# Wuh-Liang Hwu

List of Publications by Year
in descending order

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

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | 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 |
| 2 | Long-term efficacy and safety of eladocagene exuparvovec in patients with AADC deficiency. Molecular Therapy, 2022, 30, 509-518. | 8.2 | 58 |
| 3 | Outcome of Later-Onset Pompe Disease Identified Through Newborn Screening. Journal of Pediatrics, 2022, 244, 139-147.e2. | 1.8 | 10 |
| 4 | Advanced therapeutic strategy for hereditary neuromuscular diseases. Molecular Therapy, 2022, 30, 12-13. | 8.2 | 1 |
| 5 | 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 |
| 6 | Comparison of GATK and DeepVariant by trio sequencing. Scientific Reports, 2022, 12, 1809. | 3.3 | 26 |
| 7 | 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 |
| 8 | Duchenne muscular dystrophy newborn screening: the first 50,000 newborns screened in Taiwan. Neurological Sciences, 2022, 43, 4563-4566. | 1.9 | 13 |
| 9 | 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 |
| 10 | Thymidine Kinase 2 Deficiencyâ€“Induced Adult-Onset Ptosis and Proximal Weakness. Neurology: Clinical Practice, 2021, 11, e379-e382. | 1.6 | 0 |
| 11 | 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 |
| 12 | 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 |
| 13 | Rapid Trio Exome Sequencing for Autosomal Recessive Renal Tubular Dysgenesis in Recurrent Oligohydramnios. Frontiers in Genetics, 2021, 12, 606970. | 2.3 | 1 |
| 14 | Gene therapy in the putamen for curing AADC deficiency and Parkinson's disease. EMBO Molecular Medicine, 2021, 13, e14712. | 6.9 | 17 |
| 15 | 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 |
| 16 | Reduced Immunogenicity of Intraparenchymal Delivery of Adeno-Associated Virus Serotype 2 Vectors: Brief Overview. Current Gene Therapy, 2021, 21, . | 2.0 | 2 |
| 17 | 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 |
| 18 | RNA-seq of peripheral blood mononuclear cells of congenital generalized lipodystrophy type 2 patients. Scientific Data, 2021, 8, 265. | 5.3 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | A novel deep intronic variant strongly associates with Alkaptonuria. Npj Genomic Medicine, 2021, 6, 89. | 3.8 | 9 |
| 20 | Molecular Analysis of Vietnamese Patients with Mucopolysaccharidosis Type I. Life, 2021, 11, 1162. | 2.4 | 0 |
| 21 | Novel Compound Heterozygous Variants in TBCD Gene Associated with Infantile Neurodegenerative Encephalopathy. Children, 2021, 8, 1140. | 1.5 | 3 |
| 22 | Thyroid disorders in Taiwanese children with Down syndrome: The experience of a single medical center. Journal of the Formosan Medical Association, 2020, 119, 345-349. | 1.7 | 3 |
| 23 | 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 |
| 24 | Analysis of nondegraded and degraded DNA mixtures of close relatives using massively parallel sequencing. Legal Medicine, 2020, 42, 101631. | 1.3 | 8 |
| 25 | 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 |
| 26 | 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 |
| 27 | Composite Scores of Plasma Tau and Î²-Amyloids Correlate with Dementia in Down Syndrome. ACS Chemical Neuroscience, 2020, 11, 191-196. | 3.5 | 4 |
| 28 | Ultrastructural and diffusion tensor imaging studies reveal axon abnormalities in Pompe disease mice. Scientific Reports, 2020, 10, 20239. | 3.3 | 1 |
| 29 | Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985â€“2019). Orphanet Journal of Rare Diseases, 2020, 15, 314. | 2.7 | 16 |
| 30 | Towards a reference genome that captures global genetic diversity. Nature Communications, 2020, 11, 5482. | 12.8 | 34 |
| 31 | Diversity in heritable disorders of connective tissue at a single center. Connective Tissue Research, 2020, 62, 1-6. | 2.3 | 5 |
| 32 | Frequency and spectrum of actionable pathogenic secondary findings in Taiwanese exomes. Molecular Genetics & Genomic Medicine, 2020, 8, e1455. | 1.2 | 14 |
| 33 | Dietary intake and nutritional status of patients with phenylketonuria in Taiwan. Scientific Reports, 2020, 10, 14537. | 3.3 | 5 |
| 34 | 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 |
| 35 | 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 |
| 36 | Complications of erythropoietin in navigated brain gene therapy: A case report. Interdisciplinary Neurosurgery: Advanced Techniques and Case Management, 2020, 21, 100698. | 0.3 | 2 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | De novo mutation and skewed Xâ€inactivation in girl with BCAP31 â€related syndrome. Human Mutation, 2020, 41, 1775-1782. | 2.5 | 3 |
| 40 | 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 |
| 41 | Development of Newborn Screening for Pompe Disease. International Journal of Neonatal Screening, 2020, 6, 5. | 3.2 | 2 |
| 42 | The Timely Needs for Infantile Onset Pompe Disease Newborn Screeningâ€"Practice in Taiwan. International Journal of Neonatal Screening, 2020, 6, 30. | 3.2 | 1 |
| 43 | Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18. | 3.8 | 29 |
| 44 | Relationships among Height, Weight, Body Mass Index, and Age in Taiwanese Children with Different Types of Mucopolysaccharidoses. Diagnostics, 2019, 9, 148. | 2.6 | 11 |
| 45 | Functional independence of Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e790. | 1.2 | 6 |
| 46 | Heterogeneous nonataxic phenotypes of spinocerebellar ataxia in a Taiwanese population. Brain and Behavior, 2019, 9, e01414. | 2.2 | 10 |
| 47 | Genotypic and phenotypic correlations of biotinidase deficiency in the Chinese population. Orphanet Journal of Rare Diseases, 2019, 14, 6. | 2.7 | 15 |
| 48 | 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 |
| 49 | Clinical features of Pompe disease with motor neuronopathy. Neuromuscular Disorders, 2019, 29, 903-906. | 0.6 | 7 |
| 50 | Fabry disease cardiac variant IVS4+919 G>A is associated with multiple cardiac gene variants in patients with severe cardiomyopathy and fatal arrhythmia. Genetics in Medicine, 2019, 21, 1890-1891. | 2.4 | 3 |
| 51 | 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 |
| 52 | 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 |
| 53 | High incidence of coâ€existing GLA variants and stroke susceptibility. European Journal of Neurology, 2019, 26, e70-e70. | 3.3 | 0 |
| 54 | Gene therapy improves brain white matter in aromatic lâ€amino acid decarboxylase deficiency. Annals of Neurology, 2019, 85, 644-652. | 5.3 | 30 |

