Package: EthSEQ (via r-universe)

July 10, 2024

Type Package

Title Ethnicity Annotation from Whole-Exome and Targeted Sequencing Data

Version 3.0.1

Description Reliable and rapid ethnicity annotation from whole exome and targeted sequencing data.

License GPL-3

Depends R (>= 2.15)

Imports graphics, utils, parallel, grDevices, MASS (>= 7.3-47), geometry (>= 0.3-6), data.table (>= 1.10.0), SNPRelate (>= 1.8.0), gdsfmt (>= 1.10.1), plot3D (>= 1.1), Rcpp (>= 0.11.0)

LinkingTo Rcpp

RoxygenNote 7.2.1

Suggests knitr, rmarkdown

VignetteBuilder knitr

Repository https://cibiobcg.r-universe.dev

RemoteUrl https://github.com/cibiobcg/ethseq

RemoteRef HEAD

RemoteSha 14e763514d3cad5a726ee345a2e0429fdb6d12ad

Contents

`ethseq.Analysis` *Ancestry analysis from whole-exome and targeted sequencing data*

Description

This function performs ancestry analysis of a set of samples ad reports the results.

Usage

```
ethseq.Analysis(
  target.vcf = NA,
  target.gds = NA,
  bam.list = NA,
  out.dir = tempdir(),
  model.gds = NA,
  model.available = NA,
  model.assembly = "hg38",
  model.pop = "All",
  model.folder = tempdir(),
  run.genotype = FALSE,
  aseq.path = tempdir(),
  mbq = 20,
  mrq = 20,
  mdc = 10,
  cores = 1,
  verbose = TRUE,
  composite.model.call.rate = 1,
  refinement.analysis = NA,
  space = "2D",
  bam.chr.encoding = FALSE
)
```

Arguments

`target.vcf`	Path to the sample's genotypes in VCF format
`target.gds`	Path to the sample's genotypes in GDS format
`bam.list`	Path to a file containing a list of BAM files paths
`out.dir`	Path to the folder where the output of the analysis is saved
`model.gds`	Path to a GDS file specifying the reference model
`model.available`	String specifying the pre-computed reference model to use
`model.assembly`	String value indicating the assembly version to download for the pre-build models
`model.pop`	String value indicating the population to download for the pre-build models

`model.folder`	Path to the folder where reference models are already present or downloaded when needed
`run.genotype`	Logical values indicating whether the ASEQ genotype should be run
`aseq.path`	Path to the folder where ASEQ binary is available or is downloaded when needed
`mbq`	Minmum base quality used in the pileup by ASEQ
`mrq`	Minimum read quality used in the piluep by ASEQ
`mdc`	Minimum read count acceptable for genotype inference by ASEQ
`cores`	Number of parallel cores used for the analysis
`verbose`	Print detailed information
`composite.model.call.rate`	SNP call rate used to run Principal Component Analysis (PCA)
`refinement.analysis`	Matrix specifying a tree of ancestry sets
`space`	Dimensions of PCA space used to infer ancestry (2D or 3D)
`bam.chr.encoding`	Logical value indicating whether input BAM files have chromosomes encoded with "chr" prefix

Value

Logical value indicating the success of the analysis

`ethseq.RM` *Create Reference Model for Ancestry Analysis*

Description

This function creates a GDS reference model that can be used to performe EthSEQ ancestry analysis

Usage

```
ethseq.RM(
  vcf.fn,
  annotations,
  out.dir = "./",
  model.name = "Reference.Model",
  bed.fn = NA,
  verbose = TRUE,
  call.rate = 1,
  cores = 1
)
```

Arguments

vcf.fn	vector of paths to genotype files in VCF format
annotations	data.frame with mapping of all samples names, ancestries and gender
out.dir	Path to output folder
model.name	Name of the output model
bed.fn	path to BED file with regions of interest
verbose	Print detailed information
call.rate	SNPs call rate cutoff for inclusion in the final reference model
cores	How many parallel cores to use in the reference model generation

Value

Logical value indicating the success of the analysis

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getModelsList *List the models available*

Description

This function prints the list of all available models.

Usage

```
getModelsList()
```

Value

data.frame of all available models to use with specified assembly and population

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getSamplesInfo *List the samples annotation*

Description

This function prints the list of all 1,000 Genomes Project samples used to build the reference models.

Usage

```
getSamplesInfo()
```

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