	1	1	1	0
Ependymoma	SJH98QNKIU		0	1	1	1	1	1	0
Ependymoma	SJH98QNKIU		0	1	1	1	1	1	0
Ependymoma	SJH98QNKIU	JUDeceased	0	1	1	1	1	1	0
Ependymoma	SJH9YML-	Diagnosis	1	1	1	1	1	1	0
	FOTI								
Ependymoma	SJH9YML-	Remission	1	1	1	1	1	1	0
	FOTI								
Ependymoma	SJH9YML-	Relapse	1	1	1	1	1	1	0
	FOTI								
Ependymoma	SJH9YML-	Deceased	1	1	1	1	1	1	0
	FOTI								
Ependymoma	SJHA6KC56J	J6Diagnosis	1	1	1	1	1	1	0
Ependymoma	SJHA6KC56J	I6Remission	1	1	1	1	1	1	0
Ependymoma	SJHA6KC56.		1	1	1	1	1	1	0
Ependymoma	SJHA6KC56.		1	1	1	1	1	1	0
Ependymoma	SJH-	Diagnosis	1	1	1	1	1	1	0
	BKS7QSFO								
Ependymoma	SJH-	Remission	1	1	1	1	1	1	0
	BKS7QSFO								
Ependymoma	SJH-	Relapse	1	1	1	1	1	1	0
	BKS7QSFO								
Ependymoma	SJH-	Deceased	1	1	1	1	1	1	0
	BKS7QSFO								
Ependymoma	SJHC70DZR.		1	1	1	1	1	1	0
Ependymoma	SJHC70DZR.		1	1	1	1	1	1	0
Ependymoma	SJHC70DZR.	•	1	1	1	1	1	1	0
Ependymoma	SJHC70DZR.		1	1	1	1	1	1	0
Ependymoma	SJHCN03RC	-	1	1	1	1	1	1	0
Ependymoma	SJHCN03RC		1	1	1	1	1	1	0
Ependymoma	SJHCN03RC	V Deceased	1	1	1	1	1	1	0

can- cer_type_brain	SJUID	longitudi- nal	WES	WGBS	WGS	ChIPseq	Methyla- tion	RNAseq	ATAC- seq
Ependymoma	SJHD1KI- UMM6	Diagnosis	1	1	1	1	1	1	0
Ependymoma	SJHD1KI- UMM6	Remission	1	1	1	1	1	1	0
Ependymoma	SJHD1KI- UMM6	Relapse	1	1	1	1	1	1	0
Ependymoma	SJHD1KI- UMM6	Deceased	1	1	1	1	1	1	0
Ependymoma	SJHFG- GJRHYO	Remission	1	1	1	1	1	0	0
Ependymoma	SJHFG- GJRHYO	Relapse	1	1	1	1	1	0	0
Ependymoma	SJHFG- GJRHYO	Deceased	1	1	1	1	1	0	0
Ependymoma	SJH- HZH67WMF	Relapse	1	1	1	0	1	1	0
Ependymoma	SJH- HZH67WMF	Deceased	1	1	1	0	1	1	0
Ependymoma	SJHI8CC7QT	8Diagnosis	1	1	1	1	1	1	0
Ependymoma	SJHI8CC7QT		1	1	1	1	1	1	0
Ependymoma	SJHI8CC7QT		1	1	1	1	1	1	0
Ependymoma	SJHI8CC7QT		1	1	1	1	1	1	0
Ependymoma	SJHIQT- NAYIF	Diagnosis	1	1	1	1	1	1	0
Ependymoma	SJHIQT- NAYIF	Remission	1	1	1	1	1	1	0
Ependymoma	SJHIQT- NAYIF	Relapse	1	1	1	1	1	1	0
Ependymoma	SJHIQT- NAYIF	Deceased	1	1	1	1	1	1	0
Ependymoma	SJHKVQE- BOCP	Diagnosis	1	1	1	1	0	1	0
Ependymoma	SJHKVQE- BOCP	Relapse	1	1	1	1	0	1	0
Ependymoma	SJHKVQE- BOCP	Deceased	1	1	1	1	0	1	0
Ependymoma	SJHKYUIYA	M Diagnosis	1	1	1	1	0	1	0
Ependymoma	SJHKYUIYA	~	1	1	1	1	0	1	0
Ependymoma	SJHKYUIYA		1	1	1	1	0	1	0
Ependymoma	SJH-	Diagnosis	1	1	1	1	1	1	0
_penaj mema	PVXLA- CLM	2108110010	-	-	-	-	-	-	
Ependymoma	SJH- PVXLA- CLM	Remission	1	1	1	1	1	1	0
