



# Gene Therapy for Niemann-Pick Disease Type C

## NHGRI INVENTION:

Number: E-185-2014

## PATENT STATUS

PCT Application PCT/US2016/026524 filed April 7, 2016 National Phases Entered in the U.S., Europe (allowed) and Canada in October 2017

## KEY WORDS

Niemann-Pick Disease Type C, NPC1, Gene Therapy

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## PATENT-PENDING TECHNOLOGY AVAILABLE FOR LICENSING

### SUMMARY

Investigators at the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH) are seeking collaborators to further develop viral gene therapy to treat Niemann-Pick Disease Type C (NPC). NPC is a rare and fatal, autosomal recessive, neurodegenerative disease that can present in infants, children, or adults. Most patients with NPC have mutations in NPC1, a gene implicated in intracellular cholesterol trafficking, which results in intracellular accumulation of unesterified cholesterol in late endosomal/lysosomal structures and of glycosphingolipids, especially in neuronal tissue. Thus, NPC patients generally suffer from enlargement of liver and spleen and neurological degeneration.

NHGRI investigators have generated adeno-associated (AAV) constructs that are able to correct cellular defects of certain cholesterol diseases or disorders, such as NPC, in vivo. These therapeutic constructs include human NPC1 sequence under control of a constitutive promoter elongation factor 1 alpha (EF1alpha) or a neuronal-specific promoter calcium/calmodulin-dependent protein kinase II (CamKII). The constructs were able to significantly increase survival and weight of the NPC mouse model (Npc knockout).

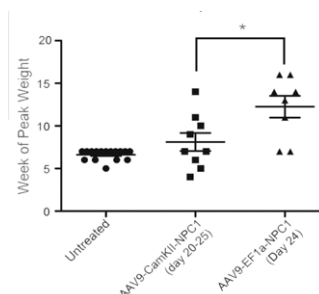
### POTENTIAL COMMERCIAL APPLICATIONS

Results of the construct experiments in mice demonstrate pre-clinical efficacy of AAV gene therapy as a therapeutic approach to NPC.

### RELATED ARTICLES

Chandler et al., Systemic AAV9 gene therapy improves the lifespan of mice with Niemann-Pick disease, type C1, Human Molecular Genetics 26(1):52-64, 2017.

<https://academic.oup.com/hmg/article/26/1/52/2623374>



*AAV-9-EF1alpha-NPC1 treatment significantly increases growth in Npc knockout mice*

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