Ron A Wevers

List of Publications by Year
in descending order

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541 papers	25,539 citations	5896 81 h-index	17105 122 g-index
570 all docs	570 docs citations	570 times ranked	21747 citing authors

#	Article	IF	Citations
1	Pre- versus post-operative untargeted plasma nuclear magnetic resonance spectroscopy metabolomics of pheochromocytoma and paraganglioma. Endocrine, 2022, 75, 254-265.	2.3	3
2	ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences. Brain, 2022, 145, 2602-2616.	7.6	5
3	Lactate infusion as therapeutical intervention: a scoping review. European Journal of Pediatrics, 2022, , 1.	2.7	8
4	DTYMK is essential for genome integrity and neuronal survival. Acta Neuropathologica, 2022, 143, 245-262.	7.7	11
5	How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681.	3.6	20
6	Application of metabolite set enrichment analysis on untargeted metabolomics data prioritises relevant pathways and detects novel biomarkers for inherited metabolic disorders. Journal of Inherited Metabolic Disease, 2022, 45, 682-695.	3.6	6
7	The novel P330L pathogenic variant of aromatic amino acid decarboxylase maps on the catalytic flexible loop underlying its crucial role. Cellular and Molecular Life Sciences, 2022, 79, 305.	5.4	8
8	Clinical presentation and longâ€•term followâ€•up of dopamine beta hydroxylase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 554-565.	3.6	13
9	Amadori rearrangement products as potential biomarkers for inborn errors of amino-acid metabolism. Communications Biology, 2021, 4, 367.	4.4	16
10	Peripheral decarboxylase inhibitors paradoxically induce aromatic L-amino acid decarboxylase. Npj Parkinson's Disease, 2021, 7, 29.	5.3	14
11	Cerebrotendinous xanthomatosis without neurological involvement. Journal of Internal Medicine, 2021, 290, 1039-1047.	6.0	12
12	Blood, urine and cerebrospinal fluid analysis in TH and AADC deficiency and the effect of treatment. Molecular Genetics and Metabolism Reports, 2021, 27, 100762.	1.1	3
13	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22
14	Metabolomics-Based Screening of Inborn Errors of Metabolism: Enhancing Clinical Application with a Robust Computational Pipeline. Metabolites, 2021, 11, 568.	2.9	11
15	Untargeted metabolomics and infrared ion spectroscopy identify biomarkers for pyridoxine-dependent epilepsy. Journal of Clinical Investigation, 2021, 131, .	8.2	33
16	Nicotinamide Riboside Improves Ataxia Scores and Immunoglobulin Levels in Ataxia Telangiectasia. Movement Disorders, 2021, 36, 2951-2957.	3.9	18
17	Monoamine oxidase A activity in fibroblasts as a functional confirmation of MAOA variants. JIMD Reports, 2021, 58, 114-121.	1.5	6
18	Metabolite Identification Using Infrared Ion Spectroscopyâ”€Novel Biomarkers for Pyridoxine-Dependent Epilepsy. Analytical Chemistry, 2021, 93, 15340-15348.	6.5	20

