Package ‘MyoScore’

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Title Transcriptomic Scoring for Human Skeletal Muscle Health

Version 1.0.1

Description Calculate MyoScore, a genetically informed muscle health score, from bulk RNA sequencing (RNA-seq) raw count data. MyoScore integrates results from genome-wide association studies (GWAS) and transcriptome-wide association studies (TWAS) across 28 muscle-related phenotypes to quantify muscle health along five dimensions (Strength, Mass, LeanMuscle, Youth, Resilience), each scored from 0 to 100. The package provides preprocessing via counts per million (CPM) normalization, dimension-level and composite scoring, and visualization utilities including radar charts and grouped boxplots. For more information, see <`https://github.com/Hirriririir/MyoScore`>.

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URL `https://github.com/Hirriririir/MyoScore`

BugReports `https://github.com/Hirriririir/MyoScore/issues`

Depends R (>= 4.0.0)

Imports stats, utils, graphics, grDevices

Suggests ggplot2 (>= 3.4.0), fmsb, patchwork, testthat (>= 3.0.0), knitr, rmarkdown

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Contents

`myoscore-constants` *MyoScore Dimension Weights and Constants*

Description

Data-driven weights for the five MyoScore dimensions, derived from GWAS-TWAS integration of 28 muscle-related phenotypes.

Details

Weights represent the relative contribution of each dimension to overall muscle health, determined by variance explained in the 1,722-sample training cohort.

`myoscore_colors` *Get MyoScore color palette*

Description

Get MyoScore color palette

Usage

```
myoscore_colors(type = c("dimensions", "spectrum", "all"))
```

Arguments

`type`	One of `"dimensions"` (5 dimension colors), `"spectrum"` (unhealthy-to-healthy gradient), or `"all"`.

Value

Named character vector of hex color codes.

Examples

```
myoscore_colors("dimensions")
myoscore_colors("spectrum")
```

`myoscore_dimensions` *Get MyoScore dimension names*

Description

Get MyoScore dimension names

Usage

```
myoscore_dimensions()
```

Value

Character vector of the five dimension names.

Examples

```
myoscore_dimensions()
```

`myoscore_genes` *MyoScore Gene Weights*

Description

A data frame containing 591 gene-dimension entries (417 unique genes) used in MyoScore calculation, filtered to genes detectable in bulk RNA-seq datasets.

Usage

```
myoscore_genes
```

Format

A data frame with 591 rows and 4 columns:

ID Gene symbol (HGNC).

weight Gene weight derived from TWAS Z-scores (|mean_Z| / n_phenotypes).

direction_v3 Direction of effect: +1 means high expression indicates health; -1 means high expression indicates disease.

dimension One of five dimensions: Strength, Mass, LeanMuscle, Youth, Resilience.

Details

Genes were identified through TWAS (Transcriptome-Wide Association Study) using FUSION with GTEx v8 skeletal muscle eQTL weights and 28 GWAS phenotypes covering grip strength, body composition, MRI fat infiltration, telomere length, and myopathy diagnoses.

Source

Myopathy Spectrum Project, GWAS-TWAS integration pipeline.

Examples

```
data(myoscore_genes)
table(myoscore_genes$dimension)
```

`myoscore_plot_boxplot` *Boxplot of MyoScore by Groups*

Description

Create grouped boxplots comparing MyoScore or individual dimension scores across conditions. Uses base R graphics by default, or ggplot2 if available.

Usage

```
myoscore_plot_boxplot(
  scores,
  groups,
  which = "MyoScore",
  colors = NULL,
  use_ggplot = TRUE,
  title = NULL,
  ...
)
```

Arguments

`scores`	A data.frame from `myoscore_score()`.
`groups`	A factor or character vector of group labels (one per sample).
`which`	Which score to plot. One of `"MyoScore"`, `"Strength"`, `"Mass"`, `"LeanMuscle"`, `"Youth"`, `"Resilience"`, or `"all"` for a multi-panel figure. Default `"MyoScore"`.
`colors`	Optional named or positional color vector.
`use_ggplot`	Logical. Use ggplot2 if available. Default `TRUE`.
`title`	Optional main title.
`...`	Additional arguments passed to `boxplot()` or `ggplot2::geom_boxplot()`.

Value

If `use_ggplot = TRUE` and ggplot2 is available, returns a ggplot object. Otherwise, invisible `NULL`.

Examples

```
# Create example scores and groups
scores_df <- data.frame(
  Strength_score  = c(rnorm(5, 55, 5), rnorm(5, 40, 5)),
  Mass_score      = c(rnorm(5, 50, 5), rnorm(5, 45, 5)),
  LeanMuscle_score = c(rnorm(5, 48, 5), rnorm(5, 38, 5)),
  Youth_score     = c(rnorm(5, 52, 5), rnorm(5, 35, 5)),
  Resilience_score = c(rnorm(5, 50, 5), rnorm(5, 45, 5)),
  MyoScore        = c(rnorm(5, 50, 3), rnorm(5, 40, 3))
)
groups <- rep(c("Healthy", "Disease"), each = 5)

myoscore_plot_boxplot(scores_df, groups = groups)
```

`myoscore_plot_radar` *Radar Chart of MyoScore Dimensions*

Description

Plot a radar (spider) chart showing the five MyoScore dimensions. Supports plotting one or more groups (e.g., disease stages) as overlaid or faceted panels.

