Wuh-Liang Hwu

List of Publications by Year
in descending order

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280 papers	9,434 citations	47006 47 h-index	56724 83 g-index
287 all docs	287 docs citations	287 times ranked	9517 citing authors

#	Article	IF	Citations
1	A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. Journal of Pediatrics, 2006, 148, 671-676.e2.	1.8	500
2	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856.	0.6	401
3	PSORS2 Is Due to Mutations in CARD14. American Journal of Human Genetics, 2012, 90, 784-795.	6.2	365
4	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
5	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
6	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
7	Early Detection of Pompe Disease by Newborn Screening Is Feasible: Results From the Taiwan Screening Program. Pediatrics, 2008, 122, e39-e45.	2.1	207
8	Gene Therapy for Aromatic <scp>l</scp> -Amino Acid Decarboxylase Deficiency. Science Translational Medicine, 2012, 4, 134ra61.	12.4	195
9	Pompe Disease in Infants: Improving the Prognosis by Newborn Screening and Early Treatment. Pediatrics, 2009, 124, e1116-e1125.	2.1	185
10	Incidence of the mucopolysaccharidoses in Taiwan, 1984â€“2004. American Journal of Medical Genetics, Part A, 2009, 149A, 960-964.	1.2	145
11	Pompe Disease: Early Diagnosis and Early Treatment Make a Difference. Pediatrics and Neonatology, 2013, 54, 219-227.	0.9	135
12	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
13	Nationwide survey of extended newborn screening by tandem mass spectrometry in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 295-305.	3.6	128
14	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
15	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
16	Presymptomatic Diagnosis of Spinal Muscular Atrophy Through Newborn Screening. Journal of Pediatrics, 2017, 190, 124-129.e1.	1.8	113
17	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
18	How well does urinary lyso-Gb3 function as a biomarker in Fabry disease?. Clinica Chimica Acta, 2010, 411, 1906-1914.	1.1	94

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19	Genetic heterozygosity and pseudodeficiency in the Pompe disease newborn screening pilot program. Molecular Genetics and Metabolism, 2010, 99, 379-383.	1.1	91
20	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
21	Diagnoses of newborns and mothers with carnitine uptake defects through newborn screening. Molecular Genetics and Metabolism, 2010, 100, 46-50.	1.1	86
22	Parkin Mutations and Early-Onset Parkinsonism in a Taiwanese Cohort. Archives of Neurology, 2005, 62, 82.	4.5	84
23	Diagnosing mucopolysaccharidosis IVA. Journal of Inherited Metabolic Disease, 2013, 36, 293-307.	3.6	77
24	Brain Development in Infantile-Onset Pompe Disease Treated by Enzyme Replacement Therapy. Pediatric Research, 2006, 60, 349-352.	2.3	75
25	Algorithm for Pompe disease newborn screening: Results from the Taiwan screening program. Molecular Genetics and Metabolism, 2012, 106, 281-286.	1.1	72
26	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
27	Screening of nine SLC25A13 mutations: their frequency in patients with citrin deficiency and high carrier rates in Asian populations. Molecular Genetics and Metabolism, 2003, 80, 356-359.	1.1	69
28	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
29	The use of dried blood spot samples in the diagnosis of lysosomal storage disorders â€" Current status and perspectives. Molecular Genetics and Metabolism, 2011, 104, 144-148.	1.1	69
30	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
31	Cloning of Dimethylglycine Dehydrogenase and a New Human Inborn Error of Metabolism, Dimethylglycine Dehydrogenase Deficiency. American Journal of Human Genetics, 2001, 68, 839-847.	6.2	66
32	A validated disease severity scoring system for adults with type 1 Gaucher disease. Genetics in Medicine, 2010, 12, 44-51.	2.4	66
33	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
34	Phenotypic Analyses and Mutation Screening of the <i>SLC26A4</i> and <i>FOXI1</i> Genes in 101 Taiwanese Families with Bilateral Nonsyndromic Enlarged Vestibular Aqueduct (DFNB4) or Pendred Syndrome. Audiology and Neuro-Otology, 2010, 15, 57-66.	1.3	60
35	Enzyme replacement therapy for mucopolysaccharidosis VIâ€"experience in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 421-427.	3.6	59
36	Molecular genetic study of Pompe disease in Chinese patients in Taiwan. Human Mutation, 1999, 13, 380-384.	2.5	58

