Ronald J A Wanders

List of Publications by Year
in descending order

333 papers	24,139 citations	4942 84 h-index	10424 139 g-index
346 all docs	346 docs citations	346 times ranked	20458 citing authors

#	Article	IF	Citations
1	Biochemistry of Mammalian Peroxisomes Revisited. Annual Review of Biochemistry, 2006, 75, 295-332.	5.0	853
2	Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies. Nature Medicine, 2014, 20, 616-623.	15.2	733
3	A general introduction to the biochemistry of mitochondrial fatty acid Î²â€oxidation. Journal of Inherited Metabolic Disease, 2010, 33, 469-477.	1.7	678
4	A Lethal Defect of Mitochondrial and Peroxisomal Fission. New England Journal of Medicine, 2007, 356, 1736-1741.	13.9	665
5	Carnitine biosynthesis in mammals. Biochemical Journal, 2002, 361, 417-429.	1.7	527
6	The Biochemistry and Physiology of Mitochondrial Fatty Acid Î²-Oxidation and Its Genetic Disorders. Annual Review of Physiology, 2016, 78, 23-44.	5.6	490
7	Mutations in MVK, encoding mevalonate kinase, cause hyperimmunoglobulinaemia D and periodic fever syndrome. Nature Genetics, 1999, 22, 175-177.	9.4	480
8	X-linked adrenoleukodystrophy (X-ALD): clinical presentation and guidelines for diagnosis, follow-up and management. Orphanet Journal of Rare Diseases, 2012, 7, 51.	1.2	403
9	Defective Remodeling of Cardiolipin and Phosphatidylglycerol in Barth Syndrome. Biochemical and Biophysical Research Communications, 2000, 279, 378-382.	1.0	352
10	Functions and biosynthesis of plasmalogens in health and disease. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2004, 1636, 219-231.	1.2	329
11	Refsum disease is caused by mutations in the phytanoylâ€“CoA hydroxylase gene. Nature Genetics, 1997, 17, 190-193.	9.4	302
12	Zellweger spectrum disorders: clinical overview and management approach. Orphanet Journal of Rare Diseases, 2015, 10, 151.	1.2	286
13	Human disorders of peroxisome metabolism and biogenesis. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 922-933.	1.9	281
14	Metabolic Interplay between Peroxisomes and Other Subcellular Organelles Including Mitochondria and the Endoplasmic Reticulum. Frontiers in Cell and Developmental Biology, 2015, 3, 83.	1.8	270
15	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. Human Mutation, 2001, 18, 499-515.	1.1	261
16	Rhizomelic chondrodysplasia punctata is a peroxisomal protein targeting disease caused by a non-functional PTS2 receptor. Nature Genetics, 1997, 15, 377-380.	9.4	260
17	Cardiolipin provides an essential activating platform for caspase-8 on mitochondria. Journal of Cell Biology, 2008, 183, 681-696.	2.3	258
18	X-linked cardioskeletal myopathy and neutropenia (Barth syndrome): An update. American Journal of Medical Genetics Part A, 2004, 126A, 349-354.	2.4	244

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19	Mutations in the gene encoding peroxisomal Î±-methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. Nature Genetics, 2000, 24, 188-191.	9.4	241
20	Peroxisomal disorders: The single peroxisomal enzyme deficiencies. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 1707-1720.	1.9	240
21	Systematic mapping of contact sites reveals tethers and a function for the peroxisome-mitochondria contact. Nature Communications, 2018, 9, 1761.	5.8	222
22	Disorders of mitochondrial long-chain fatty acid oxidation and the carnitine shuttle. Reviews in Endocrine and Metabolic Disorders, 2018, 19, 93-106.	2.6	215
23	Proteomics Characterization of Mouse Kidney Peroxisomes by Tandem Mass Spectrometry and Protein Correlation Profiling. Molecular and Cellular Proteomics, 2007, 6, 2045-2057.	2.5	210
24	The human peroxisomal ABC half transporter ALDP functions as a homodimer and accepts acylâ€“CoA esters. FASEB Journal, 2008, 22, 4201-4208.	0.2	200
