# An Introduction to  $biovizBase$

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## Contents



## <span id="page-2-0"></span>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.

## <span id="page-2-1"></span>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](#page-2-3)</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  $2$  or use palette from package *dichromat* or use color-blind safe color palette checked by ColorBrewer website[3](#page-2-5) . 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)
>
```
## <span id="page-2-2"></span>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.

<span id="page-2-3"></span><sup>1</sup><http://colorbrewer2.org/>

<span id="page-2-4"></span> $^{2}$ <http://www.vischeck.com/>

<span id="page-2-5"></span><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)



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())
```


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

- > ## 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.

## <span id="page-5-0"></span>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 *geneplotter*, 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 getBioColor 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



```
"#212121" "#1E1E1E" "#1C1C1C" "#191919" "#171717" "#141414" "#121212"
  gpos94 gpos95 gpos96 gpos97 gpos98 gpos99 gpos100
"#0F0F0F" "#0C0C0C" "#0A0A0A" "#070707" "#050505" "#020202" "#000000"
> getBioColor("CYTOBAND")
    gneg stalk acen gpos gvar gpos1 gpos2
"grey100" "brown3" "brown4" "grey0" "grey0" "#FFFFFF" "#FCFCFC"
   gpos3 gpos4 gpos5 gpos6 gpos7 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 gpos25 gpos26 gpos27 gpos28 gpos29 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 gpos46 gpos47 gpos48 gpos49 gpos50 gpos51
"#8D8D8D" "#8B8B8B" "#888888" "#858585" "#838383" "#808080" "#7E7E7E"
  gpos52 gpos53 gpos54 gpos55 gpos56 gpos57 gpos58
"#7B7B7B" "#797979" "#767676" "#737373" "#717171" "#6E6E6E" "#6C6C6C"
  gpos59 gpos60 gpos61 gpos62 gpos63 gpos64 gpos65
"#696969" "#676767" "#646464" "#616161" "#5F5F5F" "#5C5C5C" "#5A5A5A"
  gpos66 gpos67 gpos68 gpos69 gpos70 gpos71 gpos72
"#575757" "#545454" "#525252" "#4F4F4F" "#4D4D4D" "#4A4A4A" "#484848"
  gpos73 gpos74 gpos75 gpos76 gpos77 gpos78 gpos79
"#454545" "#424242" "#404040" "#3D3D3D" "#3B3B3B" "#383838" "#363636"
  gpos80 gpos81 gpos82 gpos83 gpos84 gpos85 gpos86
"#333333" "#303030" "#2E2E2E" "#2B2B2B" "#292929" "#262626" "#242424"
  gpos87 gpos88 gpos89 gpos90 gpos91 gpos92 gpos93
"#212121" "#1E1E1E" "#1C1C1C" "#191919" "#171717" "#141414" "#121212"
  gpos94 gpos95 gpos96 gpos97 gpos98 gpos99 gpos100
"#0F0F0F" "#0C0C0C" "#0A0A0A" "#070707" "#050505" "#020202" "#000000"
> ## differece source from default or options.
```
gpos80 gpos81 gpos82 gpos83 gpos84 gpos85 gpos86 "#333333" "#303030" "#2E2E2E" "#2B2B2B" "#292929" "#262626" "#242424" gpos87 gpos88 gpos89 gpos90 gpos91 gpos92 gpos93

```
> opts <- getOption("biovizBase")
> 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"

> ## get default fixed color > getBioColor("DNA\_BASES\_N", source = "default")

```
A T G C N
"#ABD9E9" "#2C7BB6" "#D7191C" "#FDAE61" "#FFFFBF"
> seqs <- c("A", "C", "T", "G", "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

## <span id="page-8-0"></span>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 Figur[e3](#page-8-1) 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.

