

# Paul Gissen

## List of Publications by Year in descending order

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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	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
2	Hematopoietic Stem-Cell Gene Therapy for Cerebral Adrenoleukodystrophy. <i>New England Journal of Medicine</i> , 2017, 377, 1630-1638.	13.9	412
3	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
4	Study of Intraventricular Cerliponase Alfa for CLN2 Disease. <i>New England Journal of Medicine</i> , 2018, 378, 1898-1907.	13.9	348
5	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
6	Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogyrosis-renal dysfunction-cholestasis (ARC) syndrome. <i>Nature Genetics</i> , 2004, 36, 400-404.	9.4	313
7	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. <i>Lancet Neurology</i> , The, 2011, 10, 721-733.	4.9	290
8	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. <i>Nature Genetics</i> , 2006, 38, 191-196.	9.4	266
9	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
10	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
11	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism-dystonia. <i>Nature Communications</i> , 2016, 7, 11601.	5.8	233
12	Structural and functional hepatocyte polarity and liver disease. <i>Journal of Hepatology</i> , 2015, 63, 1023-1037.	1.8	229
13	Phenotypic spectrum of neurodegeneration associated with mutations in the <i>PLA2G6</i> gene (PLAN). <i>Neurology</i> , 2008, 70, 1623-1629.	1.5	215
14	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. <i>Nature Genetics</i> , 2005, 37, 221-224.	9.4	201
15	Consensus clinical management guidelines for Niemann-Pick disease type C. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 50.	1.2	200
16	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017, 49, 223-237.	9.4	186
17	Clinical and molecular characterisation of hereditary dopamine transporter deficiency syndrome: an observational cohort and experimental study. <i>Lancet Neurology</i> , The, 2011, 10, 54-62.	4.9	179
18	Mutations in SLC29A3, Encoding an Equilibrative Nucleoside Transporter ENT3, Cause a Familial Histiocytosis Syndrome (Faisalabad Histiocytosis) and Familial Rosai-Dorfman Disease. <i>PLoS Genetics</i> , 2010, 6, e1000833.	1.5	174

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19	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
20	Mutations in VIPAR cause an arthrogyriposis, renal dysfunction and cholestasis syndrome phenotype with defects in epithelial polarization. <i>Nature Genetics</i> , 2010, 42, 303-312.	9.4	162
21	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. <i>Nature Communications</i> , 2015, 6, 8038.	5.8	160
22	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. <i>American Journal of Human Genetics</i> , 2011, 88, 499-507.	2.6	158
23	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. <i>Brain</i> , 2014, 137, 1350-1360.	3.7	151
24	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	13.7	148
25	Requirement of VPS33B, a member of the Sec1/Munc18 protein family, in megakaryocyte and platelet Å-granule biogenesis. <i>Blood</i> , 2005, 106, 4159-4166.	0.6	143
26	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
27	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
28	Mutations in TTC37 Cause Trichohepatoenteric Syndrome (Phenotypic Diarrhea of Infancy). <i>Gastroenterology</i> , 2010, 138, 2388-2398.e2.	0.6	124
29	Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. <i>Haematologica</i> , 2016, 101, 1170-1179.	1.7	119
30	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. <i>Neurology: Clinical Practice</i> , 2017, 7, 499-511.	0.8	119
31	Clinical and molecular genetic features of ARC syndrome. <i>Human Genetics</i> , 2006, 120, 396-409.	1.8	118
32	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
33	Lamin and the heart. <i>Heart</i> , 2018, 104, 468-479.	1.2	113
34	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
35	Molecular pathology and genetics of congenital hepatorenal fibrocystic syndromes. <i>Journal of Medical Genetics</i> , 2003, 40, 311-319.	1.5	100
36	Recessive germline SDHA and SDHB mutations causing leukodystrophy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 569-577.	1.5	100

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37	Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. <i>Brain</i> , 2010, 133, 2964-2970.	3.7	95
38	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
39	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
40	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
41	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
42	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
43	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
44	A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. <i>Journal of Medical Genetics</i> , 2003, 40, 431-435.	1.5	67
45	Mice Lacking the ITIM-Containing Receptor G6b-B Exhibit Macrothrombocytopenia and Aberrant Platelet Function. <i>Science Signaling</i> , 2012, 5, ra78.	1.6	65
46	Evidence for genetic heterogeneity in D-2-hydroxyglutaric aciduria. <i>Human Mutation</i> , 2010, 31, 279-283.	1.1	64
47	Use of next-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
48	Zebrafish <i>vps33b</i> , an ortholog of the gene responsible for human arthrogyrosis-renal dysfunction-cholestasis syndrome, regulates biliary development downstream of the onecut transcription factor <i>hnf6</i> . <i>Development (Cambridge)</i> , 2005, 132, 5295-5306.	1.2	61
49	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
50	Clinical applications for exosomes: Are we there yet?. <i>British Journal of Pharmacology</i> , 2021, 178, 2375-2392.	2.7	57
51	Comparative evolutionary analysis of <i>VPS33</i> homologues: genetic and functional insights. <i>Human Molecular Genetics</i> , 2005, 14, 1261-1270.	1.4	56
52	<i>VPS33B</i> regulates protein sorting into and maturation of $\gamma$ -granule progenitor organelles in mouse megakaryocytes. <i>Blood</i> , 2015, 126, 133-143.	0.6	56
53	A novel locus for Meckel-Gruber syndrome, <i>MKS3</i> , maps to chromosome 8q24. <i>Human Genetics</i> , 2002, 111, 456-461.	1.8	55
54	Expanding the phenotype in argininosuccinic aciduria: need for new therapies. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 357-368.	1.7	55

