# An Introduction to biovizBase

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## 1 Introduction

The biovizBase package is designed to provide a set of utilities and color schemes serving as the basis for visualizing biological data, especially genomic data. Two other packages are currently built on this package, a static version of graphics is provided by the package ggbio, and an interactive version of graphics is provided by visnab (Currently not released).

In this vignette, we will introduce those color schemes and different utilities functions using simple examples and data sets. Utilities includes functions that precess the raw data, validate names, add attributes, and generate summaries such as fragment length, GC content, and mismatch information.

## 2 Color Schemes

The biovizBase package aims to provide a set of default color schemes for biological data, based on the following principles.

- Make biological sense. Data is displayed in a way that is similar to observed results under the microscope. (Example: giemsa stain results)
- Generate aesthetically pleasing colors based on well-defined color sets like *color brewer* <sup>1</sup>. Produce the appropriate color for *sequential*, *diverging*, and *qualitative* color schemes.
- Accommodate colorblind vision by creating color pallets that pass the color blind check on the Vischeck website <sup>2</sup> or use palette from package dichromat or use color-blind safe color palette checked by ColorBrewer website<sup>3</sup>. There are three types of colorblind checking strategy defined on these website.

**Deuteranope** a form of red/green color deficit;

**Protanope** another form of red/green color deficit;

**Tritanope** a blue/yellow deficit- very rare.

Our color scheme try to pass color-blind checking points to make sure all the users can tell the difference between groups of data displayed. To make the implementation easy, we most time just use dichromat to check this, dichromat collapses red-green color distinctions to approximate the effect of the two common forms of red/green color blindness, protanopia and deuteranopia. Or we could simply implement proved color-blind safe palette from dichromat or RColorBrewer.

All color schemes have a general color generating function and a default color generating function. They are automatically stored in options as default when loading the package. Other packages built on biovizBase can use the default color scheme, ensuring consistent color themes across all static and interactive graphics. Users may also change the default color in the options to personalize the global color scheme to fit their needs.

```
> library(biovizBase)
> ## library(scales)
>
```

#### 2.1 Colorblind Safe Palette

For graphics, it's important to make sure most people can tell the difference between colors on the plots, even for people with deficient or anomalous red-green vision.

<sup>1</sup>http://colorbrewer2.org/

<sup>2</sup>http://www.vischeck.com/

<sup>3</sup>http://colorbrewer2.org/

We will add more and more colorblind safe palette gradually, now we only supported palettes from two packages, dichromat or RColorBrewer. However, RColorBrewer doesn't provide information about colorblind palette. So we need to check manually on ColorBrewer website, and add this information with the palette information. For dichromat package, it doesn't have a palette information like brewer.pal.info, which contains three different types, qual, div, seq representing quality, divergent and sequential respectively, and also missing max colors information, so we integrate all these information and generate three palette information.

- brewer.pal.blind.info provides only colorblind safe palette subset.
- dichromat.pal.blind.info provides colorblind safe palette with category information and max color allowed.
- blind.pal.info integrate first two, provides a general palette information with extra column like pal.id, which used for function colorBlindSafePal as index for arguments palette or maxcolors for allowed number of color. *pkg* providing information about which package it is defined.

#### > head(blind.pal.info)

|                 | ${\tt maxcolors}$ | category | pkg                  | pal.id |
|-----------------|-------------------|----------|----------------------|--------|
| BluetoGray.8    | 8                 | div      | dichromat            | 1      |
| BluetoOrange.8  | 8                 | div      | dichromat            | 2      |
| BrowntoBlue.10  | 10                | div      | dichromat            | 3      |
| BluetoOrange.10 | 10                | div      | dichromat            | 4      |
| PiYG            | 11                | div      | ${\tt RColorBrewer}$ | 5      |
| PRGn            | 11                | div      | RColorBrewer         | 6      |

Then we defined a color generating function colorBlindSafePal, this function reading in a palette argument which could be a index number or names for palette defined in blind.pal.info. And return a color generating function, a repeatable argument will control, for number over max color numbers required, does it simply repeat it or just providing limited number of colors.

- > ## with no arguments, return blind.pal.info
- > head(colorBlindSafePal())

|                 | ${\tt maxcolors}$ | category | pkg                  | pal.id |
|-----------------|-------------------|----------|----------------------|--------|
| BluetoGray.8    | 8                 | div      | dichromat            | 1      |
| BluetoOrange.8  | 8                 | div      | dichromat            | 2      |
| BrowntoBlue.10  | 10                | div      | dichromat            | 3      |
| BluetoOrange.10 | 10                | div      | dichromat            | 4      |
| PiYG            | 11                | div      | ${\tt RColorBrewer}$ | 5      |
| PRGn            | 11                | div      | RColorBrewer         | 6      |

- > ##
- > mypalFun <- colorBlindSafePal("Set2")</pre>
- > ## mypalFun(12, repeatable = FALSE) #only three
- > mypalFun(11, repeatable = TRUE) #repeat

```
[1] "#66C2A5" "#FC8D62" "#8DAOCB" "#66C2A5" "#FC8D62" "#8DAOCB"
```

To Collapses red-green color distinctions to approximate the effect of the two common forms of red-green color blindness, protanopia and deuteranopia, we can use function dichromat from package dichromat, this save us the time to

We only show this as an examples and won't compare all other color schemes in the following sections. Please notice that

<sup>[7] &</sup>quot;#66C2A5" "#FC8D62" "#8DA0CB" "#66C2A5" "#FC8D62"

```
> ## for palette "Paried"
> mypalFun <- colorBlindSafePal(21)
> par(mfrow = c(1, 3))
> showColor(mypalFun(4))
> library(dichromat)
> showColor(dichromat(mypalFun(4), "deutan"))
> showColor(dichromat(mypalFun(4), "protan"))
```

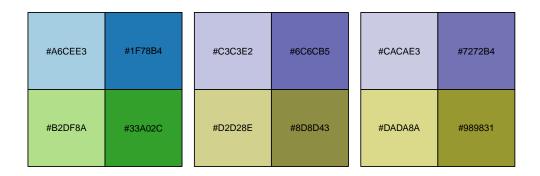


Figure 1: Checking colors with two common type of color blindness. The first one is normal perception, second one for deuteranopia and last one for protanopia. Since we are using selected color palettes in this package, it should be fine with those types of blindness.

