Name：Dau-Ming Niu, MD, PhD

Present Position

- Chairman, Department of Pediatrics, Taipei Veterans General Hospital, Taipei, Taiwan
- Director, Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taipei, Taiwan
- Professor, Institute of Clinical Medicine, National Yang-Ming University, Taipei, Taiwan

Education

1. M.D. of Kaohsiung Medical University, 1980-1987
2. Clinical and molecular researcher "Medical genetics" Duke University, USA, 1995-1996
3. PhD, National Yang-Ming University, 2001-2004
4. Pediatrics certification, Medical genetics/ metabolism certified by Taiwan pediatric association, Endocrinology certified by Taiwan pediatric association.

Honors

1. 31st Medical Dedication Awards, Health and Welfare Committee, 2021
2. 25th Academic Distinguished Outstanding Alumni, Kaohsiung Medical University, 2021
3. 「Top of the Fabry disease research and treatment center」Symbol of National Quality (SNQ) -Golden Award, 2021
4. Outstanding Alumni-National Yang Ming Chiao Tung University, 2020
5. Outstanding Research Award, Ministry of Science and Technology, 2017
6. Steering committee of Fabry Outcome Survey (FOS), 2015-present.
7. Asian Congress for inherited metabolic disease (Committee).
8. 2010 Veterans Affairs Commission, Executive Yuan, R.O.C. Best Doctor.
9. Bureau of Health Promotion, Department of Health, R.O.C.「No.1 in 2010 Outstanding Medicinal Personnel Award of Research and Caring rare diseases」.
10. 2010 National Yung-Ming University Institute of Clinical Medicine Best Teacher
11. 2007 Veterans Affairs Commission, Executive Yuan, R.O.C. Best Doctor.
12. 2006 2nd National Presidential Award for Education.
13. 2006 Veterans Affairs Commission, Executive Yuan, R.O.C. Best Doctor.

Representative Publication (*, Corresponding author)

1. Hsu TR, Hung SC, Chang FP, Yu WC, Sung SH, Hsu CL, Dzhagalov I, Yang CF, Chu TH, Lee HJ, Chang SK, Liao HC, Lin HY, Liao TC, Lee PC, Li HY, Yang AH, Ho HC, Chiang CC, Lin CY, Desnick RJ, **Niu DM*.** Later Onset Fabry Disease, Cardiac Damage Progress in Silence: Experience With a Highly Prevalent Mutation. *J Am Coll Cardiol*. 2017. 68:2554-2563.
2. 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, **Niu DM***. 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.

3. Lin HY, Chong KW, Hsu JH, Yu HC, Shih CC, Huang CH, Lin SJ, Chen CH, Chiang CC, Ho HJ, Lee PC, Kao CH, Cheng KH, Hsueh C, **Niu DM*.** High incidence of the cardiac variant of Fabry disease revealed by newborn screening in the Taiwan Chinese population. *Circ Cardiovasc Genet*. 2009. 2(5):450-6.
4. Yang CF, Yang CC, Liao HC, Huang LY, Chiang CC, Ho HC, Lai CJ, Chu TH, Yang TF, Hsu TR, Soong WJ, **Niu DM*.** Very Early Treatment for Infantile-Onset Pompe Disease Contributes to Better Outcomes. *J Pediatr*. 2016. 169:174-80.
5. Liu KM, Liu TT, Lee NC, Cheng LY, Hsiao KJ, **Niu DM*.** Long-term follow-up of Taiwanese Chinese patients treated early for 6-pyruvoyl-tetrahydropterin synthase deficiency. *Arch Neurol*. 2008. 65(3):387-92.
6. **Niu DM**, Hsu JH, Chong KW, Huang CH, Lu YH, Kao CH, Yu HC, Lo MY, Jap TS. Six new mutations of the thyroglobulin gene discovered in Taiwanese children presenting with thyroid dyshormonogenesis. J Clin Endocrinol Metab. 2009. 94(12):5045-52.
7. **Niu DM**, Hwang B, Chu YK, Liao CJ, Wang PL, Lin CY. High prevalence of a novel mutation (2268 insT) of the thyroid peroxidase gene in Taiwanese patients with total iodide organification defect, and evidence for a founder effect. J Clin Endocrinol Metab. 2002. 87(9):4208-12.
8. **Niu DM**, Lin CY, Hwang B, Jap TS, Liao CJ, Wu JY. Contribution of genetic factors to neonatal transient hypothyroidism. Arch Dis Child Fetal Neonatal Ed. 2005. 90(1):F69-72.
9. **Niu DM**, Hwang B, Hwang HW, Wang NH, Wu JY, Lee PC, Chien JC, Shieh RC, Chen YT. A common SCN5A polymorphism attenuates a severe cardiac phenotype caused by a nonsense SCN5A mutation in a Chinese family with an inherited cardiac conduction defect. J Med Genet. 2006. 43(10):817-21.
10. **Niu DM**, Pan CC, Lin CY, Hwang B, Chung MY. Mosaic or chimera? Revisiting an old hypothesis about the cause of the 46,XX/46,XY hermaphrodite. J Pediatr. 2002. 140(6):732-5.
11. Lu YH, Cheng LM, Huang YH, Lo MY, Wu TJ, Lin HY, Hsu TR, **Niu DM*.** Heterozygous carriers of classical homocystinuria tend to have higher fasting serum homocysteine concentrations than non-carriers in the presence of folate deficiency. Clin Nutr. 2015. 34(6):1155-8.
12. Lu YH, Huang YH, Cheng LM, Yu HC, Hsu JH, Wu TJ, Lo MY, Lin A, Lin CY, Wu JY, **Niu DM***. Homocystinuria in Taiwan: an inordinately high prevalence in an Austronesian aboriginal tribe, Tao. Mol Genet Metab. 2012. 105(4):590-5.

