# Paul Gissen

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 172 papers | 10,575 citations | 36303 51 h-index | 36028 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. Nature Genetics, 2006, 38, 752-754. | 21.4 | 497 |
| 2 | Hematopoietic Stem-Cell Gene Therapy for Cerebral Adrenoleukodystrophy. New England Journal of Medicine, 2017, 377, 1630-1638. | 27.0 | 412 |
| 3 | Î±-synuclein oligomers interact with ATP synthase and open the permeability transition pore in Parkinsonâ€™s disease. Nature Communications, 2018, 9, 2293. | 12.8 | 351 |
| 4 | Study of Intraventricular Cerliponase Alfa for CLN2 Disease. New England Journal of Medicine, 2018, 378, 1898-1907. | 27.0 | 348 |
| 5 | Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90. | 6.2 | 343 |
| 6 | Mutations in VPS33B, encoding a regulator of SNARE-dependent membrane fusion, cause arthrogryposisâ€“renal dysfunctionâ€“cholestasis (ARC) syndrome. Nature Genetics, 2004, 36, 400-404. | 21.4 | 313 |
| 7 | The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurology, The, 2011, 10, 721-733. | 10.2 | 290 |
| 8 | The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nature Genetics, 2006, 38, 191-196. | 21.4 | 266 |
| 9 | Alpha-Synuclein Oligomers Interact with Metal Ions to Induce Oxidative Stress and Neuronal Death in Parkinson's Disease. Antioxidants and Redox Signaling, 2016, 24, 376-391. | 5.4 | 266 |
| 10 | Î±-Synuclein binds to the ERâ€“mitochondria tethering protein VAPB to disrupt Ca2+ homeostasis and mitochondrial ATP production. Acta Neuropathologica, 2017, 134, 129-149. | 7.7 | 262 |
| 11 | Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601. | 12.8 | 233 |
| 12 | Structural and functional hepatocyte polarity and liver disease. Journal of Hepatology, 2015, 63, 1023-1037. | 3.7 | 229 |
| 13 | Phenotypic spectrum of neurodegeneration associated with mutations in the PLA2G6 gene (PLAN). Neurology, 2008, 70, 1623-1629. | 1.1 | 215 |
| 14 | Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224. | 21.4 | 201 |
| 15 | Consensus clinical management guidelines for Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2018, 13, 50. | 2.7 | 200 |
| 16 | Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237. | 21.4 | 186 |
| 17 | Clinical and molecular characterisation of hereditary dopamine transporter deficiency syndrome: an observational cohort and experimental study. Lancet Neurology, The, 2011, 10, 54-62. | 10.2 | 179 |
| 18 | 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. | 3.5 | 174 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Homozygous loss-of-function mutations in the gene encoding the dopamine transporter are associated with infantile parkinsonism-dystonia. Journal of Clinical Investigation, 2009, 119, 1595-603. | 8.2 | 173 |
| 20 | Mutations in VIPAR cause an arthrogryposis, renal dysfunction and cholestasis syndrome phenotype with defects in epithelial polarization. Nature Genetics, 2010, 42, 303-312. | 21.4 | 162 |
| 21 | Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. Nature Communications, 2015, 6, 8038. | 12.8 | 160 |
| 22 | Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 499-507. | 6.2 | 158 |
| 23 | Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. Brain, 2014, 137, 1350-1360. | 7.6 | 151 |
| 24 | Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95. | 27.8 | 148 |
| 25 | Requirement of VPS33B, a member of the Sec1/Munc18 protein family, in megakaryocyte and platelet Â-granule biogenesis. Blood, 2005, 106, 4159-4166. | 1.4 | 143 |
