

A new allele of protocadherin 15 (*Pcdh15*)

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Mutation (allele) symbol: *av-jfb* (originally *av-4J*)

Mutation (allele) name: Ames waltzer James F Battey

Gene Symbol: *Pcdh15*

Strain of Origin: B6;129-*Grpr*^{*tm1.Jfb*}

Current Strain name: B6;129-*Pcdh15*^{*av-jfb*}

Phenotype categories: Hyperactive Circling, Head-bobbing, and Deafness

Origin and Description:

The mutation arose spontaneously on the B6,129-*Grpr*^{*tm1.Jfb*} strain. The *Grpr* targeted mutation was selected against and bred out of the mutant line, and this was confirmed by PCR analysis. Mutants exhibit the classic circling behavior with abnormal head tossing movement. They fail the swim test by repeated rolling and sinking and are also deaf by four weeks as confirmed by ABR analysis.

Genetic Analysis:

An F2 intercross was performed with CAST/Ei and 30 animals were analyzed. Linkage with Chr 10 was established and the mutation was localized between D10Mit31 and *D10Mit175*. A complementation test with *av-3J* (a mutation of *Pcdh15*) confirmed allelism. A heterozygous female *av-3J* mouse was mated with a homozygous male *av-4J* producing a total of 13 pups of which 10 survived to weaning age. In the surviving pups, 3 exhibited the mutant phenotype. The mutant allele was originally designated *av-4J* (Ames Waltzer 4 Jackson). DNA from a homozygous mutant (The Jackson Laboratory DNA Resource #4622) showed the identical PCR- RFLP pattern as *Pcdh15*^{*av-jfb*} (Hampton et al.).

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References

Hampton LL, Wright CG, Alagramam KN, Battey JF, Noben-Trauth K, A new spontaneous mutation in the mouse Ames waltzer gene, *Pcdh15* Hear Res. 2003 Jun;180(1-2):67-75 PMID: 12782354