25	Metabolic functions of peroxisomes in health and disease. Biochimie, 2014, 98, 36-44.	1.3	189
26	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency: Clinical Presentation and Follow-Up of 50 Patients. Pediatrics, 2002, 109, 99-104.	1.0	184
27	Modern theories of metabolic control and their applications. Bioscience Reports, 1984, 4, 1-22.	1.1	183
28	Fatty acid omegaâ€oxidation as a rescue pathway for fatty acid oxidation disorders in humans. FEBS Journal, 2011, 278, 182-194.	2.2	181
29	Peroxisomal disorders I: biochemistry and genetics of peroxisome biogenesis disorders. Clinical Genetics, 2004, 67, 107-133.	1.0	177
30	Peroxisomal alterations in Alzheimerâ€™s disease. Acta Neuropathologica, 2011, 122, 271-283.	3.9	176
31	Clinical and biochemical spectrum of D-bifunctional protein deficiency. Annals of Neurology, 2006, 59, 92-104.	2.8	175
32	Acyl-CoA Dehydrogenase 9 Is Required for the Biogenesis of Oxidative Phosphorylation Complex I. Cell Metabolism, 2010, 12, 283-294.	7.2	172
33	Sodium taurocholate cotransporting polypeptide (SLC10A1) deficiency: Conjugated hypercholanemia without a clear clinical phenotype. Hepatology, 2015, 61, 260-267.	3.6	169
34	The enzymology of mitochondrial fatty acid betaâ€oxidation and its application to followâ€up analysis of positive neonatal screening results. Journal of Inherited Metabolic Disease, 2010, 33, 479-494.	1.7	166
35	Identification of the peroxisomal Î²-oxidation enzymes involved in the biosynthesis of docosahexaenoic acid. Journal of Lipid Research, 2001, 42, 1987-1995.	2.0	165
36	Identification of PEX7 as the Second Gene Involved in Refsum Disease. American Journal of Human Genetics, 2003, 72, 471-477.	2.6	151

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37	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
38	Metabolite transport across the peroxisomal membrane. Biochemical Journal, 2007, 401, 365-375.	1.7	142
39	Metabolic and molecular basis of peroxisomal disorders: A review. American Journal of Medical Genetics Part A, 2004, 126A, 355-375.	2.4	140
40	The role of ELOVL1 in very longâ€chain fatty acid homeostasis and Xâ€linked adrenoleukodystrophy. EMBO Molecular Medicine, 2010, 2, 90-97.	3.3	140
41	Mitochondrial protein acetylation is driven by acetyl-CoA from fatty acid oxidation. Human Molecular Genetics, 2014, 23, 3513-3522.	1.4	140
42	Analysis of very long-chain fatty acids using electrospray ionization mass spectrometry. Molecular Genetics and Metabolism, 2003, 79, 189-196.	0.5	138
43	Intrinsic acyl-CoA thioesterase activity of a peroxisomal ATP binding cassette transporter is required for transport and metabolism of fatty acids. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 1279-1284.	3.3	134
44	Pristanic acid and phytanic acid: naturally occurring ligands for the nuclear receptor peroxisome proliferator-activated receptor Î±. Journal of Lipid Research, 2000, 41, 1801-1807.	2.0	133
45	Molecular basis of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: identification of the major disease-causing mutation in the Î±-subunit of the mitochondrial trifunctional protein. Lipids and Lipid Metabolism, 1994, 1215, 347-350.	2.6	130
46	Pex11p Plays a Primary Role in Medium-Chain Fatty Acid Oxidation, a Process That Affects Peroxisome Number and Size in Saccharomyces cerevisiae. Journal of Cell Biology, 2000, 150, 489-498.	2.3	127
47	A novel defect of peroxisome division due to a homozygous non-sense mutation in thePEX11Î²gene. Journal of Medical Genetics, 2012, 49, 307-313.	1.5	127
48	Peroxisomal L-bifunctional enzyme (Ehhadh) is essential for the production of medium-chain dicarboxylic acids. Journal of Lipid Research, 2012, 53, 1296-1303.	2.0	127