 $@$ 

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



<span id="page-8-1"></span>Figure 3: Colorblind vision check for color of strand

#### <span id="page-9-0"></span>2.4 Nucleotides Color

We start with the five most used nucleotides,  $A, T, C, G, N$ , 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,  $GC\text{-}content$  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 G,C and cold colors for A,T, and a color in between to represent 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](#page-10-0) 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"
```
>

## <span id="page-9-1"></span>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.

## <span id="page-9-2"></span>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")



<span id="page-10-0"></span>Figure 4: Colorblind vision check for color of nucleotide

## <span id="page-11-0"></span>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.

## <span id="page-11-1"></span>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.

## <span id="page-11-2"></span>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 *gname* 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 object with 6 ranges and 5 metadata columns:
     seqnames ranges strand | value score group
       <Rle> <IRanges> <Rle> | <numeric> <numeric> <character>
 chr1 chr1 113-182 + | 11.46229 58.8187 Tumor
 chr1 chr1 2-76 + | 13.37479 87.8879 Tumor
 chr1 chr1 109-180 * | 10.22369 73.4545 Normal
 chr1 chr1 102-176 + | 9.82206 138.4521 Tumor
 chr1 chr1 57-131 * | 13.30008 118.0667 Normal
 chr1 chr1 160-234 * | 9.50643 87.4475 Tumor
          pair stepping
     <character> <numeric>
 chr1 q 8
 chr1 r 1
 chr1 m 18
 chr1 c 5
 chr1 p 13
 chr1 w 28
 -------
 seqinfo: 5 sequences from an unspecified genome; no seqlengths
> head(addStepping(gr, group.name = "pair"))
GRanges object with 6 ranges and 5 metadata columns:
     seqnames ranges strand | value score group
       <Rle> <IRanges> <Rle> | <numeric> <numeric> <character>
 chr1 chr1 113-182 + | 11.46229 58.8187 Tumor
 chr1 chr1 2-76 + | 13.37479 87.8879 Tumor
 chr1 chr1 109-180 * | 10.22369 73.4545 Normal
 chr1 chr1 102-176 + | 9.82206 138.4521 Tumor
 chr1 chr1 57-131 * | 13.30008 118.0667 Normal
 chr1 chr1 160-234 * | 9.50643 87.4475 Tumor
          pair stepping
     <character> <numeric>
 chr1 q 17
 chr1 r 18
 chr1 m 13
 chr1 c 3
 chr1 p 16
 chr1 w 23
 -------
 seqinfo: 5 sequences from an unspecified genome; no seqlengths
> gr.close <- GRanges(c("chr1", "chr1"), IRanges(c(10, 20), width = 9))
> addStepping(gr.close)
GRanges object with 2 ranges and 1 metadata column:
     seqnames ranges strand | stepping
       <Rle> <IRanges> <Rle> | <numeric>
 chr1 chr1 10-18 * | 1
 chr1 chr1 20-28 * | 1
```

```
-------
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
> addStepping(gr.close, extend.size = 5)
GRanges object 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
 -------
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
```
#### <span id="page-13-0"></span>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

```
> gr.temp <- GRanges("chr1", IRanges(start = c(100, 250),
                                   end = c(200, 300))> maxGap(gaps(gr.temp, start = min(start(gr.temp))))
[1] 0.1225
> maxGap(gaps(gr.temp, start = min(start(gr.temp))), ratio = 0.5)
```
[1] 24.5

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](#page-15-0) and [6,](#page-16-0) because it's clipped. It's just for visualization purpose.

```
> gr1 <- 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)
> head(shrink.fun1(gr1))
```

```
GRanges object with 3 ranges and 1 metadata column:
     seqnames ranges strand | .ori
       <Rle> <IRanges> <Rle> | <GRanges>
 [1] chr1 91-191 * | chr1:100-200
 [2] chr1 282-382 * | chr1:300-400
 [3] chr1 473-673 * | chr1:600-800
 -------
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
> head(shrink.fun2(gr1))
GRanges object with 3 ranges and 1 metadata column:
     seqnames ranges strand | .ori
       <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
 -------
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
> gr2 < - 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 object with 3 ranges and 1 metadata column:
     seqnames ranges strand | .ori
       <Rle> <IRanges> <Rle> | <GRanges>
 [1] chr1 100-200 * | chr1:100-200
 [2] chr1 222-322 * | chr1:300-400
 [3] chr1 474-674 * | chr1:600-800
 -------
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
> head(shrink.fun(gr2))
GRanges object 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
 -------
 seqinfo: 1 sequence from an unspecified genome; no seqlengths
```
## <span id="page-14-0"></span>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.



<span id="page-15-0"></span>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



<span id="page-16-0"></span>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.