- If the categorical data contains many levels like amino acid, people cannot easily tell the difference anyway, we did the trick to simply repeat the colors. This might be useful for many other cases like grand linear view for chromosomes, since if the viewed orders of chromosomes is fixed it's OK to use repeated colors since they are not going to be layout as neighbors anyway.
- For schemes like cytobands, we try to follow the biological sense, in this case, we don't really check the color blindness.

#### 2.2 Cytobands Color

Chemically staining the metaphase chromosomes results in a alternating dark and light banding pattern, which could provide information about abnormalities for chromosomes. Cytogenetic bands could also provide potential predictions of chromosomal structural characteristics, such as repeat structure content, CpG island density, gene density, and GC content.

biovizBase package provides utilities to get ideograms from the UCSC genome browser, as a wrapper around some functionality from rtracklayer. It gets the table for cytoBand and stores the table for certain species as a GRanges object.

We found a color setting scheme in package *qeneplotter*, and we implemented it in biovisBase.

The function .cytobandColor will return a default color set. You could also get it from options after you load biovizBase package.

And we recommended function <code>getBioColor</code> to get the color vector you want, and names of the color is biological categorical data. This function hides interval color genenerators and also the complexity of getting color from options. You could specify whether you want to get colors by default or from options, in this way, you can temporarily edit colors in options and could change or all the graphics. This give graphics a uniform color scheme.

#### > getOption("biovizBase")\$cytobandColor

| gpos2     | gpos1     | gvar      | gpos      | acen      | stalk     | gneg      |
|-----------|-----------|-----------|-----------|-----------|-----------|-----------|
| "#FCFCFC" | "#FFFFFF" | "grey0"   | "grey0"   | "brown4"  | "brown3"  | "grey100" |
| gpos9     | gpos8     | gpos7     | gpos6     | gpos5     | gpos4     | gpos3     |
| "#EAEAEA" | "#ECECEC" | "#EFEFEF" | "#F2F2F2" | "#F4F4F4" | "#F7F7F7" | "#F9F9F9" |
| gpos16    | gpos15    | gpos14    | gpos13    | gpos12    | gpos11    | gpos10    |
| "#D8D8D8" | "#DADADA" | "#DDDDDD" | "#E0E0E0" | "#E2E2E2" | "#E5E5E5" | "#E7E7E7" |
| gpos23    | gpos22    | gpos21    | gpos20    | gpos19    | gpos18    | gpos17    |
| "#C6C6C6" | "#C8C8C8" | "#CBCBCB" | "#CECECE" | "#DODODO" | "#D3D3D3" | "#D5D5D5" |
| gpos30    | gpos29    | gpos28    | gpos27    | gpos26    | gpos25    | gpos24    |
| "#B4B4B4" | "#B6B6B6" | "#B9B9B9" | "#BCBCBC" | "#BEBEBE" | "#C1C1C1" | "#C3C3C3" |
| gpos37    | gpos36    | gpos35    | gpos34    | gpos33    | gpos32    | gpos31    |
| "#A2A2A2" | "#A4A4A4" | "#A7A7A7" | "#AAAAAA" | "#ACACAC" | "#AFAFAF" | "#B1B1B1" |
| gpos44    | gpos43    | gpos42    | gpos41    | gpos40    | gpos39    | gpos38    |
| "#909090" | "#929292" | "#959595" | "#979797" | "#9A9A9A" | "#9D9D9D" | "#9F9F9F" |
| gpos51    | gpos50    | gpos49    | gpos48    | gpos47    | gpos46    | gpos45    |
| "#7E7E7E" | "#808080" | "#838383" | "#858585" | "#888888" | "#8B8B8B" | "#8D8D8D" |
| gpos58    | gpos57    | gpos56    | gpos55    | gpos54    | gpos53    | gpos52    |
|           | "#6E6E6E" |           |           |           |           | "#7B7B7B" |
| gpos65    | gpos64    | gpos63    | gpos62    | gpos61    | gpos60    | gpos59    |
| "#5A5A5A" | "#5C5C5C" | "#5F5F5F" | "#616161" | "#646464" | "#676767" | "#696969" |
|           | gpos71    |           |           |           |           | gpos66    |
|           | "#4A4A4A" |           |           |           |           | "#575757" |
|           | gpos78    |           |           |           |           | gpos73    |
| "#363636" | "#383838" | "#3B3B3B" | "#3D3D3D" | "#404040" | "#424242" | "#454545" |

```
gpos82
                                gpos83
   gpos80
             gpos81
                                           gpos84
                                                      gpos85
                                                                gpos86
"#333333" "#303030" "#2E2E2E" "#2B2B2B" "#292929" "#262626" "#242424"
   gpos87
                       gpos89
                                           gpos91
                                                      gpos92
                                                                gpos93
             gpos88
                                 gpos90
"#212121" "#1E1E1E" "#1C1C1C" "#191919" "#171717" "#141414" "#121212"
   gpos94
                       gpos96
                                gpos97
                                           gpos98
                                                      gpos99
             gpos95
"#0F0F0F" "#0C0C0C" "#0A0A0A" "#070707" "#050505" "#020202" "#000000"
> getBioColor("CYTOBAND")
              stalk
                         acen
                                   gpos
                                             gvar
                                                       gpos1
                                                                 gpos2
"grey100"
           "brown3"
                     "brown4"
                                "grey0"
                                          "grey0" "#FFFFFF" "#FCFCFC"
    gpos3
                                            gpos7
              gpos4
                        gpos5
                                 gpos6
                                                       gpos8
                                                                 gpos9
"#F9F9F9" "#F7F7F7" "#F4F4F4" "#F2F2F2" "#EFEFEF" "#ECECEC" "#EAEAEA"
   gpos10
             gpos11
                       gpos12
                                 gpos13
                                           gpos14
                                                      gpos15
                                                                gpos16
"#E7E7E7" "#E5E5E5" "#E2E2E2" "#E0E0E0" "#DDDDDD" "#DADADA" "#D8D8D8"
   gpos17
             gpos18
                       gpos19
                                 gpos20
                                            gpos21
                                                      gpos22
                                                                gpos23
"#D5D5D5" "#D3D3D3" "#D0D0D0" "#CECECE" "#CBCBCB" "#C8C8C8" "#C6C6C6"
   gpos24
                                                      gpos29
             gpos25
                       gpos26
                                 gpos27
                                            gpos28
                                                                gpos30
"#C3C3C3" "#C1C1C1" "#BEBEBE" "#BCBCBC" "#B9B9B9" "#B6B6B6" "#B4B4B4"
   gpos31
             gpos32
                       gpos33
                                 gpos34
                                            gpos35
                                                      gpos36
                                                                gpos37
"#B1B1B1" "#AFAFAF" "#ACACAC" "#AAAAAA" "#A7A7A7" "#A4A4A4" "#A2A2A2"
   gpos38
             gpos39
                       gpos40
                                 gpos41
                                           gpos42
                                                      gpos43
                                                                gpos44
"#9F9F9F" "#9D9D9D" "#9A9A9A" "#979797" "#959595" "#929292" "#909090"
   gpos45
                       gpos47
                                 gpos48
                                           gpos49
                                                      gpos50
             gpos46
                                                                gpos51
"#8D8D8D" "#8B8B8B" "#888888" "#858585" "#838383" "#808080" "#7E7E7E"
   gpos52
             gpos53
                       gpos54
                                gpos55
                                           gpos56
                                                      gpos57
                                                                gpos58
"#7B7B7B" "#797979" "#767676" "#737373" "#717171" "#6E6E6E" "#6C6C6C"
   gpos59
                                           gpos63
             gpos60
                       gpos61
                               gpos62
                                                      gpos64
                                                                gpos65
"#696969" "#676767" "#646464" "#616161" "#5F5F5F" "#5C5C5C" "#5A5A5A"
   gpos66
                                gpos69
                                           gpos70
             gpos67
                       gpos68
                                                      gpos71
                                                                gpos72
"#575757" "#545454" "#525252" "#4F4F4F" "#4D4D4D" "#4A4A4A" "#484848"
   gpos73
             gpos74
                       gpos75
                                           gpos77
                                                      gpos78
                                                                gpos79
                                gpos76
"#454545" "#424242" "#404040" "#3D3D3D" "#3B3B3B" "#383838" "#363636"
   gpos80
                       gpos82
                                 gpos83
                                           gpos84
                                                      gpos85
             gpos81
                                                                gpos86
"#333333" "#303030" "#2E2E2E" "#2B2B2B" "#292929" "#262626" "#242424"
                       gpos89
   gpos87
             gpos88
                                 gpos90
                                            gpos91
                                                      gpos92
                                                                gpos93
"#212121" "#1E1E1E" "#1C1C1C" "#191919" "#171717" "#141414" "#121212"
                       gpos96
                                 gpos97
   gpos94
             gpos95
                                                      gpos99
                                                               gpos100
                                           gpos98
"#0F0F0F" "#0C0C0C" "#0A0A0A" "#070707" "#050505" "#020202" "#000000"
> ## differece source from default or options.
> opts <- getOption("biovizBase")</pre>
> opts$DNABasesNColor[1] <- "red"
> options(biovizBase = opts)
> ## get from option(default)
> getBioColor("DNA_BASES_N")
```

