Special Issue

Diagnosis and Management of Lysosomal Storage Diseases in Specific Body Organs and Systems

Message from the Guest Editors

Lysosomal storage diseases (LSDs) are a group of monogenic metabolic disorders associated with the dysfunction of lysosomal apparatus. Virtually every cell in the body possesses lysosomes, yet storage in these organelles can vary even among various cells. The clinical phenotype is quite heterogeneous, affecting multiple organs and systems, reflecting also the complexity of their pathomechanism. Even though our knowledge of LSD has increased, and many therapies have been established, many aspects of their presentation remain ambiguous. The aim of this Special Issue is to provide a comprehensive overview of clinical presentation, diagnostics and treatment, focusing on specific body organs and systems. Potential topics include, but are not limited to, the following:

- Liver in LSD;
- Cardiac involvement in LSD;
- Skeletal complications in LSD;
- Respiratory system involvement in LSD;
- Central and peripheral nervous systems abnormalities in LSD;
- Biomarkers in LSD diagnostics;
- Genetics, inheritance, founder effects and genotype-phenotype correlation.

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