# **ISGNIE 2021** Scientific Program

# October 22

# Room 101

### **PC1**

Introduction of Neurometabolic Diseases in Infants and Children

Chair: Katsunori Konbayashi (Japan), Chao-Ching Huang (Taiwan)

- 08:30-09:00 Overview of Neurometabolic Diseases in Infants Ingrid Tein (The Hospital for Sick Children, Canada)
- 09:00-09:25 Mitochondrial Diseases: Infantile Epilepsy from Genetic Testing to Precision Management Ching-Shiang Chi (Tungs' Taichung MetroHarbor Hospital, Taiwan)
- 09:25-09:50 Investigations in Pediatric Neurometabolic Diseases Yann-Jang Chen (Taipei Veterans General Hospital, Taiwan)
- 09:50-10:10 Neurophysiological Monitoring on Neurometabolic Disorders Douglas R. Nordli (University of Chicago, USA)

## PC2

Mitochondrial Encephalopathy

Chair: Jun Natsume (Japan), Kun-Long Hung (Taiwan)

10:40-11:10	Approach to Mitochondrial Disorders in Children
	Haluk Topaloglu (Hacettepe University School of Medicine, Turkey)
11:10-11:35	Mitochondrial Encephalopathy: Clinical and Genetic Features
	Kei Murayama (Chiba Children's Hospital, Japan)
11:35-12:00	Molecular Basis of Neurometabolic and Neurogenetic Diseases
	Henrike O Hevne (University of Helsinki, Finland)

## L1

Luncheon Symposium - Phalanx Biotech

Chair: Dar-Shong Lin (Taiwan)

12:10-13:30 Dual Analysis Application of Whole Exome Sequencing and Microarray for the Diagnosis of Developmental Delay Children Li-Ping Tsai (Taipei Tzu Chi Hospital, Taiwan)

#### GMT+8

### **Room 101**

#### PC3

Amino Acid Metabolic Diseases

Chair: Cheuk Wing Fung (Hong Kong), Hsiu-Fen Lee (Taiwan)

- 13:30-14:00 Overview of Amino Acid Metabolic Diseases: Clinical Presentations and Genetic Roles
  - Cheuk Wing Fung (Hong Kong Children's Hospital, Hong Kong)
- 14:00-14:30 MSUD and Related Disorders: Clinical Presentations in Infants and Children Sylvia Estrada (Philippine General Hospital, Philippines)
- 14:30-15:00 Sulfite Oxidase Deficiency and Related Disorders: Neuroimaging Findings and Genetic Roles Syuan-Yu Hong (China Medical University Children's Hospital, Taiwan)

### **PC4**

#### **Miscellaneous Etiologies and Treatment**

Chair: Jao-Shwann Liang (Taiwan), Pi-Chuan Fan (Taiwan)

15:40-16:05	Artificial Intelligence Application in EEG Analysis: A New Strategy for the Neurophysiological Management of Neurometabolic Disorders Noboru Yoshida (Juntendo University Nerima Hospital, Japan)
16:05-16:35	Peroxisomal Disorders in Infants and Children Hsi Chang (Taipei Medical University Hospital, Taiwan)
16:35-17:00	Metbolic Imbalances in Fatty Acid Oxidation Disorders. Implications for Pathophysiology and Treatment Jerry Vockley (University of Pittsburgh, USA)
17:00-17:30	Lysosomal Diseases in Infancy and Children: Diagnostic Approach Jonathan Mink (University of Rochester Medical Center, USA)

# Room 102

#### L2

Luncheon Symposium - Sanofi

Chair: Wang-Tso Lee (Taiwan)

12:10-13:30 Streamlining the Diagnosis Pathway from Genotype to Phenotype in Hereditary Myopathy: Example from Pompe Disease Andrew Kornberg (The Royal Children's Hospital, Australia)

### CS1

#### **Cross-Strait Forum**

Chair: Yuwu Jiang (China), Nan-Chang Chiu (Taiwan)

