

# Paul Gissen

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7043154/publications.pdf>

Version: 2024-02-01

172  
papers

10,575  
citations

36203

51  
h-index

35952

97  
g-index

222  
all docs

222  
docs citations

222  
times ranked

15289  
citing authors

#	ARTICLE	IF	CITATIONS
1	Efficacy and safety of N-acetyl-L-leucine in Niemann-Pick disease type C. <i>Journal of Neurology</i> , 2022, 269, 1651-1662.	1.8	24
2	A survival analysis of ventricular access devices for delivery of cerliponase alfa. <i>Journal of Neurosurgery: Pediatrics</i> , 2022, 29, 115-121.	0.8	1
3	Rare missense variants in Tropomyosin-4 (TPM4) are associated with platelet dysfunction, cytoskeletal defects, and excessive bleeding. <i>Journal of Thrombosis and Haemostasis</i> , 2022, 20, 478-485.	1.9	3
4	Clinical disease characteristics of patients with Niemann-Pick Disease Type C: findings from the International Niemann-Pick Disease Registry (INPDR). <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 51.	1.2	14
5	Gene therapies targeting the liver. <i>Journal of Hepatology</i> , 2021, 74, 235-236.	1.8	9
6	Clinical Pharmacokinetics and Pharmacodynamics of Cerliponase Alfa, Enzyme Replacement Therapy for CLN2 Disease by Intracerebroventricular Administration. <i>Clinical and Translational Science</i> , 2021, 14, 635-644.	1.5	4
7	Urine proteomics analysis of patients with neuronal ceroid lipofuscinoses. <i>iScience</i> , 2021, 24, 102020.	1.9	12
8	Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 185.	1.2	17
9	Gene Therapy for Lysosomal Storage Disorders: Ongoing Studies and Clinical Development. <i>Biomolecules</i> , 2021, 11, 611.	1.8	27
10	Investigating health-related quality of life in rare diseases: a case study in utility value determination for patients with CLN2 disease (neuronal ceroid lipofuscinosis type 2). <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 217.	1.2	8
11	Clinical applications for exosomes: Are we there yet?. <i>British Journal of Pharmacology</i> , 2021, 178, 2375-2392.	2.7	57
12	Diagnosing Mitochondrial Disorders Remains Challenging in the Omics Era. <i>Neurology: Genetics</i> , 2021, 7, e597.	0.9	13
13	An ERG and OCT study of neuronal ceroid lipofuscinosis CLN2 Battens retinopathy. <i>Eye</i> , 2021, 35, 2438-2448.	1.1	6
14	Cerebrospinal fluid neurofilament light levels in CLN2 disease patients treated with enzyme replacement therapy normalise after two years on treatment. <i>F1000Research</i> , 2021, 10, 614.	0.8	4
15	Safety and efficacy of an engineered hepatotropic AAV gene therapy for ornithine transcarbamylase deficiency in cynomolgus monkeys. <i>Molecular Therapy - Methods and Clinical Development</i> , 2021, 23, 135-146.	1.8	21
16	A Case Series on Genotype and Outcome of Liver Transplantation in Children with Niemann-Pick Disease Type C. <i>Children</i> , 2021, 8, 819.	0.6	1
17	Rab35 controls formation of luminal projections required for bile canalicular morphogenesis. <i>Journal of Cell Biology</i> , 2021, 220, .	2.3	0
18	Cerliponase Alfa for the Treatment of Atypical Phenotypes of CLN2 Disease: A Retrospective Case Series. <i>Journal of Child Neurology</i> , 2021, 36, 468-474.	0.7	10

