RHAPSODY WP3 Pre-Diabetes Analysis Plan

(Version 0.9.1)

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Files description

At this stage, you were provided with the following files from the (hidden) Gist (don't share outside RHAPSODY):

- README.Rmd, the file you are actually reading;
- RHAPSODY_WP3_PreDiab.Rmd, a Rmarkdown script which perform:
 - opal node access,
 - phenotype QC,
 - preliminary modelling,
 - VCFs formatting/filtering,
 - variants analysis;
- RHAPSODY_WP3_PreDiab_DEBUG.R, a small part of the main Rmarkdown script, which includes only node access and phenotype QC;
- RunAnalysis.sh, a shell script to run the whole analysis (parameters have to be filled properly according to your informations);
- handleVCF.sh, the shell script used to format/filter VCFs files prior to the variant analysis. This shell script is also included within the main script, but you can use it directly as a standalone, if so (or if your already done formatting and don't want to do it again), parameter format_vcfs must be set to FALSE).
- opal_credentials.txt, the server, login and password informations to access phenotype data on your local node;
- Install_Rpackages.R, a R script which install a predefined version of all R packages loaded in the Rmarkdown script, only if needed (also avaiable within the main script).

Run the analysis

R Setup

Please use R version 3.5.1 (2018-07-02) (if not possible, newer version "3.4.*" can be used). If VCFtools is not available on your machine, please install it: VCFtools. Be sure the needed packages are installed (this is also done within the Rmarkdown script) or copy/paste the following command. session_info.csv provided should be used, by default in the working directory.

```
check_packages_version <- function(list_packages, session_info_csv = "session_info.csv") {</pre>
  check_packages <- function(package) {</pre>
    if (!package%in%installed.packages()[, "Package"]) {
      install.packages(
            pkgs = package,
            repos = c(
              "https://rhap-fdb01.vital-it.ch/repo/",
              "http://cran.us.r-project.org",
              "https://cran.rstudio.com/",
              "http://cran.obiba.org"
          ),
            dependencies = TRUE
    }
    library(
      package = package,
      character.only = TRUE,
      quietly = TRUE,
      warn.conflicts = FALSE
  }
  invisible(sapply(c("devtools", "tidyverse"), check_packages))
  read_csv(file = session_info_csv) %>%
    dplyr::mutate(
      local_version = purrr::map(
        .x = package,
        .f = function(x)  {
          if (x %in% utils::installed.packages()[, 'Package']) {
            utils::packageDescription(pkg = x, fields = 'Version')
        }
      )
    filter(local_version!=version) %>%
    dplyr::mutate(
      install = purrr::map2(
        .x = package,
        y = version,
        .f = function(x, y) {
          devtools::install_version(
            package = x,
            version = as.character(y),
            repos = c(
```

```
"https://rhap-fdb01.vital-it.ch/repo/",
                  "http://cran.us.r-project.org",
                  "https://cran.rstudio.com/",
                  "http://cran.obiba.org"
          )
       }
      )
    )
  invisible(sapply(list_packages, check_packages))
  tidyverse::tidyverse_conflicts()
}
list_packages <- c(</pre>
 "devtools",
  "parallel",
  "grid",
  "scales",
  "broom",
  "viridis",
  "readxl",
  "writexl",
  "cowplot",
  "knitr",
  "kableExtra",
  "lme4",
  "lmerTest",
  "Hmisc",
  "data.tree",
  "opal",
  "tidyverse"
check_packages_version(
 list_packages = list_packages,
  session_info_csv = "session_info.csv"
)
```

Run the Rmarkdown script

- 1. First, create (or change) the opal_credentials.txt with your informations.
- 2. Fill in the proper informations.
- 3. Set the analysis_step:
 - 1. Node access
 - 2. Phenotype QC
 - 3. Run Mixed Model without variants
 - 4. Check VCFs files
 - 5. Format VCFs files
 - 6. Run Mixed Model with (imputed) variants
 - 7. Check output files
- 4. Run the R code below within interactive R session.
- 5. a/ If everything worked, you should have a HTML report. Increment analysis step and repeat steps 3 to 5.

b/ If something went wrong, you should have an error message, for example:

```
label: list_vcf (with options)
List of 2
$ eval : language params_steps$step_5
$ engine: chr "sh"

Quitting from lines 1597-1598 (RHAPSODY_WP3_PreDiab.Rmd)
Error in if (options$eval) { : argument is of length zero
```

Either try to solve the issue yourself and/or send the informations to mickael.canouil@cnrs.fr

```
# set the output directory or leave as is;
# output will be generated where the Rmarkdown file is
working directory <- './'
reportName <- 'RHAPSODY_WP3_PreDiab'</pre>
# Analysis
rmarkdown::render(
  input = "RHAPSODY_WP3_PreDiab.Rmd",
  output_format = "html_document",
  output_file = paste0(reportName, ".html"),
  output_dir = working_directory,
  params = list(
    cohort_name = 'CohortName',
    author_name = 'Firstname LASTNAME',
    opal_credentials = 'opal_credentials.txt',
   vcf_input_directory = './vcfs',
   imputation_quality_tag = 'INFO', # To be set according to VCF (could also be "R2")
   vcftools_binary_path = '/usr/local/bin',
    output_directory = working_directory,
    analysis_step = 1,
   format vcfs = TRUE,
   variants_analysis = TRUE,
    n_{cpu} = 2,
    echo = FALSE, # Should R code be printed in the report
   warning = TRUE, # Should warnings be printed in the report
   message = FALSE # Should messages be printed in the report
  ),
  encoding = "UTF-8"
```