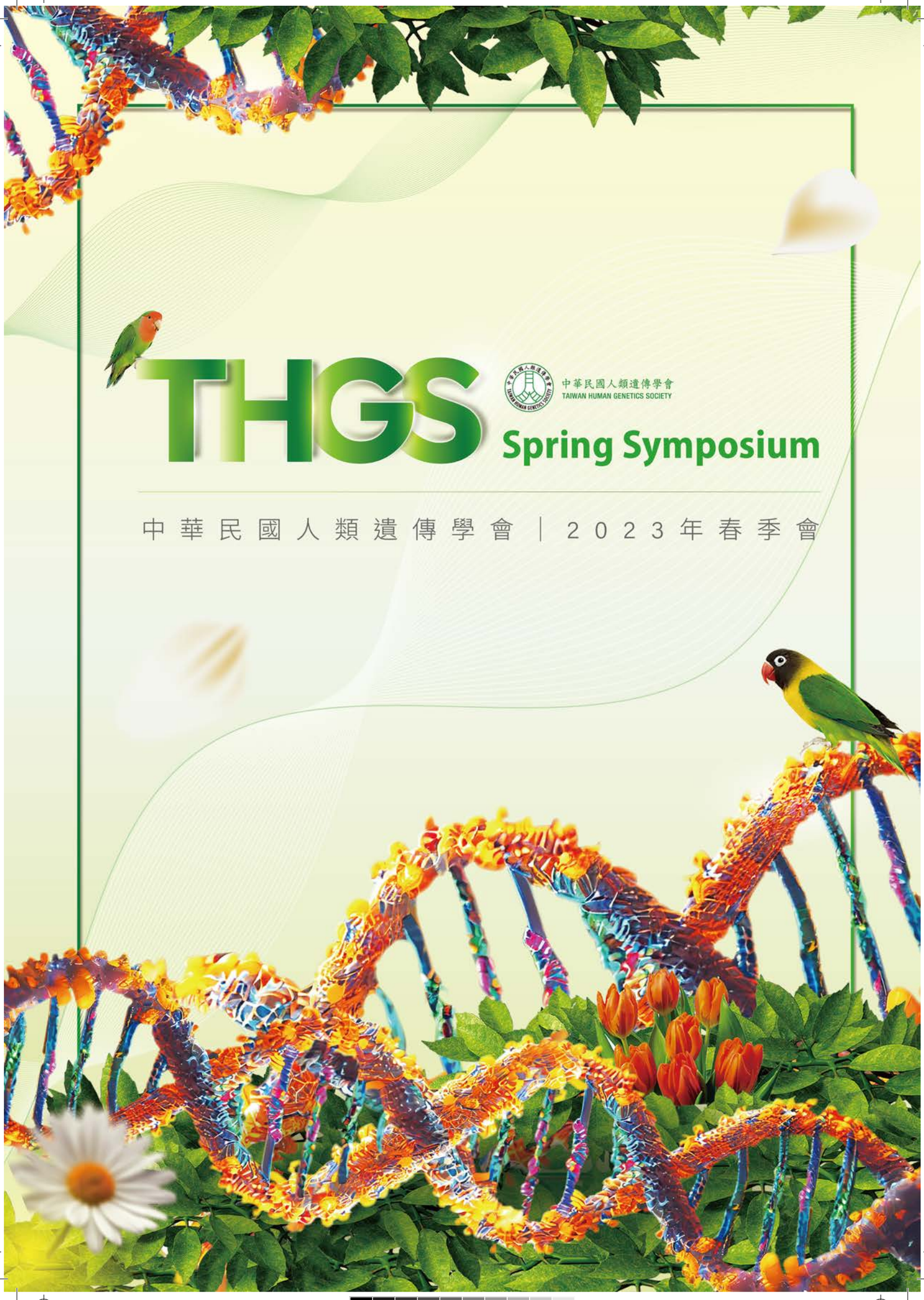

# Table of Contents

# MEMO

# 2023 THGS Spring Symposium

**LOCATION:** NTUH International Convention Center (NTHCC)
No. 2, Xu-zhou Rd., Zhong-zheng District, Taipei City, 100

| April 30th (SUNDAY) | | | |
|---|---|---|---|
| **Time** | **Topic** | **Speaker / Panelists** | **Moderator** |
| 08:20-08:50 | Registration | | |
| 08:50-09:00 | Opening Remarks | **蔡輔仁 理事長** | |
| **09:00-11:00** | **Session I - The Challenges of Next Generation Sequencing (NGS) from Multiple Perspectives** | | |
| 09:00-09:35 | The Challenges of NGS from the Perspective of Ethical Practice | **蔡甫昌 教授** | **蔡立平 主任** |
| 09:35-10:10 | The Challenges of NGS from the Perspective of Legal Practice | **吳振吉 教授** | |
| 10:10-10:45 | The Challenges of NGS from the Perspective of Consulting Practice | **鄭逸如 主任** | |
| 10:45-11:05 | Panel Discussion | | |
| **11:05-11:25** | **Coffee Break** | | |
| 11:25-12:00 | 2023 THGS General Assembly | **林翔宇 秘書長** | |
| **12:00-13:00** | **Lunch Time & Voting** | | |
| **13:00-14:40** | **Session II - New Trends in the Newborn Screening Program** | | |
| 13:00-13:35 | What's new in newborn screening | **Prof. Wendy Chung** | **胡務亮 教授** |
| 13:35-14:10 | Review and Prospect of Newborn Screening-Taiwan Experience | **簡穎秀 教授** | |
| 14:10-14:40 | Panel Discussion | | |
| **14:40-15:00** | **Coffee Break** | | |
| **15:00-17:00** | **Session III - New Trends in the Diagnosis and Treatment of Gaucher Disease (25th Anniversary)** | | |
| 15:00-15:35 | Venglustat Clinical Trial Update –A New Era Toward Promising Gaucher Disease Therapy | **Prof. Pramod Mistry** | **林炫沛 教授** |
| 15:35-16:10 | Molecular mechanism underlying ERT unresponsiveness in Gaucher disease | **Prof. Beom Hee Lee** | |
| 16:10-16:45 | Changing Clinical Manifestations of Gaucher Disease in Taiwan | **李妮鍾 教授** | |
| 16:45-17:00 | Panel Discussion | | |
| 17:00-17:10 | Closing Remark | **蔡輔仁 理事長** | |

# President of THGS


## Fuu-Jen Tsai 蔡輔仁

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## Present Position

中國醫藥大學副校長暨特聘教授

## Education

MD, China Medical University, Taichung, Taiwan
Ph.D., Institute of Chinese Medicine, China Medical University

## Brief Chronology of Employment

| | |
|---|---|
| 1998-Present | Chief, Department of Medical Genetics, China Medical University Hospital |
| 2001/03-2003/10 | Chief, Department of Pediatrics, China Medical University Hospital |
| 2001/10-Present | Professor of Pediatrics, China Medical University |
| 2002/03-2005/03 | President, Taiwan Human Genetics Society |
| 2003/10-2009/10 | Dean, College of Chinese Medicine, China Medical University |
| 2006/8-Present | Chief, Genetic Center, China Medical University Hospital |
| 2009/08-2015/08 | Dean, Office of Research and Development, China Medical University |
| 2009/08-Present | Chief, Department of Medical Research, China Medical University Hospital |
| 2013/08-Present | Vice President, China Medical University |
| 2015/08-Present | Distinguished Professor of Pediatrics, China Medical University |
| 2020/07-Present | President, Taiwan Human Genetics Society |

## Selective Publications


1. Chiou JS, Cheng CF, Liang WM, Chou CH, Wang CH, Lin WD, Chiu ML, Cheng WC, Lin CW, Lin TH, Liao CC, Huang SM, Tsai CH, Lin YJ, Tsai FJ*. Your height affects your health: genetic determinants and health-related outcomes in Taiwan. BMC Med. 2022 Jul 13;20(1):250.
2. Chang CY, Chiang AJ, Lai MT, Yan MJ, Tseng CC, Lo LC, Wan L, Li CJ, Tsui KH, Chen CM, Hwang T, Tsai FJ*, Sheu JJ. A More Diverse Cervical Microbiome Associates with Better Clinical Outcomes in Patients with Endometriosis: A Pilot Study. Biomedicines. 2022 Jan 14;10(1):174.
3. Cheng CF, Liao KY, Lee KJ, Tsai FJ*. A Study to Evaluate Accuracy and Validity of the EFAI Computer-Aided Bone Age Diagnosis System Compared With Qualified Physicians. Front Pediatr. 2022 Apr 8;10:829372.
4. Liao WL, Liu TY, Cheng CF, Chou YP, Wang TY, Chang YW, Chen SY, Tsai FJ*. Analysis of HLA Variants and Graves' Disease and Its Comorbidities Using a High Resolution Imputation System to

Examine Electronic Medical Health Records. Front Endocrinol (Lausanne). 2022 Mar 7;13:842673.

5. Lin CY, Chang YS, Liu TY, Huang CM, Chung CC, Chen YC, Tsai FJ*, Chang JG, Chang SJ. Genetic contributions to female gout and hyperuricemia using genome-wide association study and polygenic risk score analyses. Rheumatology (Oxford). 2022 Jun 27:keac369.
6. Mahajan A, Spracklen CN, Zhang W, Ng MCY, Petty LE, Kitajima H, Yu GZ, Rüeger S, Speidel L, Kim YJ, Horikoshi M, Mercader JM, Taliun D, Moon S, Kwak SH, Robertson NR, Rayner NW, Loh M, Kim BJ, Chiou J, Miguel-Escalada I, Della Briotta Parolo P, Lin K, Bragg F, Preuss MH, Takeuchi F, Nano J, Guo X, Lamri A, Nakatochi M, Scott RA, Lee JJ, Huerta-Chagoya A, Graff M, Chai JF, Parra EJ, Yao J, Bielak LF, Tabara Y, Hai Y, Steinthorsdottir V, Cook JP, Kals M, Grarup N, Schmidt EM, Pan I, Sofer T, Wuttke M, Sarnowski C, Gieger C, Nousome D, Trompet S, Long J, Sun M, Tong L, Chen WM, Ahmad M, Noordam R, Lim VJY, Tam CHT, Joo YY, Chen CH, Raffield LM, Lecoeur C, Prins BP, Nicolas A, Yanek LR, Chen G, Jensen RA, Tajuddin S, Kabagambe EK, An P, Xiang AH, Choi HS, Cade BE, Tan J, Flanagan J, Abaitua F, Adair LS, Adeyemo A, Aguilar-Salinas CA, Akiyama M, Anand SS, Bertoni A, Bian Z, Bork-Jensen J, Brandslund I, Brody JA, Brummett CM, Buchanan TA, Canouil M, Chan JCN, Chang LC, Chee ML, Chen J, Chen SH, Chen YT, Chen Z, Chuang LM, Cushman M, Das SK, de Silva HJ, Dedoussis G, Dimitrov L, Doumatey AP, Du S, Duan Q, Eckardt KU, Emery LS, Evans DS, Evans MK, Fischer K, Floyd JS, Ford I, Fornage M, Franco OH, Frayling TM, Freedman BI, Fuchsberger C, Genter P, Gerstein HC, Giedraitis V, González-Villalpando C, González-Villalpando ME, Goodarzi MO, Gordon-Larsen P, Gorkin D, Gross M, Guo Y, Hackinger S, Han S, Hattersley AT, Herder C, Howard AG, Hsueh W, Huang M, Huang W, Hung YJ, Hwang MY, Hwu CM, Ichihara S, Ikram MA, Ingelsson M, Islam MT, Isono M, Jang HM, Jasmine F, Jiang G, Jonas JB, Jørgensen ME, Jørgensen T, Kamatani Y, Kandeel FR, Kasturiratne A, Katsuya T, Kaur V, Kawaguchi T, Keaton JM, Kho AN, Khor CC, Kibriya MG, Kim DH, Kohara K, Kriebel J, Kronenberg F, Kuusisto J, Läll K, Lange LA, Lee MS, Lee NR, Leong A, Li L, Li Y, Li-Gao R, Ligthart S, Lindgren CM, Linneberg A, Liu CT, Liu J, Locke AE, Louie T, Luan J, Luk AO, Luo X, Lv J, Lyssenko V, Mamakou V, Mani KR, Meitinger T, Metspalu A, Morris AD, Nadkarni GN, Nadler JL, Nalls MA, Nayak U, Nongmaithem SS, Ntalla I, Okada Y, Orozco L, Patel SR, Pereira MA, Peters A, Pirie FJ, Porneala B, Prasad G, Preissl S, Rasmussen-Torvik LJ, Reiner AP, Roden M, Rohde R, Roll K, Sabanayagam C, Sander M, Sandow K, Sattar N, Schönherr S, Schurmann C, Shahriar M, Shi J, Shin DM, Shriner D, Smith JA, So WY, Stančáková A, Stilp AM, Strauch K, Suzuki K, Takahashi A, Taylor KD, Thorand B, Thorleifsson G, Thorsteinsdottir U, Tomlinson B, Torres JM, Tsai FJ, Tuomilehto J, Tusie-Luna T, Udler MS, Valladares-Salgado A, van Dam RM, van Klinken JB, Varma R, Vujkovic M, Wacher-Rodarte N, Wheeler E, Whitsel EA, Wickremasinghe AR, van Dijk KW, Witte DR, Yajnik CS, Yamamoto K, Yamauchi T, Yengo L, Yoon K, Yu C, Yuan JM, Yusuf S, Zhang L, Zheng W; FinnGen; eMERGE Consortium, Raffel LJ, Igase M, Ipp E, Redline S, Cho YS, Lind L, Province MA, Hanis CL, Peyser PA, Ingelsson E, Zonderman AB, Psaty BM, Wang YX, Rotimi CN, Becker DM, Matsuda F, Liu Y, Zeggini E, Yokota M, Rich SS, Kooperberg C, Pankow JS, Engert JC, Chen YI, Froguel P, Wilson JG, Sheu WHH, Kardia SLR, Wu JY, Hayes MG, Ma RCW, Wong TY, Groop L, Mook-Kanamori DO, Chandak GR,

Collins FS, Bharadwaj D, Paré G, Sale MM, Ahsan H, Motala AA, Shu XO, Park KS, Jukema JW, Cruz M, McKean-Cowdin R, Grallert H, Cheng CY, Bottinger EP, Dehghan A, Tai ES, Dupuis J, Kato N, Laakso M, Köttgen A, Koh WP, Palmer CNA, Liu S, Abecasis G, Kooner JS, Loos RJF, North KE, Haiman CA, Florez JC, Saleheen D, Hansen T, Pedersen O, Mägi R, Langenberg C, Wareham NJ, Maeda S, Kadowaki T, Lee J, Millwood IY, Walters RG, Stefansson K, Myers SR, Ferrer J, Gaulton KJ, Meigs JB, Mohlke KL, Gloyn AL, Bowden DW, Below JE, Chambers JC, Sim X, Boehnke M, Rotter JI, McCarthy MI, Morris AP. Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation. Nat Genet. 2022 May;54(5):560-572.

