Oarleg: A new autosomal recessive mutation causing a limb phenotype

Belinda S. Harris, Patricia F. Ward-Bailey, Muriel T. Davisson-Fahey, and Roderick T. Bronson

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Mutation (allele) symbol: *oar*

Mutation (allele) name: oarleg

Strain of origin: BALB/cByJ

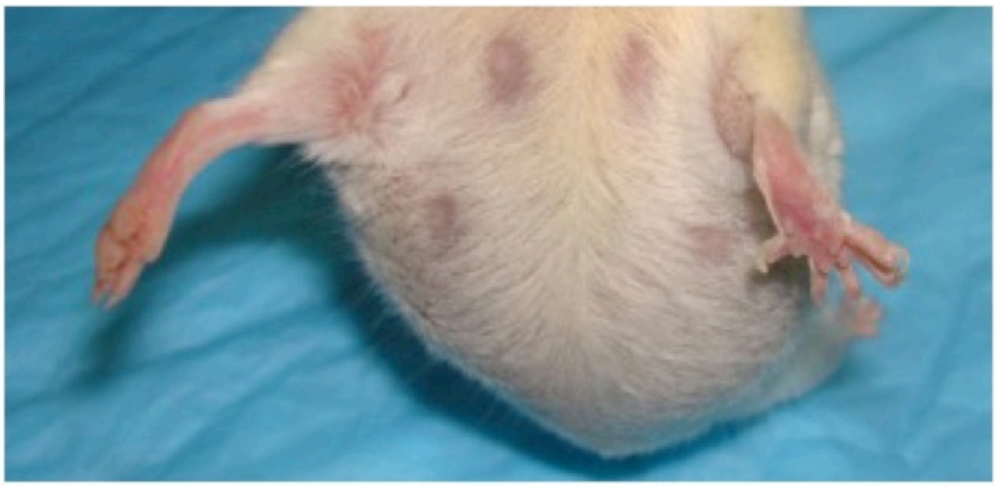
Current strain name: BALB/cByJ-*oar*/GrsrJ

Stock #010637 (jaxmice.jax.org)

Phenotype categories: skeletal/limbs

Abstract
We have identified a new recessive mouse mutation that causes a varying limb position and abnormal gait that can be observed at about two weeks of age. Mice homozygous for the oarleg mutation may have both rear legs affected or just one. They may be smaller in size than their littermate controls. The new oarleg mutation has been mapped to Chromosome 5.

Origin and Description
Oarleg was discovered by Marianne Urquhart in a production colony of inbred BALB/cByJ mice at The Jackson Laboratory in 2001. It was first recognized in an animal that was holding a rear leg at an odd angle as it walked around in its cage. When held by the tail oarleg mutant mice point their toes straight out and hold their limbs stiffly, resembling an oar, unlike littermate control mice who lift their toes upward. When both hind limbs are affected the mice walk with a stiff gait that makes the mouse look like it has a tremor. Both female and male homozygotes are fertile and live a normal lifespan.



The rear legs of a seven-month-old female are shown. Note the affected leg on the left compared to the normal leg on the right.

Genetic Analysis

Using the standard mapping protocols of The Mouse Mutant Resource, an intercross of homozygous oarleg mice mated to inbred C57BL/6J mice was set up and generated 57 F2 progeny that were utilized for linkage analysis. The oarleg mutation was found to be on Chromosome 5 between *D5Mit424*, which had 4 recombinants out of 110 meioses assessed, and *D5Mit95*, which had 1 recombinant out of 110 meioses assessed. A search in Mouse Genome Informatics for candidate genes located between the oarleg flanking markers, and using the terms skeleton and limbs indicated a QTL named femur geometry 4 (*Fmgty4*) as a potential candidate.

Pathology

A pathological screen of two oarleg homozygotes and two controls showed no lesions in somatic organs. X-rays and alizarin preparations of the skeleton showed no obvious reason for the leg extension and the hip joints appeared normal.

Hearing as assessed by auditory brainstem testing (ABR) on two oarleg homozygotes and two controls showed all were normal.

All oarleg and control animals passed a swim test successfully.

Discussion

We have discovered a new recessive mutation located on Chromosome 5 that affects the rear leg or legs of homozygotes. There is no obvious explanation for the odd limb phenotype from the pathology and tests done to date.

Acknowledgements

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References

Mouse Genome Database (MGD) at the Mouse Genome Informatics website, The Jackson Laboratory, Bar Harbor, Maine. (http://www.informatics.jax.org).