# QCumber documentation Gimelbrant lab v 0.3.2

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**QCumber** is a set of R tools for quantification of allele-specific expression. It relies on two or more technical replicate RNA-seq libraries to calculate  $Quality\ Correction\ Constant\ (QCC)$  and use it to correct for allelic imbalance overdipserion.

**QCumber** analysis starts with a table of allelic counts per gene, calculated from RNA-seq data using any analysis pipeline such as ASEReadCounter\* (https://github.com/gimelbrantlab/ASEReadCounter\_star).

### Paper

 $Mendelevich\ A.*,\ Vinogradova\ S.*,\ Gupta\ S.,\ Mironov\ A.,\ Sunyaev\ S.,\ Gimelbrant\ A.\ "Unexpected\ variability\ of\ allelic\ imbalance\ estimates\ from\ RNA\ sequencing"$ 

Source GitHub repository: https://github.com/gimelbrantlab/QCumber

**Installation** To install current version of this package in R:

devtools::install\_github("gimelbrantlab/QCumber")

# Contents

Functions	<b>2</b>
BuildDesign	2
ComputeAICIs	
ComputeCorrConstantFor2Reps	
ComputeCorrConstantsForAllPairsReps	
CountsToAI	
GetGatkPipelineTabs	
MeanCoverage	
MergeSumCounts	
MixBetaBinomialFit	
MixBetaBinomialFitStep	
NameColumns	
PerformBinTestAIAnalysisForConditionNPoint_knownCC	
PerformBinTestAIAnalysisForConditionNPoint	
$Perform Bin Test AIA nalysis For Condition NPoint Vect\_known CC \ . \ . \ . \ . \ . \ . \ . \ . \ . $	
PerformBinTestAIAnalysisForConditionNPointVect	
PerformBinTestAIAnalysisForTwoConditions_knownCC	
PerformBinTestAIAnalysisForTwoConditions	
ThresholdingCounts	
Input requirements	14
Output format	14

# **Functions**

# BuildDesign

Creates a design matrix for the experiment

#### Usage

```
BuildDesign(experimentNames, techReps, corrConst = NA)
```

### Arguments

```
techReps Vector with names of the experiments

techReps Vector with number of technical replicates in each experiment

corrConst Optional, a vector with correction constants for each experiment
```

#### Value

Dataframe with experiments numbered and numbers of columns

### Examples

```
BuildDesign(c("clone1", "clone2", "clone3"), c(2, 2, 3), c(1.7, 1.8, mean(1.55, 1.6, 1.57)))
```

# COMPUTEAICIS

Calculates Binomial and QCC-corrected binomial CIs for a given vector of AI estimates, and calculates test statistics for comparison with a point or vector of points, for given QCC.

## Usage

```
ComputeAICIs(
  inDF,
  vectReps,
  vectRepsCombsCC,
  pt = 0.5,
  Q = 0.95,
  BF = T,
  thr = NA,
  thrUP = NA,
  thrType = "each"
)
```

### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vectReps A vector of replicate numbers for which the analysis should be applied

vectRepsCombsCC A vector of pairwise-computed correction constants for given replicates (QCC=1 is no correction)

- pt Optional (default=0.5), a value or a vector of values to compare with (if second, should be compatible with the order and size of genes vector in table of allelic counts)
- Q Optional (default=0.95), confidence level, quantile
- BF Optional (default=True), Bonferroni correction for multiple testing, set False ONLY IF Q is alredy corrected
- thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

```
    thrUP Optional (default=NA), threshold for max gene coverage (default = NA)
    thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type
```

#### Value

A table with IDs, AI estimates, coverage, test p-value, and Confidence Intervals

# COMPUTECORRCONSTANTFOR2REPS

Computes QCC for one pair of replicates.