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|---|---|---|---|
| 55 | Ophthalmologic manifestations in Taiwanese patients with mucopolysaccharidoses. Molecular Genetics & Genomic Medicine, 2019, 7, e00617. | 1.2 | 13 |
| 56 | 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 |
| 57 | 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 |
| 58 | 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 |
| 59 | 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 |
| 60 | Next-generation sequencing identifies TRPV4-related skeletal dysplasia in a boy with progressive bowlegs. Pediatrics and Neonatology, 2019, 60, 102-104. | 0.9 | 3 |
| 61 | 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 |
| 62 | Electrical Abnormalities in Dopaminergic Neurons of the Substantia Nigra in Mice With an Aromatic L-Amino Acid Decarboxylase Deficiency. Frontiers in Cellular Neuroscience, 2019, 13, 9. | 3.7 | 3 |
| 63 | Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). Molecular Genetics and Metabolism, 2019, 126, 98-105. | 1.1 | 56 |
| 64 | 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 |
| 65 | Long-term effects of enzyme replacement therapy for Taiwanese patients with mucopolysaccharidosis IVA. Pediatrics and Neonatology, 2019, 60, 342-343. | 0.9 | 9 |
| 66 | Congenital generalized lipodystrophy in Taiwan. Journal of the Formosan Medical Association, 2019, 118, 142-147. | 1.7 | 11 |
| 67 | Newborn screening: Taiwanese experience. Annals of Translational Medicine, 2019, 7, 281-281. | 1.7 | 23 |
| 68 | 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 |
| 69 | Comprehensive human leukocyte antigen genotyping of patients with type 1 diabetes mellitus in Taiwan. Pediatric Diabetes, 2018, 19, 699-706. | 2.9 | 7 |
| 70 | SHOX deficiency in short Taiwanese children: A single-center experience. Journal of the Formosan Medical Association, 2018, 117, 909-914. | 1.7 | 6 |
| 71 | 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 |
| 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 | 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 |
| 74 | 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 |
| 75 | 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 |
| 76 | 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 |
| 77 | 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 |
| 78 | 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 |
| 79 | 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 |
| 80 | Gene therapy with modified U1 small nuclear RNA. Expert Review of Endocrinology and Metabolism, 2017, 12, 171-175. | 2.4 | 5 |
| 81 | 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 |
| 82 | 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 |
| 83 | Natural History of Aromatic l-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Reports, 2017, 40, 1-6. | 1.5 | 26 |
| 84 | 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 |
| 85 | Presymptomatic Diagnosis of Spinal Muscular Atrophy Through Newborn Screening. Journal of Pediatrics, 2017, 190, 124-129.e1. | 1.8 | 113 |
| 86 | A Review of Biomarkers for Alzheimerâ€™s Disease in Down Syndrome. Neurology and Therapy, 2017, 6, 69-81. | 3.2 | 19 |
| 87 | Introduction to the Newborn Screening, Diagnosis, and Treatment for Pompe Disease Guidance Supplement. Pediatrics, 2017, 140, S1-S3. | 2.1 | 11 |
| 88 | Management of Confirmed Newborn-Screened Patients With Pompe Disease Across the Disease Spectrum. Pediatrics, 2017, 140, S24-S45. | 2.1 | 43 |
| 89 | 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 |
| 90 | Newborn Screening for Severe Combined Immunodeficiency in Taiwan. International Journal of Neonatal Screening, 2017, 3, 16. | 3.2 | 38 |

