Identification of 129S6/SvEvTac-Specific Polymorphisms on Mouse Chromosome 11.

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Abstract: Polymorphisms such as single-nucleotide polymorphisms (SNPs) and insertions/deletions (Indels) can be associated with phenotypic traits and be used as markers for disease diagnosis. Identification of these genetic variations within laboratory mice is crucial to improve our understanding of the genetic background of the mice used for research. As part of a positional cloning project, we sequenced six genes (Mettl16, Evi2a, Psmd11, Cct6d, Rffl, and Ap2b1) within a 6.8-Mb domain of mmu chr 11 in the C57BL/6J and 129S6/SvEvTac inbred strains. Although 129S6/SvEvTac is widely used in the mouse community, there is very little current (or projected future) sequence information available for this strain. We identified 6 Indels and 21 novel SNPs and confirmed genotype information for 114 additional SNPs in these 6 genes. Mettl16 and Ap2b1 contained the largest numbers of variants between the C57BL/6J and 129S6/SvEvTac strains. In addition, we found five new SNPs between 129S6/SvEvTac and 129S1/SvImJ within the Ap2b1 locus. Although we did not detect differences between C57BL/6J and 129S6/SvEvTac within Evi2a, this locus contains a relatively high SNP density compared with the surrounding sequence. Our study highlights the genetic differences among three inbred mouse strains (C57BL/6J, 129S6/SvEvTac, and 129S1/SvImJ) and provides valuable sequence information that can be used to track alleles in genomics-based studies. [ABSTRACT FROM AUTHOR]

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