# Package 'GxEprsDummy'

June 8, 2023

Title What the Package Does (One Line, Title Case)

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<b>Description</b> What the package does (one paragraph).	
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2 Bcov\_discovery

Bcov_discovery	Covariate data file of the discovery dataset when the outcome is binary This contains covariate information of the individuals in the discovery
	dataset following confounders

# Description

Covariate data file of the discovery dataset when the outcome is binary This contains covariate information of the individuals in the discovery dataset following confounders

# Usage

```
Bcov_discovery
```

# **Format**

A dataframe with 7916 rows and 18 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Standardized covariate

Column 4 Square of the standardized covariate

Column 5 Confounder 1

Column 6 Confounder 2

Column 7 Confounder 3

Column 8 Confounder 4

Column 9 Confounder 5

Column 10 Confounder 6

Column 11 Confounder 7

Column 12 Confounder 8

Column 13 Confounder 9

Column 14 Confounder 10

Column 15 Confounder 11

Column 16 Confounder 12

Column 17 Confounder 13

Column 18 Confounder 14

Bcov\_target 3

Bcov_target	Covariate data file of the target dataset when the outcome is binary This contains covariate information of the individuals in the target dataset following confounders

# Description

Covariate data file of the target dataset when the outcome is binary This contains covariate information of the individuals in the target dataset following confounders

# Usage

Bcov\_target

# **Format**

A dataframe with 1939 rows and 18 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Standardized covariate

Column 4 Square of the standardized covariate

Column 5 Confounder 1

Column 6 Confounder 2

Column 7 Confounder 3

Column 8 Confounder 4

Column 9 Confounder 5

Column 10 Confounder 6

Column 11 Confounder 7

Column 12 Confounder 8

Column 13 Confounder 9

Column 14 Confounder 10

Column 15 Confounder 11

Column 16 Confounder 12

Column 17 Confounder 13

Column 18 Confounder 14

4 Bphe\_target

3phe_discovery	Phenotype data file of the discovery dataset when the outcome is binary This contains phenotype information of the individuals in the dis-
	covery dataset

# Description

Phenotype data file of the discovery dataset when the outcome is binary This contains phenotype information of the individuals in the discovery dataset

# Usage

```
Bphe_discovery
```

#### **Format**

A dataframe with 7916 rows and 3 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Phenotype (1=controls, 2=cases)

Bphe\_target Phenotype data file of the target dataset when the outcome is binary
This contains phenotype information of the individuals in the target
dataset

# Description

Phenotype data file of the target dataset when the outcome is binary This contains phenotype information of the individuals in the target dataset

# Usage

```
Bphe_target
```

#### **Format**

A dataframe with 1939 rows and 3 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Phenotype (0=controls, 1=cases)

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DummyData	PLINK binary data files This contains DummyData.fam, Dummy-	
	Data.bim and DummyData.bed files, all in one	

#### **Description**

PLINK binary data files This contains DummyData.fam, DummyData.bim and DummyData.bed files, all in one

### Usage

DummyData

#### **Format**

The data files follow the general PLINK format

GWAS\_binary function This function performs GWAS using plink2 and outputs the GWAS summary statistics file with additive SNP effects named  $B_{\rm out.trd.sum}$ 

# **Description**

GWAS\_binary function This function performs GWAS using plink2 and outputs the GWAS summary statistics file with additive SNP effects named B\_out.trd.sum

# Usage

```
GWAS_binary(
  plink_path,
  b_file,
  Bphe_discovery,
  Bcov_discovery,
  thread = 20,
  summary_output = "B_out.trd.sum")
```

# **Arguments**

Name (with file extension) of the phenotype file containing family ID, individual ID and phenotype of the discovery dataset as columns, without heading

Bcov\_discovery

Name (with file extension) of the covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the discovery dataset as columns, without heading

6 *GWAS\_quantitative* 

```
thread Number of threads used summary_output
```

Name of the SNP effects of the GWAS summary statistics file specified by the user

