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	Recombinant human acid Â-glucosidase: Major clinical benefits in infantile-onset Pompe disease. Neurology, 2007, 68, 99-109.	1.1	696
2	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
3	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
4	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
5	Early Detection of Pompe Disease by Newborn Screening Is Feasible: Results From the Taiwan Screening Program. Pediatrics, 2008, 122, e39-e45.	2.1	207
6	Gene Therapy for Aromatic <scp>l</scp> -Amino Acid Decarboxylase Deficiency. Science Translational Medicine, 2012, 4, 134ra61.	12.4	195
7	Pompe Disease in Infants: Improving the Prognosis by Newborn Screening and Early Treatment. Pediatrics, 2009, 124, e1116-e1125.	2.1	185
8	Pompe Disease: Early Diagnosis and Early Treatment Make a Difference. Pediatrics and Neonatology, 2013, 54, 219-227.	0.9	135
9	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
10	Nationwide survey of extended newborn screening by tandem mass spectrometry in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 295-305.	3.6	128
11	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
12	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
13	Presymptomatic Diagnosis of Spinal Muscular Atrophy Through Newborn Screening. Journal of Pediatrics, 2017, 190, 124-129.e1.	1.8	113
14	Deconstructing Pompe Disease by Analyzing Single Muscle Fibers: â€œTo See a World in a Grain of Sandâ€¦â€• Autophagy, 2007, 3, 546-552.	9.1	102
15	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
16	Genetic heterozygosity and pseudodeficiency in the Pompe disease newborn screening pilot program. Molecular Genetics and Metabolism, 2010, 99, 379-383.	1.1	91
17	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
18	Diagnoses of newborns and mothers with carnitine uptake defects through newborn screening. Molecular Genetics and Metabolism, 2010, 100, 46-50.	1.1	86

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19	Diagnosing mucopolysaccharidosis IVA. Journal of Inherited Metabolic Disease, 2013, 36, 293-307.	3.6	77
20	Brain Development in Infantile-Onset Pompe Disease Treated by Enzyme Replacement Therapy. Pediatric Research, 2006, 60, 349-352.	2.3	75
21	Algorithm for Pompe disease newborn screening: Results from the Taiwan screening program. Molecular Genetics and Metabolism, 2012, 106, 281-286.	1.1	72
22	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
23	Differences in the predominance of lysosomal and autophagic pathologies between infants and adults with Pompe disease: implications for therapy. Molecular Genetics and Metabolism, 2010, 101, 324-331.	1.1	69
24	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
25	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
26	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
27	Enzyme replacement therapy for mucopolysaccharidosis VIâ€"experience in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 421-427.	3.6	59
28	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
29	Long-term efficacy and safety of eladocagene exuparvovec in patients with AADC deficiency. Molecular Therapy, 2022, 30, 509-518.	8.2	58
30	Identification of novel mutations in theSLC25A15gene in hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome: A clinical, molecular, and functional study. Human Mutation, 2009, 30, 741-748.	2.5	57
31	Cystathionine Î³-lyase: Clinical, metabolic, genetic, and structural studies. Molecular Genetics and Metabolism, 2009, 97, 250-259.	1.1	57
32	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
33	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
34	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
35	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
36	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

