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	Quantitative retrospective natural history modeling of WDR45-related developmental and epileptic encephalopathy â€“ a systematic cross-sectional analysis of 160 published cases. Autophagy, 2022, 18, 1715-1727.	9.1	5
2	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
3	Robust and durable serological response following pediatric SARS-CoV-2 infection. Nature Communications, 2022, 13, 128.	12.8	54
4	Î²-Ureidopropionase deficiency due to novel and rare UPB1 mutations affecting pre-mRNA splicing and protein structural integrity and catalytic activity. Molecular Genetics and Metabolism, 2022, 136, 177-185.	1.1	3
5	The calcium-sensing receptor stabilizes podocyte function in proteinuric humans and mice. Kidney International, 2022, 101, 1186-1199.	5.2	6
6	Sudden neonatal death in individuals with medium-chain acyl-coenzyme A dehydrogenase deficiency: limit of newborn screening. European Journal of Pediatrics, 2022, 181, 2415-2422.	2.7	3
7	Second-tier strategies in newborn screening â€“ potential and limitations. Medizinische Genetik, 2022, 34, 21-28.	0.2	2
8	From newborn screening to genomic medicine: challenges and suggestions on how to incorporate genomic newborn screening in public health programs. Medizinische Genetik, 2022, 34, 13-20.	0.2	2
9	Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. Annals of Neurology, 2022, 92, 292-303.	5.3	3
10	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
11	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	2.4	8
12	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
13	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
14	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
15	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	3.6	16
16	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
17	Newborn screening and disease variants predict neurological outcome in isovaleric aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 857-870.	3.6	18
18	De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28.	2.0	64

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19	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	4.1	20
20	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
21	Genomic newborn screening: Proposal of a <scp>twoâ€stage</scp> approach. Journal of Inherited Metabolic Disease, 2021, 44, 518-520.	3.6	6
22	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
23	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
24	Prevalence of SARS-CoV-2 Infection in Children and Their Parents in Southwest Germany. JAMA Pediatrics, 2021, 175, 586.	6.2	124
25	Next-generation sequencing diagnostics of bacteremia in pediatric sepsis. Medicine (United States), 2021, 100, e26403.	1.0	7
26	Maternal vitamin deficiency mimicking multiple acyl-CoA dehydrogenase deficiency on newborn screening. Molecular Genetics and Metabolism Reports, 2021, 27, 100738.	1.1	1
27	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	2.5	9
28	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	3.6	7
29	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	3.8	10
30	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
31	A Novel UPLC-MS/MS Method Identifies Organ-Specific Dipeptide Profiles. International Journal of Molecular Sciences, 2021, 22, 9979.	4.1	7
32	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
33	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	12.8	21
34	Optimized Trientineâ€dihydrochloride Therapy in Pediatric Patients With Wilson Disease. Journal of Pediatric Gastroenterology and Nutrition, 2021, 72, 115-122.	1.8	6
35	Differences of Phenylalanine Concentrations in Dried Blood Spots and in Plasma: Erythrocytes as a Neglected Component for This Observation. Metabolites, 2021, 11, 680.	2.9	3
36	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

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37	An Integrated clinical pathway for diagnosis, treatment and care of rare diseases: model, operating procedures, and results of the project TRANSLATE-NAMSE funded by the German Federal Joint Committee. Orphanet Journal of Rare Diseases, 2021, 16, 474.	2.7	7
38	The Biochemical High Excreter Phenotype Is the Major Risk Factor for Cognitive Impairment in Early Diagnosed Individuals with Glutaric Aciduria Type 1. Neuropediatrics, 2021, 52, .	0.6	0
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	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
41	Defining clinical subgroups and genotypeâ€"phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
42	Targeted Metabolomic Profiling of Total Fatty Acids in Human Plasma by Liquid Chromatography-Tandem Mass Spectrometry. Metabolites, 2020, 10, 400.	2.9	5
43	Long-term Outcomes of Individuals With Metabolic Diseases Identified Through Newborn Screening. Pediatrics, 2020, 146, .	2.1	37
44	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Scientific Reports, 2020, 10, 11948.	3.3	11
45	Glycogen accumulation, central carbon metabolism, and aging of hematopoietic stem and progenitor cells. Scientific Reports, 2020, 10, 11597.	3.3	12
46	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
47	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
48	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
49	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
50	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
51	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23
52	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
53	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
54	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

