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The Future Perspective: Metabolomics in Laboratory Medicine for Inborn Errors of Metabolism

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Abbreviations: GCMS=gas chromatography mass spectrometry, LC-MS=liquid chromatography mass spectrometry, CSF= Cerebrospinal fluid, IEM=inborn errors of metabolism, MALDI=matrix assisted laser desorption ionization, SELDI= Surface-enhanced laser desorption/ionization, NMR=nuclear magnetic resonance, NBS=newborn screening, PKU=phenylketonuria, LCHAD= Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency, TFP=Trifunctional protein deficiency, MMA=methylmalonic acidemia, PA= propionic acidemia, OTC= Ornithine transcarbamylase deficiency, RDC=respiratory chain deficiencies, SLOS=Smith-Lemli Opitz syndrome, CTX= Cerebrotendinous xanthomatosis, RP=Reverse phase, HILIC= hydrophilic interaction liquid chromatography, ROC= receiver-operating characteristic, AUC=Area under the curve

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