#	ARTICLE	IF	CITATIONS
19	The Genetics of Inherited Cholestatic Disorders in Neonates and Infants: Evolving Challenges. <i>Genes</i> , 2021, 12, 1837.	1.0	7
20	International consensus on clinical severity scale use in evaluating Niemann-Pick disease Type C in paediatric and adult patients: results from a Delphi Study. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 482.	1.2	8
21	Free urinary glycosylated hydroxylysine as an indicator of altered collagen degradation in the mucopolysaccharidoses. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 309-317.	1.7	10
22	Gene therapy for global brain diseases: one small step for mice, one giant leap for humans. <i>Brain</i> , 2020, 143, 1964-1966.	3.7	1
23	An In-Vitro Whole-Organ Liver Engineering for Testing of Genetic Therapies. <i>iScience</i> , 2020, 23, 101808.	1.9	8
24	Clinical and Molecular Features of Early Infantile Niemann Pick Type C Disease. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5059.	1.8	21
25	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. <i>Brain Communications</i> , 2020, 2, fcaa178.	1.5	17
26	Loss-of-Function Variants in <i>HOPS</i> Complex Genes <i>VPS16</i> and <i>VPS41</i> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , 2020, 88, 867-877.	2.8	70
27	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	13.7	148
28	Myostatin inhibition in combination with antisense oligonucleotide therapy improves outcomes in spinal muscular atrophy. <i>Journal of Cachexia, Sarcopenia and Muscle</i> , 2020, 11, 768-782.	2.9	49
29	Alpha synuclein aggregation drives ferroptosis: an interplay of iron, calcium and lipid peroxidation. <i>Cell Death and Differentiation</i> , 2020, 27, 2781-2796.	5.0	142
30	G476(P)-Aminoacylase 1 deficiency, a clinical prospect. , 2020, , .		0
31	Age-Related Seroprevalence of Antibodies Against AAV-LK03 in a UK Population Cohort. <i>Human Gene Therapy</i> , 2019, 30, 79-87.	1.4	51
32	Seeding Induced Pluripotent Stem Cell-Derived Neurons onto 384-Well Plates. <i>Methods in Molecular Biology</i> , 2019, 1994, 159-164.	0.4	3
33	High-Content Analysis of Mitochondrial Function in iPSC-Derived Neurons. <i>Methods in Molecular Biology</i> , 2019, 1994, 175-184.	0.4	4
34	High-Content Autophagy Analysis in iPSC-Derived Neurons Using Immunofluorescence. <i>Methods in Molecular Biology</i> , 2019, 1994, 165-174.	0.4	1
35	Measurement of Bile Acids as a Marker of the Functionality of iPSC-Derived Hepatocytes. <i>Methods in Molecular Biology</i> , 2019, 1994, 141-147.	0.4	0
36	Surveillance for variant CJD: should more children with neurodegenerative diseases have autopsies?. <i>Archives of Disease in Childhood</i> , 2019, 104, 360-365.	1.0	7

#	ARTICLE	IF	CITATIONS
37	One centre's experience of sedation regimes for ICV and IT drug delivery in early phase paediatric clinical trials. <i>Molecular Genetics and Metabolism</i> , 2019, 126, S25.	0.5	1
38	Using stem cell-derived neurons in drug screening for neurological diseases. <i>Neurobiology of Aging</i> , 2019, 78, 130-141.	1.5	38
39	Argininosuccinic aciduria: Recent pathophysiological insights and therapeutic prospects. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 1147-1161.	1.7	35
40	Persistent treatment effect of cerliponase alfa in children with CLN2 disease: A 3 year update from an ongoing multicenter extension study. <i>Molecular Genetics and Metabolism</i> , 2019, 126, S133.	0.5	2
41	Urea Cycle Related Amino Acids Measured in Dried Bloodspots Enable Long-Term In Vivo Monitoring and Therapeutic Adjustment. <i>Metabolites</i> , 2019, 9, 275.	1.3	4
42	Phase I/II Trial of Liver-derived Mesenchymal Stem Cells in Pediatric Liver-based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liver-derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients. <i>Transplantation</i> , 2019, 103, 1903-1915.	0.5	47
43	An Optimized Method for the Proteomic Analysis of Low Volumes of Cell Culture Media and the Secretome: The Application and the Demonstration of Altered Protein Expression in iPSC-Derived Neuronal Cell Lines from Parkinson's Disease Patients. <i>Journal of Proteome Research</i> , 2019, 18, 1198-1207.	1.8	2
44	Preparation of iPSCs for Targeted Proteomic Analysis. <i>Methods in Molecular Biology</i> , 2019, 1994, 131-139.	0.4	1
45	Persistent Treatment Effect of Cerliponase Alfa in Children with CLN2 Disease: A 3 Year Update from an Ongoing Multicenter Extension Study. , 2019, 50, .		0
46	Study of Intraventricular Cerliponase Alfa for CLN2 Disease. <i>New England Journal of Medicine</i> , 2018, 378, 1898-1907.	13.9	348
47	VPS33B and VIPAR are essential for epidermal lamellar body biogenesis and function. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 1609-1621.	1.8	37
48	Lamin and the heart. <i>Heart</i> , 2018, 104, 468-479.	1.2	113
49	Oculomotor abnormalities in children with Niemann-Pick type C. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 159-168.	0.5	14
50	Reproducibility of Molecular Phenotypes after Long-Term Differentiation to Human iPSC-Derived Neurons: A Multi-Site Omics Study. <i>Stem Cell Reports</i> , 2018, 11, 897-911.	2.3	135
51	Argininosuccinic aciduria fosters neuronal nitrosative stress reversed by Asl gene transfer. <i>Nature Communications</i> , 2018, 9, 3505.	5.8	34
52	Spectrum of movement disorders and neurotransmitter abnormalities in paediatric <i>POLG</i> disease. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 1275-1283.	1.7	12
53	Markers of cognitive function in individuals with metabolic disease: Morquio syndrome and tyrosinemia type III. <i>Cognitive Neuropsychology</i> , 2018, 35, 120-147.	0.4	8
54	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	2.6	36

