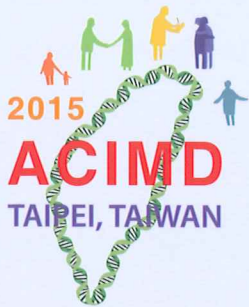


Taipei
Taiwan

Diagnosis

Prevention

Treatment



The 4th Asian Congress for Inherited Metabolic Diseases

March 19-22, 2015

Evergreen International Convention Center, Taipei, Taiwan

PROGRAM BOOK



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, Yukihiko 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 Aisyah 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 HSD10 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 Porphyrrias 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 Chiu¹, Mei-Ying Liu², Yu-Ning Liu¹, 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-threatening 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*