Yin-Hsiu Chien

List of Publications by Year
in descending order

261 papers	7,623 citations	61984 43 h-index	71685 76 g-index
281 all docs	281 docs citations	281 times ranked	8114 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	Advanced therapeutic strategy for hereditary neuromuscular diseases. Molecular Therapy, 2022, 30, 12-13.	8.2	1
6	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
7	Comparison of GATK and DeepVariant by trio sequencing. Scientific Reports, 2022, 12, 1809.	3.3	26
8	Hepatic Steatosis Assessment as a New Strategy for the Metabolic and Nutritional Management of Duchenne Muscular Dystrophy. Nutrients, 2022, 14, 727.	4.1	4
9	OP016: Mini-COMET: Safety and efficacy of â‰¥97 weeksâ€™ avalglucosidase alfa in infantile-onset Pompe disease participants previously treated with alglucosidase alfa. Genetics in Medicine, 2022, 24, S348-S349.	2.4	4
10	eP146: Application of exome sequencing in patients of congenital anomalies with or without intellectual disability. Genetics in Medicine, 2022, 24, S90.	2.4	2
11	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
12	Duchenne muscular dystrophy newborn screening: the first 50,000 newborns screened in Taiwan. Neurological Sciences, 2022, 43, 4563-4566.	1.9	13
13	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
14	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
15	Nusinersen in spinal muscular atrophy type 1 from neonates to young adult: 1-year data from three Asia-Pacific regions. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1244-1246.	1.9	12
16	STIG study: real-world data of long-term outcomes of adults with Pompe disease under enzyme replacement therapy with alglucosidase alfa. Journal of Neurology, 2021, 268, 2482-2492.	3.6	21
17	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
18	Mini-COMET: effects of avalglucosidase alfa on ptosis in participants with infantile-onset Pompe disease previously treated with alglucosidase alfa. Molecular Genetics and Metabolism, 2021, 132, S137.	1.1	0

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19	Rapid Trio Exome Sequencing for Autosomal Recessive Renal Tubular Dysgenesis in Recurrent Oligohydramnios. Frontiers in Genetics, 2021, 12, 606970.	2.3	1
20	Molecular basis of mucopolysaccharidosis IVA (Morquio A syndrome): A review and classification of GALNS gene variants and reporting of 68 novel variants. Human Mutation, 2021, 42, 1384-1398.	2.5	14
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	RNA-seq of peripheral blood mononuclear cells of congenital generalized lipodystrophy type 2 patients. Scientific Data, 2021, 8, 265.	5.3	3
23	A novel deep intronic variant strongly associates with Alkaptonuria. Npj Genomic Medicine, 2021, 6, 89.	3.8	9
24	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
25	Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. Lancet Neurology, The, 2021, 20, 1027-1037.	10.2	42
26	Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. Lancet Neurology, The, 2021, 20, 1012-1026.	10.2	59
27	Novel Compound Heterozygous Variants in TBCD Gene Associated with Infantile Neurodegenerative Encephalopathy. Children, 2021, 8, 1140.	1.5	3
28	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
29	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
30	Early initiation of high-dose oral ambroxol in combination with enzyme replacement therapy in a neuropathic Gaucher infant. Blood Cells, Molecules, and Diseases, 2020, 81, 102402.	1.4	7
31	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
32	Composite Scores of Plasma Tau and Î²-Amyloids Correlate with Dementia in Down Syndrome. ACS Chemical Neuroscience, 2020, 11, 191-196.	3.5	4
33	Ultrastructural and diffusion tensor imaging studies reveal axon abnormalities in Pompe disease mice. Scientific Reports, 2020, 10, 20239.	3.3	1
34	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€“2019). Orphanet Journal of Rare Diseases, 2020, 15, 314.	2.7	16
35	Diversity in heritable disorders of connective tissue at a single center. Connective Tissue Research, 2020, 62, 1-6.	2.3	5
36	Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes. Molecular Genetics & Genomic Medicine, 2020, 8, e1455.	1.2	14