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55	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
56	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
57	Axenfeld-Rieger Anomaly and Neuropsychiatric Problemsâ€"More than Meets the Eye. Neuropediatrics, 2020, 51, 192-197.	0.6	5
58	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	2.5	14
59	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
60	Primary carnitine deficiency â€" diagnosis after heart transplantation: better late than never!. Orphanet Journal of Rare Diseases, 2020, 15, 87.	2.7	4
61	Chronic hyperammonemia causes a hypoglutamatergic and hyperGABAergic metabolic state associated with neurobehavioral abnormalities in zebrafish larvae. Experimental Neurology, 2020, 331, 113330.	4.1	12
62	FDA orphan drug designations for lysosomal storage disorders â€" a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	2.5	17
63	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
64	FDA orphan drug designations for lysosomal storage disorders â€" a cross-sectional analysis. , 2020, 15, e0230898.		0
65	FDA orphan drug designations for lysosomal storage disorders â€" a cross-sectional analysis. , 2020, 15, e0230898.		0
66	FDA orphan drug designations for lysosomal storage disorders â€" a cross-sectional analysis. , 2020, 15, e0230898.		0
67	FDA orphan drug designations for lysosomal storage disorders â€" a cross-sectional analysis. , 2020, 15, e0230898.		0
68	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
69	Allelic phenotype values: a model for genotype-based phenotype prediction in phenylketonuria. Genetics in Medicine, 2019, 21, 580-590.	2.4	48
70	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
71	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
72	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26

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73	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
74	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
75	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
76	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
77	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
78	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
79	QDPR homologues in Danio rerio regulate melanin synthesis, early gliogenesis, and glutamine homeostasis. PLoS ONE, 2019, 14, e0215162.	2.5	12
80	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
81	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
82	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
83	High blood pressure, a red flag for the neonatal manifestation of urea cycle disorders. Orphanet Journal of Rare Diseases, 2019, 14, 80.	2.7	4
84	Clinical and biochemical footprints of inherited metabolic diseases. I. Movement disorders. Molecular Genetics and Metabolism, 2019, 127, 28-30.	1.1	35
85	Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. Molecular Genetics and Metabolism, 2019, 127, 12-22.	1.1	66
86	Bioenergetic dysfunction in a zebrafish model of acute hyperammonemic decompensation. Experimental Neurology, 2019, 314, 91-99.	4.1	16
87	Neurometabolic hereditary diseases of adults. Journal of Inherited Metabolic Disease, 2019, 42, 389-389.	3.6	2
88	Generation of an iPSC line from a patient with infantile liver failure syndrome 2 due to mutations in NBAS: DHMCi004-A. Stem Cell Research, 2019, 35, 101398.	0.7	1
89	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
90	AB1029â€…FIBRODYSPLASIA OSSIFICANS PROGRESSIVA: A CHALLENGE TO DIAGNOSE AND TO TREAT. , 2019, , .		0

