Ni-Chung Lee

List of Publications by Year
in descending order

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178 papers	4,735 citations	126901 33 h-index	123420 61 g-index
183 all docs	183 docs citations	183 times ranked	5394 citing authors

#	Article	IF	Citations
1	Clinical and molecular features of idiopathic hypogonadotropic hypogonadism in Taiwan: A single center experience. Journal of the Formosan Medical Association, 2022, 121, 218-226.	1.7	8
2	Long-term efficacy and safety of eladocagene exuparvovec in patients with AADC deficiency. Molecular Therapy, 2022, 30, 509-518.	8.2	58
3	CTLA-4 gene mutation and multiple sclerosis: A case report and literature review. Journal of Microbiology, Immunology and Infection, 2022, 55, 545-548.	3.1	3
4	Outcome of Later-Onset Pompe Disease Identified Through Newborn Screening. Journal of Pediatrics, 2022, 244, 139-147.e2.	1.8	10
5	Short stature leads to a diagnosis of Jansenâ€“de Vries syndrome in two unrelated Taiwanese girls: A case report and literature review. Journal of the Formosan Medical Association, 2022, 121, 856-860.	1.7	2
6	Comparison of GATK and DeepVariant by trio sequencing. Scientific Reports, 2022, 12, 1809.	3.3	26
7	A case of senile-onset progressive hemiballism and cognitive decline with diffuse brain iron accumulations. Parkinsonism and Related Disorders, 2022, , .	2.2	2
8	Safety and efficacy of eliglustat combined to enzyme replacement therapy for lymphadenopathy in patients with Gaucher disease type 3. Molecular Genetics and Metabolism Reports, 2022, 31, 100867.	1.1	4
9	Duchenne muscular dystrophy newborn screening: the first 50,000 newborns screened in Taiwan. Neurological Sciences, 2022, 43, 4563-4566.	1.9	13
10	Asymptomatic <scp>ASS1</scp> carriers with high blood citrulline levels. Molecular Genetics & Genomic Medicine, 2022, 10, .	1.2	3
11	Improved diagnosis of citrin deficiency by newborn screening using a molecular second-tier test. Molecular Genetics and Metabolism, 2022, 136, 330-336.	1.1	10
12	Thymidine Kinase 2 Deficiencyâ€“Induced Adult-Onset Ptosis and Proximal Weakness. Neurology: Clinical Practice, 2021, 11, e379-e382.	1.6	0
13	CMAP changes upon symptom onset and during treatment in spinal muscular atrophy patients: lessons learned from newborn screening. Genetics in Medicine, 2021, 23, 415-420.	2.4	13
14	Quantitative examination of early diabetes by light-emitting diodes light-induced pupillary light reflex. Review of Scientific Instruments, 2021, 92, 014101.	1.3	2
15	A systematic review of late-onset and very-late-onset multiple acyl-coenzyme A dehydrogenase deficiency: Cohort analysis and patient report from Taiwan. Neuromuscular Disorders, 2021, 31, 218-225.	0.6	3
16	Eyeâ€ofâ€Tiger Sign in Globus Pallidus: A Novel Radiological Feature of Spinocerebellar Ataxia Type 28. Movement Disorders, 2021, 36, 2200-2202.	3.9	0
17	Rapid Trio Exome Sequencing for Autosomal Recessive Renal Tubular Dysgenesis in Recurrent Oligohydramnios. Frontiers in Genetics, 2021, 12, 606970.	2.3	1
18	Impact of genetic tests on survivors of paediatric sudden cardiac arrest. Archives of Disease in Childhood, 2021, , archdischild-2020-321532.	1.9	0

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19	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation: A Novel DARS2 Mutation and Intraâ€Familial Heterogeneity. Movement Disorders Clinical Practice, 2021, 8, 1116-1122.	1.5	2
20	Lactate peak in muscle disclosed by magnetic resonance spectroscopy in a patient with CPEO-plus syndrome. ENeurologicalSci, 2021, 24, 100360.	1.3	1
21	A pilot study shows the positive effects of continuous airway pressure for treating hypernasal speech in children with infantile-onset Pompe disease. Scientific Reports, 2021, 11, 18826.	3.3	1
