Georg Friedrich Hoffmann

List of Publications by Year
in descending order

204	6,896	61984	91884
papers	citations	43 h-index	69 g-index

214	214	214	10494
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5â€²-phosphate oxidase. Human Molecular Genetics, 2005, 14, 1077-1086.	2.9	281
2	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. Nature Communications, 2016, 7, 12039.	12.8	178
3	Proposed recommendations for diagnosing and managing individuals with glutaric aciduria type I: second revision. Journal of Inherited Metabolic Disease, 2017, 40, 75-101.	3.6	173
4	Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet Journal of Rare Diseases, 2017, 12, 12.	2.7	172
5	L-2-hydroxyglutaric acidemia: A novel inherited neurometabolic disease. Annals of Neurology, 1992, 32, 66-71.	5.3	144
6	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
7	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	6.2	127
8	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. Brain, 2016, 139, 317-337.	7.6	126
9	Impaired Mitochondrial Dynamics and Mitophagy in Neuronal Models of Tuberous Sclerosis Complex. Cell Reports, 2016, 17, 1053-1070.	6.4	125
10	Prevalence of SARS-CoV-2 Infection in Children and Their Parents in Southwest Germany. JAMA Pediatrics, 2021, 175, 586.	6.2	124
11	Progressive Infantile Neurodegeneration Caused by 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency: A Novel Inborn Error of Branched-Chain Fatty Acid and Isoleucine Metabolism. Pediatric Research, 2000, 48, 852-855.	2.3	121
12	Tyrosine hydroxylase deficiency causes progressive encephalopathy and dopa-nonresponsive dystonia. Annals of Neurology, 2003, 54, S56-S65.	5.3	117
13	Biochemical hallmarks of tyrosine hydroxylase deficiency. Clinical Chemistry, 1998, 44, 1897-1904.	3.2	111
14	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	6.2	110
15	Leukotrienes: Biosynthesis, Metabolism, and Pathophysiologic Significance. Pediatric Research, 1995, 37, 1-9.	2.3	109
16	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
17	Diversity in the incidence and spectrum of organic acidemias, fatty acid oxidation disorders, and amino acid disorders in Asian countries: Selective screening vs. expanded newborn screening. Molecular Genetics and Metabolism Reports, 2018, 16, 5-10.	1.1	94
18	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	3.6	92

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19	Safety and efficacy of mTOR inhibitor treatment in patients with tuberous sclerosis complex under 2â€‰years of age â€“ a multicenter retrospective study. Orphanet Journal of Rare Diseases, 2019, 14, 96.	2.7	90
20	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 695-703.	6.2	87
21	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
22	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
23	Design principles for virtual patients: a focus group study among students. Medical Education, 2009, 43, 580-588.	2.1	81
24	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. American Journal of Human Genetics, 2016, 98, 358-362.	6.2	77
25	Contribution of Reactive Oxygen Species to 3-Hydroxyglutarate Neurotoxicity in Primary Neuronal Cultures from Chick Embryo Telencephalons. Pediatric Research, 2001, 50, 76-82.	2.3	74
26	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	6.2	73
27	Knowledge base and mini-expert platform for the diagnosis of inborn errors of metabolism. Genetics in Medicine, 2018, 20, 151-158.	2.4	67
28	Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. Molecular Genetics and Metabolism, 2019, 127, 12-22.	1.1	66
29	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
30	Newborn screening: A diseaseâ€changing intervention for glutaric aciduria type 1. Annals of Neurology, 2018, 83, 970-979.	5.3	65
31	De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28.	2.0	64
32	Improving Pediatric Basic Life Support Performance Through Blended Learning With Web-Based Virtual Patients: Randomized Controlled Trial. Journal of Medical Internet Research, 2015, 17, e162.	4.3	58
33	Î²-Ureidopropionase deficiency: A novel inborn error of metabolism discovered using NMR spectroscopy on urine. Magnetic Resonance in Medicine, 2001, 46, 1014-1017.	3.0	57
34	Iron and vitamin D levels among autism spectrum disorders children. Annals of African Medicine, 2017, 16, 186.	0.5	57
35	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
36	Maturation-Dependent Neurotoxicity of 3-Hydroxyglutaric and Glutaric Acids In Vitro : A New Pathophysiologic Approach to Glutaryl-CoA Dehydrogenase Deficiency. Pediatric Research, 2000, 47, 495-503.	2.3	54

