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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

External Quality Assurance Program for Neonatal Screening of Glucose-6-Phosphate Dehydrogenase Deficiency

Mei-Ling Fan[1], *Szu-Hui Chiang*[1], *Charity M. Jomento*[3], *Carmencita D. Padilla*[4], *Kwang-Jen Hsiao*[1,2]

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Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common human enzymopathic disease. The national neonatal screening program for G6PD deficiency was started in 1987 and 1996 in Taiwan and Philippines, respectively. To assess the reliability and assure the quality of the screening and confirmatory tests, external quality assurance (EQA) programs for G6PD screening and confirmatory tests were developed. For screening test, the QC materials were prepared from whole blood by spotted on Guthrie cards. For confirmatory test, lyophilized QC materials were prepared from human red blood cells. Bimonthly, 10 QC blood spots and 3 lyophilized QC samples were sent to screening and referral laboratories, respectively. The test results were submitted online and the summary reports were published on the website (g6pd.qap.tw) within 14 days after the survey started.

Forty-three screening laboratories from 16 countries and 38 referral laboratories in Taiwan and Phillippines are participating in the EQA program at the present time. From 1999 to 2014, 97 EQA surveys for screening test were performed, 174 (10.4%) unsatisfactory EQA reports were found from 1469 reports recieved. The unsatisfactory results were mainly caused by inappropiate cut-offs. From 1988 to 2014, 188 EQA surveys were sent to referral laboratories in Taiwan, 306 (8.8%) unsatisfactory reports were found from 3,459 reports received. Inter-laboratory C.V. for the quantitative test has reached < 10% in recent years. The long term (7 years) intra-laboratory presicion (C.V.) of the referral laboratories in Taiwan has reached 6.1% (0~25.0%). Since July 2009, 19 EQA surveys have been carried out for the newly established network of referral laboratories in Philippines. From 2009 to 2014, 69 (19.8%) unsatisfactory EQA reports were found from 349 reports. Inter-laboratory C.V. in Philippines were between 6.6% and 25.0%, which is lower than those found in other EQA programs (*e.g.* CAP, RCPA) for G6PD quantitative test. These G6PD EQA programs have been useful for monitoring and to improve the G6PD tests quality, and might be a reference for the participating laboratories to adjust the cut-offs for the screening test.

Keywords: *Glucose-6-phosphate dehydrogenase deficiency, G6PD, external quality assurance program, EQA*

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