Medical Institute for Research And Innovation for Children

The 10th Azalea Festival Symposium in Japan

2026 3.27 (Fri) ▸ 29 (Sun)

Main Theme

International Symposium on "The Molecular Pathomechanisms and Therapeutic Strategies of Rare and Intractable Epilepsies"

Programs & Abstracts

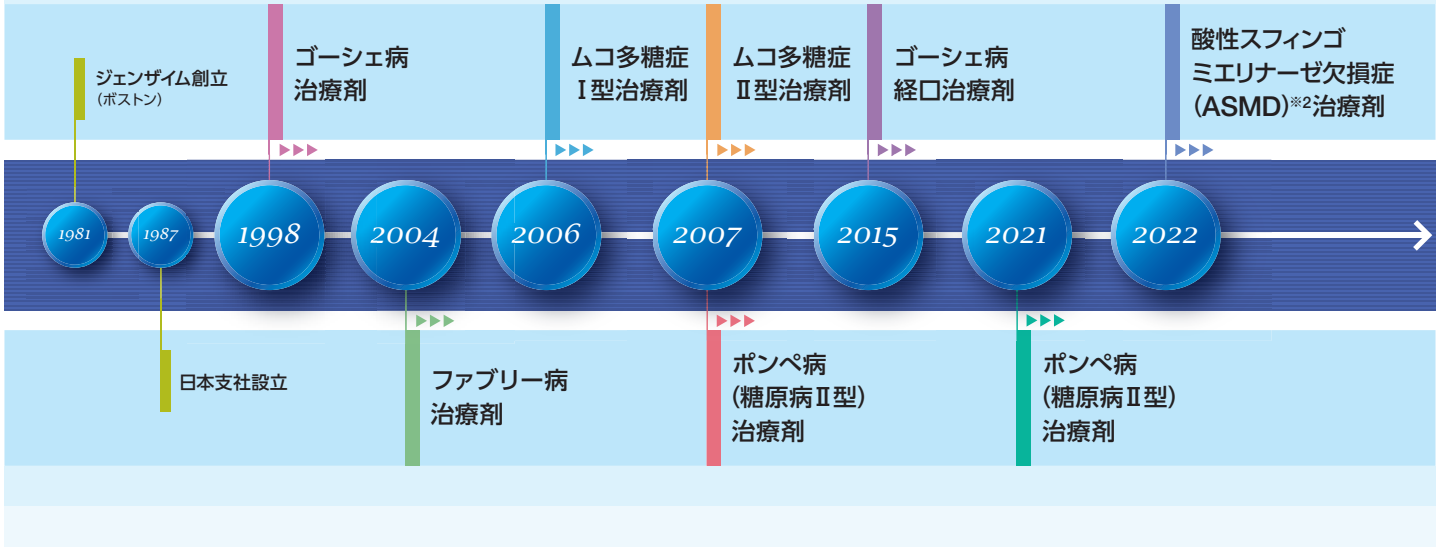
Venue

Medical Hall, Fukuoka University Hospital

President

Shinichi Hirose, M.D., Ph.D.
Professor, General Medical Research Center,
School of Medicine, Fukuoka University
JAPAN

サノフィは
スペシャルティケアのリーダーとして、
患者さんとそのご家族に
希望をお届けしていきます。



ジェンザイム※1として始まり30余年、
希少疾患の患者さんと共に　これからも。

※1：現・サノフィ スペシャルティケアビジネスユニット
※2：別名ニーマン・ピック病A型、B型　他に中間型がある

2022年4月作成
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サノフィ株式会社

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The 10th Azalea Festival Symposium in Japan (Azalea2026)

The Molecular Pathomechanisms and Therapeutic Strategies of Rare and Intractable Epilepsies

Program and Abstracts

Date: March 27 (Fri.) – March 29 (Sun.), 2026

Venue: Medical Hall, Fukuoka University Hospital

President: Shinichi Hirose, M.D., PhD
Professor, General Medical Research Center,
School of Medicine, Fukuoka University

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Welcome Message

President
Shinichi Hirose, M.D., Ph.D.

Professor, General Medical Research Center,
School of Medicine, Fukuoka University

Warmest Greeting!

I am greatly honored to host the 10th Azalea Festival symposium, which is the International Symposium on the Molecular Pathomechanisms and Therapeutic Strategies of Rare and Intractable Epilepsies and shall be held at Medical Hall of Fukuoka University Hospital, Japan on March 27 – 29, 2026.

The Azalea Festival Symposium has been held annually since 2017 at National Taiwan University, coinciding with the Azalea Festival when azaleas bloom in Taiwan. Its primary focus has been on research and treatment development for intractable pediatric neurological disorders. Over the years, many domestic and international experts have gathered at this symposium to present their latest research findings and discuss prospects for clinical applications.

As we approach the 10th anniversary, it has been decided to hold the event outside of Taiwan for the first time, with Fukuoka, Japan, chosen as the commemorative venue. Japan plays a globally significant role in molecular pathology and genetic research, with its studies on rare intractable epilepsies receiving high acclaim both domestically and internationally. This symposium will invite renowned researchers and clinicians from around the world to share cutting-edge insights under the theme "The Molecular Pathomechanisms and Therapeutic Strategies of Rare and Intractable Epilepsies" a first for the Azalea Festival Symposium.

Epilepsy is the second most common neurological disorder after headaches and has recently gained public attention through traffic accidents and other incidents. However, 30% of epilepsy cases are resistant to multiple anti-epileptic drugs and are classified as intractable epilepsy. In particular, designated rare intractable epilepsies are the most severe, with virtually no effective treatments. They not only cause severe delays in mental and motor development but can also be life-threatening. As a result, the burden on patients and their families is immeasurable.

Nevertheless, the advent of next-generation sequencing, an innovative technology in molecular biology research, has enabled the identification of genetic abnormalities in intractable epilepsies, including designated rare diseases, over the past decade. This has finally made treatment research possible. Consequently, there has been a strong demand for a symposium where researchers from around the world can gather to discuss genetic research on rare intractable epilepsies and related treatment studies. We are confident that this 10th Azalea Festival Symposium will be a milestone in the history of research on elucidating the pathophysiology and treatment strategies for rare intractable epilepsies.

Additionally, to deepen the understanding of patients and their families, we will hold a public lecture to communicate research findings in an accessible manner. Furthermore, we will introduce poster presentations and a travel grant system aimed at nurturing young researchers, creating an environment where a new generation of scientists can contribute to this field.

I am confident we can work together to make the most of this opportunity and I welcome the chance to meet and exchange ideas with many of you as I can.

Thank you all for coming!

General Information

■ Meeting Name

The 10th Azalea Festival Symposium

■ Theme

The Molecular Pathomechanisms and Therapeutic Strategies of Rare and Intractable Epilepsies

■ Date

March 27 – 29, 2026

■ Venue

Medical Hall, Fukuoka University Hospital
7-45-1 Nanakuma, Jonan, Fukuoka, 814-0180
Tel: 092-801-1011

■ Official Language

English

■ President

Shinichi Hirose
Professor, General Medical Research Center,
School of Medicine, Fukuoka University

■ Sponsored by

公益財団法人
難病医学研究財団　**Japan Intractable Diseases Research Foundation**

主催：公益財団法人　難病医学研究財団

■ Cosponsored by

Medical Institute for Research And Innovation for Children

共催：一般社団法人　小児医学研究所

■ Congress Secretariat

JTB Communication Design Inc.
1-1-35-4F, Nagahama, Chuo-ku, Fukuoka 810-0072, Japan
TEL: +81-92-751-3244　FAX: +81-92-751-3250
E-mail: azalea2026@jtbcom.co.jp

Chairs and Speakers

■ Guideline for Chairs

1. Please come to see “Invited” desk of the Registration area in Foyer 30minutes before the start of the session.
2. Please show the invitation email titled “Important _Final Notice Regarding the Syposium” sent by the secretariat. (or you may show the e-mail record on your device). We will provide you a name badge.
3. Please be seated in the “Next-chair’s seat” 10 minutes before your session time.
4. Kindly be sure to make the session finished in scheduled time.

■ Guideline for Speakers

1. Please come to see “Invited” desk of the Registration area in Foyer 30minutes before the start of the session.
2. Please show the invitation email titled “Important _ Final Notice Regarding the Symposium” sent by the secretariat. (or you may show the e-mail record on your device). We will provide you a name badge.
3. Please be seated in the “Next-speaker’s seat” 10 minutes before your presentation time.
4. Speakers are required to bring their own laptops for presentations. The projector is only compatible with HDMI terminal (Type A: standard type), and other types are not supported. If you need a conversion cable, please bring your own.
5. The screen ratio is 16:9.
6. There will be no preview room. However, you will be able to check connections before each session begins or during breaks in the session when the chair changes.
7. Please be sure to have a USB flash drive with a backup of your data in case of computer malfunction.

■ Guideline for Poster Presentation

1. Schedule and Venue

Poster Session Venue

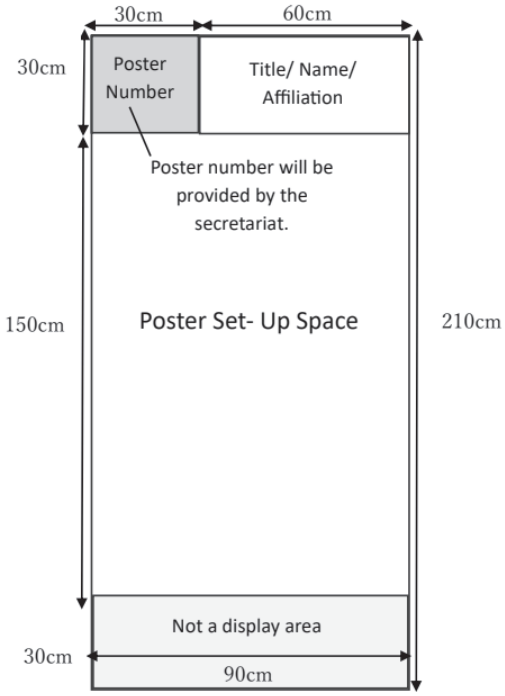
B1F Multipurpose Hall, Fukuoka University Hospital

Schedule

Poster Mounting: March 27 (Fri.) 12:00-13:00
Poster Viewing: March 27 (Fri.) 13:00-16:30,
March 28 (Sat.) 9:30-17:00
Poster Removal: March 28 (Sat.) 16:30-17:10
March 29 (Sun.) 9:00-10:30

2. Poster Session Procedures

- Poster panel size (prepared at the site): height 210cm × width 90cm.
- Poster number (30cm × 30cm) will be placed on the top left of the panel in advance.
- Each presenter should prepare the title, presenter's name and affiliation (height 30cm × width 60cm)
- Pushpins will be provided by the Secretariat. Please use the pushpins to place your poster on the panel.
- Any poster left after the scheduled removal time will be disposed of by the Secretariat.



■ Guideline for Travel Grant Recipients

The 10th Azalea Festival Symposium offers travel grants to authors of outstanding abstracts submitted by non-Japanese residents living abroad.

- Travel grants are awarded to the first author of the submitted abstract.
- The grants will be awarded at the symposium venue, and all recipients must attend the 10th Azalea Festival Symposium in person.
- Recipients are responsible for arranging their own transportation and accommodation during the meeting.
- The award money is prepared to cover recipients' travel expenses.
- Each recipient needs to visit the secretariat room in the symposium venue during the time requested by the secretariat, to receive the grant.

Information for Participants

IMPORTANT NOTICE:

Registration is accepted exclusively through the online system. On-site registration in-person will not be available at the venue. Payment can only be made by credit card.

To avoid congestion at the registration desk, **please complete your registration online before arriving at the venue.**

■ Registration time and venue

1. Invited guests

   Please come to see "Invited" desk of the Registration area in Foyer and show the invitation email titled "final announcement" sent by the registration office. (or you may show the e-mail record on your device). We will provide you a name badge.
2. Already registered participants

   Please come to see "Advance registration" desk of the Registration area in Foyer and show the email titled "[The 10th Azalea Festival Symposium] Payment Confirmation" automatically sent from the registration system. (or you may show the e-mail record on your device). We will provide you a name badge.
3. Any inquiries and those who has problem in registration

   Please visit "Information" desk.
4. Registration desk

   Registration desk will be open during the following hours at the 1st floor foyer of the symposium venue.

March 27 (Fri.)	9:00-15:00
March 28 (Sat.)	9:00-16:30
March 29 (Sun.)	9:00-10:30

5. Registration fee

Registration Category	Late & On-site (From Feb. 4, 2026)
Member (AOCNA) *1	45,000 JPY
Non-Member	60,000 JPY
Student *2	25,000 JPY
Least Developed Countries (LDC) *3	25,000 JPY
Accompanying Person *4	25,000 JPY

*1: Member rate is applicable to the person belong to the Asian Oceanian Child Neurology Association (AOCNA). Member must input his/her membership number. If you do not know your membership number, please check at https://www.aocna.org/member/search/.

*2: Student are required to upload a scanned copy (in either jpeg or pdf format) of their student ID

via online registration form. Please be advised that "Student" rate will not be applied for students who have full-time job.

*3: The applicable countries are listed below.
https://azalea2026.org/doc/ldc_list_2024undata.pdf

*4: Accompanying person is restricted to the family members of the participants.

■ Badge Policy

Name badge is required to wear for everyone to enter the symposium venue, poster room and every social program site.

■ Cloakroom (Baggage Storage)

Cloakroom is available during the symposium at the following place. Please note that we cannot accept responsibility for any loss or damage of valuables in items stored in the cloakroom.

March 27 (Fri.)	9:00-16:30
March 28 (Sat.)	9:00-17:30
March 29 (Sun.)	9:00-11:30

■ Pre-congress

All registered participants are available to join the Pre-congress lectures held on March 27 for 10:00-12:00.

■ Lunch

Lunch is available at the lunch seminar time. The box lunch is prepared. Vegetarian menu lunch box is also available. Please be noted that the lunch is served as first come basis.

■ Coffee Break & Poster

There are two slots of coffee break time. Coffee is served at the next room of poster venue. Please get some drink and snacks and enjoy poster viewing.

■ Photography/Recording

Photography taking and recording are strictly prohibited. Please also mind setting your mobile phone to silent mode during the session time.

■ Welcome Reception

Date Friday, March 27
Time 18:00-20:00
Venue The Luigans Spa and Resort, Marine World
(Add: 18-25 Saitozaki, Higashi-ku, Fukuoka/ Tel:092-603-2435)

■ Banquet

Date Saturday, March 28
Time 18:30-20:30
Venue Hotel New Otani Hakata
(Add: 1-1-2 Watanabe dori, Chuo-ku, Fukuoka / Tel: 092-714-1111)

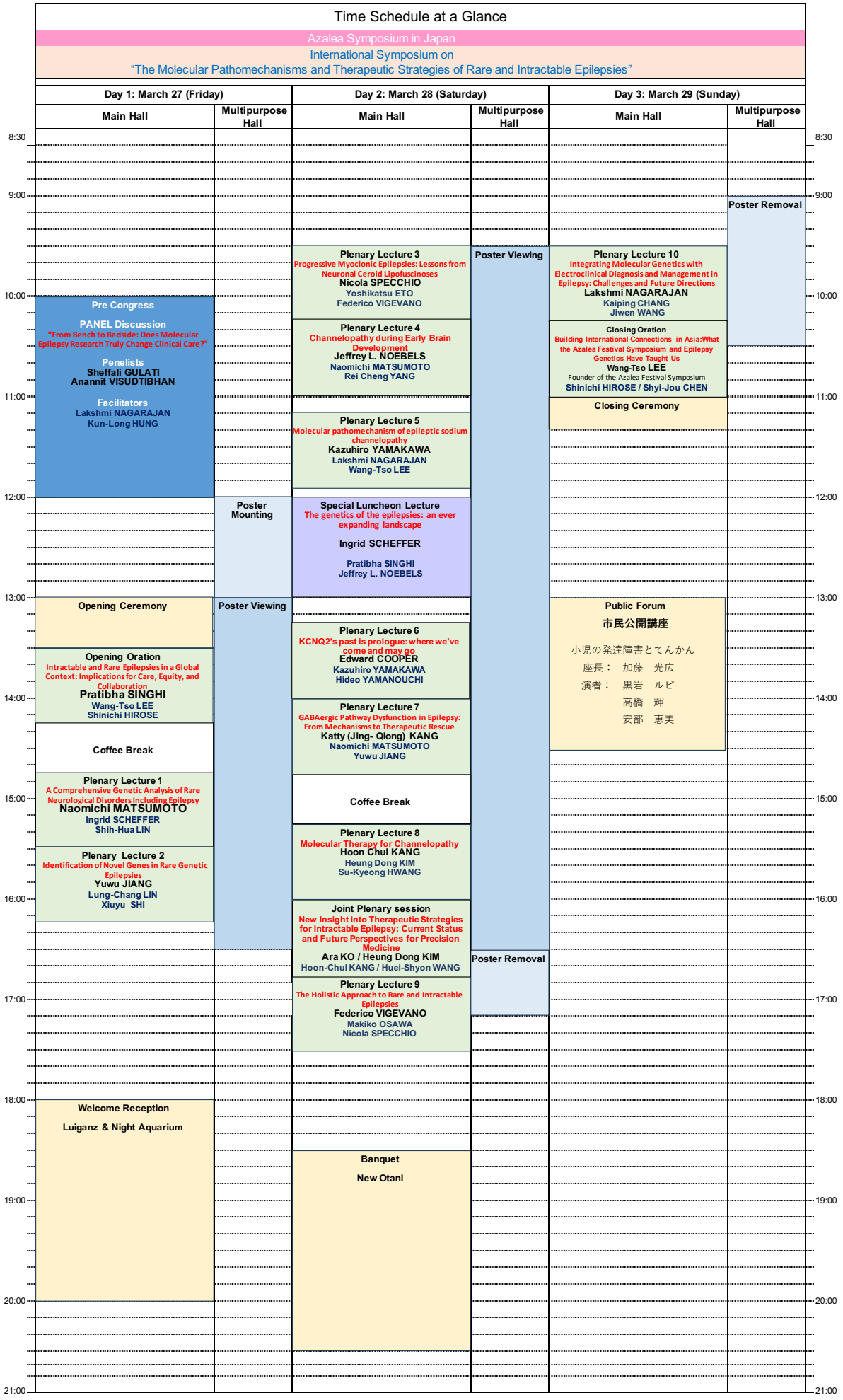
■ Shuttle Bus Service

There are shuttle services for a welcome reception dinner and for a banquet dinner. Please find following information.

