

The Lost Enzyme Project

Mission Statement

Creating treatment to transform the lives of individuals and families affected by Beta-mannosidosis, an ultra-rare lysosomal disease.

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The road to a rare disease diagnosis is often long and grueling. Those diagnosed with Beta-mannosidosis arrive at a devastating prognosis; **Beta-mannosidosis is a progressive disease with no treatment.** Patients and their families have struggled to find hope for this under-researched and poorly understood disease. The Lost Enzyme Project was created by the families of those patients diagnosed in the United States, in collaboration with **Kimonis Lab** at the University of California, Irvine to complete the preclinical studies necessary for the development of an enzyme replacement therapy for Beta-mannosidosis.



What is Beta-Mannosidosis?

Beta-mannosidosis (β -mannosidosis) is an extremely rare genetic disease that belongs to a larger group of lysosomal storage disorders (NORD, 2023). Changes in the *MANBA* gene cause metabolic abnormality, specifically a loss of a functional β -Mannosidase enzyme, resulting in the build-up of toxic materials in the cells throughout the body and brain.

The severity and progression of the disease vary widely in individuals and symptoms can appear in infancy through adulthood. Most affected individuals experience some degree of intellectual disability and delayed motor development. Other signs and symptoms include speech delay, swallowing difficulties, hearing impairment, chronic infections, seizures, gross and fine motor delay, low muscle tone, scoliosis, optic nerve atrophy, nystagmus, white matter changes to the brain (leukoencephalopathy), painful peripheral neuropathy, and behavioral and psychiatric challenges.

How to Donate

Please make all checks payable to: **UCI Foundation**, write "**Lost Enzyme Project**" in the Memo Line and mail to:

UCI Health Advancement
Attn: Carley Fox
3800 West Chapman Ave. Ste #3300
Orange, CA 92868



To donate online, please visit bit.ly/lostenzyme or scan the QR code.

For More Information: If you have any questions concerning this project, please email them to Laurel Gregier at laurelgregier@gmail.com.