A T G C N "red" "#2C7BB6" "#D7191C" "#FDAE61" "#FFFFBF"

<sup>&</sup>gt; ## get default fixed color

<sup>&</sup>gt; getBioColor("DNA\_BASES\_N", source = "default")

```
A T G C N
"#ABD9E9" "#2C7BB6" "#D7191C" "#FDAE61" "#FFFBF"

> seqs <- c("A", "C", "T", "G", "G", "C")

> ## get colors for a sequence.

> getBioColor("DNA_BASES_N")[seqs]

A C T G G G C
"red" "#FDAE61" "#2C7BB6" "#D7191C" "#D7191C" "#D7191C" "#FDAE61"
```

You can check the color scheme by calling the plotColorLegend function. or the showColor.

> cols <- getBioColor("CYTOBAND")
> plotColorLegend(cols, title = "cytoband")

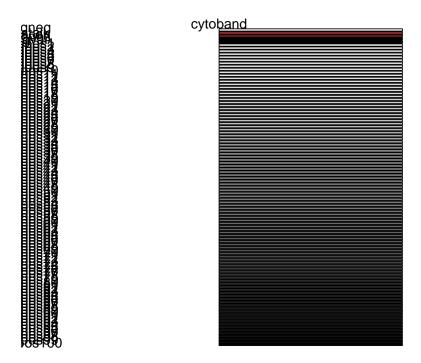


Figure 2: Legend for cytoband color

## 2.3 Strand Color

In the GRanges object, we have strand which contains three levels, +, -, \*. We are using a qualitative color set from *Color Brewer* and check with *dichromat* as Figure 3 shows, and we can see that this color set passes all three types of colorblind test. Therefore it should be a safe color set to use to color strand.

