

CURRICULUM VITAE

Dau-Ming Niu, M.D., Ph. D. 牛道明

連絡電話: 02-77368488

Email: dmniu1111@yahoo.com.tw

地址: 台北榮民總醫院科技大樓 8 樓遺傳諮詢中心

現職：

- | | | |
|------------------|------|-------------|
| 1. 國立陽明大學臨床醫學研究所 | 教授 | (2011/02 起) |
| 2. 台北榮民總醫院遺傳諮詢中心 | 主任 | (2009 起) |
| 3. 台北榮民總醫院兒童醫學部 | 主治醫師 | (1994/11 起) |

學歷：

學校名稱	國別	主修	學位	起訖年月(西元年/月)
國立陽明大學	台灣	臨床醫學所	博士	自 2001/9 月至 2004/7 月
Duke 大學	美國	醫學遺傳學進修	Research fellow	自 1995/9 月至 1996/7 月
台灣大學附設醫院	台灣	小兒部	Research fellow	自 1993/9 月至 1994/7 月
高雄醫學院	台灣	醫學系	學士	自 1980/9 月至 1987/7 月

經歷：

服務機構	服務部門	職稱	起訖年月(西元年/月)
陽明大學	臨床醫學所	副教授	自 2008/2 月至 2011/02 月
陽明大學	臨床醫學所	助理教授	自 2004/8 月至 2008/01 月
台北榮民總醫院	兒童醫學部	主治醫師	自 1994/11 月迄今
台北榮民總醫院	兒童醫學部	住院醫師	自 1989/6 月至 1994/10 月

專長：

1. 分子生物學
2. 小兒內分泌學
3. 小兒遺傳學
4. 小兒新陳代謝學

獲獎:

1. 99 年度行政院國軍退除役官兵輔導委員會優良醫師
2. 國民健康局「99 年度照護罕見疾病個案之績優醫事人員獎」第一名
3. 99 年陽明大學臨床醫學所優良教師
4. 96 年度行政院國軍退除役官兵輔導委員會優良醫師
5. 95 年度第二屆教育百人團總統獎
6. 95 年度行政院國軍退除役官兵輔導委員會優良醫師

學術論文著作目錄:

(*Corresponding author)

1. Tai CL, Liu MY, Yu HC, Chiang CC, Chiang H, Suen JH, Kao SM, Huang YH, Wu TJ, Yang CF, Tsai FC, Lin CY, Chang JG, Chen HD, Niu DM*. (2012). The use of high resolution melting analysis to detect Fabry mutations in heterozygous females via dry bloodspots. **Clinica Chimica Acta**. 413(3-4):422-7.
2. Niu DM*. (2011). Disorders of BH4 metabolism and the treatment of patients with 6-pyruvoyl-tetrahydropterin synthase deficiency in Taiwan. **Brain & Development**. 33(10):847-55
3. Niu DM, Lin SY, Li MJ, Cheng WT, Pan CC, Lin CC. (2011). Idiopathic calcinosis cutis in a child: chemical composition of the calcified deposits. **Dermatology**, **222**(3), 201-5.
4. Weng HJ, Niu DM, Turale S, Tsao LI, Shih FJ, Yamamoto-Mitani N, Chang CC, Shih FJ. (2011). Family caregiver distress with children having rare genetic disorders: a qualitative study involving Russell-Silver Syndrome in Taiwan. **Journal of Clinical Nursing**. 21(1-2):160-169
5. Jap TS, Chiu CY, Niu DM, Levine MA. (2011). Three novel mutations in the PHEX gene in Chinese subjects with hypophosphatemic rickets extends genotypic variability. **Calcified Tissue International**, **88**(5), 370-7.
6. Liao HM, Niu DM, Chen YJ, Fang JS, Chen SJ, Chen CH. (2011). Identification of a microdeletion at Xp22.13 in a Taiwanese family presenting with Nance-Horan syndrome. **Journal of Human Genetics**, **56**(1), 8-11. (equal to 1st author)
7. Lin SY, Hsieh SC, Lin YC, Lee CN, Tsai MH, Lai LC, Chuang EY, Chen PC, Hung CC, Chen LY, Hsieh WS, Niu DM, Su YN, Ho HN. (2011). A whole genome methylation analysis of systemic lupus erythematosus: hypomethylation of the IL10 and IL1R2 promoters is associated with disease activity. **Genes & Immunity**. 2011 Nov 3. [Epub ahead of print]
8. Chiang CY, Huang KH, Fang WL, Wu CW, Chen JH, Lo SS, Hsieh MC, Shen KH, Li AF, Niu DM, Chiou SH. (2011). Factors associated with recurrence within 2 years after curative surgery for gastric adenocarcinoma. **World Journal of Surgery**, **35**(11), 2472-8.
9. Fang WL, Huang KH, Chen JH, Lo SS, Hsieh MC, Shen KH, Li AF, Niu DM, Chiou SH, Wu CW. (2011).