22	Suboptimal trophectoderm mitochondrial DNA level is associated with delayed blastocyst development. Journal of Assisted Reproduction and Genetics, 2021, 38, 587-594.	2.5	11
23	RNA-seq of peripheral blood mononuclear cells of congenital generalized lipodystrophy type 2 patients. Scientific Data, 2021, 8, 265.	5.3	3
24	NAXE gene mutation-related progressive encephalopathy. Medicine (United States), 2021, 100, e27548.	1.0	5
25	A novel deep intronic variant strongly associates with Alkaptonuria. Npj Genomic Medicine, 2021, 6, 89.	3.8	9
26	Improved Motor Function in Children with AADC Deficiency Treated with Eladocagene Exuparvovec (PTC-AADC): Interim Findings from a Phase 2 Trial. Neuropediatrics, 2021, 52, .	0.6	0
27	PAX2 Mutation-Related Renal Hypodysplasia: Review of the Literature and Three Case Reports. Frontiers in Pediatrics, 2021, 9, 765929.	1.9	4
28	Novel Compound Heterozygous Variants in TBCD Gene Associated with Infantile Neurodegenerative Encephalopathy. Children, 2021, 8, 1140.	1.5	3
29	Episodic weakness and axonal sensorimotor neuropathy caused by a mitochondrial MT-ATP6 mutation. Journal of the Formosan Medical Association, 2021, , .	1.7	0
30	Clinical and electrophysiological characteristics of a type 1 sialidosis patient with a novel deletion mutation in NEU1 gene. Journal of the Formosan Medical Association, 2020, 119, 406-412.	1.7	9
31	Thyroid disorders in Taiwanese children with Down syndrome: The experience of a single medical center. Journal of the Formosan Medical Association, 2020, 119, 345-349.	1.7	3
32	REM sleep and sleep apnea are associated with language function in Down syndrome children: An analysis of a community sample. Journal of the Formosan Medical Association, 2020, 119, 516-523.	1.7	14
33	Cardiac manifestations and gene mutations of patients with RASopathies in Taiwan. American Journal of Medical Genetics, Part A, 2020, 182, 357-364.	1.2	8
34	Composite Scores of Plasma Tau and Î²-Amyloids Correlate with Dementia in Down Syndrome. ACS Chemical Neuroscience, 2020, 11, 191-196.	3.5	4
35	Autosomal dominant cerebellar ataxia, deafness, and narcolepsy with amenorrhea, subclinical optic atrophy, and electroencephalographic abnormality: A case report. ENeurologicalSci, 2020, 21, 100271.	1.3	0
36	Ultrastructural and diffusion tensor imaging studies reveal axon abnormalities in Pompe disease mice. Scientific Reports, 2020, 10, 20239.	3.3	1

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37	Mitochondrial UQCRC1 mutations cause autosomal dominant parkinsonism with polyneuropathy. Brain, 2020, 143, 3352-3373.	7.6	37
38	Towards a reference genome that captures global genetic diversity. Nature Communications, 2020, 11, 5482.	12.8	34
39	Diversity in heritable disorders of connective tissue at a single center. Connective Tissue Research, 2020, 62, 1-6.	2.3	5
40	Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes. Molecular Genetics & Genomic Medicine, 2020, 8, e1455.	1.2	14
41	Dietary intake and nutritional status of patients with phenylketonuria in Taiwan. Scientific Reports, 2020, 10, 14537.	3.3	5
42	Lessons for the clinical nephrologist: dietary management of adult-onset type II citrullinemia in chronic kidney disease: a nutritional dilemma. Journal of Nephrology, 2020, 33, 1111-1113.	2.0	0
43	Earlier and higher dosing of alglucosidase alfa improve outcomes in patients with infantile-onset Pompe disease: Evidence from real-world experiences. Molecular Genetics and Metabolism Reports, 2020, 23, 100591.	1.1	23
44	Novel Phenotype of 6p25 Deletion Syndrome Presenting Juvenile Parkinsonism and Brain Calcification. Movement Disorders, 2020, 35, 1457-1462.	3.9	9
45	UNC13D mutation presenting as fulminant familial hemophagocytic lymphohistiocytosis. Journal of Microbiology, Immunology and Infection, 2020, 53, 1039-1041.	3.1	1
46	Clinical, radiological, and genetic characteristics in patients with Huntington's disease in a Taiwanese cohort. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 352-359.	1.7	1