Note: time is tentative and subject to change

Social Program	Shuttle Service (going)	Shuttle Service (return)
Welcome Reception	From Medical Hall Depart after 16:30	From the Luigans Depart around 20:00
Banquet	From Medical Hall Depart after 17:45	No operation

Time Table


Time Schedule at a Glance
Azalea Symposium in Japan
International Symposium on
"The Molecular Pathomechanisms and Therapeutic Strategies of Rare and Intractable Epilepsies"
Day 1: March 27 (Friday)
Day 2: March 28 (Saturday)
Day 3: March 29 (Sunday)
Main Hall
Multipurpose Hall
Pre Congress
PANEL Discussion
"From Bench to Bedside: Does Molecular Epilepsy Research Truly Change Clinical Care?"
Penelists
Sheffali GULATI
Anannit VISUDTIBHAN
Facilitators
Lakshmi NAGARAJAN
Kun-Long HUNG
Opening Ceremony
Opening Oration
Intractable and Rare Epilepsies in a Global Context: Implications for Care, Equity, and Collaboration
Pratibha SINGHI
Wang-Tso LEE
Shinichi HIROSE
Coffee Break
Plenary Lecture 1
A Comprehensive Genetic Analysis of Rare Neurological Disorders Including Epilepsy
Naomichi MATSUMOTO
Ingrid SCHEFFER
Shih-Hua LIN
Plenary Lecture 2
Identification of Novel Genes in Rare Genetic Epilepsies
Yuwu JIANG
Lung-Chang LIN
Xiuyu SHI
Welcome Reception
Luiganz & Night Aquarium
Poster Mounting
Poster Viewing
Plenary Lecture 3
Progressive Myoclonic Epilepsies: Lessons from Neuronal Ceroid Lipofuscinoses
Nicola SPECCHIO
Yoshikatsu ETO
Federico VIGEVANO
Plenary Lecture 4
Channelopathy during Early Brain Development
Jeffrey L. NOEBELS
Naomichi MATSUMOTO
Rei Cheng YANG
Plenary Lecture 5
Molecular pathomechanism of epileptic sodium channelopathy
Kazuhiro YAMAKAWA
Lakshmi NAGARAJAN
Wang-Tso LEE
Special Luncheon Lecture
The genetics of the epilepsies: an ever expanding landscape
Ingrid SCHEFFER
Pratibha SINGHI
Jeffrey L. NOEBELS
Plenary Lecture 6
KCNQ2's past is prologue: where we've come and may go
Edward COOPER
Kazuhiro YAMAKAWA
Hideo YAMANOUCHI
Plenary Lecture 7
GABAergic Pathway Dysfunction in Epilepsy: From Mechanisms to Therapeutic Rescue
Katty (Jing- Qiong) KANG
Naomichi MATSUMOTO
Yuwu JIANG
Coffee Break
Plenary Lecture 8
Molecular Therapy for Channelopathy
Hoon Chul KANG
Heung Dong KIM
Su-Kyeong HWANG
Joint Plenary session
New Insight into Therapeutic Strategies for Intractable Epilepsy: Current Status and Future Perspectives for Precision Medicine
Ara KO / Heung Dong KIM
Hoon-Chul KANG / Huei-Shyon WANG
Plenary Lecture 9
The Holistic Approach to Rare and Intractable Epilepsies
Federico VIGEVANO
Makiko OSAWA
Nicola SPECCHIO
Banquet
New Otani
Poster Viewing
Poster Removal
Plenary Lecture 10
Integrating Molecular Genetics with Electroclinical Diagnosis and Management in Epilepsy: Challenges and Future Directions
Lakshmi NAGARAJAN
Kaiping CHANG
Jiwen WANG
Closing Oration
Building International Connections in Asia:What the Azalea Festival Symposium and Epilepsy Genetics Have Taught Us
Wang-Tso LEE
Founder of the Azalea Festival Symposium
Shinichi HIROSE / Shyi-Jou CHEN
Closing Ceremony
Public Forum
市民公開講座
小児の発達障害とてんかん
座長： 加藤 光広
演者： 黒岩 ルビー
高橋 輝
安部 恵美
Poster Removal
8:30
9:00
10:00
11:00
12:00
13:00
14:00
15:00
16:00
17:00
18:00
19:00
20:00
21:00

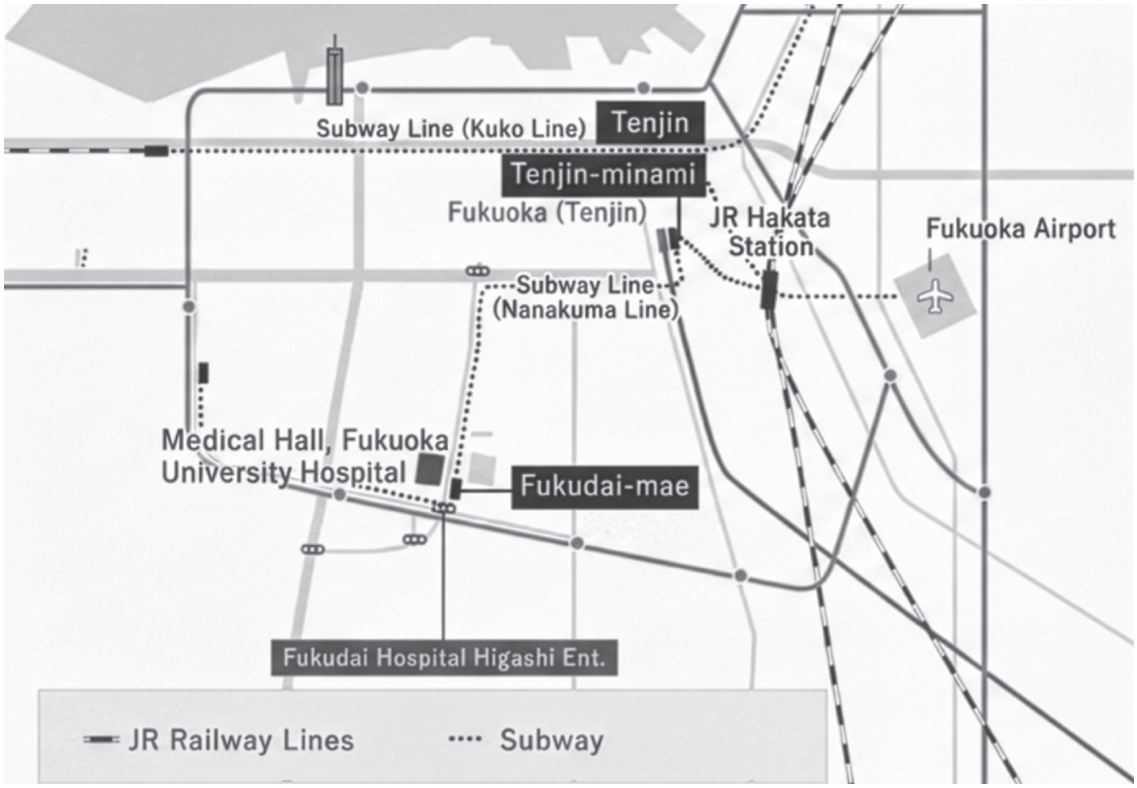
Venue and Access

Venue

Symposium Venue

Medical Hall, Fukuoka University Hospital
Add: 7-45-1 Nanakuma, Jonan, Fukuoka, 814-0180
Tel: 092-801-1011

Access



From Fukuoka Airport

Fukuoka Airport Station → Subway Kuko Line (approx. 5 min.) → JR Hakata Station → Subway Nanakuma Line (approx. 20 min.) → Fukudaimae Station

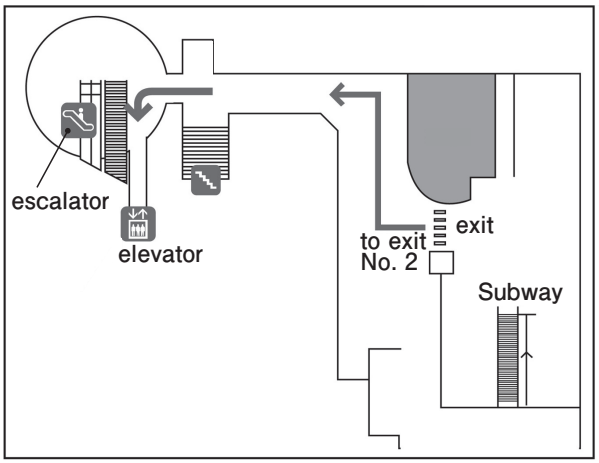
* Change trains at Hakata Station

From JR Hakata Station

JR Hakata Station → Subway Nanakuma Line (approx. 20 min.) → Fukudaimae Station

Location Map

B2F



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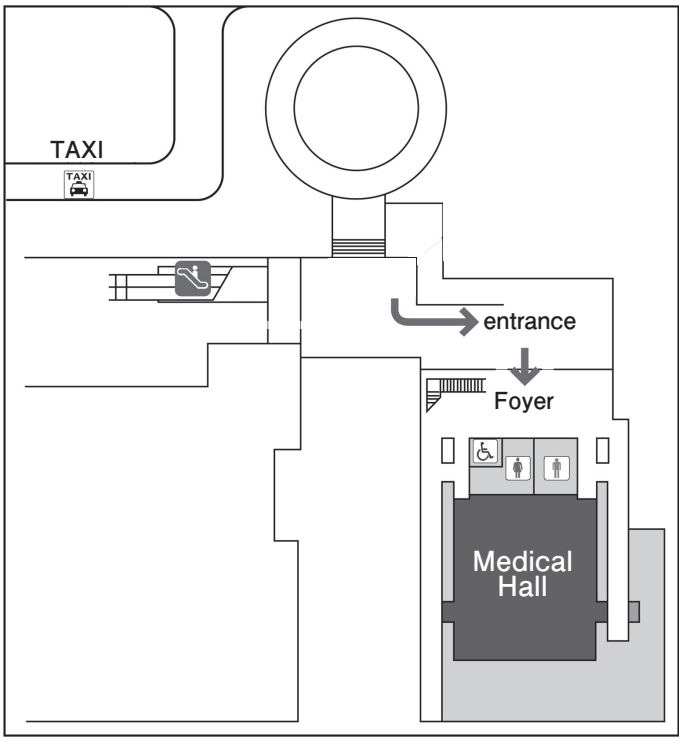

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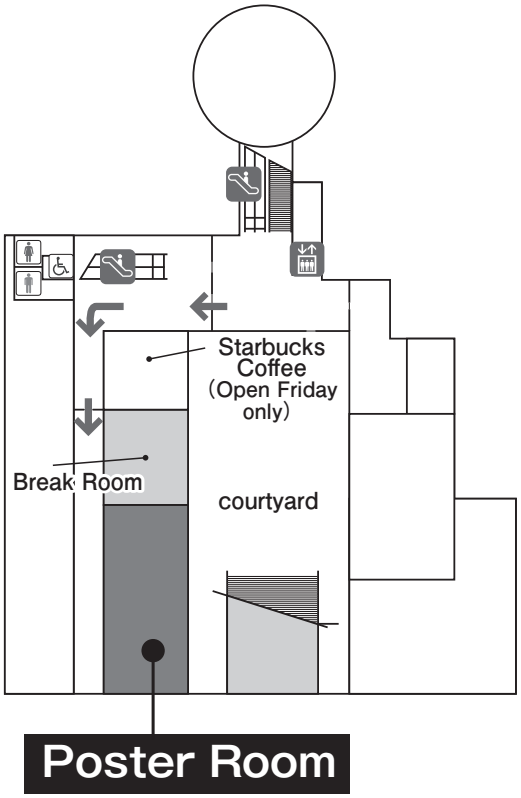

Floor Map

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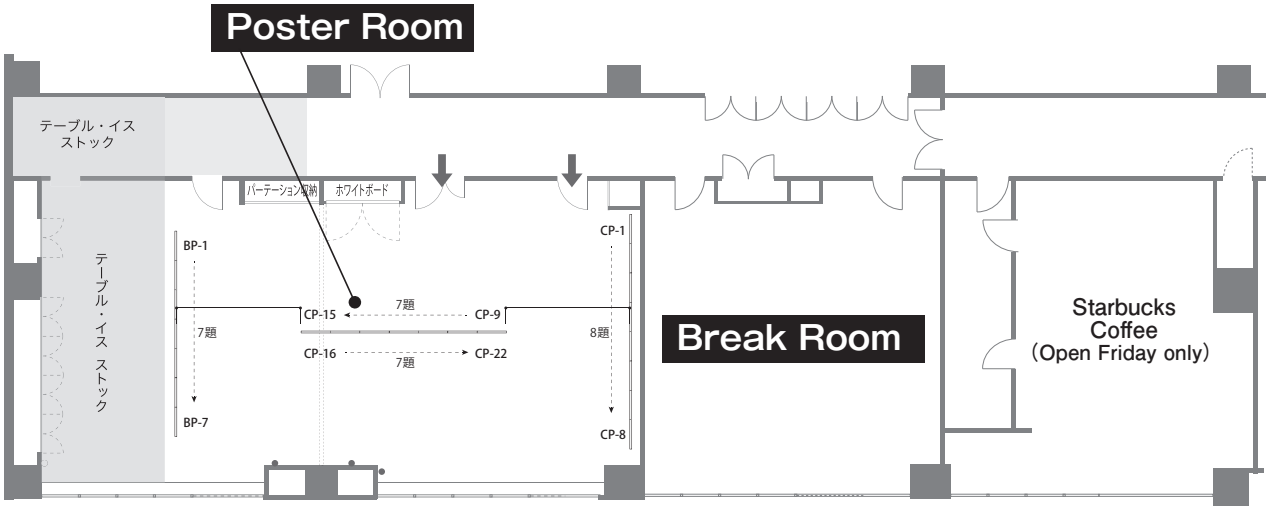

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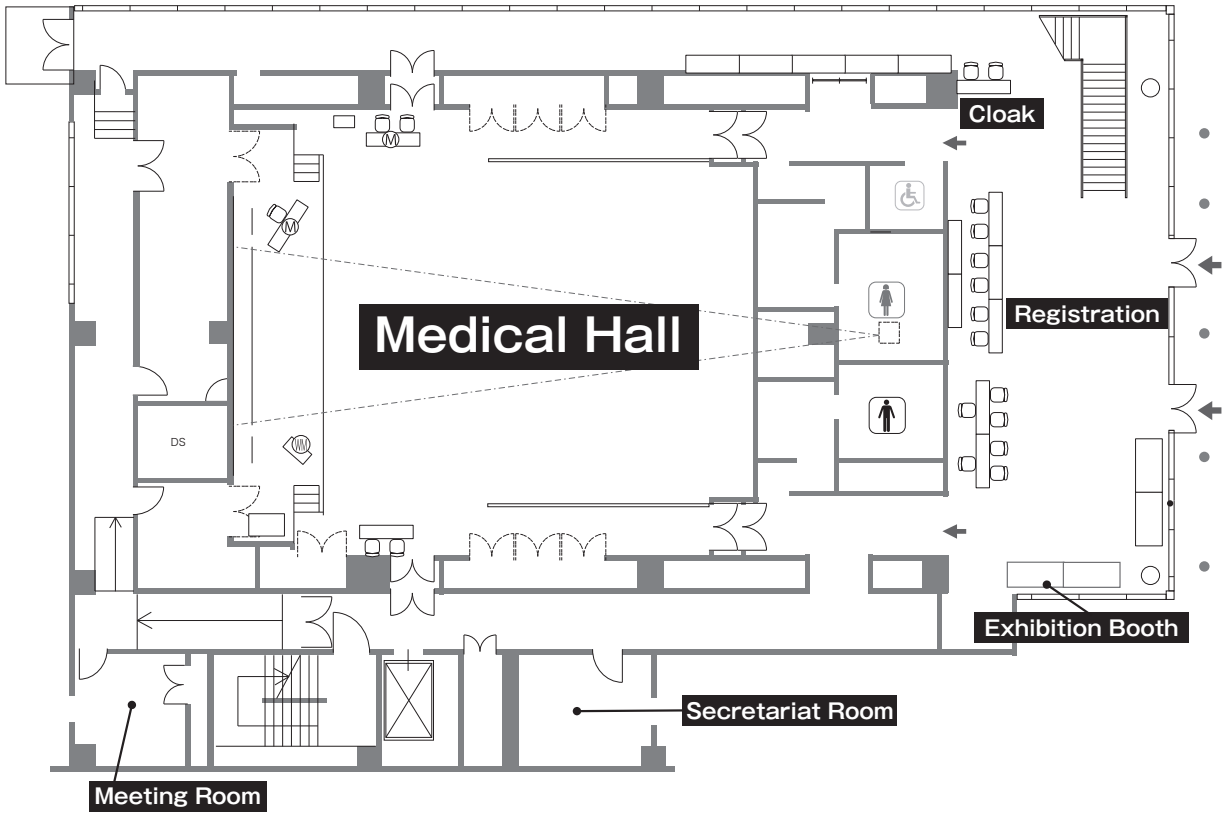


Program

Invited Speakers

Edward C. Cooper M.D., Ph.D.

