



Ninth Asian-European Workshop of Inborn Errors of Metabolism is organized by Slovak Medical Association under the auspices of Minister of Health of Slovakia, Dr. Rudolf Zajac.

Foreword

After the publication of the major presentations of the first Asian-European Workshop of Inborn Errors of Metabolism (AEWIEM) in the 1994 edition of Journal of Inherited Metabolic Disease, we have put together a proceeding for those of the 2nd and 3rd meetings containing 34 articles. Now after a long interval, we are indeed very pleased to be able to publish articles and abstracts of the 9th meeting in Slovakian Medical Journal.

Since the foundation of the Workshop 12 years ago, we have tried to keep our promise by organizing meetings in different parts of the world, establishing coherent relationships between scientists and clinicians of Asia and Europe as well as arranging collaboration studies. Still we know that this is just a start and there are many works ahead of us. In that sense, publishing this proceedings is a small step toward our goal of achieving a better health to all children of the world.

In this occasion, I would like to express my sincere thanks to the presidents and the organizers of the 9 meetings, who did excellent jobs to hold such friendly and fruitful workshops.

Yoon S. Shin

Welcome address

Dear Friends and Colleagues,

We are looking forward to welcoming you to the 9th Asian-European Workshop of Inborn Errors of Metabolism (AEWIEM) in Slovakia. The meeting will be held in Stará Lesná, on the southern side of the High Tatras mountains, which are the most beautiful part of the country. It was declared by the UNESCO to be the "Biospheric reservation".

The programme covers many interesting topics and hopefully will contribute to an enhancement of patient care. The meeting timetable will also provide ample opportunity to meet and talk with your colleagues in a relaxed atmosphere during an attractive social and excursion programme.

I hope you will find the 9th AEWIEM to be very useful and that you will enjoy visiting the most famous and exciting places of our country.

The meeting is made possible by generous sponsors for which we express our sincere gratitude.

Vladimír Bzdúch

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Scientific Programme of the 9th AEWIEM

Stará Lesná, Slovakia

26th September – 1st October 2004

Monday, 27th September 2004

8.30 h – WELCOME ADDRESS

Shin YS and Bzdúch V

I. PHENYLKETONURIA

Chairpersons: Hyánek J and Lee DH

9.00 h

Hyánek J, Kozák L, Hrabincová E, Doležal A, Soukup K, Zeman J, Šťastná S, Vaňurová L, Krijt J, Viletová H, Červená M, Paterová T (Czech Republic)

Maternal hyperphenylalaninemia in healthy Czech population of pregnant women: 30 years experience with screening prevention and treatment

9.30 h

Lee DH, Kim YS (Korea)

Tetrahydrobiopterine (BH4) responsive phenylketonuria

9.50 h

Potočnáková E, Šaligová J, Vasilíková M, Szaboová A (Slovakia)

Phenylketonuria and adulthood – what is the actual situation in the East Slovakia region?

10.05 h

Šaligová J, Potočnáková E, Vasilíková M, Bratský L, Figulová E, Szabóová A (Slovakia)

Clinical outcome of PKU patients – correlation to the type of HPA metabolic compensation and age

10.20 h – Coffee break

II. OTHER AMINOACIDOPATHIES

Chairpersons: Saheki T and Behúlová D

10.50 h

Behúlová D, Bzdúch V, Fabriciová K, Hlavatá A, Kolníková M, Škodová J, Šaligová A, Ponec J, Zeman J, Gregová E, Dvořáková L, (Slovakia, Czech Republic)

Urea cycle disorders – pitfalls in diagnosis

11.05 h

Saheki T, Kobayashi K, Begumi L, Iijimal M, Jalill A, Moriyama M (Japan)

Accumulation of cytosolic NADH as the prime cause of various symptoms in deficiency of citrin a liver-type mitochondrial aspartate glutamate carrier

11.25 h

Kobayashi K, Lu YB, Ushikai M, Li MX, Hsiao KJ, Yang Y, Lee DH, Saheki T (Japan, Taiwan, China, Korea)

Population analysis in East Asia of twelve SLC25A13 mutations found in Japanese patients with citrin deficiency (CTLN2 and NICCD)

11.45 h

Škodová J, Holešová I, Fabriciová K, Bzdúch V, Petrovič R, Ponec J, Behúlová D (Slovakia)

Plasma levels of total homocysteine and methylenetetrahydrofolate reductase C677T polymorphism

12.00 h

Vajnerová Z, Behúlová D, Holešová D, Škodová J, Holešová J, Ponec J, Bzdúch V, Fabriciová K, Gregová E, Hálová K (Slovakia)

Nonketotic hyperglycinaemia in Slovakia

12.15 h

Han-Wook Yoo, Sung-Soo Kim, Sang-Wook Park, Gu-Whan Kim (Korea)

Genotype – phenotype correlation in Korean patients with ornithine transcarbamylase deficiency

12.35 h

Bio-Rad – Presentation

12.50 h – Lunch

III. GALACTOSEMIAS AND GLYKOGENOSES

Chairpersons: Shin YS and Fateen E

14.30 h

Fateen E, El Shafei S, El Karakasy H, Mahmoud M, Roshdy S, El Temtamy S, Shin Y (Egypt, Germany)

Diagnosis and management of galactosemia: An Egyptian experience

14.55 h

Ak Basol G, Tuncel T, Yaprak I, Tanyalcin T (Turkey)

Quantitative analysis and reference value determination of galactose-1-phosphate uridyl-transferase activity in healthy babies

