Group 3 Variant_Analysis

2025-07-10

```
#get working directory
getwd() #"/Users/sequencingplatform/Documents/Linux_Basics_to_Mastery_training/Module-15"
```

```
## [1] "/Users/sequencingplatform/Documents/Linux_Basics_to_Mastery_training/Module-15"
```

```
#load libraries
library(dplyr)
```

```
##
## Attaching package: 'dplyr'
```

```
## The following objects are masked from 'package:stats':
##
##     filter, lag
```

```
## The following objects are masked from 'package:base':
##
##     intersect, setdiff, setequal, union
```

```
library(openxlsx)
library(janitor)
```

```
##
## Attaching package: 'janitor'
```

```
## The following objects are masked from 'package:stats':
##
##     chisq.test, fisher.test
```

```
library(stringi)
library(stringr)
library(ggplot2)
library(data.table)
```

```
##
## Attaching package: 'data.table'
```

```
## The following objects are masked from 'package:dplyr':
##
##     between, first, last
```

```r
#load hq_allvariants.tsv
data_allvariants <- fread("vcf/hq_allvariants.tsv")
```

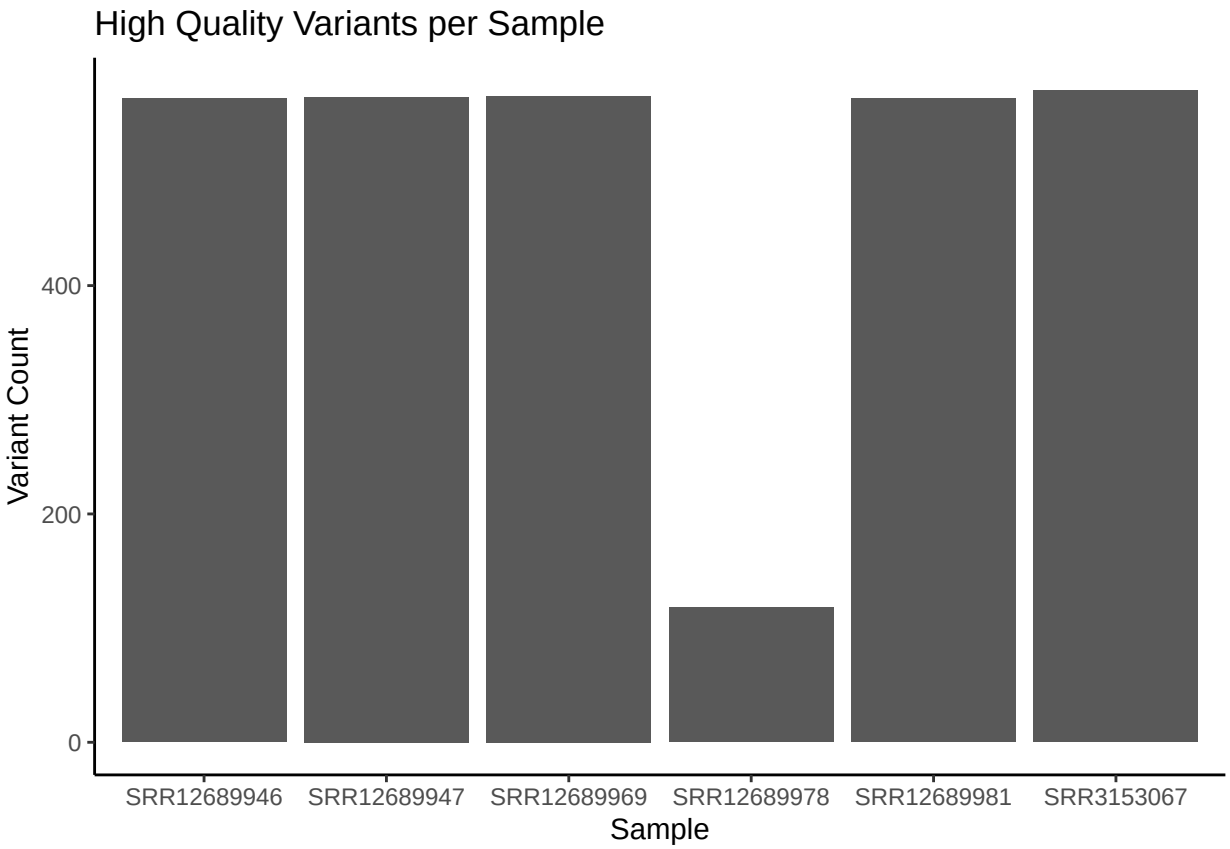
```r
#Calculate and plot the total number of high quality variants per sample

high_qual_vars <- data_allvariants %>%
  group_by(Sample) %>%
  summarise(count = n(), .groups = "drop")

high_qual_vars
```

```
## # A tibble: 6 x 2
##   Sample      count
##   <chr>       <int>
## 1 SRR12689946   564
## 2 SRR12689947   565
## 3 SRR12689969   566
## 4 SRR12689978   118
## 5 SRR12689981   564
## 6 SRR3153067    571
```

```r
plot1 <- high_qual_vars %>%
ggplot(aes(x = Sample, y = count)) +
  geom_bar(stat = "identity") +
  theme(axis.text.x = element_text(angle = 45, hjust = 1)) +
  labs(title = "High Quality Variants per Sample",
       x = "Sample",
       y = "Variant Count") +
  theme_classic()
plot1
```




```
#Merge REF and ALT column to a new column mutation
data_allvariants <- data_allvariants %>%
  mutate(mutation = paste0(REF,ALT))
```

```
#Determine the type of each variant (Transition:A<->C, OR G<->C, or Transversion) and add them to a new
