EpiSNP Report (with EpiGEN)

Ways of evaluating epistasis detection algorithm performance:

1. Number of interacting pairs (true positives) identified.
2. Number of false positives.
3. Position of true positive(s) (averaged if multiple) when ranked by p-value. If any true positives are missing their position is considered to be the “last” in the list.
   1. The above definition will be referred to as penalized true positive position.
   2. Alternatively, average true positive position can be computed only using the detected true positives. This will be referred to as unpenalized true positive position.
4. Runtime (~1second for 1000 SNPs, NA for 100000 SNPs)

Compare these performance parameters across:

1. Multiplicative vs Dominant vs Recessive vs XOR Interaction.
2. Interaction Alpha: 1.25, 1.5, 2, 3, 8, 16.
3. Pure vs Impure.

Each EpiGEN dataset is named as follows: PURE/IMPURE (indicates presence of marginal effects) \_ MULTIPLICATIVE/DOMINANT/RECESSIVE/XOR (indicates type of interaction) \_ ONEPAIR / TWOPAIRS / EIGHTPAIRS (indicates number of interacting disease snp pairs) \_ BASELINEALPHA10 (indicates quantitative phenotype value in absence of marginal and interaction effects) \_ InteractionAlpha 1.25/1.5/2/3/8/16 (indicates strength of interaction effect)

**RESULTS (pages 1-11)**

**MULTIPLICATIVE DATASETS**

Impure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha2

Number of true positives: 0

Number of false positives: 145

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha3

Number of true positives: 0

Number of false positives: 101

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha15

Number of true positives: 0

Number of false positives: 119

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha125

Number of true positives: 0

Number of false positives: 19

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha2

Number of true positives: 0

Number of false positives: 301

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha3

Number of true positives: 0

Number of false positives: 265

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha15

Number of true positives: 0

Number of false positives: 564

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha125

Number of true positives: 0

Number of false positives: 58

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha2

Number of true positives: 0

Number of false positives: 106

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha3

Number of true positives: 0

Number of false positives: 55

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha15

Number of true positives: 0

Number of false positives: 10

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_OnePair\_BaselineAlpha10\_InteractionAlpha125

Number of true positives: 0

Number of false positives: 0

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha2

Number of true positives: 0

Number of false positives: 159

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha3

Number of true positives: 0

Number of false positives: 533

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha15

Number of true positives: 0

Number of false positives: 125

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_TwoPairs\_BaselineAlpha10\_InteractionAlpha125

Number of true positives: 0

Number of false positives: 0

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_EightPairs\_BaselineAlpha10\_InteractionAlpha3

Number of true positives: 0

Number of false positives: 940

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Multiplicative\_EightPairs\_BaselineAlpha10\_InteractionAlpha2

Number of true positives: 0

Number of false positives: 1138

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_EightPairs\_BaselineAlpha10\_InteractionAlpha3

Number of true positives: 0

Number of false positives: 1092

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Multiplicative\_EightPairs\_BaselineAlpha10\_InteractionAlpha2

Number of true positives: 0

Number of false positives: 2124

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

**DOMINANT DATASETS**

Impure\_Dominant\_OnePair\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 297

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Dominant\_OnePair\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 816

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Dominant\_TwoPairs\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 220

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Dominant\_TwoPairs\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 441

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Dominant\_OnePair\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 304

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Dominant\_OnePair\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 821

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Dominant\_TwoPairs\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 376

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_Dominant\_TwoPairs\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 321

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

**RECESSIVE DATASETS**

Impure\_Recessive\_OnePair\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 5

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Recessive\_OnePair\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 7

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_Recessive\_TwoPairs\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 1

Number of false positives: 2956

F1 Score: 0.00067

Average true positive position (unpenalized) ranked by p-value: 27

Average true positive position (penalized) ranked by p-value: 1492.5

Impure\_Recessive\_TwoPairs\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 1

Number of false positives: 1926

F1 Score: 0.0010

Average true positive position (unpenalized) ranked by p-value: 19

Average true positive position (penalized) ranked by p-value: 973.5

Pure\_Recessive\_OnePair\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 1

Number of false positives: 1933

F1 Score: 0.0010

Average true positive position (unpenalized) ranked by p-value: 8

Average true positive position (penalized) ranked by p-value: 8

Pure\_Recessive\_OnePair\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 1

Number of false positives: 3812

F1 Score: 0.00052

Average true positive position (unpenalized) ranked by p-value: 61

Average true positive position (penalized) ranked by p-value: 61

Pure\_Recessive\_TwoPairs\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 2

Number of false positives: 1168

F1 Score: 0.0034

Average true positive position (unpenalized) ranked by p-value: 342

Average true positive position (penalized) ranked by p-value: 342

Pure\_Recessive\_TwoPairs\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 2

Number of false positives: 1912

F1 Score: 0.0021

Average true positive position (unpenalized) ranked by p-value: 126.5

Average true positive position (penalized) ranked by p-value: 126.5

**XOR DATASETS:**

Pure\_XOR\_EightPairs\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 693

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Pure\_XOR\_EightPairs\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 1212

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_XOR\_EightPairs\_BaselineAlpha10\_InteractionAlpha16

Number of true positives: 0

Number of false positives: 776

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

Impure\_XOR\_EightPairs\_BaselineAlpha10\_InteractionAlpha8

Number of true positives: 0

Number of false positives: 776

F1 Score: 0.0

Average true positive position (unpenalized) ranked by p-value: 0

Average true positive position (penalized) ranked by p-value: 0

**DISCUSSION (Page 12-13)**

Unpenalized true positive positions by interaction type.

Multiplicative: 0,0,0,0,0,0,0,0,0,0,0,0,0,0,0,0,0,0,0,0

Dominant: 0,0,0,0,0,0,0,0

Recessive: 0,0,27,19,8,61,342,126.5

XOR: 0,0,0,0

F1 Score by grouped by interaction type.

Multiplicative: 0,0,0,0,0, 0,0,0,0,0, 0,0,0,0,0, 0,0,0,0,0 => 0

Dominant: 0,0,0,0,0,0,0,0 => 0

Recessive: 0, 0, 0.00067, 0.0010, 0.0010, 0.00052, 0.0034, 0.0021 => 0.0011

XOR: 0, 0, 0, 0 => 0

From this point onward assume all true positive positions are penalized.

Multiplicative Interaction Average True Positive Positions (note that 0 means no true positives were detected): 0,0,0,0,0, 0,0,0,0,0, 0,0,0,0,0, 0,0,0,0,0

Dominant Interaction Average True Positive Positions: 0,0,0,0,0,0,0,0

Recessive Interaction Average True Positive Positions: 0,0, 1492.5, 973.5, 8, 61, 342, 126.5

XOR Interaction Average True Positive Positions: 0,0,0,0

EpiSNP could only detect recessive interactions.

Pairs Detected (# of SNP pairs correctly detected / total # of interacting disease SNP pairs) – only count if at least one pair was detected: ½, ½, 1/1, 1/1, 2/2, 2/2