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|---|---|---|---|
| 91 | 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 |
| 92 | 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 |
| 93 | 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 |
| 94 | 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 |
| 95 | 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 |
| 96 | Mutation-adapted U1 snRNA corrects a splicing error of the dopa decarboxylase gene. Human Molecular Genetics, 2016, 25, ddw323. | 2.9 | 18 |
| 97 | 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 |
| 98 | Integrated care for Down syndrome. Congenital Anomalies (discontinued), 2016, 56, 104-106. | 0.6 | 2 |
| 99 | Advances in newborn screening for Pompe disease and resulting clinical outcomes. Expert Opinion on Orphan Drugs, 2016, 4, 21-29. | 0.8 | 0 |
| 100 | Hypothermia improves disease manifestations in SMA mice via SMN augmentation. Human Molecular Genetics, 2016, 25, 631-641. | 2.9 | 5 |
| 101 | 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 |
| 102 | Long-term outcome for Down syndrome patients with hematopoietic disorders. Journal of the Formosan Medical Association, 2016, 115, 94-99. | 1.7 | 5 |
| 103 | Blood Beta-Amyloid and Tau in Down Syndrome: A Comparison with Alzheimerâ€™s Disease. Frontiers in Aging Neuroscience, 2016, 8, 316. | 3.4 | 44 |
| 104 | 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 |
| 105 | Congenital Malformations in Newbornsâ€”A Challenge Unmet for Decades. Pediatrics and Neonatology, 2015, 56, 5-6. | 0.9 | 0 |
| 106 | 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 |
| 107 | Benefits of Neuronal Preferential Systemic Gene Therapy for Neurotransmitter Deficiency. Molecular Therapy, 2015, 23, 1572-1581. | 8.2 | 25 |
| 108 | Mortality, disability, and intensive care in patients with mitochondrial 3243A>G mutation. Intensive Care Medicine, 2015, 41, 1493-1495. | 8.2 | 1 |