<u>(1)</u>

```
> par(mfrow = c(1, 3))
> cols <- getBioColor("STRAND")
> showColor(cols)
> showColor(dichromat(cols, "deutan"))
> showColor(dichromat(cols, "protan"))
```

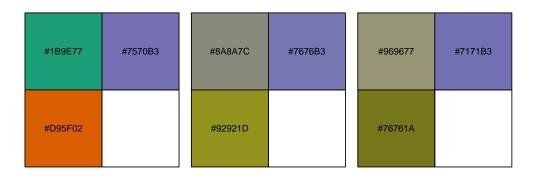


Figure 3: Colorblind vision check for color of strand

#### 2.4 Nucleotides Color

We start with the five most used nucleotides,  $\mathbf{A}, \mathbf{T}, \mathbf{C}, \mathbf{G}, \mathbf{N}$ , most genome browsers have their own color scheme to represent nucleotides, We chose our color scheme based on the principles introduced above. Since in genetics, GC-content usually has special biological significance because GC pair is bound by three hydrogen bonds instead of two like AT pairs. So it has higher thermostability which could result in different significance, like higher annealing temperature in PCR. So we hope to choose warm colors for  $\mathbf{G}, \mathbf{C}$  and cold colors for  $\mathbf{A}, \mathbf{T}$ , and a color in between to represent  $\mathbf{N}$ . They are chosen from a diverging color set of color brewer. So we should be able to easily tell the GC enriched region. Figure 4 shows the results from dichromat, and we can see this color set passes all two types of the colorblind test. It should be a safe color set to use to color the five most used nucleotides.

```
> getBioColor("DNA_BASES_N")

A         T         G         C         N
"red" "#2C7BB6" "#D7191C" "#FDAE61" "#FFFFBF"
>
```

#### 2.5 Amino Acid Color and Other Schemes

We also include some other color schemes created based on existing object in package *Biostrings* and other customized color scheme. Please notice that the object name is not the same as the name in the options. On the left of =, it's name of object, most of them are defined in *Biostrings* and on the right, it's the name in options.

```
DNA_BASES_N = "DNABasesNColor"

DNA_BASES = "DNABasesColor"

DNA_ALPHABET = "DNAAlphabetColor"

RNA_BASES_N = "RNABasesNColor"

RNA_BASES = "RNABasesColor"

RNA_ALPHABET = "RNAAlphabetColor"

IUPAC_CODE_MAP = "IUPACCodeMapColor"

AMINO_ACID_CODE = "AminoAcidCodeColor"

AA_ALPHABET = "AAAlphabetColor"

STRAND = "strandColor"

CYTOBAND = "cytobandColor"
```

They all could be retrieved by calling function getBioColor.

#### 2.6 Future Schemes

Current color schemes are most generated based on known object in R, which has a clear definition and classification. But we do have more interesting events or biological significance need to be color coded. Like most genome browser, they try to color code many events, for instance, color the insertion size which is larger/smaller than the estimated size; for paired RNA-seq data, we may color the paired reads mapped to a different chromosome.

We may include more color coded events in this package in next release.

```
> par(mfrow = c(1, 3))
> cols <- getBioColor("DNA_BASES_N", "default")
> showColor(cols, "name")
> cols.deu <- dichromat(cols, "deutan")
> names(cols.deu) <- names(cols)
> cols.pro <- dichromat(cols, "protan")
> names(cols.pro) <- names(cols)
> showColor(cols.deu, "name")
> showColor(cols.pro, "name")
```

| А | т | G | А | Т | G | А | т | G |
|---|---|---|---|---|---|---|---|---|
| С | N |   | С | N |   | С | N |   |

Figure 4: Colorblind vision check for color of nucleotide

## 3 Utilities

biovizBase serves as a basis for the visualization of biological data, especially for genomic data. IRanges and GenomicRanges are the two most important infrastructure packages to manipulate genomic data. They already have lots of useful and fast utilities for processing genomic data. Some other package such as rtracklayer, Rsamtools, ShortRead, GenomicFeatures provide common I/O for certain types of biological data and utilities for processing those raw data. Most of our utilities to be introduced in this section only manipulate the data in a simple and different way to get them ready for visualization. Most cases are only useful for visualization work, like adding brush color attributes to a GRanges object. Some of the other utilities are responsible for summarizing certain types of raw data, getting it ready to be visualized. Some of those utilities may be moved to a separate package later.

## 3.1 GRanges Related Manipulation

biovizBase mainly focuses on visualizing the genomic data, so we have some utilities for manipulating GRanges object. We are going to introduce these functions in the flow wing sub-sections. Overall, we hope to reduce people's work through these common utilities.

#### 3.1.1 Adding Disjoint Levels

```
> library(GenomicRanges)
> set.seed(1)
> N <- 500
> gr <- GRanges(seqnames =
                sample(c("chr1", "chr2", "chr3", "chrX", "chrY"),
                       size = N, replace = TRUE),
                IRanges (
                        start = sample(1:300, size = N, replace = TRUE),
                        width = sample(70:75, size = N, replace = TRUE)),
                strand = sample(c("+", "-", "*"), size = N,
                  replace = TRUE),
                value = rnorm(N, 10, 3), score = rnorm(N, 100, 30),
                group = sample(c("Normal", "Tumor"),
                  size = N, replace = TRUE),
                pair = sample(letters, size = N,
                  replace = TRUE))
```

This is a tricky question. For example, for pair-end RNA-seq data, we may want to put the reads with the same *qname* on the same level, with nothing falling in between. For better visualization of the data, we may hope that adding invisible extensions to the reads will prevent closely neighbored reads from showing up on the same level.

addStepping function takes a GenomicRanges object and will add an extra column called .levels to the object. This function is essentially a wrapper around a function disjointBins but allows a more flexible way to assign levels to each entry. For example, if the arguments group.name is specified to one of the column in elementMetadata, the function will make sure

- Grouped intervals are in the same levels (if they are not overlapped each other).
- No entry is following between the grouped intervals.
- If extend size is provided, it buffers the intervals and then computes the disjoint levels, thus ensuring that two closely positioned intervals will be assigned to different levels, a good practice for visualization.

For now, this function is only useful for visualization purposes.

