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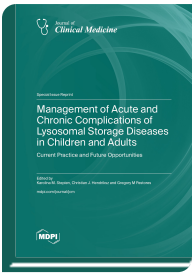
Special Issue Reprint

Management of Acute and Chronic Complications of Lysosomal Storage Diseases in Children and Adults: Current Practice and Future Opportunities



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This reprint, entitled “Management of Acute and Chronic Complications of Lysosomal Storage Diseases in Children and Adults: Current Practice and Future Opportunities”, covers broad aspects of this group of rare diseases. The first theme includes articles on the management of pain, corneal clouding, gastrointestinal and airway diseases in Mucopolysaccharidoses and Mucolipidoses, and the evaluation of dysphonia in Pompe disease. Additionally, attention deficit and ADHD and the management of atherosclerosis in Fabry disease are considered here. The second theme includes diagnostic conundrums in lysosomal storage diseases, e.g., the prenatal diagnosis of Gaucher disease during pregnancy, the diagnosis of secondary hyperparathyroidism in Mucolipidoses, and the application of the new biomarker “plasma neurofilament light” in Niemann Pick C disease, as well as the correlation of enzyme activity with molecular diagnosis in Pompe disease. The articles in the third theme describe the impact of therapies on clinical outcomes. For example, a systematic review of the impact of enzyme replacement therapy on late-onset Pompe disease and trehalose use on Niemann Pick A and B is included. The remaining articles describe the NHS processes for overcoming challenges resulting from the COVID-19 pandemic and the differences in the transition from paediatric to adult services between different countries.

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