Hans R Waterham

List of Publications by Year
in descending order

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168 papers	11,955 citations	25034 57 h-index	29157 104 g-index
176 all docs	176 docs citations	176 times ranked	10757 citing authors

#	Article	IF	Citations
1	Biochemistry of Mammalian Peroxisomes Revisited. Annual Review of Biochemistry, 2006, 75, 295-332.	11.1	853
2	A Lethal Defect of Mitochondrial and Peroxisomal Fission. New England Journal of Medicine, 2007, 356, 1736-1741.	27.0	665
3	Mutations in MVK, encoding mevalonate kinase, cause hyperimmunoglobulinaemia D and periodic fever syndrome. Nature Genetics, 1999, 22, 175-177.	21.4	480
4	Functions and biosynthesis of plasmalogens in health and disease. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2004, 1636, 219-231.	2.4	329
5	Mutations in the 3Î²-Hydroxysterol Î”24-Reductase Gene Cause Desmosterolosis, an Autosomal Recessive Disorder of Cholesterol Biosynthesis. American Journal of Human Genetics, 2001, 69, 685-694.	6.2	318
6	Human disorders of peroxisome metabolism and biogenesis. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 922-933.	4.1	281
7	Infevers: An evolving mutation database for auto-inflammatory syndromes. Human Mutation, 2004, 24, 194-198.	2.5	277
8	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
9	Metabolic Interplay between Peroxisomes and Other Subcellular Organelles Including Mitochondria and the Endoplasmic Reticulum. Frontiers in Cell and Developmental Biology, 2015, 3, 83.	3.7	270
10	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. Human Mutation, 2001, 18, 499-515.	2.5	261
11	Mutations in the gene encoding peroxisomal Î±-methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. Nature Genetics, 2000, 24, 188-191.	21.4	241
12	Genetics and molecular basis of human peroxisome biogenesis disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1430-1441.	3.8	234
13	Systematic mapping of contact sites reveals tethers and a function for the peroxisome-mitochondria contact. Nature Communications, 2018, 9, 1761.	12.8	222
14	Autosomal Recessive HEM/Greenberg Skeletal Dysplasia Is Caused by 3Î²-Hydroxysterol Î”14-Reductase Deficiency Due to Mutations in the Lamin B Receptor Gene. American Journal of Human Genetics, 2003, 72, 1013-1017.	6.2	206
15	The human peroxisomal ABC half transporter ALDP functions as a homodimer and accepts acylâ€“CoA esters. FASEB Journal, 2008, 22, 4201-4208.	0.5	200
16	Brownâ€Vialettoâ€Van Laere and Fazio Londe syndrome is associated with a riboflavin transporter defect mimicking mild MADD: a new inborn error of metabolism with potential treatment. Journal of Inherited Metabolic Disease, 2011, 34, 159-164.	3.6	194
17	A role for geranylgeranylation in interleukin-1Î² secretion. Arthritis and Rheumatism, 2006, 54, 3690-3695.	6.7	169
18	Lack of isoprenoid products raises ex vivo interleukin-1? secretion in hyperimmunoglobulinemia D and periodic fever syndrome. Arthritis and Rheumatism, 2002, 46, 2794-2803.	6.7	165

