

## **Discovery of diagnostic markers for inborn errors of metabolism using LC/ESI-MS/MS of urinary conjugated cholesterol metabolites**

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In various inborn errors of metabolism and hepatobiliary diseases related to the some cholesterol metabolite pathways, the metabolite profiles in blood, urine, and cells differ from healthy conditions. Accordingly, analysis of cholesterol metabolite profile in various diseases could find the disease specific molecules. Comprehensive analysis method of urinary conjugated cholesterol metabolites using liquid chromatography/electrospray ionization tandem mass spectrometry (LC/ESI-MS/MS) was developed and the method was applied for biomarker search. Focused metabolomics method was developed with precursor ion scan and neutral loss scan based on MS/MS patterns of each conjugated forms. Chromatographic separation of all isomers was achieved with ODS column and various conjugated metabolites were selectively detected using characteristic MS/MS conditions every conjugated forms. The method was applied for the urine of patients with Niemann-Pick disease type C and the characteristic conjugated cholesterol metabolites were found. An LC/ESI-MS/MS method for the characteristic conjugated metabolites was developed and the method was applied for the urine samples. The metabolites were useful for NPC chemical diagnosis. In future, the method is hoped to apply the other marker search and the drug discovery study.