Treatment for Restoring Ureagenesis in Carbamoyl Phosphate Synthetase 1 Deficiency

Tech ID: 30356 / UC Case 2019-600-0

SUMMARY

UCLA researchers in the Department of Surgery have developed a gene therapy to treat carbamoyl phosphate synthetase 1 deficiency.

BACKGROUND

Carbamoyl phosphate synthetase 1 deficiency (CPS1 deficiency) is an inherited autosomal recessive urea cycle disorder characterized by complete or partial loss of carbamoyl phosphate synthetase 1 enzyme. The urea cycle cannot proceed normally in the absence of CPS1, and nitrogen accumulates in the form of ammonia in the bloodstream. Ammonia is especially damaging to the central nervous system, and excess ammonia causes neurological problems associated with CPS1 deficiency. Complete lack of CPS1 results in the severe form of the disorder, and the symptoms may occur within 24-72 hours after birth, regardless of exposure to dietary protein. An infant with the severe form of CPS1 deficiency may experience seizures, abnormal body movements, and respiratory distress that are mostly attributable to the swelling of the brain caused by hyperammonemia. The disorder may result in neurological abnormalities, including developmental delays and intellectual disability and can be lethal. Patients with the milder form of CPS1 deficiency show symptoms later during infancy, childhood, or adulthood, and also can experience hyperammonemic coma and life-threatening complications. Current treatment for CPS1 deficiency consists primarily of dietary protein restriction, which is only marginally effective and leaves patients vulnerable to recurrent hyperammonemia and progressive, irreversible neurological decline.

INNOVATION

Researchers at UCLA have developed a gene therapy that replenishes CPS1 enzyme in the liver for treating CPS1 deficiency.

APPLICATIONS

- Gene therapy for carbamoyl phosphate synthetase 1 deficiency

ADVANTAGES

- The described gene therapy overcomes the limit of packaging a large cDNA into adeno-associated viral vectors

STATE OF DEVELOPMENT

The described gene therapy has been validated in a conditional CPS1 knock out mouse model.

PATENT STATUS

Country	Type	Number	Dated	Case
United States Of America	Issued Patent	12,281,323	04/22/2025	2019-600

ADDITIONAL TECHNOLOGIES BY THESE INVENTORS

- A Method to Prevent the Myelin Abnormalities Associated with Arginase Deficiency

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OTHER INFORMATION

KEYWORDS

Carbamoyl phosphate synthetase 1 deficiency, CPS1 deficiency, urea cycle disorder, gene therapy

CATEGORIZED AS

- **Medical**
  - Disease: Central Nervous System
  - Disease: Genetic Diseases and Dysmorphic Syndromes
  - Disease: Metabolic/Endocrinology
  - Gene Therapy
  - Therapeutics

RELATED CASES

2019-600-0

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