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19	Infrared ion spectroscopy: New opportunities for small-molecule identification in mass spectrometry - A tutorial perspective. Analytica Chimica Acta, 2020, 1093, 1-15.	5.4	57
20	CLPB (caseinolytic peptidase B homolog), the first mitochondrial protein refoldase associated with human disease. Biochimica Et Biophysica Acta - General Subjects, 2020, 1864, 129512.	2.4	17
21	metPropagate: network-guided propagation of metabolomic information for prioritization of metabolic disease genes. Npj Genomic Medicine, 2020, 5, 25.	3.8	13
22	A newborn screening approach to diagnose 3â€hydroxyâ€3â€methylglutarylâ€CoA lyase deficiency. JIMD Reports, 2020, 54, 79-86.	1.5	12
23	Variable Selection in Untargeted Metabolomics and the Danger of Sparsity. Metabolites, 2020, 10, 470.	2.9	5
24	Evaluation of cyclooxygenase oxylipins as potential biomarker for obesity-associated adipose tissue inflammation and type 2 diabetes using targeted multiple reaction monitoring mass spectrometry. Prostaglandins Leukotrienes and Essential Fatty Acids, 2020, 160, 102157.	2.2	21
25	Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. Genetics in Medicine, 2020, 22, 1589-1597.	2.4	19
26	A comparison of high-throughput plasma NMR protocols for comparative untargeted metabolomics. Metabolomics, 2020, 16, 64.	3.0	18
27	Novel defect in phosphatidylinositol 4â€kinase type 2â€alpha (<scp>PI4K2A</scp>) at the membraneâ€enzyme interface is associated with metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2020, 43, 1382-1391.	3.6	7
28	Confirmation of neurometabolic diagnoses using ageâ€dependent cerebrospinal fluid metabolomic profiles. Journal of Inherited Metabolic Disease, 2020, 43, 1112-1120.	3.6	16
29	Disturbed brain ether lipid metabolism and histology in <scp>SjÃ¶grenâ€Larsson</scp> syndrome. Journal of Inherited Metabolic Disease, 2020, 43, 1265-1278.	3.6	25
30	The membrane protein ANKH is crucial for bone mechanical performance by mediating cellular export of citrate and ATP. PLoS Genetics, 2020, 16, e1008884.	3.5	45
31	Title is missing!. , 2020, 16, e1008884.		0
32	Title is missing!. , 2020, 16, e1008884.		0
33	Title is missing!. , 2020, 16, e1008884.		0
34	Title is missing!. , 2020, 16, e1008884.		0
35	Movement disorders in cerebrotendinous xanthomatosis. Parkinsonism and Related Disorders, 2019, 58, 12-16.	2.2	42
36	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46

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37	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	7.6	67
38	Isocitrate dehydrogenase 1-mutated cancers are sensitive to the green tea polyphenol epigallocatechin-3-gallate. Cancer & Metabolism, 2019, 7, 4.	5.0	18
39	N-Glycosylation Defects in Humans Lower Low-Density Lipoprotein Cholesterol Through Increased Low-Density Lipoprotein Receptor Expression. Circulation, 2019, 140, 280-292.	1.6	26
40	Reference-standard free metabolite identification using infrared ion spectroscopy. International Journal of Mass Spectrometry, 2019, 443, 77-85.	1.5	32
41	The P274S Mutation of Lecithin-Cholesterol Acyltransferase (LCAT) and Its Clinical Manifestations in a Large Kindred. American Journal of Kidney Diseases, 2019, 74, 510-522.	1.9	10
42	Functional disruption of pyrimidine nucleoside transporter CNT1 results in a novel inborn error of metabolism with high excretion of uridine and cytidine. Journal of Inherited Metabolic Disease, 2019, 42, 494-500.	3.6	6
43	The GCâ€“MS metabolomics signature in patients with fibromyalgia syndrome directs to dysbiosis as an aspect contributing factor of FMS pathophysiology. Metabolomics, 2019, 15, 54.	3.0	30
44	Mutated SUCLG1 causes mislocalization of SUCLG2 protein, morphological alterations of mitochondria and an early-onset severe neurometabolic disorder. Molecular Genetics and Metabolism, 2019, 126, 43-52.	1.1	20
45	SLC13A3 variants cause acute reversible leukoencephalopathy and Î±â€ketoglutarate accumulation. Annals of Neurology, 2019, 85, 385-395.	5.3	22
46	Response to â€œLeigh-like syndrome with mild mtDNA depletion due to the SUCLG1 variant c.626C>Aâ€• Molecular Genetics and Metabolism Reports, 2019, 18, 10.	1.1	1
47	Long-term treatment effect in cerebrotendinous xanthomatosis depends on age at treatment start. Neurology, 2019, 92, e83-e95.	1.1	73
48	Evaluation of chitotriosidase as a biomarker for adipose tissue inflammation in overweight individuals and type 2 diabetic patients. International Journal of Obesity, 2019, 43, 1712-1723.	3.4	6
49	GCâ€“MS-based urinary organic acid profiling reveals multiple dysregulated metabolic pathways following experimental acute alcohol consumption. Scientific Reports, 2018, 8, 5775.	3.3	17
50	Fast and accurate quantitative organic acid analysis with LCâ€QTOF/MS facilitates screening of patients for inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2018, 41, 415-424.	3.6	37
51	Nextâ€generation metabolic screening: targeted and untargeted metabolomics for the diagnosis of inborn errors of metabolism in individual patients. Journal of Inherited Metabolic Disease, 2018, 41, 337-353.	3.6	145
52	Targeted versus untargeted omics â€” the CAFSA story. Journal of Inherited Metabolic Disease, 2018, 41, 447-456.	3.6	10
53	Mutations in CYB561 Causing a Novel Orthostatic Hypotension Syndrome. Circulation Research, 2018, 122, 846-854.	4.5	22
54	The role of the clinician in the multiâ€omics era: are you ready?. Journal of Inherited Metabolic Disease, 2018, 41, 571-582.	3.6	55