#	ARTICLE	IF	CITATIONS
55	Severe renal Fanconi and management strategies in Arthrogryposis-Renal dysfunction-Cholestasis syndrome: a case report. <i>BMC Nephrology</i> , 2018, 19, 144.	0.8	15
56	Consensus clinical management guidelines for Niemann-Pick disease type C. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 50.	1.2	200
57	Molecular architecture of the multifunctional collagen lysyl hydroxylase and glycosyltransferase LH3. <i>Nature Communications</i> , 2018, 9, 3163.	5.8	46
58	A single cell high content assay detects mitochondrial dysfunction in iPSC-derived neurons with mutations in SNCA. <i>Scientific Reports</i> , 2018, 8, 9033.	1.6	50
59	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 3-18.	2.6	46
60	Î±-synuclein oligomers interact with ATP synthase and open the permeability transition pore in Parkinson's disease. <i>Nature Communications</i> , 2018, 9, 2293.	5.8	351
61	Vps33b is crucial for structural and functional hepatocyte polarity. <i>Journal of Hepatology</i> , 2017, 66, 1001-1011.	1.8	51
62	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017, 100, 334-342.	2.6	26
63	Multiplex High-Throughput Targeted Proteomic Assay To Identify Induced Pluripotent Stem Cells. <i>Analytical Chemistry</i> , 2017, 89, 2440-2448.	3.2	15
64	Delivering efficient liver-directed AAV-mediated gene therapy. <i>Gene Therapy</i> , 2017, 24, 263-264.	2.3	11
65	Expanding the phenotype in argininosuccinic aciduria: need for new therapies. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 357-368.	1.7	55
66	The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. <i>Genetics in Medicine</i> , 2017, 19, 1217-1225.	1.1	45
67	Gene therapy for monogenic liver diseases: clinical successes, current challenges and future prospects. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 497-517.	1.7	89
68	Adjuvant oral salbutamol in treatment of juvenile Pompe disease: novel outcome assessment tool and initial report one-year efficacy in single case. <i>Molecular Genetics and Metabolism</i> , 2017, 120, S142.	0.5	0
69	Î±-Synuclein binds to the ER-mitochondria tethering protein VAPB to disrupt Ca <sup>2+</sup> homeostasis and mitochondrial ATP production. <i>Acta Neuropathologica</i> , 2017, 134, 129-149.	3.9	262
70	Direct Modulation of the Mitochondrial Permeability Transition Pore by Oligomeric Alpha-Synuclein Causes Toxicity in PD. <i>Biophysical Journal</i> , 2017, 112, 440a.	0.2	0
71	Autosomal Recessive Keratoderma-Ichthyosis-Deafness (ARKID) Syndrome Is Caused by VPS33B Mutations Affecting Rab Protein Interaction and Collagen Modification. <i>Journal of Investigative Dermatology</i> , 2017, 137, 845-854.	0.3	37
72	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	2.6	343

