

Gviz visualization: preparing GRanges object

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Installation of BiocManager and Gviz (= Bioconductor package)

```
if (!requireNamespace("BiocManager", quietly = TRUE)){
  install.packages("BiocManager")}
if (! requireNamespace("Gviz", quietly = TRUE)) {
  BiocManager::install("Gviz")
}
library(BiocManager)
```

Loading of Gviz package

```
library(Gviz)
library(GenomicRanges)
```

Writing function to create a table with GRCh37 coordinates

```
## separate function requires tidy package
library(tidyr)
Gviz_table <- function(filename) {
  X <- read.csv(file=filename, header = TRUE, sep=";")
  ##### GRCh37 coordinates in first assembly
  tablepart1 <- X[grep("^GRCh37", X$assembly1),]
  id1 <- tablepart1[1]
  Chr_1a <- tablepart1[["Chr_1"]]
  Chr_1a <- gsub("[A-Za-z]", "", Chr_1a)
  Chr_1a <- sub("", "chr", Chr_1a)
  assembly1 <- tablepart1[["assembly1"]]
  assembly1 <- gsub(".*?:", "", assembly1)
  assembly1 <- separate(data = as.data.frame(assembly1),
    col = assembly1, into = c("start", "end"), sep = "-")
  GRCh37_assembly1 <- data.frame(id1, Chr_1a, assembly1)
  names(GRCh37_assembly1)[names(GRCh37_assembly1) == "assembly1"] <- "assembly"
  names(GRCh37_assembly1)[names(GRCh37_assembly1) == "Chr_1a"] <- "chr"
  ##### GRCh37 coordinates in second assembly
  tablepart2 <- X[grep("^GRCh37", X$assembly2),]
  id2 <- tablepart2[1]
```

```

Chr_1b <- tablepart2[["Chr_2"]]
Chr_1b <- gsub("[A-Za-z]", "", Chr_1b)
Chr_1b <- sub("", "chr", Chr_1b)
assembly2 <- tablepart2[["assembly2"]]
assembly2 <- gsub(".*?:", "", assembly2)
assembly2 <- separate(data = as.data.frame(assembly2),
                      col = assembly2, into = c("start", "end"), sep = "-")
GRCh37_assembly2 <- data.frame(id2, Chr_1b, assembly2)
names(GRCh37_assembly2)[names(GRCh37_assembly2) == "assembly2"] <- "assembly"
names(GRCh37_assembly2)[names(GRCh37_assembly2) == "Chr_1b"] <- "chr"
##### Gviz table
GRCh37_Gviz <- rbind(GRCh37_assembly1, GRCh37_assembly2)
}

```

Converting tables to GRanges objects

No coordinates were available for inversions.

```

### results-CNV-dbVar.csv
CNVdbVar <- Gviz_table("results-CNV-dbVar.csv")
makeGRangesFromDataFrame(CNVdbVar)
### results-inversion-dbVar.csv
#inversiondbVar <- Gviz_table("results-inversion-dbVar.csv")
### results-insertion-dbVar.csv
insertiondbVar <- Gviz_table("results-insertion-dbVar.csv")
makeGRangesFromDataFrame(insertiondbVar)
### results-STR-dbVar.csv
STRdbVar <- Gviz_table("results-STR-dbVar.csv")
makeGRangesFromDataFrame(STRdbVar)
### results-ClinVar.csv
ClinVar <- Gviz_table("results-ClinVar.csv")
makeGRangesFromDataFrame(ClinVar)

```