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37	Poor Outcome for Neonatal-Type Nonketotic Hyperglycinemia Treated With High-Dose Sodium Benzoate and Dextromethorphan. Journal of Child Neurology, 2004, 19, 39-42.	1.4	48
38	Identification of variations in the human phosphoinositide 3-kinase p110? gene in children with primary B-cell immunodeficiency of unknown aetiology. International Journal of Immunogenetics, 2006, 33, 361-369.	1.8	48
39	Detection and imaging of non-contractile inclusions and sarcomeric anomalies in skeletal muscle by second harmonic generation combined with two-photon excited fluorescence. Journal of Structural Biology, 2008, 162, 500-508.	2.8	48
40	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
41	Newborn screening for Fabry disease by measuring GLA activity using tandem mass spectrometry. Clinica Chimica Acta, 2010, 411, 1428-1431.	1.1	47
42	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
43	Congenital intracranial teratoma. Pediatric Neurology, 2000, 22, 72-74.	2.1	45
44	Rehospitalization of extremely-low-birth-weight infants in first 2 years of life. Early Human Development, 2002, 66, 33-40.	1.8	44
45	Blood Beta-Amyloid and Tau in Down Syndrome: A Comparison with Alzheimerâ€™s Disease. Frontiers in Aging Neuroscience, 2016, 8, 316.	3.4	44
46	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
47	Treatment of Niemannâ€“Pick disease type C in two children with miglustat: Initial responses and maintenance of effects over 1Âyear. Journal of Inherited Metabolic Disease, 2007, 30, 826-826.	3.6	42
48	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
49	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
50	Rapid progressive course of later-onset Pompe disease in Chinese patients. Molecular Genetics and Metabolism, 2011, 104, 284-288.	1.1	39
51	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
52	Mapping of psoriasis to 17q terminus. Journal of Medical Genetics, 2005, 42, 152-158.	3.2	38
53	Newborn Screening for Severe Combined Immunodeficiency in Taiwan. International Journal of Neonatal Screening, 2017, 3, 16.	3.2	38
54	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

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55	A Promoter Sequence Variant of ZNF750 Is Linked with Familial Psoriasis. Journal of Investigative Dermatology, 2008, 128, 1662-1668.	0.7	35
56	Stabilization of blood methylmalonic acid level in methylmalonic acidemia after liver transplantation. Pediatric Transplantation, 2010, 14, 337-341.	1.0	35
57	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
58	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
59	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
60	Spectrum of hypermethioninemia in neonatal screening. Early Human Development, 2005, 81, 529-533.	1.8	32
61	Tandem Mass Neonatal Screening in Taiwanâ€”Report from One Center. Journal of the Formosan Medical Association, 2006, 105, 882-886.	1.7	32
62	Hypertrophic Cardiomyopathy in Pompe Disease Is Not Limited to the Classic Infantile-Onset Phenotype. JIMD Reports, 2014, 17, 71-75.	1.5	30
63	Gene therapy improves brain white matter in aromatic lâ€amino acid decarboxylase deficiency. Annals of Neurology, 2019, 85, 644-652.	5.3	30
64	Promising outcomes in glutaric aciduria type I patients detected by newborn screening. Metabolic Brain Disease, 2013, 28, 61-67.	2.9	29
65	Analysis of Lyso-Globotriaosylsphingosine in Dried Blood Spots. Annals of Laboratory Medicine, 2013, 33, 274-278.	2.5	29
66	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
67	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
68	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
69	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
70	Mutation spectrum in Taiwanese patients with phenylalanine hydroxylase deficiency and a founder effect for the R241C mutation. Human Mutation, 2004, 23, 206-206.	2.5	28
71	Enzymatic activity of methionine adenosyltransferase variants identified in patients with persistent hypermethioninemia. Molecular Genetics and Metabolism, 2010, 101, 172-177.	1.1	28
72	A Neuron-Specific Gene Therapy Relieves Motor Deficits in Pompe Disease Mice. Molecular Neurobiology, 2018, 55, 5299-5309.	4.0	28

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73	Treatment and outcome of Taiwanese patients with 6-pyruvoyltetrahydropterin synthase gene mutations. Journal of Inherited Metabolic Disease, 2001, 24, 815-823.	3.6	27
74	Lung toxicity of hydroxypropyl-Î²-cyclodextrin infusion. Molecular Genetics and Metabolism, 2013, 109, 231-232.	1.1	27
75	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
76	Fatty Acid Oxidation Disorders in a Chinese Population in Taiwan. JIMD Reports, 2013, 11, 165-172.	1.5	26
77	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
78	Natural History of Aromatic l-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Reports, 2017, 40, 1-6.	1.5	26
79	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
80	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
81	Comparison of GATK and DeepVariant by trio sequencing. Scientific Reports, 2022, 12, 1809.	3.3	26
82	The Genetics of Atopic Dermatitis. Clinical Reviews in Allergy and Immunology, 2007, 33, 178-190.	6.5	25
83	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
84	Benefits of Neuronal Preferential Systemic Gene Therapy for Neurotransmitter Deficiency. Molecular Therapy, 2015, 23, 1572-1581.	8.2	25
85	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
86	Association between levels of TNF-alpha and TNF-alpha promoter -308 A/A polymorphism in children with Kawasaki disease. Journal of the Formosan Medical Association, 2003, 102, 147-50.	1.7	25
87	Brain Damage by Mild Metabolic Derangements in Methylmalonic Acidemia. Pediatric Neurology, 2008, 39, 325-329.	2.1	24
88	Computational analysis of a novel mutation in ETFDH gene highlights its long-range effects on the FAD-binding motif. BMC Structural Biology, 2011, 11, 43.	2.3	24
89	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
90	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