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37	Copy number analysis of survival motor neuron genes by multiplex ligation-dependent probe amplification. Genetics in Medicine, 2007, 9, 241-248.	2.4	58
38	Long-term efficacy and safety of eladocagene exuparvovec in patients with AADC deficiency. Molecular Therapy, 2022, 30, 509-518.	8.2	58
39	Cystathionine Î³-lyase: Clinical, metabolic, genetic, and structural studies. Molecular Genetics and Metabolism, 2009, 97, 250-259.	1.1	57
40	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
41	Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). Molecular Genetics and Metabolism, 2019, 126, 98-105.	1.1	56
42	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
43	Establishing a standardized therapeutic testing protocol for spinal muscular atrophy. Neurobiology of Disease, 2006, 24, 286-295.	4.4	54
44	Prevalence of theFMR1 mutation in Taiwan assessed by large-scale screening of newborn boys and analysis of DXS548-FRAXAC1 haplotype. American Journal of Medical Genetics, Part A, 2005, 133A, 37-43.	1.2	53
45	Newborn Screening for Pompe Disease: Synthesis of the Evidence and Development of Screening Recommendations. Pediatrics, 2007, 120, e1327-e1334.	2.1	53
46	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
47	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
48	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
49	Screening Assay of Very Long Chain Fatty Acids in Human Plasma with Multiwalled Carbon Nanotube-Based Surface-Assisted Laser Desorption/Ionization Mass Spectrometry. Analytical Chemistry, 2010, 82, 6814-6820.	6.5	50
50	Dopa-responsive dystonia is induced by a dominant-negative mechanism. Annals of Neurology, 2000, 48, 609-613.	5.3	48
51	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
52	Mutation analysis of Gaucher disease patients in Taiwan: High prevalence of the RecNciI and L444P mutations. Blood Cells, Molecules, and Diseases, 2006, 36, 422-425.	1.4	47
53	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
54	Newborn screening for Fabry disease by measuring GLA activity using tandem mass spectrometry. Clinica Chimica Acta, 2010, 411, 1428-1431.	1.1	47

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55	Clinical,18F-dopa PET, and genetic analysis of an ethnic Chinese kindred with early-onset parkinsonism andparkin gene mutations. Movement Disorders, 2002, 17, 670-675.	3.9	44
56	Blood Beta-Amyloid and Tau in Down Syndrome: A Comparison with Alzheimerâ€™s Disease. Frontiers in Aging Neuroscience, 2016, 8, 316.	3.4	44
57	Management of Confirmed Newborn-Screened Patients With Pompe Disease Across the Disease Spectrum. Pediatrics, 2017, 140, S24-S45.	2.1	43
58	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
59	Systemic Administration of a Recombinant AAV1 Vector Encoding IGF-1 Improves Disease Manifestations in SMA Mice. Molecular Therapy, 2014, 22, 1450-1459.	8.2	42
60	IGF-1 delivery to CNS attenuates motor neuron cell death but does not improve motor function in type III SMA mice. Neurobiology of Disease, 2012, 45, 272-279.	4.4	41
61	Somatic and germâ€line mosaicism in Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1463-1467.	1.2	40
62	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
63	Rapid progressive course of later-onset Pompe disease in Chinese patients. Molecular Genetics and Metabolism, 2011, 104, 284-288.	1.1	39
64	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
65	Mapping of psoriasis to 17q terminus. Journal of Medical Genetics, 2005, 42, 152-158.	3.2	38
66	Newborn Screening for Severe Combined Immunodeficiency in Taiwan. International Journal of Neonatal Screening, 2017, 3, 16.	3.2	38
67	Neonatal type of nonketotic hyperglycinemia. Pediatric Neurology, 1999, 20, 295-300.	2.1	37
68	Hepatic steatosis and neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD) in Taiwanese infants. Journal of Pediatrics, 2006, 148, 642-646.	1.8	37
69	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
70	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
71	A Promoter Sequence Variant of ZNF750 Is Linked with Familial Psoriasis. Journal of Investigative Dermatology, 2008, 128, 1662-1668.	0.7	35
72	Stabilization of blood methylmalonic acid level in methylmalonic acidemia after liver transplantation. Pediatric Transplantation, 2010, 14, 337-341.	1.0	35

