Exon-skipping Therapy for ADNP Syndrome

Tech ID: 34305 / UC Case 2025-585-0

ABSTRACT

Researchers at the University of California, Davis have developed novel antisense oligonucleotide (ASO) therapies that enhance ADNP protein expression to address haploinsufficiency in ADNP syndrome.

FULL DESCRIPTION

Activity-Dependent Neuroprotective Protein (ADNP) syndrome is a neurodevelopmental genetic disorder caused by loss-of-function mutations in one allele of the ADNP gene, leading to intellectual disability, developmental delays, autism spectrum features, and multi-systemic symptoms. Current treatments are lacking, prompting the development of antisense oligonucleotides that target upstream open reading frames (uORFs) in the ADNP mRNA 5’-UTR to increase translation efficiency from the healthy allele. By inhibiting translation of uORFs or excluding exon 2 from the mRNA, these ASOs increase ADNP protein levels, potentially improving symptoms. The compositions include chemically modified oligonucleotides designed for stability and efficacy, offering a novel therapeutic avenue for ADNP syndrome.

APPLICATIONS

- Treatment of ADNP syndrome and related neurodevelopmental disorders.
- Therapies targeting genetic haploinsufficiency in rare diseases.
- Precision medicine approaches for autism spectrum disorders with known genetic origins.
- Pharmaceutical development of antisense oligonucleotide drugs.
- Potential expansion to other genetic conditions involving uORF-mediated translational regulation.

FEATURES/BENEFITS

- Increases ADNP protein expression from the healthy allele.
- Targets uORFs to enhance translation efficiency.
- Potential to increase ADNP levels by up to 400%.
- Uses chemically modified oligonucleotides for improved stability and safety.
- Addresses underlying genetic cause rather than symptoms alone.
- Widely accepted molecular approach with potential for broad applicability.
- Overcomes the lack of approved treatments for ADNP syndrome.
- Resolves haploinsufficiency issues related to ADNP gene mutations.
- Potential to mitigate neurodevelopmental delays and intellectual disabilities associated with ADNP syndrome.
- Effect is not dependent on differing genetic mutations.
- Improves safety and compliance compared to previous therapies.

CONTACT

Prabakaran Soundararajan
psoundararajan@ucdavis.edu
tel: .

INVENTORS

- Caputo, Claire
- Fink, Kyle
- Guglielmi, Adele
- O'Geen, Henriette
- Salter-Cid, Tomas
- Segal, David J.

OTHER INFORMATION

KEYWORDS

ADNP syndrome, antisense oligonucleotide, autism spectrum disorder, gene therapy, genetic disease, haploinsufficiency, neurodevelopmental disorder, neuroprotective protein, rare disease treatment, uORFs

CATEGORIZED AS

- Biotechnology

PATENT STATUS

Patent Pending

- Genomics
- **Medical**
- Disease: Genetic Diseases and Dysmorphic Syndromes
- Gene Therapy
- Therapeutics
- **Research Tools**
- Nucleic Acids/DNA/RNA

RELATED CASES

2025-585-0

ADDITIONAL TECHNOLOGIES BY THESE INVENTORS

- Epigenetic Prevention and Treatment of CDKL5 Deficiency Disorder
- Multiplex Epigenetic Editing using a Split-dCas9 System

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