7. Lin WD, Cheng CF, Wang CH, Liang WM, Chen CH, Hsieh AR, Chiu ML, Lin TH, Liao CC, Huang SM, Tsai CH, Chang CY, Lin YJ, Tsai FJ*. Genetic factors of idiopathic central precocious puberty and their polygenic risk in early puberty. Eur J Endocrinol. 2021 Aug 27;185(4):441-451.
8. Cheng CF, Lin YJ, Lin MC, Liang WM, Chen CC, Chen CH, Wu JY, Lin TH, Liao CC, Huang SM, Hsieh AR, Tsai FJ*. Genetic risk score constructed from common genetic variants is associated with cardiovascular disease risk in type 2 diabetes mellitus. J Gene Med. 2021 Feb;23(2):e3305.
9. Chiu ML, Liang WM, Li JP, Cheng CF, Chiou JS, Ho MW, Wu YC, Lin TH, Liao CC, Huang SM, Tsai FJ*, Lin YJ. Timing, Dosage, and Adherence of Antiretroviral Therapy and Risk of Osteoporosis in Patients With Human Immunodeficiency Virus Infection in Taiwan: A Nested Case-Control Study. Front Pharmacol. 2021 Apr 30;12:631480
10. Cheng J, Liu HP, Lin WY, Tsai FJ*. Machine learning compensates fold-change method and highlights oxidative phosphorylation in the brain transcriptome of Alzheimer's disease. Sci Rep. 2021 Jul 1;11(1):13704.
11. Huang YC, Chang YW, Cheng CW, Wu CM, Liao WL, Tsai FJ*. Causal Relationship between Adiponectin and Diabetic Retinopathy: A Mendelian Randomization Study in an Asian Population. Genes (Basel). 2020 Dec 24;12(1):17.
12. Huang YC, Chang YW, Cheng CW, Wu CM, Liao WL, Tsai FJ*. Causal Relationship between Adiponectin and Diabetic Retinopathy: A Mendelian Randomization Study in an Asian Population. Genes (Basel). 2020 Dec 24;12(1):17.
13. Chang WS, Tsai CW, Yang JS, Hsu YM, Sh ih LC, Chiu HY, Bau DT, Tsai FJ*. Resveratrol inhibited the metastatic behaviors of cisplatin-resistant human oral cancer cells via phosphorylation of ERK/p-38 and suppression of MMP-2/9. J Food Biochem. 2021 Jun;45(6):e13666.
14. Cheng CF, Hsieh AR, Liang WM, Chen CC, Chen CH, Wu JY, Lin TH, Liao CC, Huang SM, Huang YC, Ban B, Lin YJ, Tsai FJ*. Genome-Wide and Candidate Gene Association Analyses Identify a 14-SNP Combination for Hypertension in Patients With Type 2 Diabetes. Am J Hypertens. 2021 Jun 22;34 (6):651-661.
15. Chen CJ, Chiu ML, Hung CH, Liang WM, Ho MW, Lin TH, Liu X, Tsang H, Liao CC, Huang SM, Wu YF, Wu YC, Li TM, Tsai FJ*, Lin YJ. Effect of Xanthium Strumarium on HIV-1 5'-LTR Transcriptional Activity and Viral Reactivation in Latently Infected Cells. Front Pharmacol. 2021 Aug 6;12:720821.
16. Chen YT, Lin WD, Liao WL, Tsai YC, Liao JW, Tsai FJ*. NT5C2 methylation regulatory interplay

between DNMT1 and insulin receptor in type 2 diabetes. Sci Rep. 2020 Sep 30;10(1):16087.

17. Spracklen CN, Horikoshi M, Kim YJ, Lin K, Bragg F, Moon S, Suzuki K, Tam CHT, Tabara Y, Kwak SH, Takeuchi F, Long J, Lim VJY, Chai JF, Chen CH, Nakatochi M, Yao J, Choi HS, Iyengar AK, Perrin HJ, Brotman SM, van de Bunt M, Gloyn AL, Below JE, Boehnke M, Bowden DW, Chambers JC, Mahajan A, McCarthy MI, Ng MCY, Petty LE, Zhang W, Morris AP, Adair LS, Akiyama M, Bian Z, Chan JCN, Chang LC, Chee ML, Chen YI, Chen YT, Chen Z, Chuang LM, Du S, Gordon-Larsen P, Gross M, Guo X, Guo Y, Han S, Howard AG, Huang W, Hung YJ, Hwang MY, Hwu CM, Ichihara S, Isono M, Jang HM, Jiang G, Jonas JB, Kamatani Y, Katsuya T, Kawaguchi T, Khor CC, Kohara K, Lee MS, Lee NR, Li L, Liu J, Luk AO, Lv J, Okada Y, Pereira MA, Sabanayagam C, Shi J, Shin DM, So WY, Takahashi A, Tomlinson B, Tsai FJ, van Dam RM, Xiang YB, Yamamoto K, Yamauchi T, Yoon K, Yu C, Yuan JM, Zhang L, Zheng W, Igase M, Cho YS, Rotter JI, Wang YX, Sheu WHH, Yokota M, Wu JY, Cheng CY, Wong TY, Shu XO, Kato N, Park KS, Tai ES, Matsuda F, Koh WP, Ma RCW, Maeda S, Millwood IY, Lee J, Kadowaki T, Walters RG, Kim BJ, Mohlke KL, Sim X. Identification of type 2 diabetes loci in 433,540 East Asian individuals. Nature. 2020 Jun;582(7811):240-245.
18. Huang CP, Lin YW, Huang YC, Tsai FJ*. Mitochondrial Dysfunction as a Novel Target for Neuroprotective Nutraceuticals in Ocular Diseases. Nutrients. 2020 Jun 30;12(7):1950.
19. Lan YC, Wang YH, Chen HH, Lo SF, Chen SY, Tsai FJ*. Effects of Casein Kinase 2 Alpha 1 Gene Expression on Mice Liver Susceptible to Type 2 Diabetes Mellitus and Obesity. Int J Med Sci. 2020 Jan 1;17(1):13-20.
20. Cheng J, Liu HP, Lin WY, Tsai FJ*. Identification of contributing genes of Huntington's disease by machine learning. BMC Med Genomics. 2020 Nov 23;13(1):176.

**SESSION I: The Challenges of Next Generation Sequencing (NGS) from Multiple Perspectives**

## MODERATOR


### Li-Ping Tsai 蔡立平

Tel：+886-2-66289779 ext 5710
E-mail：tsaiped@gmail.com


### Present Position

Taipei Tzu Chi Hospital and Tzu Chi University

### Education

| | |
|---|---|
| 1981-1988 | School of Medicine, Taipei Medical University |

### Brief Chronology of Employment

| | |
|---|---|
| 07/1992 ~ Present | Senior Attending Physician, Department of Pediatrics, Mackay Memorial Hospital, Taipei, Taiwan |
| 07/1994 ~ Present | Director, Division of Genetics & Metabolism, Department of Medical Research, Mackay Memorial Hospital, Taipei, Taiwan |
| 02/1998 ~ Present | Board Director, Down Syndrome Foundation of R.O.C., Taipei, Taiwan |
| 07/2000 ~ Present | Committee Member, Committee of Early Intervention Program, MacKay Memorial Hospital, Taipei, Taiwan |
| 06/2004 ~ Present | Member of Medical Consulting Committee, International Prader-Willi Syndrome Organization (IPWSO) |
| 11/2005 ~ Present | Committee Member, Committee of Academic Affairs, Taipei Medical Association, Taipei, Taiwan |
| 01/2014 ~ Present | Professor, Department of Medicine, Mackay Medical College and Department of Infant and Child Care, National Taipei University of Nursing and Health Sciences |
| 03/2017 ~ Present | Taiwan Foundation of Rare Disorders, Taipei, Taiwan |
| 1989-1993 | Pediatric Resident, Taipei Municipal Women/Children Hospital |
| 1993-1995 | Medical Genetic Fellow, National Taiwan University Hospital |
| 1996-2005 | Visiting Staff in Pediatrics, Taipei Municipal Women/Children Hospital |
| 1999-2000 | Visiting Scientist, Department of Human Genetics, UCLA, USA |
| 2005-2010 | Visiting Staff in Pediatrics, Taipei Tzu Chi Hospital |
| 2009-2020 | Deputy Director, Department of Medical Education, Taipei Tzu Chi Hospital |
| 2010-2020 | Director, Department of Pediatrics, Taipei Tzu Chi Hospital |
| 2013-2021 | Assistant Professor, Department of Pediatrics, School of Medicine, Tzu Chi University |
| 2022-Present | Associate Professor, Department of Pediatrics, School of Medicine, Tzu Chi University |
| 2020-Present | Director, Medical Genetic Center, Taipei Tzu Chi Hospital |

## Awards

| | |
|---|---|
| 2013 | New Taipei City Medical Dedication Award |
| 2020 | Certificate for Symbol of National Quality「Holistic Health Care for Prader Willi Syndrome」 |

## Selective Publications


1. Chang SJ, Tsai LP, Hsu CK, Yang SS. Elevated postvoid residual urine volume predicting recurrence of urinary tract infections in toilet-trained children. Pediatr Nephrol. 2015 Jul;30(7):1131-7. doi: 10.1007/s00467-014-3009-y. Epub 2015 Feb 12. (SCI)
2. Lan MC, Hsu YB, Lan MY, Chiu TJ, Huag TT, Wong SB, Chen YC, Tsai LP. Drug-induced sleep endoscopy in children with Prader-Willi syndrome. Sleep Breath.2016 Sep;20(3):1029-34. Doi:10.1007/s11325-016-1338-8. Epub 2016 Apr 8. (SCI) (Correspondence)
3. Tzeng CC, Tsai LP, Chang YK, Humg YJ, Chang YY, Su YP, Jiang JJ, Liang HM. A 15-year-long southern blotting of FMR1 to detect female carriers and for prenatal diagnosis of fragile X syndrome in Taiwan. Clinic Genetics, 2017 Aug;92(2):217-220. (SCI)
4. Lin YJ, Liao WL, Wang CH, Tsai LP, Tang CH, Chen CH, Wu JY, Liang WM, Hsieh AR, Cheng CF, Chen JH, Chien WK, Lin TH, Wu CM, Liao CC, Huang SM, Tsai FJ. Association of human height-related genetic variants with familial short stature in Han Chinese in Taiwan. Sci Rep. 2017 Jul 25;7 (1):6372. doi: 10.1038/s41598-017-06766-z. (SCI)
5. Chiu VJ, Tsai LP, Wei JT, Tzeng IS, Wu HC. Motor performance in Prader-Willi syndrome patients and its potential influence on caregiver's quality of life. Peer J. 2017 Dec 13;5:e4097. doi: 10.7717/peer j.4097. eCollection 2017. (SCI)
6. Lee CL, Lin HY, Tsai LP, Chiu HC, Tu RY, Huang YX, Chien YH, Lee NC, Niu DM, Chao MC, Tsai FJ, Chou YY, Chuang CK, Lin SP. Functional independence of Taiwanese children with Prader-Willi syndrome. Am J Med Genet, 2018. doi: 10.1002/ajmg.a.38705. (SCI) (Co-first Author)
7. Chen FC, Ho SY, Tsai LP. Effect of aerobic dance training in adults with Prader-Willi Syndrome: A Pilot study. Taiwan J Phys Med Rehabil 2018;46(2):91-9. http://doi: 10.6315/TJPMR.201812_46 (2).0005 (Correspondence)
8. Wong SB, Zhao LL, Tsai WH, Yu CH, Tsai LP. Is prone sleeping dangerous for neonate? Polysomographic characteristics and NDN gene analysis. Tzu Chi Med J 2019;31(2):113-7 (Correspondence)
9. Chen SH, Kuo YT, Tsai LP, Liu YL, Miser JS. A 2-year-old boy with pancytopenia caused due to nutritional cobalamin deficiency. Pediatr Neonatol. 2019 Aug;53(4):264-8. http://doi.org/ 10.1016/j.pedneo.2019.06.004. Epub 2019 Jul 22. (SCI)
10. Chiu V, Chou SH, Wu HC, Tzeng IS, Chao CY, Huang PJ, Tsai LP. Elastic band training improves adiposity and physical performance in adults with Prader-Willi syndrome: A pilot study. Taiwan J Phys Med Rehabil 2019;47:133-143. (Correspondence)
11. Wang TS, Tsai WH, Tsai LP, Wong SB. Clinical characteristics and epilepsy in genomic imprinting disorders: Angelman syndrome and Prader-Willi syndrome. Tzu Chi Med J 2020;32(2):137-144