### Usage

```
ComputeCorrConstantFor2Reps(
  inDF,
  reps,
  binNObs = 40,
  fitCovThr = 50,
  EPS = 1.05,
  thr = NA,
  thrUP = NA,
  thrType = "each"
)
```

### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

reps A vector of 2 replicate numbers for which the analysis should be applied

binNObs Optional (default=40), threshold on number of observations per bin

fitCovThr Optional (default=50), threshold on coverage for genes that will be included in Beta-Bin fitting

EPS Optional (default=1.05), base of exponent for the coverage binning, setting greater base (1.1 or 1.2 or 1.3) would result in fewer number of coverage bins in fitting process, thus will increase the computational speed, but may potentially reduce accuracy

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrup Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

## Value

List with (1) fitted QCC (\$fittedCC) and (2) a table with proportions of observed to expected quantiles per coverage bin (\$QObsExpPropsTable).

# COMPUTECORRCONSTANTSFORALLPAIRSREPS

Computes QCC for all possible pairs of given replicates.

### Usage

```
ComputeCorrConstantsForAllPairsReps(
  inDF,
  vectReps,
  binNObs = 40,
  fitCovThr = 50,
  EPS = 1.05,
  thr = NA,
  thrUP = NA,
  thrType = "each"
)
```

#### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vectReps A vector of  $\geq 2$  replicate numbers for which the analysis should be applied

binNObs Optional (default=40), threshold on number of observations per bin

fitCovThr Optional (default=50), threshold on coverage for genes that will be included in Beta-Bin fitting

EPS Optional (default=1.05), base of exponent for the coverage binning, setting greater base (1.1 or 1.2 or 1.3) would result in fewer number of coverage bins in fitting process, thus will increase the computational speed, but may potentially reduce accuracy

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrUP Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

#### Value

List of fitting outputs of ComputeCorrConstantFor2Reps() for each combinatorial pair of replicates (in order 1-2,1-3,...,1-N,2-3,...2-N,...,(N-1)-N): list with (1) fitted QCC (\$fittedCC) and (2) a table with proportions of observed to expected quantiles per coverage bin (\$QObsExpPropsTable).

# CountsToAI

Calculates allelic imbalances from merged counts over given replicates (ai(sum\_reps(gene))).

#### Usage

```
CountsToAI(
   df,
   reps = NA,
   meth = "mergedToProportion",
   thr = NA,
   thrUP = NA,
   thrType = "each"
)
```

### Arguments

Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

reps Optional (default=NA, all replicates), a vector of replicate numbers for which the analysis should be applied

meth Optional (default="mergedToProportion", also can be "meanOfProportions"), method to use, either sum(m)/sum(p) (default), or sum(m/p)

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrup Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

#### Value

A table with IDs and calculated AI estimate for given set of replicates

### Examples

CountsToAI(allelicCountsTable, reps=c(1,2), thr=10, thrUP=1000)

# GETGATKPIPELINETABS

Loads the working parts of tables ("ID", allele count 2x columns, "contig" if specified); concatenates (uniting merge) tables from all provided files.

#### Usage

GetGatkPipelineTabs(inFiles, nReps, contigs = vector())

### Arguments

inFiles A vector of full pathes to files with alelleic counts tables; necessary columns: "ID", pairs of ref and alt allelic counts; optionally, for filtering, "contig"

nReps A vector of numbers, either: (1) each entry is a number of replicates in the corresponding file; (2) for one file only, each entry is a number of replicates corresponding to particular experiment

contigs Optional (default=vector()), parameter defining if the resulting table should be filtered by contig column (preserving only rows corresponding to a given vector), default set to empty vector() and no filtering applied

### Value

A concatenated table with allele counts for all replicates, each row corresponds to a feature ("ID")

# MEANCOVERAGE

Calculates mean allelic coverage (mat+pat) among given replicates.

### Usage

MeanCoverage(df, reps = NA, thr = NA, thrUP = NA, thrType = "each")

### Arguments

df Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

reps Optional (default=NA, all replicates), a vector of replicate numbers for which the analysis should be applied

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrUP Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

#### Value

A table with IDs and calculated mean allelic coverage for given set of replicates

### Examples

MeanCoverage(allelicCountsTable, reps=c(3,4), thr=8)

# MERGESUMCOUNTS

Creates a table of sums of maternal and paternal alellic counts for given replicates.