#### Value

This function will perform GWAS and output

```
B_out.trd.sum
```

GWAS\_quantitative

GWAS\_quantitative function This function performs GWAS using plink2 and outputs the GWAS summary statistics file with additive SNP effects named Q\_out.trd.sum

# **Description**

GWAS\_quantitative function This function performs GWAS using plink2 and outputs the GWAS summary statistics file with additive SNP effects named Q\_out.trd.sum

#### Usage

```
GWAS_quantitative(
  plink_path,
  b_file,
  Qphe_discovery,
  Qcov_discovery,
  thread = 20,
  summary_output = "Q_out.trd.sum")
```

# **Arguments**

plink\_path Path to the PLINK executable application

b\_file Prefix of the binary files, where all .fam, .bed and .bim files have a common prefix

Qphe\_discovery

Name (with file extension) of the phenotype file containing family ID, individual ID and phenotype of the discovery dataset as columns, without heading

Qcov\_discovery

Name (with file extension) of the covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the discovery dataset as columns, without heading

thread Number of threads used summary\_output

Name of the SNP effects of the GWAS summary statistics file specified by the user

GWEIS\_binary 7

#### Value

This function will perform GWAS and output

```
Q_out.trd.sum
```

GWEIS\_binary

GWEIS\_binary function This function performs GWEIS using plink2 and outputs the GWEIS summary statistics files with additive SNP effects named B\_out.add.sum and interaction SNP effects named B\_out.gxe.sum

# **Description**

GWEIS\_binary function This function performs GWEIS using plink2 and outputs the GWEIS summary statistics files with additive SNP effects named B\_out.add.sum and interaction SNP effects named B\_out.gxe.sum

# Usage

```
GWEIS_binary(
  plink_path,
  b_file,
  Bphe_discovery,
  Bcov_discovery,
  thread = 20,
  summary_output = "B_out"
)
```

# **Arguments**

 ${\tt plink\_path} \qquad {\tt Path} \ \ to \ the \ PLINK \ executable \ application$ 

b\_file Prefix of the binary files, where all .fam, .bed and .bim files have a common prefix

Bphe\_discovery

Phenotype file containing family ID, individual ID and phenotype of the discovery dataset as columns, without heading

Bcov\_discovery

Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the discovery dataset as columns, without heading

thread Number of threads used

summary\_output

Name (prefix) of the additive or interaction SNP effects of the GWEIS summary statistics file specified by the user

8 GWEIS\_quantitative

#### Value

This function will perform GWEIS and output

```
B_out.add.sum
B out.gxe.sum
```

GWEIS\_quantitative GWEIS\_quantitative function This function performs GWEIS using plink2 and outputs the GWEIS summary statistics files with additive SNP effects named Q\_out.add.sum and interaction SNP effects named Q\_out.gxe.sum

# **Description**

GWEIS\_quantitative function This function performs GWEIS using plink2 and outputs the GWEIS summary statistics files with additive SNP effects named  $Q_{out.add.sum}$  and interaction SNP effects named  $Q_{out.gxe.sum}$ 

#### Usage

```
GWEIS_quantitative(
   plink_path,
   b_file,
   Qphe_discovery,
   Qcov_discovery,
   thread = 20,
   summary_output = "Q_out"
)
```

# Arguments

plink\_path Path to the PLINK executable application

b\_file Prefix of the binary files, where all .fam, .bed and .bim files have a common prefix

Qphe\_discovery

Phenotype file containing family ID, individual ID and phenotype of the discovery dataset as columns, without heading

Qcov\_discovery

Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the discovery dataset as columns, without heading

thread Number of threads used summary\_output

Name (prefix) of the additive or interaction SNP effects of the GWEIS summary statistics file specified by the user

PRS\_binary 9

#### Value

This function will perform GWEIS and output

```
Q_out.add.sum
Q_out.gxe.sum
```

PRS\_binary

PRS\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_out.trd.sum, B\_out.add.sum or B\_out.gxe.sum