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55	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	21.4	86
56	Integration of genomics and metabolomics for prioritization of rare disease variants: a 2018 literature review. Journal of Inherited Metabolic Disease, 2018, 41, 435-445.	3.6	35
57	Think big â€" think omics. Journal of Inherited Metabolic Disease, 2018, 41, 281-283.	3.6	19
58	Unraveling the unknown areas of the human metabolome: the role of infrared ion spectroscopy. Journal of Inherited Metabolic Disease, 2018, 41, 367-377.	3.6	44
59	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	6.2	61
60	Lithium induces aerobic glycolysis and glutaminolysis in collecting duct principal cells. American Journal of Physiology - Renal Physiology, 2018, 314, F230-F239.	2.7	8
61	Autism spectrum disorder: an early and frequent feature in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2018, 41, 641-646.	3.6	19
62	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
63	Structural elucidation of novel biomarkers of known metabolic disorders based on multistage fragmentation mass spectra. Journal of Inherited Metabolic Disease, 2018, 41, 407-414.	3.6	18
64	The 1H-NMR-based metabolite profile of acute alcohol consumption: A metabolomics intervention study. PLoS ONE, 2018, 13, e0196850.	2.5	18
65	Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. American Journal of Human Genetics, 2018, 103, 125-130.	6.2	29
66	Sialic acid catabolism by N-acetylneuraminate pyruvate lyase is essential for muscle function. JCI Insight, 2018, 3, .	5.0	36
67	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
68	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. Journal of Inherited Metabolic Disease, 2017, 40, 423-431.	3.6	14
69	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	2.8	15
70	A diagnostic biomarker profile for fibromyalgia syndrome based on an NMR metabolomics study of selected patients and controls. BMC Neurology, 2017, 17, 88.	1.8	65
71	Cerebellar Disease Mimicking Cerebrotendinous Xanthomatosis: Langerhans Cell Histiocytosis. Pediatric Neurology, 2017, 73, 98-100.	2.1	2
72	MEGDEL Syndrome: Expanding the Phenotype and New Mutations. Neuropediatrics, 2017, 48, 382-384.	0.6	7

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73	Molecular identification in metabolomics using infrared ion spectroscopy. Scientific Reports, 2017, 7, 3363.	3.3	54
74	1 H NMR spectral identification of medication in cerebrospinal fluid of pediatric meningitis. Journal of Pharmaceutical and Biomedical Analysis, 2017, 143, 56-61.	2.8	5
75	SUCNR1-mediated chemotaxis of macrophages aggravates obesity-induced inflammation and diabetes. Diabetologia, 2017, 60, 1304-1313.	6.3	126
76	A newborn screening method for cerebrotendinous xanthomatosis using bile alcohol glucuronides and metabolite ratios. Journal of Lipid Research, 2017, 58, 1002-1007.	4.2	28
77	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
78	A homozygous DPM3 mutation in a patient with alpha-dystroglycan-related limb girdle muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1043-1046.	0.6	10
79	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
80	Zileuton for Pruritus in SjÃ¶gren-Larsson Syndrome: A Randomized Double-blind Placebo-controlled Crossover Trial. Acta Dermato-Venereologica, 2016, 96, 255-256.	1.3	12
81	Monitoring creatine and phosphocreatine by 13C MR spectroscopic imaging during and after 13C4 creatine loading: a feasibility study. Amino Acids, 2016, 48, 1857-1866.	2.7	4
82	A putative urinary biosignature for diagnosis and follow-up of tuberculous meningitis in children: outcome of a metabolomics study disclosing hostâ€“pathogen responses. Metabolomics, 2016, 12, 1.	3.0	16
83	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
84	Hydrogen cyanide emission in the lung by Staphylococcus aureus. European Respiratory Journal, 2016, 48, 577-579.	6.7	10
85	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
86	Expanding the phenotype in aminoacylase 1 (ACY1) deficiency: characterization of the molecular defect in a 63-year-old woman with generalized dystonia. Metabolic Brain Disease, 2016, 31, 587-592.	2.9	12
87	Metabolic risks at birth of neonates exposed in utero to HIV-antiretroviral therapy relative to unexposed neonates: an NMR metabolomics study of cord blood. Metabolomics, 2016, 12, 1.	3.0	4
88	Neurometabolic disorders. Neurology: Clinical Practice, 2016, 6, 348-357.	1.6	11
89	Fast, robust and high-resolution glycosylation profiling of intact monoclonal IgG antibodies using nanoLC-chip-QTOF. Clinica Chimica Acta, 2016, 461, 90-97.	1.1	20
90	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110