SCI-indexed papers published in the past 5 years (*, Corresponding author)

1. Lin NC, Tsai HL, Chen CY, Yeh YT, Lei HJ, Chou SC, Chung MH, Yang CF, **Niu DM**, Loong CC, Hsia CY, Liu CS. Safety and long-term outcomes of early liver transplantation for pediatric methylmalonic acidemia patients. Pediatr Transplant. 2022 Jan 17:e14228.
2. 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.
3. Int J Med Sci. 2022 Jan 1;19(1):126-131.
4. Liu MC, Wang MT, Chen PK, **Niu DM**, Fan Chiang YH, Hsieh MH, Tsai HC.

5. Case Report: Anesthetic Management and Electrical Cardiometry as Intensive Hemodynamic Monitoring During Cheiloplasty in an Infant With Enzyme-Replaced Pompe Disease and Preserved Preoperative Cardiac Function. Front Pediatr. 2021;9:729824.
6. 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;11(11):1197.
7. Lin HY, 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;11(11):1066.
8. Chen WY, **Niu DM**, Chen LZ, Yang CF.Congenital hypopituitarism due to novel compound heterozygous POU1F1 gene mutation: A case report and review of the literature. Mol Genet Metab Rep. 2021;29:100819.
9. Lu DY, Huang WM, Wang WT, Hung SC, Sung SH, Chen CH, Yang YJ, Niu DM, Yu WC. Reduced global longitudinal strain as a marker for early detection of Fabry cardiomyopathy. Eur Heart J Cardiovasc Imaging. 2021:jeab214.
10. Wang TH, Soong WJ, **Niu DM**, Chu YL, Chen LZ, Huang LY, Yang CF. Airway abnormalities and pulmonary complications in long-term treated late-onset Pompe disease: Diagnostic and interventional by flexible bronchoscopy. Pediatr Pulmonol. 2022;57(1):185-192.
11. Hsueh CY, Huang CY, Yang CF, Chang CC, Lin WS, Cheng HL, Wu SL, Cheng YF, **Niu DM***. Hearing characteristics of infantile-onset Pompe disease after early enzyme-replacement therapy.
12. Orphanet J Rare Dis. 2021;16(1):348.
13. Huang CC, **Niu DM**, Charng MJ. Genetic Analysis in a Taiwanese Cohort of 750 Index Patients with Clinically Diagnosed Familial Hypercholesterolemia. J Atheroscler Thromb. 2021;S1933-2874 (21)00115-X.
14. Germain DP, Moiseev S, Suárez-Obando F, Al Ismaili F, Al Khawaja H, Altarescu G, Barreto FC, Haddoum F, Hadipour F, Maksimova I, Kramis M, Nampoothiri S, Nguyen KN, **Niu DM**, Politei J, Ro LS, Vu Chi D, Chen N, Kutsev S. The benefits and challenges of family genetic testing in rare genetic diseases-lessons from Fabry disease. Mol Genet Genomic Med. 2021 May;9(5):e1666.
15. Fan WP, Li HY, Tseng SY, Juan CC, Hwang B, **Niu DM**, Lee PC. Aortic regurgitation in Marfan syndrome patients who underwent prophylactic surgery: A single-center experience. J Chin Med Assoc. 2021 May 1;84(5):540-544.
16. Lin HY, Chen MR, Lee CL, Lin SM, Hung CL, **Niu DM**, Chang TM, Chuang CK, Lin SP. Natural progression of cardiac features and long-term effects of enzyme replacement therapy in Taiwanese patients with mucopolysaccharidosis II. Orphanet J Rare Dis. 2021;16(1):99.
17. Laney DA, Germain DP, Oliveira JP, Burlina AP, Cabrera GH, Hong GR, Hopkin RJ, **Niu DM**, Thomas M, Trimarchi H, Wilcox WR, Politei JM, Ortiz A. Fabry disease and COVID-19: international expert recommendations for management based on real-world experience. Clin Kidney J. 2020;13:913-925.

