

Hsiang-Yu Lin

List of Publications by Year in descending order

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Version: 2024-02-01

80
papers

1,648
citations

279798

23
h-index

315739

38
g-index

82
all docs

82
docs citations

82
times ranked

1861
citing authors

#	ARTICLE	IF	CITATIONS
1	Increased Diagnostic Yield of Array Comparative Genomic Hybridization for Autism Spectrum Disorder in One Institution in Taiwan. <i>Medicina (Lithuania)</i> , 2022, 58, 15.	2.0	5
2	Clinical Utility of Elosulfase Alfa in the Treatment of Morquio A Syndrome.. <i>Drug Design, Development and Therapy</i> , 2022, 16, 143-154.	4.3	3
3	Long-Term Cardiovascular Findings in Williams Syndrome: A Single Medical Center Experience in Taiwan. <i>Journal of Personalized Medicine</i> , 2022, 12, 817.	2.5	1
4	Newborn Screening Program for Mucopolysaccharidosis Type II and Long-Term Follow-Up of the Screen-Positive Subjects in Taiwan. <i>Journal of Personalized Medicine</i> , 2022, 12, 1023.	2.5	4
5	Rapid Weight Loss and Severe Failure to Thrive Mimicking Lipodystrophy Syndrome in a 1-Year-Old Taiwanese Girl with Costello Syndrome. <i>Children</i> , 2022, 9, 905.	1.5	1
6	Natural progression of cardiac features and long-term effects of enzyme replacement therapy in Taiwanese patients with mucopolysaccharidosis II. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 99.	2.7	10
7	Nationwide Newborn Screening Program for Mucopolysaccharidoses in Taiwan and an Update of the "Gold Standard" Criteria Required to Make a Confirmatory Diagnosis. <i>Diagnostics</i> , 2021, 11, 1583.	2.6	11
8	Molecular basis of mucopolysaccharidosis IVA (Morquio A syndrome): A review and classification of <i>GALNS</i> gene variants and reporting of 68 novel variants. <i>Human Mutation</i> , 2021, 42, 1384-1398.	2.5	14
9	The first mucopolysaccharidosis type VII in a Taiwanese girl: A case report and review of the literature. <i>Journal of the Formosan Medical Association</i> , 2021, , .	1.7	1
10	Otorhinolaryngological Management in Taiwanese Patients with Mucopolysaccharidoses. <i>International Journal of Medical Sciences</i> , 2021, 18, 3373-3379.	2.5	5
11	Aortic Root Dilatation in Taiwanese Patients with Mucopolysaccharidoses and the Long-Term Effects of Enzyme Replacement Therapy. <i>Diagnostics</i> , 2021, 11, 16.	2.6	5
12	Quantitative DNA Methylation Analysis and Epigenotype-Phenotype Correlations in Taiwanese Patients with Beckwith-Wiedemann Syndrome. <i>Journal of Personalized Medicine</i> , 2021, 11, 1066.	2.5	0
13	Wiedemann-Steiner Syndrome with a Pathogenic Variant in <i>KMT2A</i> from Taiwan. <i>Children</i> , 2021, 8, 952.	1.5	1
14	Epigenotype, Genotype, and Phenotype Analysis of Taiwanese Patients with Silver-Russell Syndrome. <i>Journal of Personalized Medicine</i> , 2021, 11, 1197.	2.5	0
15	Therapeutic strategy of patent ductus arteriosus in extremely preterm infants. <i>Pediatrics and Neonatology</i> , 2020, 61, 133-141.	0.9	45
16	Cardiac manifestations and gene mutations of patients with RASopathies in Taiwan. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 357-364.	1.2	8
17	Identification and Functional Characterization of <i>IDS</i> Gene Mutations Underlying Taiwanese Hunter Syndrome (Mucopolysaccharidosis Type II). <i>International Journal of Molecular Sciences</i> , 2020, 21, 114.	4.1	14
18	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985-2019). <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 314.	2.7	16