Graduate of Yale College and Yale University School of Medicine's Medical Scientist Training Program.

Resident in Neurology and post-doctoral research fellowship (Lily Jan laboratory) at UCSF.

Career has focused on voltage-gated ion channels, particularly Kv channels as Mendelian causes of epilepsy and neurodevelopmental disorders and therapeutic targets.

First showed that brain KCNQ2 and KCNQ3 channels co-assemble, co-localize at axon initial segments, interact with ankyrin-G, and bind ankyrin-G through conserved mechanisms.

Has led internation efforts to analyze genotype-phenotype relationships among the KCNQ-related disorders and develop mechanism-driven novel therapies, through in vitro, animal model, and patient oriented research.

Sheffali GULATI, M.D. Pediatrics, FRCPCH (UK), FAMS, FIAP, FIMSA, FNASc, FIANS, FASc, Fellow, INSAR, Distinguished Fellow, GAPIO

She co-founded South Asia's first DM Pediatric Neurology Program in 2004 and currently serves as Professor and Faculty In-charge of the Child Neurology Division at AIIMS, New Delhi.

She leads the Centre of Excellence & Advanced Research for Childhood Neurodevelopmental Disorders and serves as Adjunct Faculty at the National Brain Research Centre, with clinical and research interests in autism, epilepsy, neuromuscular disorders, cerebral palsy, and other neurodevelopmental disorders.

She has authored over 526 publications, delivered more than 950 invited talks, and is listed among Stanford University's top 2% scientists (2025).

Yuwu Jiang, M.D., PhD

Dr. Yuwu Jiang is the Chair of the Children's Medical Center and Director of the Pediatric Epilepsy Center at Peking University First Hospital, and is the Chair of the Pediatrics Faculty at Peking University Health Science Center (PUHSC). He serves as Vice Chair of the Chinese Association Against Epilepsy (CAAE) and the Chinese Pediatric Society (CPS), and is the Immediate Past Chair of the Chinese Child Neurology Society (CCNS) under CPS. He chairs the Chinese Association of Pediatric Neurologists (CAPN) under the China Neurology Association (CNA). He is an Executive Board Member of the International Child Neurology Association (ICNA) and the China national delegate to the Asian Oceanian Child Neurology Association (AOCNA). He is also a Guest Professor at the University of Manchester.

Hoon-Chul Kang, M.D., Ph.D.

Professor at Division of Pediatric Neurology, Chief, Division of Pediatric Neurology, Department of Pediatrics, Severance Children's Hospital, Yonsei University College of Medicine, Seoul, Korea

EDUCATION

1986. 3 – 1988. 2	Premedical Course, College of Liberal Arts & Sciences, Yonsei University
1988. 3 – 1992. 2	College of Medicine (Bachelor's Degree of Medical Science), Yonsei University
2003. 9 – 2006. 2	Graduate School of Medicine (Ph.D. in Medical Science) Yonsei University, titled by "Behavioral improvement after transplantation of neural precursors derived from embryonic stem cells into globally ischemic brain of adolescent rats"

PROFESSIONAL APPOINTMENTS

2002. 3 – 2008. 8	Instructor, Assistant Professor at Department of Pediatrics, Epilepsy Center, Sanggye Paik Hospital, Inje University College of Medicine, Seoul, Korea
2009. 3 – 2013. 2	Clinical Associate Professor, Severance Children's Hospital
2013. 3 – Present	Professor at Division of Pediatric Neurology, Chief, Division of Pediatric Neurology, Chief, Department of Pediatrics, Chief, Severance Children's Hospital, Yonsei University College of Medicine, Seoul, Korea

Katty (Jingqiong) Kang, MD, PhD

Professor, Department of Neurology, Vanderbilt University Medical Center

My lab is dedicated to developing more effective, mechanism-based treatments for epilepsy.

We have focused on GABAA receptor and GABA transporter 1/SLC6A1 mutation associated neurodevelopmental disorders and epilepsies

We use both cell and mouse models that bear patient mutations including patient iPSC derived neurons and astrocytes as well as knockin mouse models.

We identified that impaired trafficking of mutant protein is a major patho-mechanism for those disorders.

We identified common and differential mechanisms between the GABAA receptor and GABA transporter 1/SLC6A1 mutations

We repurposed 4 phenylbutyrate, an FDA approved drug for urea cycle disorders to treat children with SLC6A1 mutation mediated disorders with promising results.

Heung Dong Kim, M.D., PhD

Prof. Heung Dong Kim, Distinguished Professor, Department of Pediatrics, Chief, Epilepsy Center, Kangbuk Samsung Hospital, Sungkyunkwan University College of Medicine, graduated Yonsei University College of Medicine, Seoul, Korea, trained at Severance Hospital in Pediatrics and Peidatric Neurologyand spent 2 years of exchange fellowship training at the Children's Hospital of Philadelphia from 1994 to 1996. He served as the president of Korean Epilepsy Society from 2011 to 2013, and the president of Korean Bureau for Epilpesy since 2018. He participated several committees of ILAE and served the Chair, Dietary Task Force, Commission of Medical Therapies, ILAE, in 2016-2021. He receive the Outstanding Achievement of Outstanding Achievement Award, International League against in Epilepsy-Asia-Oceania, 2025

Ara Ko, M.D., PhD

Assistant Professor 2018 – 2021

Pusan National University College of Medicine, Pusan National University Children's Hospital, Department of Pediatrics, Division of Pediatric Neurology Yangsan, Korea

Researcher 2021 – 2023

Korea Advanced Institute of Science and Technology (KAIST), Graduate School of Medical Science and Engineering Laboratory of Dr. Jeong Ho Lee, Daejeon, Korea

Assistant Professor 2023 – present

Yonsei University College of Medicine, Severance Children's Hospital, Department of Pediatric Neurology, Department of Pediatrics, Seoul, Korea

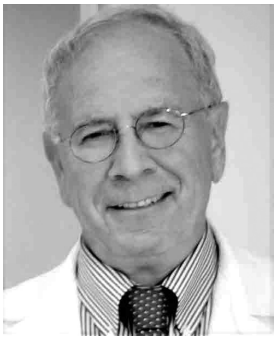
Wang-Tso Lee M.D. Ph.D.

Naomichi Matsumoto M.D. Ph.D.

Prof. Naomichi Matsumoto is Professor and Chair of the Department of Human Genetics at Yokohama City University Graduate School of Medicine.
A leading researcher in human genetics with an H-index of 95, he has identified the causative genes of more than 50 developmental genetic disorders.
His discoveries include genes responsible for Sotos syndrome, Marfan syndrome type II, Coffin-Siris syndrome, SENDA, and NIID.
He served as Editor-in-Chief of the Journal of Human Genetics (2014–2020) and continues as an advisory editor.
His work has significantly advanced the understanding and diagnosis of rare genetic diseases.
He has been serving as President of the Japan Society of Human Genetics since 2024.

Lakshmi Nagarajan, MBBS, MD, FRACP

Designation: Child Neurologist / Epileptologist
Affiliation: Perth Children's Hospital. University of Westen Australia
Under and Postgraduate Training: India, Australia, USA.
Areas of Interest: Epilepsy, Neonatal Seizures, Neurophysiology
Award: Inaugural WA Child & Adolescent Services Excellence in Clinical Care Award, 2025
Chair: Constitution and Bylaws Committee, AOCNA
Ex: ND from Australia to AOCNA
Secretary and President-Elect of ICNA



Jeffrey L. Noebels M.D. Ph.D.

Dr Noebels is Cullen Chair in Neurogenetics and Professor of Neurology, Neuroscience, and Molecular and Human Genetics, and Director of the Developmental Neurogenetics Laboratory at Baylor College of Medicine in Houston.
Received his PhD in Neuroscience at Stanford, postdoctoral training in Neuropathology at Harvard, MD at Yale, and neurology residency training at Massachusetts General Hospital.
Past President of the American Epilepsy Society, Chair of ILAE Genetics and Neurobiology Commissions
ILAE Ambassador and Fellow of the American Association of Arts and Sciences.
His discovery of the first single gene mouse model for childhood absence epilepsy in 1979 transformed the experimental approach to this disease.
Major research focus is to identify gene mechanisms of cortical network synchronization and molecular targets for treatment of epilepsy and related comorbidities.

Ingrid Scheffer, AO MBBS HonLLD PhD FRACP FAA AHMS FRS

Laureate Professor Ingrid Scheffer AO MBBS HonLLD PhD FRACP FAA AHMS FRS is a physician-scientist whose work as a paediatric neurologist and epileptologist at the University of Melbourne and Florey Institute has led epilepsy genetics research over more than 27 years. In collaboration with Professor Samuel Berkovic and molecular geneticists, they identified the first epilepsy gene and many genes subsequently. She led the first major International League Against Epilepsy revision of the classification of epilepsies in 28 years (March 2017) and was a co-recipient of the Australian Prime Minister's Prize for Science and in 2018 was elected to the Royal Society (London).

Pratibha Singhi, MBBS, MD, FIAP, FNAMS

Former Head Dept of Pediatric Neurology, Amrita Hospital, Faridabad Former Director Pediatric Neurology, Medanta, The Medicity, Gurgaon. Former Head & Chief Pediatric Neurology, and Neurodevelopment Department of Pediatrics, APC, PGIMER, Chandigarh Consultant Rehabilitation Centre for Disabled children, Chandigarh Consultant Pediatric Neurologist–The Great Ormond Street Hospital, London, UK 2004, 2008

President International Child Neurology Association (ICNA)

Former National Delegate India- Asian Oceanian Association of Child Neurology (AOCNA)

Former National President Association of Child Neurology (AOCN)

Rated among the top 2% scientists in the World by Stanford University USA

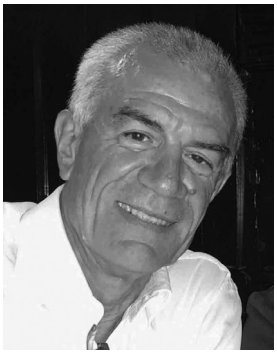
Published over 500 papers, 4 books.

Nicola Specchio, M.D, Ph.D, FRCP

Nicola Specchio is Chair of Neurology, Epilepsy and Movement Disorders Unit at Bambino Gesù Children's Hospital, and Director of Research Unit on Neurological and Neurosurgical Diseases in Rome, Italy, and Guest Professor in Pediatric Neurology at University of Leuven, Belgium.

My main interest lies with seizure semiology and the classification of epileptic seizures and syndromes, drug resistant epilepsies, developmental and epileptic encephalopathies. I published more than 280 papers in many international journals. I am also principal investigator on different clinical trials in patients with rare and complex epilepsies and Developmental and Epileptic encephalopathies.

I am recipient of the Young Investigator Award from the Commission of European Affair of the International League Against Epilepsy (2016) and of the "John Stobo Pritchard Award" from the International Child Neurology Association (ICNA) (2020). In 2021 I got the Ambassador for Epilepsy Award from the International Leagues Against Epilepsy.



Federico Vigevano, M.D.

Since 1978 to 2024 Neurologist at Children's Hospital Bambino Gesù where he held the position of head of Neuroscience DPT. Since February 2024 head of Developmental Disabilities DPT at San Raffaele Research Institute in Rome.

Research interest: Epileptic Encephalopathies and Genetic of Epilepsy, Video-EEG and Long-term monitoring in epilepsy, Pediatric Neurology, Neuro-Developmental Disabilities, Neurorehabilitation. He identified a clinical entity, currently called Self-Limited Infantile Epilepsy. Author or co-author more than 300 papers in the most important international journals.

President of the LICE (Italian League Against Epilepsy) from 1999 to 2002; Chair of the European Advisory Council of the ILAE from 2001 to 2005, Ambassador of Epilepsy by ILAE since 2001. He received the European Epileptology Award by the European ILAE Commission in Prague, 2016. Chair of the Scientific Committee of the International Congress on Epilepsy, Rome 2011, and Honorary President of the European Epilepsy Congress, Rome 2024. He is the organizer of the "International Course on Drug Resistant Epilepsy" (Tagliacozzo – Italy) and he is member of the board of European Pediatric Neurology Society.

Kazuhiro Yamakawa, Ph.D

1980 B.Sc. (March 1984) Molecular Biology, Faculty of Science, Kyoto University, Kyoto, Japan.

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1989 Researcher, Department of Biochemistry, Cancer Institute, Tokyo, Japan.

1994 Research fellow, Medical Genetics, Cedars-Sinai Medical Center, UCLA School of Medicine, USA.

1997 Laboratory Head, Laboratory for Neurogenetics, RIKEN Brain Science Institute, Saitama, Japan

2018 Laboratory Head, Laboratory for Neurogenetics, RIKEN Center for Brain Science, Saitama, Japan

2019 Professor, NDD Genetics, Nagoya City University Graduate School of Medical Sciences, Aichi, Japan

Anannit Visudtibhan, MD

Emeritus Professor (Pediatrics: Pediatric neurology)

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Medical Education & Training

1984 Doctor of Medicine, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand

1990 Diploma, Thai Board of Pediatrics, Medical Council of Thailand

1991 Diploma of Clinical Neurology, National Hospital of Neurology and Neurosurgery, University of London, London, England

1995 Certificate in Neurology & Pediatric Neurology State University of New York, Health Science Center at Brooklyn, New York 11203, USA

1996 Certificate in Neuromuscular disorders & Electromyography, State University of New York, Health Science Center at Brooklyn, New York 11203, USA

Oral Program

Day 1: March 27 (Friday)

10:00-12:00	**Pre Congress** **PANEL Discussion: From Bench to Bedside: Does Molecular Epilepsy Research Truly Change Clinical Care?**
Panelists:	**Sheffali GULATI** (Department of Pediatrics, AIIMS, New Delhi, India) **Anannit VISUDTIBHAN** (Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand)
Facilitators:	**Lakshmi NAGARAJAN** (Perth Children's Hospital. University of Westen Australia, Australia) **Kun-Long HUNG** (Department of Pediatrics, Fu-Jen Catholic University Hospital, Taiwan)
13:00-13:30	**Opening Ceremony**
13:30-14:15	**Opening Oration** **"Intractable and Rare Epilepsies in a Global Context: Implications for Care, Equity, and Collaboration"**
Chairs:	**Wang-Tso LEE** (National Taiwan University Hospital, Taiwan) **Shinichi HIROSE** (General Medical Research Center, School of Medicine, Fukuoka University, Japan)
Speaker:	**Pratibha SINGHI** (The International Child Neurology Association, Head Department of Pediatric Neurology Amrita Hospital Faridabad, India)
14:15-14:45	**Coffee Break**
14:45-15:30	**Plenary Lecture 1** **"A Comprehensive Genetic Analysis of Rare Neurological Disorders Including Epilepsy"**
Chairs:	**Ingrid SCHEFFER** (The University of Melbourne, Australia) **Shih-Hua LIN** (Department of Medicine, Tri-Service General Hospital, National Defense Medical University, Taiwan)
Speaker:	**Naomichi MATSUMOTO** (Department of Human Genetics, Yokohama City University Graduate School of Medicine, Japan)
15:30-16:15	**Plenary Lecture 2** **"Identification of Novel Genes in Rare Genetic Epilepsies"**
Chairs:	**Lung-Chang LIN** (Department of Pediatrics, Kaohsiung Medical University Hospital, Taiwan) **Xiu-Yu SHI** (People's Liberation Army General Hospital and Medical School, China)
Speaker:	**Yuwu JIANG** (Children's Medical Center, Pediatric Epilepsy Center, Peking University First Hospital, China)

