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	The frequency of lysosomal storage diseases in The Netherlands. Human Genetics, 1999, 105, 151-156.	3.8	615
2	Performance of near-infrared spectroscopy in measuring local O₂ consumption and blood flow in skeletal muscle. Journal of Applied Physiology, 2001, 90, 511-519.	2.5	477
3	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	7.6	356
4	Mitochondrial creatine kinase: a key enzyme of aerobic energy metabolism. Biochimica Et Biophysica Acta - Bioenergetics, 1992, 1102, 119-166.	1.0	344
5	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	21.4	330
6	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
7	Smith-Lemli-Opitz Syndrome Is Caused by Mutations in the 7-Dehydrocholesterol Reductase Gene. American Journal of Human Genetics, 1998, 63, 329-338.	6.2	271
8	Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis. Brain, 2010, 133, 1810-1822.	7.6	268
9	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
10	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	28.9	253
11	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	27.0	236
12	Folate Receptor Alpha Defect Causes Cerebral Folate Transport Deficiency: A Treatable Neurodegenerative Disorder Associated with Disturbed Myelin Metabolism. American Journal of Human Genetics, 2009, 85, 354-363.	6.2	228
13	Clinical and molecular genetic characteristics of patients with cerebrotendinous xanthomatosis. Brain, 2000, 123, 908-919.	7.6	219
14	Adipose tissue thickness affects in vivo quantitative near-IR spectroscopy in human skeletal muscle. Clinical Science, 2001, 101, 21-28.	4.3	213
15	Dimethylmethylene blue-based spectrophotometry of glycosaminoglycans in untreated urine: a rapid screening procedure for mucopolysaccharidoses.. Clinical Chemistry, 1989, 35, 1472-1477.	3.2	197
16	Adipose tissue thickness affects in vivo quantitative near-IR spectroscopy in human skeletal muscle. Clinical Science, 2001, 101, 21.	4.3	189
17	Elevated plasma chitotriosidase activity in various lysosomal storage disorders. Journal of Inherited Metabolic Disease, 1995, 18, 717-722.	3.6	186
18	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain, 2007, 130, 862-874.	7.6	180

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19	Deficiency of Dol-P-Man Synthase Subunit DPM3 Bridges the Congenital Disorders of Glycosylation with the Dystroglycanopathies. American Journal of Human Genetics, 2009, 85, 76-86.	6.2	178
20	Creatine deficiency syndrome caused by guanidinoacetate methyltransferase deficiency: Diagnostic tools for a new inborn error of metabolism. Journal of Pediatrics, 1997, 131, 626-631.	1.8	177
21	Role of cobalamin intake and atrophic gastritis in mild cobalamin deficiency in older Dutch subjects. American Journal of Clinical Nutrition, 1998, 68, 328-334.	4.7	176
22	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
23	The role of interlamellar chain entanglement in deformation-induced structure changes during uniaxial stretching of isotactic polypropylene. Polymer, 2007, 48, 6867-6880.	3.8	173
24	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
25	Mucopolysaccharidosis type IIIA: Clinical spectrum and genotypeâ€phenotype correlations. Annals of Neurology, 2010, 68, 876-887.	5.3	155
26	Dopa-responsive dystonia: A clinical and molecular genetic study. Annals of Neurology, 1998, 44, 649-656.	5.3	153
27	Mechanisms in Protein O-Glycan Biosynthesis and Clinical and Molecular Aspects of Protein O-Glycan Biosynthesis Defects: A Review. Clinical Chemistry, 2006, 52, 574-600.	3.2	152
28	Measuring Urinary Glycosaminoglycans in the Presence of Protein: An Improved Screening Procedure for Mucopolysaccharidoses Based on Dimethylmethylene Blue. Clinical Chemistry, 1992, 38, 803-807.	3.2	147
29	Cerebrotendinous Xanthomatosis: The Spectrum of Imaging Findings and the Correlation with Neuropathologic Findings. Radiology, 2000, 217, 869-876.	7.3	147
30	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
31	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
32	Mevalonate kinase deficiency. Neurology, 2004, 62, 994-997.	1.1	142
33	Twenty-two novel mutations in the lysosomal ?-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II. Human Mutation, 2004, 23, 47-56.	2.5	142
34	Autosomal recessive cutis laxa syndrome revisited. European Journal of Human Genetics, 2009, 17, 1099-1110.	2.8	131
35	High-resolution 1H-NMR spectroscopy of blood plasma for metabolic studies. Clinical Chemistry, 1994, 40, 1245-1250.	3.2	130
36	Autosomal Recessive Dilated Cardiomyopathy due to DOLK Mutations Results from Abnormal Dystroglycan O-Mannosylation. PLoS Genetics, 2011, 7, e1002427.	3.5	130