10. Comparison of the Survival Difference Between AJCC 6th and 7th Editions for Gastric Cancer Patients. **World Journal of Surgery**. 2011 Sep 15. [Epub ahead of print]
11. Chiang CY, Huang KH, Fang WL, Wu CW, Chen JH, Lo SS, Hsieh MC, Shen KH, Li AF, Niu DM, Chiou SH. (2011). Factors associated with recurrence within 2 years after curative surgery for gastric adenocarcinoma. **World Journal of Surgery**, **35**(11), 2472-8.
12. Lin HY, Huang CH, Yu HC, Chong KW, Hsu JH, Lee PC, Cheng KH, Chiang CC, Ho HJ, Lin SP, Chen SJ, Lin PK, Niu DM*. (2010). Enzyme assay and clinical assessment in subjects with a Chinese hotspot late-onset Fabry mutation (IVS4+919G>A). **Journal of Inherited Metabolic Disease**, **33**(5):619-24.
13. Lin HY, Wu TJ, Hsu JH, Yu HC, Chuang CK, Huang CH, Niu DM*. (2010). Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. **Human Genetics**, **127**(4), 464.
14. Niu DM, Lin HY, Wu TJ, Hsu JH, Yu HC, Lin SP, Chuang CK, Huang CH. (2010). Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. **Human Genetics**, **127**(4), 465.
15. Niu DM, Lin HY, Wu TJ, Hsu JH, Yu HC, Lin SP, Chuang CK, Huang CH. (2010). Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. **Human Genetics**, **127** (4), 465.
16. Lin HY, Wu JT, Hsu JH, Yu HC, Lin SP, Huang CH, Niu DM*. (2010). Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. **Human Genetics**, **127** (4), 464.
17. Niu DM, Lin HY, Wu TJ, Hsu JH, Yu HC, Lin SP, Chuang CK, Huang CH. (2010). Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. **Human Genetics**, **127** (4), 466.
18. Lin HY, Wu TJ, Hsu JH, Yu HC, Lin SP, Chuang CK, Huang CH, Niu DM*. (2010). Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. **Human Genetics**, **127** (4), 465.
19. Lin HY, Chen MR, Chuang CK, Chen CP, Lin DS, Chien YH, Ke YY, Tsai FJ, Pan HP, Lin SJ, Hwu WL, Niu DM, Lee NC, Lin SP. (2010). Erratum to: Enzyme replacement therapy for mucopolysaccharidosis VI-experience in Taiwan. **Journal of Inherited Metabolic Disease**. 2010 Nov 3. [Epub ahead of print]
20. Lin SY, Lee CN, Hung CC, Tsai WY, Lin SP, Li NC, Hsieh WS, Tung YC, Niu DM, Hsu WM, Chen LY, Fang MY, Tu MP, Kuo PW, Lin CY, Su YN, Ho HN. (2010). Epigenetic profiling of the H19 differentially methylated region and comprehensive whole genome array-based

analysis in Silver-Russell syndrome. **American Journal of Medical Genetics Part A**, **152A**(10), 2521-8.