49	Genetic classification and mutational spectrum of more than 600 patients with a Zellweger syndrome spectrum disorder. Human Mutation, 2011, 32, 59-69.	1.1	126
50	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.	1.1	121
51	Clinical, biochemical, and mutational spectrum of peroxisomal acylâ€“coenzyme A oxidase deficiency. Human Mutation, 2007, 28, 904-912.	1.1	121
52	Phytanic acid metabolism in health and disease. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2011, 1811, 498-507.	1.2	120
53	Clinical implications of mutation analysis in primary hyperoxaluria type 1. Kidney International, 2004, 66, 746-752.	2.6	116
54	A novel bile acid biosynthesis defect due to a deficiency of peroxisomal ABCD3. Human Molecular Genetics, 2015, 24, 361-370.	1.4	115

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55	Phytanic acid and pristanic acid are oxidized by sequential peroxisomal and mitochondrial reactions in cultured fibroblasts. Journal of Lipid Research, 1998, 39, 66-74.	2.0	115
56	Identification of the peroxisomal Î²-oxidation enzymes involved in the degradation of long-chain dicarboxylic acids. Journal of Lipid Research, 2004, 45, 1104-1111.	2.0	114
57	Peroxisomal Disorders: A Review. Journal of Neuropathology and Experimental Neurology, 1995, 54, 726-739.	0.9	113
58	Molecular basis of Refsum disease: Sequence variations in Phytanoyl-CoA Hydroxylase (PHYH) and the PTS2 receptor (PEX7). Human Mutation, 2004, 23, 209-218.	1.1	113
59	Riboflavin-responsive oxidative phosphorylation complex I deficiency caused by defective ACAD9: new function for an old gene. Brain, 2011, 134, 210-219.	3.7	113
60	Differential substrate specificities of human ABCD1 and ABCD2 in peroxisomal fatty acid Î²-oxidation. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2011, 1811, 148-152.	1.2	113
61	Lipid metabolism in peroxisomes in relation to human disease. Molecular Aspects of Medicine, 1998, 19, i-154.	2.7	111
62	ECHS1 mutations in Leigh disease: a new inborn error of metabolism affecting valine metabolism. Brain, 2014, 137, 2903-2908.	3.7	111
63	Peroxisomes in human fibroblasts have a basic pH. Nature Cell Biology, 2000, 2, 51-53.	4.6	110
64	Mitochondrial long chain fatty acid Î²-oxidation in man and mouse. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2009, 1791, 806-815.	1.2	109
65	Ataxia with loss of Purkinje cells in a mouse model for Refsum disease. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 17712-17717.	3.3	108
66	Transport of Activated Fatty Acids by the Peroxisomal ATP-binding-cassette Transporter Pxa2 in a Semi-Intact Yeast Cell System. FEBS Journal, 1997, 249, 657-661.	0.2	107
67	Alkyl-Dihydroxyacetonephosphate Synthase. Journal of Biological Chemistry, 1998, 273, 10296-10301.	1.6	106
68	MS/MS-based newborn and family screening detects asymptomatic patients with very-long-chain acyl-CoA dehydrogenase deficiency. Journal of Pediatrics, 2003, 143, 335-342.	0.9	106
69	Quantitative and Compositional Study of Cardiolipin in Platelets by Electrospray Ionization Mass Spectrometry: Application for the Identification of Barth Syndrome Patients. Clinical Chemistry, 2002, 48, 1390-1397.	1.5	105
70	Alkyl-Glycerol Rescues Plasmalogen Levels and Pathology of Ether-Phospholipid Deficient Mice. PLoS ONE, 2011, 6, e28539.	1.1	104
71	Phytanoylâ€“Coenzyme a Hydroxylase Deficiency â€” The Enzyme Defect in Refsum's Disease. New England Journal of Medicine, 1997, 337, 133-134.	13.9	103
72	Peripheral nervous system plasmalogens regulate Schwann cell differentiation and myelination. Journal of Clinical Investigation, 2014, 124, 2560-2570.	3.9	103

