

Package ‘UPDhmm’

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Title Detecting Uniparental Disomy through NGS trio data

Version 1.7.0

BugReports <https://github.com/martasevilla/UPDhmm/issues>

Description Uniparental disomy (UPD) is a genetic condition where an individual inherits both copies of a chromosome or part of it from one parent, rather than one copy from each parent. This package contains a HMM for detecting UPDs through HTS (High Throughput Sequencing) data from trio assays. By analyzing the genotypes in the trio, the model infers a hidden state (normal, father isodisomy, mother isodisomy, father heterodisomy and mother heterodisomy).

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UPDhmm-package	<i>UPDhmm: Detecting Uniparental Disomy through NGS trio data</i>
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Description

Uniparental disomy (UPD) is a genetic condition where an individual inherits both copies of a chromosome or part of it from one parent, rather than one copy from each parent. This package contains a HMM for detecting UPDs through HTS (High Throughput Sequencing) data from trio assays. By analyzing the genotypes in the trio, the model infers a hidden state (normal, father isodisomy, mother isodisomy, father heterodisomy and mother heterodisomy).

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See Also

Useful links:

- <https://github.com/martasevilla/UPDhmm>
- Report bugs at <https://github.com/martasevilla/UPDhmm/issues>

addOr	<i>Function to transform a large collapsed VCF into a dataframe, incorporating predicted states along with the log-likelihood ratio and p-value.</i>
-------	--

Description

Function to transform a large collapsed VCF into a dataframe, incorporating predicted states along with the log-likelihood ratio and p-value.

Usage

```
addOr(filtered_def_blocks_states, largeCollapsedVcf, hmm, genotypes)
```

Arguments

filtered_def_blocks_states	data.frame object containing the blocks
largeCollapsedVcf	Input VCF file
hmm	Hidden Markov Model used to infer the events. The format should adhere to the general HMM format from HMM package with a series of elements: 1. The hidden states names in the "States" vector. 2. All possible observations in the "Symbols" vector. 3. Start probabilities of every hidden state in the "startProbs" vector. 4. Transition probabilities matrix of the hidden states in "transProbs". 5. Probabilities associated between every hidden state and all possible observations in the "emissionProbs" matrix.
genotypes	Possible GT formats and its correspondence with the hmm

Value

data.frame containing the transformed information.

addRatioDepth	<i>Add depth ratios (inside vs outside events) per sample</i>
---------------	---

Description

This function computes the average depth inside each block and compares it to the average depth outside the block, generating a ratio for each sample (proband, mother, father).

Usage

```
addRatioDepth(filtered_def_blocks_states, largeCollapsedVcf, field = NULL)
```

Arguments

filtered_def_blocks_states	data.frame object containing the block
largeCollapsedVcf	Input VCF file
field	Optional character. Read Depth field to use (e.g. "DP", "DS"). If NULL, the function will try DP first, then AD as fallback.

Value

data.frame with added depth and inside/outside ratios per sample

applyViterbi	<i>Apply the hidden Markov model using the Viterbi algorithm.</i>
--------------	---

Description

Apply the hidden Markov model using the Viterbi algorithm.

Usage

```
applyViterbi(largeCollapsedVcf, hmm, genotypes)
```

Arguments

largeCollapsedVcf	input vcf file
hmm	Hidden Markov Model used to infer the events
genotypes	Possible GT formats and its correspondence with the hmm

Value

largeCollapsedVcf

asDfVcf	<i>Function to transform a large collapsed VCF into a dataframe with predicted states, including chromosome, start position, end position and metadata.</i>
---------	---

Description

Function to transform a large collapsed VCF into a dataframe with predicted states, including chromosome, start position, end position and metadata.

Usage

```
asDfVcf(largeCollapsedVcf, genotypes)
```

Arguments

largeCollapsedVcf	Name of the large collapsed VCF file.
genotypes	Possible GT formats and its correspondence with the hmm

Value

dataframe

blocksVcf	<i>Function to simplify contiguous variants with the same state into blocks.</i>
-----------	--

Description

Function to simplify contiguous variants with the same state into blocks.

Usage

```
blocksVcf(df)
```

Arguments

df	data.frame resulting from the as_df_vcf function.
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Value

data.frame containing information on the chromosome, start #' position of the block, end position of the block, and predicted state.

calculateEvents

Calculate UPD events in trio VCFs.