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37	Apolipoprotein C-III Isofocusing in the Diagnosis of Genetic Defects in O-Glycan Biosynthesis. Clinical Chemistry, 2003, 49, 1839-1845.	3.2	128
38	Clinical and genetic spectrum of Sanfilippo type C (MPS IIIC) disease in The Netherlands. Molecular Genetics and Metabolism, 2008, 93, 104-111.	1.1	127
39	SUCNR1-mediated chemotaxis of macrophages aggravates obesity-induced inflammation and diabetes. Diabetologia, 2017, 60, 1304-1313.	6.3	126
40	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
41	Proton nuclear magnetic resonance spectroscopy of body fluids in the field of inborn errors of metabolism. Annals of Clinical Biochemistry, 2003, 40, 16-24.	1.6	123
42	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	5.3	121
43	In vivo quantitative nearâ€infrared spectroscopy in skeletal muscle during incremental isometric handgrip exercise. Clinical Physiology and Functional Imaging, 2002, 22, 210-217.	1.2	117
44	Tyrosine hydroxylase deficiency causes progressive encephalopathy and dopa-nonresponsive dystonia. Annals of Neurology, 2003, 54, S56-S65.	5.3	117
45	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	2.9	115
46	<scp>l</scp> -dopaâ€responsive infantile hypokinetic rigid parkinsonism due to tyrosine hydroxylase deficiency. Neurology, 2000, 55, 1926-1928.	1.1	113
47	Biochemical hallmarks of tyrosine hydroxylase deficiency. Clinical Chemistry, 1998, 44, 1897-1904.	3.2	111
48	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
49	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
50	Cerebrospinal neuron-specific enolase, S-100 and myelin basic protein in neurological disorders. Acta Neurologica Scandinavica, 1995, 92, 247-251.	2.1	109
51	Cerebrospinal Fluid Glucose and Lactate: Age-Specific Reference Values and Implications for Clinical Practice. PLoS ONE, 2012, 7, e42745.	2.5	109
52	Late-onset metachromatic leukodystrophy: Genotype strongly influences phenotype. Neurology, 2006, 67, 859-863.	1.1	106
53	Cerebrospinal Fluid Analysis in the Workup of GLUT1 Deficiency Syndrome. JAMA Neurology, 2013, 70, 1440.	9.0	106
54	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106

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55	Mitochondrial creatine kinase: a key enzyme of aerobic energy metabolism. BBA - Proteins and Proteomics, 1992, 1102, 119-166.	2.1	105
56	Mucopolysaccharidosis type IIIB may predominantly present with an attenuated clinical phenotype. Journal of Inherited Metabolic Disease, 2010, 33, 759-767.	3.6	105
57	A mutation in the human ortholog of the Saccharomyces cerevisiae ALG6 gene causes carbohydrate-deficient glycoprotein syndrome type-Ic. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 6982-6987.	7.1	103
58	Standardized method for high-resolution 1H-NMR of cerebrospinal fluid. Clinical Chemistry, 1995, 41, 744-751.	3.2	102
59	A common point mutation in the tyrosine hydroxylase gene in autosomal recessive L-DOPA-responsive dystonia in the Dutch population. Human Genetics, 1998, 102, 644-646.	3.8	102
60	Mutations in the sterol 27-hydoxylase gene (CYP27A) cause hepatitis of infancy as well as cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2002, 25, 501-513.	3.6	99
61	Simvastatin: a new therapeutic approach for Smith-Lemli-Opitz syndrome. Journal of Lipid Research, 2000, 41, 1339-1346.	4.2	98
62	Expanded motor and psychiatric phenotype in autosomal dominant Segawa syndrome due to GTP cyclohydrolase deficiency. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 18-23.	1.9	96
63	Dimethylmethylene blue-based spectrophotometry of glycosaminoglycans in untreated urine: a rapid screening procedure for mucopolysaccharidoses. Clinical Chemistry, 1989, 35, 1472-7.	3.2	95
64	Maternal myo-inositol, glucose, and zinc status is associated with the risk of offspring with spina bifida. American Journal of Obstetrics and Gynecology, 2003, 189, 1713-1719.	1.3	94
65	A study on the dimeric structure of creatine kinase (EC 2.7.3.2). Clinica Chimica Acta, 1977, 75, 377-385.	1.1	93
66	Uneven X inactivation in a female monozygotic twin pair with Fabry disease and discordant expression of a novel mutation in the alpha-galactosidase A gene.. Journal of Medical Genetics, 1996, 33, 682-688.	3.2	93
67	A common mutation in the COG7 gene with a consistent phenotype including microcephaly, adducted thumbs, growth retardation, VSD and episodes of hyperthermia. European Journal of Human Genetics, 2007, 15, 638-645.	2.8	93
68	Longâ€term outcome in pyridoxineâ€dependent epilepsy. Developmental Medicine and Child Neurology, 2012, 54, 849-854.	2.1	93
69	Diagnosing Inborn Errors of Lipid Metabolism with Proton Nuclear Magnetic Resonance Spectroscopy. Clinical Chemistry, 2006, 52, 1395-1405.	3.2	91
70	Heterozygosity for a Loss-of-Function Mutation in GALNT2 Improves Plasma Triglyceride Clearance in Man. Cell Metabolism, 2011, 14, 811-818.	16.2	91
71	Biochemical and genetic analysis of 3-methylglutaconic aciduria type IV: a diagnostic strategy. Brain, 2009, 132, 136-146.	7.6	90
72	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. American Journal of Human Genetics, 2011, 88, 216-225.	6.2	90