Day 2: March 28 (Saturday)

09:30-10:15 **Plenary Lecture 3**

"Progressive Myoclonic Epilepsies: Lessons from Neuronal Ceroid Lipofuscinoses"

Chairs: **Yoshikatsu ETO** (Advanced Clinical Research Center & Asian LSD Center, Institute of Neurological disorders, Japan/ The Jikei University, Japan)

Federico VIGEVANO (Developmental Disabilities Dpt, IRCCS San Raffaele, Italy)

Speaker: **Nicola SPECCHIO** (Neurology, Epilepsy and Movement Disorders Unit, Director of Research Unit on Neurological and Neurosurgical Diseases, Bambino Gesù Children's Hospital, IRCCS, Italy)

10:15-11:00 **Plenary Lecture 4**

"Channelopathy during Early Brain Development"

Chairs: **Naomichi MATSUMOTO** (Department of Human Genetics, Yokohama City University Graduate School of Medicine, Japan)

Rei Cheng YANG (Department of Physiology, School of Medicine, Kaohsiung Medical University, Taiwan)

Speaker: **Jeffrey L. NOEBELS** (Baylor College of Medicine, USA)

11:10-11:55 **Plenary Lecture 5**

"Molecular pathomechanism of epileptic sodium channelopathy"

Chairs: **Lakshmi NAGARAJAN (**Perth Children's Hospital. University of Westen Australia, Australia)

Wang-Tso LEE (National Taiwan University Hospital, Taiwan)

Speaker: **Kazuhiro YAMAKAWA** (Department of Neurodevelopmental Disorder Genetics, Institute of Brain Science, Nagoya City University Graduate School of Medical Sciences, Japan)

12:00-13:00 **Special Luncheon Lecture**

"The genetics of the epilepsies: an ever expanding landscape"

Chairs: **Pratibha SINGHI** (The International Child Neurology Association, Head Department of Pediatric Neurology Amrita Hospital Faridabad, India)

Jeffrey L. NOEBELS (Baylor College of Medicine, Houston, USA)

Speaker: **Ingrid SCHEFFER** (The University of Melbourne, Australia)

13:15-14:00 **Plenary Lecture 6**

"KCNQ2's past is prologue: where we've come and may go"

Chairs: **Kazuhiro YAMAKAWA (**Department of Neurodevelopmental Disorder Genetics, Institute of Brain Science, Nagoya City University Graduate School of Medical Sciences, Japan)

Hideo YAMANOUCHI (Department of Pediatrics, Comprehensive Epilepsy Center, Saitama Medical University, Japan)

Speaker: **Edward C. COOPER** (Neurology, Neuroscience, and Molecular and Human Genetics, Baylor College of Medicine, USA)

14:00-14:45 **Plenary Lecture 7**

"GABAergic Pathway Dysfunction in Epilepsy: From Mechanisms to Therapeutic Rescue"

Chairs: **Naomichi MATSUMOTO (**Department of Human Genetics, Yokohama City University Graduate School of Medicine, Japan)

Yuwu JIANG (Children's Medical Center, Pediatric Epilepsy Center, Peking University First Hospital, China)

Speaker: **Katty (Jing-Qiong) KANG** (Department of Neurology & Pharmacology, School of Medicine, Vanderbilt University, Vanderbilt University Medical Center, USA)

14:45-15:15 **Coffee Break**

15:15-16:00 **Plenary Lecture 8**

"Molecular therapy for channelopathy"

Chairs: **Heung Dong KIM** (Department of Pediatrics, Samsung Kangbuk Hospital, Sungkyunkwan University College of Medicine, Korea)

Su-Kyeong HWANG (Astrogen Inc., Korea)

Speaker: **Hoon-Chul KANG** (Department of Pediatrics, Severance Children's Hospital, Yonsei University College of Medicine, Korea)

16:00-16:45 **Joint Plenary session**

"New Insight into Therapeutic Strategies for Intractable Epilepsy: Current Status and Future Perspectives for Precision Medicine"

Chairs: **Hoon-Chul KANG** (Department of Pediatrics, Severance Children's Hospital, Yonsei University College of Medicine, Korea)

Huei-Shyon WANG (Linkou Chang-Gung Memorial Hospital, Taiwan)

Speakers: **Ara KO**（Yonsei University College of Medicine, Severance Children's Hospital, Department of Pediatric Neurology, Department of Pediatrics, Korea）

Heung Dong KIM (Department of Pediatrics, Samsung Kangbuk Hospital, Sungkyunkwan University College of Medicine, Korea)

16:45-17:30 **Plenary Lecture 9**

"The Holistic Approach to Rare and Intractable Epilepsies"

Chairs: **Makiko OSAWA** (St. Margaret's School, Japan)

Nicola SPECCHIO (Bambino Gesù Children's Hospital, Institute for Research and Health Care (IRCCS), Italy)

Speaker: **Federico VIGEVANO** (Developmental Disabilities Dpt, IRCCS San Raffaele, Italy)

Day 3: March 29 (Sunday)

09:30-10:15	**Plenary Lecture 10** **"Integrating Molecular Genetics with Electroclinical Diagnosis and Management in Epilepsy: Challenges and Future Directions"**
Chairs:	**Kaiping CHANG** (Department of Pediatrics, Taipei Veterans General Hospital, Taiwan / Wei Gong Memorial Hospital, Taiwan) **Jiwen WANG** (Department of Neurology, Shanghai Children's Medical Center, Shanghai Jiaotong University School of Medicine, China)
Speaker:	**Lakshmi NAGARAJAN** (Dept of Neurology, Perth Children's Hospital/ University of Westen Australia Medical School, Australia)
10:15-11:00	**Closing Oration** **"Building International Connections in Asia: What the Azalea Festival Symposium and Epilepsy Genetics Have Taught Us"**
Chairs:	**Shinichi HIROSE** General Medical Research Center, School of Medicine, Fukuoka University, Japan) **Shyi-Jou CHEN** (Department of Pediatrics, Tri-Service General Hospital, School of medicine, National Defense Medical University, Taiwan)
Speaker:	**Wang-Tso LEE** (National Taiwan University Hospital Taipei, Taiwan)
11:00-11:20	**Closing Ceremony**

Poster Program: Basic Research

BP1 SCN1A mutations alter the biophysical properties of the ion-conducting pore

Chi Hua Cheng [1], Hueng-Chuen Fan [1]

[1)]Tungs' Taichung MetroHarbor Hospital, Taiwan

BP2 Interactions between voltage-gated sodium channel Nav1.1 and fibroblast growth factor FGF12

Ikuo Ogiwara [1], Chengzhu Yin [1], Atsushi Shimohata [1], Mie Gangi [1], Makoto Kaneda [1], Daisuke Kato[1]

[1)]Nippon Medical School, Japan

BP3 KCNQ2 mutations cause distinct phenotypes: functional differences and potential Kv7.2 modulating drugs

Inn-Chi Lee [1], Shi-Bing Yang [2]

[1)]Division of Pediatric Neurology, Department of Pediatrics, Chung Shan Medical University Hospital, Taichung, Taiwan, [2)]Institute of Biomedical Sciences, Academia Sinica, Taipei, Taiwan

BP4 Novel proteolytic post-translational modification in voltage-gated potassium channel KCNQ2

Yuichi Kimura [1], Hidehiko Uchiyama [1], Koji Masuda [1], Shinichi Hirose [2]

[1)]Department of Animal Science, Tokyo University of Agriculture, Japan, [2)]General Medical Research Center, School of Medicine, Fukuoka University, Japan

BP5 Megalencephaly-causing pathogenic AKT3 activation drives neuronal hyperexcitability via mTORC1: therapeutic rescue by rapamycin

Yosuke Miyamoto [1], Eisuke Ichise [1], Tomohiro Chiyonobu [1,2]

[1)]Department of Pediatrics, Kyoto Prefectural University of Medicine, Japan, [2)]Department of Molecular Diagnostics and Therapeutics, Kyoto Prefectural University of Medicine, Japan

BP6 Therapeutic Effects of Probiotics on Postnatal Seizure Susceptibility After Premature Brain Injury

Ming-Jung Tu [2], Yi-Fang Tu [1], Jia-Shing Chen [3]

[1)]National Chang Kung University, Taiwan, [2)]Chung Shan Medical University,[3)]I-Shou University, Taiwan

BP7 Developing phenotypic and polygenic scores to improve diagnosis of ADHD and related comorbidities in the Han Taiwanese population

I-Ching Chou [1,2], Yu-Tzu Chang [1,3], Ying-Ju Lin [2,4], Ting-Yuan Liu [5], Sheng-Shing Lin [1,3], Syuan-Yu Hong[1,6], Chien-Heng Lin[7,8], Fuu-Jen Tsai[2,4,5,9]

[1)]Department of Pediatric Neurology, China Medical University Children's Hospital, Taichung, Taiwan, [2)]School of Chinese Medicine, China Medical University, Taichung, Taiwan,[3)]School of Post Baccalaureate Chinese Medicine, China Medical University, Taichung, Taiwan,[4)]Genetic Center, Department of Medical Research, China Medical University Hospital, Taichung, Taiwan,[5)]Department of Medical Research, China Medical University Hospital, Taichung, Taiwan,[6)]Department of Medicine, School of Medicine, China Medical University, Taichung, Taiwan,[7)]Department of Pediatric Pulmonology, China Medical University Children's Hospital, Taichung, Taiwan,[8)]Department of Biomedical Imaging and Radiological Science, College of Medicine, China Medial University, Taichung, Taiwan,[9)]Department of Medical Genetics, Pediatric Endocrinology and Metabolism, China Medical University Children's Hospital, Taichung, Taiwan

Poster Program: Clinical Research

CP1 Classification and outcomes of genetic epilepsy in Taiwan – A tertiary center experience

Kuan-Ting Yeh [1], Ju-Yin Hou [1], Cheng-Yen Kuo [1],Yi-Hsuan Liu [1], Jainn-Jim Lin [2,3], Meng-Ying Hsieh[1,2], Yi-Ting Cheng[1], Huei-Shyong Wang[1,2], I-Jun Chou[1,2,], Meng-Han Tsai[2,4], Lin Kuang-Lin[1,2]

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CP2 Clinical and Genetic Spectrum of Pediatric Epilepsy–Movement Disorder Syndromes: A Single-Center Cohort of 56 Patients

Meng-Fan Tai [1], Lee-Chin Wong [1], Wang-Tso Lee [1]

[1)]National Taiwan University Children's Hospital, Taiwan

CP3 The effects of sleep apnea on risks of sudden cardiac arrest in children with epilepsy

Po Ming Wu [1,2], Pei-Chun Lai [1,3], Yi-Fang Tu [1]

[1)]Department of Pediatrics, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Taiwan, [2)]Institute of Clinical Medicine, College of Medicine, National Cheng Kung University, Taiwan, [3)]Education Center, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Taiwan

CP4 The Correlation and Potential Mechanisms Between Neonatal Jaundice and Attention-Deficit/Hyperactivity Disorder and Learning Disabilities

Hsi Chang [1,2], Shih-ming Weng [3,4], Yi-Wei Kao [5], Feng-Ching Li [2], Ming-Lan Tsai [1,2]

[1)]Taipei Medical University, Taipei, Taiwan, [2)]Taipei Medical University Hospital, Taipei, Taiwan,[3)]Department of Speech and Hearing Sciences, National Taipei University of Nursing and Health Sciences, Taipei, Taiwan,[4)]Department of Pediatrics, Wan Fang Hospital, Taipei Medical University, Taipei, Taiwan,[5)]Department of Applied Statistics and Data Science, Ming Chuan University, Taipei, Taiwan

CP5 EEG-Based Classification For Tic Disorders via Complementary Deep Model Integration

Shi-Bing Wong [1], Syu-Siang Wang [2], Chao-Hsiang Hung [2]

[1)]Department of Pediatrrics, Taipei Tzu Chi Hospital, New Taipei City, Taiwan, [2)]Department of Electrical Engineering, Yuan Ze University, Taoyuan, Taiwan

CP6 GMP-Grade Umbilical MSC Secretome in Paediatric Drug-Resistant Epilepsy: Matched Case–Control Evidence of Seizure, Functional, and IL-6/hs-CRP Biomarker Shifts

Dian Kesumapramudya Nurputra [1], Andika Priamas Nugrahanto [1], Agung Triono [1],Elisabeth Siti Herini [1]

[1)]Universitas Gadjah Mada, Indonesia

CP7　Epilepsy with CDK19 variants: clinical manifestations and outcome

Yen-Ting Chen [1], Wang-Tso Lee [2]

[1)]Department of Pediatrics, National Taiwan University Children's Hospital, Taiwan, [2)]Department of Pediatric Neurology, National Taiwan University Children's Hospital, Taiwan

CP8　Epileptic phenotype in a patient with a MARK2 variant: the first detailed description and review of the literature

Young Ok Kim [1]

[1)]Chonnam National University Medical School, Chonnam National University Children's Hospital, Korea

CP9　Severe Neonatal Encephalopathy in a Boy Associated with a Novel De Novo MECP2 Nonsense Variant

Young Mi Kim [1], Yun Hee Jo [1]

[1)]Pusan National University Hospital, Korea

CP10　Perampanel Treatment in CELF2-Related Developmental and Epileptic Encephalopathy: A Case With Partial Respons

Lee Chin Wong [1], Wang-Tso Lee [1]

[1)]National Taiwan University Hospital, Taiwan

CP11　Expanding the Phenotypic Spectrum of KCNK4-Related Developmental and Epileptic Encephalopathy: A Case Report Without Classical FHEIG Features

Adlina Awanis Mamat [1], Ahmad Rithauddin Mohamed [1], Sumitha Murugesu [1], Yan Lian Teo [1], Dianah Abd Hadi [1], Haslina Hashim[2]

[1)]Hospital Tunku Azizah, Malaysia, [2)]Institut Pediatrik Hospital Kuala Lumpur, Malaysia

CP12　Clinical features of HNRNPU deletion syndrome

Shin Nabatame [1], Tomoya Yano [1], Kanami Maegawa [1]

[1)]Department of Paediatrics, National Hospital Organization, National Osaka Hospital, Japan

CP13　A Case of ACTL6B-Related Disorder Presenting with Developmental and Epileptic Encephalopathy, Responsive to Corpus Callosotomy

Konosuke Watanabe [1], Hitomi Hayashi [1], Koshiro Fujikawa [1], Hiromi Yamaguchi [1], Takako Fujita [1], Takahito Inoue[1], Fuyuki Miya[2], Mitsuhiro Kato[3], Shinichiro Nagamitsu[1], Shinichi Hirose[4]

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CP14　Navigating Failure to Thrive, Refractory Epilepsy, Global Developmental Delay and Movement Disorder in a 2-Year-Old Male: The Diagnostic Significance of Extended 15q11.2-q13.2 Microdeletion

Tzu-Hung Cheng [1], Shyi-Jou Chen [1], Chih-Fen Hu [1]

[1)]Tri-service General Hospital, Taiwan

CP15 Functional Evaluation and Mechanism-Based Rescue of a Novel Variant (A305V) In GABA Transporter 1-encoding SLC6A1 Associated With Myoclonic Atonic Epilepsy

Aiden Delahanty [1], Debbie Song [1], Juexin Wang [1],Wangzhen Shen [1], Melissa Bassett [1], Jing-Qiong Kang[1]
[1)]Vanderbilt University Medical Center, Department of Neurology, USA

CP16 Successful Treatment of Dravet Syndrome with Camellia Oil: A Case Report

Wen-Ling Yeh [1], Hueng-Chuen Fan [2], Chih-I Hung [3]
[1)]Tungs' Taichung MetroHarbor Hospital, Department of Nutrition Therapy, Taiwan, [2)]Tungs' Taichung Meroharbor Hospital, Department of Pediatrics, Taiwan,[3)]Tungs' Taichung Meroharbor Hospital, Head Nurse of Nursing Department, Taiwan

CP17 SCN1A Mutation Location Predicts Stiripentol Efficacy in Dravet Syndrome

Hueng-Chuen Fan [1], Chi-Hua Cheng [1]
[1)]Department of Pediatrics, Department of Medical research, Tungs' Taichung Metroharbor Hospital, Taiwan

CP18 Experience of Using Ketogenic Therapy with Camellia Oil in the Treatment of Infantile Spasms: A Case Series of 5 Patients

Po-Cheng Chen [1], Win-Lin Yeh [2], Hueng-Chuen Fan [1]
[1)]Department of Pediatrics, Tungs' Taichung MetroHarbor Hospital, Taichung City, Taiwan, [2)]Department of Nutrition Therapy, Tungs' Taichung MetroHarbor Hospital, Taichung City, Taiwan

CP19 ALG13-Related Congenital Disorder of Glycosylation Presenting as Infantile Epileptic Spasms Syndrome

Hsiao-Ling Liu [1], Wen-Chin Weng [1]
[1)]National Taiwan University Children's Hospital, Taiwan

CP20 Epileptic Spasms Occurred Earlier In JE Induced Anti-Nmdar Encephalitis

Aye Mya Min Aye [1], Aye Mu Saan [1], Khine Mi Mi Ko [1]
[1)]Yangon Children's Hospital, Myanmar