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73	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
74	Towards a reference genome that captures global genetic diversity. Nature Communications, 2020, 11, 5482.	12.8	34
75	Outcome of pulmonary and aortic stenosis in Williamsâ€Beuren syndrome in an Asian cohort. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 906-909.	1.5	33
76	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
77	Spectrum of hypermethioninemia in neonatal screening. Early Human Development, 2005, 81, 529-533.	1.8	32
78	Tandem Mass Neonatal Screening in Taiwanâ€"Report from One Center. Journal of the Formosan Medical Association, 2006, 105, 882-886.	1.7	32
79	Glucose-6-phosphatase gene G327A mutation is common in Chinese patients with glycogen storage disease type Ia. Human Molecular Genetics, 1995, 4, 1095-1096.	2.9	30
80	Homozygous SLC25A13 Mutation in a Taiwanese Patient with Adult-onset Citrullinemia Complicated with Steatosis and Hepatocellular Carcinoma. Journal of the Formosan Medical Association, 2006, 105, 852-856.	1.7	30
81	Hypertrophic Cardiomyopathy in Pompe Disease Is Not Limited to the Classic Infantile-Onset Phenotype. JIMD Reports, 2014, 17, 71-75.	1.5	30
82	Gene therapy improves brain white matter in aromatic lâ€amino acid decarboxylase deficiency. Annals of Neurology, 2019, 85, 644-652.	5.3	30
83	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
84	Reye's syndrome developing in an infant on treatment of Kawasaki syndrome. Journal of Paediatrics and Child Health, 2005, 41, 303-304.	0.8	29
85	Promising outcomes in glutaric aciduria type I patients detected by newborn screening. Metabolic Brain Disease, 2013, 28, 61-67.	2.9	29
86	Analysis of Lyso-Globotriaosylsphingosine in Dried Blood Spots. Annals of Laboratory Medicine, 2013, 33, 274-278.	2.5	29
87	A multicenter, open-label study evaluating safety and clinical outcomes in children (1.4â€"7.5 years) with Hunter syndrome receiving idursulfase enzyme replacement therapy. Genetics in Medicine, 2014, 16, 435-441.	2.4	29
88	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
89	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
90	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29

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91	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
92	Duplication of proteolipid protein gene: A possible major cause of Pelizaeus-Merzbacher disease. Pediatric Neurology, 1997, 17, 125-128.	2.1	28
93	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
94	Enzymatic activity of methionine adenosyltransferase variants identified in patients with persistent hypermethioninemia. Molecular Genetics and Metabolism, 2010, 101, 172-177.	1.1	28
95	A Neuron-Specific Gene Therapy Relieves Motor Deficits in Pompe Disease Mice. Molecular Neurobiology, 2018, 55, 5299-5309.	4.0	28
96	Treatment and outcome of Taiwanese patients with 6-pyruvoyltetrahydropterin synthase gene mutations. Journal of Inherited Metabolic Disease, 2001, 24, 815-823.	3.6	27
97	Lung toxicity of hydroxypropyl-Î²-cyclodextrin infusion. Molecular Genetics and Metabolism, 2013, 109, 231-232.	1.1	27
98	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
99	Fatty Acid Oxidation Disorders in a Chinese Population in Taiwan. JIMD Reports, 2013, 11, 165-172.	1.5	26
100	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
101	Natural History of Aromatic l-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Reports, 2017, 40, 1-6.	1.5	26
102	The Initial Evaluation of Patients After Positive Newborn Screening: Recommended Algorithms Leading to a Confirmed Diagnosis of Pompe Disease. Pediatrics, 2017, 140, S14-S23.	2.1	26
103	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
104	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
105	Comparison of GATK and DeepVariant by trio sequencing. Scientific Reports, 2022, 12, 1809.	3.3	26
106	The controversy regarding diagnostic criteria for early myoclonic encephalopathy. Brain and Development, 1998, 20, 530-535.	1.1	25
107	The Genetics of Atopic Dermatitis. Clinical Reviews in Allergy and Immunology, 2007, 33, 178-190.	6.5	25
108	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