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37	Robust and durable serological response following pediatric SARS-CoV-2 infection. Nature Communications, 2022, 13, 128.	12.8	54
38	Potentiation of 3-hydroxyglutarate neurotoxicity following induction of astrocytic iNOS in neonatal rat hippocampal cultures. European Journal of Neuroscience, 2001, 13, 2115-2122.	2.6	53
39	Newborn Screening for Vitamin B12 Deficiency in Germanyâ€"Strategies, Results, and Public Health Implications. Journal of Pediatrics, 2020, 216, 165-172.e4.	1.8	53
40	DNAJC12 deficiency: A new strategy in the diagnosis of hyperphenylalaninemias. Molecular Genetics and Metabolism, 2018, 123, 1-5.	1.1	52
41	Neonatal mortality and outcome at the end of the first year of life in early onset urea cycle disordersâ€"review and metaâ€analysis of observational studies published over more than 35 years. Journal of Inherited Metabolic Disease, 2016, 39, 219-229.	3.6	50
42	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	1.1	50
43	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	2.4	50
44	A framework to start the debate on neonatal screening policies in the EU: an Expert Opinion Document. European Journal of Human Genetics, 2014, 22, 12-17.	2.8	49
45	Allelic phenotype values: a model for genotype-based phenotype prediction in phenylketonuria. Genetics in Medicine, 2019, 21, 580-590.	2.4	48
46	Biochemical and Molecular Genetic Characteristics of the Severe Form of Tyrosine Hydroxylase Deficiency. Clinical Chemistry, 1999, 45, 2073-2078.	3.2	48
47	Linking mitochondrial dysfunction to neurodegeneration in lysosomal storage diseases. Journal of Inherited Metabolic Disease, 2017, 40, 631-640.	3.6	46
48	Defining clinical subgroups and genotypeâ€"phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
49	Ultra-orphan diseases: a quantitative analysis of the natural history of molybdenum cofactor deficiency. Genetics in Medicine, 2015, 17, 965-970.	2.4	45
50	Incidence, disease onset and short-term outcome in urea cycle disorders â€"cross-border surveillance in Germany, Austria and Switzerland. Orphanet Journal of Rare Diseases, 2017, 12, 111.	2.7	43
51	Simultaneous determination of 3-hydroxypropionic acid, methylmalonic acid and methylcitric acid in dried blood spots: Second-tier LC-MS/MS assay for newborn screening of propionic acidemia, methylmalonic acidemias and combined remethylation disorders. PLoS ONE, 2017, 12, e0184897.	2.5	43
52	Emerging role of autophagy in pediatric neurodegenerative and neurometabolic diseases. Pediatric Research, 2014, 75, 217-226.	2.3	42
53	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	2.4	42
54	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42