#	ARTICLE	IF	CITATIONS
73	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017, 49, 223-237.	9.4	186
74	177 VPS33B mutations cause ARKID syndrome affecting Rab protein interaction, collagen modification and epidermal structure. <i>Journal of Investigative Dermatology</i> , 2017, 137, S223.	0.3	0
75	Hematopoietic Stem-Cell Gene Therapy for Cerebral Adrenoleukodystrophy. <i>New England Journal of Medicine</i> , 2017, 377, 1630-1638.	13.9	412
76	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
77	Intracerebroventricular cerliponase alfa for children with CLN2 disease: Interim results from an ongoing phase 2 extension study. <i>European Journal of Paediatric Neurology</i> , 2017, 21, e21.	0.7	2
78	404 The Vps33b-Vipar complex is required for epidermal homeostasis. <i>Journal of Investigative Dermatology</i> , 2017, 137, S70.	0.3	0
79	An LC-MS/MS-Based Method for the Quantification of Pyridox(am)ine 5-Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. <i>Analytical Chemistry</i> , 2017, 89, 8892-8900.	3.2	24
80	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. <i>Neurology: Clinical Practice</i> , 2017, 7, 499-511.	0.8	119
81	Mutations in <i>SLC25A22</i> : hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 385-394.	1.7	16
82	Role of Intramuscular Levofolate Administration in the Treatment of Hereditary Folate Malabsorption: Report of Three Cases. <i>JIMD Reports</i> , 2017, 39, 7-12.	0.7	7
83	Mouse decellularised liver scaffold improves human embryonic and induced pluripotent stem cells differentiation into hepatocyte-like cells. <i>PLoS ONE</i> , 2017, 12, e0189586.	1.1	24
84	Optimization of Liver Decellularization Maintains Extracellular Matrix Micro-Architecture and Composition Predisposing to Effective Cell Seeding. <i>PLoS ONE</i> , 2016, 11, e0155324.	1.1	69
85	Identification of novel bile acids as biomarkers for the early diagnosis of Niemann-Pick C disease. <i>FEBS Letters</i> , 2016, 590, 1651-1662.	1.3	82
86	Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 244-254.	0.5	114
87	250. A Phase 2/3 Study of the Efficacy and Safety of Ex Vivo Gene Therapy with Lenti-D™ Lentiviral Vector for the Treatment of Cerebral Adrenoleukodystrophy. <i>Molecular Therapy</i> , 2016, 24, S98-S99.	3.7	2
88	Regulation of post-Golgi LH3 trafficking is essential for collagen homeostasis. <i>Nature Communications</i> , 2016, 7, 12111.	5.8	54
89	Diagnostic workup and management of patients with suspected Niemann-Pick type C disease. <i>Therapeutic Advances in Neurological Disorders</i> , 2016, 9, 216-229.	1.5	18
90	Novel VIPAS39 mutation in a syndromic patient with arthrogryposis, renal tubular dysfunction and intrahepatic cholestasis. <i>European Journal of Medical Genetics</i> , 2016, 59, 237-239.	0.7	8

#	ARTICLE	IF	CITATIONS
91	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. <i>Brain</i> , 2016, 139, 2844-2854.	3.7	35
92	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. <i>Haematologica</i> , 2016, 101, 1170-1179.	1.7	119
93	The <sc>CHEVI</sc> tethering complex: facilitating special deliveries. <i>Journal of Pathology</i> , 2016, 240, 249-252.	2.1	14
94	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. <i>Nature Communications</i> , 2016, 7, 11601.	5.8	233
95	Alpha-Synuclein Oligomers Interact with Metal Ions to Induce Oxidative Stress and Neuronal Death in Parkinson's Disease. <i>Antioxidants and Redox Signaling</i> , 2016, 24, 376-391.	2.5	266
96	VPS33B regulates protein sorting into and maturation of Î±-granule progenitor organelles in mouse megakaryocytes. <i>Blood</i> , 2015, 126, 133-143.	0.6	56
97	Proteomic Discovery and Development of a Multiplexed Targeted MRM-LC-MS/MS Assay for Urine Biomarkers of Extracellular Matrix Disruption in Mucopolysaccharidoses I, II, and VI. <i>Analytical Chemistry</i> , 2015, 87, 12238-12244.	3.2	20
98	Hepatic regenerative medicine. <i>Journal of Hepatology</i> , 2015, 63, 523-524.	1.8	10
99	Use of nextâ€“generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders. <i>Journal of Thrombosis and Haemostasis</i> , 2015, 13, 643-650.	1.9	63
100	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 1008-1009.	2.6	1
101	Structural and functional hepatocyte polarity and liver disease. <i>Journal of Hepatology</i> , 2015, 63, 1023-1037.	1.8	229
102	The development of a rapid, multiplexed UPLCâ€“MS/MS assay for quantitation of lyso-Gb3 and Gb3 in dried blood spots. <i>Molecular Genetics and Metabolism</i> , 2015, 114, S107.	0.5	0
103	Inherited metabolic disorders presenting as acute liver failure in newborns and young children: Kingâ€™s College Hospital experience. <i>European Journal of Pediatrics</i> , 2015, 174, 1387-1392.	1.3	46
104	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. <i>Nature Communications</i> , 2015, 6, 8038.	5.8	160
105	Glycogen storage disease. <i>Paediatrics and Child Health (United Kingdom)</i> , 2015, 25, 139-144.	0.2	6
106	Perinatal systemic gene delivery using adeno-associated viral vectors. <i>Frontiers in Molecular Neuroscience</i> , 2014, 7, 89.	1.4	18
107	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 611-621.	2.6	89
108	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. <i>Brain</i> , 2014, 137, 1350-1360.	3.7	151