Ependymoma	SJH- PVXLA- CLM	Relapse	1	1	1	1	1	1	0
Ependymoma	SJH- PVXLA- CLM	Deceased	1	1	1	1	1	1	0

can- cer_type_brain	longitudi- SJUID nal	WES	WCBS	WCS	ChIPseq	Methyla- tion	RNAseq	ATAC-
								seq
Ependymoma	SJHUMKP2L6 Diagnosis	1	1	1	1	1	1	0
Ependymoma	SJHUMKP2L6 Wemission	1	1	1	1	1	1	0
Ependymoma	SJHUMKP2L6 Deceased	1	1	1	1	1	1	0
Ependymoma	SJHXTBTQ5ZDiagnosis	1	1	1	1	1	1	0
Ependymoma	SJHXTBTQ5ZRemission	1	1	1	1	1	1	0
Ependymoma	SJHXTBTQ5ZTRelapse	1	1	1	1	1	1	0
Ependymoma	SJHXTBTQ5ZDeceased	1	1	1	1	1	1	0
HGG	SJH5QHZM4UDiagnosis	0	0	0	0	1	1	0
HGG	SJH5QHZM4USemission	0	0	0	0	1	1	0
HGG	SJH5QHZM4U\$Relapse	0	0	0	0	1	1	0
HGG	SJH5QHZM4UDeceased	0	0	0	0	1	1	0
HGG	SJHADL5OJM P iagnosis	0	0	0	0	1	1	0
HGG	SJHADL5OJMRemission	0	0	0	0	1	1	0
HGG	SJHADL5OJMERelapse	0	0	0	0	1	1	0
HGG	SJHADL5OJMDeceased	0	0	0	0	1	1	0
HGG	SJH- Diagnosis	0	0	0	0	1	1	0
	BISK5KBU							
HGG	SJH- Remission	0	0	0	0	1	1	0
	BISK5KBU							
HGG	SJH- Relapse	0	0	0	0	1	1	0
	BISK5KBU							
$_{ m HGG}$	${\rm SJHBL7CDYR}{\bf N} {\rm iagnosis}$	0	0	0	0	1	1	0
HGG	SJHBL7CDYR Remission	0	0	0	0	1	1	0
$_{ m HGG}$	SJHBL7CDYRNRelapse	0	0	0	0	1	1	0
HGG	${\rm SJHBL7CDYR} \mathbf{D} eceased$	0	0	0	0	1	1	0
HGG	SJHD- Diagnosis DTE0SYL	0	0	0	0	0	1	0
HGG	SJHD- Relapse	0	0	0	0	0	1	0
	DTE0SYL							
HGG	SJHEOVURBMDIagnosis	0	0	0	0	1	1	0
HGG	SJHEOVURB M #mission	0	0	0	0	1	1	0
$_{ m HGG}$	${\bf SJHEOVURBMR} elapse$	0	0	0	0	1	1	0
$_{ m HGG}$	SJHEOVURBMDeceased	0	0	0	0	1	1	0
$_{ m HGG}$	SJHEQG3P4F I Diagnosis	0	0	0	0	1	0	0
HGG	${\bf SJHEQG3P4FKRelapse}$	0	0	0	0	1	0	0
HGG	SJHHPN- Diagnosis	0	0	0	0	1	1	0
	QERSQ							
$_{ m HGG}$	SJHHPN- Remission	0	0	0	0	1	1	0
	QERSQ							
HGG	SJHHPN- Relapse	0	0	0	0	1	1	0
шаа	QERSQ	0	0	0	0	1	-1	0
HGG	SJHHPN- Deceased	0	0	0	0	1	1	0
1100	QERSQ	0	0		0		-	^
HGG	SJHHW23UJ9Remission	0	0	0	0	1	1	0
HGG	SJHHW23UJ9PRelapse	0	0	0	0	1	1	0
HGG	SJHJ59RLHSUDiagnosis	0	0	0	0	1	1	0
HGG	SJHJ59RLHSURemission	0	0	0	0	1	1	0
HGG	SJHJ59RLHSU Relapse	0	0	0	0	1	1	0
HGG	SJHJ59RLHSUDeceased	0	0	0	0	1	1	0
HGG	SJHKKD- Diagnosis	0	0	0	0	1	1	0
	DXOYH							

centrype_brain SJUID nal WES WGBS WGS ChIPse HGG SJHKKD- Remission DXOYH 0	Methylation RNAsec 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	0 0 0 0 0 0 0
DXOYH	1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 0 1 0 1	0 0 0 0 0