21. Liu C, Niu DM, Hsia CY, Loong CC, Lin NC, Tsai HL, Tsou MY, Chin T. (2010). Living donor liver transplantation using a graft from a donor with Dubin-Johnson syndrome. **Pediatric Transplantation**. 2010 Sep 1. [Epub ahead of print]
22. Lin HY, Chen MR, Lin CC, Chen CP, Lin DS, Chuang CK, Niu DM, Chang JH, Lee HC, Lin SP. (2010). Polysomnographic characteristics in patients with mucopolysaccharidoses. **Pediatric Pulmonology**, **45**(12), 1205-12.
23. Tsao YC, Niu DM, Chen JT, Lin CY, Lin YY, Liao KK. (2010). Gabapentin reduces neurovisceral pain of porphyria. **Acta Neurologica Taiwanica**, **19**(2), 112-5.
24. Cheng KH, Liu MY, Kao CH, Chen YJ, Hsiao KJ, Liu TT, Lin HY, Huang CH, Chiang CC, Ho HJ, Lin SP, Lee NC, Hwu WL, Lin JL, Hung PY, Niu DM*. (2010). Newborn screening for methylmalonic aciduria by tandem mass spectrometry: 7 years' experience from two centers in Taiwan. **Journal of the Chinese Medical Association**, **73**(6), 314-8.
25. Niu DM*, Chien YH, Chiang CC, Ho HC, Hwu WL, Kao SM, Chiang SH, Kao CH, Liu TT, Chiang H, Hsiao KJ. (2010). Nationwide survey of extended newborn screening by tandem mass spectrometry in Taiwan. **Journal of Inherited Metabolic Disease**, **33**(Suppl 2), S295-305.
26. Niu DM*, Chong KW, Hsu JH, Wu TJ, Yu HC, Huang CH, Lo MY, Kwok CF, Kratz LE, Ho LT. (2010). Clinical observations, molecular genetic analysis, and treatment of sitosterolemia in infants and children. **Journal of Inherited Metabolic Disease**, **33**(4), 437-43.
27. Tsai YG, Yang KD, Niu DM, Chien JW, Lin CY. (2010). TLR2 agonists enhance CD8+Foxp3+ regulatory T cells and suppress Th2 immune responses during allergen immunotherapy. **Journal of immunology**, **184**(12), 7229-37.
28. Tseng LL, Lue HC, Huang CH, Niu DM*. (2010). Severe hyponatremia due to ACTH insufficiency in a 14 year-old girl with growth hormone deficiency. **Journal of Pediatric Endocrinology & Metabolism**, **23**(1-2), 197-201.
29. Lin HY, Chong KW, Hsu JH, Yu HC, Huang CH, Niu DM*. (2010). Novel human pathological mutations. Gene symbol: GLA. Disease: Fabry disease. **Human Genetics**, **127**(1), 124.
30. Liu C, Niu DM, Loong CC, Hsia CY, Tsou MY, Tsai HL, Wei C. (2010). Domino liver graft from a patient with homozygous familial hypercholesterolemia. **Pediatric Transplantation**, **14**(3), E30-3.
31. Niu DM, Lin HY, Chong KW, Hsu JH, Yu HC, Huang CH. (2010). Novel human pathological mutations. Gene symbol: GLA. Disease: Fabry disease. **Human Genetics**, **127**(1), 122.

