


詳情參閱：

[https://www.pediatr.org.tw/meeting/inland\\_info.asp?id=19552&Page=1&keyword=&SponserFlag=&TeenFlag=](https://www.pediatr.org.tw/meeting/inland_info.asp?id=19552&Page=1&keyword=&SponserFlag=&TeenFlag=)



Agenda			
08:30-09:00	Registration 報到		
09:00-09:10	Opening Remarks	Ming-Ren Chen Superintendent of MacKay Children's Hospital 陳銘仁院長 馬偕兒童醫院	
Time	Title of the Talks	Speakers	Moderators
09:10-10:30 Session I - New Trends in the Management of Mucopolysaccharidoses			
09:10-09:45	Unmet Needs of Current Treatment in Mucopolysaccharidoses: Addressing CNS and Bone Involvement	Prof. Sung Yoon Cho (趙龍允) Samsung Medical Center, Sungkyunkwan University, Seoul, Republic of Korea	林瑋宇主任 (馬偕兒童醫院) Hsiang-Yu Lin MacKay Children's Hospital
09:45-10:20	One and Only Taiwan Newborn Screening for Mucopolysaccharidoses Updates Worldwide and a Well-Established Screening Test for Alpha-Mannosidosis	莊志光研究員 (馬偕紀念醫院) Chuang, Chih-Kuang MacKay Memorial Hospital	
10:20-10:30	Panel Discussion		
10:30-10:50 Break			
10:50-12:10 Session II - New Trends in the Management of Spinal Muscular Atrophy			
10:50-11:25	Long-Term Benefit of Oral Therapy in Managing Spinal Muscular Atrophy	Prof. Furene Wang National University Hospital, Singapore	陳麗潔主任 (三軍總醫院) Shyh-Jou Chen Tri-Service General Hospital
11:25-12:00	The Change of Spinal Muscular Atrophy Therapy After Newborn Screening (新生兒篩檢後，脊髓性肌肉萎縮症治療的改變與現況)	許庭榕醫師 (臺北榮民總醫院) Ting-Rong Hsu Taipei Veterans General Hospital	
12:00-12:10	Panel Discussion		
12:10-12:40 Lunch Time			
[Lunch Symposium] Enzyme Replacement Therapy for Children with Acid Sphingomyelinase Deficiency in the Real World: Three-Year Experience in a Single Center in Taiwan			
12:40-13:30		周宮鎮醫師 (國立成功大學醫學院附設醫院) Yen-Yin Chou National Cheng Kung University Hospital	郭雲卿主任 (邵立雙和醫院) Yung-Ting Kuo Ministry of Health and Welfare Shuang-Ho Hospital
13:30-14:50 Session III - New Trends in the Diagnosis and Management of Fabry Disease			
13:30-14:05	Multi-Disciplinary Approach in Diagnosis & Management of Fabry Disease - Japan Experience	Prof. Kazuya Tsuboi (坪井一雄) Nagoya Central Hospital, Nagoya, Japan	洪崇烈主任 (馬偕紀念醫院) Chung-Lieh Hung MacKay Memorial Hospital
14:05-14:40	Cardiac Care in Fabry Disease (法布瑞氏症之心臟醫療照護)	宋國慈醫師 (馬偕紀念醫院) Kuo-Tzu Sung MacKay Memorial Hospital	
14:40-14:50	Panel Discussion		
14:50-15:10 Break			
15:10-16:30 Session IV - The Challenges of Next Generation Sequencing			
15:10-15:45	Lessons Learned from Pediatric Clinical Exome Sequencing (從兒科臨床全外顯子定序汲取的教訓與經驗)	李純輝教授 (臺大醫院) Prof. Ri-Chung Lee National Taiwan University Hospital	林炫沛教授 (馬偕兒童醫院) Shuan-Pei Lin MacKay Children's Hospital
15:45-16:20	Decoding the Future: Next-Generation Genetic Diagnostics and the Clinical Revolution in Personalized Medicine (解碼未來：新一代基因診斷技術與個人化醫療的臨床革命)	李忠政醫師 (馬偕兒童醫院) Chung-Lin Lee MacKay Children's Hospital	
16:20-16:30	Panel Discussion		
16:30-16:40	Closing Remarks	林炫沛教授 (馬偕兒童醫院) Shuan-Pei Lin MacKay Children's Hospital	

## WELCOME MESSAGE



Ladies and gentlemen, esteemed guests,

On behalf of MacKay Children's Hospital, I warmly welcome all domestic and international guests to our International Forum on Rare Diseases.