### Usage

MergeSumCounts(df, reps = NA, thr = NA, thrUP = NA, thrType = "each")

# Arguments

df Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

reps Optional (default=NA, all replicates), a vector of replicate numbers for which the analysis should be applied

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrUP Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

# Value

A table with IDs and calculated allelic counts for each of the given replicates

### Examples

MeanCoverage(allelicCountsTable, reps=c(1,2))

# MIXBETABINOMIALFIT

Fitting beta-binomial mixture distribution of AI in particular bin.

### Usage

MixBetaBinomialFit(initials, coverage, observations)

### Arguments

initials Initials for EM algm: initials = c(w1, alpha1, alpha2), weight of first component and

alphas for both beta-binomial distributions in a mixture

coverage A number, that represents the coverage bin

observations A vector of "maternal counts" in the bin

### Value

Fitted weight of first component and alphas for both beta-binomial distributions in a mixture, plus number of steps.

# MIXBETABINOMIALFITSTEP

One step of fitting beta-binomial mixture distribution of AI in particular bin.

### Usage

MixBetaBinomialFitStep(initials\_old, coverage, observations)

### Arguments

initials\_old Initials for EM step: initials = c(w1, alpha1, alpha2), weight of first component and

alphas for both beta-binomial distributions in a mixture

coverage A number, that represents the coverage bin

observations A vector of "maternal counts" in the bin

### Value

Re-fitted initials for next EM step.

# NameColumns

Helper function to quickly rename columns in geneCountTab dataframe

### Usage

NameColumns(exp\_n, rep\_n)

### Arguments

exp\_n Experiment number

rep\_n Number of replicates for the experiment

#### Value

Vector with names

## Examples

colnames(allelicCountsTable)[2:9] <- c(NameColumns(1,2), NameColumns(2,2))

# PerformBinTestAIAnalysisForConditionNPoint\_knownCC

Performs Binomial and QCC-corrected binomial tests (with Bonferroni correction) with a given point estimate, for given QCC.

### Usage

inDF,

 ${\tt PerformBinTestAIAnalysisForConditionNPoint\_knownCC(}$ 

```
vectReps,
  vectRepsCombsCC,
  pt = 0.5,
  Q = 0.95,
  thr = NA,
  thrUP = NA,
  thrType = "each",
  minDifference = NA
Arguments
                           Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts
                   inDF
                           (rep1_ref, rep1_alt, rep2_ref, rep2_alt, ...)
                           A vector (\geq 2) of replicate numbers for which the analysis should be applied
              vectReps
      vectRepsCombsCC
                           A vector of pairwise-computed correction constants for given replicates (QCC=1 is no
                           correction)
                           Optional (default=0.5), a value to compare with
                     pt
                           Optional (default=0.95), confidence level, quantile
                      Q
                           Optional (default=NA), threshold on the overall number of counts for a gene to be
                    thr
                           considered in the analysis
                           Optional (default=NA), threshold for max gene coverage (default = NA)
                  thrUP
```

### Value

A table of gene names, AIs + CIs, p-values for both non-corrected and (BT..) and QCC corrected (BT\_CC..) tests, classification into genes demonstrating signifficant difference (TRUE) from point estimate AI and those that don't (FALSE).

# PerformBinTestAIAnalysisForConditionNPoint

threshold type

passing the test)

Calculates QCC. Performs Binomial and QCC-corrected binomial tests (with Bonferroni correction) with a given point estimate.