| 26 | Alpha synuclein aggregation drives ferroptosis: an interplay of iron, calcium and lipid peroxidation. Cell Death and Differentiation, 2020, 27, 2781-2796. | 11.2 | 142 |
| 27 | Reproducibility of Molecular Phenotypes after Long-Term Differentiation toÂHuman iPSC-Derived Neurons: A Multi-Site Omics Study. Stem Cell Reports, 2018, 11, 897-911. | 4.8 | 135 |
| 28 | Mutations in TTC37 Cause Trichohepatoenteric Syndrome (Phenotypic Diarrhea of Infancy). Gastroenterology, 2010, 138, 2388-2398.e2. | 1.3 | 124 |
| 29 | Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. Haematologica, 2016, 101, 1170-1179. | 3.5 | 119 |
| 30 | Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511. | 1.6 | 119 |
| 31 | Clinical and molecular genetic features of ARC syndrome. Human Genetics, 2006, 120, 396-409. | 3.8 | 118 |
| 32 | Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. Molecular Genetics and Metabolism, 2016, 118, 244-254. | 1.1 | 114 |
| 33 | Lamin and the heart. Heart, 2018, 104, 468-479. | 2.9 | 113 |
| 34 | Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033. | 1.6 | 111 |
| 35 | Molecular pathology and genetics of congenital hepatorenal fibrocystic syndromes. Journal of Medical Genetics, 2003, 40, 311-319. | 3.2 | 100 |
| 36 | Recessive germlineSDHAandSDHBmutations causing leukodystrophy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2012, 49, 569-577. | 3.2 | 100 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Phospholipase C beta 1 deficiency is associated with early-onset epileptic encephalopathy. Brain, 2010, 133, 2964-2970. | 7.6 | 95 |
| 38 | Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2014, 95, 611-621. | 6.2 | 89 |
| 39 | Gene therapy for monogenic liver diseases: clinical successes, current challenges and future prospects. Journal of Inherited Metabolic Disease, 2017, 40, 497-517. | 3.6 | 89 |
| 40 | Identification of novel bile acids as biomarkers for the early diagnosis of Niemannâ€Pick C disease. FEBS Letters, 2016, 590, 1651-1662. | 2.8 | 82 |
| 41 | Associations among genotype, clinical phenotype, and intracellular localization of trafficking proteins in ARC syndrome. Human Mutation, 2012, 33, 1656-1664. | 2.5 | 74 |
| 42 | Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877. | 5.3 | 70 |
| 43 | Optimization of Liver Decellularization Maintains Extracellular Matrix Micro-Architecture and Composition Predisposing to Effective Cell Seeding. PLoS ONE, 2016, 11, e0155324. | 2.5 | 69 |
| 44 | A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. Journal of Medical Genetics, 2003, 40, 431-435. | 3.2 | 67 |
| 45 | Mice Lacking the ITIM-Containing Receptor G6b-B Exhibit Macrothrombocytopenia and Aberrant Platelet Function. Science Signaling, 2012, 5, ra78. | 3.6 | 65 |
| 46 | Evidence for genetic heterogeneity in D-2-hydroxyglutaric aciduria. Human Mutation, 2010, 31, 279-283. | 2.5 | 64 |
| 47 | Use of nextâ€generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders. Journal of Thrombosis and Haemostasis, 2015, 13, 643-650. | 3.8 | 63 |
| 48 | Zebrafish vps33b, an ortholog of the gene responsible for human arthrogryposis-renal dysfunction-cholestasis syndrome, regulates biliary development downstream of the onecut transcription factor hnf6. Development (Cambridge), 2005, 132, 5295-5306. | 2.5 | 61 |
| 49 | Liver disease in infancy caused by oxysterol 7Î±â€hydroxylase deficiency: successful treatment with chenodeoxycholic acid. Journal of Inherited Metabolic Disease, 2014, 37, 851-861. | 3.6 | 58 |