12. Lin YJ, Cheng CF, Wang CH, Liang WM, Tang CH, Tsai LP, Chen CH, Wu JY, Hsieh AR, Lee MT, Lin TH, Liao CC, Huang SM, Zhang Y, Tsai CH, Tsai FJ. Genetic architecture associated with familial short stature. J Clin Endocrinol Metab 2020;105(6):1801-1813 (SCI)
13. Wu RN, Hung WC, Chen CT, Tsai LP, Lai WS, Min MY, Wong SB. Firing activity of locus coeruleus noradrenergic neurons decreases in necdin-deficienct mice, an animal model of Prader Willi syndrome. J Neurodev Disord. 2020;12(1):21. doi: 10.1186/s11689-020-09323-4.(SCI)
14. Wong SB, Wang TS, Tsai WH, Tzeng IS, Tsai LP. Parenting stress in families of children with Prader-Willi syndrome. Am J Med Genet A. 2021 Jan;185(1):83-89. doi: 10.1002/ajmg.a.61915. (SCI) (Correspondence)
15. Chao TC, Yang SS, Chang SJ, Tsai LP. High prevalence of lower urinary tract dysfunction in patients with Prader-Willi syndrome. Neurourol Urodyn. 2021 Apr;40(4):1063-1068. doi: 10.1002/nau.24669. (SCI) (Correspondence)
16. Lin CM, Yang JH, Lee HJ, Lin YP, Tsai LP, Hsu CS, Luxton GWG, Hu CF. Whole Exome Sequencing Identifies a Novel Homozygous Missense Mutation in the CSB Protein-Encoding ERCC6 Gene in a Taiwanese Boy with Cockayne Syndrome. Life (Basel). 2021 Nov 14;11(11):1230. doi: 10.3390/life11111230. (SCI)
17. Lin HY, Lee CL, Fran S, Tu RY, Chang YH, Niu DM, Chang CY, Chiu PC, Chou YY, Hsiao HP, Tsai MC, Chao MC, Tsai LP, Yang CF, Su PH, Pan YW, Lee CH, Chu TH, Chuang CK, Lin SP. Epigenotype, Genotype, and Phenotype Analysis of Taiwanese Patients with Silver-Russell Syndrome. J Pers Med. 2021 Nov 13;11(11):1197. doi: 10.3390/jpm11111197.(SCI)
18. Tsai LP, Wang SS, Chee SY, Wong SB. Dynamic Changes in the Quantitative Electroencephalographic Spectrum During Attention Tasks in Patients With Prader-Willi Syndrome. Front Genet. 2022 Mar 16;13:763244. doi: 10.3389/fgene.2022.763244. (SCI)
19. Wong SB, Yang MC, Tzeng IS, Tsai WH, Lan CC, Tsai LP. Progression of Obstructive Sleep Apnea Syndrome in Pediatric Patients with Prader-Willi Syndrome. Children (Basel). 2022 Jun 17;9 (6):912. doi: 10.3390/children9060912. (SCI) (Correspondence)
20. Wu MJ, Tsai LP, Lai TF, Cho JS, Liao Y. Accelerometer-Measured Physical Activity and Sedentary Behavior of Adults with Prader-Willi Syndrome Attending and Not Attending a Small-Scale Community Workshop. Int J Environ Res Public Health. 2022 Jul 25;19(15):9013. doi: 10.3390/ijerph19159013.(SCI)
21. Tsai LP, Tzeng ST, Hsieh TH, Li YC, Hung SS. Scoliosis and BMI in patients with Prader-Willi syndrome. J Pediatr Orthop B. 2022 Nov 14. doi: 10.1097/BPB.0000000000001031.(SCI)

**The Challenges of NGS from the Perspective of Ethical Practice**

## SPEAKER


# Daniel Fu-Chang Tsai 蔡甫昌

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E-mail：fctsai@ntu.edu.tw


## Present Position

Professor, Department & Graduate Institute of Medical Education and Bioethics, National Taiwan University College of Medicine
Attending Physician, Department of Medical Research, National Taiwan University Hospital
Director, Ethics Center of NTUH.
Director, Center for Biomedical Ethics, National Taiwan University
Head of Taiwan Unit, International Chair in Bioethics
Research Fellow, Institute for Advanced Studies in the Humanities and Social Sciences, NTU
Hastings Center Fellow
Member, Merck Ethics Advisory Panel

## Education

| | |
|---|---|
| 1999 | Ph.D. in Health care ethics, Center for Social Ethics and Policy, The University of Manchester, United Kingdom |
| 1989 | M.D., National Taiwan University College of Medicine, Taiwan, ROC |

## Brief Chronology of Employment

| | |
|---|---|
| 2014-Present | Professor, Department & Research Institute of Medical Education & Bioethics, National Taiwan University College of Medicine |
| 2010-2014 | Director, Department of Social Medicine, National Taiwan University College of Medicine |
| 2013-2014 | Professor, Department of Social Medicine, National Taiwan University College of Medicine |
| 2005-2013 | Associate Professor, Department of Social Medicine, National Taiwan University College of Medicine |
| 2001-2005 | Assistant Professor, Department of Social Medicine & Jointly Appointed at Department of Family Medicine, National Taiwan University College of Medicine |
| 2002-Present | Attending Physician, Department of Medical Research, National Taiwan University Hospital |

| | |
|---|---|
| 2001 | Lecturer, Department of Social Medicine, National Taiwan University College of Medicine |
| 1994-2001 | Attending Physician, Department of Family Medicine, National Taiwan University Hospital |
| 1992-1994 | Director & Physician, Jinshan Township Public Health Center |
| 1989-1992 | Resident, Department of Family Medicine, National Taiwan University Hospital |

## Awards

| | |
|---|---|
| 2015 | Honorary Membership by the UNESCO Chair of Bioethics |
| 2016-2017 | Vice President of the International Association of Bioethics |
| 2019 | Goldman-Berland Lectureship in Palliative Medicine, USA |
| 2015-2018 | Convener of the Medical Education Discipline, and Final reviewer of the Emerging and Other Fields Discipline, Department of Science and Education Development and International Cooperation, Ministry of Science and Technology |
| 2007, 2008, 2018 | Award for Excellent Innovation Teaching Material, National Taiwan University Hospital |

## Selective Publications


1. Chen,I .A., Tsai, D.F.C. : Proposal to Amend the Regulation of Recruiting Study Subjects. Formosan Journal of Medicine 2022;26(5):600-606.
2. Kuo,T.Y., Tsai, D.F.C. : Ethical Considerations in Rewarding and Mandatory Strategies for Vaccination. Formosan Journal of Medicine 2022;26(5):573-581.
3. Hsueh, F.C., Tsai, D.F.C. : The Scientific Evidences and Ethical Consideration for Vaccine Allocation. Formosan Journal of Medicine 2022;26(4):455-465.
4. Tsai, D.F.C. , Chou, Y.C., Wang Hung, C.Y., Huang ,M.C.: The Legislation Efforts and Ethical Considerations of Gestational Surrogacy in Taiwan. Formosan Journal of Medicine 2022;26(3):261-270.
5. Tsai, D.F.C. , Chou, Y.C., Wu,M.S. : Medical Violence and Safety of Health care Provider in Pandemic. Cheng Ching Medical Journal 2022;18(2):4-8.
6. Wu,Y.C., Tai,C.F., Tsai, D.F.C. , Chan,K,A.: Optimization of Ethics Review of Clinical Trials in Taiwan. Formosan Journal of Medicine 2022;26(2):228-236.
7. Lee,S.C., Chou, Y.C.,Tsai, D.F.C.: Shared decision-making between doctors and patients and clinical ethics. Formosan Journal of Medicine 2022;26(1):84-89.
8. Tsai, D.F.C. ,Foo, K.F., Wang, C.H., Juang, Y.C., Fu, L.C.:Ethical issues of caring for robots. Formosan Journal of Medicine 2021;25(4):441-452.
9. Tsai, D.F.C., Foo, K.F., Juang, Y.C., Huang, L.M.: Vaccine Allocation and Ethics in Pandemic. Formosan Journal of Medicine 2021;25(1):1-23.
10. Tsai, D.F.C., Juang, Y.C., Chien, Y.H., Lee, N.C., Wang Hung, C.Y., Lin, S.P., Hwu, W.L.: The Ethics and Guidelines of Next-Generation Sequencing Genetic Testing and Counseling. Formosan Journal of Medicine 2020;24(2):125-144.

## The Challenges of NGS from the Perspective of Ethical Practice

Author: Daniel Fu-Chang Tsai

Abstract:
Next-generation sequencing (NGS) has been widely applied in genetic research as well as developed for personalized medicine. However, because NGS sequences all genetic information for individual, it also raises many ethical concerns among which informed consent is essential to the protection of study subjects and patients. Two NGS expert meetings were held in July and August 2016 to understand the current application of NGS in Taiwan, and its possible impact and ethical issues in research and clinical practice. Based on the expert meeting, we have developed a guideline and standard formats for the informed consent for NGS Study, a document which is also useful for research ethics committee/Institutional Review Board review. In this guideline, researchers explain whether and how they plan to disclose primary findings, incidental findings and variants of uncertain significance (VUS) to study subjects. The informed consent form for study subjects reflects the informing plan of researchers and their right for choosing to know or not to know. We believed these two documents reflect the current practical research environment in Taiwan while still meeting international ethical requirements. By using these documents, researchers in Taiwan can be better prepared for the informed consent process while study subjects are offered more detailed information to reduce possible misunderstanding and increase autonomy and protection for them in NGS genetic studies.

**The Challenges of NGS from the Perspective of Legal Practice**

## SPEAKER


## Chen-Chi Wu 吳振吉

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### Present Position

Professor, Department of Otolaryngology, National Taiwan University College of Medicine

### Education

| | |
|---|---|
| 1992-1999 | Medical School: School of Medicine, National Taiwan University College of Medicine, Taipei, Taiwan |
| 2002-2004 | Master of Medical Science: Graduate Institute of Clinical Medicine, National Taiwan University College of Medicine, Taipei, Taiwan |
| 2004-2010 | PhD: Graduate Institute of Clinical Medicine, National Taiwan University College of Medicine, Taipei, Taiwan |
| 2012-2013 | Research fellow: Dr. Stankovic’ s Lab, Department of Otolaryngology, Massachusetts Eye and Ear Infirmary, Harvard Medical School, Boston, USA |
| 2008-2011 | LL.M: Graduate Institute of Law, National Chengchi University College of Law, Taipei, Taiwan |
| 2011-2020 | PhD: Graduate Institute of Law, National Taiwan University College of Law, Taipei, Taiwan |

### Brief Chronology of Employment

| | |
|---|---|
| 2000-2004 | Resident, Department of Otolaryngology, National Taiwan University College of Medicine, Taipei, Taiwan |
| 2004- | Attending Physician, Department of Otolaryngology, National Taiwan University Hospital, Taipei, Taiwan |
| 2006- | Attending Physician, Department of Medical Genetics, National Taiwan University Hospital, Taipei, Taiwan |
| 2019- | Professor, Department of Otolaryngology, National Taiwan University College of Medicine, Taipei, Taiwan |
| 2019- | Adjunct Professor, Graduate Institute of Clinical Medicine, National Taiwan University College of Medicine, Taipei, Taiwan |
| 2020- | Director, Departments of Otolaryngology and Medical Research, National Taiwan University Hospital Hsin-Chu Branch, Hsinchu, Taipei, Taiwan |

## Selective Publications


1. 吳振吉、姜世明。論醫療契約不完全給付可歸責性要件之舉證責任－兼評最高法院 97 年台上第 1000 號民事判決。輔仁法學，第 44 期，2012 年 12 月，91-158 頁。(TSSCI 期刊論文)
2. 吳振吉、姜世明。醫師及醫療機構就債務不履行責任之法律關係－兼評最高法院 99 年度台上字第 1055 號民事判決、臺灣高等法院 99 年度醫上更(一)字第 3 號民事判決。臺北大學法學論叢，第 86 期，2013 年 6 月，1-50 頁。(TSSCI期刊論文)
3. 吳振吉。論交通事故與醫療過失競合理論：以日本法為借鑑。政大法學評論，第 150 期，2017 年 9 月，49-111 頁。(TSSCI 期刊論文)
4. 吳振吉。醫療事故損害賠償請求權之消滅時效－從兩件以消滅時效為核心爭點的實務案例談起。臺大法學論叢，第 47 卷第 1 期，2018 年 3 月，345-404 頁。(TSSCI 期刊論文)
5. 吳振吉。人工智慧醫療傷害之損害賠償責任。臺大法學論叢，第 51 卷第 2 期，2022 年 6 月，477-536 頁。(TSSCI 期刊論文)

## The Challenges of NGS from the Perspective of Legal Practice

Author: Chen-Chi Wu

Abstract:
Recent advances in genomic medicine, especially next-generation sequencing (NGS) technology, have revolutionized the clinical management of diseases in terms of diagnosis, counseling, and treatment. However, the high development of NGS technology also brings challenges to the existing legal system. Who should enjoy the fruitful results of genome research, and how should it be managed? Can we screen out healthy fetuses through the interleaved application of genetics and genomics, so that human beings can avoid the hidden dangers of diseases in advance? When using genetic testing technology, one should pay attention to how the right to privacy should be protected, and does the protected object include family members? In this talk, I will attempt to address the topics listed above.

