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PROGRAM BOOK

台大醫院 NTUH

生技醫藥國家型科技計畫 National Research Program for Biopharmaceuticals

Poster Session

Date/Time: March 20

Venue: Room 810B, EICC

PH-02 Neurofibromatosis Type 1 Gene Mutations in Taiwan: Three Cases Report
Yi-Jing Su (Taiwan), Peng-Jun Chen, Kun-Long Hung, Sing-Chung Li

PH-03 High Genetic Heterogeneity In Indian Patients with Late Infantile Metachromatic Leukodystrophy: Report of 27 Cases
Pallavi Shukla, Shahzan Anjum, Pallavi Mishra (India), Vikram Singh,Ranjana Srivastava,Shivaram Shastri, Neerja Gupta, Sheffali Gulati, Madhulika Kabra

PI Organic Acid Disorders

PI-01 Three Patients with HSD10 Disease in Japan
Toshiyuki Fukao (Japan), Hideo Sasai, Yuka Aoyama, Kazuhisa Akiba, Masahiro Goto, Yukihiro Hasegawa, Masahisa Kobayashi, Hiroyuki Ida, Shohei Akagawa, Tomohiro Hori, Yuki Hasegawa, Seiji Yamaguchi, Yosuke Shigematsu

PI-02 Neonatal Isovaleric Acidemia Presenting as Encephalopathy Infant First Case Report from Soetomo Hospital Surabaya-Indonesia
Nur Aisiyah Widjaja (Indonesia), Martono Tri

PI-03 The Advantage of Cultured Lymphocytes in Activity Assays for Propionyl-CoA Carboxylase and Methylmalonyl-CoA Mutase
Yen-Hui Chiu (Taiwan), Mei-Ying Liu, Yu-Ning Liu, Kwang-Jen Hsiao, Tze-Tze Liu

PI-04 The First Neonatal Case of HDS10 Disease in Japan
Masahisa Kobayashi (Japan), Toshiyuki Fukao, Toya Ohashi, Hiroyuki Ida

PJ Peroxisomal Disorders

PJ-01 A Child Case of Addison Disease Only Form Adrenoleukodystrophy with Novel *ABCD1* Gene Mutation
Sang heun Lee, Ji Eun Lee (North Korea)

PK Purines and Pyrimidine Disorders

PK-01 Clinical, Biochemical and Molecular Analysis of 30 Children with β-Ureidopropionase Deficiency Demonstrates High Prevalence of the C.977G>A (P.R326Q) Mutation
Yoko Nakajima (Japan), Judith Meijer, Doreen Dobritzsch, Chunhua Zhang, Tetsuya Ito, Yoriko Watanabe, Tomiko Kuhara, André B.P. van Kuilenburg

PL Screening for Inborn Errors of Metabolism

PL-01 Second-tier Tests in Newborn Screening by Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
Yi-Lin Liu (Taiwan), Hui-chen Liu, Hsin-yun Liu, Hsuan Chieh Liao, Shu-Min Kao, Chuan-Chi Chiang

PL-02 High Risk Group Screening for Porphyrias in Taiwan
Hsuan-Chieh Liao (Taiwan), Ya-Ling Fan, Ying-Chen Chang, Shu-Min Kao, Yann-Jang Chen, Chuan-Chi Chiang

PL-03 External Quality Assurance Program for Neonatal Screening of Glucose-6-Phosphate Dehydrogenase Deficiency
Mei-Ling Fan (Taiwan), Szu-Hui Chiang, Charity M. Jomento, Carmencita D. Padilla, Kwang-Jen Hsiao

The Advantage of Cultured Lymphocytes in Activity Assays for Propionyl-CoA Carboxylase and Methylmalonyl-CoA Mutase

Yen-Hui Chiu[1], *Mei-Ying Liu*[2], *Yu-Ning Liu*[1], *Kwang-Jen Hsiao*[1,3], *Tze-Tze Liu*[1,2]

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Propionic acidemia (PA) and methylmalonic acidemia (MMA) are two of the most common life-threaten organic acidemias in most areas of the world. Patients might suffer from severe complication including developmental delay, mental retardation, seizures, and, in some instances, early death. Proper treatment in early stage is crucial to prevent the irreversible physical damages. The level of methylmalonic acid and 3-hydroxypropionate in dried blood spots measured by LC-MS/MS has been developed as the second-tier test for MMA and PA, respectively. The enzyme activity assays for propionyl-CoA carboxylase (PCC) and methylmalonyl-CoA mutase (MCM) allow conformation and differential diagnosis of these two diseases. However, without proper reservation, leukocytes in the anticoagulated blood stored at room temperature lost at least 50% of enzyme activity in 3 days which might lead to false positive results. We therefore developed an enzymatic assay for PCC and MCM enzymes using phytohemagglutinin (PHA) stimulated cultured lymphocytes. With a sufficient number of cells, five-day cultured lymphocytes were used in this study. Four PA, four mut-type MMA families and 20 self-reported normal individuals were included. There were no significant differences of enzyme activity between obligated carriers and normal individuals while that in patients was markedly reduced. Furthermore, enzyme activity was unaffected when anticoagulated blood was stored at room temperature for four days followed by lymphocyte culture. Here, we have established a practical protocol to perform the diagnosis of PA and MMA in blood samples transported from other medical centres.

Keywords: *Enzyme activity assay, Cultured lymphocyte, Propionyl-CoA carboxylase, Methylmalonyl-CoA mutase*

Home