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55	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
56	Recessive Mutations in PCBD1 Cause a New Type of Early-Onset Diabetes. Diabetes, 2014, 63, 3557-3564.	0.6	41
57	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
58	Pressure for drug development in lysosomal storage disorders â€“ a quantitative analysis thirty years beyond the US orphan drug act. Orphanet Journal of Rare Diseases, 2015, 10, 46.	2.7	40
59	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40
60	3-Ureidopropionate contributes to the neuropathology of 3-ureidopropionase deficiency and severe propionic aciduria: A hypothesis. Journal of Neuroscience Research, 2001, 66, 666-673.	2.9	39
61	Comparison of different IRT-PAP protocols to screen newborns for cystic fibrosis in three central European populations. Journal of Cystic Fibrosis, 2014, 13, 15-23.	0.7	39
62	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	6.2	39
63	Impaired Synthesis of Lipoxygenase Products in Glutathione Synthetase Deficiency. Pediatric Research, 1994, 35, 307-310.	2.3	37
64	Long-term Outcomes of Individuals With Metabolic Diseases Identified Through Newborn Screening. Pediatrics, 2020, 146, .	2.1	37
65	Clinical course of 63 patients with neonatal onset urea cycle disorders in the years 2001â€“2013. Orphanet Journal of Rare Diseases, 2016, 11, 116.	2.7	36
66	Clinical and biochemical footprints of inherited metabolic diseases. I. Movement disorders. Molecular Genetics and Metabolism, 2019, 127, 28-30.	1.1	35
67	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	3.6	35
68	Co-expression of phenylalanine hydroxylase variants and effects of interallelic complementation on in vitro enzyme activity and genotype-phenotype correlation. Molecular Genetics and Metabolism, 2016, 117, 328-335.	1.1	33
69	Clinical characteristics of 248 patients with Krabbe disease: quantitative natural history modeling based on published cases. Genetics in Medicine, 2019, 21, 2208-2215.	2.4	33
70	Atypical (Mild) Forms of Dihydropteridine Reductase Deficiency: Neurochemical Evaluation and Mutation Detection. Pediatric Research, 1992, 32, 726-730.	2.3	32
71	Relationship between genotype, phenylalanine hydroxylase expression and in vitro activity and metabolic phenotype in phenylketonuria. Molecular Genetics and Metabolism, 2018, 125, 86-95.	1.1	31
72	Impact of newborn screening and quality of therapy on the neurological outcome in glutaric aciduria type 1: a meta-analysis. Genetics in Medicine, 2021, 23, 13-21.	2.4	30

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73	Significant prevalence of sickle cell disease in Southwest Germany: results from a birth cohort study indicate the necessity for newborn screening. Annals of Hematology, 2016, 95, 397-402.	1.8	29
74	Exome sequencing results in identification and treatment of brain dopamine-serotonin vesicular transport disease. Journal of the Neurological Sciences, 2017, 379, 296-297.	0.6	29
75	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. European Journal of Human Genetics, 2018, 26, 407-419.	2.8	29
76	Heterogeneous clinical spectrum of DNAJC12-deficient hyperphenylalaninemia: from attention deficit to severe dystonia and intellectual disability. Journal of Medical Genetics, 2018, 55, 249-253.	3.2	29
77	Carnosine metabolism in diabetes is altered by reactive metabolites. Amino Acids, 2015, 47, 2367-2376.	2.7	28
78	Bridging the information gap between isotope ratio mass spectrometry and conventional mass spectrometry. Biological Mass Spectrometry, 1994, 23, 376-378.	0.5	27
79	Syndromic intellectual disability: A new phenotype caused by an aromatic amino acid decarboxylase gene (DDC) variant. Gene, 2015, 559, 144-148.	2.2	27
80	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	11.4	26
81	Ultraâ€orphan lysosomal storage diseases: A crossâ€sectional quantitative analysis of the natural history of alphaâ€mannosidosis. Journal of Inherited Metabolic Disease, 2019, 42, 975-983.	3.6	26
82	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26
83	High throughput newborn screening for aromatic ÊŸâ€aminoâ€acid decarboxylase deficiency by analysis of concentrations of 3â€Oâ€methyldopa from dried blood spots. Journal of Inherited Metabolic Disease, 2020, 43, 602-610.	3.6	26
84	Extended diagnosis of purine and pyrimidine disorders from urine: LC MS/MS assay development and clinical validation. PLoS ONE, 2019, 14, e0212458.	2.5	25
85	Newborn screening for remethylation disorders and vitamin B12 deficiency-evaluation of new strategies in cohorts from Qatar and Germany. World Journal of Pediatrics, 2017, 13, 136-143.	1.8	24
86	A cross-sectional quantitative analysis of the natural history of Farber disease: an ultra-orphan condition with rheumatologic and neurological cardinal disease features. Genetics in Medicine, 2018, 20, 524-530.	2.4	24
87	Mutation analysis in glycogen storage disease type 1 non-a. Human Genetics, 2000, 107, 285-289.	3.8	23
88	Exploring the validity and reliability of a questionnaire for evaluating virtual patient design with a special emphasis on fostering clinical reasoning. Medical Teacher, 2015, 37, 775-782.	1.8	23
89	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23
90	CRISPR RNA-guided FokI nucleases repair a PAH variant in a phenylketonuria model. Scientific Reports, 2016, 6, 35794.	3.3	22

