LYSOSOMAL STORAGE DISORDER

What Is Lysosomal Storage Disorder?

Lysosomal Storage Disorders, or simply LSD, are inherited metabolic disorders, where an enzyme deficiency leads to a buildup of toxins within cells. There are over 50 specific types, and affect differing cells within the body. The treatment varies based on the cells that are affected.

How Common Is LSD?

• It is estimated, that as a group, LSDs affect 1 in every 5,000 births

What Can We Do For Our Child?

• At birth or soon after birth, signs and symptoms will become apparent. The timeline is dependent on the type of cell affected and the length of time it takes for toxin buildup to cause symptoms. It is therefore important to identify if your child may be affected with newborn screenings or prenatal screenings.
• Some common LSDs are Cystinosis, Batten Disease, Fabry Disease and Gaucher Disease.

How Is LSD Treated?

• There are no cures for LSDs, but with prenatal screening available for all the known LSDs, treatments can be started at birth or very shortly after birth. Earliest treatments will minimize or prevent the buildup of toxins within the cells and can often improve outcomes and life expectancy.
• Bone marrow transplants and either plasma derived or synthetic enzyme replacement can minimize the storage and buildup of toxins, depending on the specific LSD the patient has

Support Group Information

You can find more information on LSD at these websites

• National Organization for Rare Disorders at https://rarediseases.org/rare-diseases/lysosomal-storage-disorders/
• Lysosomal Disease Network at http://www.lysosomaldiseasenetwork.org/

There are also many specific patient support websites for individual Lysosomal Storage Disorders on the web.