18. 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;15(1):314.
19. Lee FS, Yen HJ, **Niu DM**, Hung GY, Lee CY, Yeh YC, Chen PC, Chang SK, Yang CF. Allogeneic hematopoietic stem cell transplantation for treating severe lung involvement in Gaucher disease. Mol Genet Metab Rep. 2020;25:100652.
20. Hughes DA, Aguiar P, Deegan PB, Ezgu F, Frustaci A, Lidove O, Linhart A, Lubanda JC, Moon JC, Nicholls K, **Niu DM**, Nowak A, Ramaswami U, Reisin R, Rozenfeld P, Schiffmann R, Svarstad E, Thomas M, Torra R, Vujkovac B, Warnock DG, West ML, Johnson J, Rolfe MJ, Feriozzi S. Early indicators of disease progression in Fabry disease that may indicate the need for disease-specific treatment initiation: findings from the opinion-based PREDICT-FD modified Delphi consensus initiative. BMJ Open. 2020;10(10):e035182.
21. Wang WT, Sung SH, Liao JN, Hsu TR, **Niu DM**, Yu WC. Cardiac manifestations in patients with classical or cardiac subtype of Fabry disease. J Chin Med Assoc. 2020;83(9):825-829.
22. Liang KH, Lu YH, Niu CW, Chang SK, Chen YR, Cheng CY, Hsu TR, Yang CF, Nakamura K, **Niu DM***. The Fabry disease-causing mutation, GLA IVS4+919G>A, originated in Mainland China more than 800 years ago. J Hum Genet. 2020;65(7):619-625.
23. Lin HY, Chuang CK, Lee CL, Chen MR, Sung KT, Lin SM, Hou CJ, Niu DM, Chang TM, Hung CL, Lin SP. Diagnostics (Basel). Cardiac Evaluation using Two-Dimensional Speckle-Tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. 2020 Jan 23;10(2). pii: E62.
24. Yang CF, **Niu DM**, Tai SK, Wang TH, Su HT, Huang LY, Soong WJ. Airway Abnormalities in Very Early Treated Infantile-Onset Pompe Disease: A Large-Scale Survey by Flexible Bronchoscopy. 2020; Am J Med Genet A. [published online ahead of print]
25. Ramaswami U, Beck M, Hughes D, Kampmann C, Botha J, Pintos-Morell G, West ML, **Niu DM**, Nicholls K, Giugliani R; FOS Study Group. Cardio- Renal Outcomes With Long- Term Agalsidase Alfa Enzyme Replacement Therapy: A 10- Year Fabry Outcome Survey (FOS) Analysis. Drug Des Devel Ther. 2019;13:3705-3715.
26. Lin HY, Lee CL, Chiu PC, **Niu DM**, Tsai FJ, Hwu WL, Lin SJ, Lin JL, Chang TM, Chuang CK, Lin SP. Relationships Among Height, Weight, Body Mass Index, and Age in Taiwanese Children With Different Types of Mucopolysaccharidoses. Drug Des Devel Ther. 2019;13:3705–3715.
27. Tseng SY, **Niu DM**, Chu TH, Yeh YC, Huang MH, Yang TF, Liao HC, Chiang CC, Ho HC, Soong WJ, Yang CF. Very rare condition of multiple Gaucheroma: A case report and review of the literature. Mol Genet Metab Rep. 2019;20:100489.
28. Lee CL, Lin HY, Chuang CK, Chiu HC, Tu RY, Huang YH, Hwu WL, Tsai FJ, Chiu PC, **Niu DM**, Chen YJ, Chao MC, Chang TM, Lin JL, Chang CY, Kao YC, Lin SP. Functional independence of Taiwanese patients with mucopolysaccharidoses. Mol Genet Genomic Med. 2019;7(8):e790.
29. Lin HY, Chen MR, Lin SM, Hung CL, **Niu DM**, Chang TM, Chuang CK, Lin SP. Cardiac characteristics and natural progression in Taiwanese patients with mucopolysaccharidosis III. Orphanet J Rare Dis. 2019;14(1):140.