| 50 | Clinical applications for exosomes: Are we there yet?. British Journal of Pharmacology, 2021, 178, 2375-2392. | 5.4 | 57 |
| 51 | Comparative evolutionary analysis of VPS33 homologues: genetic and functional insights. Human Molecular Genetics, 2005, 14, 1261-1270. | 2.9 | 56 |
| 52 | VPS33B regulates protein sorting into and maturation of Î±-granule progenitor organelles in mouse megakaryocytes. Blood, 2015, 126, 133-143. | 1.4 | 56 |
| 53 | A novel locus for Meckel-Gruber syndrome, MKS3 , maps to chromosome 8q24. Human Genetics, 2002, 111, 456-461. | 3.8 | 55 |
| 54 | Expanding the phenotype in argininosuccinic aciduria: need for new therapies. Journal of Inherited Metabolic Disease, 2017, 40, 357-368. | 3.6 | 55 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Regulation of post-Golgi LH3 trafficking is essential for collagen homeostasis. Nature Communications, 2016, 7, 12111. | 12.8 | 54 |
| 56 | Vps33b is crucial for structural and functional hepatocyte polarity. Journal of Hepatology, 2017, 66, 1001-1011. | 3.7 | 51 |
| 57 | Age-Related Seroprevalence of Antibodies Against AAV-LK03 in a UK Population Cohort. Human Gene Therapy, 2019, 30, 79-87. | 2.7 | 51 |
| 58 | Folliculin interacts with p0071 (plakophilin-4) and deficiency is associated with disordered RhoA signalling, epithelial polarization and cytokinesis. Human Molecular Genetics, 2012, 21, 5268-5279. | 2.9 | 50 |
| 59 | Genetic and laboratory diagnostic approach in Niemann Pick disease type C. Journal of Neurology, 2014, 261, 569-575. | 3.6 | 50 |
| 60 | A single cell high content assay detects mitochondrial dysfunction in iPSC-derived neurons with mutations in SNCA. Scientific Reports, 2018, 8, 9033. | 3.3 | 50 |
| 61 | Myostatin inhibition in combination with antisense oligonucleotide therapy improves outcomes in spinal muscular atrophy. Journal of Cachexia, Sarcopenia and Muscle, 2020, 11, 768-782. | 7.3 | 49 |
| 62 | Ophthalmic follow-up of patients with tyrosinaemia type I on NTBC. Journal of Inherited Metabolic Disease, 2003, 26, 13-16. | 3.6 | 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. Transplantation, 2019, 103, 1903-1915. | 1.0 | 47 |
| 64 | Bile acidâ€CoA ligase deficiencyâ€”a new inborn error of bile acid metabolism. Journal of Inherited Metabolic Disease, 2012, 35, 521-530. | 3.6 | 46 |
| 65 | Inherited metabolic disorders presenting as acute liver failure in newborns and young children: Kingâ€™s College Hospital experience. European Journal of Pediatrics, 2015, 174, 1387-1392. | 2.7 | 46 |
| 66 | Molecular architecture of the multifunctional collagen lysyl hydroxylase and glycosyltransferase LH3. Nature Communications, 2018, 9, 3163. | 12.8 | 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. American Journal of Human Genetics, 2018, 103, 3-18. | 6.2 | 46 |
| 68 | The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. Genetics in Medicine, 2017, 19, 1217-1225. | 2.4 | 45 |
| 69 | Cargos and genes: insights into vesicular transport from inherited human disease. Journal of Medical Genetics, 2007, 44, 545-555. | 3.2 | 43 |
| 70 | Molecular investigations to improve diagnostic accuracy in patients with ARC syndrome. Human Mutation, 2009, 30, E330-E337. | 2.5 | 40 |
| 71 | Alagille Syndrome and Other Hereditary Causes of Cholestasis. Clinics in Liver Disease, 2013, 17, 279-300. | 2.1 | 40 |
