GENE THERAPY VECTOR FOR THE TREATMENT OF GLYCOGEN STORAGE DISEASE TYPE IA (GSD-IA)

SUMMARY

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).

REFERENCE NUMBER

E-039-2015

PRODUCT TYPE

- Therapeutics

KEYWORDS

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

COLLABORATION OPPORTUNITY

This invention is available for licensing.

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 Insitute 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)

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DEVELOPMENT STAGE

- Pre-clinical (in vivo)

PUBLICATIONS

- Lee YM et al. Hepatology 2012 Nov;56(5):1719-29. [PMID 22422504]; Lee YM, et al. Mol Genet Metab. 2013 Nov;110(3):275-80. [PMID 23856420]

PATENT STATUS

- **U.S. Filed:** US Provisional Patent Application 62/096,400 filed December 23, 2014

RELATED TECHNOLOGIES

- E-552-2013

THERAPEUTIC AREA

- Kidney and the Genitourinary