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91	Identification of a novel inactivating mutation in Isocitrate Dehydrogenase 1 (IDH1-R314C) in a high grade astrocytoma. Scientific Reports, 2016, 6, 30486.	3.3	11
92	ALG6â€CDG: a recognizable phenotype with epilepsy, proximal muscle weakness, ataxia and behavioral and limb anomalies. Journal of Inherited Metabolic Disease, 2016, 39, 713-723.	3.6	36
93	Cytosolic phosphoenolpyruvate carboxykinase deficiency presenting with acute liver failure following gastroenteritis. Molecular Genetics and Metabolism, 2016, 118, 21-27.	1.1	23
94	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
95	Identification of Pseudomonas aeruginosa and Aspergillus fumigatus mono- and co-cultures based on volatile biomarker combinations. Journal of Breath Research, 2016, 10, 016002.	3.0	46
96	TMEM199 Deficiency Is a Disorder of Golgi Homeostasis Characterized by Elevated Aminotransferases, Alkaline Phosphatase, and Cholesterol and Abnormal Glycosylation. American Journal of Human Genetics, 2016, 98, 322-330.	6.2	73
97	Lactate and its many faces. European Journal of Paediatric Neurology, 2016, 20, 3-10.	1.6	38
98	Detection of Staphylococcus aureus in cystic fibrosis patients using breath VOC profiles. Journal of Breath Research, 2016, 10, 046014.	3.0	42
99	Trimethylaminuria, Dimethylglycine Dehydrogenase Deficiency and Disorders in the Metabolism of Glutathione. , 2016, , 429-437.		0
100	A Multiplex Assay for the Diagnosis of Mucopolysaccharidoses and Mucolipidoses. PLoS ONE, 2015, 10, e0138622.	2.5	35
101	Molecular Characterization of Testicular Adrenal Rest Tumors in Congenital Adrenal Hyperplasia: Lesions With Both Adrenocortical and Leydig Cell Features. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E524-E530.	3.6	59
102	Genotype-Specific Differences in the Tumor Metabolite Profile of Pheochromocytoma and Paraganglioma Using Untargeted and Targeted Metabolomics. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E214-E222.	3.6	33
103	Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids. Journal of Inherited Metabolic Disease, 2015, 38, 99-110.	3.6	47
104	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	6.2	111
105	A hypothetical astrocyteâ€“microglia lactate shuttle derived from a 1H NMR metabolomics analysis of cerebrospinal fluid from a cohort of South African children with tuberculous meningitis. Metabolomics, 2015, 11, 822-837.	3.0	46
106	Quantitative Measurement of Immunoglobulins and Free Light Chains Using Mass Spectrometry. Analytical Chemistry, 2015, 87, 8268-8274.	6.5	27
107	Regularized MANOVA (rMANOVA) in untargeted metabolomics. Analytica Chimica Acta, 2015, 899, 1-12.	5.4	59
108	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.6	34