32. Chien JC, Niu DM, Wang MS, Liu MT, Lirng JF, Chen SJ, Hwang B. (2010). Giant congenital melanocytic nevi in neonates: report of two cases. **Pediatrics & Neonatology**, **51**(1), 61-4.
33. Lin HY, Niu DM, Chong KW, Hsu JH, Yu HC, Huang CH. (2010). Novel human pathological mutations. Gene symbol: GLA. Disease: Fabry disease. **Human Genetics**, **127**(1), 122-3
34. 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*. (2009). High incidence of the cardiac variant of Fabry disease revealed by newborn screening in the Taiwan Chinese population. **Circulation: Cardiovascular Genetics**, **2**(5), 450-6.
35. Hung CC, Lin SY, Lee CN, Cheng HY, Lin SP, Chen MR, Chen CP, Chang CH, Lin CY, Yu CC, Chiu HH, Cheng WF, Ho HN, Niu DM*, Su YN*. (2009). Mutation spectrum of the fibrillin-1 (FBN1) gene in Taiwanese patients with Marfan syndrome. **Annals of Human Genetics**, **73**(Pt 6), 559-67.
36. Niu DM*, Hsu JH, Chong KW, Huang CH, Lu YH, Kao CH, Yu HC, Lo MY, Jap TS. (2009). Six new mutations of the thyroglobulin gene discovered in taiwanese children presenting with thyroid dysmorphogenesis. **The Journal of Clinical Endocrinology & Metabolism**, **94**(12), 5045-52.
37. Tiu CM, Liu TC, Hsieh CW, Niu DM, Chen JD, Jong TL. (2009). Turner syndrome phalangeal screening based on a two-stage linear regression concept. **Pediatrics International**, **51**(4), 453-9.
38. Shen CM, Lin SC, Niu DM, Kou YR. (2009). Labour increases the surface expression of two Toll-like receptors in the cord blood monocytes of healthy term newborns. **Acta Paediatrica Taiwanica**, **98**(6), 959-62.
39. Lin HY, Lin SP, Chuang CK, Niu DM, Chen MR, Tsai FJ, Chao MC, Chiu PC, Lin SJ, Tsai LP, Hwu WL, Lin JL. (2009). Incidence of the mucopolysaccharidoses in Taiwan, 1984-2004. **American Journal of Medical Genetics Part A**, **149A**(5), 960-4.
40. Extreme hypernatremia combined with rhabdomyolysis and acute renal failure. (2009). Yang TY, Chang JW, Tseng MH, Wang HH, Niu DM, Yang LY. **Journal of the Chinese Medical Association**, **72**(10), 555-8.
41. Kao CH, Liu MY, Liu TT, Hsiao KJ, Cheng KH, Huang CH, Lin HY, Niu DM*. (2009). Growth hormone therapy in neonatal patients with methylmalonic acidemia. **Journal of the Chinese Medical Association**, **72**(9), 462-7.
42. Lin HY, Lin SP, Chuang CK, Chen MR, Chang CY, Niu DM*. (2009). Clinical features of osteogenesis imperfecta in Taiwan. **Journal of the Formosan Medical Association**, **108**(7), 570-6.

