Dau-Ming Niu

List of Publications by Year
in descending order

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120 papers	2,483 citations	218677 26 h-index	265206 42 g-index
126 all docs	126 docs citations	126 times ranked	2881 citing authors

#	Article	IF	Citations
1	Genetic Analysis in a Taiwanese Cohort of 750 Index Patients with Clinically Diagnosed Familial Hypercholesterolemia. Journal of Atherosclerosis and Thrombosis, 2022, 29, 639-653.	2.0	11
2	Reduced global longitudinal strain as a marker for early detection of Fabry cardiomyopathy. European Heart Journal Cardiovascular Imaging, 2022, 23, 487-495.	1.2	19
3	Airway abnormalities and pulmonary complications in longâ€term treated lateâ€onset Pompe disease: Diagnostic and interventional by flexible bronchoscopy. Pediatric Pulmonology, 2022, 57, 185-192.	2.0	1
4	Safety and longâ€term outcomes of early liver transplantation for pediatric methylmalonic acidemia patients. Pediatric Transplantation, 2022, 26, e14228.	1.0	1
5	Prevalence of lower urinary tract symptoms in children with earlyâ€treated infantileâ€onset Pompe disease: A singleâ€centre crossâ€sectional study. Neurourology and Urodynamics, 2022, , .	1.5	0
6	Twenty years of the Fabry Outcome Survey (FOS): insights, achievements, and lessons learned from a global patient registry. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	14
7	Natural progression of cardiac features and long-term effects of enzyme replacement therapy in Taiwanese patients with mucopolysaccharidosis II. Orphanet Journal of Rare Diseases, 2021, 16, 99.	2.7	10
8	Aortic regurgitation in Marfan syndrome patients who underwent prophylactic surgery: A single-center experience. Journal of the Chinese Medical Association, 2021, 84, 540-544.	1.4	1
9	The benefits and challenges of family genetic testing in rare genetic diseasesâ€"lessons from Fabry disease. Molecular Genetics & Genomic Medicine, 2021, 9, e1666.	1.2	26
10	Genetic basis and hematologic manifestations of sitosterolemia in a group of Turkish patients. Journal of Clinical Lipidology, 2021, , .	1.5	2
11	Hearing characteristics of infantile-onset Pompe disease after early enzyme-replacement therapy. Orphanet Journal of Rare Diseases, 2021, 16, 348.	2.7	7
12	Aortic Root Dilatation in Taiwanese Patients with Mucopolysaccharidoses and the Long-Term Effects of Enzyme Replacement Therapy. Diagnostics, 2021, 11, 16.	2.6	5
13	Quantitative DNA Methylation Analysis and Epigenotype-Phenotype Correlations in Taiwanese Patients with Beckwith-Wiedemann Syndrome. Journal of Personalized Medicine, 2021, 11, 1066.	2.5	0
14	Epigenotype, Genotype, and Phenotype Analysis of Taiwanese Patients with Silverâ€"Russell Syndrome. Journal of Personalized Medicine, 2021, 11, 1197.	2.5	0
15	Congenital hypopituitarism due to novel compound heterozygous POU1F1 gene mutation: A case report and review of the literature. Molecular Genetics and Metabolism Reports, 2021, 29, 100819.	1.1	2
16	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. Frontiers in Pediatrics, 2021, 9, 729824.	1.9	1
17	Ultrasonography-Based Qualitative and Quantitative Evaluation Approaches for Pompe Disease. Journal of Medical and Biological Engineering, 2020, 40, 189-203.	1.8	2
18	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€"2019). Orphanet Journal of Rare Diseases, 2020, 15, 314.	2.7	16