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109	Absence of Î±- and Î²-dystroglycan is associated with Walker-Warburg syndrome. Neurology, 2015, 84, 2177-2182.	1.1	40
110	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. European Journal of Human Genetics, 2015, 23, 1051-1061.	2.8	24
111	SUCLA2 Deficiency: A Deafness-Dystonia Syndrome with Distinctive Metabolic Findings (Report of a) Tj ETQq1 1 0.784314 rgBT /Over	1.5	8
112	High-resolution mass spectrometry glycoprofiling of intact transferrin for diagnosis and subtype identification in the congenital disorders of glycosylation. Translational Research, 2015, 166, 639-649.e1.	5.0	73
113	Cerebral lipid accumulation in Chanarinâ€“Dorfman Syndrome. Molecular Genetics and Metabolism, 2015, 114, 51-54.	1.1	18
114	Volatile organic compounds emitted bypseudomonas aeruginosaandaspergillus fumigatusmono-cultures and in co-culture. , 2015, , .		0
115	Hypercholesterolaemia and hepatosplenomegaly: two manifestations of cholesteryl ester storage disease. Netherlands Journal of Medicine, 2015, 73, 129-32.	0.5	9
116	Longâ€term clinical outcome, therapy and mild mitochondrial dysfunction in hyperprolinemia. Journal of Inherited Metabolic Disease, 2014, 37, 383-390.	3.6	26
117	Single point mutation in Rabenosyn-5 in a female with intractable seizures and evidence of defective endocytotic trafficking. Orphanet Journal of Rare Diseases, 2014, 9, 141.	2.7	26
118	Early life adversity and serotonin transporter gene variation interact at the level of the adrenal gland to affect the adult hypothalamo-pituitary-adrenal axis. Translational Psychiatry, 2014, 4, e409-e409.	4.8	49
119	Clinical and biochemical features guiding the diagnostics in neurometabolic cutis laxa. European Journal of Human Genetics, 2014, 22, 888-895.	2.8	34
120	A novel phenotype associated with cutis laxa, abnormal fat distribution, cardiomyopathy and cataract. American Journal of Medical Genetics, Part A, 2014, 164, 1049-1055.	1.2	5
121	Characterization of acute myeloid leukemia based on levels of global hydroxymethylation. Blood, 2014, 124, 1110-1118.	1.4	80
122	Cutis laxa, fat pads and retinopathy due to ALDH18A1 mutation and review of the literature. European Journal of Paediatric Neurology, 2014, 18, 511-515.	1.6	33
123	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	7.6	41
124	Correlation Between In Vivo ¹⁸F-FDG PET and Immunohistochemical Markers of Glucose Uptake and Metabolism in Pheochromocytoma and Paraganglioma. Journal of Nuclear Medicine, 2014, 55, 1253-1259.	5.0	67
125	Leucine Loading Test is Only Discriminative for 3-Methylglutaconic Aciduria Due to AUH Defect. JIMD Reports, 2014, 16, 1-6.	1.5	17
126	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	27.0	236

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127	Alpha-fetoprotein, a fascinating protein and biomarker in neurology. European Journal of Paediatric Neurology, 2014, 18, 243-248.	1.6	57
128	Towards the Disease Biomarker in an Individual Patient Using Statistical Health Monitoring. PLoS ONE, 2014, 9, e92452.	2.5	25
129	Proton NMR Spectroscopy of Body Fluids. , 2014, , 795-801.		0
130	Lipid Profiling in Health and Disease. , 2013, , 317-332.		2
131	3â€Methylglutaconic aciduriaâ€"lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	3.6	74
132	Inborn errors of metabolism with 3â€methylglutaconic aciduria as discriminative feature: proper classification and nomenclature. Journal of Inherited Metabolic Disease, 2013, 36, 923-928.	3.6	84
133	Increased mitochondrial activity in a novel IDH1-R132H mutant human oligodendroglioma xenograft model: in situ detection of 2-HG and Î±-KG. Acta Neuropathologica Communications, 2013, 1, 18.	5.2	54
134	Disclosure of a putative biosignature for respiratory chain disorders through a metabolomics approach. Metabolomics, 2013, 9, 379-391.	3.0	25
135	A novel mutation in COQ2 leading to fatal infantile multisystem disease. Journal of the Neurological Sciences, 2013, 326, 24-28.	0.6	45
136	Thrombotic complications in patients with PMM2-CDG. Molecular Genetics and Metabolism, 2013, 109, 107-111.	1.1	44
137	Perinatal and early infantile symptoms in congenital disorders of glycosylation. American Journal of Medical Genetics, Part A, 2013, 161, 578-584.	1.2	39
138	From discrete dilated cardiomyopathy to successful cardiac transplantation in congenital disorders of glycosylation due to dolichol kinase deficiency (DK1-CDG). Heart Failure Reviews, 2013, 18, 187-196.	3.9	36
139	Symptomatic lipid storage in carriers for the PNPLA2 gene. European Journal of Human Genetics, 2013, 21, 807-815.	2.8	25
140	Child Neurology: Differential diagnosis of a low CSF glucose in children and young adults. Neurology, 2013, 81, e178-81.	1.1	18
141	Genotype-Specific Abnormalities in Mitochondrial Function Associate with Distinct Profiles of Energy Metabolism and Catecholamine Content in Pheochromocytoma and Paraganglioma. Clinical Cancer Research, 2013, 19, 3787-3795.	7.0	53
142	Intellectual disability and bleeding diathesis due to deficient CMPâ€"sialic acid transport. Neurology, 2013, 81, 681-687.	1.1	42
143	Cerebrospinal Fluid Analysis in the Workup of GLUT1 Deficiency Syndrome. JAMA Neurology, 2013, 70, 1440.	9.0	106
144	Novel proton MR spectroscopy findings in adenylosuccinate lyase deficiency. Journal of Magnetic Resonance Imaging, 2013, 37, 974-980.	3.4	7