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91	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
92	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
93	Newborn screening: Taiwanese experience. Annals of Translational Medicine, 2019, 7, 281-281.	1.7	23
94	Newborn screening for citrin deficiency and carnitine uptake defect using second-tier molecular tests. BMC Medical Genetics, 2013, 14, 24.	2.1	22
95	Outcome of early-treated type III Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2014, 53, 105-109.	1.4	22
96	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
97	Early Pathologic Changes and Responses to Treatment in Patients With Later-Onset Pompe Disease. Pediatric Neurology, 2012, 46, 168-171.	2.1	21
98	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
99	Newborn screening for neuropathic lysosomal storage disorders. Journal of Inherited Metabolic Disease, 2010, 33, 381-386.	3.6	20
100	SLC25A13 gene mutations in Taiwanese patients with non-viralhepatocellular carcinoma. Molecular Genetics and Metabolism, 2011, 103, 293-296.	1.1	20
101	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
102	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
103	A Review of Biomarkers for Alzheimerâ€™s Disease in Down Syndrome. Neurology and Therapy, 2017, 6, 69-81.	3.2	19
104	Web-Based Newborn Screening System for Metabolic Diseases: Machine Learning Versus Clinicians. Journal of Medical Internet Research, 2013, 15, e98.	4.3	19
105	Eye anomalies and neurological manifestations in patients with PAX6 mutations. Molecular Vision, 2009, 15, 2139-45.	1.1	19
106	De novo mutation in the BTK gene of atypical X-linked agammaglobulinemia in a patient with recurrent pyoderma. Annals of Allergy, Asthma and Immunology, 2006, 96, 744-748.	1.0	18
107	Early Detection of Glutaric Aciduria Type I by Newborn Screening in Taiwan. Journal of the Formosan Medical Association, 2008, 107, 139-144.	1.7	18
108	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

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109	Plasma chitotriosidase activity and malaria. Clinica Chimica Acta, 2005, 353, 215.	1.1	17
110	Time course of acylcarnitine elevation in neonatal intrahepatic cholestasis caused by citrin deficiency. Journal of Inherited Metabolic Disease, 2006, 29, 551-555.	3.6	17
111	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
112	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
113	Diffusion tensor images in children with early-treated, chronic, malignant phenylketonuric: correlation with intelligence assessment. American Journal of Neuroradiology, 2004, 25, 1569-74.	2.4	17
114	Caloric restriction in AlstrÃ¶m syndrome prevents hyperinsulinemia. American Journal of Medical Genetics, Part A, 2009, 149A, 666-668.	1.2	16
115	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
116	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€“2019). Orphanet Journal of Rare Diseases, 2020, 15, 314.	2.7	16
117	KIF21A gene c.2860C>T mutation in congenital fibrosis of extraocular muscles type 1 and 3. Molecular Vision, 2005, 11, 245-8.	1.1	16
118	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
119	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
120	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
121	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
122	Genotypic and phenotypic correlations of biotinidase deficiency in the Chinese population. Orphanet Journal of Rare Diseases, 2019, 14, 6.	2.7	15
123	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
124	Myostatin and Insulin-Like Growth Factor I: Potential Therapeutic Biomarkers for Pompe Disease. PLoS ONE, 2013, 8, e71900.	2.5	15
125	Clinical Aspects and Genetic Analysis of Taiwanese Patients with Wiskottâ€“Aldrich Syndrome Protein Mutation: The First Identification of X-Linked Thrombocytopenia in the Chinese with Novel Mutations. Journal of Clinical Immunology, 2010, 30, 593-601.	3.8	14
126	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

