MUSCLE DISEASES

Use of small molecules to rescue folding defective proteins

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What's needed for?

The use of cystic fibrosis transmembrane conductance regulator (CFTR) molecules for the treatment of rare genetic muscular diseases are the object of this patent, specifically targeted for sarcoglycanopathies, catecholaminergic polymorphic ventricular tachycardia (CPVT) and Brody myopathy.

Sarcoglycanopathies, catecholaminergic polymorphic ventricular tachycardia (CPVT) and Brody myopathy are rare inherited striated muscle diseases that lead to severe disability and are potentially lethal. Currently no treatment is available. All three diseases are caused by the loss of function of a potentially active protein that is unable to retain its correct structure. To re-establish its function, this invention focuses on favouring the folding of the faulty protein by using molecules known as CFTR (Cystic fibrosis transmembrane conductance regulator) correctors, already selected for the treatment of cystic fibrosis. Although on its own each disease is rare, the three put together affect a significant number of the population, notwithstanding the incidence of Brody myopathy is likely largely underestimated.

Advantages

- Drug treatment for Sarcoglycanopathies
- Drug treatment for Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)
- Drug treatment for Brody Myopathy
- Potential treatment for other diseases with the same pathogenetic mechanism

Applications

Treatment of rare inherited muscular diseases, currently without treatment: Sarcoglycanopathies, Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) and Brody Myopathy .

TRL scale

Discovery | **Lead Optimization** | **Preclinical** | **Clinical Phases**

Health and biomedical - Pharmaceuticals