15:30-15:50	Genetic Role in Guiding Epilepsy Treatment in Infants
	Yi-Fang Tu (National Cheng Kung University Hospital, Taiwan)
15:50-16:10	嬰兒期早發癲癇病因及臨床-單中心回顧性研究
	方方(首都醫科大學附屬北京兒童醫院)
16:10-16:30	The Predictors and Associated Factors for Ketogenic Diet Effectiveness-From the View of Metabolome
	Pi-Lien Hung (Kaohsiung Chang Gung Memorial Hospital, Taiwan)
16:30-16:50	生酮飲食治療線粒體病相關癲癇研究進展
	孫丹(華中科技大學同濟醫學院附屬武漢兒童醫院)
16:50-17:10	Epileptic Seizures in Patients with Glutaric Aciduria Type I
	Wei-Sheng Lin (Taipei Veterans General Hospital, Taiwan)
17:10-17:30	WDR45 變異相關的嬰幼兒癲癇
	周水珍(復旦大學附屬兒科醫院)
17:30-17:50	The Behavioral Phenotype and Developmental Trajectory of Autism Spectrum Disorder in Children Born Very Preterm
	Li-Wen Chen (National Cheng Kung University, Taiwan)
17:50-18:10	CLCN4 相關癲癇
	彭鏡(中南大學湘雅醫院)
18:10-18:30	With the Advance in the Field of Neuroinflammation, What Should We Know When We Approach a Neurologic Case in Pediatric Population?
	Chih-Fen Hu (Tri-Service General Hospital, Taiwan)

## **Room 101**

#### BS1

Update in Neurophysiology

Chair: Rei-Cheng Yang (Taiwan), Noboru Yoshida (Japan)

- 07:15-07:45 EEG Monitoring Approaches in Critically III Infants and Children Nicholas S. Abend (University of Pennsylvania, USA)
- 07:45-08:15 Update Applications of Neurophysiological Monitoring in Neonates Ronit Pressler (Great Ormond Street Hospital for Children, UK)

### **K1**

Prof. Yu-Zen Shen's Memorial Lecture

Chair: Wang-Tso Lee (Taiwan)

08:40-09:20 Recent Advance in Neurometabolic Diseases: The Genetic Role in Modern Era Ingrid Tein (The Hospital for Sick Children, Canada)

### K2

Taiwan Child Neurology Society 25th Anniversary Lecture

Chair: Hideo Yamanouchi (Japan)

09:20-10:00 Recent Advances in the Treatment of Neurometabolic Diseases *Phillip Pearl (Harvard Medical School, USA)* 

# Room 101

### **S1**

Neurometabolic Diseases and Epilepsy (I)

Chair: I-Ching Chou (Taiwan), Inn-Chi Lee (Taiwan)

10:30-10:50	Ion-Channel Disorders V.S. Neurometabolic Disorders in Newborns Inn-Chi Lee (Chung Shan Medical University Hospital, Taiwan)
10:50-11:10	Novel Treatment of Epileptic Encephalopathy in Neurometabolic Diseases Kazuhiro Muramatsu (Jichi Medical University, Japan)
11:10-11:30	Diet Therapy for Infants with Neurometabolic Disorders and Genetic Epilepsy Heung Dong Kim (Yonsei University College of Medicine, Korea)
11:30-12:00	Precision Medicine in Infantile Seizures Henrike O. Heyne (University of Helsinki, Finland)

## **L3**

Luncheon Sym	iposium - Biogen
Chair: Wang-Ts	o Lee (Taiwan)
12:10-13:30	Advances in Treatment of SMA and Taiwan SPINRAZA Treatment Experience Sharing

Yuh-Jyh Jong (Kaohsiung Medical University Chung-Ho Memorial Hospital, Taiwan)

# Room 101

### **S2**

Investigations in Neurometabolic Disease

Chair: Yuh-Jyh Jong (Taiwan), Yung-Ting Kuo (Taiwan)

13:30-14:00	Full Genome Analysis for Rare Genetic Diseases: as Applied to Infants with Neurometabolic Diseases
	Pui-Yan Kwok (Academia Sinica, Taiwan)
14:00-14:30	Pyridoxine-Dependent Epilepsy (PDE-ALDH7A1): Implications for Newborn Screening
	Laura Tseng (Amsterdam University Medical Center, The Netherlands)
14:30-15:00	Neuroimaging Features in Infants with Neurometabolic Diseases
	Kshitij Mankad (Great Ormond Street Hospital for Children & University College London, UK)