# **Description**

PRS\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named  $B_{\text{out.trd.sum}}$ ,  $B_{\text{out.add.sum}}$  or  $B_{\text{out.gxe.sum}}$ 

## Usage

```
PRS_binary(
  plink_path,
  b_file,
  summary_input = "B_out.trd.sum",
  summary_output = "B_trd"
)
```

# Arguments

 ${\tt plink\_path} \qquad {\tt Path} \ \ to \ the \ {\tt PLINK} \ executable \ application$ 

b\_file Prefix of the binary files, where all .fam, .bed and .bim files have a common prefix

summary\_input

Name of the summary statistics file specified by the user

summary\_output

Name of the PRS file generated using provided summary statistics file specified by the user

# Value

This function will output

```
B_trd.sscore
```

10 Qcov\_discovery

PRS\_quantitative

PRS\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named Q\_out.trd.sum, Q\_out.add.sum or Q\_out.gxe.sum

# **Description**

PRS\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named Q\_out.trd.sum, Q\_out.add.sum or Q\_out.gxe.sum

# Usage

```
PRS_quantitative(
  plink_path,
  b_file,
  summary_input = "Q_out.trd.sum",
  summary_output = "Q_trd"
)
```

# **Arguments**

plink\_path Path to the PLINK executable application

b\_file Prefix of the binary files, where all .fam, .bed and .bim files have a common prefix

summary\_input

Name of the summary statistics file specified by the user

summary\_output

Name of the PRS file generated using provided summary statistics file specified by the user

#### Value

This function will output

```
Q_trd.sscore
```

Qcov\_discovery

Covariate data file of the discovery dataset when the outcome is quantitative This contains covariate information of the individuals in the discovery dataset following confounders

# **Description**

Covariate data file of the discovery dataset when the outcome is quantitative This contains covariate information of the individuals in the discovery dataset following confounders

Qcov\_target 11

# Usage

```
Qcov_discovery
```

# **Format**

A dataframe with 6426 rows and 18 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Standardized covariate

Column 4 Square of the standardized covariate

Column 5 Confounder 1

Column 6 Confounder 2

Column 7 Confounder 3

Column 8 Confounder 4

Column 9 Confounder 5

Column 10 Confounder 6

Column 11 Confounder 7

Column 12 Confounder 8

Column 13 Confounder 9

Column 14 Confounder 10

Column 15 Confounder 11

Column 16 Confounder 12

Column 17 Confounder 13

Column 18 Confounder 14

Qcov\_target

Covariate data file of the target dataset when the outcome is quantitative This contains covariate information of the individuals in the target dataset following confounders

# Description

Covariate data file of the target dataset when the outcome is quantitative This contains covariate information of the individuals in the target dataset following confounders

# Usage

Qcov\_target

12 Qphe\_discovery

#### **Format**

A dataframe with 1579 rows and 18 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Standardized covariate

Column 4 Square of the standardized covariate

Column 5 Confounder 1

Column 6 Confounder 2

Column 7 Confounder 3

Column 8 Confounder 4

Column 9 Confounder 5

Column 10 Confounder 6

Column 11 Confounder 7

Column 12 Confounder 8

Column 13 Confounder 9

Column 14 Confounder 10

Column 15 Confounder 11

Column 16 Confounder 12

Column 17 Confounder 13

Column 18 Confounder 14

Qphe\_discovery

Phenotype data file of the discovery dataset when the outcome is quantitative This contains phenotype information of the individuals in the discovery dataset

#### **Description**

Phenotype data file of the discovery dataset when the outcome is quantitative This contains phenotype information of the individuals in the discovery dataset

# Usage

Qphe\_discovery

# **Format**

A dataframe with 6426 rows and 3 columns

Column 1 Family ID

Column 2 Individual ID

Column 3 Phenotype

Qphe\_target 13

Qphe\_target

Phenotype data file of the target dataset when the outcome is quantitative This contains phenotype information of the individuals in the target dataset