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73	A novel carbohydrate-deficient glycoprotein syndrome characterized by a deficiency in glucosylation of the dolichol-linked oligosaccharide.. Journal of Clinical Investigation, 1998, 102, 647-652.	8.2	90
74	Effect of simvastatin in addition to chenodeoxycholic acid in patients with cerebrotendinous xanthomatosis. Metabolism: Clinical and Experimental, 1999, 48, 233-238.	3.4	89
75	Tyrosine hydroxylase deficiency: Clinical manifestations of catecholamine insufficiency in infancy. Movement Disorders, 2002, 17, 354-359.	3.9	88
76	Smithâ€Lemliâ€Opitz Syndrome and the DHCR7 Gene. Annals of Human Genetics, 2003, 67, 269-280.	0.8	88
77	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
78	Intestinal permeability is increased after major vascular surgery. Journal of Vascular Surgery, 1993, 17, 734-737.	1.1	87
79	High-resolution proton nuclear magnetic resonance spectroscopy of ovarian cyst fluid. NMR in Biomedicine, 2000, 13, 297-305.	2.8	87
80	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. Brain, 2010, 133, 3210-3220.	7.6	87
81	Presence of Diarrhea and Absence of Tendon Xanthomas in Patients With Cerebrotendinous Xanthomatosis. Archives of Neurology, 2000, 57, 520.	4.5	86
82	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	21.4	86
83	1H-NMR Spectroscopy of Body Fluids: Inborn Errors of Purine and Pyrimidine Metabolism. Clinical Chemistry, 1999, 45, 539-548.	3.2	85
84	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
85	Post-synthetic changes in creatine kinase isozymes (EC 2.7.3.2). Clinica Chimica Acta, 1978, 86, 323-327.	1.1	83
86	Vitamin B12 and folate concentrations in serum and cerebrospinal fluid of neurological patients with special reference to multiple sclerosis and dementia.. Journal of Neurology, Neurosurgery and Psychiatry, 1990, 53, 951-954.	1.9	82
87	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
88	Simvastatin. A new therapeutic approach for Smith-Lemli-Opitz syndrome. Journal of Lipid Research, 2000, 41, 1339-46.	4.2	81
89	Arts Syndrome Is Caused by Loss-of-Function Mutations in PRPS1. American Journal of Human Genetics, 2007, 81, 507-518.	6.2	80
90	Characterization of acute myeloid leukemia based on levels of global hydroxymethylation. Blood, 2014, 124, 1110-1118.	1.4	80