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109	Mutation screening of the EYA1, SIX1, and SIX5 genes in an east asian cohort with branchioâ€otoâ€renal syndrome. Laryngoscope, 2012, 122, 1130-1136.	2.0	25
110	Natural history and clinical assessment of Taiwanese patients with mucopolysaccharidosis IVA. Orphanet Journal of Rare Diseases, 2014, 9, 21.	2.7	25
111	Benefits of Neuronal Preferential Systemic Gene Therapy for Neurotransmitter Deficiency. Molecular Therapy, 2015, 23, 1572-1581.	8.2	25
112	Brain Damage by Mild Metabolic Derangements in Methylmalonic Acidemia. Pediatric Neurology, 2008, 39, 325-329.	2.1	24
113	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
114	Genetic analysis of mucopolysaccharidosis type VI in Taiwanese patients. Clinica Chimica Acta, 2008, 394, 89-93.	1.1	23
115	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
116	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
117	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
118	Newborn screening: Taiwanese experience. Annals of Translational Medicine, 2019, 7, 281-281.	1.7	23
119	Newborn screening for citrin deficiency and carnitine uptake defect using second-tier molecular tests. BMC Medical Genetics, 2013, 14, 24.	2.1	22
120	Outcome of early-treated type III Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2014, 53, 105-109.	1.4	22
121	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
122	Early Pathologic Changes and Responses to Treatment in Patients With Later-Onset Pompe Disease. Pediatric Neurology, 2012, 46, 168-171.	2.1	21
123	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
124	Newborn screening for neuropathic lysosomal storage disorders. Journal of Inherited Metabolic Disease, 2010, 33, 381-386.	3.6	20
125	Long-term follow-up of a girl with Maroteaux-Lamy syndrome after bone marrow transplantation. World Journal of Pediatrics, 2008, 4, 152-154.	1.8	19
126	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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127	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
128	A Review of Biomarkers for Alzheimerâ€™s Disease in Down Syndrome. Neurology and Therapy, 2017, 6, 69-81.	3.2	19
129	Web-Based Newborn Screening System for Metabolic Diseases: Machine Learning Versus Clinicians. Journal of Medical Internet Research, 2013, 15, e98.	4.3	19
130	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
131	Application of SNaPshot multiplex assays for simultaneous multigene mutation screening in patients with idiopathic sensorineural hearing impairment. Laryngoscope, 2009, 119, 2411-2416.	2.0	18
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	Mutation-adapted U1 snRNA corrects a splicing error of the dopa decarboxylase gene. Human Molecular Genetics, 2016, 25, ddw323.	2.9	18
134	Children Conceived by Assisted Reproductive Technology Prone to Low Birth Weight, Preterm Birth, and Birth Defects: A Cohort Review of More Than 50,000 Live Births During 2011â€“2017 in Taiwan. Frontiers in Pediatrics, 2020, 8, 87.	1.9	18
135	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
136	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
137	Gene therapy in the putamen for curing AADC deficiency and Parkinson's disease. EMBO Molecular Medicine, 2021, 13, e14712.	6.9	17
138	Molecular Genetics of Glycogen-Storage Disease Type 1a in Chinese Patients of Taiwan. Molecular Genetics and Metabolism, 2001, 72, 175-180.	1.1	16
139	Caloric restriction in AlstrÃ¶m syndrome prevents hyperinsulinemia. American Journal of Medical Genetics, Part A, 2009, 149A, 666-668.	1.2	16
140	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
141	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€“2019). Orphanet Journal of Rare Diseases, 2020, 15, 314.	2.7	16
142	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
143	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
144	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

