

Special Issue

Diagnostic and Therapeutic Developments in Lysosomal Storage Disorders

Message from the Guest Editors

LSDs are a heterogeneous group of more than 70 inherited metabolic conditions caused by defects in genes that encode proteins required for lysosomal homeostasis. Clinical manifestations can be observed from the prenatal period to adulthood; The spectrum of clinical symptoms usually does not have a clear phenotype–genotype correlation, and intrafamilial variability is frequently described. Due to the multisystemic nature, non-metabolic clinicians may first suspect an LSD diagnosis.

Newborn screening and massive parallel sequencing have facilitated earlier diagnoses, as well as the identification of attenuated forms and pre-symptomatic individuals. Therapeutic advances such as enzyme replacement therapy have improved healthcare outcomes and life expectancy over the last few decades. However, new challenges arise in the management of LSDs: transition of pediatric to adult care, coordination of multidisciplinary teams, reproductive planning, and newly recognized long-term complications.

This Special Issue aims to attract original research articles, reviews, and short communications on understanding recent advances in the natural history, diagnosis, and management of LSDs.

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There has been an explosion of gene and target based research and therapeutics in the multitude of fields that compose clinical medicine. The *Journal of Clinical Medicine's* (JCM) staff and editorial board are dedicated to providing cutting edge, timely, and peer-reviewed articles covering the diverse subspecialties of clinical medicine. The journal publishes concise, innovative, and exciting research articles as well as clinically significant articles and reviews that are pertinent to the myriad of disciplines within medicine. The articles published are relevant to both primary care physicians and specialists. The journal's full-texts are archived in PubMed Central and indexed in PubMed. Please consider submitting your manuscripts for publication to our journal and check us out on-line!

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