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91	Genetic characteristics of myoadenylate deaminase deficiency. Annals of Neurology, 1998, 44, 140-143.	5.3	79
92	Dimethyl sulfone in human cerebrospinal fluid and blood plasma confirmed by one-dimensional 1H and two-dimensional 1H-13C NMR. NMR in Biomedicine, 2005, 18, 331-336.	2.8	79
93	Protein Complexes in the Archaeon Methanothermobacter thermautotrophicus Analyzed by Blue Native/SDS-PAGE and Mass Spectrometry. Molecular and Cellular Proteomics, 2005, 4, 1653-1663.	3.8	79
94	Cobalamin disorder Cbl-C presenting with late-onset thrombotic microangiopathy. American Journal of Medical Genetics Part A, 2002, 111, 195-201.	2.4	78
95	Î²-Ureidopropionase deficiency: an inborn error of pyrimidine degradation associated with neurological abnormalities. Human Molecular Genetics, 2004, 13, 2793-2801.	2.9	78
96	Intestinal permeability is increased after major vascular surgery. Journal of Vascular Surgery, 1993, 17, 734-737.	1.1	78
97	Spinal xanthomatosis: a variant of cerebrotendinous xanthomatosis. Brain, 1999, 122, 1589-1595.	7.6	77
98	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). American Journal of Human Genetics, 2006, 79, 807-819.	6.2	77
99	Mutations in ACY1, the Gene Encoding Aminoacylase 1, Cause a Novel Inborn Error of Metabolism. American Journal of Human Genetics, 2006, 78, 401-409.	6.2	76
100	Leukoencephalopathy associated with a disturbance in the metabolism of polyols. Annals of Neurology, 1999, 46, 925-928.	5.3	75
101	Treatment and follow-up of children with cerebrotendinous xanthomatosis. European Journal of Pediatrics, 1998, 157, 313-316.	2.7	74
102	Defect in Dimethylglycine Dehydrogenase, a New Inborn Error of Metabolism: NMR Spectroscopy Study. Clinical Chemistry, 1999, 45, 459-464.	3.2	74
103	Long-term course of <scp>l</scp> -dopa-responsive dystonia caused by tyrosine hydroxylase deficiency. Neurology, 2004, 63, 1524-1526.	1.1	74
104	Defective protein glycosylation in patients with cutis laxa syndrome. European Journal of Human Genetics, 2005, 13, 414-421.	2.8	74
105	3â€Methylglutaconic aciduriaâ€"lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	3.6	74
106	Lactobacilli and Acidosis in Children With Short Small Bowel. Journal of Pediatric Gastroenterology and Nutrition, 2000, 30, 288-293.	1.8	74
107	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	3.8	73
108	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

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109	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
110	Long-term treatment effect in cerebrotendinous xanthomatosis depends on age at treatment start. Neurology, 2019, 92, e83-e95.	1.1	73
111	The 3â€methylglutaconic acidurias: what's new?. Journal of Inherited Metabolic Disease, 2012, 35, 13-22.	3.6	72
112	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
113	Isoelectric focusing and hybridisation experiments on creatine kinase (EC 2.7.3.2). Clinica Chimica Acta, 1977, 78, 271-276.	1.1	67
114	Ocular and Systemic Manifestations of Cerebrotendinous Xanthomatosis. American Journal of Ophthalmology, 1995, 120, 597-604.	3.3	67
115	Defining the phenotype in an autosomal recessive cutis laxa syndrome with a combined congenital defect of glycosylation. European Journal of Human Genetics, 2008, 16, 28-35.	2.8	67
116	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
117	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	7.6	67
118	Clinical and biochemical characteristics of congenital disorder of glycosylation type Ic, the first recognized endoplasmic reticulum defect in N-glycan synthesis. Annals of Neurology, 2000, 47, 776-781.	5.3	66
119	Cloning of Dimethylglycine Dehydrogenase and a New Human Inborn Error of Metabolism, Dimethylglycine Dehydrogenase Deficiency. American Journal of Human Genetics, 2001, 68, 839-847.	6.2	66
120	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
121	Ca2+ homeostasis in Brody's disease. A study in skeletal muscle and cultured muscle cells and the effects of dantrolene an verapamil.. Journal of Clinical Investigation, 1994, 94, 741-748.	8.2	65
122	N-Acetylated Metabolites in Urine: Proton Nuclear Magnetic Resonance Spectroscopic Study on Patients with Inborn Errors of Metabolism. Clinical Chemistry, 2004, 50, 58-66.	3.2	64
123	Metabolic cutis laxa syndromes. Journal of Inherited Metabolic Disease, 2011, 34, 907-916.	3.6	64
124	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
125	THE SECOND WIND PHENOMENON IN McARDLE'S DISEASE. Brain, 1986, 109, 1087-1101.	7.6	62
126	Multi-allelic origin of congenital disorder of glycosylation (CDG)-Ic. Human Genetics, 2000, 106, 538-545.	3.8	62

