Hsiang-Yu Lin

List of Publications by Year
in descending order

80	1,648	279798 23	315739 38
papers	citations	h-index	g-index

82	82	82	1861
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	High Incidence of the Cardiac Variant of Fabry Disease Revealed by Newborn Screening in the Taiwan Chinese Population. Circulation: Cardiovascular Genetics, 2009, 2, 450-456.	5.1	214
2	Incidence of the mucopolysaccharidoses in Taiwan, 1984â€“2004. American Journal of Medical Genetics, Part A, 2009, 149A, 960-964.	1.2	145
3	Intrauterine inflammation, infection, or both (Triple I): A new concept for chorioamnionitis. Pediatrics and Neonatology, 2018, 59, 231-237.	0.9	87
4	A pilot newborn screening program for Mucopolysaccharidosis type I in Taiwan. Orphanet Journal of Rare Diseases, 2013, 8, 147.	2.7	69
5	Enzyme replacement therapy for mucopolysaccharidosis VIâ€”experience in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 421-427.	3.6	59
6	A modified liquid chromatography/tandem mass spectrometry method for predominant disaccharide units of urinary glycosaminoglycans in patients with mucopolysaccharidoses. Orphanet Journal of Rare Diseases, 2014, 9, 135.	2.7	55
7	Characterization of pulmonary function impairments in patients with mucopolysaccharidosesâ€”changes with age and treatment. Pediatric Pulmonology, 2014, 49, 277-284.	2.0	51
8	Status of newborn screening and follow up investigations for Mucopolysaccharidoses I and II in Taiwan. Orphanet Journal of Rare Diseases, 2018, 13, 84.	2.7	50
9	Polysomnographic characteristics in patients with mucopolysaccharidoses. Pediatric Pulmonology, 2010, 45, 1205-1212.	2.0	47
10	Taiwan National Newborn Screening Program by Tandem Mass Spectrometry for Mucopolysaccharidoses Types I, II, and VI. Journal of Pediatrics, 2019, 205, 176-182.	1.8	46
11	Therapeutic strategy of patent ductus arteriosus in extremely preterm infants. Pediatrics and Neonatology, 2020, 61, 133-141.	0.9	45
12	Circulatory Management Focusing on Preventing Intraventricular Hemorrhage and Pulmonary Hemorrhage in Preterm Infants. Pediatrics and Neonatology, 2016, 57, 453-462.	0.9	40
13	Association of preterm birth and small for gestational age with metabolic outcomes inÂchildren and adolescents: A population-based cohort study from Taiwan. Pediatrics and Neonatology, 2018, 59, 147-153.	0.9	38
14	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
15	Assessment of hearing loss by pure-tone audiometry in patients with mucopolysaccharidoses. Molecular Genetics and Metabolism, 2014, 111, 533-538.	1.1	36
16	Detection of hunter syndrome (mucopolysaccharidosis type II) in Taiwanese: Biochemical and linkage studies of the iduronate-2-sulfatase gene defects in MPS II patients and carriers. Clinica Chimica Acta, 2006, 369, 29-34.	1.1	33
17	Assessment of bone mineral density by dual energy x-ray absorptiometry in patients with mucopolysaccharidoses. Orphanet Journal of Rare Diseases, 2013, 8, 71.	2.7	33
18	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

