

Visualization of customized XCI escape gene annotations

Step 1: Create a template

The default template (template 1) includes columns for gene, escape state, and start position.

1. CREATE A TEMPLATE

Template Type

- ☒ Template 1 (gene, state)
- ☐ Template 2 (gene, state, tiss_samp)
- ☒ Include 'start' position

 Download Template

Template 1:

gene	samp_state	start

Example data:

gene	samp_state	start
CD99	escape	154021573
CD99	escape	154021573
CD99	inactive	154021573

The term “samp_state” refers to the state of the gene within a sample or tissue. The sample or tissue can be specified by using template 2.

Template 2:

gene	samp_state	sample	start

Example data:

gene	samp_state	sample	start
CD99	escape	STMACH	154021573
CD99	escape	SKINS	154021573
CD99	inactive	LUNG	154021573

The “start” position is optional and only necessary for plotting the genes on the x chromosome coordinate system. Deselecting “Include ‘start’ position” will remove the ‘start’ column for both templates.

Template Type

- ☒ Template 1 (gene, state)
- ☐ Template 2 (gene, state, tiss_samp)
- ☐ Include 'start' position

Click the  Download Template button to download the template in csv format.

Step 2: Upload data set

2. UPLOAD DATA SET

Choose CSV File

Browse...

No file selected

Populate the downloaded template with the necessary data. Note, the final file **must include the column names which reflect one of the available templates.**

Example csv files:

Example A

gene	samp_state	sample	start
ASMTL	escape	S100	1403139
ASMTL	escape	S200	1403139
ASMTL	escape	S300	1403139
ASMTL	escape	S400	1403139
ASMTL	inactive	S500	1403139
ASMTL	escape	S600	1403139
MECP2	inactive	S100	154021573
MECP2	inactive	S200	154021573
MECP2	inactive	S300	154021573
MECP2	escape	S400	154021573
MECP2	escape	S500	154021573
MECP2	escape	S600	154021573

Example B

gene	samp_state	sample
SHOX	escape	AORT
SHOX	escape	AORT
SHOX	escape	AORT
SHOX	inactive	AORT
SHOX	escape	AORT
SHOX	escape	AORT
SHOX	escape	AORT
MECP2	inactive	AORT
MECP2	inactive	AORT
MECP2	inactive	AORT
MECP2	escape	AORT
MECP2	escape	AORT
MECP2	escape	AORT

Click

Browse...

 to select your csv file and upload to the application. Once uploaded, a preview of your data set should be displayed.

Preview of upload:

Download Template

2. UPLOAD DATA SET

Choose CSV File

Browse...

EXAMPLE.csv

Upload complete

3. SUBMIT

Submit Uploaded Data Set

Last published: 2021-05-27 11:13:39 EDT

genes)

Show

25

 entries

Search:

gene	samp_state	sample	start
ASMTL	escape	S100	1403139
ASMTL	escape	S200	1403139
ASMTL	escape	S300	1403139
ASMTL	escape	S400	1403139
ASMTL	inactive	S500	1403139
ASMTL	escape	S600	1403139
MECP2	inactive	S100	154021573
MECP2	inactive	S200	154021573
MECP2	inactive	S300	154021573
MECP2	escape	S400	154021573
MECP2	escape	S500	154021573
MECP2	escape	S600	154021573

gene

samp_state

sample

start

Step 3: Submit data set

Once you've verified your data set, select

Submit Uploaded Data Set

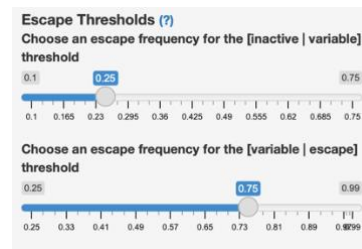
3. SUBMIT

Submit Uploaded Data Set

The application will produce the following levels of information:

■ Escape frequencies and escape statuses based upon samp_state

gene	samp_state	sample	start	escape_freq	state
ASMTL	escape	S100	1403139	0.8333333	escape
ASMTL	escape	S200	1403139	0.8333333	escape
ASMTL	escape	S300	1403139	0.8333333	escape
ASMTL	escape	S400	1403139	0.8333333	escape
ASMTL	inactive	S500	1403139	0.8333333	escape
ASMTL	escape	S600	1403139	0.8333333	escape
MECP2	inactive	S100	154021573	0.5000000	variable
MECP2	inactive	S200	154021573	0.5000000	variable
MECP2	inactive	S300	154021573	0.5000000	variable
MECP2	escape	S400	154021573	0.5000000	variable
MECP2	escape	S500	154021573	0.5000000	variable
MECP2	escape	S600	154021573	0.5000000	variable



Note: the default thresholds applied are:

- $\% \text{escape} \leq 25\% \rightarrow \text{Silenced (S)}$
- $25\% < \% \text{escape} < 75\% \rightarrow \text{Variable escape (VE)}$
- $\% \text{escape} \geq 75\% \rightarrow \text{Escape (E)}$

These can be changed with the sliders in the left side panel.

■ GWAS associations of selected genes of interest

1. Select the Association Data tab

2. Select genes of interest in left side panel

Search Type

Gene

Gene of Interest:

ASMTL MECP2

3. Results:

GWAS Catalog Search (?)

Searching "All Associations v1.02"

Show 25 entries

Search:

Date	Mapped Gene	Disease/Trait	Link	UK Bio Desc.	Ratio (f/m)	Bias
6/25/19	MECP2	snoring	www.ncbi.nlm.nih.gov/pubmed/30804565	snoring		
7/28/18	MECP2	non-albumin protein levels	www.ncbi.nlm.nih.gov/pubmed/29403010			
9/23/16	MECP2	systemic lupus erythematosus	www.ncbi.nlm.nih.gov/pubmed/26502338	systemic lupus erythematosus/sle	7.39622641509434	Female
8/5/14	MECP2	rheumatoid arthritis	www.ncbi.nlm.nih.gov/pubmed/24390342	seropositive rheumatoid arthritis		
8/5/14	MECP2	rheumatoid arthritis	www.ncbi.nlm.nih.gov/pubmed/24390342	rheumatoid arthritis	2.25826086956522	no
8/5/14	MECP2	rheumatoid arthritis	www.ncbi.nlm.nih.gov/pubmed/24390342	other rheumatoid arthritis		

- Genomic positions of genes and escape statuses (if “start” position is included)