### > head(addStepping(gr))

```
GRanges with 6 ranges and 5 metadata columns:
       seqnames
                    ranges strand |
                                         value
                                                                group
                                                   score
          <Rle>
                <IRanges>
                           <Rle> | <numeric> <numeric> <character>
           chr1 [160, 232]
                                - | 12.994463 118.45194
  chr1
                                                              Normal
  chr1
           chr1 [141, 213]
                                - | 9.350873
                                               92.11885
                                                              Normal
           chr1 [ 39, 113]
  chr1
                                * | 12.289738 65.92753
                                                               Tumor
           chr1 [242, 314]
  chr1
                                - |
                                     6.971489 102.75172
                                                              Normal
  chr1
           chr1 [ 84, 158]
                                + | 9.309577 133.85467
                                                               Tumor
  chr1
           chr1 [102, 174]
                                - | 12.610017 150.03296
                                                              Normal
              pair stepping
       <character> <numeric>
  chr1
                a
  chr1
                 У
                          19
  chr1
                          12
                 S
  chr1
                 u
                          24
  chr1
                           3
                 q
  chr1
                           9
                 У
  ___
  seqlengths:
  chr1 chr2 chr3 chrX chrY
     NA
          NA
               NA
                    NA
                         NA
> head(addStepping(gr, group.name = "pair"))
GRanges with 6 ranges and 5 metadata columns:
                    ranges strand |
       segnames
                                         value
                                                   score
                                                                group
          <Rle> <IRanges>
                            <Rle> | <numeric> <numeric> <character>
  chr1
           chr1 [160, 232]
                                - | 12.994463 118.45194
                                                              Normal
                                - | 9.350873 92.11885
  chr1
           chr1 [141, 213]
                                                              Normal
  chr1
           chr1 [ 39, 113]
                                * | 12.289738 65.92753
                                                               Tumor
           chr1 [242, 314]
                                - | 6.971489 102.75172
  chr1
                                                              Normal
                                + | 9.309577 133.85467
  chr1
           chr1 [ 84, 158]
                                                               Tumor
                                - | 12.610017 150.03296
  chr1
           chr1 [102, 174]
                                                              Normal
              pair stepping
       <character> <numeric>
  chr1
                а
  chr1
                          25
                 У
 chr1
                          19
                 s
  chr1
                 u
                          21
  chr1
                          17
                 q
  chr1
                          25
                 У
  seqlengths:
  chr1 chr2 chr3 chrX chrY
     NA
          NA
               NA
                    NA
> gr.close \leftarrow GRanges(c("chr1", "chr1"), IRanges(c(10, 20), width = 9))
> addStepping(gr.close)
GRanges with 2 ranges and 1 metadata column:
       seqnames
                  ranges strand | stepping
```

```
<Rle> <IRanges>
                             <Rle> | <numeric>
  chr1
            chr1
                  [10, 18]
                                              1
                  [20, 28]
                                 * |
                                              1
  chr1
           chr1
  seqlengths:
   chr1
     NA
> addStepping(gr.close, extend.size = 5)
GRanges with 2 ranges and 1 metadata column:
       seqnames
                    ranges strand |
                                       stepping
          <Rle> <IRanges>
                             <Rle> | <numeric>
  chr1
           chr1
                  [10, 18]
                                 * |
                                              1
  chr1
           chr1
                  [20, 28]
                                 * |
                                              2
  seqlengths:
   chr1
     NA
```

## 3.2 Shrink the Gaps

Sometime, in a gene centric view, we hope to truncate or shrink the gaps to better visualize the short reads or annotation data. It's **DANGEROUS** to shrink the gaps, since it only make sense in visualization. And even in the visualization the x-scale will be discontinued, and labels became somehow meaningless. **Make sure** you are not using the shrunk version of data when performing the down stream analysis.

This is a tricky question too, we hope to provide a flexible way to shrink the gaps. When we have multiple tracks, users would be responsible to shrink all the tracks based on the common gaps, otherwise there will be mis-aligned tracks.

maxGap computes a suitable estimated gap based on passed GenomicRanges

shrinkageFun function will read in a GenomicRanges object which represents the gaps, and returns a function which alters a different GenomicRanges object, to shrink that object based on previously specified gaps shrinking information. You could use this function to treat multiple tracks(e.g. GRanges) to make sure they are shrunk based on the common gaps and the same ratio.

Be careful in the following situations.

- When use the same shrinkage function to shrink multiple tracks, make sure the gaps passed to shrinkageFun function is the common gaps across all tracks, otherwise, it doesn't make sense to cut a overlapped gap within one of the tracks.
- The default max gap is not 0, just for visualization purpose. If for estimation purpose, you might want to make sure you cut all the gaps.