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91	Defects in amino acid catabolism and the urea cycle. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1755-1773.	1.8	21
92	Quantitative natural history characterization in a cohort of 142 published cases of patients with galactosialidosisâ€"A crossâ€sectional study. Journal of Inherited Metabolic Disease, 2019, 42, 295-302.	3.6	21
93	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	12.8	21
94	Hereditary Orotic Aciduria with Epilepsy and without Megaloblastic Anemia. Neuropediatrics, 2015, 46, 123-125.	0.6	20
95	Novel Treatments for Rare Cancers: The U.S. Orphan Drug Act Is Deliveringâ€"A Cross-Sectional Analysis. Oncologist, 2016, 21, 487-493.	3.7	20
96	Electronic assessment of clinical reasoning in clerkships: A mixed-methods comparison of long-menu key-feature problems with context-rich single best answer questions. Medical Teacher, 2017, 39, 476-485.	1.8	20
97	Newborn screening for severe combined immunodeficiency using a novel and simplified method to measure T-cell excision circles (TREC). Clinical Immunology, 2017, 175, 51-55.	3.2	20
98	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	4.1	20
99	Newborn Screening for Vitamin B6 Non-responsive Classical Homocystinuria: Systematical Evaluation of a Two-Tier Strategy. JIMD Reports, 2016, 32, 87-94.	1.5	19
100	Pharmacologic rescue of hyperammonemia-induced toxicity in zebrafish by inhibition of ornithine aminotransferase. PLoS ONE, 2018, 13, e0203707.	2.5	19
101	Assessment of methylcitrate and methylcitrate to citrate ratio in dried blood spots as biomarkers for inborn errors of propionate metabolism. Scientific Reports, 2019, 9, 12366.	3.3	19
102	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	2.4	19
103	Alanine Aminotransferase Elevation in Obese Infants and Children: A Marker of Early Onset Non Alcoholic Fatty Liver Disease. Hepatitis Monthly, 2014, 14, e14112.	0.2	18
104	50 years of newborn screening. Journal of Inherited Metabolic Disease, 2014, 37, 163-164.	3.6	18
105	Ten Years after the International Committee of Medical Journal Editorsâ€™ Clinical Trial Registration Initiative, One Quarter of Phase 3 Pediatric Epilepsy Clinical Trials Still Remain Unpublished: A Cross Sectional Analysis. PLoS ONE, 2016, 11, e0144973.	2.5	18
106	Identification of SLC20A1 and SLC15A4 among other genes as potential risk factors for combined pituitary hormone deficiency. Genetics in Medicine, 2018, 20, 728-736.	2.4	18
107	Newborn screening and disease variants predict neurological outcome in isovaleric aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 857-870.	3.6	18
108	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	3.6	17