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19	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. <i>Diagnostics</i> , 2020, 10, 854.	2.6	3
20	Evidence base multi-discipline critical strategies toward better tomorrow for very preterm infants. <i>Pediatrics and Neonatology</i> , 2020, 61, 371-377.	0.9	2
21	Cardiac Evaluation Using Two-Dimensional Speckle-Tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. <i>Diagnostics</i> , 2020, 10, 62.	2.6	9
22	Long-term outcomes of enzyme replacement therapy for Taiwanese patients with Mucopolysaccharidosis I. <i>Pediatrics and Neonatology</i> , 2019, 60, 577-578.	0.9	7
23	Normalization of glycosaminoglycan-derived disaccharides detected by tandem mass spectrometry assay for the diagnosis of mucopolysaccharidosis. <i>Scientific Reports</i> , 2019, 9, 10755.	3.3	16
24	Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with Different Types of Mucopolysaccharidoses. <i>Diagnostics</i> , 2019, 9, 148.	2.6	11
25	An At-Risk Population Screening Program for Mucopolysaccharidoses by Measuring Urinary Glycosaminoglycans in Taiwan. <i>Diagnostics</i> , 2019, 9, 140.	2.6	10
26	Functional independence of Taiwanese patients with mucopolysaccharidoses. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e790.	1.2	6
27	Cardiac characteristics and natural progression in Taiwanese patients with mucopolysaccharidosis III. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 140.	2.7	14
28	Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00617.	1.2	13
29	Taiwan National Newborn Screening Program by Tandem Mass Spectrometry for Mucopolysaccharidoses Types I, II, and VI. <i>Journal of Pediatrics</i> , 2019, 205, 176-182.	1.8	46
30	Array-CGH increased the diagnostic rate of developmental delay or intellectual disability in Taiwan. <i>Pediatrics and Neonatology</i> , 2019, 60, 453-460.	0.9	8
31	Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. <i>Pediatrics and Neonatology</i> , 2019, 60, 342-343.	0.9	9
32	Awareness of attenuated mucopolysaccharidoses in a pediatric orthopedic clinic. <i>Pediatrics and Neonatology</i> , 2019, 60, 100-101.	0.9	2
33	Functional independence of Taiwanese children with Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1309-1314.	1.2	2
34	Intrauterine inflammation, infection, or both (Triple I): A new concept for chorioamnionitis. <i>Pediatrics and Neonatology</i> , 2018, 59, 231-237.	0.9	87
35	Association of preterm birth and small for gestational age with metabolic outcomes in children and adolescents: A population-based cohort study from Taiwan. <i>Pediatrics and Neonatology</i> , 2018, 59, 147-153.	0.9	38
36	The relationships between urinary glycosaminoglycan levels and phenotypes of mucopolysaccharidoses. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 982-992.	1.2	24

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37	Cardiac features and effects of enzyme replacement therapy in Taiwanese patients with Mucopolysaccharidosis IVA. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 148.	2.7	18
38	Mucopolysaccharidosis III in Taiwan: Natural history, clinical and molecular characteristics of 28 patients diagnosed during a 21-year period. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1799-1809.	1.2	24
39	Status of newborn screening and follow up investigations for Mucopolysaccharidoses I and II in Taiwan. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 84.	2.7	50
40	Clinical characteristics and surgical history of Taiwanese patients with mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS). <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 89.	2.7	10
41	Awareness of Mucopolysaccharidosis in an Otorhinolaryngologic Clinic. <i>Pediatrics and Neonatology</i> , 2017, 58, 198-199.	0.9	5
42	Causes of death and clinical characteristics of 34 patients with Mucopolysaccharidosis II in Taiwan from 1995-2012. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 85.	2.7	26
43	Efficacy of Medical Treatment for Infantile Hypertrophic Pyloric Stenosis: A Meta-analysis. <i>Pediatrics and Neonatology</i> , 2016, 57, 515-521.	0.9	12
44	Long-term galsulfase enzyme replacement therapy in Taiwanese mucopolysaccharidosis VI patients: A case series. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 7, 63-69.	1.1	27
45	Epigenotype, genotype, and phenotype analysis of patients in Taiwan with Beckwith-Wiedemann syndrome. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 8-13.	1.1	13
46	Functional independence of Taiwanese children with Down syndrome. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 502-507.	2.1	13
47	Cardiac structure and function and effects of enzyme replacement therapy in patients with mucopolysaccharidoses I, II, IVA and VI. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 431-437.	1.1	37
48	Circulatory Management Focusing on Preventing Intraventricular Hemorrhage and Pulmonary Hemorrhage in Preterm Infants. <i>Pediatrics and Neonatology</i> , 2016, 57, 453-462.	0.9	40
49	Early Diagnosis for Mucopolysaccharidosis I - A 6-month-old Female Infant Presenting with Gibbus, Hirsutism and Mongolian Spots in a Well Baby Clinic. <i>Journal of Mucopolysaccharidosis and Rare Disease</i> , 2016, 2, 23-26.	0.0	1
50	Mucopolysaccharidosis I (Scheie syndrome): A rare cause of severe aortic stenosis in a 31-year-old man. <i>Journal of the Formosan Medical Association</i> , 2015, 114, 1015-1016.	1.7	4
51	Gastric Residuals, Feeding Intolerance, and Necrotizing Enterocolitis in Preterm Infants. <i>Pediatrics and Neonatology</i> , 2015, 56, 136-137.	0.9	0
52	A modified liquid chromatography/tandem mass spectrometry method for predominant disaccharide units of urinary glycosaminoglycans in patients with mucopolysaccharidoses. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 135.	2.7	55
53	Overcoming the barriers to diagnosis of Morquio A syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 192.	2.7	21
54	Pulmonary Hemorrhage in Very-low-birth-weight Infants. <i>Pediatrics and Neonatology</i> , 2014, 55, 326-327.	0.9	7