**The Challenges of NGS from the Perspective of Consulting Practice**

## SPEAKER


## Yih-Ru Cheng 鄭逸如

Tel：+886-2-2312-3456 ext. 67171
E-mail：yrcheng@ntuh.gov.tw


### Present Position

Director and Clinical Psychologist, Clinical Psychology Center, National Taiwan University Hospital

### Education

Ph.D. (Department of Psychology, National Taiwan University)

### Brief Chronology of Employment

| | |
|---|---|
| 1989- | Clinical Psychologist, National Taiwan University Hospital |
| 2007- | Director, Clinical Psychology Center, National Taiwan University Hospital |

### Awards

| | |
|---|---|
| 2013- | 台大醫院「教學優良獎」 |
| 2017, 2018, 2021 | 台大醫院「教材著作優良獎」 |
| 2018 | 中華民國臨床心理師公會全國聯合會「傑出人士貢獻獎」 |

### Selective Publications


1. 鄭逸如、曾嫦嫦（主編）曾嫦嫦、吳治勳、吳文珺、陳思臻、陳品樺、陳奕靜、洪家暐、張煥、洪瑞可、簡靖維、洪國倫、鄭逸如（合著）（2021）。癌症病人術前心理衛教團體手冊－臨床的現場與實務。ISBN：978-986-522-514-8。臺北市，五南圖書出版公司。
2. 鄭逸如、曾嫦嫦（主編）鄭逸如、曾嫦嫦、張琦郁、楊于婷、黃揚文、李素貞、黃柏蒼（合著）（2018）。心理腫瘤照護的實務與解析—生命交會中的療癒契機。ISBN：978-957-11-9666-4。臺北市，五南圖書出版公司。
3. 鄭逸如、何雪綾、陳秀蓉（2017）。醫病溝通之鑰－醫療人員同理心五大心法。ISBN：978-957-11-9060-0。臺北市，五南圖書出版公司。
4. Yun-Hsin Huang, Chih-Hsun Wu, Hsiu-Jung Chen, Yih-Ru Cheng, Fu-Chien Hung, Kai-Kuan Leung, Bee-Horng Lue, Ching-Yu Chen, Tai-Yuan Chiu, Yin-Chang Wu (2018). Quick Screening Tool for Patients with Severe Negative Emotional Reactions to Chronic Illness—Psychometric Study of the Negative Emotions due to Chronic Illness Screening Test (NECIS). Family Practice, 35(1), 34-40.

## The Challenges of NGS from the Perspective of Psychological Assessment and Counseling
## 從心理衡鑑與心理諮商的觀點看次世代定序的挑戰

Author: Yih-Ru Cheng 鄭逸如

Abstract:
曾有針對基因檢測進行15年長期追蹤的研究論文指出，接受檢測的個案對他們的決定都表示滿意，且沒有個案發生像遭遇創傷災難般的反應，但前提是這些基因檢測針對每位個案都做了量身訂製的服務，因為準備接受檢測與聽取結果是複雜的心理歷程，是否適時擁有適切支持是很關鍵的影響因素。如今，當次世代定序能帶來更多、更快的基因訊息，於是人們便可能期待更有機會、更迅速獲得更具體準確的資訊，但這也使心理層面的需求或問題更需被關注與協助，例如：對檢測前與後之相關心理衝擊與可能效應的認知、對自身心理強度與因應能力的檢視與強化、對獲知結果後可能的身心家庭社會反應的瞭解與預備、對處理實際適應問題所需資源的盤點與增納等。演講內容預計整理與省思這些層面的心理衡鑑與心理諮商，期待在享有先進醫療科技的時代裡，透過跨領域合作，持續落實與促進以人為本的全人醫療。

## Secretary general of THGS


# Hsiang-Yu Lin 林翔宇

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## Present Position

Director, Division of Genetics and Metabolism, Department of Pediatrics, MacKay Memorial Hospital, Taipei, Taiwan
Director, Rare Disease Center, MacKay Memorial Hospital, Taipei, Taiwan

## Education

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

## Brief Chronology of Employment

| | |
|---|---|
| 2010/07-2011/06 | Postdoctoral researcher, Ohio State University, USA |
| 2006/07-Present | Attending Physician, Department of Pediatrics, MacKay Memorial Hospital, Taipei, Taiwan |
| 2015/10-Present | Director, Division of Genetics and Metabolism, Department of Pediatrics, MacKay Memorial Hospital, Taipei, Taiwan |
| 2020/10-Present | Director, Rare Disease Center, MacKay Memorial Hospital, Taipei, Taiwan |
| 2020/02-Present | Professor, Department of Medicine, MacKay Medical College, New Taipei City, Taiwan and MacKay Junior College of Medicine, Nursing and Management, Taipei, Taiwan |

## Board Certification

Licensed Medical Doctor by the Department of Health, Taiwan, R.O.C. (License No. 029736).
Board certified specialist in Pediatrics by the Taiwan Pediatric Association (Certificate No. 003064).
Board certified specialist in Medical Genetics and Metabolism, Taiwan Pediatric Association (Certificate No. 045)

## Selective Publications


1. Lee CL, Chuang CK, Chiu HC, Tu RY, Lo YT, Chang YH, **Hsiang-Yu Lin* (co-corresponding author)**, Lin SP*. Wiedemann-Steiner Syndrome with a Pathogenic Variant in *KMT2A* from Taiwan. ***Children (Basel).*** 2021 Oct 22;8(11):952. (SCI)

2. **Hsiang-Yu Lin**, Lee CL, Fran S, Tu RY, Chang YH, Niu DM, Chang CY, Chiu PC, Chou YY, Hsiao HP, Yang CF, Tsai MC, Chu TH, Chuang CK, Lin SP. Quantitative DNA Methylation Analysis and Epigenotype-Phenotype

Correlations in Taiwanese Patients with Beckwith-Wiedemann Syndrome. ***J Pers Med.*** 2021 Oct 22;11 (11):1066. (SCI)

3. **Hsiang-Yu Lin**, Lee CL, Fran S, Tu RY, Chang YH, Niu DM, Chang CY, Chiu PC, Chou YY, Hsiao HP, Tsai MC, Chao MC, Tsai LP, Yang CF, Su PH, Pan YW, Lee CH, Chu TH, Chuang CK, Lin SP. Epigenotype, Genotype, and Phenotype Analysis of Taiwanese Patients with Silver-Russell Syndrome. ***J Pers Med.*** 2021 Nov 13;11 (11):1197. (SCI)

4 Lee CL, Lin SP, Niu DM*, **Hsiang-Yu Lin* (co-corresponding author)**. Fabry Disease and the Effectiveness of Enzyme Replacement Therapy (ERT) in Left Ventricular Hypertrophy (LVH) Improvement: A Review and Meta-Analysis. ***Int J Med Sci.*** 2022 Jan 1;19(1):126-131. (SCI)

5. Chen CP, Chern SR, Lin CH, Hsu CY, **Hsiang-Yu Lin**, Wu FT, Chen SW, Wang W. Detection of hypermethylation at H19DMR at amniocentesis in a fetus with overgrowth, distended abdomen and Beckwith-Wiedemann syndrome. ***Taiwan J Obstet Gynecol.*** 2021 Nov;60(6):1103-1106. (SCI)

6. Lee CL, Chuang CK, Chiu HC, Tu RY, Lo YT, Chang YH, Lin SP*, **Hsiang-Yu Lin* (co-corresponding author)**. Clinical Utility of Elosulfase Alfa in the Treatment of Morquio A Syndrome. ***Drug Des Devel Ther.*** 2022 Jan 10;16:143-154. (SCI)

7. Lee CL, Chuang CK, Tu RY, Chiu HC, Lo YT, Chang YH, Chen YJ, Chou, CL Wu PS, Chen CP, **Hsiang-Yu Lin* (co-corresponding author)**, Lin SP*. Increased Diagnostic Yield of Array Comparative Genomic Hybridization for Autism Spectrum Disorder in One Institution in Taiwan. ***Medicina.*** 2022, 58, 15. (SCI)

8. Lee CL, Lin SP, Niu DM*, **Hsiang-Yu Lin* (co-corresponding author)**. Fabry Disease and the Effectiveness of Enzyme Replacement Therapy (ERT) in Left Ventricular Hypertrophy (LVH) Improvement: A Review and Meta-Analysis. ***Int J Med Sci.*** 2022 Jan 1;19(1):126-131. (SCI)

9. Lee CL, Chuang CK, Chiu HC, Tu RY, Lo YT, Chang YH, Lin SP*, **Hsiang-Yu Lin* (co-corresponding author)**. Clinical Utility of Elosulfase Alfa in the Treatment of Morquio A Syndrome. ***Drug Des Devel Ther.*** 2022 Jan 10;16:143-154. (SCI)

10. Lee CL, Lin SM, Chen MR, Chuang CK, Syu YM, Chiu HC, Tu RY, Lo YT, Chang YH, **Hsiang-Yu Lin* (co-corresponding author)**, Lin SP*. Long-Term Cardiovascular Findings in Williams Syndrome: A Single Medical Center Experience in Taiwan. ***J Pers Med.*** 2022 May 18;12(5):817. (SCI)

11. Lee YH, Hsieh LC, Su CH, **Hsiang-Yu Lin**, Lin SP, Lee KS. Airway Management of the Deformed Trachea Using T-Tube Stents in Patients with Mucopolysaccharidosis Type IVA. ***Ann Otol Rhinol Laryngol.*** 2022 May;131 (5):562-566. (SCI)

12. Syu YM, Lee HC, Chang JH, Lee CL, Chuang CK, Chiu HC, Chang YH, **Hsiang-Yu Lin* (co-corresponding author)**, Lin SP*. Rapid weight loss and severe failure to thrive mimicking lipodystrophy syndrome in a 1-year-old Taiwanese girl with Costello syndrome. ***Children (Basel).*** 2022 Jun 16;9(6):905. (SCI)

13. **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 Jun 21;12(7):1023. (SCI)

14. Hung YC, Cheng KY, **Hsiang-Yu Lin**, Lin SP, Yang CY, Liu SC. Surgical Strategy to Decrease the Revision Rate of Fassier–Duval Nailing in the Lower Limbs of Osteogenesis Imperfecta. ***J Pers Med.*** 2022 Jul 15;12(7):1151. (SCI)

15. Lin CY, **Hsiang-Yu Lin* (co-first author)**, Chuang CK, Zhang PH, Tu RY, Lin SP, Tsai HJ. Quantification of Idua Enzymatic Activity Combined with Observation of Phenotypic Change in Zebrafish Embryos Provide a Preliminary Assessment of Mutated *idua* Correlated with Mucopolysaccharidosis Type I. ***J Pers Med.*** 2022 Jul 23;12(8):1199. (SCI)

16. Syu YM, Lee CL, Chuang CK, Chiu HC, Chang YH, **Hsiang-Yu Lin* (co-corresponding author)**, Lin S-P*. Functional Independence of Taiwanese Children with Osteogenesis Imperfecta. ***J Pers Med.*** 2022 Jul 24;12 (8):1205. (SCI)

17. Lee CL, Chuang CK, Syu YM, Chiu HC, Tu YR, Lo YT, Chang YH, **Hsiang-Yu Lin* (co-corresponding author)**, Lin S-P*. Efficacy and Safety of Intravenous Elosulfase Alfa for Mucopolysaccharidosis Type IVA: A Systematic Review and Meta-Analysis. ***J Pers Med.*** 2022, 12, 1338. (SCI)

18. Lao HC, Lin YC, Liang ML, Yang YW, Huang YH, Chan YL, Hsu YW, Lin SP, Chuang CK, Cheng JK*, **Hsiang-Yu Lin* (co-corresponding author)**. The Anesthetic Strategy for Patients with Mucopolysaccharidoses: A Retrospective Cohort Study. ***J Pers Med.*** 2022, 12, 1343. (SCI)

19. 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 S-P*. Updated Confirmatory Diagnosis for Mucopolysaccharidoses in Taiwanese Infants and the Application of Gene Variants. ***Int J Mol Sci.*** 2022, 23, 9979. (SCI)

20. Syu YM, Ma JY, Ou TH, Lee CL, **Hsiang-Yu Lin**, Lin SP, Lee CJ, Chen CP. De Novo Mosaic 6p23-p25.3 Tetrasomy Caused by a Small Supernumerary Marker Chromosome Presenting Trisomy Distal 6p Phenotype: A Case Report and Literature Review. ***Diagnostics (Basel).*** 2022 Sep 24;12(10):2306. (SCI)

21. Lin HY, Chang SY, Teng HH, Wu HJ, Li HY, Cheng CC, Chuang CK, **Hsiang-Yu Lin**, Lin SP, Cheng WC*. Discovery of small-molecule protein stabilizers toward exogenous alpha-l-iduronidase to reduce the accumulated heparan sulfate in mucopolysaccharidosis type I cells. ***Eur J Med Chem.*** 2022 Dec 9;247:115005.

**Session II - New Trends in the Newborn Screening Program**

## MODERATOR


# Paul Wuh-Liang Hwu 胡務亮

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E-mail：hwuwlntu@ntu.edu.tw


## Present Position

國立台灣大學醫學院小兒科教授：2010/08 起 ( 教 018017)

## Education

| | |
|---|---|
| 1977/09-1984/0 | 國立台灣大學醫學院、醫學系醫學士 |
| 1992/09-1997/07 | 國立台灣大學醫學院、分子醫學研究所博士 |

## Brief Chronology of Employment

| | |
|---|---|
| 1986/07-1989/06 | 台大醫院小兒部住院醫師 |
| 1990- | 台大醫院小兒部主治醫師 |
| 1991-2005 | 國立台灣大學醫學院小兒科講師及助理教授 |
| 2005/08-2010/07 | 國立台灣大學醫學院小兒科副教授 |
| 1989/07-1990/06 | 美國約翰霍浦金斯大學遺傳學系研究員 |
| 2000/07-2000/09 | 美國梅爾醫學中心遺傳學系研究員 |

## Selective Publications


1. Hwu WL, Chien YH. Development of Newborn Screening for Pompe Disease. Int J Neonatal Screen. 2020 Jan 24;6(1):5.
2. Chien YH, Lee NC, Chen PW, Yeh HY, Gelb MH, Chiu PC, Chu SY, Lee CH, Lee AR, Hwu WL. Newborn screening for Morquio disease and other lysosomal storage diseases: results from the 8-plex assay for 70,000 newborns. Orphanet J Rare Dis. 2020 Feb 3;15(1):38.
3. Wu ET, Hwu WL, Chien YH, Hsu C, Chen TF, Chen NQ, Chou HC, Tsao PN, Fan PC, Tsai IJ, Lin SP, Hsieh WS, Chang TM, Chen CN, Lee CH, Chou YY, Chiu PC, Tsai WH, Hsiung HC, Lai F, Lee NC. Critical Trio Exome Benefits In-Time Decision-Making for Pediatric Patients With Severe Illnesses. Pediatr Crit Care Med. 2019 Nov;20(11):1021-1026.
4. Tseng CH, Chien YH, Lee NC, Hsu YC, Peng SF, Tseng WI, Hwu WL. Gene therapy improves brain white matter in aromatic l-amino acid decarboxylase deficiency. Ann Neurol. 2019 May;85(5):644-652.
5. Lee NC, Chien YH, Hwu WL. A review of aromatic l-amino acid decarboxylase (AADC) deficiency in Taiwan. Am J Med Genet C Semin Med Genet. 2019 Jun;181(2):226-229.