#	ARTICLE	IF	CITATIONS
109	Arthrogyrosis, Renal Dysfunction, and Cholestasis Syndrome Caused By <i>VIPAR</i> Mutation. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2014, 58, e29-32.	0.9	17
110	P1248 IDENTIFYING INCIDENCE OF INHERITED METABOLIC DISORDERS IN PATIENTS WITH INFANTILE LIVER DISEASE. <i>Journal of Hepatology</i> , 2014, 60, S505.	1.8	3
111	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875.	1.5	33
112	Genetic and laboratory diagnostic approach in Niemann Pick disease type C. <i>Journal of Neurology</i> , 2014, 261, 569-575.	1.8	50
113	Liver disease in infancy caused by oxysterol 7 $\alpha$ -hydroxylase deficiency: successful treatment with chenodeoxycholic acid. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 851-861.	1.7	58
114	Disorders of Bile Acid Synthesis and Biliary Transport. , 2014, , 555-576.		2
115	A combination of mutations in <i>AKR1D1</i> and <i>SKIV2L</i> in a family with severe infantile liver disease. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 74.	1.2	17
116	Alagille Syndrome and Other Hereditary Causes of Cholestasis. <i>Clinics in Liver Disease</i> , 2013, 17, 279-300.	1.0	40
117	Polymorphisms in <i>ABCB11</i> and <i>ATP8B1</i> Associated with Development of Severe Intrahepatic Cholestasis in Hodgkin's Lymphoma. <i>Journal of Clinical and Experimental Hepatology</i> , 2013, 3, 159-161.	0.4	14
118	Glomerular involvement in the arthrogyrosis, renal dysfunction and cholestasis syndrome. <i>CKJ: Clinical Kidney Journal</i> , 2013, 6, 183-188.	1.4	7
119	Mutation detection in cholestatic patients using microarray resequencing of <i>ATP8B1</i> and <i>ABCB11</i> . <i>F1000Research</i> , 2013, 2, 32.	0.8	6
120	Folliculin interacts with p0071 (plakophilin-4) and deficiency is associated with disordered RhoA signalling, epithelial polarization and cytokinesis. <i>Human Molecular Genetics</i> , 2012, 21, 5268-5279.	1.4	50
121	Mice Lacking the ITIM-Containing Receptor G6b-B Exhibit Macrothrombocytopenia and Aberrant Platelet Function. <i>Science Signaling</i> , 2012, 5, ra78.	1.6	65
122	Recessive germline <i>SDHA</i> and <i>SDHB</i> mutations causing leukodystrophy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 569-577.	1.5	100
123	Associations among genotype, clinical phenotype, and intracellular localization of trafficking proteins in ARC syndrome. <i>Human Mutation</i> , 2012, 33, 1656-1664.	1.1	74
124	Disorders of Tyrosine Metabolism. , 2012, , 265-276.		13
125	Bile acid-CoA ligase deficiency—a new inborn error of bile acid metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 521-530.	1.7	46
126	Rapid genetic diagnosis of heritable platelet function disorders with next-generation sequencing: proof of principle with Hermansky-Pudlak syndrome. <i>Journal of Thrombosis and Haemostasis</i> , 2012, 10, 306-309.	1.9	25



