恭賀！林翔宇教授受邀出席在韓國首爾舉辦之〔2026 亞洲兒科研究學會〕[The Asian Society for Pediatric Research (ASPR 2026 SEOUL)] ，並發表〔台灣黏多醣症與高雪氏症新生兒篩檢〕之專題演講。

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林翔宇教授





S18-03

A Blueprint for Universal LSD Screening: Strategic Insights from Taiwan's Decades of Experience with MPS and Gaucher Disease



Hsiang-Yu Lin[1,2,3,4,5*], Chih-Kuang Chuang[2,3], Chung-Lin Lee[1,2,3,4,5], Yuan-Rong Tu[2], Yun-Ting Lo[3], Ya-Hui Chang[1,3], Jun-Yi Wu[3], Hsin-Yun Liu[6], Hsiao-Jan Chen[6], Li-Yun Wang[7], Huey-Jane Ho[7], Shuan-Pei Lin[1,2,3,4]

[1]Department of Pediatrics, MacKay Memorial Hospital, Taipei, Taiwan
[2]Division of Genetics and Metabolism, Department of Medical Research, MacKay Memorial Hospital, Taipei, Taiwan
[3]International Rare Disease Center, MacKay Memorial Hospital, Taipei, Taiwan
[4]Department of Medicine, MacKay Medical University, New Taipei City, Taiwan
[5]Department of Early Childhood Care and Education, MacKay Junior College of Medicine, Nursing and Management, Taipei, Taiwan
[6]The Chinese Foundation of Health, Neonatal Screening Center, Taipei, Taiwan
[7]Taipei Institute of Pathology, Neonatal Screening Center, Taipei, Taiwan

Lysosomal storage diseases (LSDs) represent a complex group of rare genetic disorders that, while individually infrequent, collectively impose a significant public health burden. Early intervention is paramount, as many LSDs are progressive and irreversible; however, the "diagnostic odyssey" often delays life-altering treatment. This lecture presents a strategic blueprint for universal LSD screening, drawing on over twenty years of pioneering experience in Taiwan with Mucopolysaccharidosis (MPS) and Gaucher disease. Taiwan's journey serves as a global benchmark for integrating rare disease screening into public health infrastructure. Our analysis identifies three critical pillars of a successful universal program. The first pillar is **Technological Innovation**. The transition from traditional enzymatic assays to high-throughput tandem mass spectrometry (MS/MS) and digital microfluidics has enabled cost-effective, large-scale newborn screening (NBS). Secondly, **Confirmatory Infrastructure**. Establishing a seamless pipeline from initial dried blood spot (DBS) suspicion to genetic validation and biomarker monitoring is essential to minimize parental anxiety and false positives. Furthermore, **Sustainable Policy Frame**works. Taiwan's success is rooted in robust legislation—such as the Prevention of Rare Diseases and Orphan Drug Act—which ensures government-subsidized treatment and fosters a multi-disciplinary "care network" involving specialized centers. We examine specific longitudinal data from Taiwan's MPS and Gaucher cohorts, demonstrating that patients identified via universal screening exhibit significantly better clinical outcomes, including preserved organ function and improved growth trajectories, compared to those diagnosed symptomatically. Furthermore, we address the ethical and economic challenges of screening for late-onset variants and the necessity of "watchful waiting" protocols. By synthesizing these strategic insights, this blueprint provides a scalable roadmap for healthcare systems worldwide to implement universal LSD screening, ultimately shifting the paradigm from reactive management to proactive, life-saving intervention.