CP21 Case Series of New-Onset Refractory Status Epilepticus

Ei Sandy Kyaw [1], Aye Mya Min Aye [1], Aye Mu Saan [1], Khine Mi Mi Ko [1], Soe Soe Maw [1]
[1)]Yangon Children's Hospital, Myanmar

CP22 A novel frameshift DMD mutation (p.Leu2017Profs5) disrupts dystrophin spectrin-like repeats and destabilizes protein conformation: structural insights from AlphaFold-based modeling

Chih-Fen Hu [1], Yu-Chin Lin [1], Yu-Yang Lu [1],Shyi-Jou Chen [1]
[1)]Tri-Service General Hospital, Taiwan

Poster Abstracts

BP1

SCN1A mutations alter the biophysical properties of the ion-conducting pore

Chi Hua Cheng[1], Hueng-Chuen Fan[1]
[1]Tungs' Taichung MetroHarbor Hospital

Purpose: SCN1A encodes the Nav1.1 voltage-gated sodium channel, with mutations causing epilepsy phenotypes ranging from mild to severe. Understanding how specific mutations affect channel structure and function is crucial for predicting clinical outcomes. This study investigates the relationship between structural cavity alterations and ligand-binding affinity changes across SCN1A functional domains to elucidate molecular mechanisms underlying phenotypic variability.
Methods: SCN1A variants were classified into good function (GF) and poor function (PF) groups based on clinical phenotypes. Computational analysis quantified cavity volume changes (ΔCavity) in the ion-conducting pore. Molecular docking assessed binding affinity alterations (ΔVina scores) for each variant. Correlation analyses between ΔCavity and ΔVina were performed for both groups. Mutations were mapped across five functional domains (N-terminus, Domains I-IV, C-terminus) to identify domain-specific patterns.
Results: 100 SCN1A variants were collected from internet. The GF group showed weak negative correlation between ΔCavity and ΔVina ($R^2 \approx 0.09$), indicating moderate structural changes minimally affect ligand-binding affinity. The PF group demonstrated stronger negative correlation ($R^2 \approx 0.44$), showing severe cavity distortions substantially reduce binding affinity and cause disruptive structural remodeling. Domain analysis revealed striking differences: PF variants concentrated in Domain I (>50%), while GF variants distributed evenly with peak in Domain III (~27%), identifying Domain I as critical for channel function. Two distinct pathological mechanisms emerged: pore enlargement mutations increased cavity volume, enabling aberrant ion permeation and destabilizing gating; pore constriction mutations reduced cavity volume, impairing conductance and decreasing opening probability.
Conclusion: SCN1A mutations alter ion-conducting pore properties through domain-specific mechanisms. Domain I mutations are particularly disruptive, causing constricted cavities and impaired conduction, while Domain III mutations retain partial stability with milder consequences. The correlation between cavity alterations and binding affinity provides a quantitative framework for predicting mutation severity with applications in clinical risk stratification and personalized treatment for SCN1A-related epilepsies.

BP2

Interactions between voltage-gated sodium channel Nav1.1 and fibroblast growth factor FGF12

Ikuo Ogiwara[1], Chengzhu Yin[1], Atsushi Shimohata[1], Mie Gangi[1], Makoto Kaneda[1], Daisuke Kato[1]
[1]Nippon Medical School

Voltage-gated sodium channels underlie electrical signaling in neurons. Mutations in the SCN1A gene encoding a brain types of sodium channel, Nav1.1, are associated with an infantile epileptic disorder, Dravet syndrome, characterized by intractable recurrent seizures and neurodevelopmental delay. Loss of Nav1.1 function causes repetitive firing impairments in GABAergic inhibitory cells. Fibroblast growth factors, FGF11-14, are involved in neural excitability by associating with voltage-gated sodium channels. A recurrent mutation in the gene encoding FGF12 is associated with infantile epilepsy. We here examined whether the disease-associated FGF12 mutant affects electrophysiological properties of Nav1.1 co-expressed in 293T cell. We first examined interactions between Nav1.1 and wild-type FGF12, and found that there were no significant differences in steady-state activation, steady-state voltage dependence of inactivation and recovery from inactivated state between the cells expressing Nav1.1 alone and those co-expressing Nav1.1 and wild-type FGF12. We next tested interactions between Nav1.1 and disease-associated FGF12 mutant. We found no significant differences in steady-state activation and steady-state voltage dependence of inactivation between the cells co-expressing Nav1.1 and wild-type FGF12 and those co-expressing Nav1.1 and mutated FGF12. We however found that the mutated FGF12 slowed recovery from inactivated state of Nav1.1, compared to wild-type FGF12. These observations suggested that the pathophysiology of FGF12-assciated epileptic disorder might involve altered Nav1.1 function, implicating the pathophysiology of the infantile epilepsies.

BP3

KCNQ2 mutations cause distinct phenotypes: functional differences and potential Kv7.2 modulating drugs

Inn-Chi Lee[1], Shi-Bing Yang[2]
[1]Division of Pediatric Neurology, Department of Pediatrics, Chung Shan Medical University Hospital, Taichung, Taiwan,
[2]Institute of Biomedical Sciences, Academia Sinica, Taipei, Taiwan

Background: Pediatric epilepsy caused by KCNQ2 mutations can manifest as self-limiting familial neonatal epilepsy (SLFNE) or neonatal-onset developmental epileptic encephalopathy (DEE). Patients may exhibit varying degrees of neurodevelopmental disabilities; however, effective treatment for cognitive dysfunction in patients with KCNQ2 mutations associated DEE (KCNQ2 DEE) are lacking.
Methods: Two patients with KCNQ2 c.635A>G (p.Asp212Gly) and c.902G>A (p.Gly301Asp) mutations presented with SLFNE and DEE, respectively. We investigated the phenotypes associated with the changes functional currents and KCNQ2 protein expression in HEK293 cells. Furthermore, we explored drugs capable opening damaged Kv7.2 channels.
Results: Neurodevelopmental outcomes were more severe in patients with D212E mutations than in those with D212G mutations. A significant loss of function was observed in mutated homomeric channels in both variants. The Kv7.2 conduction curves of the heteromeric KCNQ2+KCNQ3 variant channel in D212G were superior to those in D212E. Heteromeric KCNQ2+KCNQ3+D212G cells exhibited a left-shift of 7.6 mV compared to the heteromeric KCNQ2+KCNQ3+D212E. Phenotypic differences correlated with the half-maximal activation voltage in the heteromeric channels, indicating that D212G opens the channel faster than D212E. Protein expression on the cell membranes of the two mutations and the wild-type KCNQ2 were not significantly different. Retigabine, capable of opening damaged Kv7.2 channels, can improve Kv7.2 conductions in both the wild-type and D212E mutation variants associated with DEE.
Conclusions: Current changes were more significant in cells with homomeric transfections of D212E and D212G than in the wild-type KCNQ2. Our findings suggest that homomeric current changes are common in KCNQ2 DEE and SLFNE; however, the heteromeric characteristics of conduction correlate with long-term neurodevelopmental outcomes. Retigabine can modulate Kv7.2 currents in D212E and wild-type cells, particularly in D212G associated with SLFNE.

BP4

Novel proteolytic post-translational modification in voltage-gated potassium channel KCNQ2

Yuichi Kimura[1], Hidehiko Uchiyama[1], Koji Masuda1, Shinichi Hirose[2]
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KCNQ2 is a member of the voltage-gated potassium (Kv) channel family and regulates neuronal activity through potassium ion efflux. Pathogenic variants of KCNQ2 lead to aberrant neuronal activity and cause two types of epilepsy: self-limited familial neonatal epilepsy (SLFNE) and developmental and epileptic encephalopathies (DEE). However, how these pathogenic variants influence KCNQ2 expression remains unclear. Here we show a short isoform of mouse KCNQ2 ($KCNQ2^S$), whose expression levels differed significantly across variants compared with wild type, whereas those of full-length KCNQ2 ($KCNQ2^F$) remained unchanged. Of particular interest, two variants at the same codon, Y284C and Y284D, which are associated with distinct clinical phenotypes—self-limited familial neonatal epilepsy (SLFNE) and developmental and epileptic encephalopathy (DEE), respectively—exerted opposite effects on the short isoform: Y284C increased the level of $KCNQ2^S$, whereas Y284D decreased it compared with the wild type. As $KCNQ2^S$ was found to be localized in the plasma membrane, it is suggested that $KCNQ2^S$ is a post-translational product resulting from a cleavage of full-length KCNQ2. This novel post-translational cleavage generating KCNQ2}^{S} was observed in neuronal cells and appears to be evolutionarily conserved. Although the role of this post-translational modification in epilepsy remains unknown, it may be elucidated through future studies.

BP5

Megalencephaly-causing pathogenic AKT3 activation drives neuronal hyperexcitability via mTORC1: therapeutic rescue by rapamycin

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Rationale: AKT3, a key component of the PI3K–AKT–mTOR pathway, is highly expressed in the brain, and activating variants cause megalencephaly and cortical malformations frequently associated with intractable epilepsy. However, no disease-modifying therapy exists, and symptomatic treatments are often insufficient. Although the AKT inhibitor capivasertib, developed as an anticancer agent, could theoretically provide targeted therapy, long-term administration from infancy is impractical due to adverse effects. Therefore, identification of safer downstream therapeutic targets is required.
Methods: We identified a heterozygous activating variant in AKT3, NM_005465.7:c.233A>G, p.(Q78R), in a patient who exhibited megalencephaly and cortical malformations. During infancy, the patient exhibited severe developmental and epileptic encephalopathy. Induced pluripotent stem cell (iPSC) lines were generated from peripheral blood mononuclear cells of the patient. Isogenic control lines were established using CRISPR/Cas9 genome editing. These iPSCs were differentiated into glutamatergic neurons by Neurogenin 2 induction protocol. Phosphorylation of S6, a marker of mTORC1 activity, was assessed by western blotting at 4 weeks after neuronal differentiation, and soma size was evaluated by immunostaining. RNA sequencing of neurons at week 5 was performed to assess differentiation and maturation status. Spontaneous electrophysiological activity was recorded weekly at 4–6 weeks after differentiation using a microelectrode array. From week 4, neurons were treated with capivasertib (0.1, 0.3, or 1 μmol/L) or rapamycin (50, 100, or 300 nmol/L) for 2 weeks. Cell viability following drug treatment was evaluated using a WST-8 assay.
Results: Patient-derived neurons exhibited significantly enlarged somas and increased phospho-S6 levels compared with isogenic controls. RNA-seq analysis demonstrated comparable neuronal differentiation and maturation states between patient-derived and isogenic control neurons. At 5 weeks after differentiation, patient-derived neurons showed a significant increase in burst frequency. By week 6, total spike counts and firing rates per active electrode were also elevated. Treatment with either capivasertib or rapamycin suppressed the increased numbers of total spikes and bursts. Rapamycin had a smaller impact on cell viability compared with capivasertib.
Conclusions: Patient-derived glutamatergic neurons carrying an activating AKT3 variant exhibit soma hypertrophy, mTORC1 hyperactivation, and enhanced spontaneous neuronal activity. Both AKT inhibition and mTORC1 inhibition attenuated abnormal neuronal firing; however, rapamycin showed fewer adverse effects on cell viability, suggesting that mTORC1 inhibition may represent a safer and promising targeted therapeutic strategy for epilepsy associated with activating AKT3 variants.

BP6

Therapeutic Effects of Probiotics on Postnatal Seizure Susceptibility After Premature Brain Injury

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Purpose: Postnatal epilepsy frequently arises as a sequela of early-life brain injury in premature infants. Emerging evidence suggests that probiotics may ameliorate microbial dysbiosis and attenuate systemic inflammatory responses. These properties indicate a potential therapeutic role for probiotics in mitigating the susceptibility to epilepsy following premature brain injury (PBI).
Methods: PBI was modeled by inducing hypoxic-ischemic insult and administering lipopolysaccharide to rat pups on postnatal day 5. Seizure activity following the insult was monitored using electroencephalography. Histological analyses were performed to evaluate alterations in GABAergic neurons and mossy fiber sprouting (MFS), both prior to and following probiotic intervention. Neuroinflammatory responses were assessed by quantifying Iba-1-positive microglial activation in the cerebral cortex and measuring associated cytokine levels in cerebrospinal fluid (CSF). Additionally, fecal microbiota composition and concentrations of short-chain fatty acids were analyzed in PBI-exposed rats with and without probiotic treatment.
Results: PBI rats did not have significant ischemic brain damage but had a reduction of myelin expression. Between postnatal days 85 and 90, PBI rats showed increased electrographic seizure activity and MFS. Histological analysis revealed reduced cortical GABAergic neurons, notably glutamic acid decarboxylase (GAD)-positive and somatostatin-positive subtypes, while parvalbumin-positive neurons remained unchanged. Probiotic treatment significantly reduced electrographic seizure frequency and duration, decreased MFS, and restored GAD-positive neuron density. Fecal microbiome analysis showed increased Lactobacillaceae abundance and altered fecal microbial composition, with elevated butyrate levels in both feces and CSF, although alpha diversity, acetate, and propionate levels remained unchanged. Neuroinflammation was alleviated by probiotics, as evidenced by reduced Iba-1-positive microglial activation and lower CSF levels of TNF-α.
Conclusion: Elevated neuroinflammation and the loss of GABAergic neurons may underlie the heightened seizure susceptibility observed in immature brains following early-life brain injury. Probiotic intervention has the potential to attenuate seizure vulnerability and neuroinflammatory responses in premature brain injury by modulating gut microbiota composition, enhancing short-chain fatty acid production, and restoring inhibitory neuronal circuitry.

BP7

Developing phenotypic and polygenic scores to improve diagnosis of ADHD and related comorbidities in the Han Taiwanese population.

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Objective: Given the underexplored genetic architecture and associated health outcomes of attention-deficit/hyperactivity disorder (ADHD) among Han Taiwanese populations, this study utilized electronic health records (EHR) and genome-wide association studies (GWAS) to identify genetic loci, health conditions, and to establish phenotypic and polygenic predictors for clinical diagnosis.

Methods: We utilized electronic health records (EHR) to identify conditions associated with ADHD and developed disease phenotype risk scores (Disease PheRS) validated in an independent clinical cohort. Additionally, a genome-wide association study (GWAS) was performed to establish ADHD polygenic risk scores (PRS) for diagnostic prediction in the Han Taiwanese population.

Results: We included 664 ADHD patients and 25,771 controls from the CMUH database, finding that ADHD patients were younger, predominantly male, had more comorbidities, and showed higher usage of anti-anxiety medications. An ADHD status-PheWAS revealed multiple comorbidities of mental disorders and neurological conditions, highlighting ADHD's complex clinical presentation. An ADHD PRS-PheWAS showed that higher polygenic risk for ADHD was strongly linked to ADHD diagnosis itself and nominally associated with pervasive developmental disorders (PDDs). Finally, clinically validated ADHD and PDDs cases displayed elevated phenotype risk scores (PheRS) and polygenic risk scores (PRS). Integrating disease status-PheRS with PRS enhanced the accuracy of ADHD and PDDs clinical diagnosis predictions in the Han Taiwanese population.

Conclusion: These findings indicate that combining phenotypic (PheRS) and genetic (PRS) predictors derived from EHR and GWAS data effectively captures their value as quantitative tools for clustering ADHD and related comorbidities, thereby enhancing clinical prediction.

CP1

Classification and outcomes of genetic epilepsy in Taiwan – A tertiary center experience

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Purpose: The genetic factor is one of the crucial causes of epilepsy. Even with the same gene dysfunction, the clinical features can have a diverse spectrum. To expand the clinical features and Taiwan's experience, our study aimed at knowing the clinical phenotypes, clinical course, and prognosis of genetic epilepsy patients in Taiwan.
Methods: We enrolled patients with epilepsy who ever received a genetic exam in our hospital in a 3-years period. Information was collected by chart review. We also reviewed genetic testing reports on medical records. The potential pathogenicity of mutations was analyzed according to the guidelines from the American College of Medical Genetics and Genomics (ACMG).
Results: 250 epilepsy patients received a genetic exam. 77 patients (47 female) were diagnosed as genetic epilepsy, including 68 monogenic epilepsy and 9 copy number variants involving more than one gene. 28 were fever-sensitive epilepsy. The median age of seizure onset was 8 months (0.1-156 months). 66 (85.7%) had developmental delay (DD), and 41/46 (89.1%) had intellectual disability (ID). 74 % abnormal EEG and 44 % abnormal MRI were noted, respectively. 75 % were pharmaco-resistant, and 20 % ever received epilepsy surgery and/or neuromodulations.
Of the 68 patients with molecular diagnoses of monogenic epilepsy, the presumed modes of inheritance included 68% autosomal dominant, 16% autosomal recessive, 1% X-linked dominant, 6% X-linked recessive, and 9% X-linked mode of inheritance sparing transmitting males. They were further classified into six different groups according to function of genes: ion channels (n=30), regulators for DNA transcription and translation (n=6), cell membrane proteins including transporters and receptors (n=7), cytoskeleton and related structural protein (n=11), adhesion molecules (n=7), mitochondria stability and oxidative stress (n=7). Genetic variants involving mitochondria stability and oxidative stress had earlier seizure onset, with a median onset of one-month-old.
Among the 28 patients with fever-sensitive epilepsies, 22 (78.6%) patients had *SCN1A* mutations and presented with a younger age at seizure onset when compared with *PCDH19* mutations (median: 6.5 *vs.* 15.5 months, p =0.005). Status epilepticus was more common in patients with *SCN1A* mutations (72.7% *vs.* 16.7%, p =0.013).
Conclusions: In this study, patients with genetic epilepsy were found to have early seizure onset and high association with DD/ID. However, variable clinical presentations were noted in different functions of genes, different variants, different locations of the same gene, and intrafamilial variable phenotype. Genetic testing helps to guide clinical management and explore the genotype-phenotype relationships. Further studies are necessary.