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145	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
146	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
147	Genotypic and phenotypic correlations of biotinidase deficiency in the Chinese population. Orphanet Journal of Rare Diseases, 2019, 14, 6.	2.7	15
148	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
149	Myostatin and Insulin-Like Growth Factor I: Potential Therapeutic Biomarkers for Pompe Disease. PLoS ONE, 2013, 8, e71900.	2.5	15
150	Fundus abnormalities in a patient with type I Gaucher's disease with 12-year follow-up. American Journal of Ophthalmology, 2005, 139, 359-362.	3.3	14
151	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
152	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
153	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
154	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
155	Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes. Molecular Genetics & Genomic Medicine, 2020, 8, e1455.	1.2	14
156	Preimplantation and prenatal genetic diagnosis of aromatic L-amino acidÂdecarboxylase deficiency with an amplification refractory mutation system-quantitative polymerase chain reaction. Taiwanese Journal of Obstetrics and Gynecology, 2011, 50, 468-473.	1.3	13
157	Carnitine Uptake Defect (Primary Carnitine Deficiency): Risk in Genotype-Phenotype Correlation. Human Mutation, 2013, 34, 655-655.	2.5	13
158	Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e00617.	1.2	13
159	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
160	Duchenne muscular dystrophy newborn screening: the first 50,000 newborns screened in Taiwan. Neurological Sciences, 2022, 43, 4563-4566.	1.9	13
161	Allele distribution at the FMR1 locus in the general Chinese population. Clinical Genetics, 1999, 55, 353-356.	2.0	12
162	Regulation of GTP cyclohydrolase I by alternative splicing in mononuclear cells. Biochemical and Biophysical Research Communications, 2003, 306, 937-942.	2.1	12

#	Article	IF	Citations
163	Detection of a homozygous D645E mutation of the acidÎ±-glucosidase gene and glycogen deposition in tissues in a second-trimester fetus with infantile glycogen storage disease type II. Prenatal Diagnosis, 2004, 24, 231-232.	2.3	12
164	Genome-Wide Gene Expression Analysis Implicates the Immune Response and Lymphangiogenesis in the Pathogenesis of Fetal Chylothorax. PLoS ONE, 2012, 7, e34901.	2.5	12
165	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
166	Throwing a spotlight on under-recognized manifestations of Gaucher disease: Pulmonary involvement, lymphadenopathy and Gaucheroma. Molecular Genetics and Metabolism, 2021, 133, 335-344.	1.1	12
167	Linkage disequilibrium and linkage analysis of the glucose-6-phosphatase gene. Human Genetics, 1998, 103, 199-203.	3.8	11
168	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
169	Cyclic Pamidronate Infusion for Neonatal-onset Osteogenesis Imperfecta. Pediatrics and Neonatology, 2014, 55, 306-311.	0.9	11
170	Introduction to the Newborn Screening, Diagnosis, and Treatment for Pompe Disease Guidance Supplement. Pediatrics, 2017, 140, S1-S3.	2.1	11
171	Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with Different Types of Mucopolysaccharidoses. Diagnostics, 2019, 9, 148.	2.6	11
172	Congenital generalized lipodystrophy in Taiwan. Journal of the Formosan Medical Association, 2019, 118, 142-147.	1.7	11
173	Cranial MR spectroscopy of tetrahydrobiopterin deficiency. American Journal of Neuroradiology, 2002, 23, 1055-8.	2.4	11
174	Neonatal screening for congenital adrenal hyperplasia in Taiwan: a pilot study. Journal of the Formosan Medical Association, 2002, 101, 691-4.	1.7	11
175	Pamidronate treatment of severe osteogenesis imperfecta in a newborn infant. Journal of Inherited Metabolic Disease, 2002, 25, 593-595.	3.6	10
176	Molecular chaperones affect GTP cyclohydrolase I mutations in dopa-responsive dystonia. Annals of Neurology, 2004, 55, 875-878.	5.3	10
177	Congenital Hypopituitarism due to POU1F1 Gene Mutation. Journal of the Formosan Medical Association, 2011, 110, 58-61.	1.7	10
178	AADC Deficiency. Advances in Pharmacology, 2013, 68, 273-284.	2.0	10
179	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
180	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

