# Dominant short tail: A new dominant skeletal mutation

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Mutation (allele) symbol: *Dsht*

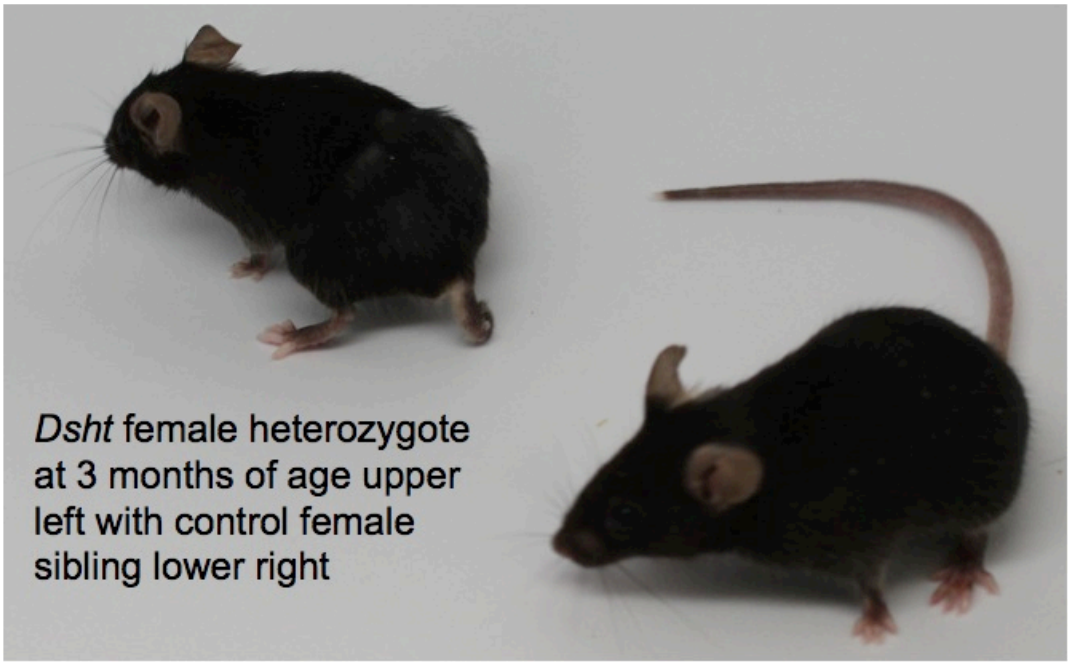
Mutation (allele) name: Dominant Short Tail

Strain of origin: B6.129S2-*Tap1*$^{tm1Arp}$/J

Stock #012654 (jaxmice.jax.org)

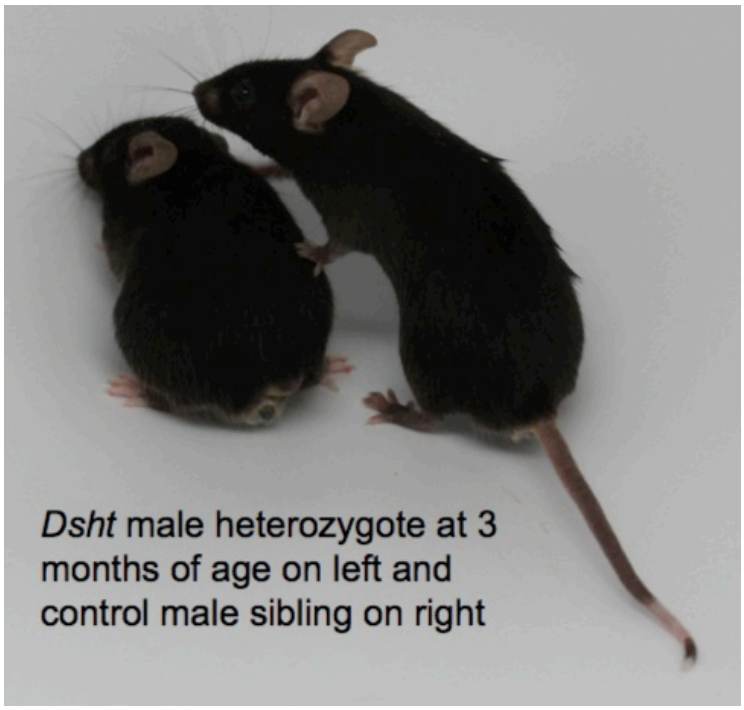
Phenotype categories: skeletal

**Abstract**
We have identified a new dominant mouse mutation that causes a short tail, thin vertebral bones, slightly smaller overall size than littermates, and an occasional belly spot. We have named this mutation dominant short tail (*Dsht*), have mapped it to Chromosome 1, and have found a genetic background dependence for expression.