43. Hung CC, Lin SY, Lee CN, Cheng HY, Lin CY, Chang CH, Chiu HH, Yu CC, Lin SP, Cheng WF, Ho HN, Niu DM*, Su YN*. (2009). Identification of fibrillin-1 gene mutations in Marfan syndrome by high-resolution melting analysis. **Analytical Biochemistry**, **389**(2), 102-6.
44. Lee YJ, Tsai LP, Niu DM, Shu SG, Chao MC, Lee HH. (2009). The gene founder effect of two spontaneous mutations in ethnic Chinese (Taiwanese) CAH patients with 21-hydroxylase deficiency. **Molecular Genetics and Metabolism**, **97**(1), 75-9.
45. Hung CC, Lin SY, Lin SP, Niu DM, Lee NC, Cheng WF, Chen CP, Lin WL, Lee CN, Su YN. (2009). Identification of CpG methylation of the SNRPN gene by methylation-specific multiplex PCR. **ELECTROPHORESIS**, **30**(2):410-6.
46. Liu KM, Liu TT, Lee NC, Cheng LY, Hsiao KJ, Niu DM*. (2008). Long-term follow-up of Taiwanese Chinese patients treated early for 6-pyruvoyl-tetrahydropterin synthase deficiency. **Archives of Neurology**, **65**(3), 387-92.
47. Lee HH, Lee YJ, Wang YM, Chao HT, Niu DM, Chao MC, Tsai FJ, Lo FS, Lin SJ. (2008). Low frequency of the CYP21A2 deletion in ethnic Chinese (Taiwanese) patients with 21-hydroxylase deficiency. **Molecular Genetics and Metabolism**, **93**(4), 450-7.
48. Lee YY, Wong TT, Fang YT, Chang KP, Chen YW, Niu DM. (2008). Comparison of hypothalamopituitary axis dysfunction of intrasellar and third ventricular craniopharyngiomas in children. **Brain & Development**, **30**(3), 189-94.
49. Cheng KH, Hung MC, Chen SJ, Kao CH, Niu DM*. (2007). Lenticular subluxation in a patient with homocystinuria undetected by neonatal screening. **Journal of the Chinese Medical Association**, **70**(12), 562-4.
50. Lin HY, Lin SP, Chuang CK, Chen MR, Yen JL, Lee YJ, Huang CY, Tsai LP, Niu DM, Chao MC, Kuo PL. (2007). Genotype and phenotype in patients with Prader-Willi syndrome in Taiwan. **Acta Paediatrica**, **96**(6), 902-5.
51. Lin HY, Lin SP, Yen JL, Lee YJ, Huang CY, Hung HY, Hsu CH, Kao HA, Chang JH, Chiu NC, Ho CS, Chao MC, Niu DM, Tsai LP, Kuo PL. (2007). Prader-Willi syndrome in Taiwan. **Pediatrics International**, **49**(3), 375-9.
52. Chiang MC, Chen HM, Lee YH, Chang HH, Wu YC, Soong BW, Chen CM, Wu YR, Liu CS, Niu DM, Wu JY, Chen YT, Chern Y. (2007). Dysregulation of C/EBPalpha by mutant Huntingtin causes the urea cycle deficiency in Huntington's disease. **Human Molecular Genetics**, **16**(5), 483-98.
53. Niu DM, Huang JY, Li HY, Liu KM, Wang ST, Chen YJ, Udaka T, Izumi K, Kosaki K. (2006). Paternal gonadal mosaicism of NIPBL mutation in a father of siblings with Cornelia de Lange syndrome. **Prenatal Diagnosis**, **26**(11), 1054-7.

54. Tsai CP, Lin PY, Lee NC, Niu DM, Lee SM, Hsu WM. (2006). Corneal lesion as the initial manifestation of tyrosinemia type II. **Journal of the Chinese Medical Association**, **69**(6), 286-8.
55. Yen JL, Lin SP, Chen MR, Niu DM. (2006). Clinical features of Ehlers-Danlos syndrome. **Journal of the Formosan Medical Association**, **105**(6), 475-80.
56. Niu DM, Hwang B, Hwang HW, Wang NH, Wu JY, Lee PC, Chien JC, Shieh RC, Chen YT. (2006). 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. **Journal of Medical Genetics**, **43**(10), 817-21.
57. Lee NC, Hwang B, Chen CH, Niu DM*. (2006). Intrafamilial phenotype variation in Marfan syndrome ascertained by intragenic linkage analysis. **Journal of the Formosan Medical Association**, **104**(12), 964-7.
58. Lee NC, Cheng LY, Liu TT, Hsiao KJ, Chiu PC, Niu DM*. (2006). Long-term follow-up of Chinese patients who received delayed treatment for 6-pyruvoyl-tetrahydropterin synthase deficiency. **Molecular Genetics and Metabolism**, **87**(2), 128-34.
59. Lee NC, Niu DM, Lin CY, Hsiao KJ, Yang AH, Ng YY. (2006). Alpha-galactosidase activity should be examined in patients with proteinuria: what have we learned from a family affected with Fabry disease? **Nephrology Dialysis Transplantation**, **21**(2), 549-50.
60. Lu CK, Chen SJ, Niu DM, Tsai CC, Lee FL, Hsu WM. (2005). Electrophysiological changes in lipaemia retinalis. **American Journal of Ophthalmology**, **139**(6), 1142-5.
61. Chien JC, Chen SJ, Tiu CM, Chen YJ, Hwang B, Niu DM*. (2005). Is urorectal septum malformation sequence a variant of the vertebral defects, anal atresia, tracheo-oesophageal fistula, renal defects and radial dysplasia association? Report of a case and a review of the literature. **European Journal of Pediatrics**, **164**(6), 350-4.
62. Hwang HW, Chen JJ, Lin YJ, Shieh RC, Lee MT, Hung SI, Wu JY, Chen YT, Niu DM, Hwang BT. (2005). R1193Q of SCN5A, a Brugada and long QT mutation, is a common polymorphism in Han Chinese. **Journal of Medical Genetics**, **42**(2), e7, author reply e8.
63. Niu DM, Lin CY, Hwang B, Jap TS, Liao CJ, Wu JY. (2005). Contribution of genetic factors to neonatal transient hypothyroidism. **Arch Dis Child - Fetal Neonatal Ed**, **90**(1), F69-72.
64. Niu DM, Hwang B, Tiu CM, Tsai LP, Yen JL, Lee NC, Lin CY. (2004). Contributions of bone maturation measurements to the differential diagnosis of neonatal transient hypothyroidism versus dysmorphogenetic congenital hypothyroidism. **Acta Paediatrica Taiwanica**, **93**(10), 1301-6.
65. Guo YC, Liao KK, Soong BW, Tsai CP, Niu DM, Lee HY, Lin KP. (2004). Congenital