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19	Allogeneic hematopoietic stem cell transplantation for treating severe lung involvement in Gaucher disease. Molecular Genetics and Metabolism Reports, 2020, 25, 100652.	1.1	2
20	Cardiac manifestations in patients with classical or cardiac subtype of Fabry disease. Journal of the Chinese Medical Association, 2020, 83, 825-829.	1.4	6
21	Diagnosis of Arboleda-Tham syndrome by whole genome sequencing in an Asian boy with severe developmental delay. Molecular Genetics and Metabolism Reports, 2020, 25, 100686.	1.1	5
22	Audiological and otologic manifestations of glutaric aciduria type I. Orphanet Journal of Rare Diseases, 2020, 15, 337.	2.7	4
23	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, e035182.	1.9	20
24	The Fabry disease-causing mutation, GLA IVS4+919G>A, originated in Mainland China more than 800 years ago. Journal of Human Genetics, 2020, 65, 619-625.	2.3	9
25	Airway abnormalities in very early treated infantileâ€onset Pompe disease: A largeâ€scale survey by flexible bronchoscopy. American Journal of Medical Genetics, Part A, 2020, 182, 721-729.	1.2	9
26	Fabry disease and COVID-19: international expert recommendations for management based on real-world experience. CKJ: Clinical Kidney Journal, 2020, 13, 913-925.	2.9	11
27	Cardiac Evaluation Using Two-Dimensional Speckle-Tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. Diagnostics, 2020, 10, 62.	2.6	9
28	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, 2019, 21, 224-232.	2.4	10
29	Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with Different Types of Mucopolysaccharidoses. Diagnostics, 2019, 9, 148.	2.6	11
30	An At-Risk Population Screening Program for Mucopolysaccharidoses by Measuring Urinary Glycosaminoglycans in Taiwan. Diagnostics, 2019, 9, 140.	2.6	10
31	Functional independence of Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e790.	1.2	6
32	Response to Juang et al.. Genetics in Medicine, 2019, 21, 1892-1893.	2.4	0
33	Cardiac characteristics and natural progression in Taiwanese patients with mucopolysaccharidosis III. Orphanet Journal of Rare Diseases, 2019, 14, 140.	2.7	14
34	Very rare condition of multiple Gaucheroma: A case report and review of the literature. Molecular Genetics and Metabolism Reports, 2019, 20, 100473.	1.1	10
35	Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e00617.	1.2	13
36	Methylmalonic acidemia/propionic acidemia â€“ the biochemical presentation and comparing the outcome between liver transplantation versus non-liver transplantation groups. Orphanet Journal of Rare Diseases, 2019, 14, 73.	2.7	26

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37	Cardio- Renal Outcomes With Long- Term Agalsidase Alfa Enzyme Replacement Therapy: A 10-Year Fabry Outcome Survey (FOS) Analysis . Drug Design, Development and Therapy, 2019, Volume 13, 3705-3715.	4.3	19
38	Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. Pediatrics and Neonatology, 2019, 60, 342-343.	0.9	9
39	Functional independence of Taiwanese children with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1309-1314.	1.2	2
40	Functional and biological studies of Î±-galactosidase A variants with uncertain significance from newborn screening in Taiwan. Molecular Genetics and Metabolism, 2018, 123, 140-147.	1.1	14
41	Fabry disease: Review and experience during newborn screening. Trends in Cardiovascular Medicine, 2018, 28, 274-281.	4.9	47
42	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. Journal of Human Genetics, 2018, 63, 1-8.	2.3	18
43	The relationships between urinary glycosaminoglycan levels and phenotypes of mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2018, 6, 982-992.	1.2	24
44	Cardiac features and effects of enzyme replacement therapy in Taiwanese patients with Mucopolysaccharidosis IVA. Orphanet Journal of Rare Diseases, 2018, 13, 148.	2.7	18
45	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. Molecular Genetics and Metabolism Reports, 2018, 16, 5-10.	1.1	94
46	Mucopolysaccharidosis III in Taiwan: Natural history, clinical and molecular characteristics of 28 patients diagnosed during a 21â€year period. American Journal of Medical Genetics, Part A, 2018, 176, 1799-1809.	1.2	24
47	Clinical characteristics and surgical history of Taiwanese patients with mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS). Orphanet Journal of Rare Diseases, 2018, 13, 89.	2.7	10
48	Energy utilization of induced pluripotent stem cell-derived cardiomyocyte in Fabry disease. International Journal of Cardiology, 2017, 232, 255-263.	1.7	33
49	Biomarkers associated with clinical manifestations in Fabry disease patients with a late-onset cardiac variant mutation. Clinica Chimica Acta, 2017, 466, 185-193.	1.1	44
50	Mass Spectrometry but Not Fluorimetry Distinguishes Affected and Pseudodeficiency Patients in Newborn Screening for Pompe Disease. Clinical Chemistry, 2017, 63, 1271-1277.	3.2	29
51	Amelioration of serum 8-OHdG level by enzyme replacement therapy in patients with Fabry cardiomyopathy. Biochemical and Biophysical Research Communications, 2017, 486, 293-299.	2.1	12
52	Experiences during newborn screening for glutaric aciduria type 1: Diagnosis, treatment, genotype, phenotype, and outcomes. Journal of the Chinese Medical Association, 2017, 80, 253-261.	1.4	26
53	Muscle ultrasound. Medicine (United States), 2017, 96, e8415.	1.0	14
54	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 Neurology, 2017, 17, 25.	1.8	13

