Quick Guide Mouse Nomenclature

Inbred and Hybrid:

Inbred* Fi C57BL/6J=B6 Fi 129S1/SvImJ=129S



*Standard Strain Abbreviations: www.jax.org/jaxmice/support/nomenclature/hints



Site-Directed Endonuclease Mutagenesis: Naming Mutations

ABSTRACT

The Mouse Genome Informatics (MGI, www.informatics.jax.org) Database serves as the authoritative repository of official symbols and names for mouse genes, alleles, and strains, implementing the rules and guidelines established by the International Committee on Standardized Genetic Nomenclature for Mice. MGI hosts the website, http://www.informatics.jax.org/nomen, where the most current version of the guidelines Similar to transgenes, many endonuclease-mutations are often made in an experiment, for mouse and rat nomenclature for genes, alleles and strains can be found. The application of powerful new site-directed endonuclease mutagenesis systems, such as zinc finger nuclease (ZFN), transcription activator-like effector nuclease (TALEN) and clustered regularly interspaced short palindromic repeat, CRISPR associated protein 9 (CRISPR/Cas9), is resulting in a flood of publications reporting the creation of new mutant http://www.informatics.jax.org/mgihome/submissions/amsp_submission.cgi. alleles and strains of mice.

The current guidelines established by the International Committee to designate these endonuclease-mediated mutant alleles use the following format: *Gene^{em#Labcode}* where "Gene" is the gene symbol, "em" designates the allele as endonuclease-mediated, "#" is

a serial number from the laboratory of origin, and "Labcode" is the ILAR-registered laboratory code of the investigator or institution where the mutation was produced. For example, *Scn8a^{em1Mm}* is the first endonuclease-mediated mutation of the *Scn8a* gene produced in the laboratory of Miriam Meisler.

Not every endonuclease-mediated mutation created requires official nomenclature. but only one or a few of the mutations are actually studied or maintained. Those that are published, phenotyped, and maintained are the most important for establishing official allele symbols and names. You can reserve nomenclature for your endonucleasemediated mutations by submitting to MGI at

Nomenclature Guidelines are available at <u>www.informatics.jax.org/nomen</u>. Authors can email MGI nomenclature staff at nomen@jax.org to reserve new nomenclature prepublication or for help with determining correct nomenclature. Supported by NIH grant HG000330.

NOMENCLATURE

When does an 'em' allele receive official nomenclature?

- 1. Unambiguously defined at the molecular level.
- 2. Germ line transmission is achieved.
- 3. A strain with a defined allele is actively maintained or cleanly cryopreserved.

Allele

New 'em' Allele

Name endonuclease-mediated mutation 1, Jackson

Abca16^{em1J} Symbol 3. lab code 1. Mutation type 2. number



Gene^{em3Lab}

1. Endonuclease-mediated mutation (em) 2. Serial number for all em alleles of this gene generated by a specific lab

3. Unique 1 to 5 letter ILAR-registered lab code for generating laboratory

Revertant Allele

Endonuclease-mediated mutations that revert an existing mutant phenotype receive revertant nomenclature.

endonuclease-mediated reversion 1, Name Jinsong Li Cryqc^{M1Sbao+em1Jsli} Symbol

Mutation being reverted

Targeting Existing Alleles

Endonuclease-mediated mutations of existing alleles and mutations that do not revert the phenotype receive standard 'em' nomenclature with the next serial number for 'em' mutations

$X \ge 10$	C57BL/6J
congenic)	
DC 120C1	Compempilat

B6.129S1-Gene^{em2Lab}

Generation Definitions

- Parental mouse in which mutation occurs (not included in the generation definition of subsequent generations)
- **p** (lower case) indicates cryo**p**reservation generation
- N backcross generation number
- Filial or inbreeding generation number (sister X brother)
- + generation numbers before the "+" took place prior to mutation; after the "+" are generation numbers after mutation (*e.g.* F12+F6)

Coisogenic C57BL/6J-A <i>lb^{em8Mvw}/</i> MvwJ		<u>Congenic</u> B6.Cg-Alb ^{em12Mvw} Fcgrt ^{tm1Dcr} /MvwJ		
Strain generated and	Strain lab code	Strain of origin (Cg for unk	-	

given lab.

*Mosaicism has been	reported in ZFN	I and CRISPR/Cas	s founders (Olive	er D <i>et. Al.</i>	2015 PLoS
One 10(6):e0129457)					

Strain

Mouse Colony GENERATION DEFINITIONS JAX labs

	Number of backcross generations.
	Examples: N1, first backcross generation; N2, second
Ν	backcross generation.
	Filial or inbreeding (sister X brother) generations.
	Examples: F1, first filial generation; F2, second filial
F	generation.
	Designates the generation when a strain was
	cryo p reserved.
	Example: F10p indicates a strain that was inbred for
р	10 generations and then cryopreserved.
	Indicates the generation of a strain upon arrival at
	The Jackson Laboratory. Generation numbers before
	the "+" took place in the lab of the donating
+	investigator; after the "+", at The Jackson Laboratory.
	Used when the prior breeding history of a strain is not
	known.
	Example: F?+F12, after arrival at The Jackson
	Laboratory, we interbred for 12 generations, prior to
?	that remains unknown.
	N Equivalent is used when a mouse from a
	N-E quivalent is used when a mouse from a
	subsequent backcross generation is crossed back to a
NE	mouse from a prior backcross generation.
	G eneration is used primarily in mutagenesis schemes.
	Examples: G0 is the mutagenized generation; G1 is
G	the first generation after mutagenesis.
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https://www.jax.org/jax-mice-and-services/customer-support/technical-support/breeding-and-husbandry-support/generation-definitions