THERAPEUTIC USE OF CERAMIDASE ACID IN THE TREATMENT OF NIEMANN-PICK TYPE C DISEASE

A research group from CIBER and CSIC has revealed a new role for acid ceramidase (ACDase) in experimental models and in patients with Niemann Pick type C disease that reduces cholesterol in mitochondrial membranes and oxidative stress.

The Need

Lysosomal storage diseases (LSDs) are a group of inherited metabolic disorders that result from defects in lysosomal function. Niemann–Pick type C disease (NPC) is a subgroup of LSDs characterized by the accumulation of cholesterol and sphingolipids in the lysosomes of various tissues, mainly brain and liver, which causes neurological deterioration and liver disease. This accumulation is due to an induction of the STARD1 protein, responsible for the transport of cholesterol to the mitochondrial inner membrane.

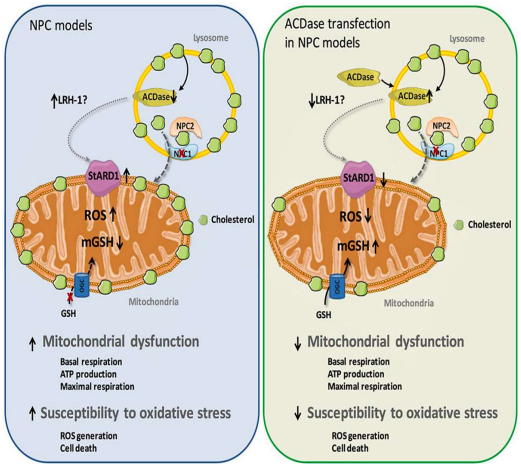
The Solution

The efficacy of most of the therapies available for patients with LSDs is limited, particularly because the treatments are usually initiated when organ damage has already occurred. Therefore, alternate targets for the treatment of NPC are required.

The present invention relates to the therapeutic use of acid ceramidase (ACDase), which inhibits STARD1 expression levels leading to the reduction of cholesterol in mitochondrial membranes and improving the performance and oxidative stress of fibroblasts of patients with NPC.

Innovative Aspects

- The therapeutic use of ACDase recovers mitocondrial function and reduces oxidative stress in samples of patients with NPC.
- Currently, targeted therapies in NPC disease focuses on preventing the accumulation of cholesterol in affected tissues. The proposed strategy is independent of the alteration of the cholesterol level and is aimed at preventing the consequences caused by said accumulation in the mitochondria.
- The action of ACDase maintains adequate mitochondrial function and reduces oxidative stress, characteristic of NPC disease.



Acid ceramidase improves mitochondrial function and oxidative stress in Niemann-Pick type C disease.

Stage of Development:

Demonstrated action in in vivo models and in patient samples.

Intellectual Property:

- Priority EP patent application filed (December 16, 2020)

Aims

Looking for a partner interested in a license and/or a collaboration agreement to develop and exploit this asset.

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