Curriculum Vitae

Name	Hsiang-Yu Lin
Country	Taiwan
Organization	MacKay Memorial Hospital
Current Position	(1) Director, International Rare Disease Center, MacKay Memorial Hospital, Taipei, Taiwan (2) Director, Division of Genetics and Metabolism, Department of Pediatrics, MacKay Children's Hospital, Taipei, Taiwan (3) Director, Department of Medical Research, MacKay Children's Hospital, Taipei, Taiwan (4) Professor, Department of Medicine, MacKay Medical University, New Taipei City, Taiwan



Educational Background

- Institute of Clinical Medicine, National Yang-Ming University, Taipei, Taiwan-PhD degree
- School of Medicine, Taipei Medical University, Taipei, Taiwan

Professional Experiences

- Postdoctoral researcher, Ohio State University, USA
- Board certified specialist in Pediatrics by the Taiwan Pediatric Association
- Board certified specialist in Medical Genetics and Metabolism, Taiwan Pediatric Association
- Board certified specialist in Pediatric Emergency Medicine, Taiwan Pediatric Association

Professional Organizations

- Board certified specialist in Pediatric Emergency Medicine, Taiwan Pediatric Association
- Member of Taiwan Pediatric Association (No. 4247)
- Member of Association of Medical Genetics and Metabolism, Taiwan (No. 046)
- Member of Taiwan Human Genetics Society (No. G0202)