6. Lee NC, Hwu WL, Muramatsu SI, Falk DJ, Byrne BJ, Cheng CH, Shih NC, Chang KL, Tsai LK, Chien YH. A Neuron-Specific Gene Therapy Relieves Motor Deficits in Pompe Disease Mice. Mol Neurobiol. 2018 Jun;55(6):5299-5309.
7. Chien YH, Lee NC, Tseng SH, Tai CH, Muramatsu S, Byrne BJ, Hwu WL. Efficacy and safety of AAV2 gene therapy in children with aromatic L-amino acid decarboxylase deficiency: an open-label, phase 1/2 trial. Lancet Child Adolesc Health 2017 Dec; 1: 265–73
8. Chien YH, Chiang SC, Weng WC, Lee NC, Lin CJ, Hsieh WS, Lee WT, Jong YJ, Ko TM, Hwu WL. Presymptomatic Diagnosis of Spinal Muscular Atrophy Through Newborn Screening. J Pediatr. 2017 Nov;190:124-129.e1
9. Lee NC, Lee YM, Chen PW, Byrne BJ, Hwu WL. Mutation-adapted U1 snRNA corrects a splicing error of the dopa decarboxylase gene. Hum Mol Genet. 2016 Dec 1;25(23):5142-5147
10. Lee NC, Muramatsu S, Chien YH, Liu WS, Wang WH, Cheng CH, Hu MK, Chen PW, Tzen KY, Byrne BJ, Hwu WL. Benefits of Neuronal Preferential Systemic Gene Therapy for Neurotransmitter Deficiency. Mol Ther. 2015 Oct;23(10):1572-81.
11. Chien YH, Lee NC, Chen CA, Tsai FJ, Tsai WH, Shieh JY, Huang HJ, Hsu WC, Tsai TH, Hwu WL. Long-term prognosis of patients with infantile-onset Pompe disease diagnosed by newborn screening and treated since birth. J Pediatr. 2015 Apr;166(4):985-91.
12. Lee NC, Chien YH, Hu MH, Liu WS, Chen PW, Wang WH, Tzen KY, Byrne BJ, Hwu WL. Treatment of Congenital Neurotransmitter Deficiencies by Intracerebral Ventricular Injection of an Adeno-Associated Virus Serotype 9 Vector. Hum Gene Ther. 2014 Mar;25(3):189-98.
13. Lee NC, Shieh YD, Chien YH, Tzen KY, Yu IS, Chen PW, Hu MH, Hu MK, Muramatsu S, Ichinose H, Hwu WL. Regulation of the dopaminergic system in a murine model of aromatic L-amino acid decarboxylase deficiency. Neurobiol Dis. 2013 Apr;52:177-90.
14. Hwu WL, Muramatsu S, Tseng SH, Tzen KY, Lee NC, Chien YH, Snyder RO, Byrne BJ, Tai CH, Wu RM. Gene therapy for aromatic L-amino acid decarboxylase deficiency. Sci Transl Med. 2012 May 16;4(134):134ra61.
15. Chien YH, Lee NC, Huang HJ, Thurberg BL, Tsai FJ, Hwu WL. Later-onset Pompe disease: early detection and early treatment initiation enabled by newborn screening. J Pediatr. 2011 Jun;158(6):1023-1027.e1.
16. Hwu WL, Chien YH, Lee NC, Chiang SC, Dobrovolny R, Huang AC, Yeh HY, Chao MC, Lin SJ, Kitagawa T, Desnick RJ, Hsu LW. Newborn screening for Fabry disease in Taiwan reveals a high incidence of the later-onset GLA mutation c.936+919G>A (IVS4+919G>A). Hum Mutat. 2009 Oct;30(10):1397-405.
17. Chien YH, Lee NC, Thurberg BL, Chiang SC, Zhang XK, Keutzer J, Huang AC, Wu MH, Huang PH, Tsai FJ, Chen YT, Hwu WL. Pompe disease in infants: improving the prognosis by newborn screening and early treatment. Pediatrics. 2009 Dec;124(6):e1116-25.

**What's new in newborn screening**

## SPEAKER


# Wendy Chung

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## Present Position

| | |
|---|---|
| 2020/01-Present | Chief of Clinical Genetics Columbia University |
| 2019/01-Present | Medical co-director, Genetic Counseling Graduate Program, Columbia University |
| 2019/01-Present | Associate Director for Education, Herbert Irving Comprehensive Cancer Center, Columbia University |
| 2017/02-Present | Affiliate Member of the New York Genome Center |
| 2017/03-Present | Kennedy Family Professor of Pediatrics in Medicine, with tenure Columbia University |
| 2014/02-Present | Director of Precision Medicine Resource, Irving Institute for Translational Research Columbia University |
| 2010/07-Present | Co-director, Medical Genetics Training Fellowship Columbia University |

## Education

| | |
|---|---|
| 1990/09-1998/05 | Cornell University Medical College, NY M.D. |
| 1992/06-1996/05 | Rockefeller University, NY Ph.D. Genetics of Non-insulin dependent diabetes in rodents and man (Dr. Rudolph Leibel) |
| 1986/09-1990/05 | Cornell University, Ithaca, NY B.A. Biochemistry; Economics |

## Brief Chronology of Employment

| | |
|---|---|
| 2015/09-2017/03 | Kennedy Family Associate Professor of Pediatrics in Medicine, with tenure Columbia University |
| 2013/02-2015/08 | Associate Professor of Pediatrics in Medicine, with tenure Columbia University |
| 2006/07-2017/06 | Director, Molecular and Cytogenetics Fellowship Program Columbia University |
| 2003/01-2013/02 | Director of Clinical Genetics Columbia University |
| 2002/07-2013/02 | Assistant Professor of Pediatrics in Medicine Division of Molecular Genetics, Dept. of Pediatrics, Columbia University |
| 1998/07-2002/06 | Guest Investigator, Laboratory of Human Behavior and Metabolism, Rockefeller University |
| 1998/07-2002/06 | Associate Research Scientist, Division of Molecular Genetics, Dept. of Pediatrics, Columbia University |
| 1997/07-1998/06 | Lecturer, Division of Molecular Genetics, Department of Pediatrics, Columbia Univ. |

## Selective Publications


1. Ganapathi, M., Matsuoka, L.S., March, M., Li, D., Brokamp, E., Benito-Sanz, S., White, S.M., Lachlan, K., Ahimaz. P., Sewda, A., Bastarache, L., Wilson, A.T., Stoler, J.M., Baptista, J., Stals, K., Demurger, F., Cogne, B., Isidor, B., Bedeschi, M.F., Peron, A., Amiel, J., Zackai, E., Schacht, J.P., Iglesias, A.D., Morton, J., Seidel, V., Lucia, S., Beaskin, S.M., THiffault, I., Cogan, J.D., Gordon, C.T., Chung, W.K., Bowdin, S., Bhoj, E. Heterozygous variants NRF2 cause a recognizable multiple congenital syndrome with developmental delays. European Journal of Human Genetics. Submitted. February 17, 2023.
2. Saffari, A., Lau, T., Tajsharghi, H., Karimiani, E.G., Kariminejad, A., Efthymiou, S., Zifarelli, G., Sultan, T., Toosi, M.B., Sedighzadeh, S., Siu, V.M., Ortigoza-Escobar, J.D., AlShamsi, A.M., Ibrahim, S., Al-Sannaa, N.A., Al-Hertani, W., Sandra, W., Tarnopolsky, M., Alavi, S., Li, C., Day-Salvatore, D.L., Martínez-González, M.J., Levandoski, K.M., Bedoukian, E., Madan-Khetarpal, S., Idleburg, M.J., Menezes, M.J., Siddharth, A., Platzer, K., Oppermann, H., Smitka, M., Collins, F., Lek, M., Shahrooei, M., Ghavideldarestani, M., Herman, I., Rendu, J., Faure, J., Baker, J., Bhambhani, V., Calderwood, L., Akhondian, J., Imannezhad, S., Mirzadeh, H.S., Hashemi, N., Doosti, M., Safi, M., Ahangari, N., Torbati, P.N., Abedini, S., Salpietro, V., Gulec, E.Y., Eshaghian, S., Ghazavi, M., Pascher, M.T., Vogel, M., Abicht, A., Moutton, S., Bruel, A.L., Rieubland, C., Gallati, S., Strom, T.M., Lochmüller, H., Mohammadi, M.H., Alvi, J.R., Zackai, E.H., Keena, B.A., Skraban, C.M., Berger, S.I., Andrew, H.E., Rahimian, E., Morrow, M.M., Wentzensen, I.M., Millan, F., Henderson, L.B., Dafsari, H.S., Jungbluth, H., Gomez-Ospina, N., McRae, A., Peter, M., Veltram, D., Marinakis, N.M., Sofocleous, C., Ashrafzadeh, F., Pehlivan, D., Lemke, J.R., Melki, J., Benezit, A., Bauer, P., Weis, D., Lupski, J.R., Senderek, J., Christodoulou, J., Chung, W.K., Goodchild, R., Offiah, A.C., Moreno-De-Luca, A., Mohnish, S., Ebrahimi-Fakhari, D., Houlden, H., Maroofian, R. The clinical and genetic spectrum of autosomal-recessive TOR1A-related disorders. Brain. 2023 Feb 9:awad039. doi: 10.1093/brain/awad039. PMID: 36757831.
3. Casillan, A., Florido, M.E., Cornejo, J.G., Bakken, S., Lynch, J.A., Chung, W.K., Mittendorf, K.F., Berner, E.S., Connolly, J.J., Weng, C., Holm, I.A., Khan, A., Kiryluk, K., Limdi, N.A., Petukhova, L., Sabatello, M., Wynn, J. Participant-guided development of bilingual genomic educational infographics for Electronic Medical Records and Genomics Phase IV study. Journal of Patient Education and Counseling. Submitted. January 31, 2023.
4. Applebaum, P.S., Berger, S.M., Brokamp, E., Brown, S., Burke, W., Clayton, E.W., Evans, B.J., Hamid, R., Marchant, G.E., Martin, D.M., O' Connor, B.C., Pagan, J.A., Parenas, E., Roberts, J.L., Rowe, J., Schneider, J., Siegel, K., Veenstra, D.L. Chung. W.K. Practical Considerations for Reinterpretation of Individual Genetic Variants. Genet Med. 2023 Feb 4:100801. doi: 10.1016/j.gim.2023.100801. PMID: 36748709.
5. Morton, S.U., Norris, A.N., Cunningham, S., King, E., Goldmuntz, E., Brueckner, M., Miller, T.A., Thomas, N.H., Liu, C., Adams, H.R., Bellinger, D.C., Cleveland, J., Cnota, J.F., Dale, A.M., Frommelt, M., Gelb, B.D., Grant P.E., Goldberg, C.S., Huang, H., Kuperman, J.M., Li, J.S., McQuillen, P.S., Panigraphy, A., Porter, G.A., Roberts, A.E., Russell, M.W., Seidman CE., Tivarus, M.E., Anagnoustou, E., Hagler, D.J.,

Chung, W.K., Newburger, J.W. Association of Potentially Damaging De Novo Gene Variants with Neurologic Outcomes in Congenital Heart Disease. JAMA Netw Open. 2023 Jan 3;6(1):e2253191. doi: 10.1001/jamanetworkopen.2022.53191. PMID: 36701153.