And notice, after shrinking, the x-axis labes only provide approximate position as shown in Figure 5 and 6, because it's clipped. It's just for visualization purpose.

```
> gr1 \leftarrow GRanges("chr1", IRanges(start = c(100, 300, 600),
                                  end = c(200, 400, 800))
> shrink.fun1 <- shrinkageFun(gaps(gr1), max.gap = maxGap(gaps(gr1), 0.15))
> shrink.fun2 <- shrinkageFun(gaps(gr1), max.gap = 0)</pre>
> head(shrink.fun1(gr1))
GRanges with 3 ranges and 1 metadata column:
      segnames
                   ranges strand |
         <Rle> <IRanges> <Rle> |
                                            <GRanges>
          chr1 [ 91, 191]
  [1]
                           * | chr1:*:[100, 200]
          chr1 [282, 382]
  [2]
                               * | chr1:*:[300, 400]
  [3]
          chr1 [473, 673]
                               * | chr1:*:[600, 800]
  seqlengths:
  chr1
     NA
> head(shrink.fun2(gr1))
GRanges with 3 ranges and 1 metadata column:
      seqnames
                   ranges strand |
         <Rle> <IRanges> <Rle> |
                                            <GRanges>
  [1]
          chr1 [ 1, 101]
                               * | chr1:*:[100, 200]
  [2]
          chr1 [102, 202]
                               * | chr1:*:[300, 400]
  [3]
          chr1 [203, 403]
                              * | chr1:*:[600, 800]
  seqlengths:
  chr1
     NA
> gr2 \leftarrow GRanges("chr1", IRanges(start = c(100, 350, 550),
                                  end = c(220, 500, 900)))
> gaps.gr <- intersect(gaps(gr1, start = min(start(gr1))),
                       gaps(gr2, start = min(start(gr2))))
> shrink.fun <- shrinkageFun(gaps.gr, max.gap = maxGap(gaps.gr))
> head(shrink.fun(gr1))
GRanges with 3 ranges and 1 metadata column:
      segnames
                   ranges strand |
                                                 .ori
         <Rle>
               <IRanges> <Rle> |
                                            <GRanges>
          chr1 [100, 200]
  [1]
                               * | chr1:*:[100, 200]
  [2]
          chr1 [222, 322]
                               * | chr1:*:[300, 400]
  [3]
          chr1 [474, 674]
                             * | chr1:*:[600, 800]
  seqlengths:
  chr1
     NA
> head(shrink.fun(gr2))
```

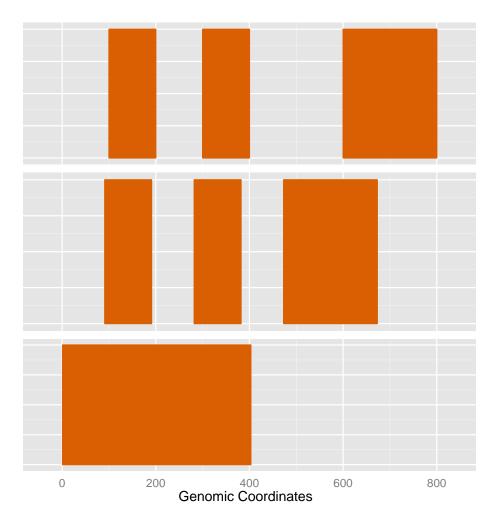


Figure 5: Shrink single GRanges. The first track is original GRanges, the second one use a ratio which shrink the GRanges a little bit, and default is to remove all gaps shown as the third track

```
GRanges with 3 ranges and 1 metadata column:
      seqnames
                   ranges strand |
                                                  .ori
         <Rle> <IRanges> <Rle> |
                                            <GRanges>
  [1]
          chr1 [100, 220]
                               * | chr1:*:[100, 220]
  [2]
          chr1 [272, 422]
                                * | chr1:*:[350, 500]
  [3]
          chr1 [424, 774]
                                * | chr1:*:[550, 900]
  seqlengths:
   chr1
     NA
```

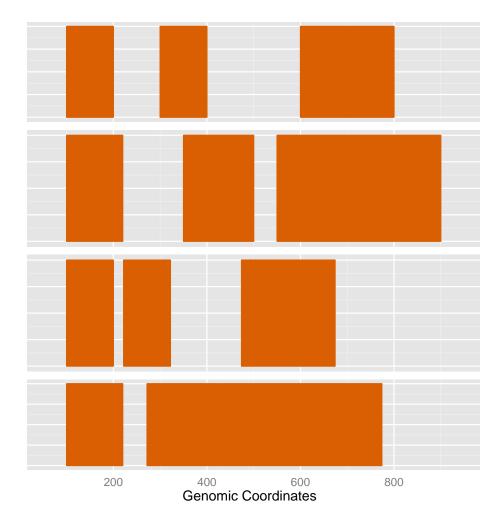


Figure 6: shrinkageFun demonstration for multiple GRanges, the top two tracks are the original tracks, please note how we clipped common gaps for those two tracks and shown as bottom two tracks.

#### 3.3 GC content

As mentioned before, GC content is an interesting variable which may be related to various biological questions. So we need a way to compute GC content in a certain region of a reference genome.

GCcontent function is a wrapper around getSeq function in *BSgenome* package and letterFrequency in *Biostrings* package. It reads a BSgenome object and returns count/probability for GC content in specified region.

```
> library(BSgenome.Hsapiens.UCSC.hg19)
> GCcontent(Hsapiens, GRanges("chr1", IRanges(1e6, 1e6 + 1000)))
> GCcontent(Hsapiens, GRanges("chr1", IRanges(1e6, 1e6 + 1000)), view.width = 300)
```

#### 3.4 Mismatch Summary

Compared to short-read alignment visualization, it's more useful to just show the summary of nucleotides of short reads per base and compare with the reference genome. We need a way to show the mismatched nucleotides, coverage at each position and proportion of mismatched nucleotides, and use the default color to indicate the type of nucleotide.

pileupAsGRanges function summarizes reads from bam files for nucleotides on single base units in a given region, which allows the downstream mismatch summary analysis. It's a wrapper around applyPileup function in Rsamtools package and more detailed control could be found under manual of PileupParam function in Rsamtools. pileupAsGRanges function returns a GRanges object which includes a summary of nucleotides, depth, and bam file path. This object could be read directly into the pileupGRangesAsVariantTable function for a mismatch summary.

This function returns a GRanges object with extra elementMetadata, counts for A,C,T,G,N and depth for coverage. bam indicates the bam file path. Each row is single base unit.

pileupGRangesAsVariantTable performs comparisons to the reference genome (a BSgenome object) and computes the mismatch summary for a certain region of reads. User need to make sure to pass the right reference genome to this function to get the right summary. This function drops the positions that have no reads and only keeps the regions with coverage in the summary. The result could be used to show stacked barchart for the mismatch summary.

This function returns a GRanges with the following elementMetadata information.

ref Reference base.

read Sequenced read at that position. Each type of A,C,T,G,N summarize counts at one position, if no counts detected, will not show it.

count Count for each nucleotide.

