Dr. Dietrich Matern and his colleagues at the Mayo Clinic Biochemical Genetics Laboratory in Rochester, MN, are conducting an IRB-approved research study to develop a testing strategy for the most effective and efficient newborn screening for lysosomal storage disorders (LSD). An effective newborn screening program would allow for the diagnosis and treatment of affected individuals prior to the onset of symptoms and development of irreversible damage. In addition, we will try to identify biomarkers that could indicate the most appropriate time to initiate treatment.

We are currently seeking volunteers who have a confirmed diagnosis of any one of the LSDs and/or first degree relatives of these individuals. In addition, families who have a child who screened positive for an LSD through newborn screening are also eligible to participate. We welcome samples from patients and their parents, siblings, and children.

Participation will include the following:

1) Allowing Mayo Clinic access to any leftover newborn screening samples that may still be available in the newborn screening laboratory of the state where an LSD patient or family member of an LSD patient was born.

2) At least annual collection by a finger stick or regular blood draw of a few drops of blood which will be used to measure LSD enzyme levels and to identify relevant biomarkers.

If you would like to participate, please contact Dr. Matern or a biochemical genetic counselor at 507-266-8158 or by e-mail at biochemicalgenetics@mayo.edu for assistance in requesting a new blood sample and/or a leftover newborn screening sample from the respective newborn screening laboratory. Please understand that your current or future medical care at the Mayo Clinic will not be jeopardized if you choose not to participate.

If you have any questions or concerns, we would be happy to discuss the study with you in more detail. Please call with any inquiries or to assist with initiating participation in the study. Thank you in advance for considering this request.