6. Mueller, S.H., Lai, A.G., Valkovskaya, M., Michailidou, K., Bolla, M.K., Wang, Q., Dennis, J., Lush, M., Abu-Ful, Z., Ahearn, T.U., Andrulis, I.L., Culver, H.A., Antonenkova, N.N., Arndt, V., Aronson, K.J., Agustinsson, A., Baert, T., Freeman, L.E.B., Beckmann, M.W., Behrens, S., Benitez, J., Bermisheva, M., Blomqvist, C., Bogdanova, N.V., Bojesen, S.E., Bonanni, B., Brenner, H., Brucker, S.Y., Buys, S.S., Castelao, J.E., Chan, T.L., Chang, C.J., Chanock, S.J., Choi, J.Y., Chung, W.K., Colonna, S.V., Cornelissen, S., Couch, F.G., Czene, K., Daly, M.B., Devilee, P., Dork, T., Dossus, L., Dwek, M., Eccles, D.M., Ekici, A.B., Eliassen, A.H., Engel, C., Evans, D.G., Fashching, P.A., Fletcher, O., Flyger, H., Gago, D.M., Gao, Y.T., Garcia, C.M., Garcia, S.J.A., Genkinger, J., Gentry, M.A., Grassmann, F., Guenel, P., Gundert, M., Haeberle, L., Hahnen, E., Hairman, C.A., Hakansson, N., Hall, P., Harkness, E.F., Harrington, P.A., Hartikainen, J.M., Hartman, M., Hein, A., Ho, W.K., Hooning, M.J., Hoppe, R., Hopper, J.L., Houlston, R.S., Howell, A., Hunter, D.J., Huo, D., Ito, H., Iwasaki, M., Jakubowska, A., Janni, W., John, E.M., Jones, M.E., Jung, A., Kaaks, R., Kang, D., Khusnutinova, E.K., Kim, S.W., Kitahara, C.M., Koutros, S., Kraft, P., Kristensen, V.N., Kubelka, S.K., Kurian, A.W., Kwong, A., Lacey, J.V., Lambreschts, D., Le Marchand, L., Li, J., Linet, M., Lo, W.Y., Long, J., Lophatananon, A., Mannermaa, A., Manoochehri, M., Margoin, S., Masuo, K., Mavroudis, D., Menon, U., Muir, K., Murphy, R.A., Nevanlinna H., Newman, W.G., Niederacher, D., Obrien, K.M., Obi, N., Offit, K., Olopade, O.I., Olshan, A.F., Olsson, H., Park, S.K., Patel, A.V., Patel, A., Perou, C.M., Peto, J., Pharoah, P.D.P., Plaseka, K.D., Presneau, N., Rack, B., Radice, P., Ramachandran, D., Rashid, M.U., Rennert, G., Romero, A., Ruddy, K.J., Ruebner, M., Saloustros, E., Sandler, D.P., Sawyer, E.J., Schmidt, M.K., Schmutzler, R.K., Schneider, M.O., Scott, C., Shah, M., Sharma, P., Shen, C.Y., Shu, X.O., Simard, J., Surowy, H., Tamimi, R.M., Tapper, W.J., Taylor, J.A., Teo, S.H., Teras, L.R., Toland, A.E., Tollenaar, R.A.E.M., Torres, D., Torres, M.G., Troester, M.A., Truong, T., Vachon, C.M., Vijai, J., Weinberg, C.R., Wendt, C., Winqvist, R., Wolk, A., Wu, A.H., Yamaji, T., Yang, X.R., Yu, J.C., Zheng, W., Ziogas, A., Ziv, E., Dunning, A.M., Easton, D.F., Heimingway, Hamann, U., Kuchenbaecker, K.B. Aggregation tests identify new gene associations with breast cancer in populations with diverse ancestry. Genome Med. 2023 Jan 26;15(1):7. doi: 10.1186/s13073-022-01152-5. PMID: 36703164.
7. Ding, Z., Huang, G., Wang, T., Duan, W., Li, H., Yang, Z., Wang, K., Chu, X., Nelson, E.C.K., Ahlers, K., Earl, R.K., Han, Y., Feliciano, P., Chung, W.K., Eichler, E.E., Jiang, M., Xiong, B. Genetic Ablation of GIGYF1, associated with autism, causes behavorial and neurodevelopmental defects in zebrafish and mice. Biological Psychiatry. Submitted. January 15, 2023.
8. Wang, B.Z., Nash, T.R., Zhang, X., Rao, J., Abriola, L., Kim, Y., Zakharov, S., Kim, M., Lui, L., Morsink, M., Liu, B., Lock, R.I., Fleischer, S., Tamargo, M.J., Welch, C.L., Chung, W.K., Marx, S.O., Surovsteva Y.V., Novakovic, G.V., Fine, B.M. Engineered cardiac tissue model of restrictive cardiomyopathy for drug discovery. Cells Reports Medicine. Accepted. January 3, 2023.

**What's new in newborn screening?**

Author: Wendy K. Chung

Abstract:
Prof. Chung will first review the past US experience in newborn screening. Next, she will discuss the current infrastructure in place for newborn screening and how new conditions are evaluated for the Recommended Uniform Screening panel (RUSP). Following the discussion on RUSP, she will review the experience of a pilot study for spinal muscular atrophy and how the pilot study led to adoption into US newborn screening. Lastly, she will discuss the current pilot study using genome sequencing in GUARDIAN to expand the conditions that can be screened.

**Review and Prospect of Newborn Screening-Taiwan Experience**

## SPEAKER


### Chien, Yin-Hsiu 簡穎秀

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E-mail：chienyh@ntu.edu.tw


### Present Position

Visiting Staff, National Taiwan University Hospital
Clinical professor, National Taiwan University College of Medicine

### Education

Graduate Institute of Clinical Medicine, National Taiwan University College of Medicine, Taipei, Taiwan.
Chang Gung Collage of Medicine and Technology, Taoyuan, Taiwan.

### Brief Chronology of Employment

Pediatrics, National Taiwan University Hospital, Taipei, Taiwan
Fellowship, Division of Pediatrics Genetics and metabolisms, Department of Pediatrics, National Taiwan University Hospital, Taipei, Taiwan

### Selective Publications


1. Chien YH, Chiang SC, Zhang XK, Keutzer J, Lee NC, Huang AC, Chen CA, Wu MH, Huang PH, Tsai FJ, Chen YT, Hwu WL. Early detection of Pompe disease by newborn screening is feasible: results from the Taiwan screening program. Pediatrics. 2008 Jul;122(1):e39-45.
2. Wang LY, Chen NI, Chen PW, Chiang SC, Hwu WL, Lee NC, Chien YH.* Newborn screening for citrin deficiency and carnitine uptake defect using second tier molecular testing. BMC Med Genet. 2013;14(1):24.
3. Chien YH, Lee NC, Chen CA, Tsai FJ, Tsai WH, Shieh JY, Huang HJ, Hsu WC, Tsai TH, Hwu WL*. Long-term efficacy of enzyme replacement therapy on Pompe disease in patients detected by newborn screening. J Pediatr. 2015 Apr;166(4):985-91.e1-2.
4. Chien YH, Chiang SC, Weng WC, Lee NC, Lin CJ, Hsieh WS, Lee WT, Jong YJ, Ko TM, Hwu WL. Presymptomatic Diagnosis of Spinal Muscular Atrophy through Newborn Screening. J Pediatr. 2017 Nov;190:124-129.e1.
5. Chien YH, Hwu WL. The modern face of newborn screening. (Review) Pediatr Neonatol. 2022 Nov 14:S1875-9572(22)00247-9.
6. Chen HA, Hsu RH, Chen YH, Hsu LW, Chiang SC, Lee NC, Hwu WL, Chiu PC, Chien YH *. Improved Diagnosis of Citrin Deficiency by Newborn Screening Using a Molecular Second-Tier Test. Mol Genet Metab. 2022 Aug;136(4):330-336.

**Review and Prospect of Newborn Screening-Taiwan Experience**

Author: Yin-Hsiu Chien

Abstract:
A more and more genetic diseases can be treated, even by gene replacement therapy or gene modifying therapy. Newborn metabolites screening using dried blood spots offer the potential for the early detection of severe conditions. Therefore more and more conditions could be and have been added to the newborn screening recommended panels. In Taiwan, we have initiated the first newborn screening worldwide for Pompe and spinal muscular atrophy. We also introduce screening for severe combined immunodeficiency in 2010, the first program outside the USA. Of course, each test's sensitivity and specificity shall be reviewed carefully. Genome and exome sequencing has now opened more opportunities for early identification and thus decreased the lag between disease onset and diagnosis. Even throughout the efforts of applying every method we have now, a more significant challenge, coupled with the advance of new technology, comes from the spectrum of severity of those diseases. The dissection of the molecular, biochemical phenotypes, and clinical manifestations of those diseases is inevitable. Further studies and collaboration will explore our understanding and shape a new phenotype of those conditions.

**SESSION III: New Trends in the Diagnosis and Treatment of Gaucher Disease (25th Anniversary)**

## MODERATOR


### Shuan-Pei Lin 林炫沛

Tel：+886-2-2543-3535 ext. 3089~3090
E-mail：4535lin@gmail.com


### Present Professional Assignment

Senior Attending Physician, Division of Genetics and Metabolism, Department of Pediatrics, Founding Director of Rare Disease Center, Director of Division of Biochemical Genetics, Department of Medical Research, MacKay Memorial Hospital, Taipei, Taiwan; Professor, Department of Medicine, MacKay Medical College, Department of Infant and Early Childhood Care, National Taipei University of Nursing and Health Sciences, Taipei, Taiwan

### Education

| | |
|---|---|
| 1998/02-Present | Board Director, Down Syndrome Foundation of R.O.C., Taipei, Taiwan |
| 2000/07-Present | Committee Member, Committee of Early Intervention Program, MacKay Memorial Hospital, Taipei, Taiwan |
| 2004/06-2022/09 | Member of Medical Consulting Committee, International Prader-Willi Syndrome Organization (IPWSO) |
| 2005/11-Present | Committee Member, Committee of Academic Affairs, Taipei Medical Association, Taipei, Taiwan |
| 10/2008-Present | Honorary President, Taiwan MPS Society, Taipei, Taiwan |
| 2009/01-2010/05 | President, International Prader-Willi Syndrome Organization (IPWSO) |
| 2014/01-Present | Professor, Department of Medicine, Mackay Medical College and Department of Infant and Child Care, National Taipei University of Nursing and Health Sciences |
| 2017/03-2020/09 | Director, Rare Disease Center, MacKay Memorial Hospital<br>President, Taiwan Human Genetics Society |
| 2017/03-Present | Taiwan Foundation of Rare Disorders, Taipei, Taiwan |

### Awards and Honor

| | |
|---|---|
| 2005 | Physician of The Year Award, The Kyorin Award |
| 2010 | Special Contribution Award from IPWSO |
| 2014 | Best Director(s) of the Year, MacKay Memorial Hospital |
| 2018 | Best Teacher(s) of the Year for Senior Medical Students, Taipei Medical University |
| 2018 | Silver Award of Symbol of National Quality (SNQ), R.O.C. |
| 2019 | The 29th Taiwan Medical Dedication Awards |

## Selective Publications


1. Braun DA, et al. Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nat Genet 2017 Oct;49(10):1529-1538. doi: 10.1038/ng.3933.
2. Yang CF, Lin SP, Chiang CP, Wu YH, H'ng WS, Chang CP, Chen YT, Wu JY. Loss of GPNMB Causes Autosomal-Recessive Amyloidosis Cutis Dyschromica in Humans. Am J Hum Genet. 2018 Feb 1;102 (2):219-232. doi: 10.1016/j.ajhg.2017.12.012.
3. Cutiongco-de la Paz EM, Chung BH, Faradz SMH, Thong MK, David-Padilla C, Lai PS, Lin SP, Chen YH, Sura T, Laurino M. Training in clinical genetics and genetic counseling in Asia. Am J Med Genet C Semin Med Genet. 2019 Jun;181(2):177-186. doi: 10.1002/ajmg.c.31703.
4. Lin HY, 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.
5. Lin CY, Lin HY, 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 Muco polysaccharidosis Type II Occurrence in Humans. Diagnostics (Basel). 2020 Oct 21;10(10):854.
6. Giugliani R, Harmatz P, Lin SP, Scarpa M. Assessing the impact of the five senses on quality of life in mucopolysaccharidoses. Orphanet J Rare Dis. 2020 Apr 19;15(1):97.
7. Lee CL, Tan LTH, Lin HY, Hwu WL, Lee NC, Chien YH, Chuang CK, Wu MH, Wang JK, Chu SY, Lin JL, Lo FS, Su PH, Hsu CC, Ko YY, Chen MR, Chiu HC, Lin SP. Cardiac manifestations and gene mutations of patients with RASopathies in Taiwan. Am J Med Genet A. 2020 Feb;182(2):357 364.
8. Lee CL, Lee KS, Chuang CK, Su CH, Chiu HC, Tu RY, Lo YT, Chang YH, Lin HY, Lin SP. Otorhinolaryngological Management in Taiwanese Patients with Mucopolysaccharidoses. Int J Med Sci 2021 Jul 25;18(15):3373-3379. doi: 10.7150/ijms.61827.
9. Syu YM, Lee CL, Chuang CK, Chiu HC, Chang YH, Lin HY, Lin SP. Functional independence of Taiwanese children with osteogenesis imperfect. J Pers Med. 2022 Jul 24;12(8):1205. doi: 10.3390/jpm12081205
10. Okur I, Ezgu F, Giugliani R, Muschol N, Koehn A, Amartino H, Harmatz P, de Castro Lopez MJ, Couce ML, Lin SP, Batzios S, Cleary M, Solano M, Peters H, Lee J, Nestrasil I, Shaywitz AJ, Maricich SM, Kuca B, Kovalchin J, Zanelli E. Longitudinal natural history of pediatric subjects affected with mucopolysaccharidosis IIIB. J Pediatr 2022 Oct;249:50-58.e2.doi: 10.1016/j.jpeds.2022.06.005


*Dr. Shuan-Pei Lin has published more than 320 papers since 1984.