#	ARTICLE	IF	CITATIONS
127	Abstract LB-145: Folliculin interacts with p0071 (Plakophilin-4) and deficiency is associated with disordered RhoA signalling, epithelial polarization and cytokinesis. , 2012, , .		1
128	Glycogen storage disease. Paediatrics and Child Health (United Kingdom), 2011, 21, 84-89.	0.2	4
129	P10 Whole-exome-Sequencing-based discovery of novel syndromic form of neonatal cholestasis. Gut, 2011, 60, A5-A5.	6.1	0
130	Clinical and molecular characterisation of hereditary dopamine transporter deficiency syndrome: an observational cohort and experimental study. Lancet Neurology, The, 2011, 10, 54-62.	4.9	179
131	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurology, The, 2011, 10, 721-733.	4.9	290
132	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 499-507.	2.6	158
133	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 678.	2.6	2
134	MR spectroscopy-based brain metabolite profiling in propionic acidaemia: metabolic changes in the basal ganglia during acute decompensation and effect of liver transplantation. Orphanet Journal of Rare Diseases, 2011, 6, 19.	1.2	34
135	Magnetic Resonance Spectroscopy in the Diagnostic Evaluation of Brainstem Lesions in Alexander Disease. Journal of Child Neurology, 2011, 26, 356-360.	0.7	10
136	Submaximal Inhibition of Protein Kinase C Restores ADP-induced Dense Granule Secretion in Platelets in the Presence of Ca <sup>2+</sup> . Journal of Biological Chemistry, 2011, 286, 21073-21082.	1.6	23
137	Nephrotic range albuminuria as a feature of arthrogyrosis, renal dysfunction and cholestasis (ARC) syndrome: three new cases. Archives of Disease in Childhood, 2011, 96, A70-A71.	1.0	0
138	10 Differential Diagnosis in Patients with Developmental Delay: Focus on Inborn Errors of Metabolism. Pediatric Research, 2010, 68, 6-7.	1.1	0
139	Evidence for genetic heterogeneity in D-2-hydroxyglutaric aciduria. Human Mutation, 2010, 31, 279-283.	1.1	64
140	Design and validation of a metabolic disorder resequencing microarray (BRUM1). Human Mutation, 2010, 31, 858-865.	1.1	16
141	Mutations in VIPAR cause an arthrogyrosis, renal dysfunction and cholestasis syndrome phenotype with defects in epithelial polarization. Nature Genetics, 2010, 42, 303-312.	9.4	162
142	Mutations in SLC29A3, Encoding an Equilibrative Nucleoside Transporter ENT3, Cause a Familial Histiocytosis Syndrome (Faisalabad Histiocytosis) and Familial Rosai-Dorfman Disease. PLoS Genetics, 2010, 6, e1000833.	1.5	174
143	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. Brain, 2010, 133, 2964-2970.	3.7	95
144	Trafficking and Transporter Disorders in Pediatric Cholestasis. Clinics in Liver Disease, 2010, 14, 619-633.	1.0	17

