FIREVAT Introduction

Andy Jinseok Lee, Hyunbin Kim

00. Using FIREVAT

00-1. Variant refinement

Using `RunFIREVAT`, you can perform variant refinement using mutational signatures. With 2 cores (each with 3.5GHz clock speed) this sample script takes about 10 minutes to run.

```
library(FIREVAT)

# Assign output directory
output.dir <- ""

sample.vcf.file <- system.file("extdata", "DCC_PCAWG_Cell_Lines_HCC1954.vcf", package = "FIREVAT")
config.file <- system.file("config", "PCAWG_DKFZ_Cell_Line_Filtering_Params.json", package = "FIREVAT")

results <- RunFIREVAT(vcf.file = sample.vcf.file,
                      vcf.file.genome = 'hg19',
                      config.file = config.file,
                      df.ref.mut.sigs = GetPCAWGMutSigs(),
                      target.mut.sigs = GetPCAWGMutSigsNames(),
                      sequencing.artifact.mut.sigs = PCAWG.All.Sequencing.Artifact.Signatures,
                      output.dir = output.dir,
                      objective.fn = Default.Obj.Fn,
                      num.cores = 2,
                      ga.pop.size = 100,
                      ga.max.iter = 5,
                      ga.run = 5,
                      ga.pmutation = 0.1,
                      perform.strand.bias.analysis = TRUE,
                      ref.forward.strand.var = "TumorDPRefForward",
                      ref.reverse.strand.var = "TumorDPRefReverse",
                      alt.forward.strand.var = "TumorDPAltForward",
                      alt.reverse.strand.var = "TumorDPAltReverse",
                      annotate = FALSE)
```

00-2. Manual filtering

You can also perform manual variant filtering.

```
## TODO NEEDS WORKS

library(FIREVAT)

# Assign output directory
output.dir <- "/home/jinseoklee/Documents/Projects/Temp/FIREVAT_TEMP/"

sample.vcf.file <- system.file("extdata", "DCC_PCAWG_Cell_Lines_HCC1954.vcf", package = "FIREVAT")
config.file <- system.file("config", "PCAWG_DKFZ_Cell_Line_Filtering_Params.json", package = "FIREVAT")
```

```
results <- RunFIREVAT(vcf.file = sample.vcf.file,
                      vcf.file.genome = 'hg19',
                      config.file = config.file,
                      mode = "manual",
                      df.ref.mut.sigs = GetPCAWGMutSigs(),
                      target.mut.sigs = GetPCAWGMutSigsNames(),
                      sequencing.artifact.mut.sigs = PCAWG.All.Sequencing.Artifact.Signatures,
                      output.dir = output.dir,
                      num.cores = 2,
                      perform.strand.bias.analysis = TRUE,
                      ref.forward.strand.var = "TumorDPRefForward",
                      ref.reverse.strand.var = "TumorDPRefReverse",
                      alt.forward.strand.var = "TumorDPAltForward",
                      alt.reverse.strand.var = "TumorDPAltReverse",
                      annotate = FALSE)
```

00-3. Mutational signature analysis

Using FIREVAT, you can run Mutalisk?

01. Introduction

FIREVAT (FInding REliable Variants without ArTifacts) uses mutational signatures to perform variant refinement and generates accurate signature analysis.

The R package FIREVAT provides

This is an R Markdown document. Markdown is a simple formatting syntax for authoring HTML, PDF, and MS Word documents. For more details on using R Markdown see http://rmarkdown.rstudio.com.

When you click the **Knit** button a document will be generated that includes both content as well as the output of any embedded R code chunks within the document. You can embed an R code chunk like this:

02. Installing FIREVAT

03. Inputs

03-X. Loading example data

03-X. Genetic Algorithm (GA) parameters

03-X. Objective function

Refer to 04-X.

03-X. Preparing your own VCF file

03-X. Preparing your own configuration file

03-X. Preparing your own mutational signature reference matrix

03-X. Selecting target mutational signatures by cancer type

03-X. Mutalisk parameters

Refer to 05-X. Mutalisk parameters

03-X. Variant annotation parameters

03-X-Y. Downloading ClinVar, COSMIC etc

03-X-Y. Preparing annotation data.frame

04. Variant refinement

04-X. Modes

04-Y. Optimization parameters

05. Mutational signature analysis

05-X. Mutalisk

05-X-Y. Mutalisk parameters

05-X-Y. Mutalisk results

05-X. MutationalPatterns

05-X-Y. MutationalPatterns parameters

05-X-Y. MutationalPatterns results

06. Strand bias analysis

06-1. Filtering variants by strand bias analysis results

07. Variant annotation

07-1. Annotating variants

07-2. White/black list

08. FIREVAT report

09. Generating individual FIREVAT report plots

10. Advanced FIREVAT examples

10-1. Additional plot generation

Cohort-level signature probability distribution plot Correlation plot Sequence logo plot Bubble chart plot

10-2. Custom pipeline

11. Session info