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37	Dietary intake and nutritional status of patients with phenylketonuria in Taiwan. Scientific Reports, 2020, 10, 14537.	3.3	5
38	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
39	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
40	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
41	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
42	Development of Newborn Screening for Pompe Disease. International Journal of Neonatal Screening, 2020, 6, 5.	3.2	2
43	The Timely Needs for Infantile Onset Pompe Disease Newborn Screeningâ€"Practice in Taiwan. International Journal of Neonatal Screening, 2020, 6, 30.	3.2	1
44	Turner syndrome and cardiovascular anomalies: Care for girls and women. Pediatrics and Neonatology, 2020, 61, 129-130.	0.9	0
45	Monitoring of liver stiffness by transient elastography during the treatment of Gaucher disease. Pediatrics and Neonatology, 2019, 60, 221-223.	0.9	2
46	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
47	GAA variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. Human Mutation, 2019, 40, 2146-2164.	2.5	51
48	Heterogeneous nonataxic phenotypes of spinocerebellar ataxia in a Taiwanese population. Brain and Behavior, 2019, 9, e01414.	2.2	10
49	Genotypic and phenotypic correlations of biotinidase deficiency in the Chinese population. Orphanet Journal of Rare Diseases, 2019, 14, 6.	2.7	15
50	Clinical features of Pompe disease with motor neuronopathy. Neuromuscular Disorders, 2019, 29, 903-906.	0.6	7
51	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
52	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
53	Rare Concurrence of Two Congenital Disorders: Miller-Dieker Syndrome and T-Cell Lymphopenia. Cytogenetic and Genome Research, 2019, 157, 227-230.	1.1	0
54	High incidence of coâ€existing GLA variants and stroke susceptibility. European Journal of Neurology, 2019, 26, e70-e70.	3.3	0

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55	International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria. Molecular Genetics and Metabolism, 2019, 127, 1-11.	1.1	44
56	Gene therapy improves brain white matter in aromatic lâ€amino acid decarboxylase deficiency. Annals of Neurology, 2019, 85, 644-652.	5.3	30
57	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
58	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
59	Front Cover, Volume 40, Issue 11. Human Mutation, 2019, 40, i.	2.5	0
60	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
61	Next-generation sequencing identifies TRPV4-related skeletal dysplasia in a boy with progressive bowlegs. Pediatrics and Neonatology, 2019, 60, 102-104.	0.9	3
62	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
63	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
64	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
65	Congenital generalized lipodystrophy in Taiwan. Journal of the Formosan Medical Association, 2019, 118, 142-147.	1.7	11
66	Newborn screening: Taiwanese experience. Annals of Translational Medicine, 2019, 7, 281-281.	1.7	23
67	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
68	SHOX deficiency in short Taiwanese children: A single-center experience. Journal of the Formosan Medical Association, 2018, 117, 909-914.	1.7	6
69	Functional independence of Taiwanese children with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1309-1314.	1.2	2
70	Clinical characteristics of Taiwanese children with congenital adrenal hyperplasia due to 21-hydroxylase deficiency detected by neonatal screening. Journal of the Formosan Medical Association, 2018, 117, 126-131.	1.7	10
71	A Neuron-Specific Gene Therapy Relieves Motor Deficits in Pompe Disease Mice. Molecular Neurobiology, 2018, 55, 5299-5309.	4.0	28
72	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

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73	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
74	Results of Fabry Disease Screening in Male Pre-End Stage Renal Disease Patients with Unknown Etiology Found Through the Platform of a Chronic Kidney Disease Education Program in a Northern Taiwan Medical Center. Kidney and Blood Pressure Research, 2018, 43, 1636-1645.	2.0	23
75	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
76	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
77	Russellâ€“Silver syndrome presenting with ambiguous genitalia. Journal of the Formosan Medical Association, 2017, 116, 645-646.	1.7	2
78	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
79	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
80	Risk assessments in infants suspect having later-onset Pompe disease identified through newborn screening. Molecular Genetics and Metabolism, 2017, 120, S36.	1.1	0
81	Genetic epidemiological study doesn't support GLA IVS4 + 919G > A variant is a significant mutation in Fabry disease. Molecular Genetics and Metabolism, 2017, 121, 22-27.	1.1	9
82	Natural History of Aromatic l-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Reports, 2017, 40, 1-6.	1.5	26
83	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
84	Presymptomatic Diagnosis of Spinal Muscular Atrophy Through Newborn Screening. Journal of Pediatrics, 2017, 190, 124-129.e1.	1.8	113
85	A Review of Biomarkers for Alzheimerâ€™s Disease in Down Syndrome. Neurology and Therapy, 2017, 6, 69-81.	3.2	19
86	Consensus recommendations for the diagnosis, treatment and followâ€up of inherited methylation disorders. Journal of Inherited Metabolic Disease, 2017, 40, 5-20.	3.6	47
87	Gestational age, not transient hyperthyrotropinemia impacts brain white matter diffusion tensor imaging in premature infants. Experimental and Therapeutic Medicine, 2017, 15, 1013-1020.	1.8	1
88	Newborn Screening for Severe Combined Immunodeficiency in Taiwan. International Journal of Neonatal Screening, 2017, 3, 16.	3.2	38
89	Family History of Early Infant Death Correlates with Earlier Age at Diagnosis But Not Shorter Time to Diagnosis for Severe Combined Immunodeficiency. Frontiers in Immunology, 2017, 8, 808.	4.8	34
90	AB104. Evaluation of a new non-derivatized MS/MS kit in newborn screening program. Annals of Translational Medicine, 2017, 5, AB104-AB104.	1.7	1

