Lightning bolt tail (*Bolt*): a new dominant mutation that affects the tail and spinal vertebrae in the mouse

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Mutation (allele) symbol: *Bolt*

Mutation (allele) name: Lightning bolt tail

Gene symbol: *Bolt*

Strain of origin: (BALB/cJ x A/J) F1

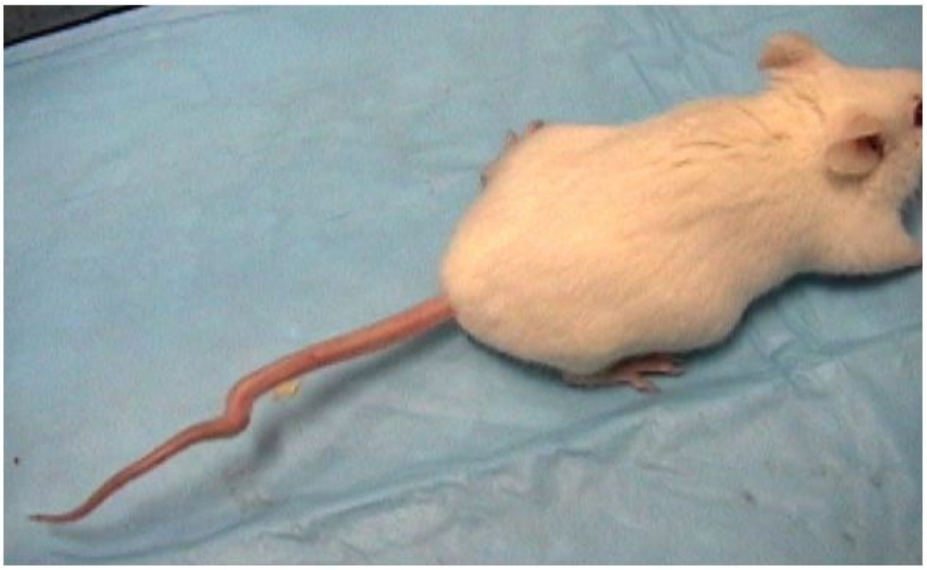
Current strain name: B6.Cg-*Bolt*/GrsrJ

Stock #11075 (jaxmice.jax.org)

