National Human Genome Research Institute

Murine Model of Niemann-Pick C Disease

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KEY WORDS

Niemann-Pick Disease type C, NPC1

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PATENT-PENDING TECHNOLOGY AVAILABLE FOR LICENSING

SUMMARY

NPC1 gene mutations result in Niemann-Pick disease type C (NPC) which is an autosomal recessive neurodegenerative disorder characterized by intracellular accumulation of cholesterol and gangliosides. The Niemann-Pick C deficient mouse model, Npc1-/-, recapitulates the disease phenotype of human patients. These mice, however, die around 8 weeks of age, making it a difficult model to study the observed visceral cholesterol accumulation. NHGRI investigators generated a transgenic (Tg) (Npc1) mouse that when mated to the Npc1-/- model, rescues the disease effects related to neurologic degeneration, allowing for a normal lifespan. The new mouse strain, defined as Tg (Npc1); BALB/c npcnih/nih rescues NPC1 disease but maintains the defects that correspond to the Niemann-Pick C visceral phenotype associated with cholesterol accumulation.

POTENTIAL COMMERCIAL APPLICATIONS

This model would be a useful tool to study both the visceral disease aspects of Niemann-Pick C and the biology of cholesterol accumulation with its effect on visceral organ systems.

RELATED ARTICLES

Loftus et al., Rescue of neurodegeneration in Niemann-Pick C mice by a prion-promoter-driven Npc1 cDNA transgene, 11 Human Molecular Genetics 3107 (2002). http://hmg.oxfordjournals.org/content/11/24/3107.long

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