

## Blunt tail, a spontaneous mouse mutation

Authors: Son Yong Karst, Melissa L. Berry, David E. Bergstrom, and Leah Rae Donahue

Source of Support: This research was supported by NIH/NCRR grants RR01183 and OD010972-35 to the Mouse Mutant Resource (Leah Rae Donahue, PI) and Cancer Center Core Grant CA34196

Mutation (allele) symbol: *blnt*

Mutation (allele) name: blunt tail

Current strain name: B6(C)-*blnt*/GrsrJ

Stock #024009 (jaxmice.jax.org)

Phenotype categories: tail

### Origin and Description:

A novel mutation that alters the tail tip morphology arose spontaneously in the laboratory of Dr. Derry Roopenian in a predominantly C57BL/6J strain congenic for a portion of Chromosome 9 from BALB/cJ. This mutation results in a slightly shorter than normal tail with a blunted, not tapered, tip. This mutation also causes irregular regions of reduced pigment on the tail. This tail defect can be discerned by two or three weeks of age. Mutants are fertile and live a normal life span. This mutant subline has been maintained via backcross-intercross breeding to C57BL/6J such that most or all of the BALB/cJ derived sequence has likely been bred out. We have named this mutation blunt tail, *blnt*.



The distal tail from a *blnt* homozygote on the left and a sibling control on the right

**Genetic Analysis:**

Mutant animals were outcrossed to FVB/NJ mice to establish heritability. No affected mice were found in the F1 generation. Intercrossing unaffected F1 animals produced affected F2 animals, indicating a recessive mode of inheritance. A population of F2 mice was generated for linkage analysis and fine mapping. Using standard SNP protocols, linkage analysis for this mutation was completed in the Fine Mapping Laboratory at The Jackson Laboratory. This mutation mapped to Chromosome 10, between position 53,898,997 bp and position 91,119,681 bp (MGSCv37).

**Pathology:**

A routine pathological screen of three mutants and a control mouse at 21 to 22 weeks of age found no distinct abnormalities. The eyes of one homozygote and one control at age 20 weeks were tested by electroretinography (ERG) and showed normal ERG response and normal retinal function. Hearing assessments by auditory brainstem response testing (ABR) of one homozygote and one control at age 28 weeks showed normal hearing.

**Acknowledgements:**

We thank Dr. Derry Roopenian for discovery of the mutant, Dr. Roderick Bronson and Coleen Kane for pathological screening, Chantal Longo-Guess for hearing assessment, and Ron Hurd for eye examinations.