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Newborn Screening for Pompe Disease

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Message from the Guest Editors

Newborn screening is important for the early diagnosis and treatment of Pompe disease. The initial successes of Pompe disease newborn screening were from Taiwan, and currently, a good number of newborn screening programs have already started or are in the planning stages. However, screening for Pompe disease is not without its difficulties. There are a few assays available for screening. The accuracy of the assay as well as the screening algorithm determine the false positive and negative rates. Confirming diagnosis can be difficult, and genotype–phenotype correlation may not be established. Decision about when to initiate treatment, especially for late-onset Pompe disease, is difficult. To add to this, all these problems are further diverse among different ethnic groups. Through this Special Issue, experts in the field of newborn screening can share experiences in Pompe disease and help to accumulate data concerning prevalence, genotype, and phenotypes. More general readers of the Journal can also have the chance to understand Pompe disease and its screening. We think this Special Issue is just in time, and we thank all contributing authors in advance.