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HGG SJHMI643DMDRelapse 0 0 0 HGG SJHMI643DMIDeceased 0 0 0 HGG SJHQBOPS9FDiagnosis 0 0 0 HGG SJHQBOPS9FDemission 0 0 0 HGG SJHQBOPS9FDeceased 0 0 0 0 HGG SJHRO- Diagnosis 0 0 0 0 0 HGG SJHRO- Remission 0	$egin{array}{cccccccccccccccccccccccccccccccccccc$	0 0
HGG SJHMI643DMIDeceased 0 0 0 0 HGG SJHQBOPS9FDiagnosis 0 0 0 0 HGG SJHQBOPS9FDeceased 0 0 0 0 HGG SJHQBOPS9FDeceased 0 0 0 0 HGG SJHRO- Diagnosis 0 0 0 0 HGG SJHRO- Remission 0 0 0 0 JUQZAP Diagnosis 0 0 0 0 HGG SJHUT- Diagnosis 0 0 0 0 PISXFQ HGG SJHUT- Remission 0 0 0 0 HGG SJHUT- Relapse 0 0 0 0 0 PISXFQ HGG SJHVT- Deceased 0 0 0 0 HGG SJHVIS5Q8HXDiagnosis 0 0 0 0 HGG SJHVIS5Q8HXRemission 0 0 0 0	$egin{array}{cccccccccccccccccccccccccccccccccccc$	0
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HGG SJHQBOPS9FRemission 0 0 0 0 HGG SJHQBOPS9FIRelapse 0 0 0 0 HGG SJHRO- Diagnosis 0 0 0 0 HGG SJHRO- Remission 0 0 0 0 JUQZAP HGG SJHRO- Relapse 0 0 0 0 HGG SJHUT- Diagnosis 0 0 0 0 0 PISXFQ HGG SJHUT- Remission 0 0 0 0 0 PISXFQ HGG SJHUT- Deceased 0	$egin{array}{ccc} 0 & & 1 \\ 0 & & 1 \\ 0 & & 1 \\ \end{array}$	0
HGG SJHQBOPS9FIRelapse 0 0 0 0 HGG SJHQBOPS9FIDeceased 0 0 0 0 HGG SJHRO- Diagnosis 0 0 0 0 JUQZAP HGG SJHRO- Relapse on the street of the str	$egin{array}{ccc} 0 & & 1 \ 0 & & 1 \end{array}$	o 0
HGG SJHQBOPS9FDeceased 0 0 0 0 HGG SJHRO- Diagnosis 0 0 0 0 JUQZAP HGG SJHRO- Remission 0 0 0 0 HGG SJHRO- Relapse 0 0 0 0 0 JUQZAP HGG SJHUT- Diagnosis 0 0 0 0 0 HGG SJHUT- Remission 0 <td>0 1</td> <td>Ö</td>	0 1	Ö
HGG SJHRO- Diagnosis JUQZAP O 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0		o 0
JUQZAP	1 0	0
HGG	1	
JUQZAP HGG SJHUT- Diagnosis 0 0 0 0 PISXFQ Remission 0 0 0 0 HGG SJHUT- Relapse 0 0 0 0 PISXFQ PISXFQ 0 0 0 0 HGG SJHUT- Deceased 0 0 0 0 PISXFQ 0 0 0 0 0 HGG SJHVIS5Q8HXDiagnosis 0 0 0 0 HGG SJHVIS5Q8HXRemission 0 0 0 0 HGG SJHVIS5Q8HXDeceased 0 0 0 0 HGG SJHVIS5Q8HXDeceased 0 0 0 0 LGG SJH2W47P7D@emission 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH2W47P7D@ece	1 0	0
HGG SJHUT- Diagnosis 0 0 0 0 PISXFQ SJHUT- Remission 0 0 0 0 HGG SJHUT- Relapse 0 0 0 0 PISXFQ Deceased 0 0 0 0 HGG SJHUT- Deceased 0 0 0 0 PISXFQ PISXFQ 0 0 0 0 HGG SJHVIS5Q8HXDiagnosis 0 0 0 0 HGG SJHVIS5Q8HXRemission 0 0 0 0 HGG SJHVIS5Q8HXDeceased 0 0 0 0 HGG SJHVIS5Q8HXDeceased 0 0 0 0 LGG SJH2W47P7D@iagnosis 1 0 1 1 LGG SJH2W47P7D@emission 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH2W47P7D@eceased 1 0	1 0	0