**Venglustat Clinical Trial Update - A New Era Toward Promising Gaucher Disease Therapy**

## SPEAKER


# Pramod Mistry

Tel：+203-785-3412
E-mail：pramod.mistry@yale.edu


## Present Position

| | |
|---|---|
| 2013/07-Present | Professor of Medicine (Digestive diseases) and Pediatrics (Gastroenterology) and Cellular & Molecular Physiology. |
| 2013/07-Present | Yale New Haven Hospital, Attending Physician/ Transplant Hepatologist (Hepatology) |

## Education

| | |
|---|---|
| 1972/08-1975/06 | B.Sc. (Hons) Queen Elizabeth College, University of London. Physiology/ Biochemi |
| 1975/08-1978/10 | PhD, St Thomas' s Hospital School of Medicine, University of London |
| 1978/08-1983/06 | MB BS Royal Free UCL School of Medicine |
| 1987 | Member of Royal College of Physicians (by examination) – elected Fellow of the Royal College of Physicians 1999 |
| 2007 | MA (privatum) Yale University |
| 2015 | M.D. University of the State of New York, MD conferral by the Boards of Regents |

## Brief Chronology of Employment

| | |
|---|---|
| | University of Cambridge Clinical School, Cambridge |
| 1989-1994 | Clinical Lecturer in Medicine, Department of Medicine, Royal Free UCL School of Medicine, London |
| 1994-1998 | Senior Lecturer (Hon Consultant) in Medicine, Department of Medicine, Mount Sinai School of Medicine |
| 1998-2001 | Associate Professor, Departments of Human Genetics and Medicine, Yale University School of Medicine |
| 2001-2005 | Associate Professor of Medicine (Section of Digestive Diseases) |
| 2005-2013 | Professor of Pediatrics and Medicine, Chief, Section of Pediatric Gastroenterology and Hepatology |
| 2013/07-Present | Professor of Medicine (Digestive diseases) and Pediatrics (Gastroenterology) and Cellular & Molecular Physiology. |

## Awards

| | |
|---|---|
| 1980 | Foulke's Foundation Fellowship |
| 1995 | Alan Gordon Memorial Award of the Gaucher Association (UK) |
| 1997/08 | Founding Director, National Center of Excellence for Gaucher Disease, Royal Free Hospital, London |
| 1998 | Elected Fellow of the Royal College of Physicians (London) |
| 1999 | Physician of the Year Award, Genetic Disease Foundation, New York |
| 2006/08 | Chairman of Medical Advisory Board, American Liver Foundation (Connecticut Chapter |
| 2002 | Max Millman Memorial Lecturer, Baystate Medical Center, Springfield, MA |
| 2003 | Visiting Professor, University of Manitoba Medical School, Winnipeg. Ca |
| 2004 | Visiting Professor, University of Connecticut GCRC |
| 2005,2010 | Nominated for Alice Bohmfalk Teaching Award, Yale University. |
| 2005,2008 | Award from National Gaucher Foundation for contributions to Gaucher disease |
| 2007 | MA (privatim) Yale University |
| 2010 | Fellow of the Royal Society of Medicine |
| 2004-Present | Listed in The Best Doctors in America |
| 2015-Present | Chairman of MAB of Project HOPE Gaucher Initiative for children in Egypt |
| 2017 | Elected Fellow of the American Association for Study of the Liver |
| 2020 | Nominated for Leonard Tow Humanism in Medicine Award |

## Selective Publications


1. Nair S, Sng J, Boddupalli CS, Seckinger A, Chesi M, Fulciniti M, Zhang L, Rauniyar N, Lopez M, Neparidze N, Parker T, Munshi NC, Sexton R, Barlogie B, Orlowski R, Bergsagel L, Hose D, Flavell RA, Mistry PK, Meffre E, Dhodapkar MV. Antigen-Mediated Regulation in Monoclonal Gammopathies and Myeloma. JCI Insight. 2018; 3(8). PMID 29669929
2. Mistry PK, Balwani M, Baris HN, Turkia HB, Burrow TA, Charrow J, Cox GF, Dada S, Dragosky M, Drelichman G, El-Beshlawy A, Fraga C, Freisens S, Gaemers S, Hadjiev E, Kishnani PS, Lukina E, Maison-Blanche P, Martins AM, Pastores G, Petakov M, Peterschmitt MJ, Rosenbaum H, Rosenbloom B, Underhill LH, Cox TM. Safety,Efficacy, and Authorization of Eliglustat as a First-Line Therapy in Gaucher Disease Type 1. Blood Cells Mol Dis. 2018; 71:71-74. No abstract available. PMID:29680197
3. Hakim A, Zhang X, DeLisle A, Oral EA, Dykas D, Drzewiecki K, Assis DN, Silveira M, Batisti J, Jain D, Bale A, Mistry PK, Vilarinho S. Clinical utility of genomic analysis in adults with idiopathic liver disease. J Hepatol. 2019 Jun;70(6):1214-1221. doi: 10.1016/j.jhep.2019.01.036. Epub 2019 Apr 15. PMID: 31000363
4. Afinogenova, Y., Ruan, J., Yang, R., ...Lischuk, A., Mistry, P.K. Aberrant progranulin, YKL-40, cathepsin D and cathepsin S in Gaucher disease.

5. Molecular Genetics and Metabolism, 2019, 128(1-2), pp.62-67
6. Beshlawy, A.E., Murugesan, V., Mistry, P.K., Eid, K. Reversal of life-threatening hepatopulmonary syndrome in Gaucher disease by imiglucerase enzyme replacement therapy. Molecular Genetics and Metabolism Reports, 2019, 20, 100490
7. Vujosevic, S., Medenica, S., Vujicic, V., ...Liu, J., Mistry, P.K. Gaucher disease in Montenegro - genotype/phenotype correlations: Five cases report.
8. World Journal of Clinical Cases, 2019, 7(12), pp.1475-1482
9. Nair, S., Bar, N., Xu, M.L., ...Dhodapkar, M., Mistry, P.K. Glucosylsphingosine but not Saposin C, is the target antigen in Gaucher disease-associated gammopathy. Molecular Genetics and Metabolism, 2020, 129(4), pp.286-291
10. Gao, E., Cheema, H., Waheed, N., ...Lifton, R.P., Vilarinho, S. Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. Hepatology (Baltimore, Md.), 2020, 71(5), pp.1879-1882
11. Raskovalova, T., Deegan, P.B., Mistry, P.K., ...Labarère, J., Berger, M.G.
12. Accuracy of chitotriosidase activity and CCL18 concentration in assessing type I Gaucher disease severity. A systematic review with meta-analysis of individual participant data. Haematologica, 2020, 105(5)
13. Mistry, P.K., Balwani, M., Charrow, J., ...Underhill, L.H., McClain, M.R. Real-world effectiveness of eliglustat in treatment-naïve and switch patients enrolled in the International Collaborative Gaucher Group Gaucher Registry.American Journal of Hematology, 2020, 95(9), pp.1038-1046
14. Fierro L, Nesheiwat N, Naik H, Narayanan P, Mistry PK, Balwani M. Gaucher disease and SARS-CoV-2 infection: Experience from 181 patients in New York. Mol Genet Metab. 2021 Jan;132 (1):44-48. doi: 10.1016/j.ymgme.2020.12.288. Epub 2020 Dec 15. PMID: 33353808; PMCID: PMC7834197.
15. Mistry PK, Balwani M, Charrow J, Kishnani P, Niederau C, Underhill LH, McClain MR. Real-world effectiveness of eliglustat in treatment-naïve and switch patients enrolled in the International Collaborative Gaucher Group Gaucher Registry. Am J Hematol. 2020 Sep;95(9):1038-1046. doi: 10.1002/ajh.25875. Epub 2020 Jun 24. PMID: 32438452; PMCID: PMC7497238.
16. Weinreb NJ, Camelo JS Jr, Charrow J, McClain MR, Mistry P, Belmatoug N; International Collaborative Gaucher Group (ICGG) Gaucher Registry (NCT00358943) investigators. Gaucher disease type 1 patients from the ICGG Gaucher Registry sustain initial clinical improvements during twenty years of imiglucerase treatment. Mol Genet Metab. 2021 Feb;132(2):100-111. doi: 10.1016/j.ymgme.2020.12.295. Epub 2021 Jan 8. PMID: 33485799.

## Venglustat Clinical Trial Update –A New Era Toward Promising Gaucher Disease Therapy

Author: Prof. Pramod K. Mistry

Abstract:
Gaucher disease type 3 is a chronic neuronopathic disorder with wide-ranging effects, including hepatosplenomegaly, anaemia, thrombocytopenia, skeletal disease and diverse neurological manifestations. Biallelic mutations in GBA1 reduce lysosomal acid β-glucosidase activity, and its substrates, glucosylceramide and glucosylsphingosine, accumulate. Enzyme replacement therapy and substrate reduction therapy ameliorate systemic features of Gaucher disease, but no therapies are approved for neurological manifestations. Venglustat is an investigational, brain-penetrant, glucosylceramide synthase inhibitor with potential to improve the disease by rebalancing influx of glucosylceramide with impaired lysosomal recycling. The Phase 2, open-label LEAP trial (NCT02843035) evaluated orally administered venglustat 15 mg once-daily in combination with maintenance dose of imiglucerase enzyme replacement therapy during 1 year of treatment in 11 adults with Gaucher disease type 3. Primary endpoints were venglustat safety and tolerability and change in concentration of glucosylceramide and glucosylsphingosine in CSF from baseline to Weeks 26 and 52. Secondary endpoints included change in plasma concentrations of glucosylceramide and glucosylsphingosine, venglustat pharmacokinetics in plasma and CSF, neurologic function, infiltrative lung disease and systemic disease parameters. Exploratory endpoints included changes in brain volume assessed with volumetric MRI using tensor-based morphometry, and resting functional MRI analysis of regional brain activity and connectivity between resting state networks. After 1 year of treatment, median (inter-quartile range) glucosylceramide decreased 78% (72, 84) in plasma and 81% (77, 83) in CSF; median (inter-quartile range) glucosylsphingosine decreased 56% (41, 60) in plasma and 70% (46, 76) in CSF. Ataxia improved slightly in nine patients. Whole brain volume increased slightly in patients with venglustat exposure and biomarker reduction in CSF. Functional MRI indicated stronger connectivity at Weeks 26 and 52 relative to baseline between a broadly distributed set of brain regions in patients with venglustat exposure. There were no deaths, serious adverse events or discontinuations. In adults with Gaucher disease type 3 receiving imiglucerase, addition of once-daily venglustat showed acceptable safety and tolerability and preliminary evidence of clinical stability with intriguing but intrinsically inconsistent signals in selected biomarkers, which need to be validated and confirmed in future research.

**Molecular mechanism underlying ERT unresponsiveness in Gaucher disease**

## SPEAKER


# Beom Hee Lee

Tel：++82 2 3010 5950
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## Present Position

| | |
|---|---|
| 2021- | Professor-University of Ulsan College of Medicine, Seoul, Korea |

## Education

| | |
|---|---|
| 1993-1999 | Seoul National University M.D. |
| 2007-2011 | Seoul National University Ph.D., Molecular Genetics |

## Brief Chronology of Employment

| | |
|---|---|
| 2000-2004 | Residency - Department of Pediatrics, Seoul National University Children’ s Hospital |
| 2007-2009 | Fellow - Division of Pediatric Nephrology, Department of Pediatrics, Seoul National University Children’ s Hospital |
| 2009-2011 | Fellow - Medical Genetics, Department of Pediatrics, Asan Medical Center |
| 2011–2015 | Assistant Professor - University of Ulsan College of Medicine, Seoul, Korea |
| 2016-2021 | Associate Professor - University of Ulsan College of Medicine, Seoul, Korea |
| 10/2013-09/2015 | PostDoc Fellowship - Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mout Sinai, New York, NY (Dr. Robert J. Desnick’ s Lab.) |

## Selective Publications


1. Kim EN, Do HS, Jeong H, Kim T, Heo SH, Kim YM, Cheon CK, Lee Y, Choi Y, Choi IH, Choi J, Yoo HW, Kim CJ, Zimran A, Kim K, Lee BH. Identification of a novel therapeutic target underlying atypical manifestation of Gaucher disease. Clin Transl Med. 2022 May;12(5):e862
2. Kim YM, Yum MS, Heo SH, Kim T, Jin HK, Bae JS, Seo GH, Oh A, Yoon HM, Lim HT, Kim HW, Ko TS, Lim HS, Osborn MJ, Tolar J, Cozma C, Rolfs A,Zimran A, Lee BH, Yoo HW. Pharmacologic properties of high-dose ambroxol in four patients with Gaucher disease and myoclonic epilepsy. J Med Genet. 2020 Feb;57(2):124-131.
3. Lee J, Kang E, Byeon JH, Yu HJ, Shin YL, Oh A, Kim WJ, Yum MS, Lee BH, Eun BL. Diagnostic performance of automated, streamlined, daily updated exome analysis in patients with neurodevelopmental delay. Mol Med. 2022 Mar 26;28(1):38.

4. Hwang J, Yoon HM, Lee BH, Kim PH, Kim KW. Efficacy and Safety of Selumetinib in Pediatric Patients With Neurofibromatosis Type 1: A Systematic Review and Meta-analysis. Neurology. 2022 Mar 1;98(9):e938-e946.
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9. Seo GH, Kim YM, Oh SH, Chung SJ, Choi IH, Kim GH, Yum MS, Choi JH, Kim KM, Ko TS, LEE BH, Yoo HW. Biochemical and molecular characterisation of neurological Wilson disease. Journal of medical genetics. 2018;55(9):587-93.
10. LEE BH, Aggarwal A, Slavotinek A, Edelmann L, Chen B, Desnick RJ. The focal facial dermal dysplasias: phenotypic spectrum and molecular genetic heterogeneity. Journal of medical genetics. 2017;54(9):585-90.
11. Heo SH, Kang E, Kim YM, Go H, Kim KY, Jung JY, Kang M, Kim GH, Kim JM, Choi IH, Choi JH, Jung SC, Desnick RJ, Yoo HW, LEE BH. Fabry disease: characterisation of the plasma proteome pre- and post-enzyme replacement therapy. Journal of medical genetics. 2017;54(11):771-80.

*And more than 150 publications.