30. 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, **Niu DM*.** Methylmalonic acidemia/propionic acidemia - the biochemical presentation and comparing the outcome between liver transplantation versus non-liver transplantation groups. Orphanet J Rare Dis. 2019;14(1):73.
31. Lin HY, Chan WC, Chen LJ, Lee YC, Yeh SI, **Niu DM**, Chiu PC, Tsai WH, Hwu WL, Chuang CK, Lin SP. Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. Mol Genet Genomic Med. 2019;7(5):e00617.
32. Lin HY, Lee CL, Lo YT, Wang TJ, Huang SF, Chen TL, Wang YS, **Niu DM**, Chuang CK, Lin SP. The relationships between urinary glycosaminoglycan levels and phenotypes of mucopolysaccharidoses. Mol Genet Genomic Med. 2018;6(6):982-992.
33. Lin HY, Chuang CK, Ke YY, Hsu CC, Chiu PC, **Niu DM**, Tsai FJ, Hwu WL, Lin JL, Lin SP. Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. 2018. pii: S1875-9572(18)30270-5.
34. Lin HY, Chen MR, Lin SM, Hung CL, **Niu DM**, Chuang CK, Lin SP. Cardiac features and effects of enzyme replacement therapy in Taiwanese patients with Mucopolysaccharidosis IVA. Orphanet J Rare Dis. 2018;13(1):148.
35. Lin HY, Chuang CK, Lee CL, Tu RY, Lo YT, Chiu PC, **Niu DM**, Fang YY, Chen TL, Tsai FJ, Hwu WL, Lin SJ, Chang TM, Lin SP. Mucopolysaccharidosis III in Taiwan: Natural history, clinical and molecular characteristics of 28 patients diagnosed during a 21-year period. Am J Med Genet A. 2018;176:1799-1809.
36. Shibata N, Hasegawa Y, Yamada K, Kobayashi H, Purevsuren J, Yang Y, Dung VC, Khanh NN, Verma IC, Bijarnia-Mahay S, Lee DH, **Niu DM**, Hoffmann GF, Shigematsu Y, Fukao T, Fukuda S, Taketani T, Yamaguchi S. Diversity in the incidence and spectrum of organic acidemias, fatty acid oxidation disorders, and amino acid disorders in Asian countries: Selective screening vs. expanded newborn screening. Mol Genet Metab Rep. 2018;16:5-10.
37. 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, **Niu DM*.** 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..
38. Lin HY, Chuang CK, Chen MR, Lin SJ, Chiu PC, **Niu DM**, Tsai FJ, Hwu WL, Chien YH, Lin JL, Lin SP. Clinical characteristics and surgical history of Taiwanese patients with mucopolysaccharidosis type II: data from the hunter outcome survey (HOS). Orphanet J Rare Dis. 2018 Jun 4;13(1):89
39. Lee CL, Lin HY, Tsai LP, Chiu HC, Tu RY, Huang YH, 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 A. 2018;176(6):1309-1314.
40. Yung-Hsiu Lu, Po-Hsun Huang, Li-Yun Wang, Ting-Rong Hsu, Hsing-Yuan Li, Pi-Chang Lee, Yu-Ping Hsieh, Sheng-Che Hung, Yu-Chen Wang, Sheng-Kai Chang, Ya-Ting Lee, Ping-Hsun Ho, Hui-Chen Ho, **Dau-Ming Niu*.** 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;63(1):1-8.