| 72 | Using stem cellâ€“derived neurons in drug screening for neurological diseases. Neurobiology of Aging, 2019, 78, 130-141. | 3.1 | 38 |

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|---|---|---|---|
| 73 | Autosomal Recessive Keratoderma-Ichthyosis-Deafness (ARKID) Syndrome IsÂCaused by VPS33B Mutations AffectingÂRab Protein Interaction andÂCollagen Modification. Journal of Investigative Dermatology, 2017, 137, 845-854. | 0.7 | 37 |
| 74 | VPS33B and VIPAR are essential for epidermal lamellar body biogenesis and function. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 1609-1621. | 3.8 | 37 |
| 75 | De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153. | 6.2 | 36 |
| 76 | Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. Brain, 2016, 139, 2844-2854. | 7.6 | 35 |
| 77 | Argininosuccinic aciduria: Recent pathophysiological insights and therapeutic prospects. Journal of Inherited Metabolic Disease, 2019, 42, 1147-1161. | 3.6 | 35 |
| 78 | 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. | 2.7 | 34 |
| 79 | Argininosuccinic aciduria fosters neuronal nitrosative stress reversed by Asl gene transfer. Nature Communications, 2018, 9, 3505. | 12.8 | 34 |
| 80 | Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. Neurology, 2014, 83, 1873-1875. | 1.1 | 33 |
| 81 | Gene Therapy for Lysosomal Storage Disorders: Ongoing Studies and Clinical Development. Biomolecules, 2021, 11, 611. | 4.0 | 27 |
| 82 | Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342. | 6.2 | 26 |
| 83 | Rapid genetic diagnosis of heritable platelet function disorders with nextâ€generation sequencing: proofâ€ofâ€principle with Hermanskyâ€“Pudlak syndrome. Journal of Thrombosis and Haemostasis, 2012, 10, 306-309. | 3.8 | 25 |
| 84 | An LCâ€“MS/MS-Based Method for the Quantification of Pyridox(am)ine 5â€²-Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. Analytical Chemistry, 2017, 89, 8892-8900. | 6.5 | 24 |
| 85 | Mouse decellularised liver scaffold improves human embryonic and induced pluripotent stem cells differentiation into hepatocyte-like cells. PLoS ONE, 2017, 12, e0189586. | 2.5 | 24 |
| 86 | Efficacy and safety of N-acetyl-l-leucine in Niemannâ€“Pick disease type C. Journal of Neurology, 2022, 269, 1651-1662. | 3.6 | 24 |
| 87 | Submaximal Inhibition of Protein Kinase C Restores ADP-induced Dense Granule Secretion in Platelets in the Presence of Ca2+. Journal of Biological Chemistry, 2011, 286, 21073-21082. | 3.4 | 23 |
| 88 | Clinical and Molecular Features of Early Infantile Niemann Pick Type C Disease. International Journal of Molecular Sciences, 2020, 21, 5059. | 4.1 | 21 |
| 89 | Safety and efficacy of an engineered hepatotropic AAV gene therapy for ornithine transcarbamylase deficiency in cynomolgus monkeys. Molecular Therapy - Methods and Clinical Development, 2021, 23, 135-146. | 4.1 | 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. Analytical Chemistry, 2015, 87, 12238-12244. | 6.5 | 20 |

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|---|---|---|---|
| 91 | Perinatal systemic gene delivery using adeno-associated viral vectors. Frontiers in Molecular Neuroscience, 2014, 7, 89. | 2.9 | 18 |
| 92 | Diagnostic workup and management of patients with suspected Niemann-Pick type C disease. Therapeutic Advances in Neurological Disorders, 2016, 9, 216-229. | 3.5 | 18 |
| 93 | Trafficking and Transporter Disorders in Pediatric Cholestasis. Clinics in Liver Disease, 2010, 14, 619-633. | 2.1 | 17 |