#	ARTICLE	IF	CITATIONS
145	Mutations in TTC37 Cause Trichohepatoenteric Syndrome (Phenotypic Diarrhea of Infancy). <i>Gastroenterology</i> , 2010, 138, 2388-2398.e2.	0.6	124
146	Quantitative in vivo brain magnetic resonance spectroscopic monitoring of neurological involvement in mucopolysaccharidosis type II (Hunter Syndrome). <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 395-399.	1.7	14
147	Phenotypic approaches to gene mapping in platelet function disorders - identification of new variant of P2Y12, TxA2 and GPVI receptors. <i>Hamostaseologie</i> , 2010, 30, 29-38.	0.9	6
148	Molecular investigations to improve diagnostic accuracy in patients with ARC syndrome. <i>Human Mutation</i> , 2009, 30, E330-E337.	1.1	40
149	Homozygous loss-of-function mutations in the gene encoding the dopamine transporter are associated with infantile parkinsonism-dystonia. <i>Journal of Clinical Investigation</i> , 2009, 119, 1595-603.	3.9	173
150	Phenotypic spectrum of neurodegeneration associated with mutations in the <i>PLA2G6</i> gene (PLAN). <i>Neurology</i> , 2008, 70, 1623-1629.	1.5	215
151	Cargos and genes: insights into vesicular transport from inherited human disease. <i>Journal of Medical Genetics</i> , 2007, 44, 545-555.	1.5	43
152	A novel VPS33B mutation in an ARC syndrome patient presenting with osteopenia and fractures at birth. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2835-2837.	0.7	16
153	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpr rat. <i>Nature Genetics</i> , 2006, 38, 191-196.	9.4	266
154	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. <i>Nature Genetics</i> , 2006, 38, 752-754.	9.4	497
155	Increased nuchal translucency in arthrogryposis, renal dysfunction and cholestasis (ARC) syndrome and discovery of a Portuguese specific mutation in the VPS33B gene. <i>Ultrasound in Obstetrics and Gynecology</i> , 2006, 28, 233-234.	0.9	10
156	Successful treatment of pyridoxine-unresponsive homocystinuria with betaine in pregnancy. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 688-689.	1.7	16
157	Clinical and molecular genetic features of ARC syndrome. <i>Human Genetics</i> , 2006, 120, 396-409.	1.8	118
158	ZEBRAFISH VPS33B, AN ORTHOLOG OF THE GENE RESPONSIBLE FOR HUMAN ARTHROGRYPOSIS-RENAL DYSFUNCTION-CHOLESTASIS SYNDROME, REGULATES BILIARY DEVELOPMENT DOWNSTREAM OF THE ONECUT TRANSCRIPTION FACTOR HNF-6. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2005, 41, 513.	0.9	1
159	Requirement of VPS33B, a member of the Sec1/Munc18 protein family, in megakaryocyte and platelet $\alpha$ -granule biogenesis. <i>Blood</i> , 2005, 106, 4159-4166.	0.6	143
160	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. <i>Nature Genetics</i> , 2005, 37, 221-224.	9.4	201
161	Comparative evolutionary analysis of VPS33 homologues: genetic and functional insights. <i>Human Molecular Genetics</i> , 2005, 14, 1261-1270.	1.4	56
162	Zebrafish <i>vps33b</i> , an ortholog of the gene responsible for human arthrogryposis-renal dysfunction-cholestasis syndrome, regulates biliary development downstream of the <i>onecut</i> transcription factor <i>hnf6</i> . <i>Development (Cambridge)</i> , 2005, 132, 5295-5306.	1.2	61

#	ARTICLE	IF	CITATIONS
163	O0009 A LOCUS FOR ARTHROGRYPOSIS, RENAL DYSFUNCTION AND CHOLESTASIS SYNDROME MAPS TO CHROMOSOME 15Q. Journal of Pediatric Gastroenterology and Nutrition, 2004, 39, S10.	0.9	0
164	Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogyrosisâ€“renal dysfunctionâ€“cholestasis (ARC) syndrome. Nature Genetics, 2004, 36, 400-404.	9.4	313
165	New hope for treatment of neonatal haemochromatosis. Lancet, The, 2004, 364, 1644-1645.	6.3	9
166	Ophthalmic follow-up of patients with tyrosinaemia type I on NTBC. Journal of Inherited Metabolic Disease, 2003, 26, 13-16.	1.7	47
167	A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. Journal of Medical Genetics, 2003, 40, 431-435.	1.5	67
168	Molecular pathology and genetics of congenital hepatorenal fibrocystic syndromes. Journal of Medical Genetics, 2003, 40, 311-319.	1.5	100
169	Intentional overdose of warfarin in an adolescent: need for follow up. Emergency Medicine Journal, 2002, 19, 90-90.	0.4	4
170	A novel locus for Meckel-Gruber syndrome, MKS3 , maps to chromosome 8q24. Human Genetics, 2002, 111, 456-461.	1.8	55
171	Cerebrospinal fluid neurofilament light chain levels in CLN2 disease patients treated with enzyme replacement therapy normalise after two years on treatment. F1000Research, 0, 10, 614.	0.8	2
172	Niemannâ€“Pick type C disease as proofâ€“ofâ€“concept for intelligent biomarker panel selection in neurometabolic disorders. Developmental Medicine and Child Neurology, 0, , .	1.1	6