Main Scientific Publications

(1) Chuang CK, Tu YR, Lee CL, Lo YT, Chang YH, Wu JY, Liu MY, Liu HY, Chen HJ, Kao SM, Wang LY, Ho HJ, Hsiang-Yu Lin* (co-corresponding author), Lin SP*. Nationwide Newborn Screening for Mucopolysaccharidoses in Taiwan: Impact, Early Diagnosis, and Clinical Advances over the Past Decade. Genet Med. 2026. Accept. (SCI)
(2) Lee CL, Chang SW, Fang HH, Chuang CK, Chiu HC, Chang YH, Tu YR, Lo YT, Wu JY, Hsiang-Yu Lin* (co-corresponding author), Lin SP*. Systematic Analysis of Multiple Imaging Modalities in Infants Diagnosed with Mucopolysaccharidosis by Newborn Screening. Diagnostics (Basel). 2025 Apr 12;15(8):980. (SCI)
(3) Hsiang-Yu Lin, Lee CL, Chang YH, Tu YR, Lo YT, Wu JY, Niu DM, Liu MY, Liu HY, Chen HJ, Kao SM, Wang LY, Ho HJ, Chuang CK, Lin SP. Implementation of Newborn Screening for Mucopolysaccharidosis Type IVA and Long-Term Monitoring in Taiwan. Genet Med. 2024 Oct 4;26(12):101286. (SCI)
(4) Chuang CK, Tu YR, Lee CL, Lo YT, Chang YH, Liu MY, Liu HY, Chen HJ, Kao SM, Wang LY, Ho HJ, Hsiang-Yu Lin* (co-corresponding author), Lin SP*. Updated Confirmatory Diagnosis for Mucopolysaccharidoses in Taiwanese Infants and the Application of Gene Variants. Int J Mol Sci. 2022 Sep 1;23(17):9979. (SCI)
(5) Chuang CK, Lee CL, Tu RY, Lo YT, Sisca F, Chang YH, Liu MY, Liu HY, Chen HJ, Kao SM, Wang LY, Ho HJ, Hsiang-Yu Lin* (co-corresponding author), Lin SP*. Nationwide newborn screening program for mucopolysaccharidoses in Taiwan and an update of the "gold standard" criteria required to make a confirmatory diagnosis. Diagnostics (Basel). 2021 Aug 31;11(9):1583. (SCI)
(6) Lee CL, Lee KS, Chuang CK, Su CH, Chiu HC, Tu RY, Lo YT, Chang YH, Hsiang-Yu Lin* (co-corresponding author), Lin SP*. Otorhinolaryngological Management in Taiwanese Patients with Mucopolysaccharidoses. Int J Med Sci. 2021 Jul 25;18(15):3373-3379. (SCI)
(7) Hsiang-Yu Lin, Chang YH, Lee CL, Tu YR, Lo YT, Hung PW, Niu DM, Liu MY, Liu HY, Chen HJ, Kao SM, Wang LY, Ho HJ, Chuang CK, Lin SP. Newborn Screening Program for Mucopolysaccharidosis Type II and Long-Term Follow-up of the Screen-Positive Subjects in Taiwan. J Pers Med. 2022;12(7):1023. (SCI)
(8) Hsiang-Yu Lin, Chen MR, Lee CL, Lin SM, Hung CL, Niu DM, Chang TM, Chuang CK, Lin SP. Aortic root dilatation in Taiwanese patients with mucopolysaccharidoses and the long-term effects of enzyme replacement therapy. Diagnostics (Basel). 2021;11(1):16. (SCI)
(9) Hsiang-Yu Lin, Chen MR, Lee CL, Lin SM, Hung CL, Niu DM, Chang TM, Chuang CK, Lin SP. Natural Progression of Cardiac Features and Long-term Effects of Enzyme Replacement Therapy in Taiwanese Patients with Mucopolysaccharidosis II. Orphanet J Rare Dis. 2021;16(1):99. (SCI)
(10) Hsiang-Yu Lin, Lee CL, Chang CY, Chiu PC, Chien YH, Niu DM, Tsai FJ, Hwu WL, Lin SJ, Lin JL, Chao MC, Chang TM, Tsai WH, Wang TJ, Chuang CK, Lin SP. Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985-2019). Orphanet J Rare Dis. 2020 Nov 7;15(1):314. (SCI)
(11) Lin CY, Hsiang-Yu Lin* (co-first author), Chuang CK, Zhang PH, Tu RY, Lin SP, Tsai HJ. Effect of Mutated ids Overexpression on IDS Enzyme Activity and Developmental Phenotypes in Zebrafish Embryos: A Valuable Index for Assessing Critical Point-Mutations Associated with Mucopolysaccharidosis Type II Occurrence in Humans. Diagnostics (Basel). 2020 Oct 21;10(10):E854. (SCI)
(12) Hsiang-Yu Lin, Chuang CK, Lee CL, Chen MR, Sung KT, Lin SM, Hou CJ, Niu DM, Chang TM, Hung CL, Lin SP. Cardiac Evaluation using Two-dimensional Speckle-tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. Diagnostics (Basel). 2020;10(2), 62. (SCI)
(13) Hsiang-Yu Lin, Tu RY, Chern SR, Lo YT, Fran S, Wei FJ, Huang SF, Tsai SY, Chang YH, Lee CL, Lin SP, Chuang CK. Identification and functional characterization of IDS gene mutations underlying Taiwanese Hunter Syndrome (mucopolysaccharidosis type II). Int J Mol Sci. 2020;21(1):114. (SCI)
(14) Lee CL, Hsiang-Yu Lin* (co-first author), Chuang CK, Chiu HC, Tu RY, Huang YH, Hwu WL, Tsai FJ, Chiu PC, Niu DM, Chen YJ, Chao MC, Chang TM, Lin JL, Chang CY, Kao YC, Lin SP. Functional independence of Taiwanese patients with mucopolysaccharidoses. Mol Genet Genomic Med. 2019 Aug;7(8):e790. (SCI)
(15) Hsiang-Yu Lin, Chen MR, Lin SM, Hung CL, Niu DM, Chang TM, Chuang CK, Lin SP. Cardiac Characteristics and Natural Progression in Taiwanese Patients with Mucopolysaccharidosis III. Orphanet J Rare Dis. 2019;14:140. (SCI)


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A Blueprint for Universal LSD Screening:
Strategic Insights from Taiwan's Decades of
Experience with MPS and Gaucher Disease
Hsiang-Yu Lin, MD, PhD


Outline
• Newborn screening (NBS) programs in Taiwan
• NBS for lysosomal storage diseases (LSDs) in Taiwan
(focus on Gaucher and MPSs)
國際罕見疾病中心
遺傳諮詢中心
黏多醣症
醫療照護中心
MPS Center
1滴血讓MPS現形
2015-10-23
2013-10-21
Pilot stud NBS for MPS I s
morial Hospital
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MacKay Memorial Hospital
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Strategic Insights from Taiwan's Decades of
Experience with MPS and Gaucher Disease
Hsiang-Yu Lin, MD, PhD
Department of Pediatrics, MacKay Memorial Hospital, Taipei 104217, Taiwan