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127	Biochemical hallmarks of tyrosine hydroxylase deficiency. Clinical Chemistry, 1998, 44, 1897-904.	3.2	62
128	Cerebrospinal Fluid Investigations for Neurometabolic Disorders. Neuropediatrics, 1998, 29, 59-71.	0.6	61
129	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
130	Juvenile Cataract Associated With Chronic Diarrhea in Pediatric Cerebrotendinous Xanthomatosis. American Journal of Ophthalmology, 1991, 112, 606-607.	3.3	60
131	Increased Cerebrospinal Fluid Glycine: A Biochemical Marker for a Leukoencephalopathy With Vanishing White Matter. Journal of Child Neurology, 1999, 14, 728-731.	1.4	59
132	Association of 3-methylglutaconic aciduria with sensori-neural deafness, encephalopathy, and Leigh-like syndrome (MEGDEL association) in four patients with a disorder of the oxidative phosphorylation. Molecular Genetics and Metabolism, 2006, 88, 47-52.	1.1	59
133	3-Methylglutaconic aciduria type I redefined. Neurology, 2010, 75, 1079-1083.	1.1	59
134	Plasma N-Glycan Profiling by Mass Spectrometry for Congenital Disorders of Glycosylation Type II. Clinical Chemistry, 2011, 57, 593-602.	3.2	59
135	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
136	Regularized MANOVA (rMANOVA) in untargeted metabolomics. Analytica Chimica Acta, 2015, 899, 1-12.	5.4	59
137	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
138	Vacuolar H+-ATPase meets glycosylation in patients with cutis laxa. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 903-914.	3.8	57
139	Alpha-fetoprotein, a fascinating protein and biomarker in neurology. European Journal of Paediatric Neurology, 2014, 18, 243-248.	1.6	57
140	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
141	Phosphomannomutase deficiency is the main cause of carbohydrate-deficient glycoprotein syndrome with type I isoelectrofocusing pattern of serum sialotransferrins. Journal of Inherited Metabolic Disease, 1997, 20, 447-449.	3.6	56
142	Clinical and biochemical presentation of siblings with COG-7 deficiency, a lethal multiple O- and N-glycosylation disorder. Journal of Inherited Metabolic Disease, 2005, 28, 707-714.	3.6	56
143	Ophthalmological abnormalities in children with congenital disorders of glycosylation type I. British Journal of Ophthalmology, 2009, 93, 350-354.	3.9	56
144	Clinical Symptoms of Adult Metachromatic Leukodystrophy and Arylsulfatase A Pseudodeficiency. Archives of Neurology, 1995, 52, 408-413.	4.5	55

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145	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
146	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
147	1H-magnetic resonance spectroscopy. Cancer, 1998, 82, 1726-1730.	4.1	54
148	Aminoacylase I deficiency: A novel inborn error of metabolism. Biochemical and Biophysical Research Communications, 2005, 338, 1322-1326.	2.1	54
149	PNPO deficiency: An under diagnosed inborn error of pyridoxine metabolism. Molecular Genetics and Metabolism, 2008, 94, 431-434.	1.1	54
150	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
151	Molecular identification in metabolomics using infrared ion spectroscopy. Scientific Reports, 2017, 7, 3363.	3.3	54
152	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
153	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
154	Four novel mutations in the Tyrosine Hydroxylase gene in patients with infantile parkinsonism. Annals of Human Genetics, 2000, 64, 25-31.	0.8	52
155	Variable clinical presentation of lysosomal Î²-mannosidosis in patients with null mutations. Molecular Genetics and Metabolism, 2002, 77, 282-290.	1.1	52
156	Measuring urinary glycosaminoglycans in the presence of protein: an improved screening procedure for mucopolysaccharidoses based on dimethylmethylene blue. Clinical Chemistry, 1992, 38, 803-7.	3.2	52
157	NMR spectroscopic studies on the late onset form of 3-methylglutaconic aciduria type I and other defects in leucine metabolism. NMR in Biomedicine, 2006, 19, 271-278.	2.8	51
158	Transferrin and Apolipoprotein C-III Isofocusing Are Complementary in the Diagnosis of N- and O-Glycan Biosynthesis Defects. Clinical Chemistry, 2007, 53, 180-187.	3.2	50
159	Cerebellar ataxia and congenital disorder of glycosylation Ia (CDG-Ia) with normal routine CDG screening. Journal of Neurology, 2007, 254, 1356-1358.	3.6	50
160	Is muscle glycogenolysis impaired in X-linked phosphorylase b kinase deficiency?. Neurology, 2008, 70, 1876-1882.	1.1	50
161	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	3.6	50
162	Tyrosine hydroxylase deficiency with severe clinical course: Clinical and biochemical investigations and optimization of therapy. Journal of Pediatrics, 2000, 136, 560-562.	1.8	49

#	Article	IF	Citations
163	Severe hypomyelination associated with increased levels of N -acetylaspartylglutamate in CSF. Neurology, 2004, 62, 1503-1508.	1.1	49
164	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
165	The calcium homeostasis and the membrane potential of cultured muscle cells from patients with myotonic dystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1990, 1096, 14-19.	3.8	48
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