## Molecular mechanism underlying ERT unresponsiveness in Gaucher disease

Author: Prof. Beom Hee Lee

Abstract:
Gaucher disease (GD) is caused by glucocerebrosidase1 (GCase) deficiency, resulting in the accumulation of glucosylceramide and its deacylated form, glucosylsphingosine (Lyso-Gb1). Despite enzyme replacement therapy (ERT), therapeutic unmet needs such as lymphadenopathy and neurological manifestations have been observed. In the current study, the GD patients' lymph nodes, plasma, and fibroblasts were examined by multi-dimensional in vitro and in vivo studies. In a type 3 GD patient with severe mesenteric lymphadenopathy despite ERT, atypical Gaucher-like cells were found with intense Lyso-Gb1 accumulation as well as multinucleation, surrounded by fibrous band-like structures. Detailed immunohistochemistry and tissue proteomic analysis revealed aberration in complement activity, macrophage polarization, and autophagy metabolism with a subsequent severe inflammatory response and the endothelial mesenchymal transition by an altered TGF-ß signaling activity. These results demonstrated that complex molecular pathways underlie the unusual progression of GD, leading to ERT unresponsiveness, and a multi-functional therapeutic approach with combination therapy is required to relieve this devastating process.

**Changing Clinical Manifestations of Gaucher Disease in Taiwan**

## SPEAKER


## Ni-Chung Lee 李妮鍾

Tel：+886-975-693-910
E-mail：ncleentu@ntu.edu.tw


### Present Position

Attending Physician, Department of Medical Genetics, NTUH
Clinical Professor, Department of Pediatrics, College of Medicine, NTU

### Education

Ph.D., Graduate Institute of Clinical Medicine, College of Medicine, National Taiwan University
M.D., Medical College, National Yang-Ming University

### Brief Chronology of Employment

Dr. Lee is a clinical associate professor at the National Taiwan University Hospital (NTUH). She had her PhD degree from NTU. Her research interests include the diagnosis and treatment of pediatric patients with rare diseases. In the diagnosis, she involves in the clinical application of next generation sequencing for pediatric rare diseases. About the treatment, she conducted gene therapy researches for AADC deficiency, Pompe disease and several rare diseases.

### Selective Publications


1. Lee NC, Chien YH, Wang CH, Wong SL, Peng SS, Tsai FJ, Hwu WL. Safety and efficacy of eliglustat combined to enzyme replacement therapy for lymphadenopathy in patients with Gaucher disease type 3. Mol Genet Metab Rep. 2022 Apr 19;31:100867.
2. Lin YL, Chang PC, Hsu C, Hung MZ, Chien YH, Hwu WL, Lai F, Lee NC. Comparison of GATK and DeepVariant by trio sequencing. Sci Rep. 2022 Feb 2;12(1):1809.
3. Lee NC, Chang KL, In 't Groen SLM, de Faria DOS, Huang HJ, Pijnappel WWMP, Hwu WL, Chien YH. Outcome of Later-Onset Pompe Disease Identified Through Newborn Screening. J Pediatr. 2022 Jan 4:S0022-3476(21)01279-8.
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5. Huang YH, Su TC, Wang CH, Wong SL, Chien YH, Wang YT, Hwu WL, Lee NC. RNA-seq of peripheral blood mononuclear cells of congenital generalized lipodystrophy type 2 patients. Sci Data. 2021 Oct 13;8(1):265.

6. Lee NC, Peng WH, Tsai LK, Lu YH, Wang HC, Shih YC, Pung ZX, Hu HY, Hwu WL, Tseng WI, Chien YH. Ultrastructural and diffusion tensor imaging studies reveal axon abnormalities in Pompe disease mice. Sci Rep. 2020 Nov 19;10(1):20239.
7. Hsu RH, Chien YH, Hwu WL, Lee NC. Diversity in heritable disorders of connective tissue at a single center. Connect Tissue Res. 2020 Sep 8:1-6. (Corresponding author)
8. Kuo CW, Hwu WL, Chien YH, Hsu C, Hung MZ, Lin IL, Lai F, Lee NC. Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes. Mol Genet Genomic Med. 2020 Aug 14:e1455. (Corresponding author)
9. Kao HJ, Chiang HL, Chen HH, Fan PC, Tu YF, Chou YY, Hwu WL, Lin CL, Kwok PY, Lee NC. De novo mutation and skewed X-inactivation in girl with BCAP31-related syndrome. Hum Mutat. 2020 Jul 11. (Corresponding author)
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11. Lee CT, Tung YC, Hwu WL, Shih JC, Lin WH, Wu MZ, Kuo KT, Yang YL, Chen HL, Chen M, Su YN, Jong YJ, Liu SY, Tsai WY, Lee NC. Mosaic paternal haploidy in a patient with pancreatoblastoma and Beckwith-Wiedemann spectrum. Am J Med Genet A. 2019 Sep;179(9):1878-1883. (Corresponding author)
12. Yu MH, Tsang MH, Lai S, Ho MS, Tse DML, Willis B, Kwong AK, Chou YY, Lin SP, Quinzii CM, Hwu WL, Chien YH, Kuo PL, Chan VC, Tsoi C, Chong SC, Rodenburg RJT, Smeitink J, Mak CC, Yeung KS, Fung JL, Lam W, Hui J, Lee NC, Fung CW, Chung BH. Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. NPJ Genom Med. 2019 Aug 5;4:18. (Corresponding author)
13. Lee NC, Hsu WC, Chang LM, Chen YC, Huang PT, Chien CC, Chien YH, Chen CL, Hwu WL, Lee PL. REM sleep and sleep apnea are associated with language function in Down syndrome children: An analysis of a community sample. J Formos Med Assoc. 2019 Aug 1..
14. Wu ET, Hwu WL, Chien YH, Hsu C, Chen TF, Chen NQ, Chou HC, Tsao PN, Fan PC, Tsai IJ, Lin SP, Hsieh WS, Chang TM, Chen CN, Lee CH, Chou YY, Chiu PC, Tsai WH, Hsiung HC, Lai F, Lee NC. Critical Trio Exome Benefits In-Time Decision-Making for Pediatric Patients With Severe Illnesses. Pediatr Crit Care Med. 2019 Jul 1. (Corresponding author)
15. Ho SY, Chien YH, Tsai LK, Muramatsu S, Hwu WL, Liou HH, Lee NC. Electrical Abnormalities in Dopaminergic Neurons of the Substantia Nigra in Mice With an Aromatic L-Amino Acid Decarboxylase Deficiency. Front Cell Neurosci. 2019 Jan (Co-Corresponding author)
16. Hsu RH, Chien YH, Hwu WL, Chang IF, Ho HC, Chou SP, Huang TM, Lee NC. Genotypic and phenotypic correlations of biotinidase deficiency in the Chinese population. Orphanet J Rare Dis. 2019 Jan 7;14(1):6. (Corresponding author)

## Changing Clinical Manifestations of Gaucher Disease in Taiwan

Author: Ni-Chung Lee

Abstract:
Gaucher disease (GD) is a lysosomal storage disorders characterized by deficient glucocerebrosidase activity, resulting from biallelic mutations in the GBA gene. Its phenotypic variability allows GD to be classified into 3 subtypes based on the presence and extent of neurological manifestations. Enzyme replacement therapy (ERT) has been available to all patients with GD in Taiwan since 1998. Newborn screening (NBS) for GD has been available since 2015. This study attempts to unveil the clinical features of patients diagnosed with GD at the different eras in Taiwan. Data from health records of two tertiary hospitals responsible for two thirds of the Gaucher patients in Taiwan were used. The study population included all patients identified as having GD between 1998 and April 2022 in these two hospitals for review. A total of 42 individuals were recorded. Compared to that reported worldwide, our cohort presents a higher proportion of GD3 individuals, both by clinical suspicion and by NBS diagnosis. The major subtypes recognized following NBS diagnosis were of GD2 and GD3. The majority of GD patients carry at least one p.Leu483Pro variant. The 5-year survival rates were 0% for GD2 and 100% for others. Those diagnosed in the post-NBS era were free of symptoms on initial presentation, except for GD2. For those diagnosed earlier, ERT proved to be effective in terms of improved hemograms and circumvented bone crisis. However, the neurological symptoms of GD3 patients progressed despite of ERT intervention. ERT is essential in reversing the hematological presentations and preventing skeletal complications of GD. Timely diagnosis of GD with NBS allows early intervention with ERT to preempt disease progression and complications. However, needs of effective intervention for neurological dysfunction remain unfulfilled.

# 人類遺傳學會第九屆理監事選舉
# 提 名 名 單

| 理 事 | |
|---|---|
| 一、 牛道明 | 二、 李妮鍾 |
| 三、 罕見疾病基金會 | 四、 林炫沛 |
| 五、 林翔宇 | 六、 邱寶琴 |
| 七、 胡務亮 | 八、 陳沛隆 |
| 九、 陳明 | 十、 陳燕彰 |
| 十一、 楊佳鳳 | 十二、 鄔哲源 |
| 十三、 趙美琴 | 十四、 蔡文暉 |
| 十五、 蔡立平 | 十六、 蔡輔仁 |
| 十七、 簡穎秀 | 十八、 鐘育志 |

# 人類遺傳學會第九屆理監事選舉

# 提 名 名 單

| 監 事 | |
|---|---|
| 一、 方菊雄 | 二、 林清淵 |
| 三、 張建國 | 四、 蔡世峯 |
| 五、 魏耀揮 | |

# 人類遺傳學會第九屆理監事選舉
# 會員大會/選舉當天注意事項與流程

| | |
|---|---|
| 報到 | 1. 請會員主動出示個人有效身分證件並告知您的會員編號，以進行會籍資料比對。<br>報到並領取選票時間：08:20-09:30 |
| | 2. 同時受委託之會員請於報到時主動出示委託書(需雙方親簽/蓋章)，經學會驗證確認後（委託人及受委託人皆須繳清常年會費）收取保留，方得代行使選舉權利。 |
| | 3. 驗證「委託書」時間：112 年4月30日(日)上午08:20-09:30，請最晚於09:30 前至報到處領取 / 驗證（以大會電腦之時間為準），逾時恕不受理，並視同放棄本人、受委託人之選舉權。 |
| 投票方式 | 1. 投票時間：112 年4月30日(日) 上午09:00-13:00【最晚請於13:00前完成投票】 |
| | 2. 投票地點：台大醫院國際會議中心3樓Lobby。 |
| | 3. 請於投票時間內，至指定地點(台大醫院國際會議中心)完成驗證身分與投票。 |
| | 4. 本次選舉採用「無記名連記法」。 |
| | 5. 圈寫方式：於圈選欄位中「○」，或在填選候選人欄位上填寫您理想中的候選人姓名。<br><br>理事：最少應圈寫 1 名，至多可圈寫 15 名。<br>監事：最少應圈寫 1 名，至多可圈寫 5 名。 |
| | 6. 在投票時間內將選舉票分別投入理事票箱及監事票箱中。 |
| | 7. 112 年 4 月 30 日 (日) 下午 13:00 進行票箱彌封作業，未能於 13:00 前投票者，視同放棄此屆選舉權。 |
| 開票 | 俟彌封票箱後，112 年4 月30 日(日) 13:00- 於台大醫院國際會議中心進行開票作業。 |
| 公告當選名次 | 1. 公告時間：112 年4 月30日(日) |
| | 2. 公告地點：台大醫院國際會議中心 3 樓 Lobby。 |
| | 3. 當選及候補當選名次以得票多寡為序。 |
| | ※本會設置理事 15 人及候補理事 5 人；監事5 人及候補監事 1 人。 |

**◎身份確認/繳清會費→簽到→投票→開票→公告。**

**◎其他詳細內容請見大會手冊內公告**

**◎其他未盡事宜，悉依本會章程規定，及內政部「人民團體選舉罷免辦法」規定辦理。**

# 人類遺傳學會第九屆理監事選舉
# 投票規範

| | |
|---|---|
| 有效票 | 1. 圈選於某一候選人上空白格內者，應屬有效票。 |
| | 2. 圈選於某一候選人欄各格內，能辨別為圈選何人者，應屬有效票。 |
| | 3. 雖部分圈選於某一候選人欄各格之內線與選舉票邊緣之間。但能辨別為圈選何人者，應屬有效票。 |
| | 4. 雖部分圈選於候選人共用空間。但能辨別為圈選何人者，應屬有效票。 |
| | 5. 圈選於某一候選人欄格與選票邊緣之間，且部分圈印加蓋於欄格線上，仍能辨別為圈選何人者，應屬有效票。 |
| | 6. 圈選一個候選人而圈二圈以上能辨別圈選何人者，應屬有效票。 |
| | 7. 因疊折反印之圈選痕跡，能辨別係摺票時所疊折反印者，應屬有效票。 |
| | 8. 使用選舉機關製備之圈選工具圈選，縱然僅部分印出，仍能辨別為圈選何人者，應屬有效票。 |
| 廢票 | 1. 不用學會秘書處製發之選舉票者，應屬無效票。 |
| | 2. 監事選票：如完全空白沒有圈選或圈選及手寫超過 5 人，應屬無效票。 |
| | 3. 理事選票：如完全空白沒有圈選或圈選及手寫超過 15 人，應屬無效票。 |
| | 4. 圈選於相鄰候選人共用空間，且圈印加蓋於欄格線外，不能辨別為圈選何人，應屬無效票。 |
| | 5. 圈選後加以塗改另行圈選者，應屬無效票。 |
| | 6. 簽名者，應屬無效票。 |
| | 7. 圈選後加以簽名者或圈選後加以蓋章者，應屬無效票。 |
| | 8. 蓋章者或按指印者，應屬無效票。 |
| | 9. 圈選後記載任何文字者，應屬無效票。 |
| | 11. 劃寫符號者/圈選後加以劃寫符號者，應屬無效票。 |
| | 12. 將選舉票任何一角撕破致不完整者，應屬無效票。 |
| | 13. 將選舉票污染致不能辨別所圈為何人，應屬無效票。 |
| | **※第九屆監事- 應選 5 人、理事-應選 15 人。** |
| | **※候選人名單係應姓氏筆劃排列，並經本學會審查通過，以供會員圈選。** |

## Acknowledgements

**感謝有您與我們一起**
**為遺傳與罕見疾病的診斷與治療共同努力**

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