**depth** Coverage at that position.

match A logical value, indicate it's matched or not.

**bam** Indicate bam file path.

Sample raw data is from SRA(Short Read Archive), Accession: SRR027894 and subset the gene at chr10:6118023-6137427, which within gene RBM17. contains junction reads.

```
> library(Rsamtools)
> data(genesymbol)
> library(BSgenome.Hsapiens.UCSC.hg19)
> bamfile <- system.file("extdata", "SRR027894subRBM17.bam", package="biovizBase")
> test <- pileupAsGRanges(bamfile, region = genesymbol["RBM17"])
> test.match <- pileupGRangesAsVariantTable(test, Hsapiens)
> head(test[,-7])
> head(test.match[,-5])
```

### 3.5 Get an Ideogram

getIdeogram function is a wrapper of some functionality from rtracklayer to get certain table like cytoBand. A full table schema can be found here at UCSC genome browser. Please click describe table schema.

This function requires a network connection and will parse the data on the fly. The first argument of getIdeogram is species. If missing, the function will give you a choice hint, so you will not have to remember the name for the database you want, or you can simply get the database name for a different genome using the ucscGenomes function in Rtracklayer. The second argument subchr is used to subset the result by chromosome name. The third argument cytoband controls if you want to get the gieStain information/band information or not, which is useful for the visualization of the whole genome or single chromosome. You can see some examples in ggbio.

```
> library(rtracklayer)
```

- > hg19IdeogramCyto <- getIdeogram("hg19", cytoband = TRUE)
- > hg19Ideogram <- getIdeogram("hg19", cytoband = FALSE)
- > unknowIdeogram <- getIdeogram()</pre>

#### Please specify genome

```
1: hg19
              2: hg18
                            3: hg17
                                         4: hg16
                                                       5: felCat4
6: felCat3
              7: galGal3
                            8: galGal2
                                         9: panTro3
                                                      10: panTro2
                                        14: bosTau2
11: panTro1
             12: bosTau4
                           13: bosTau3
                                                      15: canFam2
16: canFam1
             17: loxAfr3
                           18: fr2
                                        19: fr1
                                                      20: cavPor3
21: equCab2
             22: equCab1
                           23: petMar1
                                        24: anoCar2
                                                      25: anoCar1
26: calJac3
                           28: oryLat2
                                        29: mm9
                                                      30: mm8
             27: calJac1
31: mm7
             32: monDom5
                           33: monDom4
                                        34: monDom1
                                                      35: ponAbe2
36: ailMel1
             37: susScr2
                                        39: oryCun2
                                                      40: rn4
                           38: ornAna1
41: rn3
             42: rheMac2
                           43: oviAri1
                                        44: gasAcu1
                                                      45: tetNig2
46: tetNig1
             47: xenTro2
                           48: xenTro1
                                        49: taeGut1
                                                      50: danRer7
51: danRer6
             52: danRer5
                           53: danRer4
                                        54: danRer3
                                                      55: ci2
56: ci1
             57: braFlo1
                           58: strPur2
                                        59: strPur1
                                                      60: apiMel2
61: apiMel1
             62: anoGam1
                           63: droAna2
                                                      65: droEre1
                                        64: droAna1
66: droGri1
             67: dm3
                           68: dm2
                                        69: dm1
                                                      70: droMoj2
71: droMoj1
             72: droPer1
                           73: dp3
                                        74: dp2
                                                      75: droSec1
76: droSim1
             77: droVir2
                           78: droVir1
                                        79: droYak2
                                                      80: droYak1
81: caePb2
             82: caePb1
                           83: cb3
                                        84: cb1
                                                      85: ce6
86: ce4
             87: ce2
                           88: caeJap1
                                        89: caeRem3
                                                      90: caeRem2
91: priPac1
             92: aplCall 93: sacCer2
                                        94: sacCer1
```

#### Selection:

Here is the example on how to get the genome names.

#### > head(ucscGenomes()\$db)

```
[1] hg19 hg18 hg17 hg16 felCat4 felCat3
122 Levels: ailMel1 anoCar1 anoCar2 anoGam1 apiMel1 apiMel2 ...
```

We put the most used hg19 ideogram as our default data set, so you can simply load it and see what they look like. They are all returned by the getIdeogram function. The one with cytoband information has two special columns.

name Name of cytogenetic band

#### gieStain Giemsa stain results

- > data(hg19IdeogramCyto)
- > head(hg19IdeogramCyto)

GRanges with 6 ranges and 2 metadata columns:

```
seqnames
                            ranges strand |
                                                 name gieStain
       <Rle>
                         <IRanges>
                                    <Rle> | <factor> <factor>
[1]
        chr1 [
                      0, 2300000]
                                         * |
                                               p36.33
                                                           gneg
[2]
        chr1 [ 2300000, 5400000]
                                               p36.32
                                                         gpos25
[3]
        chr1 [ 5400000, 7200000]
                                         * |
                                               p36.31
                                                           gneg
[4]
        chr1 [ 7200000, 9200000]
                                         * |
                                               p36.23
                                                         gpos25
        chr1 [ 9200000, 12700000]
[5]
                                         * |
                                               p36.22
                                                           gneg
[6]
        chr1 [12700000, 16200000]
                                               p36.21
                                                         gpos50
```

seqlengths:

```
chr1 chr10 chr11 chr12 chr13 chr14 ...
                                          chr7
                                                 chr8
                                                        chr9
                                                              chrX
                                                                    chrY
        NA
              NA
                     NA
                           NA
                                  NA ...
                                            NA
                                                   NA
                                                         NA
                                                                NA
                                                                      NA
```

- > data(hg19Ideogram)
- > head(hg19Ideogram)