| 94 | A combination of mutations in AKR1D1 and SKIV2L in a family with severe infantile liver disease. Orphanet Journal of Rare Diseases, 2013, 8, 74. | 2.7 | 17 |
| 95 | Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome Caused By VIPAR Mutation. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, e29-32. | 1.8 | 17 |
| 96 | Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178. | 3.3 | 17 |
| 97 | Guidelines on the diagnosis, clinical assessments, treatment and management for CLN2 disease patients. Orphanet Journal of Rare Diseases, 2021, 16, 185. | 2.7 | 17 |
| 98 | Successful treatment of pyridoxine-unresponsive homocystinuria with betaine in pregnancy. Journal of Inherited Metabolic Disease, 2006, 29, 688-689. | 3.6 | 16 |
| 99 | A novel VPS33B mutation in an ARC syndrome patient presenting with osteopenia and fractures at birth. American Journal of Medical Genetics, Part A, 2007, 143A, 2835-2837. | 1.2 | 16 |
| 100 | Design and validation of a metabolic disorder resequencing microarray (BRUM1). Human Mutation, 2010, 31, 858-865. | 2.5 | 16 |
| 101 | Mutations in SLC25A22: hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. Journal of Inherited Metabolic Disease, 2017, 40, 385-394. | 3.6 | 16 |
| 102 | Multiplex High-Throughput Targeted Proteomic Assay To Identify Induced Pluripotent Stem Cells. Analytical Chemistry, 2017, 89, 2440-2448. | 6.5 | 15 |
| 103 | Severe renal Fanconi and management strategies in Arthrogryposis-Renal dysfunction-Cholestasis syndrome: a case report. BMC Nephrology, 2018, 19, 144. | 1.8 | 15 |
| 104 | Quantitative in vivo brain magnetic resonance spectroscopic monitoring of neurological involvement in mucopolysaccharidosis type II (Hunter Syndrome). Journal of Inherited Metabolic Disease, 2010, 33, 395-399. | 3.6 | 14 |
| 105 | Polymorphisms in ABCB11 and ATP8B1 Associated with Development of Severe Intrahepatic Cholestasis in Hodgkin's Lymphoma. Journal of Clinical and Experimental Hepatology, 2013, 3, 159-161. | 0.9 | 14 |
| 106 | The <scp>CHEVI</scp> tethering complex: facilitating special deliveries. Journal of Pathology, 2016, 240, 249-252. | 4.5 | 14 |
| 107 | Oculomotor abnormalities in children with Niemann-Pick type C. Molecular Genetics and Metabolism, 2018, 123, 159-168. | 1.1 | 14 |
| 108 | Clinical disease characteristics of patients with Niemann-Pick Disease Type C: findings from the International Niemann-Pick Disease Registry (INPDR). Orphanet Journal of Rare Diseases, 2022, 17, 51. | 2.7 | 14 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Disorders of Tyrosine Metabolism. , 2012, , 265-276. | | 13 |
| 110 | Diagnosing Mitochondrial Disorders Remains Challenging in the Omics Era. Neurology: Genetics, 2021, 7, e597. | 1.9 | 13 |
| 111 | Spectrum of movement disorders and neurotransmitter abnormalities in paediatric POLG disease. Journal of Inherited Metabolic Disease, 2018, 41, 1275-1283. | 3.6 | 12 |
| 112 | Urine proteomics analysis of patients with neuronal ceroid lipofuscinoses. IScience, 2021, 24, 102020. | 4.1 | 12 |
| 113 | Delivering efficient liver-directed AAV-mediated gene therapy. Gene Therapy, 2017, 24, 263-264. | 4.5 | 11 |
| 114 | Increased nuchal translucency in arthrogryposis, renal dysfunction and cholestasis (ARC) syndrome and discovery of a Portuguese specific mutation in theVPS33B gene. Ultrasound in Obstetrics and Gynecology, 2006, 28, 233-234. | 1.7 | 10 |