CP2

Clinical and Genetic Spectrum of Pediatric Epilepsy–Movement Disorder Syndromes: A Single-Center Cohort of 56 Patients

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Purpose: Epilepsy–movement disorder syndromes (EPIMDs) comprise a heterogeneous group of neurogenetic conditions characterized by the coexistence of seizures and abnormal movements. Despite insights from large multinational cohorts, region-specific data from Asia remain underrepresented. We aimed to characterize the clinical, developmental, and genetic spectrum of EPIMDs in a single tertiary pediatric hospital.
Methods: We conducted a retrospective study (2015–2025) at a single tertiary pediatric center, including children with childhood-onset epilepsy and coexisting movement disorders who harbored pathogenic or likely pathogenic variants identified through genetic testing. Clinical data included seizure and movement phenomenology, developmental outcomes, neuroimaging findings, and treatment response.
Results: Fifty-six patients were included with a mean follow-up of 7.95 years. Seizure onset occurred during infancy in 46%, and seizures preceded movement disorders in 64%. Focal seizures (48%) and epileptic spasms (25%) were most common; 10% experienced status epilepticus and 26% met criteria for drug-resistant epilepsy. Dystonia (53%), chorea (45%), and ataxia (30%) predominated, with 59% exhibiting mixed movement phenomenology and 64% showing persistent symptoms. Pathogenic variants were identified in 39 genes, most frequently PRRT2 (14.5%), FOXG1 (9.1%), and TBC1D24 (7.3%). Developmental delay was present in 75% (severe in 50%). Brain MRI abnormalities were detected in 89%, most commonly cortical malformations (61%) and corpus callosum thinning (48%). Genetic diagnosis led to treatment modification in 52% of cases.
Conclusions: This single-center cohort demonstrates substantial genetic and phenotypic heterogeneity in pediatric EPIMDs. Early-onset epilepsy frequently precedes complex movement phenotypes and contributes to significant neurodevelopmental burden. Compared with recent multinational cohorts, our study showed a lower proportion of drug-resistant epilepsy and a higher representation of PRRT2-related disorders, potentially reflecting population-related genetic patterns or regional referral characteristics. The high rate of treatment modification following genetic diagnosis underscores the clinical utility of early genomic evaluation and supports its role in precision care for epilepsy–movement disorder syndromes.

CP3

The effects of sleep apnea on risks of sudden cardiac arrest in children with epilepsy

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Purpose: Sleep apnea (SA) is a known risk factor for sudden cardiac death (SCD). However, the association between SA and SCD specifically in pediatric epilepsy patients remains understudied. Furthermore, little is known about the impact of SA subtypes—beyond obstructive sleep apnea—on this risk. This study investigated the incidence and risk of SCD in pediatric epilepsy patients with SA and evaluated the impact of different SA phenotypes.
Methods: We utilized the TriNetX Analytics network, accessing electronic health records from 108 health care organizations (HCOs). We identified a cohort of pediatric patients (age <18 years) diagnosed with epilepsy. We conducted a retrospective cohort study comparing epilepsy patients with SA to a propensity-score matched cohort of epilepsy patients without SA to control for confounders. The primary outcome was Cardiac Arrest (serving as a proxy for SCD). We calculated Hazard Ratios (HRs) for the incidence of cardiac arrest over a 10-year follow-up period.
Results: A total of 359,127 pediatric epilepsy patients were analyzed. The incidence of SCD was significantly elevated in epilepsy patients with SA (43.2 per 10,000 person-years) compared to those without. After propensity matching, the epilepsy-with-SA cohort showed a significantly increased risk of SCD compared to the epilepsy-without-SA cohort (HR: 1.913; 95% CI: 1.532–2.387; P<0.001). Subgroup analysis revealed that the risk was highest in patients with mixed SA (HR: 4.152; 95% CI: 2.083–8.275) and in individuals older than 8 years (HR: 2.804; 95% CI: 1.982–3.967).
Conclusions: This study demonstrates that comorbid SA significantly increases the risk of SCD in pediatric epilepsy patients. notably, mixed SA posed the highest risk, suggesting that the addition of a central respiratory deficit may synergistically enhance SCD susceptibility in this population. These results underscore the urgent need for early detection and aggressive management of sleep apnea—particularly mixed types—in children with epilepsy.

CP4

The Correlation and Potential Mechanisms Between Neonatal Jaundice and Attention-Deficit/Hyperactivity Disorder and Learning Disabilities

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Background: Neonatal jaundice is a common condition caused by elevated serum bilirubin levels. Current treatment guidelines are primarily designed to prevent kernicterus, an overt and severe form of bilirubin-induced brain injury. However, how these thresholds were established remains unclear, and potential risks of subtle or “minimal" brain injury have been largely overlooked. We hypothesized that even moderate neonatal hyperbilirubinemia may be associated with long-term neurodevelopmental dysfunction, including attention-deficit/hyperactivity disorder (ADHD) and learning disabilities.
Methods: A nationwide population-based cohort study was conducted using Taiwan's Longitudinal Health Insurance Database (LHID) from 2000 to 2012. Individuals were classified based on neonatal jaundice diagnosis (ICD-10: P59) and further stratified into hospitalized (requiring phototherapy or medical treatment) and non-hospitalized groups. Cox proportional hazards models were applied to estimate adjusted hazard ratios for ADHD and learning disabilities, with Kaplan–Meier analysis used to assess cumulative incidence.
To explore biological mechanisms, in vitro experiments were performed using SH-SY5Y neuronal cells and SVGp12 astrocytes exposed to clinically relevant bilirubin concentrations. Neuronal differentiation, neurite outgrowth, synapse formation (SV2A, α7-AChR), and cell proliferation were evaluated using immunofluorescence, Sholl analysis, and quantitative image analysis. To further elucidate molecular mechanisms, RNA sequencing (RNA-seq) was conducted on bilirubin-exposed neuronal cells to identify differentially expressed genes and dysregulated pathways.
Results: Children with a history of neonatal jaundice demonstrated a significantly increased risk of ADHD and learning disabilities compared with controls, with the highest risk observed in those requiring hospitalization, suggesting a severity-dependent effect. In vitro, bilirubin exposure dose-dependently suppressed neuronal and astrocytic proliferation, inhibited neurite growth, and reduced synaptic formation. Notably, these impairments persisted even after bilirubin withdrawal. Astrocytes partially preserved neuronal survival but failed to rescue neurite outgrowth.
RNA-seq analysis revealed significant dysregulation of genes involved in synaptic plasticity, axon guidance, mitochondrial function, oxidative stress, and neuroinflammatory signaling, indicating that bilirubin induces sustained transcriptional alterations that may underlie long-term neurodevelopmental dysfunction.
Conclusion: Neonatal jaundice is associated with an increased long-term risk of ADHD and learning disabilities. Bilirubin exerts persistent effects on neuronal differentiation, synaptic development, and gene expression, even at serum levels currently regarded as safe (15–18 mg/dL). These findings suggest that minimal bilirubin-induced brain injury may be underrecognized and support reconsideration of current treatment thresholds to better protect neurodevelopmental outcomes.

CP5

EEG-Based Classification For Tic Disorders via Complementary Deep Model Integration

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Purpose: To improve the robustness and generalization of EEG-based detection of tic disorders by systematically comparing multiple deep-learning (DL) architectures and evaluating ensemble strategies that integrate their complementary strengths.
Methods: EEG recordings were acquired using an EEG machine (Neurofax EEG-1200; Nihon Kohden, Tokyo, Japan) with Ag/AgCl electrodes at a sampling rate of 200 Hz. Nineteen electrodes were placed according to the International 10–20 system by a trained technologist. All electrodes were referenced to the ground electrode at Fpz during recording. After visual inspection for artifact rejection, a 10-s eye-closed waking segment was extracted for analysis. Data were split into training (40 tic, 40 control) and independent test (30 tic, 30 control) sets. We trained Convolutional Neural Networks (CNN), Deep Neural Networks (DNN), Long Short-Term Memory networks (LSTM), and Conformer models. Within the training set, we performed stratified cross-validation to examine sensitivity/specificity balance and class bias using confusion matrices. Feature-space visualizations (model embeddings) assessed decision-boundary stability. We then evaluated multiple ensemble schemes (hard voting, soft voting, learned fusion), with special focus on LSTM-anchored combinations.
Results: CNN and Conformer achieved high training sensitivity but showed notable generalization drop on the independent test set, with evidence of class bias and unstable decision boundaries. Among single models, LSTM exhibited the most balanced performance, maintaining relatively stable sensitivity and specificity across cross-validation and test sets; however, no individual model was uniformly optimal. Ensembles that included LSTM consistently outperformed other configurations. The best ensemble—integrating LSTM + CNN + DNN + Conformer—achieved accuracy = 0.90 and unweighted average recall (UAR) = 0.90 on the independent test set, while preserving a favorable sensitivity–specificity trade-off. Embedding visualizations showed that the ensemble produced more compact and separable class clusters than any single classifier.
Conclusions: Reliable EEG classification of tic disorders is better achieved through complementary model integration than by increasing the complexity of a single architecture. An LSTM-anchored ensemble mitigates model-specific biases, stabilizes decision boundaries, and enhances generalization. Using short (10-s), eye-closed EEG recorded with a standard 19-channel 10–20 montage at 200 Hz, our approach delivers strong performance on an independent cohort, supporting the feasibility of practical, robust EEG-based screening for tic disorders in clinical settings.

CP6

GMP-Grade Umbilical MSC Secretome in Paediatric Drug-Resistant Epilepsy: Matched Case–Control Evidence of Seizure, Functional, and IL-6/hs-CRP Biomarker Shifts

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Purpose: Drug-resistant epilepsy (DRE) is etiologically diverse yet increasingly conceptualized as a convergent neuroimmune network disorder, where cytokine-driven signalling—particularly IL-6—can amplify excitability, synaptic dysfunction, and injury, while systemic inflammatory tone is mirrored by high-sensitivity C-reactive protein (hs-CRP). Cell-free mesenchymal stem/stromal cell secretome (MSC-S), enriched with soluble mediators and extracellular vesicles, offers a plausible strategy to modulate these shared molecular “final pathways" without cell engraftment. We assessed whether adjunctive intravenous MSC-S is associated with clinical improvement and inflammatory biomarker downshifts in paediatric DRE.
Methods: In a matched observational case–control design, children with DRE receiving MSC-S (n=16) were compared with controls receiving standard care (n=16). MSC-S was derived from *clinical-grade/GMP-certified umbilical MSCs* (Kalbe Regenic) and delivered as *10 intravenous doses over 6 months*, alongside standardized antiseizure therapy and rehabilitation held stable across groups. Serum IL-6 and hs-CRP were measured at baseline and 6 months using standardized commercial-laboratory ELISA. Outcomes were monthly seizure frequency (primary), GMFM-66 in participants with CP phenotype, Bayley-4 cognitive score in participants with GDD phenotype, and safety/tolerability.
Results: MSC-S was well tolerated with no major safety signals. A >35% seizure reduction occurred in 62.5% (10/16) of MSC-S recipients versus 31.2% (5/16) of controls (p=0.048). Clinically meaningful GMFM-66 improvement (≥5 points) was observed in 9/16 versus 4/16 (p=0.007). Bayley-4 cognitive scores increased by +7.2±3.1 versus +2.1±2.8 points (p<0.001). Biomarker trajectories were concordant with a systemic anti-inflammatory signal: baseline IL-6 was 21.0±5.65 pg/mL across the groups; at 6 months, IL-6 was lower in MSC-S group (9.1±5.25) than controls (15.22±7.23, p<0.048). Baseline hs-CRP was 6.82±2.4 mg/L across the groups; at 6 months, hs-CRP was 0.88±0.52 in MSC-S versus 4.22±2.11 in controls (p<0.032).
Conclusions: Adjunctive GMP-grade umbilical MSC secretome was associated with improved seizure control and functional/developmental gains accompanied by marked reductions in IL-6 and hs-CRP. These findings support further biomarker-stratified randomized trials to test causality and define immunomodulatory mechanisms in rare and intractable epilepsies.

CP7

Epilepsy with CDK19 variants: clinical manifestations and outcome

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Purpose: Cyclin-dependent kinase 19 (CDK19) encodes a component of the Mediator kinase module, which plays a critical role in RNA polymerase II–dependent transcription. De novo CDK19 variants have been implicated in developmental and epileptic encephalopathy 87 (DEE87); however, reported cases remain limited, and the phenotypic spectrum is incompletely defined. This study aims to expand the clinical and molecular characterization of CDK19-related neurodevelopmental disorders by reporting additional cases and systematically reviewing published literature.
Methods: Four previously unreported individuals with de novo pathogenic CDK19 variants and 16 published cases were identified through international collaboration using GeneMatcher. Detailed clinical, neuroimaging, EEG, and genetic data were collected from contributing clinicians. A systematic literature review of PubMed was performed to identify genetically confirmed CDK19 cases with available phenotypic data.
Results: Among the 20 individuals, 55% were male. Seizures were present in approximately 70% of cases, most commonly infantile spasms (42.8%), followed by generalized (64.3%) and focal seizures (35.7%). Seizure control was poor, with 85% of affected individuals exhibiting refractory epilepsy despite polytherapy. All individuals demonstrated developmental delay, with 60% showing severe global impairment and absence of verbal communication. Hypotonia (70%), abnormal gait (40%), and autism spectrum features (45%) were frequent. Brain MRI findings were variable, with 23% reported as normal and others showing white matter abnormalities, cerebral atrophy, or corpus callosum thinning. Genetically, most variants clustered within codons 28–32 of CDK19, accounting for 60% of cases, suggesting a mutational hotspot in the N-terminal region. This combined cohort refines the clinical profile of CDK19-related neurodevelopmental disorders, highlighting refractory epilepsy, global developmental delay, hypotonia, and distinctive facial dysmorphism as core features. Compared with earlier reports, our findings demonstrate broader seizure phenotypes and occasional milder developmental outcomes, indicating greater phenotypic heterogeneity.
Conclusion: By integrating four unpublished cases with all reported individuals to date, this study provides the most comprehensive characterization of CDK19-related developmental and epileptic encephalopathy. The high burden of refractory epilepsy and the identification of a recurrent mutational hotspot have important implications for early recognition, genetic diagnosis, and future mechanistic studies.

CP8

Epileptic phenotype in a patient with a MARK2 variant: the first detailed description and review of the literature

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Purpose: The gene encoding microtubule affinity-regulating kinase 2 (*MARK2*) has recently been implicated in patients with autism spectrum disorder (ASD). Although seizures have been reported in 46.4% of a small cohort, detailed information regarding seizure phenotypes and longitudinal progression remains scarce. We'd like to present a case of *MARK2*-related epilepsy and review previously reported cases in the literature.
Methods: We describe a case with a pathogenic *de novo MARK2* variant, including detailed longitudinal information on seizure manifestations, EEG evolution, and therapeutic response. In addition, we review and summarize the clinical features of the fifteen previously reported individuals with pathogenic or likely pathogenic *MARK2* variants who experienced seizures, incorporating our current case.
Results: An 11-year-old male patient with ASD carrying a heterozygous pathogenic variant (c.888+1G>A) in *MARK2* gene experienced his first nocturnal tonic seizure at 5 years 11 months. Recurrent nocturnal focal impaired-consciousness seizures were accompanied by left temporal sharps on EEG, and episodes of focal status epilepticus were also noted. Oxcarbazepine provided only partial benefit, whereas perampanel effectively reduced nocturnal seizures and improved sleep initiation. He has remained seizure-free since 9 years 9 months on combined therapy. In our literature review, no prior reports described longitudinal changes in seizure manifestations, EEG evolution, or treatment response. EEG abnormalities were noted in 72.7% of patients (8/11), with focal epileptiform discharges being the most common finding (N=5). No major structural abnormalities were identified on brain MRI.
Conclusions: This case provides the first detailed clinical description of *MARK2*-related epilepsy, characterized by focal seizures responsive to antiseizure medication.