## 01

Oral Presentation I	
Chair: Marilyn	H. Ortiz (Phillipines), Shyi-Jou Chen (Taiwan)
15:30-15:40	A Case Series of Varied Manifestations of Glut1 Transporter Defect Harshuti Shah (Rajvee Hospital, India)
15:40-15:50	GNAO1-related Severe Involuntary Movements Treated with Deep Brain Stimulation <i>Mizuki Takagi (Aichi Medical University, Japan)</i>
15:50-16:00	COQ4 Mutations-related Infantile-onset Mitochondrial Disorder Associated with Primary Coenzyme Q10 Deficiency Chia-Jui Hsu (National Taiwan University Hospital Hsinchu Branch, Taiwan)

# **Room 101**

#### **S**3

#### Infantile Epilepsy in Mitochondrial Disorders

Chair: Norimichi Higurashi (Japan), Che-Sheng Ho (Taiwan)

16:00-16:30	Mitochondrial Transplantation in MELAS Disease
	Chin-San Liu (Changhua Christian Hospital, Taiwan)

- 16:30-16:50 Neurophysiologic Monitoring in Infants with Mitochondrial Diseases Ronit Pressler (Great Ormond Street Hospital, UK)
- 16:50-17:10 Infantile Onset Epilepsy in Mitochondrial Disorder: Clinical and Genetic Insights Jong Hee Chae (Seoul National University Hospital, Korea)
- 17:10-17:30 Paradigm Changes in the Genetics of the Infantile Epilepsies Understanding the Exome and Beyond *Ingo Helbig (Children's Hospital of Philadelphia, USA)*

### 02

### Oral Presentation II

Chair: Teik Beng Khoo (Malaysia), Yung-Ting Kuo (Taiwan)

17:30-17:40	The First Case Series of Bilateral Frontoparietal Polymicrogyria in Taiwan Cheng-Yen Kuo (Chang-Geng Medical Foundation Linkou Chang-Geng Memorial Hospital, Taiwan)
17:40-17:50	Clinical and Electrophysiological Features of Pyridoxamine 5' Phosphate Oxidase (PNPO) Deficiency Beyond the Neonatal Period Lakshmi Kalband (Children's Hospital at Westmead, Australia)
17:50-18:00	Alternating Hemiplegia of Childhood: A Malaysian Tertiary Centre Experience Lip Yuen Teng (Hospital Tunku Azizah Kuala Lumpur, Malaysia)
18:00-18:10	Focal Gyratory Seizure Associated with Ectopic Gray Matter in the Frontal Lobe Satomi Kakuta-Ohtaki (Saitama Medical University, Japan)
18:10-18:20	Therapeutic Effects and Mechanisms of Transcranial Photobiomodulation (tPBM) on Pediatric Epilepsy <i>Hsi Chang (Taipei Medical University Hospital, Taiwan)</i>

# Room 102

### L4

Luncheon Symposium - UCB

Chair: Kuang-Lin Lin (Taiwan)

12:10-13:30 A New Insight of AED Selection in Pediatric Epilepsy Ting-Rong Hsu (Taipei Veterans General Hospital, Taiwan)

## **Room 101**

#### BS2

Progressive Myoclonic Epilepsy in Infants and Children

Chair: Kuang-Lin Lin (Taiwan), Kazuhiro Muramatsu (Japan)

- 07:15-07:45 Overview of Progressive Myoclonic Epilepsy: Genetic Roles Nicola Specchio (Bambino Gesù Children's Hospital, Italy)
- 07:45-08:15 Progressive Myoclonic Epilepsies. Electro-Clinical Features Jorge Vidaurre (Nationwide Children's Hospital, USA)

### **K3**

Prof. Yukio Fukuyama's Memorial Lecture

Chair: Ching-Shiang Chi (Taiwan)

08:30-09:10 Metabolic Epilepsy in Infancy: The Role of Genes Pratibha Singhi (Medanta, India)

### 03

#### **Oral Presentation III**

Chair: Ming-Yuh Chang (Taiwan), Sung-Tse Li (Taiwan)