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)
```
### <span id="page-17-0"></span>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 ApplyPileupsParam 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])
```
#### <span id="page-18-0"></span>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](http://genome.ucsc.edu/cgi-bin/hgTables) 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()
```
Please specify genome



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 object 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
 [5] chr1 9200000-12700000 * | p36.22 gneg
 [6] chr1 12700000-16200000 * | p36.21 gpos50
 -------
 seqinfo: 24 sequences from an unspecified genome; no seqlengths
> data(hg19Ideogram)
> head(hg19Ideogram)
GRanges object with 6 ranges and 0 metadata columns:
              seqnames ranges strand
                 <Rle> <IRanges> <Rle>
 [1] chr1 1-249250621 *
 [2] chr1_g1000191_r random 1-106433[3] chr1_g1000192_random 1-547496 *
 [4] chr2 1-243199373
 [5] chr3 1-198022430 *
 [6] chr4 1-191154276
 -------
 seqinfo: 93 sequences from hg19 genome
```
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.

```
> isIdeogram(hg19IdeogramCyto)
```
[1] TRUE

> isIdeogram(hg19Ideogram)

[1] FALSE

> isSimpleIdeogram(hg19IdeogramCyto)

[1] FALSE

```
> isSimpleIdeogram(hg19Ideogram)
```
[1] TRUE

#### <span id="page-20-0"></span>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 ggbio.

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 ensembl id. 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 object with 6 ranges and 2 metadata columns:
     seqnames ranges strand | symbol ensembl_id
        <Rle> <IRanges> <Rle> | <character> <character>
 A1BG chr19 58858174-58864865 - | A1BG ENSG00000121410
  A2M chr12 9220304-9268558 - | A2M ENSG00000175899
 NAT1 chr8 18027971-18081197 + | NAT1 ENSG00000171428
 NAT1 chr8 18067618-18081197 + | NAT1 ENSG00000171428
 NAT1 chr8 18079177-18081197 + | NAT1 ENSG00000171428
 NAT2 chr8 18248755-18258723 + | NAT2 ENSG00000156006
 -------
 seqinfo: 45 sequences from an unspecified genome; no seqlengths
> genesymbol["RBM17"]
GRanges object with 1 range and 2 metadata columns:
      seqnames ranges strand | symbol ensembl_id
         <Rle> <IRanges> <Rle> | <character> <character>
 RBM17 chr10 6130949-6159420 + | RBM17 ENSG00000134453
 -------
 seqinfo: 45 sequences from an unspecified genome; no seqlengths
```
>

## <span id="page-20-1"></span>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  $\langle$ at $\rangle$  gmail dot com.

It's a new package and under active development.

Thanks in advance for any feedback.

## <span id="page-20-2"></span>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

## <span id="page-21-0"></span>6 Session Information

```
> sessionInfo()
R version 4.4.1 (2024-06-14)
Platform: x86_64-pc-linux-gnu
Running under: Ubuntu 24.04.1 LTS
Matrix products: default
BLAS: /home/biocbuild/bbs-3.20-bioc/R/lib/libRblas.so
LAPACK: /usr/lib/x86_64-linux-gnu/lapack/liblapack.so.3.12.0
locale:
 [1] LC_CTYPE=en_US.UTF-8 LC_NUMERIC=C
 [3] LC_TIME=en_GB 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
time zone: America/New_York
tzcode source: system (glibc)
attached base packages:
[1] stats4 stats graphics grDevices utils datasets
[7] methods base
other attached packages:
[1] GenomicRanges_1.58.0 GenomeInfoDb_1.42.0 IRanges_2.40.0
[4] S4Vectors_0.44.0 BiocGenerics_0.52.0 dichromat_2.0-0.1
[7] biovizBase_1.54.0
loaded via a namespace (and not attached):
 [1] DBI_1.2.3 bitops_1.0-9
 [3] gridExtra_2.3 rlang_1.1.4
 [5] magrittr_2.0.3 matrixStats_1.4.1
 [7] compiler_4.4.1 RSQLite_2.3.7
 [9] GenomicFeatures_1.58.0 png_0.1-8
[11] vctrs_0.6.5 ProtGenerics_1.38.0
[13] stringr_1.5.1 pkgconfig_2.0.3
[15] crayon_1.5.3 fastmap_1.2.0
[17] backports_1.5.0 XVector_0.46.0
[19] utf8_1.2.4 Rsamtools_2.22.0
[21] rmarkdown_2.28 UCSC.utils_1.2.0
[23] bit_4.5.0 xfun_0.48
[25] zlibbioc_1.52.0 cachem_1.1.0
[27] jsonlite_1.8.9 blob_1.2.4
[29] DelayedArray_0.32.0 BiocParallel_1.40.0
[31] parallel_4.4.1 cluster_2.1.6
[33] R6_2.5.1 VariantAnnotation_1.52.0
[35] stringi_1.8.4 RColorBrewer_1.1-3
[37] rtracklayer_1.66.0 rpart_4.1.23
```