HGG SJHUT- Remission PISXFQ HGG SJHUT- Relapse 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	0 1	0
HGG SJHUT- Relapse 0 0 0 0 PISXFQ BJHUT- Deceased 0 0 0 0 HGG SJHUT- Deceased 0 0 0 0 HGG SJHVIS5Q8HXDiagnosis 0 0 0 0 HGG SJHVIS5Q8HXRemission 0 0 0 0 HGG SJHVIS5Q8HX Relapse 0 0 0 0 HGG SJHVIS5Q8HXDeceased 0 0 0 0 LGG SJH2W47P7D@iagnosis 1 0 1 1 LGG SJH2W47P7D@emission 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 LGG SJH5HCKPC9Remission 0 0 0 0	0 1	0
HGG SJHUT- Deceased PISXFQ 0 0 0 0 0 HGG SJHVIS5Q8HXDiagnosis 0 0 0 0 0 HGG SJHVIS5Q8HXRemission 0 0 0 0 0 HGG SJHVIS5Q8HX Relapse 0 0 0 0 0 HGG SJHVIS5Q8HXDeceased 0 0 0 0 0 LGG SJH2W47P7D@biagnosis 1 0 1 1 1 LGG SJH2W47P7D@emission 1 0 1 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 1 LGG SJH2W47P7D@eceased 1 0 1 1 1 LGG SJH5HCKPC9Remission 0 0 0 0 0	0 1	0
$\begin{array}{cccccccccccccccccccccccccccccccccccc$	0 1	0
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$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	1 1	1
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	1 1	1
LGG SJH5HCKPC9Remission 0 0 0	1 1	1
	0 0	1
	0 0	1
LGG SJH5HCKPC9TDeceased 0 0 0 0	0 0	1
LGG SJHAY4GX4A D iagnosis 1 0 1 1	1 1	1
LGG SJHAY4GX4ANemission 1 0 1 1	1 1	1
LGG SJHAY4GX4ANRelapse 1 0 1 1		1
LGG SJHAY4GX4ANDeceased 1 0 1 1	1 1	1
LGG SJHBN- Remission 1 0 1 1 JSZHW6	1 0	1
LGG SJHBN- Relapse 1 0 1 1 $JSZHW6$	1 0	1
LGG SJHBN- Deceased 1 0 1 1 JSZHW6		1
LGG SJHBV3Q6UVRiagnosis 1 0 1 1	1 0	1
LGG SJHBV3Q6UVRemission 1 0 1 1	1 0 1 1	1

can-	1 .	CHID	longitudi-	MEG	WODO	Waa	CLID	Methyla-	DMA	ATAC-
cer_type_	_brain	SJUID	nal	WES		WGS	ChIPseq	tion	RNAseq	seq
$_{ m LGG}$		SJHBV3Q6U	VRelapse	1	0	1	1	1	1	1
LGG		SJHBV3Q6U	V Receased	1	0	1	1	1	1	1
LGG		SJHC70DZR	JSDiagnosis	0	0	0	0	0	0	1
$_{ m LGG}$		SJHC70DZR	JRemission	0	0	0	0	0	0	1
$_{ m LGG}$		SJHC70DZR	JS Relapse	0	0	0	0	0	0	1
$_{ m LGG}$		SJHC70DZR	JSDeceased	0	0	0	0	0	0	1
LGG		SJH- CLGFJTIG	Diagnosis	1	0	1	1	1	1	1
LGG		SJH- CLGFJTIG	Remission	1	0	1	1	1	1	1
LGG		SJH- CLGFJTIG	Relapse	1	0	1	1	1	1	1
LGG		SJH- CLGFJTIG	Deceased	1	0	1	1	1	1	1
LGG		SJHD- DTE0SYL	Diagnosis	1	0	1	1	0	0	0