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91	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
92	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
93	372. U1 snRNA-Mediated Correction of a Splicing Error of the Dopa Decarboxylase Gene. Molecular Therapy, 2016, 24, S149.	8.2	0
94	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
95	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
96	Bioevaluation of sixteen ADMDP stereoisomers toward alpha-galactosidase A: Development of a new pharmacological chaperone for the treatment of Fabry disease and potential enhancement of enzyme replacement therapy efficiency. European Journal of Medicinal Chemistry, 2016, 123, 14-20.	5.5	15
97	Integrated care for Down syndrome. Congenital Anomalies (discontinued), 2016, 56, 104-106.	0.6	2
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	201. Neuron-Specific Systemic Gene Therapy for Aromatic L-Amino Acid Decarboxylase (AADC) Deficiency. Molecular Therapy, 2015, 23, S80.	8.2	0
103	C-11. An Update on Gene Therapy for the Treatment of Aromatic L-Amino Acid Decarboxylase (AADC) Deficiency. Molecular Therapy, 2015, 23, S103.	8.2	0
104	Muddâ€™s disease (MAT I/III deficiency): a survey of data for MAT1A homozygotes and compound heterozygotes. Orphanet Journal of Rare Diseases, 2015, 10, 99.	2.7	39
105	Survival and Developmental Milestones Among Pompe Registry Patients with Classic Infantile-Onset Pompe Disease with Different Timing of Initiation of Treatment with Enzyme Replacement Therapy. Journal of Neuromuscular Diseases, 2015, 2, S61-S62.	2.6	5
106	The Pompe Registry: 10 Years of Data. Journal of Neuromuscular Diseases, 2015, 2, S22-S23.	2.6	2
107	Congenital Malformations in Newbornsâ€”A Challenge Unmet for Decades. Pediatrics and Neonatology, 2015, 56, 5-6.	0.9	0
108	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

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109	The Pompe Registry: 10years of data. Molecular Genetics and Metabolism, 2015, 114, S65.	1.1	0
110	Benefits of Neuronal Preferential Systemic Gene Therapy for Neurotransmitter Deficiency. Molecular Therapy, 2015, 23, 1572-1581.	8.2	25
111	Mortality, disability, and intensive care in patients with mitochondrial 3243A>G mutation. Intensive Care Medicine, 2015, 41, 1493-1495.	8.2	1
112	X-linked hyper-IgM syndrome with CD40LG mutation: Two case reports and literature review in Taiwanese patients. Journal of Microbiology, Immunology and Infection, 2015, 48, 113-118.	3.1	25
113	Incidence of severe combined immunodeficiency through newborn screening in a Chinese population. Journal of the Formosan Medical Association, 2015, 114, 12-16.	1.7	68
114	The Pompe Registry: 10 Years of Data. Journal of Neuromuscular Diseases, 2015, 2, S22-S23.	2.6	1
115	Survival and Developmental Milestones Among Pompe Registry Patients with Classic Infantile-Onset Pompe Disease with Different Timing of Initiation of Treatment with Enzyme Replacement Therapy. Journal of Neuromuscular Diseases, 2015, 2, S61-S62.	2.6	2
116	Hypertrophic Cardiomyopathy in Pompe Disease Is Not Limited to the Classic Infantile-Onset Phenotype. JIMD Reports, 2014, 17, 71-75.	1.5	30
117	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
118	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
119	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
120	Parental discussion of G6PD deficiency and child health: implications for clinical practice. Archives of Disease in Childhood, 2014, 99, 251-255.	1.9	1
121	Cyclic Pamidronate Infusion for Neonatal-onset Osteogenesis Imperfecta. Pediatrics and Neonatology, 2014, 55, 306-311.	0.9	11
122	The value of muscle biopsies in Pompe disease: identifying lipofuscin inclusions in juvenile- and adult-onset patients. Acta Neuropathologica Communications, 2014, 2, 2.	5.2	55
123	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
124	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
125	Clinical characteristics and outcomes of primary antibody deficiency: A 20-year follow-up study. Journal of the Formosan Medical Association, 2014, 113, 340-348.	1.7	17
126	Development of a feasible assay for the detection of GAA mutations in patients with Pompe disease. Clinica Chimica Acta, 2014, 429, 18-25.	1.1	7