Established in 2016 under the leadership of Professor Shuan-Pei Lin, the Rare Disease Center at MacKay Memorial Hospital exemplifies our commitment to specialized medical care in Taiwan. It plays a pivotal role in enhancing the medical care for rare diseases. We also pioneer the holistic healthcare approach for patients with mucopolysaccharidoses. Our efforts have been recognized with the Silver Award in the 2018 Symbol of National Quality (SNQ) for Specialty Medical Care, followed by a Bronze Award in 2023.

MacKay Children's Hospital, founded in 2014, continues to uphold the spirit of Dr. MacKay by serving numerous patients with rare diseases both domestically and internationally. Last year marked the tenth anniversary of our hospital, and under the guidance of our Director, Professor Hsiang-Yu Lin, we established the International Rare Disease Center. Our aim is to extend our care for rare diseases globally and enhance the visibility of MacKay Children's Hospital.


Today's International Forum on Rare Diseases brings together experts from around the world to discuss the latest advancements in medical care and research. We extend a special welcome to our international experts and hope you create wonderful memories during your stay in Taiwan. Wishing everyone a successful and fruitful conference. Thank you.

Kind regards,

Ming-Ren Chen, MD  
Professor and Superintendent  
MacKay Children's Hospital



## WELCOME MESSAGE



Dear attendees, guests, and colleagues,


Welcome to the 2025 International Forum on Rare Diseases at MacKay Children's Hospital! This is a gathering where the shining minds assemble to share the latest developments in medical care and research in the field. A heartfelt thank-you to all the speakers, many of you have traveled long distance to be with us for this special occasion. I would like to extend my sincere appreciation for all the attendees who take the time to make this Forum a unique and successful one.

I believe that a friendly atmosphere fosters collaboration and innovation. The goal of the Forum is to ensure that you feel rewarding and inspired throughout the meeting. The organizing team has invited a fantastic lineup of experts to give special talks with a diverse spectrum of topics covering important aspects in rare diseases, ranging from unmet needs in lysosomal storage diseases to pioneering newborn screening and therapeutic modalities, all aimed at enriching our collective understanding and propelling innovations in better care for individuals with rare disorders.

Your esteemed presence is deeply valued, and please accept my sincere gratitude for your commitment to this gathering. I am very sure that your expertise and engagement will contribute to a vibrant and productive exchange of ideas. I look forward to meeting and working with you to shape the future of rare disease management.

Warm regards,

Shuan-Pei Lin, MD  
Founding Director of Rare Disease Center  
MacKay Memorial Hospital  
Professor and Senior Attending Physician  
MacKay Children's Hospital  
Department of Medicine  
MacKay Medical College  
Taipei, Taiwan



## WELCOME MESSAGE



Welcome to the 2025 MacKay Children's Hospital International Forum on Rare Diseases, jointly organized by MacKay Memorial Hospital, MacKay Children's Hospital, and the Taiwan Pediatric Association. We are honored to have you here in Taipei for a full day of meaningful presentations and dialogue around innovation and care in the field of rare diseases.

MacKay Children's Hospital was founded in 2014. To date, it has served many patients with rare diseases in Taiwan and abroad, and accepts referrals from various branches of the MacKay system, other hospitals and clinics. It has carried out the spirit of Dr. MacKay and helped to improve the medical standards of rare diseases in the country. The purpose of holding the "2025 Mackay Children's Hospital International Forum on Rare Diseases" is to discuss and share the latest medical research results in this field. The forum will bring together experts and scholars from Taiwan and abroad to promote academic exchanges and advance the development of research cooperation and clinical applications of rare diseases. The conference will cover the latest trends in diagnosis and treatment of rare diseases and develop into the application of genetic diagnostic technology in personalized medicine. It is hoped that through this forum, the medical community will increase its attention to and ability to care for rare diseases.

We sincerely welcome you and hope you enjoy the conference.

Kind regards,

Hsiang-Yu Lin, MD, PhD  
Professor and Director  
International Rare Disease Center  
MacKay Memorial Hospital  
Taipei, Taiwan  
Director of the Organizing Committee  
2025 MacKay Children's Hospital International  
Forum on Rare Diseases

