Small molecules for the treatment of APBD and other glycogen storage diseases (Hadasit)

code: 8-2015-299

Or Kakhlon, Hadassah Ein Kerem, Neurology
Miguel Weil, Tel Aviv University

Need:

Adult Polyglucosan Body Disease (APBD) and infant Glycogen Storage Disease type IV are rare neurodegenerative disorders caused by mutations in the gene encoding glycogen-branching enzyme and ensuing deficiency of the GBE protein. The current proof of concept is relevant for treating not only other types of Glycogen storage diseases but also diseases caused by destabilizing mutations in protein surface-exposed regions. APBD and other PB-based diseases as well as glycogen storage diseases in general are Orphan Diseases, some being ultra-rare, and most lack any available treatment. Beyond the value of OD designation affording significant advantages, many orphan drugs have significant markets in the range of $100Ms, even >$1B.

Innovation:

A cell-based assay was developed to identify small molecule (SMC) inhibitors of PB formation from an APBD patient. Eleven SMC hits show favorable dose-response profiles and are non-toxic in animals and improve the symptoms in animal, and hence can be useful as a drug to treat PB-based diseases.

Findings:

• In vitro proof of concept

• In vivo safety and efficacy tests.

Competitive advantage:

Novel Compounds that can be used for a cost-effective treatment of glycogen storage disorders using an ultra-rare disease for entry, in a market dominated by expensive Enzyme Replacement Therapies.

Contact for more information:

Tal Almog ✉, 054-3187538

Hadasit Medical Research Services & Development Ltd
Mother & Child Pavilion, Hadassah Ein Kerem, Jerusalem , 91120 Israel
Phone: +972-2-6778757, Fax: +972-2-6437712, E-mail: skimhi@hadassah.org.il