data_allvariants <- data_allvariants %>%
  mutate(Type = case_when(mutation %in% c("AT","TA","GC","CG") ~ "Transition",
                          mutation %in% c("AG", "GA", "AC", "CA", "TC", "CT", "TG", "GT") ~ "Transversi
```

```
#Analyze and plot the distribution of variant types per sample both count and proportions
Var_dist <- data_allvariants %>%
  na.omit() %>%
  group_by(Sample, Type) %>%
  summarise(count = n(), .groups = "drop") %>%
  mutate(prop = count/sum(count)*100)

Var_dist
```

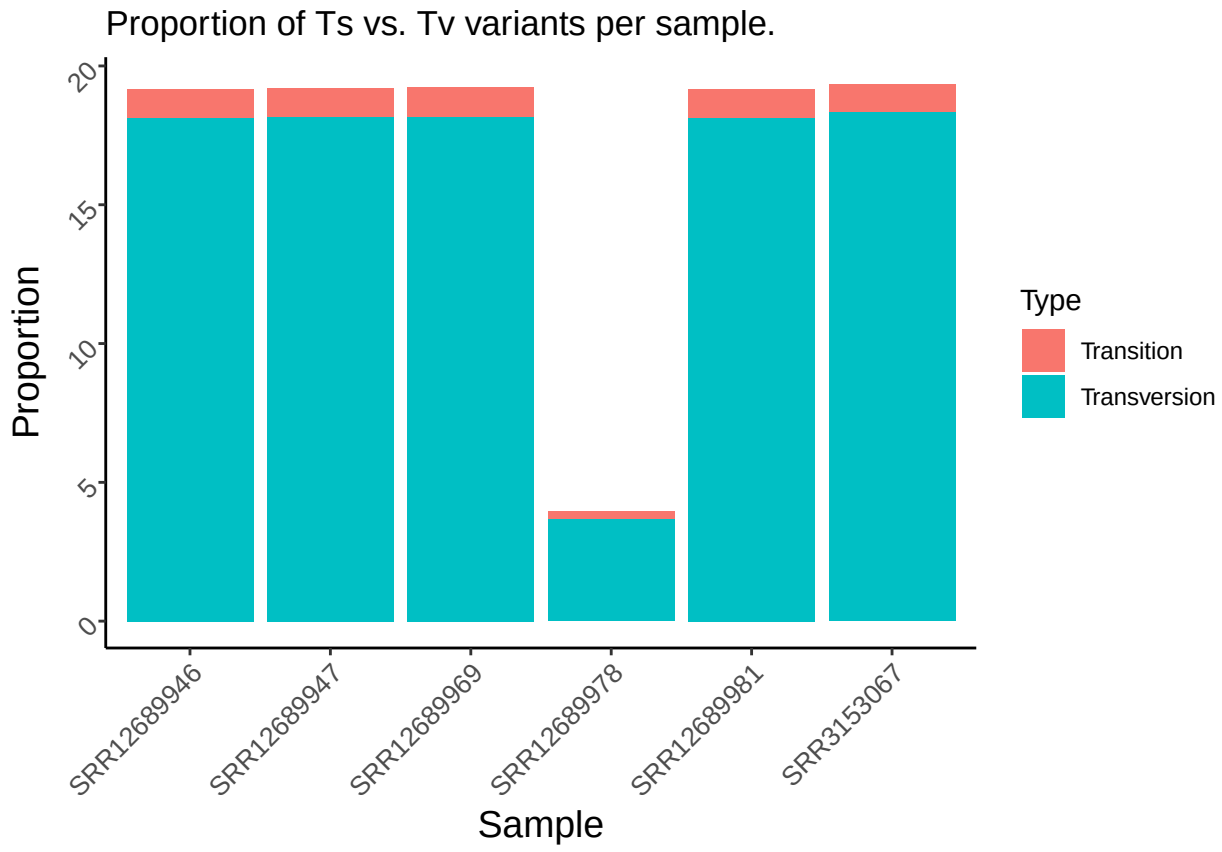
```
## # A tibble: 12 x 4
##    Sample      Type         count   prop
##    <chr>       <chr>        <int>  <dbl>
##  1 SRR12689946 Transition      30  1.02
##  2 SRR12689946 Transversion   534 18.1
##  3 SRR12689947 Transition      30  1.02
##  4 SRR12689947 Transversion   535 18.2
```

```
##  5 SRR12689969 Transition      31  1.05
##  6 SRR12689969 Transversion   535 18.2
##  7 SRR12689978 Transition       9  0.305
##  8 SRR12689978 Transversion   108  3.67
##  9 SRR12689981 Transition      30  1.02
## 10 SRR12689981 Transversion   534 18.1
## 11 SRR3153067  Transition      30  1.02
## 12 SRR3153067  Transversion   540 18.3
```

```r
plot2 <- Var_dist %>%
  ggplot(aes(x = Sample, prop, fill = Type)) +
  geom_bar(stat = "identity", position = "stack") +
  labs(title = "Proportion of Ts vs. Tv variants per sample.",
    x = "Sample",
    y = "Proportion"
  ) +
  theme_classic() +
  theme(
    axis.title = element_text(size =14),
    axis.text = element_text(size =10, angle = 45, hjust = 1)

  )

plot2
```



```r
#Identify common variants across samples or unique variants

## Create a unique identifier for each variant
data_allvariants$variant_id <- paste(data_allvariants$CHROM,
                                     data_allvariants$POS,
                                     data_allvariants$REF,
                                     data_allvariants$ALT,
                                     sep = "_")

## Keep one row per sample per variant (remove duplicates)
variant_sample_table <- data_allvariants %>%