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127	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
128	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
129	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
130	Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes. Molecular Genetics & Genomic Medicine, 2020, 8, e1455.	1.2	14
131	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
132	Carnitine Uptake Defect (Primary Carnitine Deficiency): Risk in Genotype-Phenotype Correlation. Human Mutation, 2013, 34, 655-655.	2.5	13
133	Two Frequent Mutations Associated with the Classic Form of Propionic Acidemia in Taiwan. Biochemical Genetics, 2014, 52, 415-429.	1.7	13
134	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
135	Duchenne muscular dystrophy newborn screening: the first 50,000 newborns screened in Taiwan. Neurological Sciences, 2022, 43, 4563-4566.	1.9	13
136	Slipped Capital Femoral Epiphysis as a Complication of Growth Hormone Therapy. Journal of the Formosan Medical Association, 2007, 106, S46-S50.	1.7	12
137	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
138	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
139	Rapid detection of FGFR3 gene mutation in achondroplasia by DHPLC system-coupling heteroduplex and fluorescence-enhanced primer-extension analysis. Journal of Human Genetics, 2004, 49, 399-403.	2.3	11
140	Deficiency of the carnitine transporter (OCTN2) with partial N-acetylglutamate synthase (NAGS) deficiency. Journal of Inherited Metabolic Disease, 2007, 30, 816-816.	3.6	11
141	Cyclic Pamidronate Infusion for Neonatal-onset Osteogenesis Imperfecta. Pediatrics and Neonatology, 2014, 55, 306-311.	0.9	11
142	Congenital generalized lipodystrophy in Taiwan. Journal of the Formosan Medical Association, 2019, 118, 142-147.	1.7	11
143	Cranial MR spectroscopy of tetrahydrobiopterin deficiency. American Journal of Neuroradiology, 2002, 23, 1055-8.	2.4	11
144	Neonatal screening for congenital adrenal hyperplasia in Taiwan: a pilot study. Journal of the Formosan Medical Association, 2002, 101, 691-4.	1.7	11

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145	Pamidronate treatment of severe osteogenesis imperfecta in a newborn infant. Journal of Inherited Metabolic Disease, 2002, 25, 593-595.	3.6	10
146	Transcatheter Closure of Portal-Systemic Shunt Combining Congenital Double Extrahepatic Inferior Vena Cava with Vascular Plug. Journal of Pediatrics, 2008, 153, 723.	1.8	10
147	Congenital Hypopituitarism due to POU1F1 Gene Mutation. Journal of the Formosan Medical Association, 2011, 110, 58-61.	1.7	10
148	AADC Deficiency. Advances in Pharmacology, 2013, 68, 273-284.	2.0	10
149	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
150	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
151	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
152	Heterogeneous nonataxic phenotypes of spinocerebellar ataxia in a Taiwanese population. Brain and Behavior, 2019, 9, e01414.	2.2	10
153	Outcome of Later-Onset Pompe Disease Identified Through Newborn Screening. Journal of Pediatrics, 2022, 244, 139-147.e2.	1.8	10
154	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
155	Newborn Screening Healthcare Information System Based on Service-Oriented Architecture. Journal of Medical Systems, 2010, 34, 519-530.	3.6	9
156	Elevation of urinary globotriaosylceramide (GL3) in infants with Fabry disease. Molecular Genetics and Metabolism, 2011, 102, 57-60.	1.1	9
157	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
158	A novel deep intronic variant strongly associates with Alkaptonuria. Npj Genomic Medicine, 2021, 6, 89.	3.8	9
159	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
160	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
161	Torsade de pointes ventricular tachycardia during elective intubation in a patient with Pompe disease. Paediatric Anaesthesia, 2008, 18, 346-348.	1.1	7
162	Clinical Aspects and Molecular Analysis of Chinese Patients with Wiskott-Aldrich Syndrome in Taiwan. International Archives of Allergy and Immunology, 2008, 145, 15-23.	2.1	7

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163	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
164	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
165	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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