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73	Identification of a Peroxisomal ATP Carrier Required for Medium-Chain Fatty Acid Î²-Oxidation and Normal Peroxisome Proliferation in Saccharomyces cerevisiae. Molecular and Cellular Biology, 2001, 21, 4321-4329.	1.1	101
74	PEX12, the Pathogenic Gene of Group III Zellweger Syndrome: cDNA Cloning by Functional Complementation on a CHO Cell Mutant, Patient Analysis, and Characterization of Pex12p. Molecular and Cellular Biology, 1998, 18, 4324-4336.	1.1	99
75	Impaired neuronal migration and endochondral ossification in Pex7 knockout mice: a model for rhizomelic chondrodysplasia punctata. Human Molecular Genetics, 2003, 12, 2255-2267.	1.4	97
76	Participation of Two Members of the Very Long-chain Acyl-CoA Synthetase Family in Bile Acid Synthesis and Recycling. Journal of Biological Chemistry, 2002, 277, 24771-24779.	1.6	96
77	The peroxisomal ABC transporter family. Pflugers Archiv European Journal of Physiology, 2007, 453, 719-734.	1.3	95
78	Mammalian peroxisomal ABC transporters: from endogenous substrates to pathology and clinical significance. British Journal of Pharmacology, 2011, 164, 1753-1766.	2.7	93
79	Disorders of Peroxisome Biogenesis Due to Mutations in PEX1: Phenotypes and PEX1 Protein Levels. American Journal of Human Genetics, 2001, 69, 35-48.	2.6	92
80	Mutational Spectrum in the PEX7 Gene and Functional Analysis of Mutant Alleles in 78 Patients with Rhizomelic Chondrodysplasia Punctata Type 1. American Journal of Human Genetics, 2002, 70, 612-624.	2.6	92
81	A key role for the peroxisomalABCD2transporter in fatty acid homeostasis. American Journal of Physiology - Endocrinology and Metabolism, 2009, 296, E211-E221.	1.8	91
82	Inactivation of the peroxisomal ABCD2 transporter in the mouse leads to late-onset ataxia involving mitochondria, Golgi and endoplasmic reticulum damage. Human Molecular Genetics, 2005, 14, 3565-3577.	1.4	90
83	ACBD5 deficiency causes a defect in peroxisomal very long-chain fatty acid metabolism. Journal of Medical Genetics, 2017, 54, 330-337.	1.5	90
84	Plasmalogens participate in very-long-chain fatty acid-induced pathology. Brain, 2008, 132, 482-492.	3.7	89
85	Identification and characterization of human cardiolipin synthase. FEBS Letters, 2006, 580, 3059-3064.	1.3	87
86	A role for the human peroxisomal half-transporter ABCD3 in the oxidation of dicarboxylic acids. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2014, 1841, 563-568.	1.2	87
87	Long-Chain Fatty Acid Oxidation during Early Human Development. Pediatric Research, 2005, 57, 755-759.	1.1	86
88	Alpha-Oxidation. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 1403-1412.	1.9	86
89	NAD ⁺ homeostasis in human health and disease. EMBO Molecular Medicine, 2021, 13, e13943.	3.3	86
90	Organization of the mevalonate kinase (MVK) gene and identification of novel mutations causing mevalonic aciduria and hyperimmunoglobulinaemia D and periodic fever syndrome. European Journal of Human Genetics, 2001, 9, 253-259.	1.4	85

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91	Refsum Disease, Peroxisomes and Phytanic Acid Oxidation: A Review. Journal of Neuropathology and Experimental Neurology, 2001, 60, 1021-1031.	0.9	83
92	Characterization of carnitine and fatty acid metabolism in the long-chain acyl-CoA dehydrogenase-deficient mouse. Biochemical Journal, 2005, 387, 185-193.	1.7	81
93	Peroxisomal Fatty Acid Uptake Mechanism in Saccharomyces cerevisiae. Journal of Biological Chemistry, 2012, 287, 20144-20153.	1.6	81
94	Mitochondrial trifunctional protein deficiency: A severe fatty acid oxidation disorder with cardiac and neurologic involvement. Journal of Pediatrics, 2003, 142, 684-689.	0.9	80
95	Mutational Spectrum of d-Bifunctional Protein Deficiency and Structure-Based Genotype-Phenotype Analysis. American Journal of Human Genetics, 2006, 78, 112-124.	2.6	80
