

恭賀！林翔宇教授受邀出席在美國丹佛科羅拉多兒童醫院舉行之「國際黏多醣症新生兒篩檢會議」(International Modified Delphi Process for MPS IVA & VI NBS & Follow Up) 並發表「台灣黏多醣症新生兒篩檢」之專題演講。

詳情參閱：Children's Hospital Colorado | Children's Hospital Colorado

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Newborn Screening for MPS IVA and VI in Taiwan



Hsiang-Yu Lin, MD, PhD



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Simon A. Jones MBChB, BSc, MRCPCH, was born in Northern Ireland and attended Edinburgh Univ. Medical School, qualifying with a BSc in neurosciences and MBChB. Simon worked (2005-2026) at the Willink Biochemical Genetics Unit, now part of Genetic Medicine at St Mary's Hospital, Manchester. He has been a Consultant in paediatric inherited metabolic diseases, an honorary Prof. of paediatrics and translational medicine and principal investigator in many phase I-IV international multicentre trials of ERT, small molecules or gene therapies for lysosomal storage disorders. Since 2026 Simon has been a full time employee of Denali therapeutics working to develop CNS penetrant therapies for childhood neurodegenerative disorders. He is a medical advisor to the UK MPS society, Niemann-Pick UK and Cure GM1 patient advocacy groups.



Terri L Klein, MPA, is the President and CEO of the National MPS Society. Her background and graduate degrees are in Public Administration and Nonprofit. She has been a patient advocate for 25 years. Her role in advocacy has focused on family support and outreach, research, and newborn screening. In addition to advocacy, she has contributed to significant efforts around original research on the use of dried blood spots for newborn screening, policy issues relevant to biomarkers, accelerated approval, and advocacy's role in the RUSP nomination process.



Christina Lampe, MD, is the Director of the Centre for Rare Diseases within the Dept. of Pediatric Neurology, Muscular Diseases, and Social Pediatrics at the Univ. of Giessen. She trained at the Humboldt Univ. of Berlin (Charité), where she also completed her Surgical internship. In 2007, Christina transitioned to metabolic genetics, beginning work at Villa Metabolica at the Johannes Gutenberg Univ. of Mainz. With Prof. Maurizio Scarpa, they started the Centre for Rare Diseases in 2014 at the Dr. Horst Schmidt Kliniken in Wiesbaden, focusing on lysosomal related disorders. In Giessen since 2018, she cares for children and adults with lysosomal diseases and is a PI or co-PI of several clinical trials concerning lysosomal diseases.



Hsiang-Yu "Steven" Lin, MD. Dr. Hsiang-Yu Lin is an elite expert and medical specialist based in Taiwan, recognized for his extensive clinical and research work in lysosomal storage disorders, specifically the mucopolysaccharidoses. Serving as a pediatrician and researcher including roles at Mackay Memorial Hospital, his professional contributions focus on the diagnosis, newborn screening, genetic analysis, and long-term monitoring of rare metabolic conditions. In relation to, Steven has actively led and contributed to clinical investigations involving newborn screening programs, biochemical and enzymatic evaluations, and patient health outcomes MPS Type IVA (Morquio A syndrome) and Type VI (Maroteaux-Lamy syndrome).



Andrew McFadyen, BA, BEd, MHSc, HEC-C, is Ethics Lead for Precision Child Health at The Hospital for Sick Children in Toronto and an Assistant Prof. at the Univ. of Toronto's Dalla Lana School of Public Health. Formerly Executive Director of two rare-disease patient advocacy organizations, he has advanced research, launched clinical trials, and advised governments and pharmaceutical companies on access to novel therapies, expanded access, and scarce-resource allocation. Andrew has testified before U.S. Senate and Canadian parliamentary committees and contributes to NYU ethics working groups. His interest in rare disease began after his son Isaac was diagnosed with MPS VI.



Nationwide newborn screening for MPS I, II, IVA and VI in Taiwan (since 2015)



ARTICLE

Nationwide newborn screening for mucopolysaccharidoses in Taiwan: Impact, early diagnosis, and clinical advances over the past decade

Chih-Kuang Chuang^{1,2}, Yuan-Rong Tu¹, Chung-Lin Lee^{1,3,4}, Yun-Ting Lo⁵, Ya-Hui Chang⁶, Jun-Yi Wu⁷, Mei-Ying Liu⁸, Hsin-Yun Liu⁹, Hsiao-Jan Chen⁹, Shu-Min Kao⁹, Li-Yun Wang⁹, Huey-Jane Ho⁹, Hsiang-Yu Lin^{1,3,4,7,8,9,*}, Shuan-Pei Lin^{1,3,4,8,10,*}



Chuang et al. *Genet Med*. 2026 May 7;28(7):102596.

https://youtu.be/d8kJBXL20rA?si=24xNOY00ji_uZNJI

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Implementation of Newborn Screening for MPS IVA and Long-Term Monitoring in Taiwan

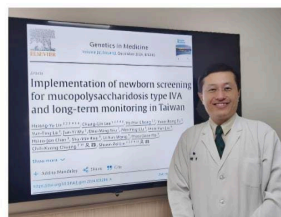
- Mucopolysaccharidosis IVA (MPS IVA) is a rare lysosomal storage disorder caused by *N*-acetylgalactosamine-6-sulfatase (**GALNS**) deficiency, leading to the accumulation of glycosaminoglycans (GAGs) in the lysosomes of many tissues and organs, leading to progressive cellular dysfunction.
- An **MPS IVA newborn screening program** has been available in Taiwan since September 2019.



ARTICLE

Implementation of newborn screening for mucopolysaccharidosis type IVA and long-term monitoring in Taiwan

Hsiang-Yu Lin^{1,2,3,4,5,6,*}, Chung-Lin Lee^{1,2,3,4,5}, Ya-Hui Chang^{1,3}, Yuan-Rong Tu², Yun-Ting Lo³, Jun-Yi Wu⁷, Dau-Ming Niu⁸, Mei-Ying Liu⁹, Hsin-Yun Liu⁹, Hsiao-Jan Chen⁹, Shu-Min Kao⁹, Li-Yun Wang⁹, Huey-Jane Ho⁹, Chih-Kuang Chuang^{2,10,*}, Shuan-Pei Lin^{1,2,3,9,11,*}



Lin et al. *Genet Med*. 2024 Oct 3:101286.

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THANKS FOR YOUR ATTENTION!



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