LGG		SJHD-	Relapse	1	0	1	1	0	0	0
LGG		DTE0SYL SJHI52BLNV	Wiomasia	1	0	1	1	1	1	1
LGG		SJHI52BLNV		1						
LGG				1	0	1	1	1 1	1	1
LGG		SJHI52BLNV SJHI52BLNV		1	0	1	1		1	1
					0	1	1	1	1	1
LGG		SJHIQT- NAYIF	Diagnosis	0	0	0	0	0	0	1
LGG		SJHIQT- NAYIF	Remission	0	0	0	0	0	0	1
LGG		SJHIQT- NAYIF	Relapse	0	0	0	0	0	0	1
LGG		SJHIQT- NAYIF	Deceased	0	0	0	0	0	0	1
LGG		SJHN- PJTQHIT	Diagnosis	1	0	1	1	1	0	1
LGG		SJHN- PJTQHIT	Relapse	1	0	1	1	1	0	1
LGG		SJHN- PJTQHIT	Deceased	1	0	1	1	1	0	1
LGG		SJHO- FORRR7C	Diagnosis	1	0	1	1	1	1	1
LGG		SJHO- FORRR7C	Remission	1	0	1	1	1	1	1
LGG		SJHO- FORRR7C	Relapse	1	0	1	1	1	1	1
LGG		SJHO- FORRR7C	Deceased	1	0	1	1	1	1	1
LGG		SJHOY05OJ	.INDiagnosis	1	0	0	0	1	1	1
LGG		SJHOY05OJ		1	0	0	0	1	1	1
LGG		SJHQBOPS9	-	1	0	1	1	1	0	1
LGG		SJHQBOPS9		1	0	1	1	1	0	1
LGG		SJHQBOPS9		1	0	1	1	1	0	1
LGG		SJHQBOPS9		1	0	1	1	1	0	1
LGG		SJHUT-	Diagnosis	1	0	1	1	1	0	1
200		PISXFQ	T-1001100110	1	J	-	1	±	V	1

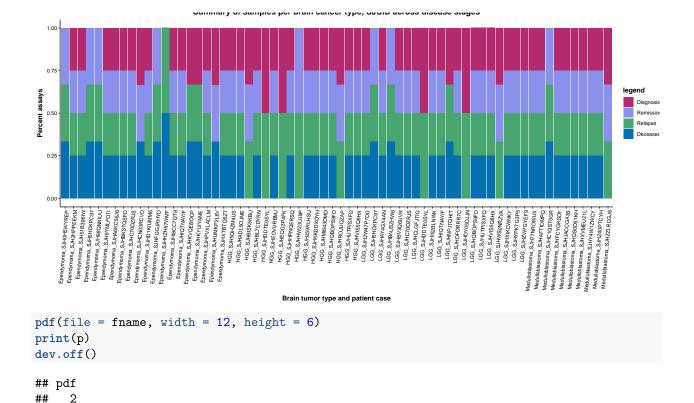
can- cer_type_brain	SJUID	longitudi- nal	WES	WGBS	WGS	ChIPseq	Methyla- tion	RNAseq	ATAC- seq
LGG	SJHUT- PISXFQ	Remission	1	0	1	1	1	0	1
LGG	SJHUT- PISXFQ	Relapse	1	0	1	1	1	0	1
LGG	SJHUT- PISXFQ	Deceased	1	0	1	1	1	0	1
LGG	SJHVIS5Q8F	HXDiagnosis	1	0	1	1	1	0	1
LGG	SJHVIS5Q8I		1	0	1	1	1	0	1
LGG	SJHVIS5Q8F		1	0	1	1	1	0	1
LGG	SJHVIS5Q8F		1	0	1	1	1	0	1
LGG	SJHWS0NRZ		1	0	0	1	1	1	1
LGG	SJHWS0NRZ		1	0	0	1	1	1	1
LGG	SJHWS0NRZ		1	0	0	1	1	1	1
LGG	SJHXIKCWI		1	0	1	1	1	1	1
LGG	SJHXIKCWI		1	0	1	1	1	1	1
LGG	SJHXIKCWI		1						
	SJHXIKCWI			0	1	1	1	1	1
LGG			1	0	1	1	1	1	1
LGG	SJHYP- KTG3P5	Diagnosis	1	0	1	1	1	1	1
LGG	SJHYP- KTG3P5	Remission	1	0	1	1	1	1	1
LGG	SJHYP- KTG3P5	Relapse	1	0	1	1	1	1	1
LGG	SJHYP- KTG3P5	Deceased	1	0	1	1	1	1	1
LGG	SJHZW7GY	FH0ingnosis	1	0	1	1	1	1	1
LGG	SJHZW7GY		1	0	1	1	1	1	1
LGG	SJHZW7GY		1	0	1	1	1	1	1
LGG	SJHZW7GY	-	1	0	1	1	1	1	1
Medulloblas-	SJH77NRD-	Diagnosis	0	0	0	0	1	0	0
toma	WUX								
Medulloblas- toma	SJH77NRD- WUX	Remission	0	0	0	0	1	0	0
Medulloblas- toma	SJH77NRD- WUX	Relapse	0	0	0	0	1	0	0
Medulloblas-	SJH77NRD-	Deceased	0	0	0	0	1	0	0
toma Medulloblas-	WUX SJHAF-	Diagnosis	0	0	0	0	1	0	0
toma	TIOMPQ								
Medulloblas-	SJHAF-	Remission	0	0	0	0	1	0	0
toma	TIOMPQ								
Medulloblas-	SJHAF-	Relapse	0	0	0	0	1	0	0
toma Medulloblas-	TIOMPQ SJHAF-	Deceased	0	0	0	0	1	0	0