47	De novo mutation and skewed Xâ€inactivation in girl with BCAP31 â€related syndrome. Human Mutation, 2020, 41, 1775-1782.	2.5	3
48	Newborn screening for Morquio disease and other lysosomal storage diseases: results from the 8-plex assay for 70,000 newborns. Orphanet Journal of Rare Diseases, 2020, 15, 38.	2.7	40
49	The Timely Needs for Infantile Onset Pompe Disease Newborn Screeningâ€"Practice in Taiwan. International Journal of Neonatal Screening, 2020, 6, 30.	3.2	1
50	SUN-033 A Rare Case: Bone Pain and Continued Linear Growth in a Young Adult Male Due to Aromatase Deficiency. Journal of the Endocrine Society, 2020, 4, .	0.2	0
51	Vohwinkel syndrome associated with a p.Gly59Arg missense mutation in GJB2. Dermatologica Sinica, 2020, 38, 228.	0.5	0
52	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
53	Heterogeneous nonataxic phenotypes of spinocerebellar ataxia in a Taiwanese population. Brain and Behavior, 2019, 9, e01414.	2.2	10
54	Genotypic and phenotypic correlations of biotinidase deficiency in the Chinese population. Orphanet Journal of Rare Diseases, 2019, 14, 6.	2.7	15

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55	Clinical features of Pompe disease with motor neuronopathy. Neuromuscular Disorders, 2019, 29, 903-906.	0.6	7
56	Fabry disease cardiac variant IVS4+919 G>A is associated with multiple cardiac gene variants in patients with severe cardiomyopathy and fatal arrhythmia. Genetics in Medicine, 2019, 21, 1890-1891.	2.4	3
57	Reply to Lutz-Bonengel et al.: Biparental mtDNA transmission is unlikely to be the result of nuclear mitochondrial DNA segments. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1823-1824.	7.1	15
58	Mosaic paternal haploidy in a patient with pancreatoblastoma and Beckwithâ€“Wiedemann spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 1878-1883.	1.2	6
59	Gene therapy improves brain white matter in aromatic lâ€amino acid decarboxylase deficiency. Annals of Neurology, 2019, 85, 644-652.	5.3	30
60	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
61	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and ureaâ€cycle disorders: On the basis of information from a European multicenter registry. Journal of Inherited Metabolic Disease, 2019, 42, 1162-1175.	3.6	30
62	Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment. Molecular Genetics and Metabolism, 2019, 126, 397-405.	1.1	26
63	Critical Trio Exome Benefits In-Time Decision-Making for Pediatric Patients With Severe Illnesses*. Pediatric Critical Care Medicine, 2019, 20, 1021-1026.	0.5	29
64	Next-generation sequencing identifies TRPV4-related skeletal dysplasia in a boy with progressive bowlegs. Pediatrics and Neonatology, 2019, 60, 102-104.	0.9	3
65	A review of aromatic <scp>l</scp>â€amino acid decarboxylase (AADC) deficiency in Taiwan. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 226-229.	1.6	12
66	Electrical Abnormalities in Dopaminergic Neurons of the Substantia Nigra in Mice With an Aromatic L-Amino Acid Decarboxylase Deficiency. Frontiers in Cellular Neuroscience, 2019, 13, 9.	3.7	3
67	Mycobacterium abscessus infection in a boy with X-linked anhidrotic ectodermal dysplasia, immunodeficiency. Journal of Microbiology, Immunology and Infection, 2019, 52, 504-506.	3.1	4
68	Congenital generalized lipodystrophy in Taiwan. Journal of the Formosan Medical Association, 2019, 118, 142-147.	1.7	11
69	Newborn screening: Taiwanese experience. Annals of Translational Medicine, 2019, 7, 281-281.	1.7	23
70	AGIL-AADC Gene Therapy Results in Sustained Improvements in Motor and Developmental Milestones over 5 Years in Children with AADC Deficiency. Neuropediatrics, 2019, 50, .	0.6	0
71	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