#	Article	IF	Citations
181	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
182	Heterogeneous nonataxic phenotypes of spinocerebellar ataxia in a Taiwanese population. Brain and Behavior, 2019, 9, e01414.	2.2	10
183	Glypican-3 induces oncogenicity by preventing IGF-1R degradation, a process that can be blocked by Grb10. Oncotarget, 2017, 8, 80429-80442.	1.8	10
184	Outcome of Later-Onset Pompe Disease Identified Through Newborn Screening. Journal of Pediatrics, 2022, 244, 139-147.e2.	1.8	10
185	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
186	Correlation of Survival Motor Neuron Expression in Leukocytes and Spinal Cord in Spinal Muscular Atrophy. Journal of Pediatrics, 2009, 154, 303-305.	1.8	9
187	Elevation of urinary globotriaosylceramide (GL3) in infants with Fabry disease. Molecular Genetics and Metabolism, 2011, 102, 57-60.	1.1	9
188	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
189	Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. Pediatrics and Neonatology, 2019, 60, 342-343.	0.9	9
190	A novel deep intronic variant strongly associates with Alkaptonuria. Npj Genomic Medicine, 2021, 6, 89.	3.8	9
191	Hereditary progressive dystonia with marked diurnal fluctuation (Segawa syndrome) in Taiwan. Brain and Development, 1994, 16, 126-131.	1.1	8
192	Lack of mutations in spinocerebellar ataxia type 2 and 3 genes in a Taiwanese (ethnic Chinese) cohort of familial and early-onset parkinsonism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 434-438.	1.7	8
193	Fibrous Dysplasia in a Child with Mitochondrial A8344G Mutation. Journal of Child Neurology, 2008, 23, 1447-1450.	1.4	8
194	Analysis of nondegraded and degraded DNA mixtures of close relatives using massively parallel sequencing. Legal Medicine, 2020, 42, 101631.	1.3	8
195	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
196	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
197	Transfusion-acquired cytomegalovirus infection in children in a hyperendemic area. Journal of Medical Virology, 1992, 36, 49-53.	5.0	7
198	Deletion of 11q24.2-qter with agenesis of unilateral internal carotid artery and total anomalous pulmonary venous return. American Journal of Medical Genetics Part A, 2001, 103, 245-248.	2.4	7

#	Article	IF	Citations
199	Bone marrow transplantation results in donor-derived hepatocytes in an animal model of inherited cholestatic liver disease. Journal of Biomedical Science, 2008, 15, 615-622.	7.0	7
200	Torsade de pointes ventricular tachycardia during elective intubation in a patient with Pompe disease. Paediatric Anaesthesia, 2008, 18, 346-348.	1.1	7
201	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
202	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
203	Comprehensive human leukocyte antigen genotyping of patients with type 1 diabetes mellitus in Taiwan. Pediatric Diabetes, 2018, 19, 699-706.	2.9	7
204	Clinical features of Pompe disease with motor neuronopathy. Neuromuscular Disorders, 2019, 29, 903-906.	0.6	7
205	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
206	A review of treatment of Pompe disease in infants. Biologics: Targets and Therapy, 2007, 1, 195-201.	3.2	7
207	Cytogenetic study of mentally retarded children in Taipei. Journal of Biomedical Science, 1994, 1, 100-104.	7.0	6
208	Spontaneous chylothorax in a case of cardio-facio-cutaneous syndrome. Clinical Dysmorphology, 2002, 11, 297-298.	0.3	6
209	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
210	Pseudogeneâ€derived IKBKG gene mutations in incontinentia pigmenti. Clinical Genetics, 2009, 76, 417-419.	2.0	6
211	X-linked Liver Glycogenosis in a Taiwanese Family: Transmission From Undiagnosed Males. Pediatrics and Neonatology, 2009, 50, 230-233.	0.9	6
212	FOXL2 mutations in Taiwanese patients with blepharophimosis, ptosis, epicanthus inversus syndrome. Clinical Chemistry and Laboratory Medicine, 2010, 48, 485-8.	2.3	6
213	Current diagnosis and management of mucopolysaccharidosis VI in the Asia-Pacific region. Molecular Genetics and Metabolism, 2012, 107, 136-144.	1.1	6
214	SHOX deficiency in short Taiwanese children: A single-center experience. Journal of the Formosan Medical Association, 2018, 117, 909-914.	1.7	6
215	Functional independence of Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e790.	1.2	6
216	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