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127	Outcome of early-treated type III Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2014, 53, 105-109.	1.4	22
128	Two Frequent Mutations Associated with the Classic Form of Propionic Acidemia in Taiwan. Biochemical Genetics, 2014, 52, 415-429.	1.7	13
129	AADC Deficiency. , 2014, , 3-4.		0
130	Promising outcomes in glutaric aciduria type I patients detected by newborn screening. Metabolic Brain Disease, 2013, 28, 61-67.	2.9	29
131	Newborn screening for citrin deficiency and carnitine uptake defect using second-tier molecular tests. BMC Medical Genetics, 2013, 14, 24.	2.1	22
132	Lysoâ€globotriaosylsphingosine (lysoâ€Gb₃) levels in neonates and adults with the Fabry disease laterâ€onset GLA IVS4+919G>A mutation. Journal of Inherited Metabolic Disease, 2013, 36, 881-885.	3.6	18
133	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
134	Pompe Disease: Early Diagnosis and Early Treatment Make a Difference. Pediatrics and Neonatology, 2013, 54, 219-227.	0.9	135
135	AADC Deficiency. Advances in Pharmacology, 2013, 68, 273-284.	2.0	10
136	Carnitine Uptake Defect (Primary Carnitine Deficiency): Risk in Genotype-Phenotype Correlation. Human Mutation, 2013, 34, 655-655.	2.5	13
137	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
138	Lung toxicity of hydroxypropyl-Î²-cyclodextrin infusion. Molecular Genetics and Metabolism, 2013, 109, 231-232.	1.1	27
139	Diagnosing mucopolysaccharidosis IVA. Journal of Inherited Metabolic Disease, 2013, 36, 293-307.	3.6	77
140	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
141	Fatty Acid Oxidation Disorders in a Chinese Population in Taiwan. JIMD Reports, 2013, 11, 165-172.	1.5	26
142	Clinical Features and Genetic Analysis of Taiwanese Patients With the Hyper IgM Syndrome Phenotype. Pediatric Infectious Disease Journal, 2013, 32, 1010-1016.	2.0	24
143	Analysis of Lyso-Globotriaosylsphingosine in Dried Blood Spots. Annals of Laboratory Medicine, 2013, 33, 274-278.	2.5	29
144	Myostatin and Insulin-Like Growth Factor I: Potential Therapeutic Biomarkers for Pompe Disease. PLoS ONE, 2013, 8, e71900.	2.5	15

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145	Web-Based Newborn Screening System for Metabolic Diseases: Machine Learning Versus Clinicians. Journal of Medical Internet Research, 2013, 15, e98.	4.3	19
146	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
147	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
148	Human Pompe disease-induced pluripotent stem cells for pathogenesis modeling, drug testing and disease marker identification. Human Molecular Genetics, 2012, 21, 2618-2618.	2.9	0
149	Multimodel assessment of BRCA1 mutations in Taiwanese (ethnic Chinese) women with early-onset, bilateral or familial breast cancer. Journal of Human Genetics, 2012, 57, 130-138.	2.3	21
150	Newborn Screening for Phenylketonuria: Machine Learning vs Clinicians. , 2012, , .		1
151	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
152	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
153	Early Pathologic Changes and Responses to Treatment in Patients With Later-Onset Pompe Disease. Pediatric Neurology, 2012, 46, 168-171.	2.1	21
154	Algorithm for Pompe disease newborn screening: Results from the Taiwan screening program. Molecular Genetics and Metabolism, 2012, 106, 281-286.	1.1	72
155	An acidic oligopeptide displayed on AAV2 improves axial muscle tropism after systemic delivery. Genetic Vaccines and Therapy, 2012, 10, 3.	1.5	4
156	Dysphagia as a risk factor for mortality in Niemann-Pick disease type C: systematic literature review and evidence from studies with miglustat. Orphanet Journal of Rare Diseases, 2012, 7, 76.	2.7	66
157	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
158	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
159	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
160	Gene Therapy for Aromatic <scp>l</scp> -Amino Acid Decarboxylase Deficiency. Science Translational Medicine, 2012, 4, 134ra61.	12.4	195
161	Integrating Human Genome Database into Electronic Health Record with Sequence Alignment and Compression Mechanism. Journal of Medical Systems, 2012, 36, 2587-2597.	3.6	7
162	A reliable password-based user authentication scheme for Web-based Human Genome Database System. , 2011, , .		0

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163	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	2.4	308
164	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
165	Human Pompe disease-induced pluripotent stem cells for pathogenesis modeling, drug testing and disease marker identification. Human Molecular Genetics, 2011, 20, 4851-4864.	2.9	129
166	Distribution, clinical features and treatment in Taiwanese patients with symptomatic primary immunodeficiency diseases (PIDs) in a nationwide population-based study during 1985â€“2010. Immunobiology, 2011, 216, 1286-1294.	1.9	51
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