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55	Correlations between Endomyocardial Biopsies and Cardiac Manifestations in Taiwanese Patients with the Chinese Hotspot IVS4+919G>A Mutation: Data from the Fabry Outcome Survey. International Journal of Molecular Sciences, 2017, 18, 119.	4.1	9
56	Modulation the alternative splicing of GLA (IVS4+919G>A) in Fabry disease. PLoS ONE, 2017, 12, e0175929.	2.5	15
57	Homozygosity Mapping and Whole-Genome Sequencing Links a Missense Mutation inPOMGNT1to Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 3601.		18
58	Later Onset Fabry Disease, Cardiac Damage Progress in Silence. Journal of the American College of Cardiology, 2016, 68, 2554-2563.	2.8	81
59	Causes of death and clinical characteristics of 34 patients with Mucopolysaccharidosis II in Taiwan from 1995â€“2012. Orphanet Journal of Rare Diseases, 2016, 11, 85.	2.7	26
60	Evaluation of Proinflammatory Prognostic Biomarkers for Fabry Cardiomyopathy With Enzyme Replacement Therapy. Canadian Journal of Cardiology, 2016, 32, 1221.e1-1221.e9.	1.7	35
61	Long-term galsulfase enzyme replacement therapy in Taiwanese mucopolysaccharidosis VI patients: A case series. Molecular Genetics and Metabolism Reports, 2016, 7, 63-69.	1.1	27
62	Cognitive Development in Infantile-Onset Pompe Disease Under Very Early Enzyme Replacement Therapy. Journal of Child Neurology, 2016, 31, 1617-1621.	1.4	11
63	Epigenotype, genotype, and phenotype analysis of patients in Taiwan with Beckwithâ€“Wiedemann syndrome. Molecular Genetics and Metabolism, 2016, 119, 8-13.	1.1	13
64	A 15-Year Perspective of the Fabry Outcome Survey. FIRE Forum for International Research in Education, 2016, 4, 232640981666629.	0.7	9
65	Fabry in the older patient: Clinical consequences and possibilities for treatment. Molecular Genetics and Metabolism, 2016, 118, 319-325.	1.1	15
66	Functional independence of Taiwanese children with Down syndrome. Developmental Medicine and Child Neurology, 2016, 58, 502-507.	2.1	13
67	Brain MR Imaging Findings of Cardiac-Type Fabry Disease with an IVS4+919G>A Mutation. American Journal of Neuroradiology, 2016, 37, 1044-1049.	2.4	15
68	Cardiac structure and function and effects of enzyme replacement therapy in patients with mucopolysaccharidoses I, II, IVA and VI. Molecular Genetics and Metabolism, 2016, 117, 431-437.	1.1	37
69	Measuring propionyl-CoA carboxylase activity in phytohemagglutinin stimulated lymphocytes using high performance liquid chromatography. Clinica Chimica Acta, 2016, 453, 13-20.	1.1	5
70	Very Early Treatment for Infantile-Onset Pompe Disease Contributes toÂBetter Outcomes. Journal of Pediatrics, 2016, 169, 174-180.e1.	1.8	85
71	Genotype and phenotype analysis of Taiwanese patients with osteogenesis imperfecta. Orphanet Journal of Rare Diseases, 2015, 10, 152.	2.7	30
72	Clinical observations and treatment of pediatric homozygous familial hypercholesterolemia due to a low-density lipoprotein receptor defect. Journal of Clinical Lipidology, 2015, 9, 234-240.	1.5	9