Optional (default = "each", also can be "average" for average coverage on replicates),

Optional (default=NA), if specified, one additional column is added to the output (T/F depending if the gene changed AI expression more than minDifference in addition to

#### Usage

```
PerformBinTestAIAnalysisForConditionNPoint(
  inDF,
  vectReps,
  pt = 0.5,
  binNObs = 40,
  Q = 0.95,
  fitCovThr = 50,
  EPS = 1.05,
  thr = NA,
  thrUP = NA,
  thrType = "each",
  minDifference = NA
)
```

thrType

minDifference

### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vectReps A vector  $(\geq 2)$  of replicate numbers for which the analysis should be applied

pt Optional (default=0.5), a value to compare with

binNObs Optional (default=40), threshold on number of observations per bin

Q Optional (default=0.95), confidence level, quantile

fitCovThr Optional (default=50), threshold on coverage for genes that will be included in Beta-Bin fitting

EPS Optional (default=1.05), base of exponent for the coverage binning, setting greater base (1.1 or 1.2 or 1.3) would result in fewer number of coverage bins in fitting process, thus will increase the computational speed, but may potentially reduce accuracy

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrup Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

minDifference Optional (default=NA), if specified, one additional column is added to the output (T/F

depending if the gene changed AI expression more than minDifference in addition to

passing the test)

### Value

List of (1) fitted QCC for all combanatorial pairs of replicates (\$CC), (2) ComputeCorrConstantsForAllPairsReps() output (\$FitDATA), and (3) PerformBinTestAIAnalysisForConditionNPoint\_knownCC() output (\$Output).

# PerformBinTestAIAnalysisForConditionNPointVect\_knownCC

Performs Binomial and QCC-corrected binomial tests (with Bonferroni correction) with a given vector of point estimates, for given QCC.

### Usage

```
PerformBinTestAIAnalysisForConditionNPointVect_knownCC(
  inDF,
  vectReps,
  vectRepsCombsCC,
  ptVect,
  Q = 0.95,
  thr = NA,
  thrUP = NA,
  thrType = "each",
  minDifference = NA
```

#### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vectReps A vector  $(\geq 2)$  of replicate numbers for which the analysis should be applied

vectRepsCombsCC A vector of pairwise-computed correction constants for given replicates (QCC=1 is no correction)

ptVect A vector of values to compare with, should be compatible with the order and size of genes vector in table of allelic counts

Q Optional (default=0.95), confidence level, quantile

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrUP Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

minDifference Optional (default=NA), if specified, one additional column is added to the output (T/F depending if the gene changed AI expression more than minDifference in addition to

passing the test)

#### Value

A table of gene names, AIs + CIs, p-values for both non-corrected and (BT..) and QCC corrected (BT\_CC..) tests, classification into genes demonstrating significant difference (TRUE) from corresponding point estimate AI and those that don't (FALSE).

# PERFORMBINTESTAIANALYSISFORCONDITIONNPOINTVECT

Calculates QCC. Performs Binomial and QCC-corrected binomial tests (with Bonferroni correction) with a given vector of point estimates.

### Usage

```
PerformBinTestAIAnalysisForConditionNPointVect(
  inDF,
  vectReps,
  ptVect,
  binNObs = 40,
  Q = 0.95,
  fitCovThr = 50,
  EPS = 1.05,
  thr = NA,
  thrUP = NA,
  thrType = "each",
  minDifference = NA
```

### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vectReps A vector  $(\geq 2)$  of replicate numbers for which the analysis should be applied

ptVect A vector of values to compare with, should be compatible with the order and size of genes vector in table of allelic counts

binNObs Optional (default=40), threshold on number of observations per bin

Q Optional (default=0.95), confidence level, quantile

fitCovThr Optional (default=50), threshold on coverage for genes that will be included in Beta-Bin fitting

EPS Optional (default=1.05), base of exponent for the coverage binning, setting greater base (1.1 or 1.2 or 1.3) would result in fewer number of coverage bins in fitting process, thus will increase the computational speed, but may potentially reduce accuracy

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrUP Optional (default=NA), threshold for max gene coverage (default = NA)

Optional (default = "each", also can be "average" for average coverage on replicates), thrType

threshold type

minDifference Optional (default=NA), if specified, one additional column is added to the output (T/F)

depending if the gene changed AI expression more than minDifference in addition to

passing the test)

#### Value

List of (1) fitted QCC for all combanatorial pairs of replicates (\$CC), (2) ComputeCorrConstantsForAllPairsReps() output (\$FitDATA), and (3) PerformBinTestAIAnalysisForConditionNPointVect\_knownCC() output (\$Output).