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145	Phenotypic variability in a dystonia family with mutations in the manganese transporter gene. Movement Disorders, 2013, 28, 685-686.	3.9	9
146	Evidence for treatable inborn errors of metabolism in a cohort of 187 Greek patients with autism spectrum disorder (ASD). Frontiers in Human Neuroscience, 2013, 7, 858.	2.0	53
147	Aberrant 5-Hydroxymethylcytosine Levels Correlate With Poor Overall Survival In Acute Myeloid Leukemia. Blood, 2013, 122, 1261-1261.	1.4	1
148	Optimized Metabolomic Approach to Identify Uremic Solutes in Plasma of Stage 3â€“4 Chronic Kidney Disease Patients. PLoS ONE, 2013, 8, e71199.	2.5	55
149	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
150	Glucose Transporter-1 (GLUT1) Deficiency Syndrome: Diagnosis and Treatment in Late Childhood. Neuropediatrics, 2012, 43, 168-171.	0.6	14
151	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
152	Defining the Phenotype in Congenital Disorder of Glycosylation Due to ALG1 Mutations. Pediatrics, 2012, 130, e1034-e1039.	2.1	35
153	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081.	6.2	159
154	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	5.3	121
155	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	3.8	73
156	SjÃ¶grenâ€“Larsson syndrome in clinical practice. Journal of Inherited Metabolic Disease, 2012, 35, 955-962.	3.6	48
157	Acid sphingomyelinase (Asm) deficiency patients in The Netherlands and Belgium: Disease spectrum and natural course in attenuated patients. Molecular Genetics and Metabolism, 2012, 107, 526-533.	1.1	71
158	Mitochondrial DNA m.3242GÂ>ÂA mutation, an under diagnosed cause of hypertrophic cardiomyopathy and renal tubular dysfunction?. European Journal of Medical Genetics, 2012, 55, 552-556.	1.3	27
159	Thyroid function in PMM2-CDG: Diagnostic approach and proposed management. Molecular Genetics and Metabolism, 2012, 105, 681-683.	1.1	12
160	Longâ€term outcome in pyridoxineâ€dependent epilepsy. Developmental Medicine and Child Neurology, 2012, 54, 849-854.	2.1	93
161	Identification of Novel Translational Urinary Biomarkers for Acetaminophen-Induced Acute Liver Injury Using Proteomic Profiling in Mice. PLoS ONE, 2012, 7, e49524.	2.5	10
162	Glycosylation defects underlying fetal alcohol spectrum disorder: a novel pathogenetic model. Journal of Inherited Metabolic Disease, 2012, 35, 399-405.	3.6	16

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163	Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10 , a Manganese Transporter in Man. American Journal of Human Genetics, 2012, 90, 457-466.	6.2	321
164	The 3â€methylglutaconic acidurias: what's new?. Journal of Inherited Metabolic Disease, 2012, 35, 13-22.	3.6	72
165	Cerebrospinal Fluid Glucose and Lactate: Age-Specific Reference Values and Implications for Clinical Practice. PLoS ONE, 2012, 7, e42745.	2.5	109
166	Trimethylaminuria and Dimethylglycine Dehydrogenase Deficiency. , 2012, , 431-435.		1
167	Heterozygosity for a Loss-of-Function Mutation in GALNT2 Improves Plasma Triglyceride Clearance in Man. Cell Metabolism, 2011, 14, 811-818.	16.2	91
168	Clinical and diagnostic approach in unsolved CDG patients with a type 2 transferrin pattern. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 691-698.	3.8	25
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