

# Package ‘UPDhmm’

December 6, 2024

**Title** Detecting Uniparental Disomy through NGS trio data

**Version** 1.2.0

**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).

**biocViews** Software, HiddenMarkovModel, Genetics

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**Encoding** UTF-8

**LazyData** false

**RoxygenNote** 7.3.1

**Depends** R (>= 4.3.0)

**Imports** HMM, utils, VariantAnnotation, GenomicRanges, S4Vectors, IRanges, stats

**Suggests** knitr, testthat (>= 2.1.0), BiocStyle, rmarkdown, markdown, karyoploteR, regioneR, dplyr

**VignetteBuilder** knitr

**Roxygen** list(markdown = TRUE)

**URL** <https://github.com/martasevilla/UPDhmm>

**Language** en-US

**git\_url** <https://git.bioconductor.org/packages/UPDhmm>

**git\_branch** RELEASE\_3\_20

**git\_last\_commit** 0ca2a87

**git\_last\_commit\_date** 2024-10-29

**Repository** Bioconductor 3.20

**Date/Publication** 2024-12-05

**Author** Marta Sevilla [aut, cre] (<<https://orcid.org/0009-0005-0179-920X>>),  
 Carlos Ruiz-Arenas [aut] (<<https://orcid.org/0000-0002-6014-3498>>)

**Maintainer** Marta Sevilla <marta.sevilla@upf.edu>

## Contents

|                           |          |
|---------------------------|----------|
| UPDhmm-package . . . . .  | 2        |
| addOr . . . . .           | 3        |
| applyViterbi . . . . .    | 3        |
| asDfVcf . . . . .         | 4        |
| blocksVcf . . . . .       | 4        |
| calculateEvents . . . . . | 5        |
| hmm . . . . .             | 6        |
| vcfCheck . . . . .        | 6        |
| <b>Index</b>              | <b>8</b> |

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UPDhmm-package

*UPDhmm: Detecting Uniparental Disomy through NGS trio data*

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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).

## Author(s)

**Maintainer:** Marta Sevilla <marta.sevilla@upf.edu> ([ORCID](#))

Authors:

- Carlos Ruiz-Arenas <cruizarenas@unav.es> ([ORCID](#))

## See Also

Useful links:

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

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|-------|--|
| 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> |
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### 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: <ol style="list-style-type: none"> <li>1. The hidden states names in the "States" vector.</li> <li>2. All possible observations in the "Symbols" vector.</li> <li>3. Start probabilities of every hidden state in the "startProbs" vector.</li> <li>4. Transition probabilities matrix of the hidden states in "transProbs".</li> <li>5. Probabilities associated between every hidden state and all possible observations in the "emissionProbs" matrix.</li> </ol> |
| genotypes                  | Possible GT formats and its correspondence with the hmm   |

### Value

data.frame containing the transformed information.

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

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### 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

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

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**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> |
|-----------|--|

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**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. |
|----|---|

**Value**

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

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|                 |   |
|-----------------|---|
| calculateEvents | <i>Calculate UPD events in trio VCFs.</i> |
|-----------------|---|

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**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)
```

**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.

**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"
)
```

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`hmm`*HMM data for predicting UPD events in trio genomic data*

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**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)
```

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`vcfCheck`*Check quality parameters (optional) and change IDs.*

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**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.<br>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")
```

# Index

\* **datasets**

hmm, [6](#)

\* **internal**

UPDhmm-package, [2](#)

addOr, [3](#)

applyViterbi, [3](#)

asDfVcf, [4](#)

blocksVcf, [4](#)

calculateEvents, [5](#)

hmm, [6](#)

UPDhmm (UPDhmm-package), [2](#)

UPDhmm-package, [2](#)

vcfCheck, [6](#)