#	Article	IF	Citations
217	Arylsulfatase A pseudodeficiency in Chinese. Human Genetics, 1996, 97, 148-149.	3.8	5
218	Two common mutations 934C to G and 937C to G of fibroblast growth factor receptor 2 (FGFR2) gene in Chinese patients with apert syndrome. Human Mutation, 1998, 11, S18-S19.	2.5	5
219	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
220	Hypothermia improves disease manifestations in SMA mice via SMN augmentation. Human Molecular Genetics, 2016, 25, 631-641.	2.9	5
221	Long-term outcome for Down syndrome patients with hematopoietic disorders. Journal of the Formosan Medical Association, 2016, 115, 94-99.	1.7	5
222	Gene therapy with modified U1 small nuclear RNA. Expert Review of Endocrinology and Metabolism, 2017, 12, 171-175.	2.4	5
223	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
224	Diversity in heritable disorders of connective tissue at a single center. Connective Tissue Research, 2020, 62, 1-6.	2.3	5
225	Dietary intake and nutritional status of patients with phenylketonuria in Taiwan. Scientific Reports, 2020, 10, 14537.	3.3	5
226	Enzyme replacement therapy with imiglucerase in Taiwanese patients with type I Gaucher disease. Journal of the Formosan Medical Association, 2002, 101, 627-31.	1.7	5
227	Molecular diagnosis of Wiskott-Aldrich syndrome in Taiwan. Journal of Microbiology, Immunology and Infection, 2004, 37, 276-81.	3.1	5
228	Mitral annuloplasty in an infant with Barth syndrome and severe mitral insufficiency: First case report and determination of annular diameter. Journal of Thoracic and Cardiovascular Surgery, 2008, 136, 1095-1097.	0.8	4
229	Myopathy in Gaucher disease. Journal of Inherited Metabolic Disease, 2008, 31, 489-491.	3.6	4
230	Schizencephaly in LEOPARD Syndrome. Pediatric Neurology, 2009, 41, 71-73.	2.1	4
231	An acidic oligopeptide displayed on AAV2 improves axial muscle tropism after systemic delivery. Genetic Vaccines and Therapy, 2012, 10, 3.	1.5	4
232	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
233	Composite Scores of Plasma Tau and Î²-Amyloids Correlate with Dementia in Down Syndrome. ACS Chemical Neuroscience, 2020, 11, 191-196.	3.5	4
234	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

#	Article	IF	Citations
235	Type I Gaucher disease with exophthalmos and pulmonary arteriovenous malformation. BMC Medical Genetics, 2005, 6, 25.	2.1	3
236	Adenoviral interneuronal transportation after retrograde gene transfer in mice. Molecular Brain Research, 2005, 142, 151-155.	2.3	3
237	Glycogen Storage Disease Type Ib: The First Case in Taiwan. Pediatrics and Neonatology, 2009, 50, 125-128.	0.9	3
238			