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55	Regulation of post-Golgi LH3 trafficking is essential for collagen homeostasis. <i>Nature Communications</i> , 2016, 7, 12111.	5.8	54
56	Vps33b is crucial for structural and functional hepatocyte polarity. <i>Journal of Hepatology</i> , 2017, 66, 1001-1011.	1.8	51
57	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
58	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
59	Genetic and laboratory diagnostic approach in Niemann Pick disease type C. <i>Journal of Neurology</i> , 2014, 261, 569-575.	1.8	50
60	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
61	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
62	Ophthalmic follow-up of patients with tyrosinaemia type I on NTBC. <i>Journal of Inherited Metabolic Disease</i> , 2003, 26, 13-16.	1.7	47
63	Phase I/II Trial of Liverâ€‘derived Mesenchymal Stem Cells in Pediatric Liverâ€‘based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liverâ€‘derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients. <i>Transplantation</i> , 2019, 103, 1903-1915.	0.5	47
64	Bile acidâ€‘CoA ligase deficiencyâ€‘a new inborn error of bile acid metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 521-530.	1.7	46
65	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
66	Molecular architecture of the multifunctional collagen lysyl hydroxylase and glycosyltransferase LH3. <i>Nature Communications</i> , 2018, 9, 3163.	5.8	46
67	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
68	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
69	Cargos and genes: insights into vesicular transport from inherited human disease. <i>Journal of Medical Genetics</i> , 2007, 44, 545-555.	1.5	43
70	Molecular investigations to improve diagnostic accuracy in patients with ARC syndrome. <i>Human Mutation</i> , 2009, 30, E330-E337.	1.1	40
71	Alagille Syndrome and Other Hereditary Causes of Cholestasis. <i>Clinics in Liver Disease</i> , 2013, 17, 279-300.	1.0	40
72	Using stem cellâ€‘derived neurons in drug screening for neurological diseases. <i>Neurobiology of Aging</i> , 2019, 78, 130-141.	1.5	38

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73	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
74	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
75	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
76	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. <i>Brain</i> , 2016, 139, 2844-2854.	3.7	35
77	Argininosuccinic aciduria: Recent pathophysiological insights and therapeutic prospects. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 1147-1161.	1.7	35
78	MR spectroscopy-based brain metabolite profiling in propionic acidemia: metabolic changes in the basal ganglia during acute decompensation and effect of liver transplantation. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 19.	1.2	34
79	Argininosuccinic aciduria fosters neuronal nitrosative stress reversed by Asl gene transfer. <i>Nature Communications</i> , 2018, 9, 3505.	5.8	34
80	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875.	1.5	33
81	Gene Therapy for Lysosomal Storage Disorders: Ongoing Studies and Clinical Development. <i>Biomolecules</i> , 2021, 11, 611.	1.8	27
82	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
83	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
84	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
85	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
86	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
87	Submaximal Inhibition of Protein Kinase C Restores ADP-induced Dense Granule Secretion in Platelets in the Presence of Ca <sup>2+</sup> . <i>Journal of Biological Chemistry</i> , 2011, 286, 21073-21082.	1.6	23
88	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
89	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
90	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

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91	Perinatal systemic gene delivery using adeno-associated viral vectors. <i>Frontiers in Molecular Neuroscience</i> , 2014, 7, 89.	1.4	18
92	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
93	Trafficking and Transporter Disorders in Pediatric Cholestasis. <i>Clinics in Liver Disease</i> , 2010, 14, 619-633.	1.0	17
94	A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 74.	1.2	17
95	Arthrogryposis, 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
96	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. <i>Brain Communications</i> , 2020, 2, fcaa178.	1.5	17
97	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
98	Successful treatment of pyridoxine-unresponsive homocystinuria with betaine in pregnancy. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 688-689.	1.7	16
99	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
100	Design and validation of a metabolic disorder resequencing microarray (BRUM1). <i>Human Mutation</i> , 2010, 31, 858-865.	1.1	16
101	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
102	Multiplex High-Throughput Targeted Proteomic Assay To Identify Induced Pluripotent Stem Cells. <i>Analytical Chemistry</i> , 2017, 89, 2440-2448.	3.2	15
103	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
104	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
105	Polymorphisms in ABCB11 and ATP8B1 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
106	The <i>CHEV1</i> tethering complex: facilitating special deliveries. <i>Journal of Pathology</i> , 2016, 240, 249-252.	2.1	14
107	Oculomotor abnormalities in children with Niemann-Pick type C. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 159-168.	0.5	14
108	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