# **Description**

Phenotype data file of the target dataset when the outcome is quantitative This contains phenotype information of the individuals in the target dataset

# Usage

```
Qphe_target
```

#### **Format**

A dataframe with 1579 rows and 3 columns

Column 1 Family IDColumn 2 Individual IDColumn 3 Phenotype

```
results_permuted_binary
```

results\_permuted\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

# Description

results\_permuted\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

#### Usage

```
results_permuted_binary(
   Bphe_target,
   Bcov_target,
   n_confounders,
   input_score1 = "B_trd.sscore",
   input_score2 = "B_add.sscore",
   input_score3 = "B_gxe.sscore")
```

#### **Arguments**

Bphe\_target Phenotype file containing family ID, individual ID and phenotype of the target dataset as columns, without heading

Bcov\_target Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the target dataset as columns, without heading

n\_confounders

Number of confounding variables in the target dataset

#### Value

### This function will output

```
Individual_risk_values.txt
```

```
results_regular_binary
```

results\_regular\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

# Description

results\_regular\_binary function This function uses plink2 and outputs PRSs of each individual in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

# Usage

```
results_regular_binary(
   Bphe_target,
   Bcov_target,
   n_confounders,
   input_score1 = "B_trd.sscore",
   input_score2 = "B_add.sscore",
   input_score3 = "B_gxe.sscore")
```

# **Arguments**

Bphe\_target Phenotype file containing family ID, individual ID and phenotype of the target dataset as columns, without heading

Bcov\_target Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the target dataset as columns, without heading

n\_confounders

Number of confounding variables in the target dataset

#### Value

# This function will output

```
Individual_risk_values.txt
```

```
summary_permuted_binary
```

summary\_permuted\_binary function This function uses plink2 and outputs the p value of permuted model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

# Description

summary\_permuted\_binary function This function uses plink2 and outputs the p value of permuted model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

# Usage

```
summary_permuted_binary(
   Bphe_target,
   Bcov_target,
   iterations = 1000,
   add_score = "B_add.sscore",
   gxe_score = "B_gxe.sscore")
```

# **Arguments**

Bphe_target	Phenotype file containing family ID, individual ID and phenotype of the target dataset as columns, without heading
Bcov_target	Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the target dataset as columns, without heading
iterations	Number of iterations used in permutation
add_score	The .sscore file generated using additive SNP effects of GWEIS summary statistics
gxe_score	The .sscore file generated using interaction SNP effects of GWEIS summary statistics

#### Value

# This function will output

```
B_permuted_p.txt
```

```
summary_permuted_quantitative
```

summary\_permuted\_quantitative function This function uses plink2 and outputs the p value of permuted model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named Q\_trd.sum, Q\_add.sum and Q\_gxe.sum

# Description

summary\_permuted\_quantitative function This function uses plink2 and outputs the p value of permuted model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named Q\_trd.sum, Q\_add.sum and Q\_gxe.sum

# Usage

```
summary_permuted_quantitative(
    Qphe_target,
    Qcov_target,
    iterations = 1000,
    add_score = "Q_add.sscore",
    gxe_score = "Q_gxe.sscore")
```

# **Arguments**

Qphe_target	Phenotype file containing family ID, individual ID and phenotype of the target dataset as columns, without heading
Qcov_target	Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the target dataset as columns, without heading
iterations	Number of iterations used in permutation
add_score	The .sscore file generated using additive SNP effects of GWEIS summary statistics
gxe_score	The .sscore file generated using interaction SNP effects of GWEIS summary statistics

# Value

This function will output

```
Q_permuted_p.txt
```

```
summary_regular_binary
```

summary\_regular\_binary function This function uses plink2 and outputs the summary of regular model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named B\_trd.sum, B\_add.sum and B\_gxe.sum

# **Description**

summary\_regular\_binary function This function uses plink2 and outputs the summary of regular model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named  $B_{trd.sum}$ ,  $B_{add.sum}$  and  $B_{gxe.sum}$ 