15.15 h

Tanyalcin T, Reclos G (Turkey, Greece)

The effect of blood spot card variability on the detection limit analysis and the linearity of galactose newborn screening kit

15.35 h

Lee HK, Podskarbi T, Shin YS (Germany)

Clinical biochemical and molecular heterogeneity of branching enzyme deficiency (Anderson disease)

15.55 h

Perkin-Elmer – Presentation

16.10 h – Coffee break

16.40 h

I. Round table – Galactosemia (Shin YS)

Tuesday 28th September 2004

IV. LYSOSOMAL STORAGE DISEASES

Chairpersons: Elleder M and Podskarbi T

9.00 h

Linhart A, Karetová D, Paleček T, Bultas J (Czech Republic)

The heart and Fabry disease: current knowledge

9.35 h

Yoon HR, Lee K, Cho K, Kang S, Lee DH, Yoo HW (Korea)

Quantification of globotriaosylceramide in human plasma and urine by electrospray ionization tandem mass spectrometry for therapeutic monitoring of Fabry disease

9.55 h

Podskarbi T (Germany)

Genotype and phenotype correlation of Pompe disease

10.20 h – Coffee break

10.50 h

Elleder M, Pavlů-Pereira H, Asfaw B, Poupětová H, Ledvinová J, Sikora J, Vanier MT, Zeman J, Novotná Z, Chudoba D (Czech Republic, France)

Acid sphingomyelinase deficiency: molecular studies of a series of 25 Czech and Slovak patients with prevalence of intermediate phenotype

11.15 h

Dietze M, Shin YS (Germany)

Biochemical diagnosis of Niemann-Pick disease A,B and C: The important role of sphingomyelinase

11.35 h

Fisher L, Shin YS (Germany)

Four enzymes involved in mucopolysaccharidosis III (Sanfilippo syndrome): Expression in various tissues and the diagnostic possibility using leucocytes

11.55 h

Aboul Nasr A, Fateen E (Egypt)

Prenatal diagnosis of mucopolysaccharidoses (MPS): The first Egyptian experience

12.15 h – Lunch

V. DISORDERS OF FATTY ACID BETA-OXIDATION AND ORGANIC ACIDEMIAS

Chairpersons: Gregersen N and Lehnert W

14.00 h

Lehnert W, Matern D (Germany, USA)

Mitochondrial fatty acid oxidation deficiencies: an overview

14.45 h

Gregersen N (Denmark)

Genotyping patients with fatty acid oxidation defects: Why When and How

15.15 h

Šalingová A, Bzdúch V, Fabriciová K, Kolníková M, Ponec J, Behúlová D (Slovakia)

Free L-carnitine in diagnosis and monitoring of inherited metabolic diseases

15.30 h – Coffee break

16.00 h

Gündüz M, Ezgü FS, Tümer L, Hasanoglu A, Biberoglu G, Fowler B (Turkey, Switzerland)

Unexplained familial methylmalonic aciduria: a benign situation or an unidentified inborn error?

16.20 h

Liu TT, Teng YT, Lee SF, Lam WF, Liu MY, Wu SJ, Hsiao KJ (Taiwan)

Methylmalonyl CoA mutase gene mutations in Chinese methylmalonic acidemia

16.40 h

Liu MY, Liu YN, Wu SJ, Liu TT, Hsiao KJ (Taiwan)

Identification of the PCCA and PCCB Gene mutations in Chinese propionic acidemia patients

17.00 h – Coffee break

17.30 h

II. Round table – Dysmorphology and Inborn errors of metabolism (Bzdúch V)

Wednesday 29th September 2004

VI. NEONATAL SCREENING AND VARIA

Chairpersons: Hsiao KJ and Bzdúch V

9.00 h

Hsiao KJ, Chiang SH, Liu TT (Taiwan)

Development of neonatal screening in Taiwan

9.25 h

Šimurka P, Rosipal Š, Dluholucký S, Ďatelová M, Hrubá F (Slovakia)

Universal screening of cholesterolaemia in Slovakia – the current state

9.45 h

Lugovska R, Krumina Z, Vevere P, Pronina N, Muceniece Z, Purina G, Lace B, Grauduma I (Latvia)

Inborn errors of metabolism in Latvia

10.05 h

Bzdúch V, Záhoráková D, Grechanina E, Zdibskaja EP, Goldfarb IG, Zeman J, Martásek P (Slovakia, Czech Republic, Ukraine)

A case of Rett syndrome from Ukraine - clinical diagnosis confirmed by the mutation analysis of the MECP2 gene

10.20 h – Coffee break

10.50 h

Huijmans JGM, de Klerk JBC, Williams M, Cairo WI, van der Meijden E, Duran M (The Netherlands)

Glutamine and glutamate in the differential diagnosis of hyperammonemias, organic acidurias, the hyperinsulinism/hyperammonemia (HIHA) syndrome and hyperglutamic acidemia

11.15 h

Grechanina EY, Grechanina JB, Gusar VA, Novikova IV (Ukraine)

The experience of diagnosis of mitochondriopathies in Ukraine

11.40 h

Lelovics Z (Hungary)

Modern dietary therapy of maple syrup urine disease

12.00 h

Klčová V, Bzdúch V, Sýkora P, Kolníková M, Behúlová D (Slovakia)

Ketogenic diet

12.15 h

Fabriciová K, Bzdúch V, Bibza J, Králik R, Klčová V (Slovakia)

Percutaneous endoscopic gastrostomy in patients with inborn errors of metabolism

12.30 h – Lunch and end of scientific programme