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55	Cardiovascular abnormalities in Taiwanese patients with mucopolysaccharidosis. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 493-498.	1.1	26
56	Natural history and clinical assessment of Taiwanese patients with mucopolysaccharidosis IVA. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 21.	2.7	25
57	Characterization of pulmonary function impairments in patients with mucopolysaccharidoses—changes with age and treatment. <i>Pediatric Pulmonology</i> , 2014, 49, 277-284.	2.0	51
58	Assessment of hearing loss by pure-tone audiometry in patients with mucopolysaccharidoses. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 533-538.	1.1	36
59	Assessment of bone mineral density by dual energy x-ray absorptiometry in patients with mucopolysaccharidoses. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 71.	2.7	33
60	A pilot newborn screening program for Mucopolysaccharidosis type I in Taiwan. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 147.	2.7	69
61	Effects of enzyme replacement therapy for cardiac-type Fabry patients with a Chinese hotspot late-onset Fabry mutation (IVS4+919G>A). <i>BMJ Open</i> , 2013, 3, e003146.	1.9	16
62	Functional independence of Taiwanese children with VACTERL association. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 3101-3105.	1.2	7
63	Enzyme assay and clinical assessment in subjects with a Chinese hotspot late-onset Fabry mutation (IVS4+919G>A). <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 619-624.	3.6	24
64	Polysomnographic characteristics in patients with mucopolysaccharidoses. <i>Pediatric Pulmonology</i> , 2010, 45, 1205-1212.	2.0	47
65	Enzyme replacement therapy for mucopolysaccharidosis VI—experience in Taiwan. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 421-427.	3.6	59
66	Novel human pathological mutations. Gene symbol: COL1A2. Disease: Osteogenesis imperfecta III. <i>Human Genetics</i> , 2010, 127, 467.	3.8	0
67	Novel human pathological mutations. Gene symbol: OTC. Disease: ornithine transcarbamylase deficiency. <i>Human Genetics</i> , 2010, 127, 475.	3.8	0
68	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. <i>Human Genetics</i> , 2010, 127, 465.	3.8	0
69	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta IV. <i>Human Genetics</i> , 2010, 127, 466.	3.8	0
70	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. <i>Human Genetics</i> , 2010, 127, 464.	3.8	0
71	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta IV. <i>Human Genetics</i> , 2010, 127, 466.	3.8	2
72	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta I. <i>Human Genetics</i> , 2010, 127, 467.	3.8	0

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73	Novel human pathological mutations. Gene symbol: GAA. Disease: glycogen storage disease 2. Human Genetics, 2010, 127, 464.	3.8	1
74	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta IV. Human Genetics, 2010, 127, 467.	3.8	0
75	Novel human pathological mutations. Gene symbol: COL1A2. Disease: osteogenesis imperfecta I. Human Genetics, 2010, 127, 468.	3.8	7
76	Incidence of the mucopolysaccharidoses in Taiwan, 1984-2004. American Journal of Medical Genetics, Part A, 2009, 149A, 960-964.	1.2	145
77	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
78	Prader-Willi syndrome in Taiwan. Pediatrics International, 2007, 49, 375-379.	0.5	16
79	Genotype and phenotype in patients with Prader-Willi Syndrome in Taiwan. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 902-905.	1.5	23
80	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