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Proceeding

Advances in Cardiology and Cardiovascular Disorders

Cardiac manifestations in lysosomal storage disorders

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Received: November 10, 2017 | **Published:** November 14, 2017

Lysosomal storage diseases (LSDs) are a heterogeneous group of rare inherited disorders characterized by the accumulation of undigested or partially digested macromolecules that ultimately results in cellular dysfunction and clinical abnormalities. The LSDs occur as a result of specific genetic mutations that result in deficiencies of lysosomal hydrolases. More recently, the concept of LSDs has been expanded to include deficiencies or defects in proteins necessary for the normal post-translational modification of lysosomal enzymes, activator proteins, protein defects in intracellular trafficking etc.

More than 50 lysosomal storage diseases have been described. Depending on the specific enzyme deficiency, LSDs may affect different parts of the body, including the liver, spleen, skeleton, brain, skin, heart, and central nervous system. Age of onset and clinical manifestations may vary widely among patients, and significant phenotypic heterogeneity is seen. The constellation of dysmorphic features (coarse facies, macroglossia), bony abnormalities (dysostosis multiplex), cardiac involvement (arrhythmia or cardiomegaly), hepatosplenomegaly, ophthalmologic signs (corneal clouding or macular cherry-red spot), and neurological features may lead to clinical suspicion of a lysosomal storage disease. Symptoms are typically gradually progressive. With the advent of ERT therapies for various LSDs there is more emphasis on understanding the multi-systemic nature of these disorders. There is significant cardiac involvement in several LSDS involving the endocardium, myocardium, conduction system and blood vessels.

In this presentation the varying cardiac manifestations of different lysosomal storage diseases will be presented. The challenges and successes with treatment interventions and new emerging natural history on treatment will also be described for the various LSDs.