# Usage

```
summary_regular_binary(
   Bphe_target,
   Bcov_target,
   trd_score = "B_trd.sscore",
   add_score = "B_add.sscore",
   gxe_score = "B_gxe.sscore",
   Model,
   summary_output = "Bsummary.txt",
   risk_output = "Individual_risk_values.txt")
```

# Arguments

Bphe_target	Phenotype file containing family ID, individual ID and phenotype of the target dataset as columns, without heading	
Bcov_target	Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the target dataset as columns, without heading	
trd_score	The .sscore file generated using additive SNP effects of GWAS summary statistics	
add_score	The .sscore file generated using additive SNP effects of GWEIS summary statistics $% \left( 1\right) =\left( 1\right) \left( 1\right)$	
gxe_score	The .sscore file generated using interaction SNP effects of GWEIS summary statistics	
Model	Specify the model number (1: $y = PRS_t + E + PRS_t + x + CONFOUNDERS$ , 2: $y = PRS_a + E + PRS_a + E + CONFOUNDERS$ , 3: $y = PRS_a + E + CONFOUNDERS$ , 4: $y = PRS_a + E + CONFOUNDERS$ , 5: $y = PRS_a + E + CONFOUNDERS$ , 5: $y = PRS_a + E + E^2 + PRS_a + PRS_a + E + CONFOUNDERS$ , where $y + E + E + E + E + E + E + E + E + E + $	
summary_output		

Name of the model summary file specified by the user

risk\_output Name of the file containing risk scores of the target individuals specified by the user

#### Value

# This function will output

```
Bsummary.txt
Individual_risk_values.txt
```

```
summary_regular_quantitative
```

summary\_regular\_quantitative function This function uses plink2 and outputs the summary of regular model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named Q\_trd.sum, Q\_add.sum and Q\_gxe.sum

# **Description**

summary\_regular\_quantitative function This function uses plink2 and outputs the summary of regular model in the target dataset, using pre-generated GWAS and GWEIS summary statistics files named  $Q_{trd.sum}$ ,  $Q_{add.sum}$  and  $Q_{gxe.sum}$ 

#### Usage

```
summary_regular_quantitative(
    Qphe_target,
    Qcov_target,
    trd_score = "Q_trd.sscore",
    add_score = "Q_add.sscore",
    gxe_score = "Q_gxe.sscore",
    Model,
    summary_output = "Qsummary.txt",
    risk_output = "Individual_risk_values.txt")
```

# **Arguments**

Qphe_target	Phenotype file containing family ID, individual ID and phenotype of the target dataset as columns, without heading
Qcov_target	Covariate file containing family ID, individual ID, standardized covariate, square of standardized covariate, and/or confounders of the target dataset as columns, without heading
trd_score	The .sscore file generated using additive SNP effects of GWAS summary statistics
add_score	The .sscore file generated using additive SNP effects of GWEIS summary statistics
gxe_score	The .sscore file generated using interaction SNP effects of GWEIS summary statistics

Model

Specify the model number (1: y = PRS\_trd + E + PRS\_trd x E + confounders, 2: y = PRS\_add + E + PRS\_add x E + confounders, 3: y = PRS\_add + E + PRS\_gxe x E + confounders, 4: y = PRS\_add + E + PRS\_gxe + PRS\_gxe x E + confounders, where y is the outcome variable, E is the covariate of interest, PRS\_trd and PRS\_add are the polygenic risk scores computed using additive SNP effects of GWAS and GWEIS summary statistics respectively, and PRS\_gxe is the polygenic risk scores computed using GxE interaction SNP effects of GWEIS summary statistics.)

summary\_output

Name of the model summary file specified by the user

 $\verb|risk_output| Name of the file containing risk scores of the target individuals specified by the$ 

#### Value

#### This function will output

```
Qsummary.txt
Individual_risk_values.txt
```

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