| 115 | Magnetic Resonance Spectroscopy in the Diagnostic Evaluation of Brainstem Lesions in Alexander Disease. Journal of Child Neurology, 2011, 26, 356-360. | 1.4 | 10 |
| 116 | Hepatic regenerative medicine. Journal of Hepatology, 2015, 63, 523-524. | 3.7 | 10 |
| 117 | Free urinary glycosylated hydroxylysine as an indicator of altered collagen degradation in the mucopolysaccharidoses. Journal of Inherited Metabolic Disease, 2020, 43, 309-317. | 3.6 | 10 |
| 118 | Cerliponase Alfa for the Treatment of Atypical Phenotypes of CLN2 Disease: A Retrospective Case Series. Journal of Child Neurology, 2021, 36, 468-474. | 1.4 | 10 |
| 119 | New hope for treatment of neonatal haemochromatosis. Lancet, The, 2004, 364, 1644-1645. | 13.7 | 9 |
| 120 | Gene therapies targeting the liver. Journal of Hepatology, 2021, 74, 235-236. | 3.7 | 9 |
| 121 | Novel VIPAS39 mutation in a syndromic patient with arthrogryposis, renal tubular dysfunction and intrahepatic cholestasis. European Journal of Medical Genetics, 2016, 59, 237-239. | 1.3 | 8 |
| 122 | Markers of cognitive function in individuals with metabolic disease: Morquio syndrome and tyrosinemia type III. Cognitive Neuropsychology, 2018, 35, 120-147. | 1.1 | 8 |
| 123 | An InÂVitro Whole-Organ Liver Engineering for Testing of Genetic Therapies. IScience, 2020, 23, 101808. | 4.1 | 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). Orphanet Journal of Rare Diseases, 2021, 16, 217. | 2.7 | 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. Orphanet Journal of Rare Diseases, 2021, 16, 482. | 2.7 | 8 |
| 126 | Glomerular involvement in the arthrogryposis, renal dysfunction and cholestasis syndrome. CKJ: Clinical Kidney Journal, 2013, 6, 183-188. | 2.9 | 7 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Role of Intramuscular Levofolinate Administration in the Treatment of Hereditary Folate Malabsorption: Report of Three Cases. JIMD Reports, 2017, 39, 7-12. | 1.5 | 7 |
| 128 | Surveillance for variant CJD: should more children with neurodegenerative diseases have autopsies?. Archives of Disease in Childhood, 2019, 104, 360-365. | 1.9 | 7 |
| 129 | The Genetics of Inherited Cholestatic Disorders in Neonates and Infants: Evolving Challenges. Genes, 2021, 12, 1837. | 2.4 | 7 |
| 130 | Glycogen storage disease. Paediatrics and Child Health (United Kingdom), 2015, 25, 139-144. | 0.4 | 6 |
| 131 | An ERG and OCT study of neuronal ceroid lipofuscinosis CLN2 Battens retinopathy. Eye, 2021, 35, 2438-2448. | 2.1 | 6 |
| 132 | Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. F1000Research, 2013, 2, 32. | 1.6 | 6 |
| 133 | Phenotypic approaches to gene mapping in platelet function disorders - identification of new variant of P2Y12, TxA2 and GPVI receptors. Hamostaseologie, 2010, 30, 29-38. | 1.9 | 6 |
| 134 | Niemannâ€“Pick type C disease as proofâ€ofâ€concept for intelligent biomarker panel selection in neurometabolic disorders. Developmental Medicine and Child Neurology, 0, , . | 2.1 | 6 |
| 135 | Intentional overdose of warfarin in an adolescent: need for follow up. Emergency Medicine Journal, 2002, 19, 90-90. | 1.0 | 4 |
| 136 | Glycogen storage disease. Paediatrics and Child Health (United Kingdom), 2011, 21, 84-89. | 0.4 | 4 |
| 137 | High-Content Analysis of Mitochondrial Function in iPSC-Derived Neurons. Methods in Molecular Biology, 2019, 1994, 175-184. | 0.9 | 4 |
| 138 | Urea Cycle Related Amino Acids Measured in Dried Bloodspots Enable Long-Term In Vivo Monitoring and Therapeutic Adjustment. Metabolites, 2019, 9, 275. | 2.9 | 4 |
