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Murine Model of Niemann-Pick C Disease

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NPC1

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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 Article

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://hmq.oxfordjournals.org/content/11/24/3107.long>