

A new allele, *pcd-5J*, of the *Agtbp1* gene

Susan A. Cook, Roderick T. Bronson, Sandra J. Gray, and Muriel T. Davisson

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Mutation (allele) symbol: *pcd-5J*

Mutation (allele) name: Purkinje cell degeneration 5 Jackson

Gene symbol: *Agtbp1*

Strain of origin: DBA/2J

Current strain name: DBA/2J-*Agtbp1*^{*pcd-5J*}/GrsrJ

Stock: #004518 (jaxmice.jax.org)

Origin and Description

The spontaneous *pcd-5J* mutation arose in the DBA/2J production colony (Stock #000671) at The Jackson Laboratory in 2000. Mutant mice were recovered in the second generation from a putative mutation-carrying sire mated to a DBA/2J wildtype mouse, which indicates that *pcd-5J* is inherited as a recessive allele. Penetrance of the mutant phenotype is ~100% as heterozygous parents produced 11 mutant mice from a total of 41 progeny (27%). *pcd-5J/pcd-5J* mice are visibly classified by weaning age and live through adulthood. They present the ataxic gait and severe deficiency of Purkinje cells characteristic of other *Agtbp1* alleles. *pcd-5J/pcd-5J* females may breed but males do not; hence, the colony is maintained by mating homozygous females x heterozygous males or by ovarian transplant with C3SnSmn.CB17-*Prkdc*^{*scid*}/J hosts (Stock # 001131). Qualitative examination by light microscopy of one sperm sample from a 3 month old mutant showed morphology and quantity similar to its sibling control. DNA has been preserved in The Jackson Laboratory DNA Resource. Sperm samples from homozygous and heterozygous males are currently being cryopreserved.

Genetic Analysis

Pathology and visible phenotype warranted a complementation test with BALB/cBy-*Agtbp1*^{*pcd-3J*} (Stock # 003237). Progeny from a +/*pcd-3J* female x +/*pcd-5J* male mating yielded 3 affected mice from a total of 15. A concurrent linkage intercross with CAST/Ei substantiated the positive test for allelism. Using spleen tip DNAs from 22 mutant F2 progeny and standard PCR protocols, we typed 2 recombinant meioses from a total of 44 with *D13Mit99* located 3 cM distal to *Agtbp1* (MGD 2003).

Acknowledgements

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