Shuan-Pei Lin

List of Publications by Year
in descending order

110 papers	1,938 citations	236925 25 h-index	315739 38 g-index
113 all docs	113 docs citations	113 times ranked	2458 citing authors

#	Article	IF	Citations
1	Efficacy and safety of enzyme replacement therapy with BMN 110 (elosulfase alfa) for Morquio A syndrome (mucopolysaccharidosis IVA): a phase 3 randomised placeboâ€controlled study. Journal of Inherited Metabolic Disease, 2014, 37, 979-990.	3.6	176
2	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
3	A pilot newborn screening program for Mucopolysaccharidosis type I in Taiwan. Orphanet Journal of Rare Diseases, 2013, 8, 147.	2.7	69
4	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
5	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
6	Characterization of pulmonary function impairments in patients with mucopolysaccharidosesâ€"changes with age and treatment. Pediatric Pulmonology, 2014, 49, 277-284.	2.0	51
7	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
8	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
9	Frequency of associated anomalies in congenital hypoplasia of depressor anguli oris muscle: A study of 50 patients. American Journal of Medical Genetics Part A, 1997, 71, 215-218.	2.4	41
10	Loss of GPNMB Causes Autosomal-Recessive Amyloidosis Cutis Dyschromica in Humans. American Journal of Human Genetics, 2018, 102, 219-232.	6.2	41
11	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
12	Assessment of hearing loss by pure-tone audiometry in patients with mucopolysaccharidoses. Molecular Genetics and Metabolism, 2014, 111, 533-538.	1.1	36
13	Skipping of exon 12 as a consequence of a point mutation (1898 + 5G â†' T) in the cystic fibrosis transmembrane conductance regulator gene found in a consanguineous Chinese family. Clinical Genetics, 1995, 47, 125-132.	2.0	35
14	Smallest terminal deletion of the long arm of chromosome 2 in a mildly affected boy. American Journal of Medical Genetics Part A, 1992, 44, 500-502.	2.4	33
15	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
16	Mutation analysis of Wilson disease in Taiwan and description of six new mutations. Human Mutation, 1998, 12, 370-376.	2.5	31
17	Genotype and phenotype analysis of Taiwanese patients with osteogenesis imperfecta. Orphanet Journal of Rare Diseases, 2015, 10, 152.	2.7	30
18	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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19	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
20	Netherton syndrome: mutation analysis of two Taiwanese families. Archives of Dermatological Research, 2007, 299, 145-150.	1.9	28
21	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
22	Molecular analysis of survival motor neuron (SMN) and neuronal apoptosis inhibitory protein (NAIP) genes of spinal muscular atrophy patients and their parents. Human Genetics, 1997, 100, 577-581.	3.8	26
23	Effects of Anticoagulants in Amino Acid Analysis: Comparisons of Heparin, EDTA, and Sodium Citrate in Vacutainer Tubes for Plasma Preparation. Clinical Chemistry, 1998, 44, 1052-1056.	3.2	26
24	Prenatal diagnosis of inherited satellited non-acrocentric chromosomes. Prenatal Diagnosis, 2000, 20, 384-389.	2.3	26
25	Cardiovascular abnormalities in Taiwanese patients with mucopolysaccharidosis. Molecular Genetics and Metabolism, 2014, 111, 493-498.	1.1	26
26	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
27	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
28	Rapid detection of trisomy 21 by homologous gene quantitative PCR (HGQ-PCR). Human Genetics, 1997, 99, 364-367.	3.8	25
29	Natural history and clinical assessment of Taiwanese patients with mucopolysaccharidosis IVA. Orphanet Journal of Rare Diseases, 2014, 9, 21.	2.7	25
30	The relationships between urinary glycosaminoglycan levels and phenotypes of mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2018, 6, 982-992.	1.2	24
31	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
32	Overcoming the barriers to diagnosis of Morquio A syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 192.	2.7	21
33	Galloway-Mowat syndrome: a glomerular basement membrane disorder?. Pediatric Nephrology, 2001, 16, 653-657.	1.7	20
34	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
35	Galloway-Mowat syndrome in Taiwan: OSGEP mutation and unique clinical phenotype. Orphanet Journal of Rare Diseases, 2018, 13, 226.	2.7	16
36	Normalization of glycosaminoglycan-derived disaccharides detected by tandem mass spectrometry assay for the diagnosis of mucopolysaccharidosis. Scientific Reports, 2019, 9, 10755.	3.3	16