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|---|---|---|---|
| 109 | 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 |
| 110 | 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 |
| 111 | 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 |
| 112 | Hypertrophic Cardiomyopathy in Pompe Disease Is Not Limited to the Classic Infantile-Onset Phenotype. JIMD Reports, 2014, 17, 71-75. | 1.5 | 30 |
| 113 | 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 |
| 114 | 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 |
| 115 | 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 |
| 116 | Parental discussion of G6PD deficiency and child health: implications for clinical practice. Archives of Disease in Childhood, 2014, 99, 251-255. | 1.9 | 1 |
| 117 | Cyclic Pamidronate Infusion for Neonatal-onset Osteogenesis Imperfecta. Pediatrics and Neonatology, 2014, 55, 306-311. | 0.9 | 11 |
| 118 | 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 |
| 119 | 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 |
| 120 | Natural history and clinical assessment of Taiwanese patients with mucopolysaccharidosis IVA. Orphanet Journal of Rare Diseases, 2014, 9, 21. | 2.7 | 25 |
| 121 | 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 |
| 122 | Outcome of early-treated type III Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2014, 53, 105-109. | 1.4 | 22 |
| 123 | | | |

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|---|---|---|---|
| 127 | Lysoâ€globotriaosylsphingosine (lysoâ€Gb<sub>3</sub>) 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 |
| 128 | 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 |
| 129 | Pompe Disease: Early Diagnosis and Early Treatment Make a Difference. Pediatrics and Neonatology, 2013, 54, 219-227. | 0.9 | 135 |
| 130 | AADC Deficiency. Advances in Pharmacology, 2013, 68, 273-284. | 2.0 | 10 |
| 131 | Carnitine Uptake Defect (Primary Carnitine Deficiency): Risk in Genotype-Phenotype Correlation. Human Mutation, 2013, 34, 655-655. | 2.5 | 13 |
| 132 | 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 |
| 133 | Lung toxicity of hydroxypropyl-Î²-cyclodextrin infusion. Molecular Genetics and Metabolism, 2013, 109, 231-232. | 1.1 | 27 |
| 134 | Diagnosing mucopolysaccharidosis IVA. Journal of Inherited Metabolic Disease, 2013, 36, 293-307. | 3.6 | 77 |
| 135 | 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 |
| 136 | Fatty Acid Oxidation Disorders in a Chinese Population in Taiwan. JIMD Reports, 2013, 11, 165-172. | 1.5 | 26 |
| 137 | Analysis of Lyso-Globotriaosylsphingosine in Dried Blood Spots. Annals of Laboratory Medicine, 2013, 33, 274-278. | 2.5 | 29 |
| 138 | Myostatin and Insulin-Like Growth Factor I: Potential Therapeutic Biomarkers for Pompe Disease. PLoS ONE, 2013, 8, e71900. | 2.5 | 15 |
| 139 | Web-Based Newborn Screening System for Metabolic Diseases: Machine Learning Versus Clinicians. Journal of Medical Internet Research, 2013, 15, e98. | 4.3 | 19 |
| 140 | Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655. | 2.4 | 117 |
| 141 | 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 |
| 142 | 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 |
| 143 | Newborn Screening for Phenylketonuria: Machine Learning vs Clinicians. , 2012, , . | | 1 |
| 144 | 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 |

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|---|---|---|---|
| 145 | Current diagnosis and management of mucopolysaccharidosis VI in the Asia-Pacific region. Molecular Genetics and Metabolism, 2012, 107, 136-144. | 1.1 | 6 |
| 146 | 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 |
| 147 | Early Pathologic Changes and Responses to Treatment in Patients With Later-Onset Pompe Disease. Pediatric Neurology, 2012, 46, 168-171. | 2.1 | 21 |
| 148 | Algorithm for Pompe disease newborn screening: Results from the Taiwan screening program. Molecular Genetics and Metabolism, 2012, 106, 281-286. | 1.1 | 72 |
| 149 | An acidic oligopeptide displayed on AAV2 improves axial muscle tropism after systemic delivery. Genetic Vaccines and Therapy, 2012, 10, 3. | 1.5 | 4 |
| 150 | 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 |
| 151 | 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 |
| 152 | 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 |
| 153 | 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 |
| 154 | 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 |
| 155 | Gene Therapy for Aromatic <scp>l</scp> -Amino Acid Decarboxylase Deficiency. Science Translational Medicine, 2012, 4, 134ra61. | 12.4 | 195 |
| 156 | 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 |
| 157 | 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 |
| 158 | PSORS2 Is Due to Mutations in CARD14. American Journal of Human Genetics, 2012, 90, 784-795. | 6.2 | 365 |
| 159 | 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 |
| 160 | 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 |
| 161 | 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 |
| 162 | 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 |

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