Evaluating AAV-mediated gene replacement for Spinal Muscular Atrophy with Respiratory Distress 1

https://neurodegenerationresearch.eu/survey/evaluating-aav-mediated-gene-replacement-for-spinal-muscular-atrophy-with-respiratory-distress-1/

Principal Investigators

LORSON, CHRISTIAN L

Institution

UNIVERSITY OF MISSOURI-COLUMBIA

Contact information of lead PI
Country

USA

Title of project or programme

Evaluating AAV-mediated gene replacement for Spinal Muscular Atrophy with Respiratory Distress 1

Source of funding information

NIH (NINDS)

Total sum awarded (Euro)

372545.8716

Start date of award

01/09/2015

Total duration of award in years

1

Keywords

Respiratory distress, gene replacement, Spinal Muscular Atrophy, Dependovirus, Distal Spinal Muscular Atrophy

Research Abstract

? DESCRIPTION (provided by applicant): Spinal Muscular Atrophy with Respiratory Distress 1 (SMARD1) or Distal Spinal Muscular Atrophy (DSMA1) is a fatal autosomal recessive genetic

disorder that is the second most common motor neuron disease in children. To date, no effective therapies or treatments exist for SMARD1. However, SMARD1 is an ideal candidate for vector-based gene therapy since it is monogenic and an animal model exists that reasonably recapitulates important features of disease. The gene responsible for SMARD1 is immunoglobulin μ-binding protein 2 (IGHMBP2). IGHMBP2 encodes a 993 amino acid protein that exhibits DNA/RNA helicase and ATPase activity, however, its normal and disease related functions are not well understood. While IGHMBP2 is ubiquitously expressed, motor neurons are a primary tissue in disease development, resulting in a pronounced wasting of skeletal muscle including the diaphragm. Recent advances in vector-based gene delivery in related areas such as Spinal Muscular Atrophy (or proximal SMA; 5q-linked SMA) have demonstrated that Adeno Associated Virus (AAV) vectors can efficiently enter a broad range of tissues within the central nervous system and the periphery, thereby effectively restoring expression of the disease gene. This proposal is designed to examine the utility of gene replacement as well as provide insight into disease development.

Further information available at:

Types:
Investments < €500k

Member States:
United States of America

Diseases:
N/A

Years:
2016

Database Categories:
N/A

Database Tags:
N/A