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109	Disorders of Tyrosine Metabolism. , 2012, , 265-276.		13
110	Diagnosing Mitochondrial Disorders Remains Challenging in the Omics Era. <i>Neurology: Genetics</i> , 2021, 7, e597.	0.9	13
111	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
112	Urine proteomics analysis of patients with neuronal ceroid lipofuscinoses. <i>IScience</i> , 2021, 24, 102020.	1.9	12
113	Delivering efficient liver-directed AAV-mediated gene therapy. <i>Gene Therapy</i> , 2017, 24, 263-264.	2.3	11
114	Increased nuchal translucency in arthrogyriposis, renal dysfunction and cholestasis (ARC) syndrome and discovery of a Portuguese specific mutation in theVPS33B gene. <i>Ultrasound in Obstetrics and Gynecology</i> , 2006, 28, 233-234.	0.9	10
115	Magnetic Resonance Spectroscopy in the Diagnostic Evaluation of Brainstem Lesions in Alexander Disease. <i>Journal of Child Neurology</i> , 2011, 26, 356-360.	0.7	10
116	Hepatic regenerative medicine. <i>Journal of Hepatology</i> , 2015, 63, 523-524.	1.8	10
117	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
118	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
119	New hope for treatment of neonatal haemochromatosis. <i>Lancet, The</i> , 2004, 364, 1644-1645.	6.3	9
120	Gene therapies targeting the liver. <i>Journal of Hepatology</i> , 2021, 74, 235-236.	1.8	9
121	Novel VIPAS39 mutation in a syndromic patient with arthrogyriposis, renal tubular dysfunction and intrahepatic cholestasis. <i>European Journal of Medical Genetics</i> , 2016, 59, 237-239.	0.7	8
122	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
123	An InÂVtro Whole-Organ Liver Engineering for Testing of Genetic Therapies. <i>IScience</i> , 2020, 23, 101808.	1.9	8
124	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
125	International consensus on clinical severity scale use in evaluating Niemannâ€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
126	Glomerular involvement in the arthrogyriposis, renal dysfunction and cholestasis syndrome. CKJ: <i>Clinical Kidney Journal</i> , 2013, 6, 183-188.	1.4	7



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127	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
128	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
129	The Genetics of Inherited Cholestatic Disorders in Neonates and Infants: Evolving Challenges. <i>Genes</i> , 2021, 12, 1837.	1.0	7
130	Glycogen storage disease. <i>Paediatrics and Child Health (United Kingdom)</i> , 2015, 25, 139-144.	0.2	6
131	An ERG and OCT study of neuronal ceroid lipofuscinosis CLN2 Battens retinopathy. <i>Eye</i> , 2021, 35, 2438-2448.	1.1	6
132	Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. <i>F1000Research</i> , 2013, 2, 32.	0.8	6
133	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
134	Niemannâ€“Pick type C disease as proofâ€“ofâ€“concept for intelligent biomarker panel selection in neurometabolic disorders. <i>Developmental Medicine and Child Neurology</i> , 0, , .	1.1	6
135	Intentional overdose of warfarin in an adolescent: need for follow up. <i>Emergency Medicine Journal</i> , 2002, 19, 90-90.	0.4	4
136	Glycogen storage disease. <i>Paediatrics and Child Health (United Kingdom)</i> , 2011, 21, 84-89.	0.2	4
137	High-Content Analysis of Mitochondrial Function in iPSC-Derived Neurons. <i>Methods in Molecular Biology</i> , 2019, 1994, 175-184.	0.4	4
138	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
139	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
140	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
141	P1248 IDENTIFYING INCIDENCE OF INHERITED METABOLIC DISORDERS IN PATIENTS WITH INFANTILE LIVER DISEASE. <i>Journal of Hepatology</i> , 2014, 60, S505.	1.8	3
142	Seeding Induced Pluripotent Stem Cell-Derived Neurons onto 384-Well Plates. <i>Methods in Molecular Biology</i> , 2019, 1994, 159-164.	0.4	3
143	Rare missense variants in Tropomyosinâ€“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
144	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. <i>American Journal of Human Genetics</i> , 2011, 88, 678.	2.6	2

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145	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
146	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
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