#	Article	IF	Citations
253	Reduced Immunogenicity of Intraparenchymal Delivery of Adeno-Associated Virus Serotype 2 Vectors: Brief Overview. Current Gene Therapy, 2021, 21, .	2.0	2
254	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
255	Gene symbol: OTC. Disease: ornithine carbamoyltransferase deficiency. Human Genetics, 2003, 113, 365.	3.8	2
256	Gene symbol: OTC. Disease: ornithine carbamoyltransferase deficiency. Human Genetics, 2003, 113, 367.	3.8	2
257	Phenylalanine hydroxylase deficiency: intelligence of patients after early dietary treatment. Acta Paediatrica Taiwanica = Taiwan Er Ke Yi Xue Hui Za Zhi, 2004, 45, 320-3.	0.1	2
258	Neonatal screening and monitoring system in Taiwan. Southeast Asian Journal of Tropical Medicine and Public Health, 2003, 34 Suppl 3, 91-3.	1.0	2
259	Newborn Screening for Phenylketonuria: Machine Learning vs Clinicians. , 2012, , .		1
260	Parental discussion of G6PD deficiency and child health: implications for clinical practice. Archives of Disease in Childhood, 2014, 99, 251-255.	1.9	1
261	Mortality, disability, and intensive care in patients with mitochondrial 3243A>G mutation. Intensive Care Medicine, 2015, 41, 1493-1495.	8.2	1
262	Ultrastructural and diffusion tensor imaging studies reveal axon abnormalities in Pompe disease mice. Scientific Reports, 2020, 10, 20239.	3.3	1
263	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
264	The Timely Needs for Infantile Onset Pompe Disease Newborn Screeningâ€”Practice in Taiwan. International Journal of Neonatal Screening, 2020, 6, 30.	3.2	1
265	Rapid Trio Exome Sequencing for Autosomal Recessive Renal Tubular Dysgenesis in Recurrent Oligohydramnios. Frontiers in Genetics, 2021, 12, 606970.	2.3	1
266	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
267	Advanced therapeutic strategy for hereditary neuromuscular diseases. Molecular Therapy, 2022, 30, 12-13.	8.2	1
268	Homozygous mutation 341delG/101X of the glucose-6-phosphatase (G6PC) gene causes glycogen storage disease type Ia (von Gierke disease) in a Chinese patient. Human Mutation, 1999, 13, 414-414.	2.5	0
269	Screening of Newborns for Pompe Disease and/or Other Lysosomal Storage Disorders. Clinical Therapeutics, 2008, 30, S8.	2.5	0
270	Alglucosidase Alfa (MyozymeÂ®) in Infants and Children with Rapidly Progressive Pompe Disease. Clinical Therapeutics, 2008, 30, S9-S10.	2.5	0

#	Article	IF	Citations
271	Hyperammonemia and positive allopurinol test in hyperinsulinismâ€“hyperammonemia syndrome: Taiwanese case report. Pediatrics International, 2009, 51, 305-307.	0.5	0
272	Can a girl with 3-hydroxy-3-methylglutaryl-CoA lyase deficiency live a normal life?. Tzu Chi Medical Journal, 2012, 24, 215-217.	1.1	0
273	Congenital Malformations in Newbornsâ€”A Challenge Unmet for Decades. Pediatrics and Neonatology, 2015, 56, 5-6.	0.9	0
274	Advances in newborn screening for Pompe disease and resulting clinical outcomes. Expert Opinion on Orphan Drugs, 2016, 4, 21-29.	0.8	0
275	High incidence of coâ€existing GLA variants and stroke susceptibility. European Journal of Neurology, 2019, 26, e70-e70.	3.3	0
276	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
277	Thymidine Kinase 2 Deficiencyâ€“Induced Adult-Onset Ptosis and Proximal Weakness. Neurology: Clinical Practice, 2021, 11, e379-e382.	1.6	0
278	AADC Deficiency. , 2014, , 3-4.		0
279	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
280	Molecular Analysis of Vietnamese Patients with Mucopolysaccharidosis Type I. Life, 2021, 11, 1162.	2.4	0