  distinct(Sample, variant_id)

##Count how many samples each variant appears in
variant_counts <- variant_sample_table %>%
  group_by(variant_id) %>%
  summarise(sample_count = n(), .groups = "drop")

## Identify unique variants (appear in only one sample)
unique_variants <- variant_counts %>%
  filter(sample_count == 1)

## Get total number of samples
total_samples <- n_distinct(data_allvariants$Sample)

## Identify common variants (appear in all samples)
common_variants <- variant_counts %>%
  filter(sample_count == total_samples)
common_variants
```

```
## # A tibble: 114 x 2
##    variant_id            sample_count
##    <chr>                        <int>
##  1 NC_002549.1_10208_C_T            6
##  2 NC_002549.1_10566_T_C            6
##  3 NC_002549.1_10569_C_A            6
##  4 NC_002549.1_10575_T_C            6
##  5 NC_002549.1_10602_A_T            6
##  6 NC_002549.1_10624_T_C            6
##  7 NC_002549.1_10979_T_A            6
##  8 NC_002549.1_11043_A_G            6
##  9 NC_002549.1_11308_C_T            6
## 10 NC_002549.1_12096_A_G            6
## # i 104 more rows
```

```r
## Join back to see which sample each unique variant belongs to
unique_variants_with_samples <- unique_variants %>%
  inner_join(variant_sample_table, by = "variant_id")
unique_variants_with_samples
```

```
## # A tibble: 24 x 3
##    variant_id             sample_count Sample
##    <chr>                         <int> <chr>
```

```
##  1 NC_002549.1_11817_T_C             1 SRR3153067
##  2 NC_002549.1_12780_C_T             1 SRR3153067
##  3 NC_002549.1_12910_A_G             1 SRR3153067
##  4 NC_002549.1_12996_C_A             1 SRR3153067
##  5 NC_002549.1_14449_C_T             1 SRR12689946
##  6 NC_002549.1_16514_G_A             1 SRR3153067
##  7 NC_002549.1_16600_A_G             1 SRR3153067
##  8 NC_002549.1_17061_C_T,A           1 SRR12689978
##  9 NC_002549.1_18467_C_T             1 SRR12689981
## 10 NC_002549.1_18622_A_C             1 SRR12689947
## # i 14 more rows
```