96	Mutations in the Gene Encoding 3-Hydroxyisobutyryl-CoA Hydrolase Results in Progressive Infantile Neurodegeneration. American Journal of Human Genetics, 2007, 80, 195-199.	2.6	80
97	Identification of an unusual variant peroxisome biogenesis disorder caused by mutations in the PEX16 gene. Journal of Medical Genetics, 2010, 47, 608-615.	1.5	80
98	Quantitative acylcarnitine profiling in fibroblasts using [U-13C] palmitic acid: an improved tool for the diagnosis of fatty acid oxidation defects. Clinica Chimica Acta, 1999, 281, 1-17.	0.5	79
99	Isolated 2-Methylbutyrylglycinuria Caused by Short/Branched-Chain Acyl-CoA Dehydrogenase Deficiency: Identification of a New Enzyme Defect, Resolution of Its Molecular Basis, and Evidence for Distinct Acyl-CoA Dehydrogenases in Isoleucine And Valine Metabolism. American Journal of Human Genetics, 2000, 67, 1095-1103.	2.6	79
100	Metabolic interactions between peroxisomes and mitochondria with a special focus on acylcarnitine metabolism. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165720.	1.8	78
101	Peroxisome biogenesis disorders with prolonged survival: Phenotypic expression in a cohort of 31 patients. American Journal of Medical Genetics Part A, 2004, 126A, 333-338.	2.4	77
102	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. Brain, 2019, 142, 3382-3397.	3.7	76
103	Mutations in PEX10 are a cause of autosomal recessive ataxia. Annals of Neurology, 2010, 68, 259-263.	2.8	74
104	Peroxisomes contribute to the acylcarnitine production when the carnitine shuttle is deficient. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2013, 1831, 1467-1474.	1.2	74
105	Proteomic analysis of mouse kidney peroxisomes: identification of RP2p as a peroxisomal nudix hydrolase with acyl-CoA diphosphatase activity. Biochemical Journal, 2006, 393, 537-543.	1.7	73
106	Peroxisomal D-bifunctional protein deficiency. Neurology, 2014, 82, 963-968.	1.5	73
107	A novel type of rhizomelic chondrodysplasia punctata, RCDP5, is caused by loss of the PEX5 long isoform. Human Molecular Genetics, 2015, 24, 5845-5854.	1.4	73
108	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. Journal of Inherited Metabolic Disease, 2016, 39, 93-106.	1.7	73

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109	Peroxisomal disorders: clinical, biochemical, and molecular aspects. Neurochemical Research, 1999, 24, 565-580.	1.6	71
110	Human phytanoyl-CoA hydroxylase: resolution of the gene structure and the molecular basis of Refsum's disease. Human Molecular Genetics, 2000, 9, 1195-1200.	1.4	71
111	The peroxisomal lumen in Saccharomyces cerevisiae is alkaline. Journal of Cell Science, 2004, 117, 4231-4237.	1.2	70
112	Ï‰-Oxidation of Very Long-chain Fatty Acids in Human Liver Microsomes. Journal of Biological Chemistry, 2006, 281, 13180-13187.	1.6	69
113	Enzymology of the branchedâ€chain amino acid oxidation disorders: the valine pathway. Journal of Inherited Metabolic Disease, 2012, 35, 5-12.	1.7	69
114	Substrate specificity of human carnitine acetyltransferase: Implications for fatty acid and branched-chain amino acid metabolism. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 773-779.	1.8	69
115	Cardiacâ€specific succinate dehydrogenase deficiency in Barth syndrome. EMBO Molecular Medicine, 2016, 8, 139-154.	3.3	69
116	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	1.2	68
117	Identification of a new complementation group of the peroxisome biogenesis disorders andPEX14 as the mutated gene. Human Mutation, 2004, 23, 552-558.	1.1	67
118	Bile acid treatment alters hepatic disease and bile acid transport in peroxisome-deficientPEX2Zellweger mice. Hepatology, 2007, 45, 982-997.	3.6	66