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37	Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€“2019). Orphanet Journal of Rare Diseases, 2020, 15, 314.	2.7	16
38	Familial transmission of recurrent 15q11.2 (BP1-BP2) microdeletion encompassing NIPA1 , NIPA2 , CYFIP1 , and TUBGCP5 associated with phenotypic variability in developmental, speech, and motor delay. Taiwanese Journal of Obstetrics and Gynecology, 2017, 56, 93-97.	1.3	15
39	Mutation identification and characterization of a Taiwanese patient with fucosidosis. Journal of Human Genetics, 2007, 52, 553-556.	2.3	14
40	Interphase FISH on uncultured amniocytes at repeat amniocentesis for rapid diagnosis of true mosaicism in a case of level II mosaicism involving trisomy 21 in a single colony from an in situ culture of amniocytes. Taiwanese Journal of Obstetrics and Gynecology, 2014, 53, 120-122.	1.3	14
41	Clinical observations on enzyme replacement therapy in patients with Fabry disease and the switch from agalsidase beta to agalsidase alfa. Journal of the Chinese Medical Association, 2014, 77, 190-197.	1.4	14
42	Cardiac characteristics and natural progression in Taiwanese patients with mucopolysaccharidosis III. Orphanet Journal of Rare Diseases, 2019, 14, 140.	2.7	14
43	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
44	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
45	PRENATAL DIAGNOSIS OF PARTIAL MONOSOMY 3p AND PARTIAL TRISOMY 2p IN A FETUS ASSOCIATED WITH SHORTENING OF THE LONG BONES AND A SINGLE UMBILICAL ARTERY. , 1996, 16, 270-275.		13
46	Epigenotype, genotype, and phenotype analysis of patients in Taiwan with Beckwithâ€“Wiedemann syndrome. Molecular Genetics and Metabolism, 2016, 119, 8-13.	1.1	13
47	Functional independence of Taiwanese children with Down syndrome. Developmental Medicine and Child Neurology, 2016, 58, 502-507.	2.1	13
48	Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e00617.	1.2	13
49	Comparison of free fatty acid content of human milk from Taiwanese mothers and infant formula. Taiwanese Journal of Obstetrics and Gynecology, 2013, 52, 527-533.	1.3	12
50	Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with Different Types of Mucopolysaccharidoses. Diagnostics, 2019, 9, 148.	2.6	11
51	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
52	Transcranial pulsed ultrasound facilitates brain uptake of laronidase in enzyme replacement therapy for Mucopolysaccharidosis type I disease. Orphanet Journal of Rare Diseases, 2017, 12, 109.	2.7	10
53	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
54	An At-Risk Population Screening Program for Mucopolysaccharidoses by Measuring Urinary Glycosaminoglycans in Taiwan. Diagnostics, 2019, 9, 140.	2.6	10