toma Medulloblas-	TIOMPQ SJHC1QST5	GRemission	0	0	0	0	1	0	0
toma Medulloblas-	SJHC1QST5	GRRelapse	0	0	0	0	1	0	0
toma Medulloblas- toma	SJHC1QST5	GRDeceased	0	0	0	0	1	0	0

can-		longitudi-					Methyla-		ATAC-
cer_type_brain	SJUID	$_{\mathrm{nal}}$	WES	WGBS	WGS	ChIPseq	tion	RNAseq	seq
Medulloblas-	SJHF-	Diagnosis	0	0	0	0	1	0	0
toma	CYGKSDY	8		ŭ		v		, and the second	, and the second
Medulloblas-	SJHF-	Remission	0	0	0	0	1	0	0
toma	CYGKSDY	1001111001011	Ŭ	Ŭ	V	v	-	v	Ü
Medulloblas-	SJHF-	Relapse	0	0	0	0	1	0	0
toma	CYGKSDY	1							
Medulloblas-	SJHF-	Deceased	0	0	0	0	1	0	0
toma	CYGKSDY								
Medulloblas-	SJHJAC-	Diagnosis	0	0	0	0	1	0	0
toma	CGA3S	Ü							
Medulloblas-	SJHJAC-	Remission	0	0	0	0	1	0	0
toma	CGA3S								
Medulloblas-	SJHJAC-	Relapse	0	0	0	0	1	0	0
toma	CGA3S	-							
Medulloblas-	SJHJAC-	Deceased	0	0	0	0	1	0	0
toma	CGA3S								
Medulloblas-	SJHQS0D511	KHDiagnosis	0	0	0	0	1	0	0
toma	•	G							
Medulloblas-	SJHQS0D511	KRemission	0	0	0	0	1	0	0
toma	•								
Medulloblas-	SJHQS0D511	KHRelapse	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHQS0D511	KHDeceased	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHXVMEU	21Diagnosis	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHXVMEU	21Remission	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHXVMEU	21IRelapse	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHXVMEU	21Deceased	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHY4W1ZV	W O Magnosis	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHY4W1ZV	V RY emission	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHY4W1ZV	VC K elapse	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHY4W1ZV	W D eceased	0	0	0	0	1	0	0
toma									
Medulloblas-	SJHZ40PT-	Diagnosis	0	0	0	0	1	0	0
toma	CYH								
Medulloblas-	SJHZ40PT-	Remission	0	0	0	0	1	0	0
toma	CYH								
Medulloblas-	SJHZ40PT-	Relapse	0	0	0	0	1	0	0
toma	СҮН			_		_			_
Medulloblas-	SJHZ40PT-	Deceased	0	0	0	0	1	0	0
toma	СҮН	D	_	•		•	_	•	_
Medulloblas-	SJHZZLR-	Diagnosis	0	0	0	0	1	0	0
toma	CGJ6	ъ				0			
Medulloblas-	SJHZZLR-	Remission	0	0	0	0	1	0	0
toma	CGJ6								

can- cer_type_brain	SJUID	longitudi- nal	WES	WGBS	WGS	ChIPseq	Methyla- tion	RNAseq	ATAC- seq
Medulloblas- toma	SJHZZLR- CGJ6	Relapse	0	0	0	0	1	0	0

7.1 Plots

```
tables1 <- df %>% count(cancer_type_brain, SJUID, longitudinal, Assay) %>%