**Origin and Description**
This mutation was found in the B6.129S2-*Tap1*$^{tm1Arp}$/J colony at The Jackson Laboratory by Theresa Fennelly and was first recognized by its short tail. Heterozygotes can be recognized immediately after birth by their short tails, and they usually have a smaller overall size than their littermates that can also be detected at birth. The tail varies

from being very small, approximately an eighth to a quarter of an inch and wound close to the body, to being approximately a half-inch and tightly kinked or curled up at only the distal end (see photos). The decrease in overall body size varies and is not generally pronounced. At two days of age one heterozygous female was found to weigh 8% less than the average of three unaffected sibling females; at 4 months of age 2 mutant females were only 8% lighter than 3 wildtype female siblings and one heterozygous male was less than 4% lighter than one wildtype male littermate; at nine months of age one heterozygous female was 78% the average weight of 2 wildtype females and one heterozygous male was 78% the weight of one wildtype male.




Both female and male heterozygotes breed and live a normal lifespan. One mating of a heterozygote to a heterozygote produced three heterozygotes, three normal, and four progeny that died in the first week of life. These may have been homozygotes dying after birth as no homozygotes have ever been identified. Normal siblings mated together produced only normal progeny, suggesting that there is complete expressivity in the heterozygote. This colony is maintained by breeding a female or male heterozygote to a normal sibling.

**Genetic Analysis**

Using the standard mapping procedure of The Mouse Mutant Resource, heterozygotes were bred to C3H/HeSnJ and the offspring were assessed for phenotype and genotyped for linkage analysis. *Dsht* mapped to Chromosome 1 near *D1Mit242*, which showed only one recombinant in 44 meioses tested (2.4%). This places *Dsht* near the proximal end of Chromosome 1. *D1Mit316*, *D1Mit66*, and *D1Mit430*, which are more distal than *D1Mit242*, each showed 8 recombinants in 42 meioses tested (19%).

Although this is a dominant mutation fewer than the expected 50% have the mutation. This is not due to incomplete expressivity because intercrossing unaffected siblings of

affected mice on a predominantly C57BL/6J background never yielded any affected offspring in more than 35 pups. This strain was outcrossed to three other strains with two different results. Outcrossing to CAST/EiJ did not produce any affected progeny in the F1 generation or in the backcross. Backcrossing to C57BL/6J and outcrossing to C3H/HeSnJ did express the short tail phenotype in the F1 generation and also in the offspring of a second backcross generation (N2), though at a percentage rate much less than the expected 50% (generally seen was 10% affected or less). Thus, some pre-wean lethality may play a factor in the number of mutants identified on these backgrounds.

**Pathology**

Routine pathological screening[1] showed two nine-month-old and one one-month-old heterozygote having abnormally thin vertebral bones. Hearing tests on one female wild type and one male heterozygote at approximately 14 weeks of age showed normal hearing. Eye examinations of three male and three female controls and four heterozygotes had normal eyes and ERGs. Five other heterozygotes ranging from three weeks to five months of age had one of their eyes affected with corneal holes and/or iris colobomas, with all having normal ERGs.

**Discussion**

We report a new dominant mutation named dominant short tail, which we have mapped to Chromosome 1. It is characterized by a short tail and is maintained by using a heterozygote x a wild type mouse, either direction as both sexes of heterozygotes are fertile.

**Acknowledgements**

We thank the late Norm Hawes for eye examinations, Heping Yu for ABR testing, and Coleen Kane for histological preparations.

[1]**Standard Histology Protocol used in The Mouse Mutant Resource**

For fixation of tissues, mice were deeply anesthetized with tribromoethanol (avertin) until they no longer displayed a withdrawal reflex in the hind limbs and then perfused intracardially with Bouin's fixative following a flush of the vasculature with saline solution. After soaking in Bouin's for one week to demineralize bones, tissues were dissected. Six segments of spine with axial muscles and spinal cord in situ, representing cervical, thoracic and lumbar spinal segments, were dissected. The brain was removed and sliced into 6 cross sectional pieces at the levels of olfactory lobes, frontal cortex, striatum, thalamus, midbrain, rostral and caudal medulla with cerebellum. Midsagittal slices of hind leg through the knees were prepared. Slices of basal skull through the pituitary and inner ears were taken. Both eyes, salivary glands and submandibular lymph node, trachea plus thyroid and sometimes parathyroid were removed and cassetted. A longitudinal slice of skin from the back was removed. The thymus, slices of lung, and a longitudinal slice of heart were cassetted. Similarly slices of liver through gall bladder, kidney with adrenal attached, pancreas and spleen were prepared. The stomach was sliced longitudinally to include both squamous and glandular portions. Loops of small intestine from 3 levels and slices of large intestine and cecum were removed, as were slices of urinary bladder. The whole uterus, with ovaries attached, was taken. In males testes were sliced longitudinally. The accessory male organs including seminal vesicles, coagulating gland and prostate were removed en block. Altogether in most cases all tissue fit into a total of 10 cassettes. The cassettes were processed in an automatic tissue processor to dehydrate tissues which were then embedded in paraffin. Six micron sections were cut and stained with hematoxylin and eosin (H&E). Sections of brain and spinal cord in vertebral bones also were stained with luxol fast blue (LFB) for myelin and cresylecht violet (CV) for cellular detail.