CP9

Severe Neonatal Encephalopathy in a Boy Associated with a Novel De Novo MECP2 Nonsense Variant

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Purpose: Mutations in the MECP2 gene cause a broad spectrum of neurodevelopmental disorders. While Rett syndrome is well known in females, severe neonatal encephalopathy associated with MECP2 variants occurs almost exclusively in males and is very rarely reported. We describe the first Korean boy with severe neonatal encephalopathy caused by a de novo novel nonsense variant in MECP2.
Methods: A boy presenting with early-onset respiratory failure, seizures, and developmental delay followed by regression from infancy. Clinical features, neuroimaging, electroencephalography, and metabolic evaluations were reviewed. Extensive genetic testing was performed, including chromosomal microarray analysis, targeted molecular tests for Prader-Willi syndrome and Fragile X syndrome, and they were negative. Whole exome sequencing was done and identified variants were interpreted according to the American College of Medical Genetics (ACMG) and Genomics guidelines. Sanger sequencing tests for the candidate variant of parents were performed to determine inheritance.
Results: The patient was born at 36 weeks and 5 days of gestation and showed apnea, stridor, and desaturation shortly after birth, requiring tracheostomy and mechanical ventilation. Brain magnetic resonance imaging (MRI) revealed bilateral cerebral white matter lesions suggestive of hypoxic injury, and electroencephalography showed the burst suppression pattern. Seizures were observed during neonatal period, and it was controlled with phenobarbital. Although brief developmental progress was observed, significant regression occurred after 6 months of age, accompanied by hypotonia and loss of eye contact. Follow-up brain MRI revealed corpus callosum thinning and diffuse cortical atrophy. Metabolic and biochemical studies were unremarkable. Whole exome sequencing identified a hemizygous nonsense variant in MECP2, c.658C>T (p.Gln220Ter), on the X chromosome. Trio Sanger sequencing confirmed the variant as de novo. This variant was classified as likely pathogenic based on ACMG criteria and has not been previously reported.
Conclusions: This report expands the mutational and clinical spectrum of MECP2-related disorders in males. Severe neonatal encephalopathy due to MECP2 mutations should be considered in male infants presenting with early-onset encephalopathy, respiratory failure, and developmental regression. Early genetic diagnosis using whole exome sequencing is crucial for accurate diagnosis, prognosis, and genetic counseling.

CP10

Perampanel Treatment in CELF2-Related Developmental and Epileptic Encephalopathy: A Case With Partial Respons

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Purpose: CELF2-related developmental and epileptic encephalopathy (DEE) is a rare condition typically characterized by early-onset, severe epilepsy, profound developmental delay, markedly abnormal EEG findings, and structural brain abnormalities. Common seizure types include infantile spasms, tonic seizures, and generalized tonic-clonic seizures, whereas myoclonic seizures have been rarely reported. We describe a patient with CELF2-related DEE presenting with prominent myoclonic seizures and a partial response to perampanel.
Methods: This case report retrospectively reviewed clinical data, including seizure semiology, developmental course, antiseizure medication (ASM) response, EEG findings, neuroimaging, and genetic results. Genetic testing was performed using next-generation sequencing.
Results: The patient is a female with global developmental delay and seizure onset at 4 months of age, manifesting tonic and prominent myoclonic seizures. Despite treatment with multiple ASMs, including levetiracetam, seizures occurred daily. Following initiation of perampanel at 2 mg twice daily, a significant reduction in myoclonic seizures was observed. By 9 years of age, myoclonic seizures became rare, with no events observed for several months, although other seizure types persisted. EEG demonstrated generalized multifocal epileptiform discharges. Brain MRI revealed diffuse structural abnormalities, including thinning of the corpus callosum, reduced cerebral and cerebellar volumes, small basal ganglia, and suspected diffuse parenchymal loss. Genetic analysis identified a heterozygous CELF2 variant (c.976G>A), inherited from a mosaic mother, consistent with autosomal dominant inheritance.
Conclusions: This case expands the phenotypic spectrum of CELF2-related DEE by highlighting myoclonic seizures as a notable feature and suggests that perampanel may provide partial seizure control, particularly for myoclonic seizures. Given emerging evidence linking CELF2 variants to neurodevelopmental disorders, perampanel may be considered a therapeutic option in selected patients with CELF2-associated epilepsy.

CP11

Expanding the Phenotypic Spectrum of KCNK4-Related Developmental and Epileptic Encephalopathy: A Case Report Without Classical FHEIG Features

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Purpose: Pathogenic variants in KCNK4, encoding the TASK-1 potassium channel, are associated with a rare neurodevelopmental disorder known as FHEIG syndrome (facial dysmorphism, hypertrichosis, epilepsy, intellectual disability, and gingival overgrowth). Fewer than ten cases have been reported worldwide, and the epileptic phenotype remains incompletely characterized. We report a child with KCNK4-related developmental and epileptic encephalopathy (DEE) without classical syndromic features, aiming to expand the recognized phenotypic spectrum.
Methods: We describe the clinical, electroencephalography(EEG), neuroimaging, and genetic findings of an 8-year-old boy with early-onset epileptic encephalopathy. Clinical evaluation, serial EEG monitoring, brain magnetic resonance imaging, targeted genetic testing for Angelman and Prader–Willi syndromes, and whole-exome sequencing were performed.
Results: The patient presented with speech delay and autistic traits prior to seizure onset at one year of age, manifesting as generalized tonic–clonic seizures. Examination revealed autistic features without facial dysmorphism, hypertrichosis, or gingival overgrowth. Brain MRI was normal. Initial EEG demonstrated diffuse background slowing without focal epileptiform discharges. Seizures became drug-resistant by three years of age despite sodium valproate therapy, requiring polytherapy with clobazam and levetiracetam. Serial EEGs evolved into a Lennox–Gastaut syndrome pattern. Whole-exome sequencing identified a heterozygous KCNK4 variant, c.698C>T (p.Pro233Leu), previously reported and classified as likely pathogenic according to American College of Medical Genetics and Genomics(ACMG) criteria. The variant affects a highly conserved residue and is predicted to be deleterious.
Conclusion: This case broadens the phenotypic spectrum of KCNK4-associated disease, demonstrating that severe DEE may occur in the absence of classical FHEIG features. Recognition of this variability is important to prompt early genetic testing in children with refractory epilepsy and normal neuroimaging. Further case accumulation is needed to clarify genotype–phenotype correlations in KCNK4-related disorders.

CP12

Clinical features of HNRNPU deletion syndrome

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Heterogeneous nuclear ribonucleoproteins (HNRNPs) are a family of proteins that bind to RNA and are involved in several processes of mRNA maturation and expression. HNRNP-related neurodevelopmental disorders present with developmental delay, epilepsy and autism spectrum disorders, as well as various symptoms affecting multiple organs. The most common subtype is HNRNPU deletion syndrome, which it caused by pathogenic variants in the *HNRNPU* gene or by a 1q44 chromosomal microdeletion. Patient 1 was a 10-year-old female who presented with global developmental delay from infancy. She developed a cluster of febrile seizures from 11 months of age and displayed hand stereotypy involving mutual hand rubbing from 18 months of age. She started walking at the age of six, can use cutlery, but cannot speak. A brain MRI scan revealed thinning of the corpus callosum. Whole exome analysis revealed a de novo pathogenic variant c.1222T>C (p.Cys408Arg) in the *HNRNPU* gene. Patient 2 and 3 were 14-year-old female homozygous twins who developed intractable focal epilepsy from two months of age. They exhibited severe developmental delay, which was limited to rolling over from the age of two, and were unable to speak. They exhibited hyperpnoea and trembling of both arms when excited. Brain MRI scans showed thinning of the corpus callosum and progressive cerebral and cerebellar atrophy. Microarray analysis revealed a 920 kb microdeletion in 1q44. Patient 4 was an 11-year-old male with refractory focal epilepsy from one month of age. He had profound global developmental delay and remained at the crawling stage. He could not speak. He had microcephaly. He exhibited hyperpnoea and trembling of both arms when excited. A brain MRI scan showed a lack of corpus callosum. Microarray analysis revealed a 6.9 Mb microdeletion in 1q44. While all patients exhibited severe intellectual disability, epilepsy and hand stereotypy, patient 1 displayed a milder phenotype. Therefore, the phenotype of the 1q44 chromosomal microdeletion was much more severe than a single-gene defect of *HNRNPU*. Patient 2-4 shared a deletion area that included the *HNRNPU*, *C1orf199*, *FAM36A*, *PPPDE1*, *C1orf101*, *ADSS*, *C1orf10* and *ZBTB18* genes. Patient 4 had a larger deletion area, including the *AKT3* gene. Agenesis of the corpus callosum may be related to the *AKT3* gene and the common features of hyperpnoea and hand trembling stereotypy may be related to these genes, which are delated in common. 1q44 deletion syndrome has characteristics that are unique to it, other than the single *HNRNPU* defects.

CP13

A Case of ACTL6B-Related Disorder Presenting with Developmental and Epileptic Encephalopathy, Responsive to Corpus Callosotomy

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Introduction: ACTL6B-related disorders are characterized by severe global developmental delay, drug-resistant epilepsy, and Rett syndrome–like stereotypies, caused by pathogenic variants of ACTL6B, which encodes a neuron-specific subunit of the nBAF chromatin remodeling complex.
Patient: During the 10-month health checkup, difficulty in maintaining a sitting position prompted medical consultation; subsequent brain MRI at 12 months revealed delayed myelination. Repetitive hand movements were also observed. Independent walking was achieved at age 3, although with an ataxic gait. Brain MRI at 7 years of age revealed cerebellar atrophy. Various types of seizures, primarily infantile epileptic spasms syndrome, began at age 5. Epilepsy gene panel testing identified compound heterozygous pathogenic variants of ACTL6B: c.709_711del:p.Lys237del (maternal origin), and c.1031_1032insGGGGAG:p.Ser344delinsArgGlySer (paternal origin). These mutations were located on different alleles, which is consistent with an autosomal recessive inheritance pattern. ACTH therapy was initially effective; however, the seizures occurred again, and the patient's condition became drug-resistant. Subsequently corpus callosotomy was performed at age 8, which resulted in seizure cessation.
Discussion: ACTL6B encodes BAF53B, an actin-related protein that is a neuron-specific subunit of the BAF chromatin remodeling complex. This protein plays crucial roles in neuronal differentiation, dendritic outgrowth, synaptic function, and long-term memory. Mutations in ACTL6B lead to a spectrum of neurodevelopmental disorders depending on the mode of inheritance. Biallelic (recessive) mutations typically result in a severe phenotype characterized by global developmental delay, profound intellectual disability, axial hypotonia, limb spasticity, epilepsy (including infantile spasms), feeding difficulties, and severely limited or absent speech and ambulation. Many affected individuals are nonverbal and no ambulatory, and brain MRI may reveal structural abnormalities such as microcephaly and a thin corpus callosum. In contrast, heterozygous de novo mutations, particularly the recurrent p.Gly343Arg variant, are associated with a distinct phenotype involving severe intellectual disability, delayed motor milestones, profound speech impairment, hypotonia, autism spectrum disorder, Rett-like stereotypies (e.g., hand-wringing), and mild craniofacial dysmorphisms such as a wide mouth, diastema, bulbous nasal tip, and hypertelorism.
Conclusion: ACTL6B-related epilepsy is often drug-resistant, and no established treatment protocol currently exists. In the case reported here, corpus callosotomy was performed, and resulted in seizure cessation. Information from additional cases is necessary to evaluate the efficacy of corpus callosotomy as a therapeutic strategy in this disorder.

CP14

Navigating Failure to Thrive, Refractory Epilepsy, Global Developmental Delay and Movement Disorder in a 2-Year-Old Male: The Diagnostic Significance of Extended 15q11.2-q13.2 Microdeletion

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Purpose: To report the clinico-genetic features of a 2-year-old child with an 15q11.2-q13.2 microdeletion presenting as atypical Angelman syndrome with refractory epilepsy and choreoathetosis.
Methods: A 35-week preterm male was longitudinally evaluated for global developmental delay, failure to thrive, and progressive motor regression. Diagnostic workup included serial neurological examination, electroencephalography (EEG), brain MRI and ^{99m}Tc-TRODAT-1 dopamine transporter SPECT. Molecular diagnosis was achieved through whole-exome sequencing (WES) for copy number variation (CNV) detection, targeting coding structural alterations.
Results: Neurological examination revealed a dissociated motor phenotype: profound axial hypotonia with retrocollis alongside appendicular hypertonia and hyperreflexia. Brain MRI revealed slow myelination of bilateral parietal periventricular white matter on T2FLAIR and T2WI. EEG demonstrated continuous spike-and-wave during sleep (CSWS), indicating evolution into a severe epileptic encephalopathy. Despite clinical choreoathetosis, ^{99m}Tc-TRODAT-1 SPECT showed robust, symmetric radiotracer uptake in the bilateral striatum, suggesting preserved presynaptic dopaminergic integrity. CNV analysis identified an 8.67-Mb pathogenic deletion at 15q11.2-q13.2, involving the imprinting-regulated *UBE3A* locus and the 15q13.3 region including *CHRNA7*.
Conclusions: The combination of dissociated muscle tone, pre-verbal stagnation, and the electrographic hallmark of CSWS supports a diagnosis of Angelman Syndrome. The normal ^{99m}Tc-TRODAT-1 SPECT results suggest that the movement disorder originates from extra-striatal synaptic dysregulation rather than nigrostriatal deficiency. Methylation-specific multiplex ligation-dependent probe amplification (MS-MLPA) is planned to confirm the imprinting status.

CP15

Functional Evaluation and Mechanism-Based Rescue of a Novel Variant (A305V) In GABA Transporter 1-encoding SLC6A1 Associated With Myoclonic Atonic Epilepsy

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Background: We have previously studied the pathophysiology of mutations in **SLC6A1**, which encodes γ-aminobutyric acid (GABA) transporter 1 (GAT-1). Here, we report a new variant in **SLC6A1** associated with myoclonic-atonic epilepsy. We used artificial intelligence (AI) tools to predict the mutational impact and to correlate AI predictions with experimental studies.
Methods: We report a novel de novo variant (c.>A (Ala305Val)) in a girl with myoclonic-atonic epilepsy. We used both AI tools and experimental approaches to characterize this novel mutation (A305V) in **SLC6A1** and compared it with another variant at the same amino acid position (A305T).
Results: Both AI and experimental approaches indicate that the GAT-1(A305V) mutation destabilizes the global protein conformation and shows increased localization within the endoplasmic reticulum (ER) in astrocytes cultured from postnatal day 1 mouse pups. A radioactive ^{3}H-labeled GABA uptake assay indicated that the mutation reduced GAT-1(A305V) function to ~25% of wild-type, while GAT-1(A305T) reduced function to ~35% of wild-type. Importantly, pharmacochaperones such as 4-phenylbutyric acid (PBA) rescued trafficking and function of the mutant GAT-1.
Conclusions: AI tools and experimental approaches consistently suggest that the A305V and A305T mutations destabilize global protein conformation and reduce mutant GAT-1 GABA uptake function. Excitingly, PBA treatment rescued mutant GAT-1 protein expression and GABA uptake function in both HEK293T cells and astrocytes.
(**Acknowledgements:** This research was supported by research grants from National Institute of Health (NIH) National Institute of Neurological Disorders and Stroke (NINDS) NS121718 to J-Q.K. The work was also supported by research grants from SLC6A1 Connect to J-Q.K.

CP16

Successful Treatment of Dravet Syndrome with Camellia Oil: A Case Report

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Purpose: Dravet syndrome (DS) is a severe genetic epileptic encephalopathy with intractable seizures. The ketogenic diet (KD) is an effective adjunctive therapy but commonly relies on costly medium-chain triglyceride (MCT) oil. This study evaluated the feasibility and clinical effects of substituting MCT oil with camellia oil in KD therapy for a child with DS. Camellia oil is rich in monounsaturated fatty acids (70–82% oleic acid) and antioxidants, with good cooking stability.
Case report: A 7-year-old female was hospitalized in late March 2016 due to frequent epileptic seizures. Following physician assessment, ketogenic diet therapy was initiated. A 6-day inpatient program provided dietary adjustment and caregiver education, with the mother continuing implementation at home after discharge. The diet was designed based on the patient's age, height, and weight, providing 1600 kcal/day with macronutrient ratios of 80% fat and 20% combined carbohydrate and protein. Camellia oil replaced traditional MCT oil as the cooking fat. The patient was followed in outpatient clinics until June 2021, during which the fat ratio was gradually reduced to 65%. Seizure frequency, growth parameters, biochemical values, and antiepileptic medication use were monitored.
Results: Seizure frequency decreased significantly, and seizure duration shortened. From treatment initiation through 2021, average seizure frequency decreased from multiple episodes per month to occasional events (2-3 times per month). Growth parameters showed stable increases in height and weight with age, with BMI maintained within normal range for age. Biochemical results (including blood glucose, liver and kidney function, lipid profile, and ketone bodies) remained within normal ranges, with no hypercholesterolemia or hypertriglyceridemia despite the high-fat diet. No adverse effects such as diarrhea, vomiting, or fat malabsorption occurred. The family reported good cooking acceptability, favorable taste, and excellent dietary compliance. Antiepileptic medication dosage was moderately reduced in later stages, with overall stable clinical control.
Conclusion: This case demonstrates that camellia oil can be successfully used in ketogenic diet therapy without causing hyperlipidemia or diarrhea despite high fat content. The patient showed improved growth parameters and reduced seizure frequency. Camellia oil's high monounsaturated fatty acid content may reduce the risk of essential fatty acid deficiency during dietary treatment. Although research on camellia oil application in ketogenic diets remains limited, this case suggests it may be a cost-effective alternative to MCT oil. Further implementation should be conducted under physician and dietitian supervision, with continued monitoring for individual variations, allergic reactions, and drug interactions to provide additional clinical evidence.