GRanges with 6 ranges and 0 metadata columns:

```
seqnames
                                   ranges strand
                    <Rle>
                                <IRanges>
                                           <Rle>
[1]
                     chr1 [1, 249250621]
[2] chr1_gl000191_random [1,
                                  106433]
[3] chr1_gl000192_random [1,
                                  547496]
[4]
                     chr2 [1, 243199373]
[5]
                     chr3 [1, 198022430]
[6]
                     chr4 [1, 191154276]
seqlengths:
                   chr1
                         chr1_gl000191_random ...
             249250621
                                        106433 ...
```

There are two simple functions to test if the ideogram is valid or not. isIdeogram simply tests if the result came from the getIdeogram function, making sure it's a GenomicRanges object with an extra column. isSimpleIdeogram only tests if it's GenomicRanges and does not require cytoband information. But it double checks to make sure there is only one entry per chromosome. This is useful to show stacked overview for genomes. Please check some examples in *ggbio* to draw stacked overview and single chromosome.

chrM

16571

- > isIdeogram(hg19IdeogramCyto)
- [1] TRUE
- > isIdeogram(hg19Ideogram)
- [1] FALSE
- > isSimpleIdeogram(hg19IdeogramCyto)
- [1] FALSE
- > isSimpleIdeogram(hg19Ideogram)
- [1] TRUE

#### 3.6 Other Utilities and Data Sets

We are not going to introduce other utilities in this vignette, please refer to the manual for more details, we have other function to transform a GRanges to a special format only for graphic purpose, such as function transformGRangesForEvenSpace and transformGRangesToDfWithTicks could be used for grand linear view or linked view as introduced in package *qqbio*.

We have introduced data sets like hg19IdeogramCyto and hg19Ideogram in the previous sections. We also have a data set called genesymbol, which is extracted from human annotation package and stored as GRanges object, with extra columns symbol and ensemblid. For fast mapping, we use symbol as row names too.

This could be used for convenient overlapped subset with other annotation, and has potential use in a auto-complement drop list for gene search bar like most gene browsers have.

```
> data(genesymbol)
> head(genesymbol)
GRanges with 6 ranges and 2 metadata columns:
       segnames
                               ranges strand |
                                                     symbol
          <Rle>
                            <IRanges>
                                       <Rle> | <character>
  A1BG
          chr19 [58858174, 58864865]
                                            - |
                                                       A1BG
   A2M
          chr12 [ 9220304, 9268558]
                                            - 1
                                                        A2M
           chr8 [18027971, 18081197]
 NAT1
                                            + |
                                                       NAT1
 NAT1
           chr8 [18067618, 18081197]
                                            + |
                                                       NAT1
           chr8 [18079177, 18081197]
                                                       NAT1
 NAT1
                                            + |
           chr8 [18248755, 18258723]
                                            + |
                                                       NAT2
 NAT2
            ensembl_id
           <character>
 A1BG ENSG00000121410
   A2M ENSG00000175899
 NAT1 ENSG00000171428
 NAT1 ENSG00000171428
 NAT1 ENSG00000171428
 NAT2 ENSG00000156006
  seqlengths:
                     chr1
                                           chr10 ...
                                                                       chrY
                                                                         NA
                       NA
                                              NA ...
> genesymbol["RBM17"]
GRanges with 1 range and 2 metadata columns:
        seqnames
                              ranges strand |
                                                    symbol
                           <IRanges> <Rle> | <character>
           <Rle>
 RBM17
           chr10 [6130949, 6159420]
                                           + |
                                                     RBM17
             ensembl id
            <character>
 RBM17 ENSG00000134453
  seqlengths:
                                           chr10 ...
                     chr1
                                                                       chrY
                       NA
                                              NA ...
                                                                         NA
```

## 4 Bugs Report and Features Request

Latest code are available on github https://github.com/tengfei/biovizBase

Please file bug/request on issue page, this is preferred way. or email me at yintengfei <at> gmail dot com.

It's a new package and under active development.

Thanks in advance for any feedback.

## 5 Acknowledgement

I wish to thank all those who helped me. Without them, I could not have started this project.

Genentech Sponsorship and valuable feed back and help for this project and my other project.

Jennifer Chang Feedback on this package

## 6 Session Information

```
> sessionInfo()
R version 3.0.2 Patched (2013-12-18 r64488)
Platform: x86_64-unknown-linux-gnu (64-bit)
locale:
 [1] LC_CTYPE=en_US.UTF-8
                                 LC_NUMERIC=C
 [3] LC_TIME=en_US.UTF-8
                                 LC_COLLATE=C
 [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
attached base packages:
[1] parallel
              stats
                        graphics grDevices utils
                                                        datasets
[7] methods
              base
other attached packages:
[1] GenomicRanges_1.14.4 XVector_0.2.0
                                               IRanges_1.20.6
[4] BiocGenerics_0.8.0
                         dichromat_2.0-0
                                               biovizBase_1.10.8
loaded via a namespace (and not attached):
 [1] AnnotationDbi_1.24.0
                             BSgenome_1.30.0
 [3] Biobase_2.22.0
                             Biostrings_2.30.1
 [5] DBI_0.2-7
                             Formula_1.1-1
 [7] GenomicFeatures_1.14.3 Hmisc_3.14-1
 [9] RColorBrewer_1.0-5
                             RCurl_1.95-4.1
[11] RSQLite_0.11.4
                             Rcpp_0.11.0
[13] Rsamtools_1.14.3
                             XML_3.98-1.1
[15] biomaRt_2.18.0
                             bitops_1.0-6
[17] cluster_1.14.4
                             colorspace_1.2-4
[19] grid_3.0.2
                             labeling_0.2
[21] lattice_0.20-27
                             latticeExtra_0.6-26
```

| [23] | munsell_0.4.2      | plyr_1.8.1      |
|------|--------------------|-----------------|
| [25] | rtracklayer_1.22.4 | scales_0.2.3    |
| [27] | splines_3.0.2      | stats4_3.0.2    |
| [29] | stringr_0.6.2      | survival_2.37-7 |
| [31] | tools_3.0.2        | zlibbioc_1.8.0  |