



Gene Therapy Vector for the Treatment of Glycogen Storage Disease Type Ia (GSD-Ia)

Summary (1024-character limit)

GSD-Ia is an inherited disorder of metabolism associated with life-threatening hypoglycemia, hepatic malignancy, and renal failure caused by the deficiency of glucose-6-phosphatase-alpha (G6Pase-alpha or G6PC). NICHD seeks parties to license this invention towards commercialization.

NIH Reference Number

E-039-2015

Product Type

- Therapeutics

Keywords

- GSD-Ia
- hypoglycemia
- hepatic malignancy
- renal failure

Collaboration Opportunity

This invention is available for licensing.

Contact

- John D. Hewes
NCI - National Cancer Institute

240-276-5515

John.Hewes@nih.gov

Description of Technology

GSD-Ia is an inherited disorder of metabolism associated with life-threatening hypoglycemia, hepatic malignancy, and renal failure caused by the deficiency of glucose-6-phosphatase-alpha (G6Pase-alpha or G6PC). Current therapy, which primarily consists of dietary modification, fails to prevent long-term complications in many patients, including growth failure, gout, pulmonary hypertension, renal dysfunction, osteoporosis, and hepatocellular adenomas (HCA). Gene therapy-based techniques, which directly address the underlying genetic deficiency driving the disorder, offer the prospect of long-term remission in patients with GSD-Ia.

Researchers at the NIH [National Institute for Childhood Health and Diseases](#) have developed an adeno-associated viral (AAV) vector for the treatment of glycogen storage disease type Ia (GSD-Ia). This new



AAV vector that expresses human G6Pase-alpha directed by the tissue-specific human G6PC promoter/enhancer incorporates two improvements: 1) it expresses a variant of G6Pase-alpha with enhanced enzymatic activity; 2) it is codon optimized to achieve higher enzyme expression levels and enhanced enzymatic activity.

In vivo data is available.

Potential Commercial Applications

- Gene therapy vector for the treatment of GSD-Ia

Competitive Advantages

- Protein coding sequence modified for enhanced enzymatic activity.
- Codon optimized for increased enzyme expression in target organs.

Inventor(s)

[Janice J. Chou \(NICHD\)](#)

Development Stage

- Pre-clinical (in vivo)

Publications

Lee YM et al. [\[\[PMID 22422504\]\]](#)

Lee YM, et al. [\[\[PMID 23856420\]\]](#)

Patent Status

- **U.S. Patent Filed:** U.S. Patent Application Number 62/096,400, Filed 23 Dec 2014

Related Technologies

- E-552-2013

Therapeutic Area

- Kidney and the Genitourinary