  as.data.frame() %>%
  arrange(cancer_type_brain, longitudinal, Assay) %>%
  mutate(match_id_brain = paste(cancer_type_brain, SJUID, sep = "_"),
         match_id_brain_longitudinal = paste(cancer_type_brain, SJUID, longitudinal, sep = "_")) %>%
  mutate_all(funs(replace_na(.,0)))
## Warning: `funs()` was deprecated in dplyr 0.8.0.
## i Please use a list of either functions or lambdas:
## # Simple named list: list(mean = mean, median = median)
##
## # Auto named with `tibble::lst()`: tibble::lst(mean, median)
##
## # Using lambdas list(~ mean(., trim = .2), ~ median(., na.rm = TRUE))
## Call `lifecycle::last_lifecycle_warnings()` to see where this warning was generated.
# Run function
fname <- paste0(figures_plots_dir, "/", "cancer-type-brain-per-assay-patient.pdf")</pre>
p <- create_stacked_barplot(plot_df = tables1,</pre>
                            x_value = tables1$match_id_brain_longitudinal,
                            use_palette = palette,
                            xtitle = "Brain tumor type and patient case",
                            legend = tables1$Assay,
                             title_value = "Summary of samples per brain cancer type, SJUID and disease
pdf(file = fname, width = 25, height = 6)
print(p)
dev.off()
## pdf
##
# Run function
fname <- paste0(figures_plots_dir, "/", "cancer-type-brain-patient-longitudinal.pdf")</pre>
p <- create_stacked_barplot(plot_df = tables1,</pre>
                             x_value = tables1$match_id_brain,
                            use_palette = longitudinal_palette,
                            xtitle = "Brain tumor type and patient case",
                            legend = tables1$longitudinal,
                            title_value = "Summary of samples per brain cancer type, SJUID across disea
```



8 Future directions

The current exploratory data analysis module can be expanded by investigating samples with paired assays. Moreover, if other metadata are available, e.g., disease_stage, treatment, this will build large, longitudinal cohorts with multi-omic sequencing data. Such an analysis permits consideration of samples according to the condition(s) of the experiment and research aims. In addition, it can be used to refine research questions and/or generate new ones.

