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大會議程及論文摘要
Programs & Abstracts

中華民國毒物學學會 Toxicology Society of Taiwan
中國生理學會 The Chinese Physiological Society
中華藥理學會 The Pharmacological Society in Taiwan
中華民國解剖學學會 The Association of Anatomists of the Republic of China
中國生物化學及分子生物學會 The Chinese Society for Biochemistry and Molecular Biology, Taipei
中華民國細胞及分子生物學學會 The Chinese Society of Cell and Molecular Biology
中華民國臨床生化學會 Chinese Association for Clinical Biochemistry

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Application of Electrospray Tandem Mass Spectrometry in Screening Inherited Disorders of Amino Acid Metabolism

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A neonatal screening method, with few false results and less potential for interference from drugs or their metabolites, of inherited disorders of amino acid metabolism using electrospray tandem mass spectrometry was developed in our study. Five amino acids, phenylalanine (m/z 222), tyrosine (m/z 238), valine (m/z 171), methionine (m/z 206), leucine (m/z 188) extracted from a dry blood spot (Guthrie card), were quantitated in neutral loss (m/z 102) scanning mode after conversion of amino acids to their butyl esters through treatment with butanolic hydrogen chloride. Each scan takes only 2 minutes.

The recoveries (93~98%) and precisions (c.v. 7.2~10.9%) in amino acids were obtained. The linearity (r^2) for phenylalanine and tyrosine was 0.997 and 0.981 respectively. The concentrations of phenylalanine (18.3~519.9 μmol/L), tyrosine (49.0~674.0 μmol/L) and valine (38.5~405.0 μmol/L) for normal neonates were determined. This simple and accurate method is suitable for the routine neonatal screening of PKU and other inherited disorders of amino acid metabolism.

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Identification of Mutations Causing 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency in Chinese

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Deficiency in 6-pyruvoyl-tetrahydropterin synthase (PTPS) activity is a major case of the tetrahydrobiopterin (BH_4) deficient hyperphenylalaninemia (PHA). In this study, four single base mutations at nucleotide IVS3+1(G>C), 200 (C>T), 226 (C>T), 430 (G>C) in the PTPS gene were detected in Chinese PTPS deficient PHA by polymerase chain reaction and solid phase sequencing. These 200C>T, 226C>T, 430G>C alterations resulted in Thr67Met, Leu70Phe, Gly144Arg amino acid substitutions, respectively. The single base transversion of G to C at IVS3+1 might render splicing error. The IVS3+1, 226C>T, 430G>C were novel mutations. However, the 200C>T mutation has been identified in four Italian families. None of 100 normal alleles were found to have 226C>T or 430G>C substitutions. This data indicated these substitutions were not polymorphisms in Chinese population but disease causing mutations.