# PerformBinTestAIAnalysisForTwoConditions\_knownCC

Performs differential tests (with Bonferroni correction) for AI values for two conditions, for given QCC.

### Usage

```
PerformBinTestAIAnalysisForTwoConditions_knownCC(
 vect1CondReps,
 vect2CondReps,
 vect1CondRepsCombsCC,
 vect2CondRepsCombsCC,
 Q = 0.95,
 thr = NA,
 thrUP = NA,
 thrType = "each",
 minDifference = NA
```

### Arguments

Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts inDF

(rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vect1CondReps A vector  $(\geq 2)$  of replicate numbers that should be considered as first condition's tech

vect2CondReps A vector  $(\geq 2)$  of replicate numbers that should be considered as second condition's tech

vect1CondRepsCombsCC A vector of pairwise-computed correction constants for first condition's tech reps (QCC=1

is no correction)

vect2CondRepsCombsCC A vector of pairwise-computed correction constants for second condition's tech reps

(QCC=1 is no correction)

Optional (default=0.95), confidence level, quantile

Optional (default=NA), threshold on the overall number of counts for a gene to be thr

considered in the analysis

Optional (default=NA), threshold for max gene coverage (default = NA) thrUP

Optional (default = "each", also can be "average" for average coverage on replicates), thrType

threshold type

Optional (default=NA), if specified, one additional column is added to the output (T/F minDifference

depending if the gene changed AI expression more than minDifference in addition to

passing the test)

#### Value

A table of gene names, AIs + CIs for both conditions, p-values for both non-corrected (BT..) and QCC corrected (BT\_CC..) differential tests, classification into genes demonstrating signifficant difference (TRUE) of AI estimates in two conditions, and those that don't (FALSE).

# PERFORMBINTESTAIANALYSISFORTWOCONDITIONS

Calculates QCC. Performs differential tests (with Bonferroni correction) for AI values for two conditions.

#### Usage

```
PerformBinTestAIAnalysisForTwoConditions(
  inDF,
  vect1CondReps,
  vect2CondReps,
  binNObs = 40,
  fitCovThr = 50,
  Q = 0.95,
  EPS = 1.05,
  thr = NA,
  thrUP = NA,
  thrType = "each",
  minDifference = NA
)
```

### Arguments

inDF Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)

vect1CondReps A vector ( $\geq 2$ ) of replicate numbers that should be considered as first condition's tech reps

vect2CondReps A vector ( $\geq 2$ ) of replicate numbers that should be considered as second condition's tech reps

binNObs Optional (default=40), threshold on number of observations per bin

fitCovThr Optional (default=50), threshold on coverage for genes that will be included in Beta-Bin fitting

Q Optional (default=0.95), confidence level, quantile

EPS Optional (default=1.05), base of exponent for the coverage binning, setting greater base (1.1 or 1.2 or 1.3) would result in fewer number of coverage bins in fitting process, thus will increase the computational speed, but may potentially reduce accuracy

thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis

thrup Optional (default=NA), threshold for max gene coverage (default = NA)

thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

Optional (default=NA), if specified, one additional column is added to the output (T/F depending if the gene changed AI expression more than minDifference in addition to passing the test)

# Value

minDifference

List of (1) fitted QCC for all combanatorial pairs of replicates for both conditions (\$CC), (2) ComputeCorrConstantsForAllPairsReps() output for both conditions (\$FitDATA), and (3) PerformBinTestAIAnalysisForTwoConditions\_knownCC() output (\$Output).

# THRESHOLDINGCOUNTS

Takes allelic counts table and returns table, where all genes that don't pass a given coverage threshold have NA coverage. Can be restricted to particular replicates.