119	Plasmalogen Phospholipids Are Involved in HDL-Mediated Cholesterol Efflux: Insights from Investigations with Plasmalogen-Deficient Cells. Biochemical and Biophysical Research Communications, 1998, 250, 369-373.	1.0	65
120	A PEX6-Defective Peroxisomal Biogenesis Disorder with Severe Phenotype in an Infant, versus Mild Phenotype Resembling Usher Syndrome in the Affected Parents. American Journal of Human Genetics, 2002, 70, 1062-1068.	2.6	65
121	Phytanic acid alpha-oxidation, new insights into an old problem: a review. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2003, 1631, 119-135.	1.2	65
122	PeroxisomeDB: a database for the peroxisomal proteome, functional genomics and disease. Nucleic Acids Research, 2007, 35, D815-D822.	6.5	65
123	Lipidomic analysis of fibroblasts from Zellweger spectrum disorder patients identifies disease-specific phospholipid ratios. Journal of Lipid Research, 2016, 57, 1447-1454.	2.0	65
124	The Arabidopsis Peroxisomal ABC Transporter, Comatose, Complements the Saccharomyces cerevisiae pxa1 pxa2Î" Mutant for Metabolism of Long-chain Fatty Acids and Exhibits Fatty Acyl-CoA-stimulated ATPase Activity. Journal of Biological Chemistry, 2010, 285, 29892-29902.	1.6	64
125	Mitochondrial NADP(H) deficiency due to a mutation in NADK2 causes dienoyl-CoA reductase deficiency with hyperlysinemia. Human Molecular Genetics, 2014, 23, 5009-5016.	1.4	63
126	Impaired amino acid metabolism contributes to fasting-induced hypoglycemia in fatty acid oxidation defects. Human Molecular Genetics, 2013, 22, 5249-5261.	1.4	61

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127	Isolated Mitochondrial Long-Chain Ketoacyl-CoA Thiolase Deficiency Resulting from Mutations in the HADHB Gene. Clinical Chemistry, 2006, 52, 530-534.	1.5	60
128	Specific combination of compound heterozygous mutations in 17Î²-hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of D-bifunctional protein deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 90.	1.2	60
129	Molecular Cloning and Expression of Human Carnitine Octanoyltransferase: Evidence for Its Role in the Peroxisomal Î²-Oxidation of Branched-Chain Fatty Acids. Biochemical and Biophysical Research Communications, 1999, 263, 213-218.	1.0	59
130	Peroxisomes in Human Health and Disease: Metabolic Pathways, Metabolite Transport, Interplay with Other Organelles and Signal Transduction. Sub-Cellular Biochemistry, 2013, 69, 23-44.	1.0	59
131	Metabolite studies in HIBCH and ECHS1 defects: Implications for screening. Molecular Genetics and Metabolism, 2015, 115, 168-173.	0.5	59
132	Lovastatin in X-Linked Adrenoleukodystrophy. New England Journal of Medicine, 2010, 362, 276-277.	13.9	58
133	Carnitine palmitoyltransferase 2 and carnitine/acylcarnitine translocase are involved in the mitochondrial synthesis and export of acylcarnitines. FASEB Journal, 2013, 27, 2039-2044.	0.2	58
134	Heterozygosity for the Common LCHAD Mutation (1528G>C) Is Not a Major Cause of HELLP Syndrome and the Prevalence of the Mutation in the Dutch Population Is Low. Pediatric Research, 2000, 48, 151-154.	1.1	57
135	Mevalonate kinase is a cytosolic enzyme in humans. Journal of Cell Science, 2004, 117, 631-639.	1.2	57
136	A mutation in an alternative untranslated exon of hexokinase 1 associated with Hereditary Motor and Sensory Neuropathy â€“ Russe (HMSNR). European Journal of Human Genetics, 2009, 17, 1606-1614.	1.4	55
137	The role of the clinician in the multiâ€omics era: are you ready?. Journal of Inherited Metabolic Disease, 2018, 41, 571-582.	1.7	55
138	Studies on phytanic acid Î±-oxidation in rat liver and cultured human skin fibroblasts. Lipids and Lipid Metabolism, 1993, 1167, 345-350.	2.6	53