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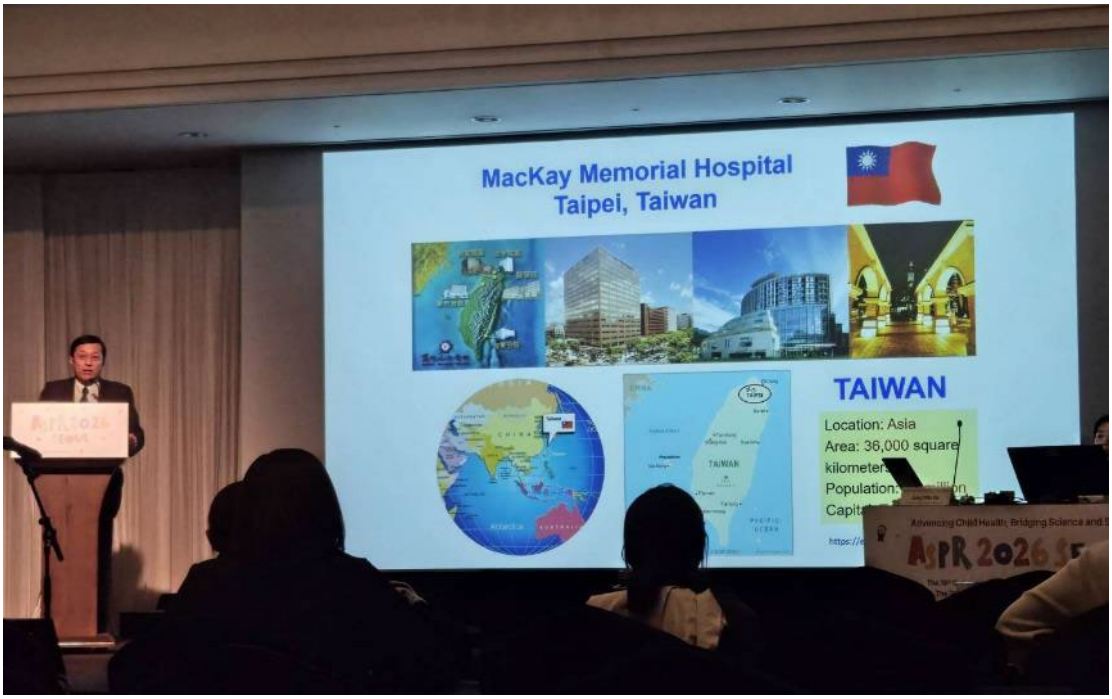
MacKay Memorial Hospital
Taipei, Taiwan
TAIWAN
Location: Asia
Area: 36,000 square
kilometers
Population:
Capita
Advancing Child Health, Bridging Science and
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羅允廷醫檢師



吳君儀醫檢師


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Dear Participants,

On behalf of the Organizing Committee, we would like to express our heartfelt gratitude for your participation in the 19th Asian Society for Pediatric Research Congress (ASPR 2026) & the 76th Annual Meeting of the Korean Pediatric Society on October 21–23, 2026, in Seoul, Korea.

Your participation and contributions made ASPR 2026 a memorable occasion for sharing knowledge, exchanging ideas, and building connections across the pediatric research community.

We hope that the congress provided valuable insights for your research and clinical practice, as well as meaningful opportunities to connect with colleagues from around the world. We also hope you took home fond memories of your time in Seoul.

Thank you once again for being part of ASPR 2026. We look forward to seeing you again at future ASPR congresses.

With warm regards,



Han-Suk Kim
Congress President, ASPR 2026
Chairman/CEO, The Korean Pediatric Society



Kyeongbae Park
Congress President,
The 76th Annual Meeting of the Korean Pediatric Society
President, The Korean Pediatric Society

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