09:10-09:20	Brain Age Prediction from Electroencephalographic Maturation Assessed by Deep Learning Algorithm Shi-Bing Wong (Taipei Tzu Chi Hospital, Taiwan)
09:20-09:30	Electroencephalographic Patterns and Clinical-radiological Correlation in Children with Lissencephaly: A Case Series Report <i>Min-Lan Tsai (Taipei Medical University Hospital, Taiwan)</i>
09:30-09:40	Infantile Epileptic Encephalopathy Caused by the SCN8A Variant in a Child with Citrine Deficiency Marina Hashiguchi (Jichi Medical University, Japan)

## **Room 101**

#### **S4**

Neurometabolic Diseases and Epilepsy (II)

Chair: Tung-Ming Chang (Taiwan), Julie Chi Chow (Taiwan)

10:30-10:55	SSADH Deficiency: Clinical, Genetic Role, and Treatment Strategies
	Phillip Pearl (Harvard Medical School, USA)
10:55-11:20	Genetic Investigation in Lipid Metabolism with Infantile Epilepsy
	Tina-Rona Hsu (Taipei Veterans General Hospital Taiwan)

- 11:20-11:45 The Role of Autophagy in the Pathomechanism and Treatment of Leukodystrophy Dar-Shong Lin (Mackay Memorial Hospital, Taiwan)
- 11:45-12:10Update Treatment for Leukodystrophy and Its Associated Epilepsy in Infants<br/>Wang-Tso Lee (National Taiwan University Children's Hospital, Taiwan)

## L5

Luncheon Symposium - Eisai

Chair: Wang-Tso Lee (Taiwan)

12:10-13:30 Role of Perampanel in Early Add-on Treatment for Pediatric Patients with Epilepsy
Hsiu-Fen Lee (Taichung Veterans General Hospital, Taiwan)
Management of LGS and Rufinamide Experience Sharing
Chu-Chin Chen (Kaohsiung Veterans General Hospital, Taiwan)

## **Room 101**

#### **S5**

#### Neurometabolic Diseases and Epilepsy (III)

Chair: Kai-Ping Chang (Taiwan), Wen-Chen Liang (Taiwan)

13:30-13:55	Congenital Disorders of Glycosylation and Infantile Epilepsy
	Hsiu-Fen Lee (Taichung Veterans General Hospital, Taiwan)

- 13:55-14:20 Infantile Epilepsy and Carnitine Inborn Errors of Metabolism: The Role of Genes *Shinichi Hirose (Fukuoka University, Japan)*
- 14:20-14:40 Organic Acid and Infantile Epilepsy: The Role of Genes Yi Wang (Children's Hospital of Fudan University, China)
- 14:40-15:00 Diagnosis of Neurometabolic Disorders Involving Basal Ganglia based on Neuroimages Shekeeb Mohammad (The Children's Hospital at Westmead, Australia)

### **S6**

### Treatable Neurometabolic Diseases and Epilepsy

Chair: Ting-Rong Hsu (Taiwan), Chih-Fen Hu (Taiwan)

15:30-15:55	Glucose Transporter 1 Deficiency : Past Experience, Current Status, and Future Challenges
	Shin Nabatame (Osaka University, Japan)
15:55-16:20	Pyridoxine-Responsive and Dependent Epilepsy
	Huei-Shyong Wang (Chang Gung Medical Center, Taiwan)
16:20-16:45	Biotinidase Deficiency and Infantile Seizures
	I-Ching Chou (China Medical University Hospital, Taiwan)
16:45-17:10	Novel Treatment of Mitochondrial Disorders
	Tsu-Kung Lin (Kaohsiung Chang Gung Memorial Hospital, Taiwan)
17:10-17:35	Creatine Transporter Deficiency and Epilepsy
	Ming-Tao Yang (Far Eastern Memorial Hospital, Taiwan)

## Room 102

#### **L6**

Luncheon Symposium - GSK

Chair: Huei-Shyong Wang (Taiwan)

12:10-13:30 Seizure Control in Times of Emergency: Start with a High-Risk Teratogenicity Medication for Immediate Control And Switch to a Low-Risk Teratogenicity Option Later?

Yen-Ju Chu (National Taiwan University Hospital, Taiwan)