139	Carnitine-acylcarnitine translocase deficiency: metabolic consequences of an impaired mitochondrial carnitine cycle. Clinica Chimica Acta, 2000, 298, 55-68.	0.5	52
140	Purification of peroxisomal acyl-CoA: dihydroxyacetonephosphate acyltransferase from human placenta. BBA - Proteins and Proteomics, 1994, 1206, 27-34.	2.1	48
141	Identification of the molecular defect in patients with peroxisomal mosaicism using a novel method involving culturing of cells at 40Â°C: Implications for other inborn errors of metabolism. Human Mutation, 2004, 24, 130-139.	1.1	48
142	Toxicity of peroxisomal C27-bile acid intermediates. Molecular Genetics and Metabolism, 2009, 96, 121-128.	0.5	48
143	Peroxisomal disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1593-1609.	1.0	48
144	Functional characterisation of peroxisomal Î²â€oxidation disorders in fibroblasts using lipidomics. Journal of Inherited Metabolic Disease, 2018, 41, 479-487.	1.7	48

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145	Cytosolic Aspartate Aminotransferase Encoded by the AAT2 Gene is Targeted to the Peroxisomes in Oleate-Grown Saccharomyces Cerevisiae. FEBS Journal, 1997, 247, 972-980.	0.2	46
146	Characterization of the human Ï‰â€oxidation pathway for Ï‰â€hydroxyâ€veryâ€longâ€chain fatty acids. FASEB Journal, 2008, 22, 2064-2071.	0.2	46
147	The Peroxisomal NAD Carrier from Arabidopsis Imports NAD in Exchange with AMP. Plant Physiology, 2016, 171, 2127-2139.	2.3	45
148	Identification of fatty aldehyde dehydrogenase in the breakdown of phytol to phytanic acid. Molecular Genetics and Metabolism, 2004, 82, 33-37.	0.5	44
149	Identification of novel mutations in classical galactosemia. Human Mutation, 2005, 25, 502-502.	1.1	44
150	The important role of biochemical and functional studies in the diagnostics of peroxisomal disorders. Journal of Inherited Metabolic Disease, 2016, 39, 531-543.	1.7	44
151	Analysis of Carnitine Biosynthesis Metabolites in Urine by HPLCâ€“Electrospray Tandem Mass Spectrometry. Clinical Chemistry, 2002, 48, 826-834.	1.5	42
152	Clinical and Biochemical Pitfalls in the Diagnosis of Peroxisomal Disorders. Neuropediatrics, 2016, 47, 205-220.	0.3	41
153	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976.	2.6	41
154	Plasma analysis of di- and trihydroxycholestanoic acid diastereoisomers in peroxisomal Î±-methylacyl-CoA racemase deficiency. Journal of Lipid Research, 2001, 42, 137-141.	2.0	41
155	2,6-Dimethylheptanoyl-CoA is a specific substrate for long-chain acyl-CoA dehydrogenase (LCAD): evidence for a major role of LCAD in branched-chain fatty acid oxidation. Lipids and Lipid Metabolism, 1998, 1393, 35-40.	2.6	40
156	Short/branched-chain acyl-CoA dehydrogenase deficiency due to an IVS3+3A>G mutation that causes exon skipping. Human Genetics, 2006, 118, 680-690.	1.8	40
157	Functional redundancy of mitochondrial enoylâ€CoA isomerases in the oxidation of unsaturated fatty acids. FASEB Journal, 2012, 26, 4316-4326.	0.2	40
158	Bezafibrate lowers very longâ€chain fatty acids in Xâ€linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation. Journal of Inherited Metabolic Disease, 2012, 35, 1137-1145.	1.7	39
159	Method for Measurement of Peroxisomal Very-Long-Chain Fatty Acid Î²-Oxidation in Human Skin Fibroblasts Using Stable-Isotope-Labeled Tetracosanoic Acid. Clinical Chemistry, 2004, 50, 1824-1826.	1.5	38
160	Carnitine-acylcarnitine translocase deficiency: phenotype, residual enzyme activity and outcome. European Journal of Pediatrics, 2001, 160, 101-104.	1.3	37
161	Evidence for two enzymatic pathways for Ï‰-oxidation of docosanoic acid in rat liver microsomes. Journal of Lipid Research, 2005, 46, 1001-1008.	2.0	37
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