Targeting the Ubiquitin Proteasome System to Treat Spinal Muscular Atrophy

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Country

USA

Title of project or programme

Targeting the Ubiquitin Proteasome System to Treat Spinal Muscular Atrophy

Source of funding information

NIH (NINDS)

Total sum awarded (Euro)

€ 1,365,219.27

Start date of award

01/04/2016

Total duration of award in years

5

The project/programme is most relevant to:

Spinal muscular atrophy (SMA)

Keywords

SMN protein , Spinal Muscular Atrophy, multicatalytic endopeptidase complex, Ubiquitin, Motor Neurons

Research Abstract
? DESCRIPTION (provided by applicant): Spinal muscular atrophy (SMA) is the most common

inherited cause of death in infants and young children. SMA is caused by the deletion or mutation in the survival of motor neuron 1 (SMN1) gene, leading to a deficiency of the ubiquitously expressed SMN protein. Currently, there is no effective treatment option available for SMA. Evidence from studies in humans and rodents suggests that increasing SMN protein levels in the central nervous system is sufficient to ameliorate the disease phenotype and prolong survival. To identify protective modifiers of SMN protein levels we performed a genome-wide RNAi screen. Genes we identified in this screen will allow us to investigate genetic modifiers and molecular pathways that regulate SMN protein levels. These targets and pathways should provide novel avenues for therapeutic development for the treatment of SMA.

Lay Summary
PUBLIC HEALTH RELEVANCE: Spinal muscular atrophy (SMA) is a motor neuron disease and one of the leading inherited causes of infant mortality, affecting about 1 in 8000 live births. The disease is caused by deficiency of the survival motor neuron (SMN) protein and there is currently no effective treatment. The studies outlined in this proposal will identify genetic modifiers of SMN protein levels and define the molecular mechanisms regulating SMN protein stability in order to find new therapeutic targets for treating SMA.

Further information available at:

Types:
Investments > €500k

Member States:
United States of America

Diseases:
Spinal muscular atrophy (SMA)

Years:
2016

Database Categories:
N/A

Database Tags:
N/A