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91	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.	3.6	4
92	Risk Factors for Childhood Overweight and Obesity in Ukraine and Germany. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 247-252.	0.9	7
93	Urea Cycle Disorders in the US and Europe â€" Evidence-based Clinical Outcomes Derived from Two Decades of Experience with Prospective Registry Studies. , 2019, 50, .		0
94	Newborn screening: A diseaseâ€changing intervention for glutaric aciduria type 1. Annals of Neurology, 2018, 83, 970-979.	5.3	65
95	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
96	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
97	Knowledge base and mini-expert platform for the diagnosis of inborn errors of metabolism. Genetics in Medicine, 2018, 20, 151-158.	2.4	67
98	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
99	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
100	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
101	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
102	Pharmacologic rescue of hyperammonemia-induced toxicity in zebrafish by inhibition of ornithine aminotransferase. PLoS ONE, 2018, 13, e0203707.	2.5	19
103	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
104	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
105	Cross-sectional analysis on publication status and age representation of clinical studies addressing mechanical ventilation and ventilator-induced lung injury in infants and children. BMJ Open, 2018, 8, e023524.	1.9	4
106	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
107	ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e49-e49.	7.6	8
108	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

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109	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
110	Endothelial progenitor cells accelerate endothelial regeneration in an in vitro model of Shigatoxin-2a-induced injury via soluble growth factors. American Journal of Physiology - Renal Physiology, 2018, 315, F861-F869.	2.7	6
111	Publication status of completed registered studies in paediatric appendicitis: a cross-sectional analysis. BMJ Open, 2018, 8, e021684.	1.9	9
112	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42
113	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
114	DNAJC12 deficiency: A new strategy in the diagnosis of hyperphenylalaninemias. Molecular Genetics and Metabolism, 2018, 123, 1-5.	1.1	52
115	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
116	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	6.2	127
117	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
118	Linking mitochondrial dysfunction to neurodegeneration in lysosomal storage diseases. Journal of Inherited Metabolic Disease, 2017, 40, 631-640.	3.6	46
119	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	2.4	42
120	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
121	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
122	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
123	Critical appraisal of genotype assessment in molybdenum cofactor deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 801-811.	3.6	13
124	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	11.4	26
125	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
126	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41

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127	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
128	Pediatric use of tetracyclines: focus on neurodevelopmental effects. Pediatric Research, 2017, 82, 725-726.	2.3	3
129	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
130	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	1.1	50
131	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
132	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
133	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
134	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
135	Iron and vitamin D levels among autism spectrum disorders children. Annals of African Medicine, 2017, 16, 186.	0.5	57
136	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
137	An Assessment of Publication Status of Pediatric Liver Transplantation Studies. PLoS ONE, 2016, 11, e0168251.	2.5	8
138	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
139	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
140	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	3.6	17
141	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
142	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
143	Newborn Screening Programmes in Europe, Arguments and Efforts Regarding Harmonisation: Focus on Organic Acidurias. JIMD Reports, 2016, 32, 105-115.	1.5	17
144	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

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145	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. Nature Communications, 2016, 7, 12039.	12.8	178
146	CRISPR RNA-guided FokI nucleases repair a PAH variant in a phenylketonuria model. Scientific Reports, 2016, 6, 35794.	3.3	22
147	Impaired Mitochondrial Dynamics and Mitophagy in Neuronal Models of Tuberous Sclerosis Complex. Cell Reports, 2016, 17, 1053-1070.	6.4	125
148	Novel treatments for rare rheumatologic disorders: analysis of the impact of 30Âyears of the US orphan drug act. Orphanet Journal of Rare Diseases, 2016, 11, 60.	2.7	6
149	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
150	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
151	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
152	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
153	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
154	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
155	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
156	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
157	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. Brain, 2016, 139, 317-337.	7.6	126
158	In vivo monitoring of urea cycle activity with 13C-acetate as a tracer of ureagenesis. Molecular Genetics and Metabolism, 2016, 117, 19-26.	1.1	5
159	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
160	The pattern of neural tube defects in a highly endogamous society: A 25-year incidence trends. Journal of Pediatric Neurology, 2015, 10, 193-198.	0.2	0
161	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
162	Variability of OTC deficiency in heterozygous carriers: Case report of a family. Journal of Pediatric Biochemistry, 2015, 04, 005-010.	0.2	0

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