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19	Identification of PEX7 as the Second Gene Involved in Refsum Disease. American Journal of Human Genetics, 2003, 72, 471-477.	6.2	151
20	Metabolite transport across the peroxisomal membrane. Biochemical Journal, 2007, 401, 365-375.	3.7	142
21	HMG-CoA reductase inhibition induces IL-1Î² release through Rac1/PI3K/PKB-dependent caspase-1 activation. Blood, 2008, 112, 3563-3573.	1.4	129
22	A novel defect of peroxisome division due to a homozygous non-sense mutation in thePEX11Î²gene. Journal of Medical Genetics, 2012, 49, 307-313.	3.2	127
23	Genetic classification and mutational spectrum of more than 600 patients with a Zellweger syndrome spectrum disorder. Human Mutation, 2011, 32, 59-69.	2.5	126
24	Clinical, biochemical, and mutational spectrum of peroxisomal acylâ€“coenzyme A oxidase deficiency. Human Mutation, 2007, 28, 904-912.	2.5	121
25	A novel bile acid biosynthesis defect due to a deficiency of peroxisomal ABCD3. Human Molecular Genetics, 2015, 24, 361-370.	2.9	115
26	Molecular basis of Refsum disease: Sequence variations in Phytanoyl-CoA Hydroxylase (PHYH) and the PTS2 receptor (PEX7). Human Mutation, 2004, 23, 209-218.	2.5	113
27	Mutational spectrum and genotypeâ€“phenotype correlations in mevalonate kinase deficiency. Human Mutation, 2006, 27, 796-802.	2.5	113
28	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.	2.4	113
29	ECHS1 mutations in Leigh disease: a new inborn error of metabolism affecting valine metabolism. Brain, 2014, 137, 2903-2908.	7.6	111
30	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. American Journal of Human Genetics, 2015, 97, 535-545.	6.2	103
31	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.	2.3	101
32	Temperature dependence of mutant mevalonate kinase activity as a pathogenic factor in Hyper-IgD and periodic fever syndrome. Human Molecular Genetics, 2002, 11, 3115-3124.	2.9	97
33	Impaired neuronal migration and endochondral ossification in Pex7 knockout mice: a model for rhizomelic chondrodysplasia punctata. Human Molecular Genetics, 2003, 12, 2255-2267.	2.9	97
34	The peroxisomal ABC transporter family. Pflugers Archiv European Journal of Physiology, 2007, 453, 719-734.	2.8	95
35	Disorders of Peroxisome Biogenesis Due to Mutations in PEX1: Phenotypes and PEX1 Protein Levels. American Journal of Human Genetics, 2001, 69, 35-48.	6.2	92
36	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.	6.2	92

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37	Defects of cholesterol biosynthesis. FEBS Letters, 2006, 580, 5442-5449.	2.8	92
38	ACBD5 deficiency causes a defect in peroxisomal very long-chain fatty acid metabolism. Journal of Medical Genetics, 2017, 54, 330-337.	3.2	90
39	Plasmalogens participate in very-long-chain fatty acid-induced pathology. Brain, 2008, 132, 482-492.	7.6	89
40	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.	2.4	87
41	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.	2.8	85
42	Peroxisomes can oxidize mediumâ€•and longâ€•chain fatty acids through a pathway involving ABCD3 and HSD17B4. FASEB Journal, 2019, 33, 4355-4364.	0.5	82
43	Mutational Spectrum of d-Bifunctional Protein Deficiency and Structure-Based Genotype-Phenotype Analysis. American Journal of Human Genetics, 2006, 78, 112-124.	6.2	80
44	Identification of an unusual variant peroxisome biogenesis disorder caused by mutations in the PEX16 gene. Journal of Medical Genetics, 2010, 47, 608-615.	3.2	80
45	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
46	Statin synergizes with LPS to induce IL-1Î² release by THP-1 cells through activation of caspase-1. Molecular Immunology, 2008, 45, 2158-2165.	2.2	77
47	Identification of the human mitochondrial FAD transporter and its potential role in multiple acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2005, 86, 441-447.	1.1	75
48	Mutations in PEX10 are a cause of autosomal recessive ataxia. Annals of Neurology, 2010, 68, 259-263.	5.3	74
49	A novel type of rhizomelic chondrodysplasia punctata, RCDP5, is caused by loss of the PEX5 long isoform. Human Molecular Genetics, 2015, 24, 5845-5854.	2.9	73
50	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. Journal of Inherited Metabolic Disease, 2016, 39, 93-106.	3.6	73
51	Peroxisomal ABC transporters: functions and mechanism. Biochemical Society Transactions, 2015, 43, 959-965.	3.4	71
52	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in GLS. New England Journal of Medicine, 2019, 380, 1433-1441.	27.0	71
53	Mutational spectrum of Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 263-284.	1.6	69
54	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	2.7	68