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19	Cardiovascular abnormalities in Taiwanese patients with mucopolysaccharidosis. Molecular Genetics and Metabolism, 2014, 111, 493-498.	1.1	26
20	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
21	Natural history and clinical assessment of Taiwanese patients with mucopolysaccharidosis IVA. Orphanet Journal of Rare Diseases, 2014, 9, 21.	2.7	25
22	Enzyme assay and clinical assessment in subjects with a Chinese hotspot lateâ€onset Fabry mutation (IVS4â€‰+â€‰919Gâ†’A). Journal of Inherited Metabolic Disease, 2010, 33, 619-624.	3.6	24
23	The relationships between urinary glycosaminoglycan levels and phenotypes of mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2018, 6, 982-992.	1.2	24
24	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
25	Genotype and phenotype in patients with Praderâ€“Willi Syndrome in Taiwan. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 902-905.	1.5	23
26	Overcoming the barriers to diagnosis of Morquio A syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 192.	2.7	21
27	Cardiac features and effects of enzyme replacement therapy in Taiwanese patients with Mucopolysaccharidosis IVA. Orphanet Journal of Rare Diseases, 2018, 13, 148.	2.7	18
28	Prader?Willi syndrome in Taiwan. Pediatrics International, 2007, 49, 375-379.	0.5	16
29	Effects of enzyme replacement therapy for cardiac-type Fabry patients with a Chinese hotspot late-onset Fabry mutation (IVS4+919G>A). BMJ Open, 2013, 3, e003146.	1.9	16
30	Normalization of glycosaminoglycan-derived disaccharides detected by tandem mass spectrometry assay for the diagnosis of mucopolysaccharidosis. Scientific Reports, 2019, 9, 10755.	3.3	16
31	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€“2019). Orphanet Journal of Rare Diseases, 2020, 15, 314.	2.7	16
32	Cardiac characteristics and natural progression in Taiwanese patients with mucopolysaccharidosis III. Orphanet Journal of Rare Diseases, 2019, 14, 140.	2.7	14
33	Identification and Functional Characterization of IDS Gene Mutations Underlying Taiwanese Hunter Syndrome (Mucopolysaccharidosis Type II). International Journal of Molecular Sciences, 2020, 21, 114.	4.1	14
34	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
35	Epigenotype, genotype, and phenotype analysis of patients in Taiwan with Beckwithâ€“Wiedemann syndrome. Molecular Genetics and Metabolism, 2016, 119, 8-13.	1.1	13
36	Functional independence of Taiwanese children with Down syndrome. Developmental Medicine and Child Neurology, 2016, 58, 502-507.	2.1	13

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37	Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e00617.	1.2	13
38	Efficacy of Medical Treatment for Infantile Hypertrophic Pyloric Stenosis: AÂMeta-analysis. Pediatrics and Neonatology, 2016, 57, 515-521.	0.9	12
39	Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with Different Types of Mucopolysaccharidoses. Diagnostics, 2019, 9, 148.	2.6	11
40	Nationwide Newborn Screening Program for Mucopolysaccharidoses in Taiwan and an Update of the â€œGold Standardâ€•Criteria Required to Make a Confirmatory Diagnosis. Diagnostics, 2021, 11, 1583.	2.6	11
41	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
42	An At-Risk Population Screening Program for Mucopolysaccharidoses by Measuring Urinary Glycosaminoglycans in Taiwan. Diagnostics, 2019, 9, 140.	2.6	10
43	Natural progression of cardiac features and long-term effects of enzyme replacement therapy in Taiwanese patients with mucopolysaccharidosis II. Orphanet Journal of Rare Diseases, 2021, 16, 99.	2.7	10
44	Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. Pediatrics and Neonatology, 2019, 60, 342-343.	0.9	9
45	Cardiac Evaluation Using Two-Dimensional Speckle-Tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. Diagnostics, 2020, 10, 62.	2.6	9
46	Array-CGH increased the diagnostic rate of developmental delay or intellectual disability in Taiwan. Pediatrics and Neonatology, 2019, 60, 453-460.	0.9	8
47	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
48	Functional independence of Taiwanese children with VACTERL association. American Journal of Medical Genetics, Part A, 2012, 158A, 3101-3105.	1.2	7
49	Pulmonary Hemorrhage in Very-low-birth-weight Infants. Pediatrics and Neonatology, 2014, 55, 326-327.	0.9	7
50	Long-term outcomes of enzyme replacement therapy for Taiwanese patients with Mucopolysaccharidosis I. Pediatrics and Neonatology, 2019, 60, 577-578.	0.9	7
51	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta I. Human Genetics, 2010, 127, 468.	3.8	7
52	Functional independence of Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e790.	1.2	6
53	Awareness of Mucopolysaccharidosis in an Otorhinolaryngologic Clinic. Pediatrics and Neonatology, 2017, 58, 198-199.	0.9	5
54	Otorhinolaryngological Management in Taiwanese Patients with Mucopolysaccharidoses. International Journal of Medical Sciences, 2021, 18, 3373-3379.	2.5	5

