

PHARMACOLOGY THERAPY FOR TREATING MCARDLE DISEASE

INNOVATION AND DESCRIPTION OF THE TECHNOLOGY

The present invention refers to a novel therapy for McArdle disease. Patients with McArdle disease are unable to mobilize ATP from glycogen degradation when doing exercise, leading a crisis together with discomfort signs, pain, fatigue and even myoglobinuria and rhabdomyolysis. The disease has a broad clinical heterogeneity, but there are some clinical features that are common to most patients.

The present invention disclosure that compounds belinostat and mocetinostat significantly reduce glycogen accumulation in myogenic precursor cells (mpcs) of knock-in (KI) McArdle mice carrying the mutation p.R50X in homozygosis. Mpcs of KI McArdle mice carrying the mutation p.R50X in homozygosis are a good “in vitro” model of McArdle disease because they mimic the glycogen accumulation that is commonly found in the skeletal muscle from individuals with McArdle disease.

MARKET AND ADVANTAGES OF THE TECHNOLOGY

At present, there is no therapy against McArdle disease. The only approach is based on controlled physical training in order to develop mitochondrial oxidation capacities in muscles, and programmed glucose intake according to exercising periods and nutritional interventions have limited benefits in the functional performance of the patients. No pharmacological treatment has so far demonstrated substantial benefits .

The present invention is capable of increasing the amount of brain and liver isoforms of glycogen phosphorylase in the muscle cells, resulting in effective reduction of glycogen.

IPRS AND CONTACT

This technology was developed by CIBERER research groups : <https://www.ciberer.es/>

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