

Visualization of customized XCI escape gene annotations


Step 1: Create a template


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

1. CREATE A TEMPLATE

Template Type

- ☒ Template 1 (gene, state, start, end)
- ☐ Template 2 (gene, state, sample, start, end)

 Download Template

 Download Example

Template 1:

GENE	STATE	START	END

Example data:

GENE	STATE	START	END
CD99	escape	2691187	2741309
ZFX	escape	24149173	24216255
IDS	inactive	149476988	149521096


For template 2, the term “SAMPLE” refers to the sample or tissue of the observed gene status.

Template 2:

GENE	STATE	SAMPLE	START	END

Example data:

GENE	STATE	SAMPLE	START	END
CD99	escape	STMACH	2691187	2741309
CD99	escape	SKINS	2691187	2741309
CD99	inactive	LUNG	2691187	2741309

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

Click the  Download Example button to download an example study 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	Element	START	END
ACSL4	Escape	109641335	109741335
AKAP17A	Escape	1591604	1691604
ASMTL	Escape	1403139	1503139
ATP6AP1	Inactive	154428677	154528677
ATP6AP2	Inactive	40580894	40680894
BTK	Inactive	50580894	50680894
CFP	Inactive	47624214	47724214
CXorf21	Escape	30558809	30658809
CXorf38	Escape	40626921	40726921
DDX3X	Escape	41333284	41433284
DHR SX	Escape	2219506	2319506
DNASE1L1	Escape	154401238	154501238
EDA2R	Inactive	66595637	66695637

Example B

GENE	STATE	SAMPLE	START	END
ACSL4	Escape	HG00146	109641335	109741335
AKAP17A	Escape	HG00146	1591604	1691604
ASMTL	Inactive	HG00146	1403139	1503139
ATP6AP1	Inactive	HG00146	154428677	154528677
ATP6AP2	Inactive	HG00146	40580894	40680894
BTK	Inactive	HG00146	101349447	101449447
CFP	Inactive	HG00146	47624214	47724214
CXorf21	Escape	HG00146	30558809	30658809
CXorf38	Escape	HG00146	40626921	40726921
DDX3X	Escape	HG00146	41333284	41433284
DHR SX	Escape	HG00146	2219506	2319506
DNASE1L1	Escape	HG00146	154401238	154501238
EDA2R	Inactive	HG00146	66595637	66695637
ELF4	Inactive	HG00146	130064874	130164874
FAM3A	Inactive	HG00146	154506159	154606159
FLNA	Inactive	HG00146	154348529	154448529

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

Download Example

2. UPLOAD DATA SET

Choose CSV File

Browse...

TEMPLATE-2-EXAMPLE.csv

Upload complete

3. SUBMIT

Submit Uploaded Data Set

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UPLOADED STUDY [\[download table\]](#)

Filter results

☒ Filter by searched genes (if no genes/diseases are selected, returns all genes)

☐ Filter by displayed genes (zoom in plot)

☐ No filters (return all genes)

Show

25

 entries

Search:

GENE	STATE	SAMPLE	START	END
ACSL4	Escape	HG00146	109641335	109733403
AKAP17A	Escape	HG00146	1591604	1602520
ASMTL	Escape	HG00146	1403139	1453762
ATP6AP1	Inactive	HG00146	154428677	154436516
ATP6AP2	Inactive	HG00146	40580894	40606848
BTK	Inactive	HG00146	101349447	101390796
CFP	Inactive	HG00146	47624214	47630305
CXorf21	Escape	HG00146	30558809	30577766
CXorf38	Escape	HG00146	40626921	40647561
DDX3X	Escape	HG00146	41333284	41364472

Step 3: Submit data set

Once you’ve verified your data set, select

Submit Uploaded Data Set

.

The application will produce the following levels of information:

■ **Escape frequencies and escape status based upon STATE**

GENE	STATE	SAMPLE	START	END	ESCAPE_FREQ
ACSL4	escape	HG00146	109641335	109733403	1
AKAP17A	escape	HG00146	1591604	1602520	1
ASMTL	escape	HG00146	1403139	1453762	1
ATP6AP1	inactive	HG00146	154428677	154436516	0
ATP6AP2	inactive	HG00146	40580894	40606848	0
BTK	inactive	HG00146	101349447	101390796	0
CFP	inactive	HG00146	47624214	47630305	0
CXorf21	escape	HG00146	30558809	30577766	1
CXorf38	escape	HG00146	40626921	40647561	1
DDX3X	escape	HG00146	41333284	41364472	1
DHRSX	escape	HG00146	2219506	2500976	1
DNASE1L1	escape	HG00146	154401238	154412090	1

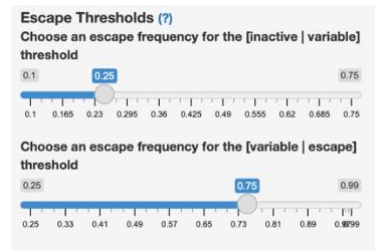
Note: the default thresholds applied are:

- %escape ≤ 25% → Silenced (S)
- 25% < %escape < 75% → Variable escape (VE)
- %escape ≥ 75% → Escape (E)

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

3. SUBMIT

Submit Uploaded Data Set



■ **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:

LAMP2 KDM6A

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
7/17/19	ATP1B4 - LAMP2	waist-to-hip ratio adjusted for bmi	www.ncbi.nlm.nih.gov/pubmed/28552196			
12/14/20	KDM6A	sex hormone-binding globulin levels adjusted for bmi	www.ncbi.nlm.nih.gov/pubmed/32042192			
12/14/20	KDM6A	sex hormone-binding globulin levels	www.ncbi.nlm.nih.gov/pubmed/32042192			

Date

Mapped Gene

Disease/Trait

Link

UK Bio Des

Ratio (f/m)

Bias

Showing 1 to 3 of 3 entries

Previous 1 Next

Visual annotation of XCI status

X-Chromosome Escape Profile

Select an area and double-click to zoom.
To reset, double-click any location, or click the 'Reset' button