72	SHOX deficiency in short Taiwanese children: A single-center experience. Journal of the Formosan Medical Association, 2018, 117, 909-914.	1.7	6

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73	Functional independence of Taiwanese children with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1309-1314.	1.2	2
74	A Neuron-Specific Gene Therapy Relieves Motor Deficits in Pompe Disease Mice. Molecular Neurobiology, 2018, 55, 5299-5309.	4.0	28
75	Performance of the Four-Plex Tandem Mass Spectrometry Lysosomal Storage Disease Newborn Screening Test: The Necessity of Adding a 2nd Tier Test for Pompe Disease. International Journal of Neonatal Screening, 2018, 4, 41.	3.2	17
76	Biparental Inheritance of Mitochondrial DNA in Humans. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 13039-13044.	7.1	349
77	Disease progression in a preâ€symptomatically treated patient with juvenileâ€onset Pompe disease â€“ need for an earlier treatment?. European Journal of Neurology, 2018, 25, e111.	3.3	5
78	Exome sequencing for paediatric-onset diseases: impact of the extensive involvement of medical geneticists in the diagnostic odyssey. Npj Genomic Medicine, 2018, 3, 19.	3.8	11
79	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
80	Russellâ€“Silver syndrome presenting with ambiguous genitalia. Journal of the Formosan Medical Association, 2017, 116, 645-646.	1.7	2
81	Longitudinal follow-up to evaluate speech disorders in early-treated patients with infantile-onset Pompe disease. European Journal of Paediatric Neurology, 2017, 21, 485-493.	1.6	14
82	A limb-girdle myopathy phenotype of RUNX2 mutation in a patient with cleidocranial dysplasia: a case study and literature review. BMC Neurology, 2017, 17, 2.	1.8	7
83	Gene therapy with modified U1 small nuclear RNA. Expert Review of Endocrinology and Metabolism, 2017, 12, 171-175.	2.4	5
84	Albuterol as an adjunctive treatment to enzyme replacement therapy in infantile-onset Pompe disease. Molecular Genetics and Metabolism Reports, 2017, 11, 31-35.	1.1	10
85	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	1.2	75
86	Natural History of Aromatic l-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Reports, 2017, 40, 1-6.	1.5	26
87	Efficacy and safety of AAV2 gene therapy in children with aromatic L-amino acid decarboxylase deficiency: an open-label, phase 1/2 trial. The Lancet Child and Adolescent Health, 2017, 1, 265-273.	5.6	96
88	Reply to: â€œPeculiarities of progressive external ophthalmoplegia due to single mtDNA deletionsâ€• Journal of the Formosan Medical Association, 2017, 116, 821-822.	1.7	0
89	Presymptomatic Diagnosis of Spinal Muscular Atrophy Through Newborn Screening. Journal of Pediatrics, 2017, 190, 124-129.e1.	1.8	113
90	A Review of Biomarkers for Alzheimerâ€™s Disease in Down Syndrome. Neurology and Therapy, 2017, 6, 69-81.	3.2	19

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91	Newborn Screening for Severe Combined Immunodeficiency in Taiwan. International Journal of Neonatal Screening, 2017, 3, 16.	3.2	38
92	Slow, progressive myopathy in neonatally treated patients with infantile-onset Pompe disease: a muscle magnetic resonance imaging study. Orphanet Journal of Rare Diseases, 2016, 11, 63.	2.7	23
93	3-O-methyldopa levels in newborns: Result of newborn screening for aromatic l-amino-acid decarboxylase deficiency. Molecular Genetics and Metabolism, 2016, 118, 259-263.	1.1	52
94	Mutation-adapted U1 snRNA corrects a splicing error of the dopa decarboxylase gene. Human Molecular Genetics, 2016, 25, ddw323.	2.9	18
95	Integrated care for Down syndrome. Congenital Anomalies (discontinued), 2016, 56, 104-106.	0.6	2
96	Histopathological and genetic analysis of extraocular muscle in chronic progressive external ophthalmoplegia. Journal of the Formosan Medical Association, 2016, 115, 1012-1014.	1.7	4