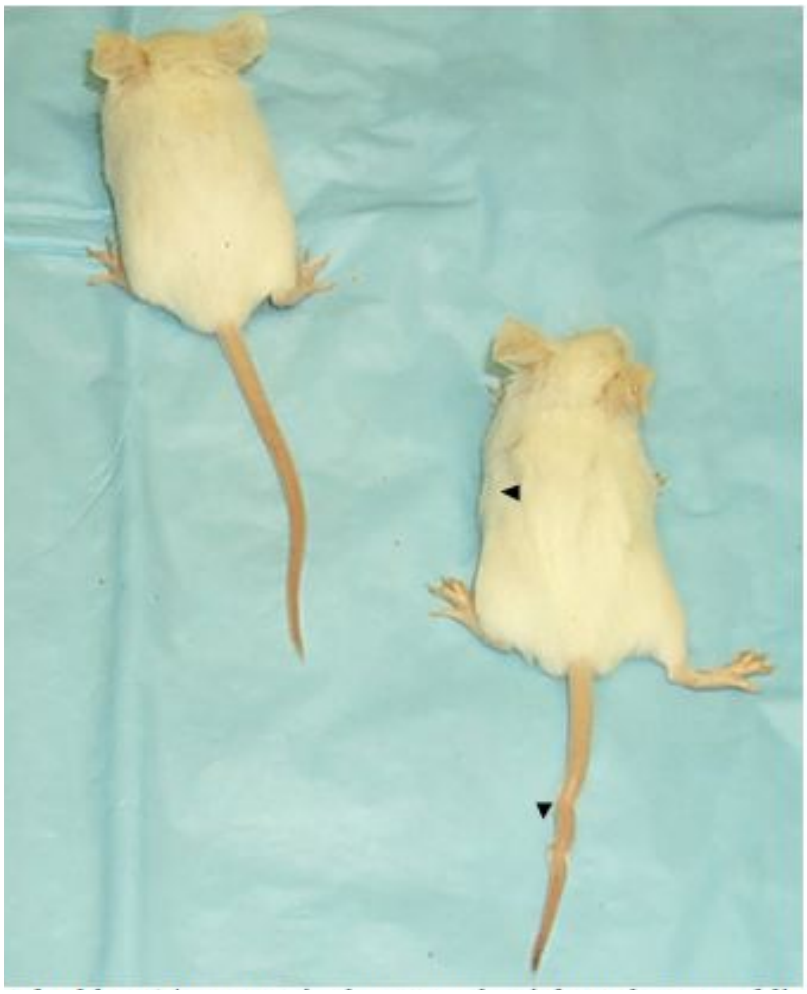
Phenotype categories: skeletal

Abstract
A new dominant skeletal mutation named lightning bolt tail (*Bolt*) has been found at The Jackson Laboratory. Mice carrying the *Bolt* mutation have kinked tails and shortened spines. This new mutation has been mapped to Chromosome 11.

Origin and Description
This novel skeletal mutation was found in 1998 in a production colony of (BALB/cJ x A/J) F1 mice at The Jackson Laboratory and was first recognized by its kinked tail. Mice carrying the *Bolt* mutation can be identified shortly after birth by their variable-length kinked tail and curved spine. Both sexes are viable and live a normal lifespan. The *Bolt* colony is maintained by mating heterozygous females or males to normal wild type littermates.



A mouse carrying the *Bolt* mutation. Note the kinked tail.



A 3-week-old *Bolt*/+ mouse is shown on the right and a control littermate on the left. Note arrows showing the kinked tail and curved spine.

Genetic Analysis

In order to determine the mode of inheritance for the *Bolt* mutation, a female C3H/HeSnJ mouse was mated to a *Bolt*/+ male. This mating produced two *Bolt*/+ and seven wild type progeny, proving this to be a dominant mutation. Heterozygotes mated to heterozygotes produce both normal and *Bolt* phenotypes. The ratio of *Bolt* phenotypes born is 25%, which is much lower than the expected 75%, suggesting prenatal lethality of *Bolt* homozygotes.

Using the standard mapping protocols of The Mouse Mutant Resource a linkage cross was performed by mating a heterozygous female *Bolt*/+ to a male CAST/Ei (Mus castaneus). This mating produced six *Bolt*/+ phenotypes out of seventeen progeny in the F1 generation. The heterozygote F1 *Bolt*/+ animals were backcrossed to the wildtype background strain. The backcross matings produced 153 progeny (49 Nm/+ and 104 +/+) of which 70 were utilized to map the *Bolt* mutation to Chromosome 11. This mutation maps between *D11Mit14* and *D11Mit128*. A search of the Mouse Genome database between these two flanking markers found no known genes with a phenotype similar to the *Bolt* phenotype. Two possible candidate genes in this interval are *Wnt3* and *Wnt9b*

because a hypomorphic mutation of another *Wnt* gene, vestigial tail *(Wnt3a*^{vt}) causes abnormal tail development.

Pathology

X-Rays were done on two heterozygous mutants. They were found to have kinked tails and abnormal spinal vertebral bodies. When mice carrying the mutation are picked up by their tails, their rear legs are oriented in abnormal positions. The lumbar spine has irregularly placed vertebral bodies.

Auditory brain stem response (ABR)[1] testing was done on three female heterozygotes and two controls. Severe hearing loss was found in all mice tested, but is likely an effect of the A/J strain background.

Eyes of two female wildtype and two male heterozygotes (*Bolt*/+) were examined using an ophthalmoscope and found to be normal for a BALB/cJ X A/J strain background. Spinal sections are being processed for pathology. Check back in the near future for additional pathological information.

Discussion

We report a new dominant mutation named lightning bolt tail that is located on mouse Chromosome 11. This new mutation affects the tail and vertebral spine of affected animals. The phenotype is clearly visible on X-Rays as well as by direct observation of the mice.

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[1] **ABR thresholds** in mice are determined using a semi-automated computer system (Intelligent Hearing Systems, Miami, Florida). Subdermal needle electrodes are inserted at the vertex and ventrolaterally to both ears of anesthetized mice. Specific auditory stimuli from 10-100 dB SPL are delivered binaurally through plastic tubes from high frequency transducers. ABR thresholds are obtained, in an acoustic chamber, for clicks and for 8, 16, and 32 kHz pure-tone pips.