A cauliflower remutation to Bmp5

Authors: Jill Giggey, Joiel Bauschatz, Michelle Curtain, Julie Hurd and Leah Rae Donahue, Ph.D.

Source of Support: This research was supported by a grant awarded to The Jackson Laboratory by The National Eye Institute titled "Gene Discovery For Craniofacial Disorders" (R01 EY015073- Dr. Leah Rae Donahue, PI).

Mutation (allele) symbol: *Bmp5*^{cfe-se8J}

Mutation (allele) name: cauliflower ear short ear 8 Jackson

Gene symbol: *Bmp5*

Strain of origin: CByB6 Hybrid

Current strain name: CBy;B6-Bmp5^{cfe-se8J}/J

Stock #005421 (JaxMice.jax.org)
Phenotype Category: craniofacial

Discoverer: Stephanie Stark

We report here a new cauliflower remutation (*cfe-se8J*) to Bmp5 that arose spontaneously in the CbyB6 hybrid colony. The recessive mutation was discovered at the Jackson Laboratory in the ENU Mutagenesis Program in 2005. The homozygous mutation is characterized by small ears and ruffles around the perimeter of the ear pinnae. The stock is currently maintained by mating a BALB/cByJ male or female to a *cfe-se8J/cfe-se8J* of the opposite sex. F1 progeny are then mated together to produce mutants in the F2 progeny. Both male and female mutants and control littermates are fertile; females have normal litter sizes and lactate frequently. An allele test with C.129S7-Gt(ROSA)26Sor-*Bmp5*^{cfe-se7J} confirmed the remutation to *Bmp5*. A female *Bmp5*^{cfe-se7J}/*Bmp5*^{cfe-se7J} was mated to a male *Bmp5*^{cfe-se8J}/*Bmp5*^{cfe-se8J} and 16 out of 16 pups had small ears with ruffled ear pinnae.

Phenotypically, mice with the short ear mutation develop characteristic skeletal defects, including reduction of the external ear, loss of several small bones, alterations in size or shape of the xiphoid process, reduction of ventral processes at the sixth cervical vertebrae and deletion of one pair of ribs (Green and Green, 1946; Green, 1951, 1968). An x-ray of $Bmp5^{cfe-se7J}/Bmp5^{cfe-se7J}$ mice did not display these characteristic phenotypes and neither does this remutation.