臺北榮民總醫院
臨床共同研究室

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學歷:

- 國立陽明大學臨床醫學所博士, 2001-2004
- 美國杜克大學醫學遺傳學研究員, 1995-1996
- 高雄醫學院醫學系學士, 1980-1987

重要經歷:

- 臺北榮民總醫院 兒童醫學部部主任
- 臺北榮民總醫院 罕見疾病治療中心主任
- 國立陽明交通大學 臨床醫學所教授
- 臺北榮民總醫院 遺傳諮詢中心主任
- 臺灣兒科醫學會 醫學遺傳學／新陳代謝學 主任委員

研究方向: (關鍵詞)

先天遺傳及代謝疾病、罕見疾病、臨床試驗、基因檢測、基因治療、精準醫療、人工智慧全基因定序分析診斷

五年內代表著作:

(*, Corresponding author)

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.

五年內 代表著作(續)	6. 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 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, **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 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, **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. 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, **Niu DM***. 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, **Niu DM***. 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, **Niu DM***, 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.
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