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109	Newborn Screening Programmes in Europe, Arguments and Efforts Regarding Harmonisation: Focus on Organic Acidurias. JIMD Reports, 2016, 32, 105-115.	1.5	17
110	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	2.5	17
111	Clinical spectrum and treatment outcome of 95 children with continuous spikes and waves during sleep (CSWS). European Journal of Paediatric Neurology, 2021, 30, 121-127.	1.6	17
112	Phenotypic diversity, disease progression, and pathogenicity of <scp>MVK</scp> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	3.6	17
113	Health Outcomes of Infants with Vitamin B12 Deficiency Identified by Newborn Screening and Early Treated. Journal of Pediatrics, 2021, 235, 42-48.	1.8	17
114	Thirty Years of Orphan Drug Legislation and the Development of Drugs to Treat Rare Seizure Conditions: A Cross Sectional Analysis. PLoS ONE, 2016, 11, e0161660.	2.5	17
115	Diagnosis and therapeutic monitoring of inborn errors of creatine metabolism and transport using liquid chromatographyâ€“tandem mass spectrometry in urine, plasma and CSF. Gene, 2014, 538, 188-194.	2.2	16
116	Quantification of methylcitrate in dried urine spots by liquid chromatography tandem mass spectrometry for the diagnosis of propionic and methylmalonic acidemias. Clinica Chimica Acta, 2018, 487, 41-45.	1.1	16
117	High incidence of maternal vitamin B12 deficiency detected by newborn screening: first results from a study for the evaluation of 26 additional target disorders for the German newborn screening panel. World Journal of Pediatrics, 2018, 14, 470-481.	1.8	16
118	Bioenergetic dysfunction in a zebrafish model of acute hyperammonemic decompensation. Experimental Neurology, 2019, 314, 91-99.	4.1	16
119	Vitamin B12 Deficiency in Newborns and their Mothersâ€”Novel Approaches to Early Detection, Treatment and Prevention of a Global Health Issue. Current Medical Science, 2020, 40, 801-809.	1.8	16
120	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	3.6	16
121	Detection of 3-O-methyldopa in dried blood spots for neonatal diagnosis of aromatic L-amino-acid decarboxylase deficiency: The northeastern Italian experience. Molecular Genetics and Metabolism, 2021, 133, 56-62.	1.1	16
122	Dopamine-Responsive Growth-Hormone Deficiency and Central Hypothyroidism in Sepiapterin Reductase Deficiency. JIMD Reports, 2015, 24, 109-113.	1.5	15
123	Human heterologous liver cells transiently improve hyperammonemia and ureagenesis in individuals with severe urea cycle disorders. Journal of Inherited Metabolic Disease, 2018, 41, 81-90.	3.6	15
124	In vivo High-Content Screening in Zebrafish for Developmental Nephrotoxicity of Approved Drugs. Frontiers in Cell and Developmental Biology, 2020, 8, 583.	3.7	15
125	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. JIMD Reports, 2018, 44, 1-7.	1.5	15
126	Point shear wave elastography (pSWE) using Acoustic Radiation Force Impulse (ARFI) imaging: a feasibility study and norm values for renal parenchymal stiffness in healthy children and adolescents. Medical Ultrasonography, 2017, 19, 366.	0.8	15

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127	A product of immunoreactive trypsinogen and pancreatitis-associated protein as second-tier strategy in cystic fibrosis newborn screening. Journal of Cystic Fibrosis, 2016, 15, 752-758.	0.7	14
128	Analysis of the functional muscleâ€“bone unit of the forearm in patients with phenylketonuria by peripheral quantitative computed tomography. Journal of Inherited Metabolic Disease, 2017, 40, 219-226.	3.6	14
129	A cross-sectional quantitative analysis of the natural history of free sialic acid storage diseaseâ€”an ultra-orphan multisystemic lysosomal storage disorder. Genetics in Medicine, 2019, 21, 347-352.	2.4	14
130	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
131	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	2.5	14
132	Severity-adjusted evaluation of newborn screening on the metabolic disease course in individuals with cytosolic urea cycle disorders. Molecular Genetics and Metabolism, 2020, 131, 390-397.	1.1	14
133	Critical appraisal of genotype assessment in molybdenum cofactor deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 801-811.	3.6	13
134	Defining the hidden evidence in autism research. Forty per cent of rigorously designed clinical trials remain unpublished â€•a crossâ€sectional analysis. International Journal of Methods in Psychiatric Research, 2017, 26, .	2.1	13
135	Impact of interventional and nonâ€interventional variables on anthropometric longâ€term development in glutaric aciduria type 1: A national prospective multiâ€centre study. Journal of Inherited Metabolic Disease, 2021, 44, 629-638.	3.6	13
136	Brain <scp>MR</scp> patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients. Journal of Inherited Metabolic Disease, 2021, 44, 1070-1082.	3.6	13
137	A novel autosomal recessive TERT T1129P mutation in a dyskeratosis congenita family leads to cellular senescence and loss of CD34+ hematopoietic stem cells not reversible by mTOR-inhibition. Aging, 2015, 7, 911-927.	3.1	13
138	Pediatric in-hospital emergencies: real life experiences, previous training and the need for training among physicians and nurses. BMC Research Notes, 2019, 12, 19.	1.4	12
139	Novel variants and clinical symptoms in four new ALG3â€CDG patients, review of the literature, and identification of AAGRPâ€ALG3 as a novel ALG3 variant with alanine and glycineâ€rich Nâ€terminus. Human Mutation, 2019, 40, 938-951.	2.5	12
140	QDPR homologues in Danio rerio regulate melanin synthesis, early gliogenesis, and glutamine homeostasis. PLoS ONE, 2019, 14, e0215162.	2.5	12
141	Glycogen accumulation, central carbon metabolism, and aging of hematopoietic stem and progenitor cells. Scientific Reports, 2020, 10, 11597.	3.3	12
142	Chronic hyperammonemia causes a hypoglutamatergic and hyperGABAergic metabolic state associated with neurobehavioral abnormalities in zebrafish larvae. Experimental Neurology, 2020, 331, 113330.	4.1	12
143	Implementing a tracking system for confirmatory diagnostic results after positive newborn screening for cystic fibrosisâ€”implications for process quality and patient care. European Journal of Pediatrics, 2021, 180, 1145-1155.	2.7	12
144	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Scientific Reports, 2020, 10, 11948.	3.3	11

