Preface

MMS Special Issue “Newborn Screening and Mass Spectrometry”

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Recently, mass spectrometry has seen expanding applications in the field of clinical medicine. Among these, GC/MS and MS/MS (tandem mass spectrometry) have been widely used to the diagnosis of inherited metabolic disorders (IMDs), and the newborn screening using MS/MS (TMS screening) is now spreading globally.

Currently the TMS screening employs flow injection analysis, which directly introduces samples extracted from newborn blood filter paper absorbed with minute amount of blood, into MS/MS. It enables simultaneous analysis of amino acids and acylcarnitines, allowing for simple and rapid screening for amino acidemias, organic acidemias, and fatty acid oxidation defects.

This special issue featured six papers, including one in “Short communication”, and five in “Research papers”, respectively. In the “Short communication”, a case report of hereditary spherocytosis found in the TMS screening was presented. In the “Research papers”, 1) development of a highly sensitive analytical method for succinylacetone which is a diagnostic marker for hereditary hypertyrosinemia type 1; 2) experience and significance of prenatal diagnosis combining mass spectrometry and genetic testing for the severest types of IMD cases detected in the NBS; 3) an attempt of screening for urea cycle disorders through the development of orotic acid detection method, 4) a summary of disease distribution and frequency in cases detected via GC/MS in clinically high-risk patients in India; and 5) development of a rapid, highly sensitive MPS screening method combining RapidFire and MS/MS, were presented.

We believe the papers featured in this special issue contribute to expanding the application scope of MS/MS in NBS and improving diagnostic accuracy.

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