CP17

SCN1A Mutation Location Predicts Stiripentol Efficacy in Dravet Syndrome

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Background: Stiripentol (STP) is an essential anticonvulsant for treating Dravet syndrome, but patient responses vary, especially among those with SCN1A mutations. Previous theories suggested that the efficacy of STP could be linked to channel pore dimensions, yet the exact molecular factors influencing its effectiveness remain elusive. This study aims to explore how the types and spatial distributions of SCN1A mutations impact STP-protein affinity and clinical outcomes.
Methods: We examined a range of SCN1A mutations, including missense, splicing, and truncation variants. Molecular docking using Vina scores was conducted to evaluate the binding affinity of STP with mutated Nav1.1 channels. Clinical responses were classified as "good" (≥50% response) or "poor/resistant" (<50% response). We analyzed the relationship between drug efficacy and two key variables: cavity size (Δcavity) and the functional domain containing the mutation.
Results: Our findings revealed that mutation types (missense, splice, and truncation) significantly influence STP's binding affinity to SCN1A. Interestingly, no strong correlation was found between cavity size and drug response. However, a notable spatial pattern was identified: the most significant differences in response rates were linked to mutations in Domain I (DI) and the Domain II-III (DII-DIII) linker regions.
Conclusions: The response to STP is primarily determined by the specific location of SCN1A mutations, not by cavity volume. Mutations in Domain I and the DII-DIII linker are critical indicators of clinical efficacy, suggesting a genetic framework for predicting STP treatment outcomes.

CP18

Experience of Using Ketogenic Therapy with Camellia Oil in the Treatment of Infantile Spasms: A Case Series of 5 Patients

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Background: Infantile spasms (IS) represent a severe epileptic encephalopathy characterized by epileptic spasms, hypsarrhythmia on electroencephalography, and psychomotor developmental delay. The ketogenic diet (KD) has emerged as an established non-pharmacologic treatment option for drug-resistant epilepsy. Recent evidence suggests that lipid composition within KD formulations may significantly influence clinical outcomes. Camellia oil, characterized by high monounsaturated fatty acid content and polyphenolic compounds, may confer additional neuroprotective and anti-inflammatory properties that could enhance therapeutic efficacy.
Purpose: To evaluate the effectiveness and tolerability of a camellia oil-enriched ketogenic diet in children with infantile spasms.
Methods: We conducted a retrospective case series of five previously KD-untreated children diagnosed with IS at a single tertiary pediatric center. The KD was initiated at a classical 4:1 fat-to-(carbohydrate plus protein) ratio utilizing a standardized medium- and long-chain triglyceride formula with camellia oil incorporated as a partial fat source. Diet ratios were subsequently down-titrated following clinical improvement. Seizure frequency was systematically recorded from caregiver seizure logs throughout a 12-month follow-up period or until KD discontinuation. Concomitant antiseizure medications and adverse events were prospectively documented.
Results: Five patients with infantile spasms (IS) were included (M:F = 4:1), with ages ranging from 18 to 66 months. At baseline, daily spasm frequency ranged from 1 to 30 episodes. Following KD initiation, all five patients (100%) demonstrated clinical improvement. Mean weekly seizure frequency decreased from 86.7 to 5.9 episodes, representing a 93.2% reduction. Median weekly seizure frequency decreased from 105 to 1.5 episodes, demonstrating a 98.6% reduction. One patient experienced a single seizure relapse within the 6-month follow-up period. No severe metabolic complications were observed, and no patients discontinued the diet due to adverse effects or intolerance.
Conclusion: Camellia oil-enriched KD demonstrated significant effectiveness and excellent tolerability in reducing seizure frequency among children with IS in this case series. This dietary modification may represent a promising optimization strategy for KD therapy in pediatric refractory epilepsy. Further prospective, controlled studies with larger sample sizes are warranted to validate these preliminary findings and elucidate potential mechanisms underlying the observed benefits.

CP19

ALG13-Related Congenital Disorder of Glycosylation Presenting as Infantile Epileptic Spasms Syndrome

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Purpose: To describe the clinical presentation, diagnostic process, and treatment course of an infant with infantile epileptic spasms syndrome (IESS) caused by ALG13-related congenital disorder of glycosylation (CDG).
Methods: We retrospectively reviewed the clinical course, electroencephalographic findings, imaging studies, laboratory data, genetic testing, and treatment response of a 6-month-old female infant.
Results: The patient developed progressive daytime paroxysmal episodes beginning at 5 months of age, characterized by blinking, upward gaze deviation, limb elevation, and neck extension without loss of consciousness. Episode frequency and duration increased over time, followed by developmental regression. EEG performed on December 29 demonstrated findings consistent with IESS. High-dose vitamin B6 and folinic acid were administered, with limited clinical response. Brain MRI and abdominal ultrasonography revealed no structural abnormalities. Lumbar puncture showed normal glucose and protein levels. Persistent elevation of liver transaminases was noted, with negative viral studies. Whole-exome sequencing identified a pathogenic variant in ALG13, confirming a diagnosis of ALG13-related CDG. Multidisciplinary evaluations, including ophthalmologic and cardiac assessments, revealed no significant abnormalities. Due to ongoing seizures, oral prednisolone was initiated and escalated, and vigabatrin was added. Follow-up 24-hour EEG demonstrated improvement in background activity, although residual nocturnal seizures persisted. The patient was discharged in stable condition with continued antiepileptic therapy and rehabilitation support.
Conclusion: ALG13-related CDG should be considered in infants presenting with IESS, developmental regression, and unexplained hepatic dysfunction. Early genetic diagnosis is essential for guiding management and counseling in this rare condition.

CP20

Epileptic Spasms Occurred Earlier in JE Induced Anti-Nmdar Encephalitis

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Purpose: Epileptic spasms are well-recognized post-encephalitis Epilepsy, but the timing of appearance can be varied depends on post infectious or autoimmune encephalitis. In 2017 Tokorodani, Chiho et al. reported 2 cases of epileptic spasms following infectious encephalitis were 21 months and 41 moths while one year later, Okanishi, Tohru et al. reported a 5-year-old girl with anti-NMDAR encephalitis developed epileptic spasms only after 5 months from the onset. We are going to report a 20-month-old infant boy developed epileptic spasms occurred much earlier in JE induced anti-NMDAR encephalitis.
Case report: A 20-month-old infant boy presented with status epilepticus following five-day history of fever. His GCS remained 4/15 despite status epilepticus was aborted. CRP was >200 mg/L, total WBC 24.37 x10^9/L, CSF protein (82 g/L) and cell count (110 cells/mcl) with 97% lymphocytes; CSF gram statin, culture and FilmArray multiplex PCR ME panel test were negative and positive JE IgM. Generalized dystonia appeared after 2 weeks of the onset of illness. Because of progressive dystonia, increased agitation and reduced sleep, CSF anti-NMDAR antibody was tested and revealed positive. Epileptic spasms developed after 3 months from the onset. It did not respond to conventional ASMs and the first cycle of IV methylprednisolone and IVIG. As second line immunotherapeutic agents are not sustainably available in Myanmar, monthly IV methylprednisolone pulse therapy were given. After 3 cycles, spasms remission and developmental skill gradually recovered.
Conclusion: JE induced anti-NMDAR encephalitis in pediatric patients is previously known and the distinctive manifestations in younger children are seizure and dyskinetic movements. We would like to highlight the earlier occurrence of epileptic spasms in cases of JE induced anti-NMDAR encephalitis in younger patient could be difficult to recognized and mis-diagnosed as dyskinetic movements resulting in delayed treatment.

CP21

Case Series of New-Onset Refractory Status Epilepticus

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Purpose: New-onset refractory status epilepticus (NORSE) is characterized by the absence of a structural, metabolic or toxic cause that can explain the presence of RSE in otherwise healthy patients, despite extensive diagnostic workup and no control of status epilepticus within 48 hours. These often include viral and autoimmune encephalitis. We are going to describe 4 cases of new-onset refractory status epilepticus admitted to ICU of our center from June 2025 to January 2026.
Case series: Case-1 is a 10-year-old girl experienced generalized tonic-clonic seizure after 8-hours onset of fever without associated respiratory or GI manifestations. Case-2 is an 8-year-old girl, started to have generalized seizure after 72-hours of febrile illness. Case-3 is a 20-months old infant girl presented with bilateral asymmetric tonic seizure after 48-hours of fever with cough and respiratory distress. Case-4 is a 4-month-old infant girl admitted for generalized tonic seizure 6-hours following pentavalent vaccination and associated with fever. Complete seizure control was achieved only after giving IV Methylprednisolone 30 mg/kg/day 5 doses and IVIG total 2 G/kg in addition to IV phenobarbitone 20 mg/kg/day, IV levetiracetam 80-100 mg/kg/day, IV continuous infusion midazolam up to 6-8 mcg/kg/min in case-1, 2 and 3 on Day-4, Day-5 and Day-7 from the onset of seizure respectively. Only case 4 achieved seizure control on Day-4 without requiring immunotherapy. None of the cases had CSF abnormal finding and negative autoimmune encephalitis antibodies. Only case 3 had raised CRP (24 mg/dl) and procalcitonin (0.3 ng/ml). CT scan was carried out in case 1, 2 and 3 and diffused cerebral oedema was reported in case 2 and 3. Among 4 cases, only case 2 had Dengue NS 1 antigen tested positive and no definite etiology could be found out in the rest of the cases. MRS score were 5, 6, 8 and 5 at discharge; 2, 4, 6 and 3 at 2 months follow-up. Only case-3 had refractory epilepsy developed at 2-months after discharge till 4 months of follow up.
Conclusion: New-onset refractory status epilepticus is not common but management is complex due to the difficulty to detect prompt identification of the etiology and limited therapeutic options available. It also often associated with high mortality and significant neurodevelopmental outcomes.

CP22

A novel frameshift DMD mutation (p.Leu2017Profs5) disrupts dystrophin spectrin-like repeats and destabilizes protein conformation: structural insights from AlphaFold-based modeling

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Background: Duchenne muscular dystrophy (DMD) is an X-linked neuromuscular disorder caused by mutations in the DMD gene, which encodes dystrophin, a key structural protein required for maintaining muscle fiber membrane integrity. With the increasing availability of disease-modifying therapies, early and accurate diagnosis of DMD has become increasingly important. In Taiwan, whole-exome sequencing (WES) is a feasible and widely accessible diagnostic approach for evaluating unexplained hyperCKemia and distinguishing DMD from other inherited neuromuscular disorders.
Methods and Results: Using WES, we identified a novel hemizygous deletion in the DMD gene (chrX:32310147; NM_004006.3: c.6050_6051del; p.Leu2017Profs5) in a Taiwanese boy presenting with markedly elevated serum creatine kinase levels and clinical features consistent with DMD. According to the American College of Medical Genetics and Genomics (ACMG) guidelines, this variant was classified as likely pathogenic. To further support its functional relevance, comparative genomic analysis was performed on the exon 42 spectrin-like repeat region encompassing the deletion site. Multiple sequence alignment across nine vertebrate species, ranging from chimpanzee to lizard, demonstrated strong evolutionary conservation of this region, suggesting that amino acid alterations at this site are likely to have a deleterious functional impact.
Structural consequences of the mutation were further investigated using AlphaFold-based modeling. A dystrophin fragment spanning Val1401 to Leu2800 was derived from the AlphaFold model (AF_AFP11532F8). Structural superimposition revealed that the truncated dystrophin protein failed to adopt the characteristic spectrin-like helix-bundle architecture observed in the wild-type protein. In addition, mutation-induced changes in protein stability were assessed by calculating free energy differences using the CHARMm force field. The predicted free energy of the p.Leu2017Profs5 variant was comparable to that of other pathogenic exon 42 variants reported in the Leiden Open Variation Database (LOVD), such as p.Glu2003Asnfs19 and p.Glu2013Ilefs11, but differed substantially from benign missense variants (p.Leu2017Ile and p.Asn2019Ser) in the same region.
Conclusions: We report the first case of Duchenne muscular dystrophy associated with the novel hemizygous deletion mutation p.Leu2017Profs5 in exon 42 of the DMD gene. By integrating genomic, evolutionary, and structural in silico analyses, this study expands the mutational spectrum of DMD and underscores the utility of combining WES with computational modeling for variant interpretation.

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てんかん発作が起きた時の対応、希少疾患の家族会の運営、生活の中で実践できる発達支援の工夫について講演します。
ご家族、教育関係者、支援者の方にも役立つプログラムです。
是非ご参加ください。

座長／加藤 光広〈昭和大学医学部小児科学講座教授〉

講演

1. 黒岩 ルビー〈ドラベ症候群患者家族会 代表〉
「発作が止まらない時どうする?～患者家族のリアル～」

2. 安部 恵美〈CDKL5遺伝子欠損症（CDD）患者家族会「CDKL5 JAPAN らぶはんず」代表〉
「希少疾患患者会にできること」

3. 高橋　輝〈静岡てんかん・神経医療センター 療育指導室保育士〉
「発達が気になる子どもさんへの関わり ～日常生活に取り入れたい発達支援の工夫～」

4. 質疑応答

公益財団法人　難病医学研究財団
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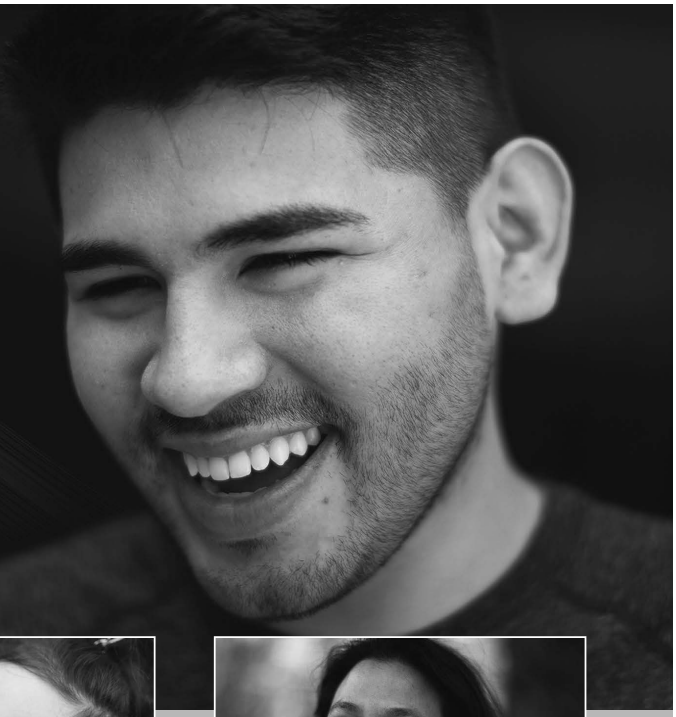
第 10 回アザリアフェスティバルシンポジウムの開催に際し、上記の企業・団体より多大なるご支援ご協力を賜りました。ここに感謝の意を表し、厚く御礼申し上げます。

第 10 回アザリアフェスティバルシンポジウム
会長　廣瀬　伸一

情熱の先にあるのは
より良い治療法の実現



アミカスは、希少疾患とともに
生きる患者さんの生活の質を
より良くすることを使命としています。

Amicus Therapeutics®
最先端の治療法で希少疾患に挑む

アミカス・セラピューティクス株式会社

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ファブリーコネクト 検索

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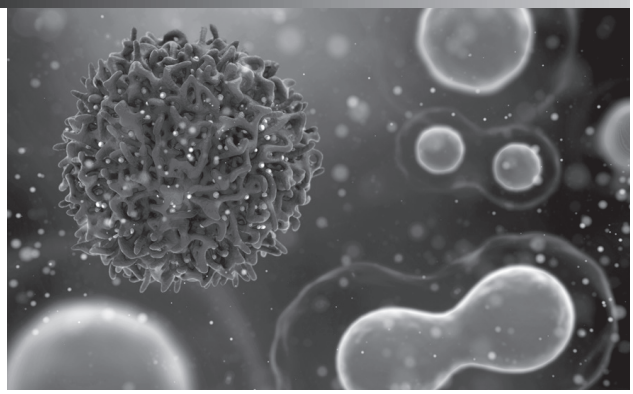
Transforming Lives. Redefining Possibilities.



Jazz Pharmaceuticals is a global biopharmaceutical company whose purpose is to innovate to transform the lives of patients and their families. We are dedicated to developing life-changing medicines for people with serious diseases — often with limited or no therapeutic options.

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