Description

This function predicts the hidden states by applying the Viterbi algorithm using the Hidden Markov Model (HMM) from the UPDhmm package. It takes the genotypes of the trio as input and includes a final step to simplify the results into blocks.

Usage

```
calculateEvents(
  largeCollapsedVcf,
  hmm = NULL,
  field_DP = NULL,
  BPPARAM = BiocParallel::SerialParam(),
  verbose = FALSE
)
```

Arguments

largeCollapsedVcf

The VCF file in the general format (largeCollapsedVcf) with VariantAnnotation package. Previously edited with vcfCheck() function from UPDhmm package.

hmm

Default = NULL. If no arguments are added, the package will use the default HMM already implemented, based on Mendelian inheritance. If an optional HMM is desired, it should adhere to the general HMM format from HMM package with the following elements inside a list:

1. The hidden state names in the "States" vector.
2. All possible observations in the "Symbols" vector.
3. Start probabilities of every hidden state in the "startProbs" vector.
4. Transition probabilities matrix between states in "transProbs".
5. Probabilities associated between every hidden state and all possible observations in the "emissionProbs" matrix.

field_DP

Default = NULL. Character string specifying which FORMAT field in the VCF contains the read depth information to use in addRatioDepth(). If NULL (default), the function will automatically try "DP" (standard depth) or "AD" (allelic depths, summed across alleles). Use this parameter if your VCF uses a non-standard field name for depth, e.g. field = "NR" or "field_DP".

BPPARAM

Parallelization settings, passed to [bplapply](#). By default BiocParallel::SerialParam() (serial execution). To enable parallelization, provide a BiocParallel backend, e.g. BiocParallel::MulticoreParam(workers = min(2, parallel::detectCores())) or BiocParallel::SnowParam(workers = 2). Note: when running under R CMD check or Bioconductor build systems, the number of workers may be automatically limited (usually less or equal to 2).

verbose

Logical, default = FALSE. If TRUE, progress messages will be printed during processing.

Value

A data.frame object containing all detected events in the provided trio. If no events are found, the function will return an empty data.frame.

Examples

```
file <- system.file(package = "UPDhmm", "extdata", "test_het_mat.vcf.gz")
vcf <- VariantAnnotation::readVcf(file)
processedVcf <- vcfCheck(vcf,
  proband = "NA19675",
  mother = "NA19678",
  father = "NA19679"
)

# Run in serial mode (default)
res <- calculateEvents(processedVcf)

# Run in parallel with 2 cores
library(BiocParallel)
param <- MulticoreParam(workers = 2)
res_parallel <- calculateEvents(processedVcf, BPPARAM = param)
```

collapseEvents

*Collapse events per sample and chromosome***Description**

This function collapses genomic events per individual, chromosome, and group, summarising the number of events, total Mendelian errors, the total span size, and a string listing all merged event coordinates.

Usage

```
collapseEvents(subset_df)
```

Arguments

subset_df	A data.frame containing event-level data with columns: ID: Sample identifier. seqnames: Chromosome name. start: Start position of the event. end: End position of the event. group: Event group/class. n_mendelian_error: Number of Mendelian errors in the event.
-----------	--

Value

A data.frame with collapsed events and columns: ID, seqnames, group n_events: Number of events collapsed total_mendelian_error: Sum of Mendelian errors across events total_size: Total genomic span size covered by events collapsed_events: Comma-separated list of collapsed events min_start, max_end: Genomic span of collapsed block

Examples

```
all_events <- data.frame(
  ID = c("S1", "S1", "S1", "S2", "S2"),
  seqnames = c("1", "1", "1", "2", "2"),
  start = c(100, 150, 300, 500, 550),
  end = c(120, 180, 320, 520, 580),
  group = c("iso_mat", "iso_mat", "het_pat", "iso_mat", "iso_mat"),
  n_mendelian_error = c(5, 10, 2, 50, 30),
  stringsAsFactors = FALSE
)
out <- collapseEvents(all_events)
```

hmm

HMM data for predicting UPD events in trio genomic data

Description

This dataset provides Hidden Markov Model (HMM) parameters for predicting uniparental disomy (UPD) events in trio genomic data.

states Five different possible states.

symbols Code symbols used for genotype combinations.

startProbs The initial probabilities of each state.

transProbs Probabilities of transitioning from one state to another.

emissionProbs Given a certain genotype combination, the odds of each possible state.