Usage

```
myoscore_plot_radar(
  scores,
  groups = NULL,
  colors = NULL,
  facet = TRUE,
  title = NULL,
  show_values = TRUE,
  ...
)
```

Arguments

`scores`	A data.frame from `myoscore_score()`, or a named numeric vector of length 5 (one value per dimension), or a matrix/data.frame where each row is a group and columns are dimension scores.
`groups`	Optional. A factor or character vector assigning each row of `scores` to a group. When provided, group means are plotted.
`colors`	Optional. Character vector of colors (one per group). Default uses `myoscore_colors()` with type `"spectrum"`.

`facet`	Logical. If `TRUE` and multiple groups exist, plot each group in a separate panel. Default `TRUE`.
`title`	Optional main title.
`show_values`	Logical. Show score values at vertices. Default `TRUE`.
`...`	Additional arguments passed to `fmsb::radarchart()`.

Details

Requires the **fmsb** package (in Suggests).

Value

Invisible `NULL`. Called for its side effect (plot).

Examples

```
# Radar chart from a named vector of dimension scores
dim_scores <- c(Strength = 55, Mass = 48, LeanMuscle = 42,
                Youth = 60, Resilience = 50)

myoscore_plot_radar(dim_scores)
```

`myoscore_preprocess` *Preprocess Raw Counts to log2(CPM+1)*

Description

Normalize raw RNA-seq count data using CPM (Counts Per Million) followed by log2 transformation. This is the standard preprocessing step before MyoScore calculation.

Usage

```
myoscore_preprocess(raw_counts, verbose = TRUE)
```

Arguments

`raw_counts`	A numeric matrix or data.frame of raw counts with genes as rows and samples as columns. Row names should be gene symbols.
`verbose`	Logical. Print progress messages. Default `TRUE`.

Details

The transformation pipeline is:

1. CPM: counts / total_counts * 1e6
2. log2(CPM + 1)

Value

A numeric matrix of log2(CPM+1) values with the same dimensions and names as the input.

Examples

```
# Create example count matrix
counts <- matrix(rpois(500, lambda = 100), nrow = 50, ncol = 10)
rownames(counts) <- paste0("Gene", 1:50)
colnames(counts) <- paste0("Sample", 1:10)

log2cpm <- myoscore_preprocess(counts)
```

`myoscore_score` *Calculate MyoScore from Raw Count Data*

Description

Main entry point for computing MyoScore (Genetic Muscle Health Score). Accepts either a file path or a count matrix, and returns per-sample scores for all five dimensions plus the composite score.

Usage

```
myoscore_score(
  input,
  gene_weights = NULL,
  sep = ",",
  min_coverage = 0.1,
  verbose = TRUE
)
```

Arguments

`input`	Either a file path (character) to a raw count CSV/TSV, or a numeric matrix/data.frame with genes as rows and samples as columns. Gene symbols must be row names.
`gene_weights`	Optional. A data.frame of gene weights with columns `ID`, `weight`, `direction_v3`, `dimension`. Default uses the built-in `myoscore_genes` dataset.
`sep`	Separator for reading CSV files. Default `","`. Use `"\t"` for tab-separated files.
`min_coverage`	Minimum fraction (0-1) of genes required per dimension. Dimensions below this threshold return `NA`. Default `0.1`.
`verbose`	Logical. Print progress messages. Default `TRUE`.

Details

Scoring Pipeline:

1. Raw counts are normalized to log2(CPM+1).
2. For each dimension, available genes are z-score standardized (gene-wise across all input samples).
3. Z-scores are multiplied by gene direction and weight, then averaged (weighted mean).
4. Raw dimension scores are min-max normalized to 0-100.
5. Composite MyoScore is a weighted sum of the five dimensions.

Interpretation:

Higher scores indicate healthier muscle. The composite MyoScore ranges from 0 (severe myopathy) to 100 (optimal muscle health).

Important Notes:

- Requires >= 20 samples for meaningful min-max normalization.
- Single-sample scoring is not recommended (use a reference cohort).
- Typical bulk RNA-seq datasets contain ~417 of the 1,116 scoring genes.

Value

A data.frame with samples as rows and columns: `Strength_score`, `Mass_score`, `LeanMuscle_score`, `Youth_score`, `Resilience_score`, `MyoScore`.

Examples

```
# Create a small example count matrix (50 genes x 10 samples)
set.seed(42)
genes <- head(MyoScore::myoscore_genes$ID, 50)
counts <- matrix(rpois(50 * 10, lambda = 100), nrow = 50,
                 dimnames = list(genes, paste0("S", 1:10)))

scores <- myoscore_score(counts, verbose = FALSE)
head(scores)
```

`myoscore_score_dimension`

Calculate Score for a Single Dimension

Description

Calculate Score for a Single Dimension

Usage

```
myoscore_score_dimension(
  log2cpm,
  gene_weights = NULL,
  dimension,
  min_coverage = 0.1,
  verbose = TRUE
)
```

Arguments

`log2cpm`	Numeric matrix of log2(CPM+1) values (genes x samples).
`gene_weights`	Data.frame with columns `ID`, `weight`, `direction_v3`, `dimension`.
`dimension`	Character. One of the five MyoScore dimensions.
`min_coverage`	Minimum gene coverage fraction. Default `0.1`.
`verbose`	Logical. Print progress. Default `TRUE`.

Value

Numeric vector of dimension scores (0-100), one per sample.

`myoscore_weights` *Get MyoScore dimension weights*

Description

Get MyoScore dimension weights

Usage

```
myoscore_weights()
```

Value

Named numeric vector of dimension weights (sum to 1.0).

Examples

```
myoscore_weights()
```

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