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145	A Global Cndp1-Knock-Out Selectively Increases Renal Carnosine and Anserine Concentrations in an Age- and Gender-Specific Manner in Mice. International Journal of Molecular Sciences, 2020, 21, 4887.	4.1	11
146	Lower plasma cholesterol, LDL-cholesterol and LDL-lipoprotein subclasses in adult phenylketonuria (PKU) patients compared to healthy controls: results of NMR metabolomics investigation. Orphanet Journal of Rare Diseases, 2020, 15, 61.	2.7	11
147	A cross-sectional controlled developmental study of neuropsychological functions in patients with glutaric aciduria type I. Orphanet Journal of Rare Diseases, 2015, 10, 163.	2.7	10
148	Targeted cerebrospinal fluid analysis for inborn errors of metabolism on an LCâ€MS/MS analysis platform. Journal of Inherited Metabolic Disease, 2020, 43, 712-725.	3.6	10
149	Transient elastography correlated to four different histological fibrosis scores in children with liver disease. European Journal of Pediatrics, 2021, 180, 2237-2244.	2.7	10
150	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	3.8	10
151	Clinical Research in Vulnerable Populations: Variability and Focus of Institutional Review Boardsâ€™ Responses. PLoS ONE, 2015, 10, e0135997.	2.5	9
152	Publication status of completed registered studies in paediatric appendicitis: a cross-sectional analysis. BMJ Open, 2018, 8, e021684.	1.9	9
153	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	2.5	9
154	The biochemical subtype is a predictor for cognitive function in glutaric aciduria type 1: a national prospective follow-up study. Scientific Reports, 2021, 11, 19300.	3.3	9
155	Complementing the phenotypical spectrum of TUBA1A tubulinopathy and its role in early-onset epilepsies. European Journal of Human Genetics, 2022, 30, 298-306.	2.8	9
156	Recurrent Stroke-Like Episodes in FBXL4-Associated Early-Onset Mitochondrial Encephalomyopathy. Pediatric Neurology, 2015, 53, 549-550.	2.1	8
157	An Assessment of Publication Status of Pediatric Liver Transplantation Studies. PLoS ONE, 2016, 11, e0168251.	2.5	8
158	ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e49-e49.	7.6	8
159	Semi-quantitative detection of a vanillactic acid/vanillylmandelic acid ratio in urine is a reliable diagnostic marker for aromatic L-amino acid decarboxylase deficiency. Molecular Genetics and Metabolism, 2020, 131, 163-170.	1.1	8
160	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	2.4	8
161	1H-NMR-based metabolic profiling identifies non-invasive diagnostic and predictive urinary fingerprints in 5q spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2021, 16, 441.	2.7	8
162	Generation of an induced pluripotent stem cell (iPSC) line, DHMCi005-A, from a patient with CALFAN syndrome due to mutations in SCYL1. Stem Cell Research, 2019, 37, 101428.	0.7	7

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163	Identification of <i>Transient Receptor Potential Channel 4-Associated Protein</i> as a Novel Candidate Gene Causing Congenital Primary Hypothyroidism. Hormone Research in Paediatrics, 2020, 93, 16-29.	1.8	7
164	Next-generation sequencing diagnostics of bacteremia in pediatric sepsis. Medicine (United States), 2021, 100, e26403.	1.0	7
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