Usage

```
data(hmm)
```

Format

A list with 5 different elements

Source

Created in-house based on basic Mendelian rules for calculating UPD events.

Examples

```
data(hmm)
```

identifyRecurrentRegions

Identify recurrent genomic regions across samples

Description

This function finds recurrent genomic regions across samples based on overlapping intervals. Regions exceeding the Mendelian error threshold are excluded.

Usage

```
identifyRecurrentRegions(
  df,
  ID_col = "ID",
  error_threshold = 100,
  min_support = 3
)
```

Arguments

df	A data.frame with columns: - ID: Sample identifier. - seqnames: Chromosome name. - start: Start coordinate of the region. - end: End coordinate of the region. - n_mendelian_error: Number of Mendelian errors in the region.
ID_col	Character string indicating the column name containing sample identifiers. Default is "ID".
error_threshold	Numeric, default = 100. Maximum number of Mendelian errors allowed for a region to be considered.
min_support	Integer, default = 3. Minimum number of unique samples required to call a region recurrent.

Value

A GRanges object containing the recurrent regions that meet the minimum support threshold.

Examples

```
df <- data.frame(
  ID = c("S1", "S2", "S3", "S3"),
  seqnames = c("chr1", "chr1", "chr1", "chr1"),
  start = c(100, 120, 500, 510),
  end = c(150, 170, 550, 560),
  n_mendelian_error = c(10, 20, 5, 5)
)
identifyRecurrentRegions(df, ID_col = "ID", error_threshold = 50, min_support = 2)
```

markRecurrentRegions *Annotate regions as recurrent or non-recurrent*

Description

Given a results data.frame and a set of recurrent genomic regions, this function labels each row as "Yes" (recurrent) or "No".

Usage

```
markRecurrentRegions(df, recurrent_regions)
```

Arguments

df Data.frame with region coordinates and sample IDs.
 recurrent_regions GRanges object from identifyRecurrentRegions().

Value

The same data.frame with two added columns:

- Recurrent: "Yes" or "No"
- n_samples: Number of supporting samples (if recurrent)

Examples

```
input <- data.frame(
  ID = c("S1", "S2", "S3", "S4"),
  seqnames = c("chr1", "chr1", "chr1", "chr2"),
  start = c(100, 120, 500, 100),
  end = c(150, 170, 550, 150),
  n_mendelian_error = c(10, 20, 5, 200)
)

recurrent_gr <- GenomicRanges::GRanges(
  seqnames = "chr1",
  ranges = IRanges::IRanges(
    start = 100,
    end = 170
  ),
  n_samples = 2
)
markRecurrentRegions(input, recurrent_gr)
```

processChromosome	<i>Process a single chromosome for UPD detection</i>
-------------------	--

Description

Internal helper function to run the full pipeline on one chromosome:

- applyViterbi
- asDfVcf
- blocksVcf

Usage

```
processChromosome(vcf_chr, hmm, genotypes)
```

Arguments

vcf_chr	CollapsedVCF object for one chromosome
hmm	Hidden Markov Model object
genotypes	Named vector mapping genotype strings to numeric states

Value

A data.frame of detected blocks for the chromosome, or NULL if error

vcfCheck	<i>Check quality parameters (optional) and change IDs.</i>
----------	--

Description

This function takes a VCF file and converts it into a largeCollapsedVcf object using the VariantAnnotation package. It also rename the sample for subsequent steps needed in UPDhmm package. Additionally, it features an optional parameter, quality_check, which triggers warnings when variants lack sufficient quality based on RD and GQ parameters in the input VCF.

Usage

```
vcfCheck(largeCollapsedVcf, father, mother, proband, check_quality = FALSE)
```

Arguments

largeCollapsedVcf	The file in largeCollapsedVcf format.
father	Name of the father's sample.
mother	Name of the mother's sample.
proband	Name of the proband's sample.
check_quality	Optional argument. TRUE/FALSE. If quality parameters want to be measured. Default = FALSE

Value

largeCollapsedVcf (VariantAnnotation VCF format).

Examples

```
f1 <- system.file("extdata", "test_het_mat.vcf.gz", package = "UPDhmm")
vcf <- VariantAnnotation::readVcf(f1)
processedVcf <-
  vcfCheck(vcf, proband = "Sample1", mother = "Sample3", father = "Sample2")
```

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