Cocoa 8 Jackson, a new spontaneous mouse mutation in the *Hps3* gene

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Mutation (allele) symbol: *Hps3*$^{coa\text{-}8J}$

Mutation (allele) name: cocoa 8 Jackson

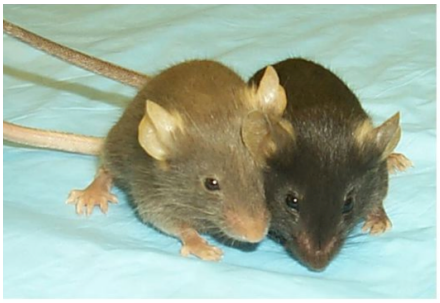
Gene symbol: *Hps3*

Strain of origin: C57BL/6J

Current strain name: C57BL/6J-*Hps3*$^{coa\text{-}8J}$/GrsrJ

Stock #007711 (jaxmice.jax.org)

Phenotype categories: Coat color



A mouse homozygous for the *Hps3*$^{coa\text{-}8J}$ mutation on the left and an unaffected control littermate on the right, at 24 days of age.

Origin and Description

The recessively inherited cocoa 8 Jackson (*Hps3*$^{coa\text{-}8J}$) mutation arose spontaneously and was found by Vicki Hutchinson in 2005 in a production colony of C57BL/6J mice at the Jackson Laboratory. Like previously described *Hps3*^{coa} mutants, mice homozygous for the *Hps3*$^{coa\text{-}8J}$ mutation have a lighter coat color than littermate controls and can be recognized when their first coat of hair comes in at about 10 days of age. Heterozygotes have a normal C57BL/6J coat color (black). Both heterozygous and homozygous mice live a normal lifespan and breed well. The originally described *coa* mutants had prolonged bleeding associated with a platelet defect. Blood work was not performed on the new *Hps3*$^{coa\text{-}8J}$ mutants, so it is uncertain that they carry the platelet defect.

Genetic Analysis

Using standard MMR mapping protocols, a linkage cross was performed by mating a female homozygous for the *Hps3*$^{coa\text{-}8J}$ mutation to a wild type CAST/Ei male mouse. The unaffected F1 progeny from this mating were intercrossed and produced 48 affected mice of which 21 were used for linkage analysis. The mutation was mapped to the region of Chromosome 3 where the *Hps3*^{coa} is located, between *D3Mit176* (NCBI 36 position 22.1 Mb) and *D3Mit268* (NCBI 36 position 28.9 Mb). Because of the similarity of phenotype of *Hps3*^{coa} mutants to this new mutation a direct test for allelism was performed. A female heterozygous for the *Hps3*$^{coa\text{-}8J}$ mutation was mated to a C3H/HeJ-*Hps3*$^{coa\text{-}7J}$/J male heterozygote. This mating produced 9 progeny of which 2 were affected with the *coa* phenotype proving allelism.

Pathology

A routine pathological screen of one homozygous *Hps3*$^{coa\text{-}8J}$ mutant mouse and a littermate control at 9 weeks age showed no gross abnormalities.

Hearing as assessed by auditory-evoked brainstem response testing of one homozygous *Hps3*$^{coa\text{-}8J}$ mutant mouse at 12 weeks age revealed no hearing loss.

The eyes of one homozygous *Hps3*$^{coa\text{-}8J}$ mutant mouse at 8 weeks age were examined with an ophthalmoscope and showed a normal eye phenotype.

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