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55	Aortic Root Dilatation in Taiwanese Patients with Mucopolysaccharidoses and the Long-Term Effects of Enzyme Replacement Therapy. Diagnostics, 2021, 11, 16.	2.6	5
56	Increased Diagnostic Yield of Array Comparative Genomic Hybridization for Autism Spectrum Disorder in One Institution in Taiwan. Medicina (Lithuania), 2022, 58, 15.	2.0	5
57	Mucopolysaccharidosis I (Scheie syndrome): A rare cause of severe aortic stenosis in a 31-year-old man. Journal of the Formosan Medical Association, 2015, 114, 1015-1016.	1.7	4
58	Newborn Screening Program for Mucopolysaccharidosis Type II and Long-Term Follow-Up of the Screen-Positive Subjects in Taiwan. Journal of Personalized Medicine, 2022, 12, 1023.	2.5	4
59	Effect of Mutated ids Overexpression on IDS Enzyme Activity and Developmental Phenotypes in Zebrafish Embryos: A Valuable Index for Assessing Critical Point-Mutations Associated with Mucopolysaccharidosis Type II Occurrence in Humans. Diagnostics, 2020, 10, 854.	2.6	3
60	Clinical Utility of Elosulfase Alfa in the Treatment of Morquio A Syndrome.. Drug Design, Development and Therapy, 2022, 16, 143-154.	4.3	3
61	Functional independence of Taiwanese children with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1309-1314.	1.2	2
62	Awareness of attenuated mucopolysaccharidoses in a pediatric orthopedic clinic. Pediatrics and Neonatology, 2019, 60, 100-101.	0.9	2
63	Evidence base multi-discipline critical strategies toward better tomorrow for very preterm infants. Pediatrics and Neonatology, 2020, 61, 371-377.	0.9	2
64	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta IV. Human Genetics, 2010, 127, 466.	3.8	2
65	The first mucopolysaccharidosis type VII in a Taiwanese girl: A case report and review of the literature. Journal of the Formosan Medical Association, 2021, , .	1.7	1
66	Early Diagnosis for Mucopolysaccharidosis I - A 6-month-old Female Infant Presenting with Gibbus, Hirsutism and Mongolian Spots in a Well Baby Clinic. Journal of Mucopolysaccharidosis and Rare Disease, 2016, 2, 23-26.	0.0	1
67	Wiedemannâ€“Steiner Syndrome with a Pathogenic Variant in KMT2A from Taiwan. Children, 2021, 8, 952.	1.5	1
68	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 464.	3.8	1
69	Long-Term Cardiovascular Findings in Williams Syndrome: A Single Medical Center Experience in Taiwan. Journal of Personalized Medicine, 2022, 12, 817.	2.5	1
70	Rapid Weight Loss and Severe Failure to Thrive Mimicking Lipodystrophy Syndrome in a 1-Year-Old Taiwanese Girl with Costello Syndrome. Children, 2022, 9, 905.	1.5	1
71	Gastric Residuals, Feeding Intolerance, and Necrotizing Enterocolitis in Preterm Infants. Pediatrics and Neonatology, 2015, 56, 136-137.	0.9	0
72	Quantitative DNA Methylation Analysis and Epigenotype-Phenotype Correlations in Taiwanese Patients with Beckwith-Wiedemann Syndrome. Journal of Personalized Medicine, 2021, 11, 1066.	2.5	0

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73	Epigenotype, Genotype, and Phenotype Analysis of Taiwanese Patients with Silverâ€“Russell Syndrome. Journal of Personalized Medicine, 2021, 11, 1197.	2.5	0
74	Novel human pathological mutations. Gene symbol: COL1A2. Disease: Osteogenesis imperfecta III. Human Genetics, 2010, 127, 467.	3.8	0
75	Novel human pathological mutations. Gene symbol: OTC. Disease: ornithine transcarbamylase deficiency. Human Genetics, 2010, 127, 475.	3.8	0
76	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 465.	3.8	0
77	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta IV. Human Genetics, 2010, 127, 466.	3.8	0
78	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 464.	3.8	0
79	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta I. Human Genetics, 2010, 127, 467.	3.8	0
80	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta IV. Human Genetics, 2010, 127, 467.	3.8	0