

Collaborations Pharmaceuticals, Inc. and collaborators announce publication on the efficacy of a treatment for CLN1

A new paper published in the Journal of Clinical Investigation describes the efficacy of an enzyme replacement therapy in multiple species for Batten disease.

RALEIGH, NORTH CAROLINA, USA, September 1, 2022 /EINPresswire.com/ -- [Collaborations Pharmaceuticals, Inc.](#) (CPI) and collaborators at Washington University in St Louis, The Roslin Institute, Amicus Therapeutics, Edinburgh University, NHS Lothian, and University of Texas Southwestern Medical Center are pleased to announce a [publication](#) led by the laboratory of Dr. Jonathan Cooper and colleagues in this prestigious journal (<https://www.jci.org/articles/view/163107>) that describes the development of an enzyme replacement therapy for CLN1 Batten disease and provides a foundation for future development.

CLN1 disease is a fatal neurodegenerative lysosomal storage disorder resulting from mutations in the CLN1 gene encoding the soluble lysosomal enzyme, palmitoyl-protein thioesterase-1 (PPT1). This neurodegenerative storage disorder primarily affects the brain and the retina of children and young adults, leading to dementia, blindness, epilepsy, and early death. There are currently no treatments available (other than palliative therapies) for this disease. Even gene therapy has proven less effective for this disease than for similar lysosomal storage diseases.

With substantial funding from NINDS we have now been able to demonstrate how monthly intracerebroventricular administration of human PPT1 produced statistically significant treatment effects in enzyme palmitoyl-protein thioesterase-1 (PPT1) deficient mice (Ppt1^{-/-}), such as rescue of PPT1 enzyme activity, decreased secondary enzyme levels, decreased the loss of



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neurons in all regions of brain and spinal cord and improved gait and rotarod results. Additional data is also presented that shows a similar efficacy in a sheep model of the disease.

“These results suggest the efficacy and feasibility of the repeated ICV delivery of the recombinant enzyme and are an important next step before clinical testing. We were recently funded by the NIH/NINDS with a nearly \$3M Phase II SBIR to manufacture the recombinant human PPT1 and conduct IND enabling toxicology studies. This work will set the stage for future clinical studies. CPI holds the Orphan Drug and rare disease designations from the Food and Drug Administration for this potential treatment which we are focused on commercializing. We welcome

discussions with other rare disease companies and potential partners that would be interested in learning more about this or other treatments in our rare disease [pipeline](#)” said CPI CEO Dr. Sean Ekins.



Collaborations Pharmaceuticals, Inc staff

The ERT was originally developed in the laboratory of Dr. Sandra Hofmann at the University of Texas Southwestern Medical Center and the team involved in this multi-year collaborative project included Hemanth R. Nelvagal, Samantha L. Eaton, Sophie H. Wang, Elizabeth M. Eultgen, Keigo Takahashi, Steven Q. Le, Rachel Nesbitt, Joshua T. Dearborn, Nicholas Siano, Ana C. Puhl, Patricia I. Dickson, Gerard Thompson, Fraser Murdoch, Paul M Brennan, Mark Gray, Stephen N Greenhalgh, Peter Tennant, Rachael Gregson, Eddie Clutton, James Nixon, Chris Proudfoot, Stefano Guido, Simon G. Lillico, C. Bruce A. Whitelaw, Jui-Yun Lu, Sandra L Hofmann, Sean Ekins, Mark S Sands, Thomas M Wishart, Jonathan D. Cooper.

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