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73	Late-Onset Pompe Disease With Left-Sided Bronchomalacia. Respiratory Care, 2015, 60, e26-e29.	1.6	20
74	Age at First Cardiac Symptoms in Fabry Disease: Association with a Chinese Hotspot Fabry Mutation (IVS4+919G>A), Classical Fabry Mutations, and Sex in a Taiwanese Population from the Fabry Outcome Survey (FOS). JIMD Reports, 2015, 22, 107-113.	1.5	7
75	Heterozygous carriers of classical homocystinuria tend to have higher fasting serum homocysteine concentrations than non-carriers in the presence of folate deficiency. Clinical Nutrition, 2015, 34, 1155-1158.	5.0	4
76	A largeâ€scale nationwide newborn screening program for pompe disease in Taiwan: Towards effective diagnosis and treatment. American Journal of Medical Genetics, Part A, 2014, 164, 54-61.	1.2	59
77	Endomyocardial biopsies in patients with left ventricular hypertrophy and a common Chinese later-onset fabry mutation (IVS4â€‰+â€‰919Gâ€‰>â€‰A). Orphanet Journal of Rare Diseases, 2014, 9, 96.	2.7	30
78	Electroencephalography and transcranial Doppler ultrasonography in neonatal citrullinemia. Journal of the Formosan Medical Association, 2014, 113, 857-861.	1.7	4
79	Detecting multiple lysosomal storage diseases by tandem mass spectrometry â€" A national newborn screening program in Taiwan. Clinica Chimica Acta, 2014, 431, 80-86.	1.1	97
80	A novel mutation of ABCG5 gene in a Turkish boy with phytosterolemia presenting with macrotrombocytopenia and stomatocytosis. Pediatric Blood and Cancer, 2014, 61, 1457-1459.	1.5	17
81	Hepatomegaly and hyperammonemia in a girl with Silverâ€"Russell syndrome caused by maternal uniparental isodisomy of chromosome 7. American Journal of Medical Genetics, Part A, 2014, 164, 2114-2117.	1.2	1
82	The mutation spectrum of the phenylalanine hydroxylase (PAH) gene and associated haplotypes reveal ethnic heterogeneity in the Taiwanese population. Journal of Human Genetics, 2014, 59, 145-152.	2.3	19
83	Globotriaosylsphingosine (lyso-Gb3) might not be a reliable marker for monitoring the long-term therapeutic outcomes of enzyme replacement therapy for late-onset Fabry patients with the Chinese hotspot mutation (IVS4+919G>A). Orphanet Journal of Rare Diseases, 2014, 9, 111.	2.7	33
84	Anesthetic management of comprehensive dental restoration in a child with glutaric aciduria type 1 using volatile sevoflurane. Journal of the Chinese Medical Association, 2014, 77, 548-551.	1.4	3
85	Two Frequent Mutations Associated with the Classic Form of Propionic Acidemia in Taiwan. Biochemical Genetics, 2014, 52, 415-429.	1.7	13
86	High-throughput detection of common sequence variations of Fabry disease in Taiwan using DNA mass spectrometry. Molecular Genetics and Metabolism, 2014, 111, 507-512.	1.1	14
87	Assessment of hearing loss by pure-tone audiometry in patients with mucopolysaccharidoses. Molecular Genetics and Metabolism, 2014, 111, 533-538.	1.1	36
88	Clinical observations on enzyme replacement therapy in patients with Fabry disease and the switch from agalsidase beta to agalsidase alfa. Journal of the Chinese Medical Association, 2014, 77, 190-197.	1.4	14
89	Plasma globotriaosylsphingosine (lysoGb3) could be a biomarker for Fabry disease with a Chinese hotspot late-onset mutation (IVS4+919G>A). Clinica Chimica Acta, 2013, 426, 114-120.	1.1	33
90	Effects of enzyme replacement therapy for cardiac-type Fabry patients with a Chinese hotspot late-onset Fabry mutation (IVS4+919G>A). BMJ Open, 2013, 3, e003146.	1.9	16