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55	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
56	Fabry Disease and the Effectiveness of Enzyme Replacement Therapy (ERT) in Left Ventricular Hypertrophy (LVH) Improvement: A Review and Meta-Analysis. International Journal of Medical Sciences, 2022, 19, 126-131.	2.5	10
57	Identifying the need for a multidisciplinary approach for early recognition of mucopolysaccharidosis VI (MPS VI). Molecular Genetics and Metabolism, 2015, 115, 41-47.	1.1	9
58	Molecular cytogenetic characterization of an inv dup(15) chromosome presenting as a small supernumerary marker chromosome associated with the inv dup(15) syndrome. Taiwanese Journal of Obstetrics and Gynecology, 2016, 55, 728-732.	1.3	9
59	Training in clinical genetics and genetic counseling in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 177-186.	1.6	9
60	Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. Pediatrics and Neonatology, 2019, 60, 342-343.	0.9	9
61	Cardiac Evaluation Using Two-Dimensional Speckle-Tracking Echocardiography and Conventional Echocardiography in Taiwanese Patients with Mucopolysaccharidoses. Diagnostics, 2020, 10, 62.	2.6	9
62	CVS-exposed limb deficiency defects with or without other birth defects: Presentation of six cases born during a period of nine years. American Journal of Medical Genetics Part A, 1996, 63, 447-453.	2.4	8
63	Mutations in Pseudohypoparathyroidism 1a and Pseudopseudohypoparathyroidism in Ethnic Chinese. PLoS ONE, 2014, 9, e90640.	2.5	8
64	A 13-year-old girl with 18p deletion syndrome presenting Turner syndrome-like clinical features of short stature, short webbed neck, low posterior hair line, puffy eyelids and increased carrying angle of the elbows. Taiwanese Journal of Obstetrics and Gynecology, 2018, 57, 583-587.	1.3	8
65	Array-CGH increased the diagnostic rate of developmental delay or intellectual disability in Taiwan. Pediatrics and Neonatology, 2019, 60, 453-460.	0.9	8
66	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
67	Array CGH characterization of an unbalanced X-autosome translocation associated with Xq27.2â€“qter deletion, 11q24.3â€“qter duplication and Xq22.3â€“q27.1 duplication in a girl with primary amenorrhea and mental retardation. Gene, 2014, 535, 88-92.	2.2	7
68	Pediatric sialendoscopy in Asians: A preliminary report. Journal of Pediatric Surgery, 2016, 51, 1684-1687.	1.6	7
69	Recurrent 2q13 microduplication encompassing MALL , NPHP1 , RGPD6 , and BUB1 associated with autism spectrum disorder, intellectual disability, and liver disorder. Taiwanese Journal of Obstetrics and Gynecology, 2017, 56, 98-101.	1.3	7
70	Bio-Plex immunoassay measuring the quantity of lysosomal N -acetylgalactosamine-6-sulfatase protein in dried blood spots for the screening of mucopolysaccharidosis IVA in newborn: a pilot study. BMJ Open, 2017, 7, e014410.	1.9	7
71	Long-term outcomes of enzyme replacement therapy for Taiwanese patients with Mucopolysaccharidosis I. Pediatrics and Neonatology, 2019, 60, 577-578.	0.9	7
72	Maternal serum screening abnormality in a fetus associated with arthrogryposis multiplex congenita and amyoplasia. , 1997, 17, 1187-1188.		6

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73	Prenatal prediction of spinal muscular atrophy in Chinese. Prenatal Diagnosis, 1999, 19, 657-661.	2.3	6
74	A novel in-frame deletion mutation (c106-111del) identified in a Taiwan Chinese patient with type IVA mucopolysaccharidosis. Human Mutation, 2001, 18, 254-254.	2.5	6
75	Molecular cytogenetic characterization and prenatal diagnosis of familial Xp22.33 microdeletion encompassing short stature homeobox gene in a male fetus with a favorable outcome. Taiwanese Journal of Obstetrics and Gynecology, 2017, 56, 264-267.	1.3	6
76	Functional independence of Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e790.	1.2	6
77	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
78	Molecular cytogenetic characterization of mosaicism for a small supernumerary marker chromosome derived from chromosome 8 or r(8)(::p11.22â†'q11.21::) in an 18-year-old female with short stature, obesity, attention deficit hyperactivity disorder, and intellectual disability. Taiwanese Journal of Obstetrics and Gynecology, 2016, 55, 856-860.	1.3	5
79	Awareness of Mucopolysaccharidosis in an Otorhinolaryngologic Clinic. Pediatrics and Neonatology, 2017, 58, 198-199.	0.9	5
80	Assessing the impact of the five senses on quality of life in mucopolysaccharidoses. Orphanet Journal of Rare Diseases, 2020, 15, 97.	2.7	5
81	Otorhinolaryngological Management in Taiwanese Patients with Mucopolysaccharidoses. International Journal of Medical Sciences, 2021, 18, 3373-3379.	2.5	5
82	Aortic Root Dilatation in Taiwanese Patients with Mucopolysaccharidoses and the Long-Term Effects of Enzyme Replacement Therapy. Diagnostics, 2021, 11, 16.	2.6	5
83	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
84	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
85	Pregnancy with de novo 9q34.3 microdeletion and Kleefstra syndrome in the fetus may be associated with an abnormal maternal serum screening result. Taiwanese Journal of Obstetrics and Gynecology, 2015, 54, 450-451.	1.3	4
86	Molecular cytogenetic characterization of mosaicism for a small supernumerary marker chromosome derived from chromosome 8 or r(8)(::p12â†'q13.1::) associated with phenotypic abnormalities. Taiwanese Journal of Obstetrics and Gynecology, 2016, 55, 852-855.	1.3	4
87	A Truncating De Novo Point Mutation in a Young Infant with Severe Menkes Disease. Pediatrics and Neonatology, 2017, 58, 89-92.	0.9	4
88	Pfeiffer syndrome with FGFR2 C342R mutation presenting extreme proptosis, craniosynostosis, hearing loss, ventriculomegaly, broad great toes and thumbs, maxillary hypoplasia, and laryngomalacia. Taiwanese Journal of Obstetrics and Gynecology, 2017, 56, 412-414.	1.3	4
89	Prenatal diagnosis of partial trisomy 12 and partial trisomy 21 due to a 3:1 segregation of maternal reciprocal translocation t(12;21) (p13.3;q21). , 1997, 17, 675-680.		3
90	Concomitant chyloperitoneum and omental cysts presenting as fetal ascites with intra-abdominal cysts on prenatal ultrasound. , 1998, 18, 984-986.		3

