

Cardiac Involvement in Lysosomal Diseases

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Lysosomal storage disorders (LSD) include more than 40 diseases caused by a deficiency of lysosomal enzymes, membrane transporters or proteins involved in lysosomal biology.

Clinical organ involvement usually occurs in the presence of substrate excess. Different diseases may affect various parts of the body, including the skeleton, brain, skin, heart, liver, kidney and central nervous system.

Cardiac disease is particularly important in lysosomal glycogen storage diseases (Pompe and Danon disease), mucopolysaccharidoses and in glycosphingolipidoses (Fabry disease).

Various disease manifestations may be observed including hypertrophic and dilated cardiomyopathy, coronary artery disease and valvular disease.

Keywords: Heart; Lysosomal disease; Treatment

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