WashU

Office of Technology Management

NIEMANN-PICK C1 P1007A MOUSE MODEL

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Mouse model for Niemann-Pick C1 (NPC1) disease

This mouse model was generated by CRISPR KI of the P1007A mutation into the NPC1 protein. Mice heterozygous or homozygous for this mutation do not have an apparent phenotype. However, when the heterozgous P1007A mouse is crossed with the NPC1 I1061T mouse, the compound heterozygous mice (P1007A/I1061T) demonstrate lipid storage typical of NPC1 disease. As the P1007A mutation is only found in late onset disease, the compound heterozygous mice likely will serve as the first late onset model of NPC1 disease.