### Usage

ThresholdingCounts(df, reps = NA, thr = NA, thrUP = NA, thrType = "each")

### Arguments

- df Allele counts dataframe: with 2n+1 columns, "ID" and 2n columns with ref & alt counts (rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ...)
- reps Optional (default=NA, all replicates), a vector of replicate numbers for which the analysis should be applied
- thr Optional (default=NA), threshold on the overall number of counts for a gene to be considered in the analysis
- thrUP Optional (default=NA), threshold for max gene coverage (default = NA)
- thrType Optional (default = "each", also can be "average" for average coverage on replicates), threshold type

### Value

Allelic counts table with masked with NA undercovered genes, for selected replicates

### Examples

ThresholdingCounts(df = allelicCountsTable, reps = c(1,2), thr = 10)

# Input requirements

Allelic Counts Tables (GetGatkPipelineTabs input)

#### • Structure:

A tab-segmented table with present column names, each line corresponds to a genomic feature (for example, gene). The first column is genomic feature ID, next  $2 \times N$  columns stores alellelic counts for reference and alternative alleles for each of N replicates (in order rep1\_ref, rep1\_alt, rep2\_ref, rep2\_alt, ... repN\_ref, repN\_alt). The additional column contig should be present in the table at any position  $\geq 2N+1$  if filtering by contig is needed.

#### • Column names:

If contig parameter is set non-empty, a table must have corresponding column named contig. The other columns may have any names.

# Output formats

#### AI CI tables

Genomic feature ID ID sumCOV Total allelic counts matCOV Maternal/reference allelic counts Allelic Imbalance point estimate AΙ  $BT_CIleft$ Left boundary of CI\* for non-corrected test  $BT\_CIright$ Right boundary of CI\* for non-corrected test Left boundary of CI\* for QCC-corrected test (condition 1)  $BT_CIleft_CC_1$ Right boundary of CI\* for QCC-corrected test (condition 1) BT\_CIright\_CC\_1  $BT_pval$ Non-corrected test statistics (p-value) QCC-corrected test statistics (p-value) BT\_pval\_CC Non-corrected test decision BTBT\_CC QCC-corrected test decision If minDifference was set, QCC-corrected test decision AND passed minDifference threshold BT\_CC\_thrDiff

### Differential AI tables

ID	Genomic feature ID
$sumCOV_1$	Total allelic counts (condition 1)
${\tt matCOV\_1}$	Maternal/reference allelic counts (condition 1)
AI_1	Allelic Imbalance point estimate (condition 1)
$BT_CIleft_1$	Left boundary of CI* for non-corrected test (condition 1)
$BT_CIright_1$	Right boundary of CI* for non-corrected test (condition 1)
$BT_CIleft_CC_2$	Left boundary of CI* for QCC-corrected test (condition 1)
$BT_CIright_CC_2$	Right boundary of CI* for QCC-corrected test (condition 1)
$sumCOV_2$	Total allelic counts (condition 2)
$\mathtt{matCOV}_2$	Maternal/reference allelic counts (condition 2)
AI_2	Allelic Imbalance point estimate (condition 2)
$BT_CIleft_2$	Left boundary of CI* for non-corrected test (condition 2)
BT_CIright_2	Right boundary of CI* for non-corrected test (condition 2)
$BT_CIleft_CC_2$	Left boundary of CI* for QCC-corrected test (condition 2)
$BT_CIright_CC_2$	Right boundary of CI* for QCC-corrected test (condition 2)
$\mathtt{BT\_pval}$	Non-corrected test statistics (p-value)
$BT_pval_CC$	QCC-corrected test statistics (p-value)
BT	Non-corrected test decision
BT_CC	QCC-corrected test decision
$BT\_CC\_thrDiff$	If minDifference was set, QCC-corrected test decision AND passed minDifference threshold

<sup>\* -</sup> for quantile Q corrected on multiple testing using Bonferroni approach  $(1 - \frac{1-Q}{\#genes})$ , where only considered genes in the analysis were counted.