Brachyury 11 Jackson

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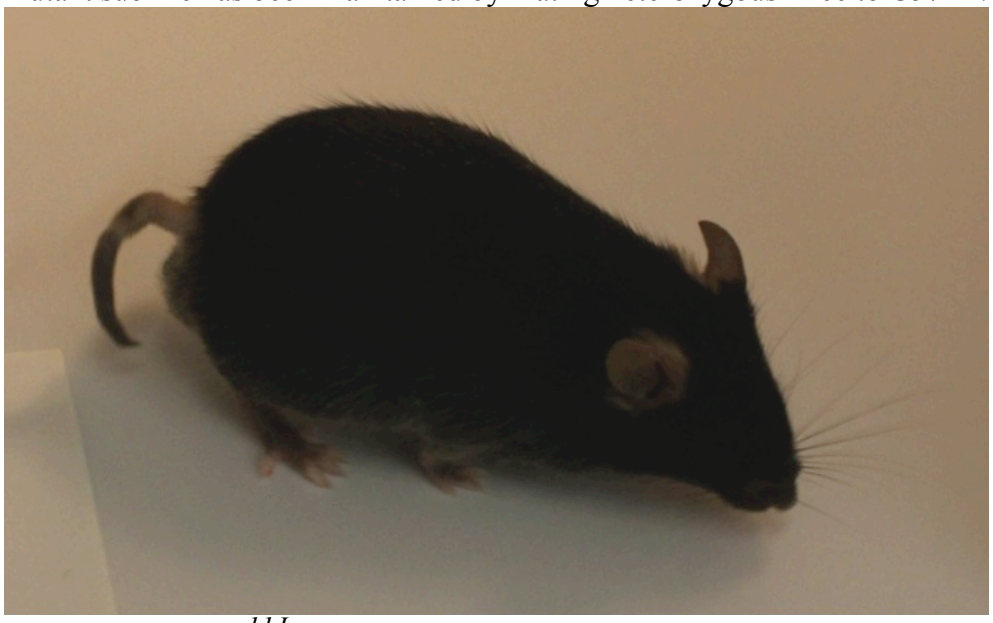
Mutation (allele) symbol: T^{11J}
Mutation (allele) name: Brachyury 11 Jackson
Gene symbol: *T*
Strain of origin: C57BL/6J
Current strain name: C57BL/6J-T^{11J}/GrsrJ
Stock #018072 (jaxmice.jax.org)
Phenotype categories: skeletal

A dominant mutation that causes mice to have variable tail lengths and defects was discovered in the progeny of an N-ethyl-N-nitrosourea (ENU) treated C57BL/6J male at the Jackson Laboratory. The tail phenotype has been found to vary and has included nearly normal length with a blunt or kinked tail, a high set tail with an abnormal point, or almost no tail at all. A routine pathological screen was done of three mutant and three control mice at the age of 3 weeks. Two of the 3-week-old mutants littermates were found to have no anal opening to the outside, and accordingly were small and had intestinal blockage with hard compacted feces in the caecum and colon. Both had reduced pelvic size and the hip bones were closer together than normal. The eyes of two mutant and two control mice at ages 5 and 6 weeks were examined by ophthalmoscopy and found to be normal. Hearing assessment by auditory brainstem response testing (ABR) of two mutant and two control mice showed normal hearing at 5 and 6 weeks of age. This mutant subline has been maintained by mating heterozygous mice to C57BL/6J.



8-week-old male T^{11J} heterozygote

For genetic analysis, a mutant was mated to 129S1/SvImJ and produced affected progeny, proving this mutation to be dominant. Crossing the affected F1 offspring to a C57BL/6J generated affected N2 offspring for linkage analysis. Using the standard mapping procedure of The Mouse Mutant Resource this mutation was mapped to Chromosome 17.

Once linkage to Chromosome 17 was established, brachyury (*T*) was the most likely candidate gene based on known phenotypic characteristics and inheritance patterns. Sanger-based sequencing of *T* exons identified a T to A transversion at position 8,629,002 on Chromosome 17 (NCBI37/mm9). This mutation in exon 3 of *T* results in the nonsense mutation Y166*. Potential effects of this mutation include the production of a truncated protein, or functional null with transcripts degraded by nonsense mediated decay. This mutation has been assigned the allele designation brachyury 11 Jackson (*T11J*).

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