

Research Study of Bone Marrow Transplant for Patients with Cystinosis from Matched Related Donor

A research study is being done by physicians at the Ronald Reagan UCLA Medical Center in Los Angeles to determine if blood stem cell transplants, with bone marrow from matched related donors, are an effective treatment that will slow down or prevent the progression of cystinosis. The study will also look into the risks associated with this treatment and if they are acceptable considering the potential benefit of avoiding the long-term consequences of cystinosis. *Bone marrow transplant has never been used to treat cystinosis in humans*, but recent studies in a mouse model of cystinosis show that this form of therapy, not only helps, but prevents disease progression in mice. In this experimental study, the most successful form of bone marrow transplant, HLA-matched related donor bone marrow transplantation, will be performed.

Eligibility criteria for participating in the study includes:

1. A) Adults, ages 18 and older with nephropathic cystinosis with stable kidney function (defined as less than 20% change in creatinine clearance from prior 12 weeks) and one or more of the following: a) muscle weakness; b) swallowing difficulties; c) progressive visual loss; d) intestinal malabsorption.

or

- B) Children ages 13-17 years who do not tolerate or do not take cysteamine (defined by leukocyte cystine levels greater than 5 nmol half-cystine/mg protein for 2 consecutive time points at least 3 months apart during the prior 6 months or parental confirmation of patient intolerance) and worsening clinical manifestations as determined by a physician who is not an investigator on this study.
2. Patients must have a related bone marrow donor who is HLA-matched on 10 of 10 alleles.
3. Patients with adequate physical function as measured by:
Pre-transplant tests of heart, lungs, kidneys, liver, and other organs and must not have a serious infection, be pregnant, or have undergone a prior stem cell transplant.

Interested subjects should ask their physician to contact Zoe Solsby at the Cystinosis Research Foundation for additional information.

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