97	Tet oncogene family member 2 gene alterations in childhood acute myeloid leukemia. Journal of the Formosan Medical Association, 2016, 115, 801-806.	1.7	8
98	Advances in newborn screening for Pompe disease and resulting clinical outcomes. Expert Opinion on Orphan Drugs, 2016, 4, 21-29.	0.8	0
99	Measuring propionyl-CoA carboxylase activity in phytohemagglutinin stimulated lymphocytes using high performance liquid chromatography. Clinica Chimica Acta, 2016, 453, 13-20.	1.1	5
100	Long-term outcome for Down syndrome patients with hematopoietic disorders. Journal of the Formosan Medical Association, 2016, 115, 94-99.	1.7	5
101	Blood Beta-Amyloid and Tau in Down Syndrome: A Comparison with Alzheimerâ€™s Disease. Frontiers in Aging Neuroscience, 2016, 8, 316.	3.4	44
102	Moyamoya disease in two patients with Noonanâ€like syndrome with loose anagen hair. American Journal of Medical Genetics, Part A, 2015, 167, 1285-1288.	1.2	19
103	Congenital Malformations in Newbornsâ€”A Challenge Unmet for Decades. Pediatrics and Neonatology, 2015, 56, 5-6.	0.9	0
104	Long-Term Prognosis of Patients with Infantile-Onset Pompe Disease Diagnosed by Newborn Screening and Treated since Birth. Journal of Pediatrics, 2015, 166, 985-991.e2.	1.8	113
105	Benefits of Neuronal Preferential Systemic Gene Therapy for Neurotransmitter Deficiency. Molecular Therapy, 2015, 23, 1572-1581.	8.2	25
106	Mortality, disability, and intensive care in patients with mitochondrial 3243A>G mutation. Intensive Care Medicine, 2015, 41, 1493-1495.	8.2	1
107	Baseline Urinary Glucose Tetrasaccharide Concentrations in Patients with Infantile- and Late-Onset Pompe Disease Identified by Newborn Screening. JIMD Reports, 2014, 19, 67-73.	1.5	29
108	Treatment of Congenital Neurotransmitter Deficiencies by Intracerebral Ventricular Injection of an Adeno-Associated Virus Serotype 9 Vector. Human Gene Therapy, 2014, 25, 189-198.	2.7	19

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109	Prominent vacuolation of the eyelid levator muscle in an earlyâ€treated child with infantileâ€onset Pompe disease. Muscle and Nerve, 2014, 50, 301-302.	2.2	7
110	Cyclic Pamidronate Infusion for Neonatal-onset Osteogenesis Imperfecta. Pediatrics and Neonatology, 2014, 55, 306-311.	0.9	11
111	Efficacy and safety of intermittent hemodialysis in infants and young children with inborn errors of metabolism. Pediatric Nephrology, 2014, 29, 111-116.	1.7	16
112	Diagnosis of aromatic l-amino acid decarboxylase deficiency by measuring 3-O-methyldopa concentrations in dried blood spots. Clinica Chimica Acta, 2014, 431, 19-22.	1.1	29
113	Outcome of early-treated type III Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2014, 53, 105-109.	1.4	22
114	AADC Deficiency. , 2014, , 3-4.		0
115	Promising outcomes in glutaric aciduria type I patients detected by newborn screening. Metabolic Brain Disease, 2013, 28, 61-67.	2.9	29
116	Newborn screening for citrin deficiency and carnitine uptake defect using second-tier molecular tests. BMC Medical Genetics, 2013, 14, 24.	2.1	22
117	Regulation of the dopaminergic system in a murine model of aromatic l-amino acid decarboxylase deficiency. Neurobiology of Disease, 2013, 52, 177-190.	4.4	37
118	Pompe Disease: Early Diagnosis and Early Treatment Make a Difference. Pediatrics and Neonatology, 2013, 54, 219-227.	0.9	135
119	AADC Deficiency. Advances in Pharmacology, 2013, 68, 273-284.	2.0	10
120	Carnitine Uptake Defect (Primary Carnitine Deficiency): Risk in Genotype-Phenotype Correlation. Human Mutation, 2013, 34, 655-655.	2.5	13
121	Longâ€term efficacy of miglustat in paediatric patients with Niemannâ€Pick disease type C. Journal of Inherited Metabolic Disease, 2013, 36, 129-137.	3.6	34
122	Serial cytokine expressions in infants with incontinentia pigmenti. Immunobiology, 2013, 218, 772-779.	1.9	11