This will facilitate collaboration across departments at St. Jude, expedite discoveries, and find cures for children with cancer and other catastrophic diseases.

9 Session Info

```
## R version 4.4.0 (2024-04-24)
## Platform: x86_64-pc-linux-gnu
## Running under: Red Hat Enterprise Linux 8.8 (Ootpa)
##
## Matrix products: default
           /research/rgs01/applications/hpcf/authorized_apps/rhel8_apps/lapack/3.10.1/install/lib64/lib
## LAPACK: /research/rgs01/applications/hpcf/authorized_apps/rhel8_apps/lapack/3.10.1/install/lib64/lib
## locale:
##
  [1] LC_CTYPE=en_US.UTF-8
                                   LC_NUMERIC=C
  [3] LC_TIME=en_US.UTF-8
                                   LC_COLLATE=en_US.UTF-8
   [5] LC_MONETARY=en_US.UTF-8
                                   LC_MESSAGES=en_US.UTF-8
   [7] LC_PAPER=en_US.UTF-8
                                   LC_NAME=C
##
  [9] LC_ADDRESS=C
                                   LC_TELEPHONE=C
## [11] LC_MEASUREMENT=en_US.UTF-8 LC_IDENTIFICATION=C
##
## time zone: America/Chicago
## tzcode source: system (glibc)
## attached base packages:
## [1] grid
                 stats
                           graphics grDevices utils
                                                          datasets methods
## [8] base
## other attached packages:
  [1] ggthemes_5.1.0
                        lubridate_1.9.3 forcats_1.0.0
##
                                                         stringr_1.5.1
   [5] dplyr_1.1.4
                        purrr_1.0.2
                                        readr_2.1.5
                                                         tidyr_1.3.1
## [9] tibble_3.2.1
                        ggplot2_3.5.1
                                        tidyverse_2.0.0
## loaded via a namespace (and not attached):
## [1] sass_0.4.9
                          utf8_1.2.4
                                             generics_0.1.3
                                                               stringi_1.8.4
## [5] hms_1.1.3
                          digest_0.6.37
                                             magrittr_2.0.3
                                                               evaluate_0.24.0
## [9] timechange_0.3.0 fastmap_1.2.0
                                             jsonlite_1.8.8
                                                               tinytex_0.52
## [13] fansi_1.0.6
                          scales_1.3.0
                                             jquerylib_0.1.4
                                                               cli_3.6.3
## [17] rlang_1.1.4
                          crayon_1.5.3
                                             bit64_4.0.5
                                                               munsell_0.5.1
## [21] withr_3.0.1
                                             yaml_2.3.10
                                                               tools_4.4.0
                          cachem_1.1.0
## [25] parallel_4.4.0
                          tzdb_0.4.0
                                             colorspace_2.1-1
                                                               vctrs_0.6.5
## [29] R6_2.5.1
                          mime_0.12
                                             lifecycle_1.0.4
                                                               bit_4.0.5
## [33] vroom_1.6.5
                                             pillar_1.9.0
                          pkgconfig_2.0.3
                                                               bslib_0.8.0
## [37] gtable_0.3.5
                          glue_1.7.0
                                             xfun 0.47
                                                               tidyselect 1.2.1
                                                               htmltools_0.5.8.1
## [41] highr_0.11
                          knitr_1.48
                                             farver_2.1.2
## [45] rmarkdown_2.28
                          labeling_0.4.3
                                             compiler_4.4.0
```