| 139 | Clinical Pharmacokinetics and Pharmacodynamics of Cerliponase Alfa, Enzyme Replacement Therapy for CLN2 Disease by Intracerebroventricular Administration. Clinical and Translational Science, 2021, 14, 635-644. | 3.1 | 4 |
| 140 | Cerebrospinal fluid neurofilament light levels in CLN2 disease patients treated with enzyme replacement therapy normalise after two years on treatment. F1000Research, 2021, 10, 614. | 1.6 | 4 |
| 141 | P1248 IDENTIFYING INCIDENCE OF INHERITED METABOLIC DISORDERS IN PATIENTS WITH INFANTILE LIVER DISEASE. Journal of Hepatology, 2014, 60, S505. | 3.7 | 3 |
| 142 | Seeding Induced Pluripotent Stem Cell-Derived Neurons onto 384-Well Plates. Methods in Molecular Biology, 2019, 1994, 159-164. | 0.9 | 3 |
| 143 | Rare missense variants in Tropomyosinâ€4 (TPM4) are associated with platelet dysfunction, cytoskeletal defects, and excessive bleeding. Journal of Thrombosis and Haemostasis, 2022, 20, 478-485. | 3.8 | 3 |
| 144 | Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 678. | 6.2 | 2 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | 250. A Phase 2/3 Study of the Efficacy and Safety of Ex Vivo Gene Therapy with Lenti-D TM Lentiviral Vector for the Treatment of Cerebral Adrenoleukodystrophy. Molecular Therapy, 2016, 24, S98-S99. | 8.2 | 2 |
| 146 | Intracerebroventricular cerliponase alfa for children with CLN2 disease: Interim results from an ongoing phase 2 extension study. European Journal of Paediatric Neurology, 2017, 21, e21. | 1.6 | 2 |
| 147 | Persistent treatment effect of cerliponase alfa in children with CLN2 disease: A 3 year update from an ongoing multicenter extension study. Molecular Genetics and Metabolism, 2019, 126, S133. | 1.1 | 2 |
| 148 | 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 Parkinsonâ€™s Disease Patients. Journal of Proteome Research, 2019, 18, 1198-1207. | 3.7 | 2 |
| 149 | Disorders of Bile Acid Synthesis and Biliary Transport. , 2014, , 555-576. | | 2 |
| 150 | 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. | 1.6 | 2 |
| 151 | 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. Journal of Pediatric Gastroenterology and Nutrition, 2005, 41, 513. | 1.8 | 1 |
| 152 | Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2015, 96, 1008-1009. | 6.2 | 1 |
| 153 | High-Content Autophagy Analysis in iPSC-Derived Neurons Using Immunofluorescence. Methods in Molecular Biology, 2019, 1994, 165-174. | 0.9 | 1 |
| 154 | One centreâ€™s experience of sedation regimes for ICV and IT drug delivery in early phase paediatric clinical trials. Molecular Genetics and Metabolism, 2019, 126, S25. | 1.1 | 1 |
| 155 | Gene therapy for global brain diseases: one small step for mice, one giant leap for humans. Brain, 2020, 143, 1964-1966. | 7.6 | 1 |
| 156 | A Case Series on Genotype and Outcome of Liver Transplantation in Children with Niemann-Pick Disease Type C. Children, 2021, 8, 819. | 1.5 | 1 |
| 157 | Preparation of iPSCs for Targeted Proteomic Analysis. Methods in Molecular Biology, 2019, 1994, 131-139. | 0.9 | 1 |
| 158 | Abstract LB-145: Folliculin interacts with p0071 (Plakophilin-4) and deficiency is associated with disordered RhoA signalling, epithelial polarization and cytokinesis. , 2012, , . | | 1 |
| 159 | A survival analysis of ventricular access devices for delivery of cerliponase alfa. Journal of Neurosurgery: Pediatrics, 2022, 29, 115-121. | 1.3 | 1 |
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