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Department:	Department of Pediatrics	Position:	Chairman	
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Education:	<ul style="list-style-type: none"> ● PhD, Institute of Clinical Medicine, National Yang-Ming University, 2001-2004 ● Clinical and molecular researcher “Medical genetics” Duke University, USA, 1995-1996 ● M.D. of Kaohsiung Medical University, 1980-1987 			
Experience:	<ul style="list-style-type: none"> ● Chairman, Department of Pediatrics, Taipei Veterans General Hospital ● Director, Rare Disease Medical Research Center, Taipei Veterans General Hospital ● Professor, Institute of Clinical Medicine, National Yang-Ming University ● Director, Genetic Counseling Center, Taipei Veterans General Hospital ● Chairperson, Taiwan Pediatric Association- Medical Genetics / Metabolism 			
Research Interests: (Key words)	Congenital Genetic and Metabolic Diseases, Rare Diseases, Clinical Trials, Genetic Testing, Gene Therapy, Precision Medicine			
Selected Publications (in recent 5 yr):	<p>(*, Corresponding author)</p> <ol style="list-style-type: none"> 1. Cheng YF, Xirasagar S, Chen CS, Niu DM*, Lin HC. Association of Fabry Disease with Hearing Loss, Tinnitus, and Sudden Hearing Loss: A Nationwide Population-Based Study. J Clin Med. 2022 Dec 13;11(24): 7396. 2. Yeh TC, Cheng HC, Li HY, Chi SC, Yang HY, Yu JY, Niu DM*, Wang AG. Ophthalmic characteristics and retinal vasculature changes in Williams syndrome, and its association with systemic diseases. Eye (Lond). 2022 Nov 28. 3. Yang CF, Liao TE, Chu YL, Chen LZ, Huang LY, Yang TF, Ho HC, Kao SM, Niu DM*. Long-term outcomes of very early treated infantile-onset Pompe disease with short-term steroid premedication: experiences from a nationwide newborn screening programme. J Med Genet. 2022 Sep 22:jmedgenet-2022-108675. (SCI, IF: 5.945, 20/191, GENETICS & HEREDITY) 4. Lee CL, Lin SP, Niu DM*, Lin HY. 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. 5. Chen YC, Huang CY, Lee YT, Wu CH, Chang SK, Cheng HL, Chang PH, Niu DM*, Cheng YF. Audiological and otologic manifestations of glutaric aciduria type I. Orphanet J Rare Dis. 2020 Dec 1;15(1):337. 			

	<ol style="list-style-type: none"> 6. Liang KH, Lu YH, Niu CW, Chang SK, Chen YR, Cheng CY, Hsu TR, Yang CF, Nakamura K, <u>Niu DM*</u>. The Fabry disease-causing mutation, GLA IVS4+919G>A, originated in Mainland China more than 800 years ago. J Hum Genet. 2020 Jul;65(7):619-625. 7. Chu TH, Chien YH, Lin HY, Liao HC, Ho HJ, Lai CJ, Chiang CC, Lin NC, Yang CF, Hwu WL, Lee NC, Lin SP, Liu CS, Hu RH, Ho MC, <u>Niu DM*</u>. Methylmalonic acidemia/propionic acidemia - the biochemical presentation and comparing the outcome between liver transplantation versus non-liver transplantation groups. Orphanet J Rare Dis. 2019 Apr 2;14(1):73. 8. Hsu MJ, Chang FP, Lu YH, Hung SC, Wang YC , Yang AH, Lee HJ, Sung SH, Wang YF, Yu WC, Hsu TR, Huang PH, Chang SK, Ivan Dzhagalov , Hsu CL, <u>Niu DM*</u>. Identification of lysosomal and extralysosomal globotriaosylceramide (Gb3) accumulations before the occurrence of typical pathological changes in the endomyocardial biopsies of Fabry disease patients. Genetics in Medicine. 2018; 21(1):224-232. 9. Lu YH, Huang PH, Wang LY, Hsu TR, Li HY, Lee PC, Hsieh YP, Hung SC, Wang YC, Chang SK, Lee YT, Ho PH, Ho HC, <u>Niu DM*</u>. Improvement in the sensitivity of newborn screening for Fabry disease among females through the use of a high-throughput and cost-effective method, DNA mass spectrometry. J Hum Genet. 2018 Jan;63(1):1-8. 10. Hsu TR, <u>Niu DM*</u>. Fabry disease: Review and experience during newborn screening. Trends Cardiovasc Med. 2018 May;28(4):274-281. 11. Liao HC, Hsu TR, Young L, Chiang CC, Huang CK, Liu HC, <u>Niu DM*</u>, Chen YJ. Functional and biological studies of α-galactosidase A variants with uncertain significance from newborn screening in Taiwan. Mol Genet Metab. 2018 Feb;123(2):140-147.
Names of Lab members:	Yung-Hsiu Lu, Yun-Ru Chen, Chi-Hya Cheng, Yu-Ting Chiang, Yu-Ping Hsieh