AAV-Mediated Gene Therapy

Tech ID: 34804 / UC Case 2021-928-0

ABSTRACT

Researchers at the University of California, Davis have developed AAV-mediated gene therapy that delivers the choline acetyltransferase (ChAT) gene to treat presynaptic congenital myasthenic syndromes and related neuromuscular disorders.

FULL DESCRIPTION

This therapy uses recombinant adeno-associated virus (AAV) virions to deliver a functional copy of the ChAT gene to patients suffering from presynaptic congenital myasthenic syndromes (CMS) caused by ChAT deficiency. By restoring ChAT enzyme activity at the neuromuscular junction and in the central nervous system (CNS), the treatment aims to improve or prevent severe muscle weakness and respiratory complications associated with CMS, as well as address other conditions linked to ChAT deficiency such as myasthenia gravis and Alzheimer's disease. Subjects can be diagnosed via genetic, electrodiagnostic, or blood tests, allowing targeted therapeutic intervention.

APPLICATIONS

- Gene therapy for genetic neuromuscular disorders like congenital myasthenic syndromes.
- Treatment of myasthenia gravis and other autoimmune-related neuromuscular diseases.
- Therapeutic intervention for Alzheimer's disease and dementia linked to ChAT deficiency.
- Biomedical research tools for studying neuromuscular junction diseases and gene editing.
- Personalized medicine approaches based on genetic and biomarker diagnostics.

FEATURES/BENEFITS

- Targets the underlying genetic cause of presynaptic congenital myasthenic syndrome (CMS).
- Restores choline acetyltransferase (ChAT) activity in peripheral neuromuscular junctions and potentially in the CNS.
- Delivers therapy non-invasively using recombinant AAV vectors.
- Extends treatment potential to multiple ChAT-related neuromuscular and neurological disorders.
- Improves motor function and survival in preclinical models.
- Reduces severe muscle weakness and respiratory failure associated with congenital myasthenic syndromes.
- Overcomes the limited effectiveness of existing symptomatic treatments for presynaptic CMS. Corrects neuromuscular junction dysfunction caused by ChAT deficiency.
- Addresses the lack of gene-targeted therapies for neuromuscular disorders and select CNS conditions.

CONTACT

Raj Gururajan
rgururajan@ucdavis.edu
tel: 530-754-7637.

INVENTORS

- Maselli, Ricardo

OTHER INFORMATION

KEYWORDS

acetylcholine, AAV vectors, central nervous system, choline acetyltransferase, congenital myasthenic syndrome, gene therapy, neuromuscular junction, presynaptic deficiency, recombinant viral vectors, therapeutics

CATEGORIZED AS

- **Biotechnology**
  - Health
- **Medical**
  - Disease: Central Nervous System
  - Disease: Genetic Diseases and

PATENT STATUS

Country	Type	Number	Dated	Case
Patent Cooperation Treaty	Reference for National Filings	WO 2024/197073	09/26/2024	2021-928

Patent Pending

Dysmorphic Syndromes

▶ Gene Therapy

▶ Therapeutics

▶ **Research Tools**

▶ Vectors

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University of California, Davis

Technology Transfer Office

1 Shields Avenue, Mrak Hall 4th Floor,

Davis,CA 95616

Tel:

530.754.8649

techtransfer@ucdavis.edu

https://research.ucdavis.edu/technology-transfer/

Fax:

530.754.7620