RARE MUSCLE DISEASES

Use of small molecules to rescue folding defective proteins

Applicant	Università degli Studi Padova
Inventors	Pompeo Volpe, Romeo Betto, Francesco Mascarello, Elisa Bianchini, Roberta Sacchetto, Dorianna Sandonà
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What it is needed for?

The use of cystic fibrosis transmembrane conductance regulator (CFTR) correctors for the treatment of rare and devastating genetic muscular diseases, namely sarcoglycanopathies and Brody myopathy, is the object of this invention.

Sarcoglycanopathies and Brody myopathy are rare inherited striated muscle diseases that lead to severe disability and are potentially lethal. Currently no available treatment exists. Both 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 correctors, already selected for the treatment of cystic fibrosis. Although on its own each disease is rare, the two put together affect a significant number of the population, notwithstanding the incidence of Brody myopathy is likely largely underestimated.

Advantages

- *A pharmacological intervention that targets the cause of the disease;*
- *Some of them are already in clinic for use in cystic fibrosis;*
- *The molecules act on the endogenous (though mutated) proteins promoting their folding and correct localization;*
- *Rare diseases.*

Applications

- *A pharmacological treatment for rare inherited muscular diseases, currently without treatment: Sarcoglycanopathies and Brody Myopathy; a treatment for other diseases linked to the same pathogenetic mechanism.*

What we are looking for

Technology is available for licensing and/or co-development.

TRL scale

Discovery | Lead Optimization | Preclinical | Clinical Phases

Health and biomedical - Pharmaceuticals

Patent@unismart.it