insensitivity to pain with anhidrosis in Taiwan: a morphometric and genetic study. **European Neurology**, **51**(4), 206-14.

66. Kao CD, Niu DM, Chen JT, Shan DE, Lin YY, Wu ZA, Liao KK. (2004). Subtle brain dysfunction in treated 6-pyruvoyl-tetrahydropterin synthase deficiency: relationship to motor tasks and neurophysiological tests. **Brain & Development** , **26**(2), 93-8.
67. Lee HH, Niu DM, Lin RW, Chan P, Lin CY. (2002). Structural analysis of the chimeric CYP21P/CYP21 gene in steroid 21-hydroxylase deficiency. **Journal of Human Genetics**, **47**(10), 517-22.
68. Niu DM, Hwang B, Chu YK, Liao CJ, Wang PL, Lin CY. (2002). 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. **The Journal of Clinical Endocrinology & Metabolism**, **87**(9),4208-12.
69. Niu DM, Pan CC, Lin CY, Hwang B, Chung MY. (2002). Mosaic or chimera? Revisiting an old hypothesis about the cause of the 46,XX/46,XY hermaphrodite. **Journal of Pediatrics**, **140**(6):732-5.
70. Niu DM, Guo WY, Pan HC, Wong TT. (2002). Rapid enlargement of a residual craniopharyngioma during short-term growth hormone replacement. **Child's Nervous System**, **18**(3-4):164-5.
71. Chung WT, Niu DM, Lin CY. (2002). Clinical aspects of X-linked hypophosphatemic rickets. **Acta Paediatrica Taiwanica**, **43**(1), 26-34.
72. Lin SY, Niu DM, Tu CP, Lin HL, Li MJ, Cheng YD. (2001). Diagnosis of congenital hypothyroidism from human anagen scalp hair by infrared microspectroscopy. **Ultrastructural Pathology**, **25**(5), 357-60.
73. Lin WD, Wu JY, Lai CC, Tsai FJ, Tsai CH, Lin SP, Niu DM. (2001). A pilot study of neonatal screening by electrospray ionization tandem mass spectrometry in Taiwan. **Acta Paediatrica Taiwanica**, **42**(4), 224-30.
74. Lin SY, Niu DM, Li MJ, Tu CP, Lin HL. (2000). Differentiation of hair growth cycle from scalp hair roots for the diagnosis of glucose-6-phosphate dehydrogenase deficiency in neonates. **Journal of Medical Genetics** , **23**(7), 693-704.
75. Chen CH, Tiu CM, Chou YH, Chen WY, Hwang B, Niu DM*. (1999). Congenital hypothyroidism with multiple ovarian cysts. **European Journal of Pediatrics**, **158**(10), 851-2.
76. Niu DM, Hsiao KJ, Wang NH, Chin LS, Chen CH. (1996). Chinese achondroplasia is also defined by recurrent G380R mutations of the fibroblast growth factor receptor-3 gene. **Human Genetics**, **98**(1), 65-7.