123	Lung toxicity of hydroxypropyl-Î²-cyclodextrin infusion. Molecular Genetics and Metabolism, 2013, 109, 231-232.	1.1	27
124	Clinical application of massively parallel sequencing in the molecular diagnosis of glycogen storage diseases of genetically heterogeneous origin. Genetics in Medicine, 2013, 15, 106-114.	2.4	65
125	Mitochondrial Depletion Causes Neonatal-Onset Leigh Syndrome, Myopathy, and Renal Tubulopathy. Journal of Child Neurology, 2013, 28, 404-408.	1.4	10
126	Fatty Acid Oxidation Disorders in a Chinese Population in Taiwan. JIMD Reports, 2013, 11, 165-172.	1.5	26

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127	Association of the Congenital Neuromuscular Form of Glycogen Storage Disease Type IV With a Large Deletion and Recurrent Frameshift Mutation. Journal of Child Neurology, 2012, 27, 204-208.	1.4	15
128	Diagnosis of Neonatal Intrahepatic Cholestasis Caused by Citrin Deficiency Using High-Resolution Melting Analysis and a Clinical Scoring System. Journal of Pediatrics, 2012, 161, 626-631.e2.	1.8	14
129	Late onset of symptoms in an atypical patient with the cblJ inborn error of vitamin B12 metabolism: Diagnosis and novel mutation revealed by exome sequencing. Molecular Genetics and Metabolism, 2012, 107, 664-668.	1.1	33
130	Early Pathologic Changes and Responses to Treatment in Patients With Later-Onset Pompe Disease. Pediatric Neurology, 2012, 46, 168-171.	2.1	21
131	Algorithm for Pompe disease newborn screening: Results from the Taiwan screening program. Molecular Genetics and Metabolism, 2012, 106, 281-286.	1.1	72
132	An acidic oligopeptide displayed on AAV2 improves axial muscle tropism after systemic delivery. Genetic Vaccines and Therapy, 2012, 10, 3.	1.5	4
133	A Novel 3670-Base Pair Mitochondrial DNA Deletion Resulting in Multi-systemic Manifestations in a Child. Pediatrics and Neonatology, 2012, 53, 264-268.	0.9	15
134	Application of Mass Spectrometry in Newborn Screening: About Both Small Molecular Diseases and Lysosomal Storage Diseases. Topics in Current Chemistry, 2012, 336, 177-196.	4.0	5
135	Fabry Disease: Incidence of the Common Later-Onset Î±-Galactosidase A IVS4+919Gâ†'A Mutation in Taiwanese Newbornsâ€"Superiority of DNA-Based to Enzyme-Based Newborn Screening for Common Mutations. Molecular Medicine, 2012, 18, 780-784.	4.4	71
136	Gene Therapy for Aromatic <scp>l</scp> -Amino Acid Decarboxylase Deficiency. Science Translational Medicine, 2012, 4, 134ra61.	12.4	195
137	Novel heterozygous tissue-nonspecific alkaline phosphatase (TNAP) gene mutations causing lethal perinatal hypophosphatasia. Journal of Bone and Mineral Metabolism, 2012, 30, 109-113.	2.7	13
138	Unusual Spinal Cord Lesions in Late-Onset Non-ketotic Hyperglycinemia. Journal of Child Neurology, 2011, 26, 900-903.	1.4	12
139	Left Ventricular Geometry, Global Function, and Dyssynchrony in Infants and Children With Pompe Cardiomyopathy Undergoing Enzyme Replacement Therapy. Journal of Cardiac Failure, 2011, 17, 930-936.	1.7	25
140	Elevation of urinary globotriaosylceramide (GL3) in infants with Fabry disease. Molecular Genetics and Metabolism, 2011, 102, 57-60.	1.1	9
141	Rapid progressive course of later-onset Pompe disease in Chinese patients. Molecular Genetics and Metabolism, 2011, 104, 284-288.	1.1	39
142	Congenital Hypopituitarism due to POU1F1 Gene Mutation. Journal of the Formosan Medical Association, 2011, 110, 58-61.	1.7	10
143	Later-Onset Pompe Disease: Early Detection and Early Treatment Initiation Enabled by Newborn Screening. Journal of Pediatrics, 2011, 158, 1023-1027.e1.	1.8	88
144	Mitochondrial DNA polymerase Â mutations: an ever expanding molecular and clinical spectrum. Journal of Medical Genetics, 2011, 48, 669-681.	3.2	140

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145	Newborn screening for neuropathic lysosomal storage disorders. Journal of Inherited Metabolic Disease, 2010, 33, 381-386.	3.6	20