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91	Identification of a novel missense mutation (T16A) in the glucose-6-phosphatase gene in a Taiwan Chinese patient with glycogen storage disease Ia (Von Gierke disease). , 2000, 15, 390-390.		3
92	A 1.37-Mb 12p11.22â€“p11.21 deletion coincident with a 367-kb 22q11.2 duplication detected by array comparative genomic hybridization in an adolescent girl with autism and difficulty in self-care of menstruation. Taiwanese Journal of Obstetrics and Gynecology, 2014, 53, 74-78.	1.3	3
93	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
94	The first SHORT syndrome in a Taiwanese boy: A case report and review of the literature. Molecular Genetics and Metabolism Reports, 2021, 27, 100768.	1.1	3
95	Airway Management of the Deformed Trachea Using T-Tube Stents in Patients with Mucopolysaccharidosis Type IVA. Annals of Otology, Rhinology and Laryngology, 2022, 131, 562-566.	1.1	3
96	Incidence and treatment of adult femoral fractures with osteogenesis imperfecta: An analysis of a center of 72 patients in Taiwan. International Journal of Medical Sciences, 2021, 18, 1240-1246.	2.5	3
97	Clinical ocular manifestations of Taiwanese patients with mucopolysaccharidoses VI (Maroteauxâ€“Lamy syndrome). Taiwan Journal of Ophthalmology, 2019, 9, 194.	0.7	3
98	Clinical Utility of Elosulfase Alfa in the Treatment of Morquio A Syndrome.. Drug Design, Development and Therapy, 2022, 16, 143-154.	4.3	3
99	Mucopolysaccharidosis Type IIâ€”An Unexpected â€œ3 in 1â€•Family. Pediatrics and Neonatology, 2016, 57, 359-360.	0.9	2
100	Functional independence of Taiwanese children with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1309-1314.	1.2	2
101	Awareness of attenuated mucopolysaccharidoses in a pediatric orthopedic clinic. Pediatrics and Neonatology, 2019, 60, 100-101.	0.9	2
102	Tetrasomy of 11q13.4-q14.3 due to an intrachromosomal triplication associated with paternal uniparental isodisomy for 11q14.3-qter, intrauterine growth restriction, developmental delay, corpus callosum dysgenesis, microcephaly, congenital heart defects and facial dysmorphism. Taiwanese Journal of Obstetrics and Gynecology, 2021, 60, 169-172.	1.3	2
103	Array comparative genomic hybridization characterization of a 3.3-Mb 17p13.3-p13.2 deletion encompassing YWHAE, CRK, HIC1 and PAFAH1B1 in an 8-year-old girl with Miller-Dieker lissencephaly syndrome, congenital heart defects, growth restriction and developmental delay. Taiwanese Journal of Obstetrics and Gynecology, 2018, 57, 765-768.	1.3	1
104	A 17-year-old boy with Klinefelter syndrome presenting Marfan syndrome-like clinical features of tall stature, scoliosis, arachnodactyly and subluxation of bilateral elbow joints. Taiwanese Journal of Obstetrics and Gynecology, 2018, 57, 608-610.	1.3	1
105	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
106	Wiedemannâ€“Steiner Syndrome with a Pathogenic Variant in KMT2A from Taiwan. Children, 2021, 8, 952.	1.5	1
107	Long-Term Cardiovascular Findings in Williams Syndrome: A Single Medical Center Experience in Taiwan. Journal of Personalized Medicine, 2022, 12, 817.	2.5	1
108	MUTATIONAL ANALYSIS OF PTPN11 AND KRAS GENES IN TAIWANESE CHILDREN WITH NOONAN SYNDROME. Pediatrics, 2008, 121, S116-S117.	2.1	0

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109	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
110	Epigenotype, Genotype, and Phenotype Analysis of Taiwanese Patients with Silverâ€“Russell Syndrome. Journal of Personalized Medicine, 2021, 11, 1197.	2.5	0