Dilute lethal 33 Jackson: A remutation to dilute lethal on Chromosome 9

Belinda Harris, Patricia F. Ward Bailey, Leah Rae Donahue, and David E. Bergstrom

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Mutation (allele) symbol: *Myo5a*$^{d\text{-}l33J}$

Mutation (allele) name: dilute lethal 33 Jackson

Gene symbol: *Myo5a*

Strain of origin B6.129S2-*Ighm*^{tm1Cgn}/J

Current strain name:B6.Cg-*Myo5a*$^{d\text{-}l33J}$ *Ighm*^{tm1Cgn}/GrsrJ

Stock #016100

Phenotype categories: pigmentation, neuromuscular, lethal

Abstract
We have identified the 33rd recessive remutation to dilute lethal to occur spontaneously at The Jackson Laboratory. Homozygotes exhibit the same abnormal body control movements and gray coat color as mice homozygous for the original dilute lethal mutation. A direct test for allelism showed this new remutation to be allelic with dilute lethal 32 Jackson.

Origin and Description
This remutation was found by Brandi Rowell in the strain B6.129S2-*Ighm*^{tm1Cgn}/J at The Jackson Laboratory in 2008, and was initially identified by its gray coat color. Subsequently, the mutant was found to have an abnormal side-to-side leaning gait before it died by approximately four weeks of age. The gray color is evident by seven days of age and the ataxic movements are evident by two weeks of age, in the time period when some homozygous animals die. Others may live to approximately one month of age. As neither female nor male homozygotes lived long enough to breed, the colony was maintained by breeding progeny tested heterozygotes, which are fertile and live a normal lifespan.

Genetic Analysis
A direct test for allelism between two female *Myo5a*$^{d\text{-}l32J}$ heterozygotes and a male heterozygous for this new mutation produced six affected homozygotes out of sixteen total progeny born, with one unclassified pup missing. This failed complementation proved this new mutation to be an allele of *Myo5a* with the dilute lethal phenotype and it was assigned the next remutation designation, *Myo5a*$^{d\text{-}l33J}$.

Pathology
A routine pathological screen of two homozygotes, one female and one male, both 3 weeks of age, showed apoptosis of the thymus and atrophic spleens. The control

littermate male was normal with no lesions.

Discussion

We report a new remutation to dilute lethal which has characteristics similar to the original dilute lethal mutation. This new remutation has been named dilute lethal 33 Jackson and is available from The Jackson Laboratory DNA Resource. No embryos were cryopreserved.

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