146	Complex rearrangements between chromosomes 6, 10, and 11 with multiple deletions at breakpoints. American Journal of Medical Genetics, Part A, 2010, 152A, 2327-2334.	1.2	14
147	Stabilization of blood methylmalonic acid level in methylmalonic acidemia after liver transplantation. Pediatric Transplantation, 2010, 14, 337-341.	1.0	35
148	Enzyme replacement therapy for mucopolysaccharidosis VIâ€"experience in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 421-427.	3.6	59
149	CCL18 as an alternative marker in Gaucher and Niemann-Pick disease with chitotriosidase deficiency. Blood Cells, Molecules, and Diseases, 2010, 44, 38-40.	1.4	15
150	Genetic heterozygosity and pseudodeficiency in the Pompe disease newborn screening pilot program. Molecular Genetics and Metabolism, 2010, 99, 379-383.	1.1	91
151	Diagnoses of newborns and mothers with carnitine uptake defects through newborn screening. Molecular Genetics and Metabolism, 2010, 100, 46-50.	1.1	86
152	Reduction in imiglucerase dosage causes immediate rise of chitotriosidase activity in patients with Gaucher disease. Molecular Genetics and Metabolism, 2010, 101, 90-91.	1.1	2
153	Newborn Screening for Methylmalonic Aciduria by Tandem Mass Spectrometry: 7 Years' Experience From Two Centers in Taiwan. Journal of the Chinese Medical Association, 2010, 73, 314-318.	1.4	15
154	FOXL2 mutations in Taiwanese patients with blepharophimosis, ptosis, epicanthus inversus syndrome. Clinical Chemistry and Laboratory Medicine, 2010, 48, 485-8.	2.3	6
155	Reversal of Cardiac Dysfunction after Enzyme Replacement in Patients with Infantile-Onset Pompe Disease. Journal of Pediatrics, 2009, 155, 271-275.e2.	1.8	56
156	Caloric restriction in AlstrÃ¶m syndrome prevents hyperinsulinemia. American Journal of Medical Genetics, Part A, 2009, 149A, 666-668.	1.2	16
157	Somatic and germâ€line mosaicism in Rubinsteinâ€"Taybi syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1463-1467.	1.2	40
158	Newborn screening for Fabry disease in Taiwan reveals a high incidence of the later-onsetGLAmutation c.936+919G>A (IVS4+919G>A). Human Mutation, 2009, 30, 1397-1405.	2.5	299
159	Identification of CpG methylation of the SNRPN gene by methylationâ€specific multiplex PCR. Electrophoresis, 2009, 30, 410-416.	2.4	6
160	Cryptic subtelomeric deletion plus inverted duplication at chromosome 18q in a fetus: molecular delineation by multicolor banding. Prenatal Diagnosis, 2009, 29, 1058-1060.	2.3	6
161	Pseudogeneâ€derived IKBKG gene mutations in incontinentia pigmenti. Clinical Genetics, 2009, 76, 417-419.	2.0	6
162	Glycogen Storage Disease Type Ib: The First Case in Taiwan. Pediatrics and Neonatology, 2009, 50, 125-128.	0.9	3

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163	Pompe Disease in Infants: Improving the Prognosis by Newborn Screening and Early Treatment. Pediatrics, 2009, 124, e1116-e1125.	2.1	185
164	Acute metabolic decompensation and sudden death in Barth syndrome: report of a family and a literature review. European Journal of Pediatrics, 2008, 167, 941-944.	2.7	47
165	Mutation of Mitochondrial DNA G13513A Presenting with Leigh Syndrome, Wolff-Parkinson-White Syndrome and Cardiomyopathy. Pediatrics and Neonatology, 2008, 49, 145-149.	0.9	52
166	Brain Damage by Mild Metabolic Derangements in Methylmalonic Acidemia. Pediatric Neurology, 2008, 39, 325-329.	2.1	24
167	Early Detection of Pompe Disease by Newborn Screening Is Feasible: Results From the Taiwan Screening Program. Pediatrics, 2008, 122, e39-e45.	2.1	207
168	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
169	Chubby Face and the Biochemical Parameters for the Early Diagnosis of Neonatal Intrahepatic Cholestasis Caused by Citrin Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2008, 47, 187-192.	1.8	19
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