41. Hsu TR, **Niu DM***. Fabry disease: Review and experience during newborn screening. Trends Cardiovasc Med. 2017. pii: S1050-1738(17)30162-7.
42. Hwang HE, Hsu TR, Lee YH, Wang HK, Chiou HJ, **Niu DM.** Muscle ultrasound: A useful tool in newborn screening for infantile onset pompe disease. 2017;96(44):e8415
43. Liao HC, Hsu TR, Young L, Chiang CC, Huang CK, Liu HC, **Niu DM***, Chen YJ. Functional and biological studies of α-galactosidase A variants with uncertain significance from newborn screening in Taiwan. *Mol Genet Metab*. 2017. pii: S1096-7192(17)30141-5.
44. Liao HC, Chan MJ, Yang CF, Chiang CC, **Niu DM**, Huang CK, Gelb MH. Mass Spectrometry but Not Fluorimetry Distinguishes Affected and Pseudodeficiency Patients in Newborn Screening for Pompe Disease. *Clin Chem.* 2017. 63(7):1271-1277.
45. Chang WH, **Niu DM**, Lu CY, Lin SY, Liu TC, Chang JG. Modulation the alternative splicing of GLA (IVS4+919G>A) in Fabry disease. PLoS One. 2017. 12(4):e0175929.
46. Tsai FC, Lee HJ, Wang AG, Hsieh SC, Lu YH, Lee MC, Pai JS, Chu TH, Yang CF, Hsu TR, Lai CJ, Tsai MT, Ho PH, Lin MC, Cheng LY, Chuang YC, **Niu DM***. Experiences during newborn screening for glutaric aciduria type 1: Diagnosis, treatment, genotype, phenotype, and outcomes. *J Chin Med Assoc*. 2017. 80(4):253-261.
47. Chen KH, Chou YC, Hsiao CY, Chien Y, Wang KL, Lai YH, Chang YL, **Niu DM**, Yu WC. Amelioration of serum 8-OHdG level by enzyme replacement therapy in patients with Fabry cardiomyopathy. *Biochem Biophys Res Commun*. 2017. 486(2):293-299.
48. Lee HJ, Hsu TR, Hung SC, Yu WC, Chu TH, Yang CF, Bizjajeva S, Tiu CM, **Niu DM*.** A comparison of central nervous system involvement in patients with classical Fabry disease or the later-onset subtype with the IVS4+919G>A mutation. *BMC Neurol*. 2017. 17(1):25.
49. Auray-Blais C, Lavoie P, Boutin M, Ntwari A, Hsu TR, Huang CK, **Niu DM*.** Biomarkers associated with clinical manifestations in Fabry disease patients with a late-onset cardiac variant mutation. *Clin Chim Acta*. 2017. 466:185-193.
50. Chou SJ, Yu WC, Chang YL, Chen WY, Chang WC, Chien Y, Yen JC, Liu YY, Chen SJ, Wang CY, Chen YH, **Niu DM**, Lin SJ, Chen JW, Chiou SH, Leu HB. Energy utilization of induced pluripotent stem cell-derived cardiomyocyte in Fabry disease. *Int J Cardiol*. 2017. pii: S0167-5273 (17) 30028-1.
51. Hsu TR, Chang FP, Chu TH, Sung SH, Bizjajeva S, Yu WC, **Niu DM*.** Correlations between Endomyocardial Biopsies and Cardiac Manifestations in Taiwanese Patients with the Chinese Hotspot IVS4+919G>A Mutation: Data from the Fabry Outcome Survey. *Int J Mol Sci*. 2017. 68:2554-2563.
52. Bu XM, **Niu DM**, Wu J, Yuan YL, Song JX, Wang JJ. Elevated levels of preβ1-high-density lipoprotein are associated with cholesterol ester transfer protein, the presence and severity of coronary artery disease. *Lipids Health Dis*. 2017. 16(1):4.