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91	Mutation Profile of the MUT Gene in Chinese Methylmalonic Aciduria Patients. JIMD Reports, 2012, 6, 55-64.	1.5	22
92	Homocystinuria in Taiwan: An inordinately high prevalence in an Austronesian aboriginal tribe, Tao. Molecular Genetics and Metabolism, 2012, 105, 590-595.	1.1	9
93	The use of high resolution melting analysis to detect Fabry mutations in heterozygous females via dry bloodspots. Clinica Chimica Acta, 2012, 413, 422-427.	1.1	21
94	Recessive congenital methemoglobinemia caused by a rare mechanism: Maternal uniparental heterodisomy with segmental isodisomy of a chromosome 22. Blood Cells, Molecules, and Diseases, 2012, 49, 114-117.	1.4	7
95	Activation of silenced tumor suppressor genes in prostate cancer cells by a novel energy restrictionâ€mimetic agent. Prostate, 2012, 72, 1767-1778.	2.3	18
96	Mutation spectrum of and founder effects affecting the PTS gene in East Asian populations. Journal of Human Genetics, 2012, 57, 145-152.	2.3	34
97	Idiopathic Calcinosis Cutis in a Child: Chemical Composition of the Calcified Deposits. Dermatology, 2011, 222, 201-205.	2.1	11
98	Disorders of BH4 metabolism and the treatment of patients with 6-pyruvoyl-tetrahydropterin synthase deficiency in Taiwan. Brain and Development, 2011, 33, 847-855.	1.1	19
99	Clinical observations, molecular genetic analysis, and treatment of sitosterolemia in infants and children. Journal of Inherited Metabolic Disease, 2010, 33, 437-443.	3.6	57
100	Nationwide survey of extended newborn screening by tandem mass spectrometry in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 295-305.	3.6	128
101	Enzyme assay and clinical assessment in subjects with a Chinese hotspot lateâ€onset Fabry mutation (IVS4â€‰+â€‰919Gâ†’A). Journal of Inherited Metabolic Disease, 2010, 33, 619-624.	3.6	24
102	Domino liver graft from a patient with homozygous familial hypercholesterolemia. Pediatric Transplantation, 2010, 14, E30-E33.	1.0	25
103	Novel human pathological mutations. Gene symbol: GLA. Disease: Fabry disease. Human Genetics, 2010, 127, 122.	3.8	2
104	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 466.	3.8	0
105	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 465.	3.8	0
106	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 465.	3.8	0
107	Six New Mutations of the Thyroglobulin Gene Discovered in Taiwanese Children Presenting with Thyroid Dyshormonogenesis. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 5045-5052.	3.6	28
108	High Incidence of the Cardiac Variant of Fabry Disease Revealed by Newborn Screening in the Taiwan Chinese Population. Circulation: Cardiovascular Genetics, 2009, 2, 450-456.	5.1	214

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109	Growth Hormone Therapy in Neonatal Patients With Methylmalonic Acidemia. Journal of the Chinese Medical Association, 2009, 72, 462-467.	1.4	7
110	Long-term Follow-up of Taiwanese Chinese Patients Treated Early for 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency. Archives of Neurology, 2008, 65, 387-92.	4.5	20
111	Lenticular Subluxation in a Patient with Homocystinuria Undetected by Neonatal Screening. Journal of the Chinese Medical Association, 2007, 70, 562-564.	1.4	8
112	Corneal Lesion as the Initial Manifestation of Tyrosinemia Type II. Journal of the Chinese Medical Association, 2006, 69, 286-288.	1.4	13
113	Long-term follow-up of Chinese patients who received delayed treatment for 6-pyruvoyl-tetrahydropterin synthase deficiency. Molecular Genetics and Metabolism, 2006, 87, 128-134.	1.1	15
114	Paternal gonadal mosaicism of NIPBL mutation in a father of siblings with Cornelia de Lange syndrome. Prenatal Diagnosis, 2006, 26, 1054-1057.	2.3	27
115	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, 2006, 43, 817-821.	3.2	41
116	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. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4208-4212.	3.6	42
117	Mosaic or chimera? Revisiting an old hypothesis about the cause of the 46,XX/46,XY hermaphrodite. Journal of Pediatrics, 2002, 140, 732-735.	1.8	34
118	Rapid enlargement of a residual craniopharyngioma during short-term growth hormone replacement. Child's Nervous System, 2002, 18, 164-165.	1.1	22
119	Diagnosis of Congenital Hypothyroidism from Human Anagen Scalp Hair by Infrared Microspectroscopy. Ultrastructural Pathology, 2001, 25, 357-360.	0.9	5
120	Chinese achondroplasia is also defined by recurrent G380R mutations of the fibroblast growth factor receptor-3 gene. Human Genetics, 1996, 98, 65-67.	3.8	8