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### Murine Model of Niemann-Pick C Disease
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**Key Words**
Niemann-Pick Disease type C, 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.

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