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55	Identification and Characterization of Three Novel Missense Mutations in Mevalonate Kinase cDNA Causing Mevalonic Aciduria, a Disorder of Isoprene Biosynthesis. Human Molecular Genetics, 1999, 8, 1523-1528.	2.9	65
56	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.	6.2	65
57	Lipidomic analysis of fibroblasts from Zellweger spectrum disorder patients identifies disease-specific phospholipid ratios. Journal of Lipid Research, 2016, 57, 1447-1454.	4.2	65
58	Regulation of Isoprenoid/Cholesterol Biosynthesis in Cells from Mevalonate Kinase-deficient Patients. Journal of Biological Chemistry, 2003, 278, 5736-5743.	3.4	62
59	A homozygous missense mutation in ERAL1, encoding a mitochondrial rRNA chaperone, causes Perrault syndrome. Human Molecular Genetics, 2017, 26, 2541-2550.	2.9	61
60	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.	2.1	59
61	Autosomal recessive cerebellar ataxia caused by mutations in the PEX2 gene. Orphanet Journal of Rare Diseases, 2011, 6, 8.	2.7	58
62	A novel case of ACOX2 deficiency leads to recognition of a third human peroxisomal acyl-CoA oxidase. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 952-958.	3.8	58
63	Mevalonate kinase is a cytosolic enzyme in humans. Journal of Cell Science, 2004, 117, 631-639.	2.0	57
64	Hyperimmunoglobulinemia D and periodic fever syndrome; treatment with etanercept and follow-up. Clinical Rheumatology, 2008, 27, 1317-1320.	2.2	55
65	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
66	Detection of nonsterol isoprenoids by HPLCâ€“MS/MS. Analytical Biochemistry, 2008, 383, 18-24.	2.4	52
67	Desmosterolosisâ€”phenotypic and molecular characterization of a third case and review of the literature. American Journal of Medical Genetics, Part A, 2011, 155, 1597-1604.	1.2	52
68	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.	2.5	48
69	Fatty acid oxidation flux predicts the clinical severity of VLCAD deficiency. Genetics in Medicine, 2015, 17, 989-994.	2.4	48
70	Functional characterisation of peroxisomal Î²â€oxidation disorders in fibroblasts using lipidomics. Journal of Inherited Metabolic Disease, 2018, 41, 479-487.	3.6	48
71	Adrenoleukodystrophy Newborn Screening in the Netherlands (SCAN Study): The X-Factor. Frontiers in Cell and Developmental Biology, 2020, 8, 499.	3.7	47
72	Phosphomevalonate kinase is a cytosolic protein in humans. Journal of Lipid Research, 2004, 45, 697-705.	4.2	46

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73	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
74	Absence of functional peroxisomes does not lead to deficiency of enzymes involved in cholesterol biosynthesis. Journal of Lipid Research, 2002, 43, 90-98.	4.2	46
75	Inherited disorders of cholesterol biosynthesis. Clinical Genetics, 2002, 61, 393-403.	2.0	45
76	Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. Prenatal Diagnosis, 2008, 28, 309-312.	2.3	45
77	Unprenylated RhoA Contributes to IL-1Î² Hypersecretion in Mevalonate Kinase Deficiency Model through Stimulation of Rac1 Activity. Journal of Biological Chemistry, 2014, 289, 27757-27765.	3.4	45
78	The Peroxisomal NAD Carrier from Arabidopsis Imports NAD in Exchange with AMP. Plant Physiology, 2016, 171, 2127-2139.	4.8	45
79	Identification of novel mutations in classical galactosemia. Human Mutation, 2005, 25, 502-502.	2.5	44
80	The important role of biochemical and functional studies in the diagnostics of peroxisomal disorders. Journal of Inherited Metabolic Disease, 2016, 39, 531-543.	3.6	44
81	Clinical and Biochemical Pitfalls in the Diagnosis of Peroxisomal Disorders. Neuropediatrics, 2016, 47, 205-220.	0.6	41
82	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976.	6.2	41
83	Diagnostic Value of Urinary Mevalonic Acid Excretion in Patients with a Clinical Suspicion of Mevalonate Kinase Deficiency (MKD). JIMD Reports, 2015, 27, 33-38.	1.5	40
84	A novel PEX12 mutation identified as the cause of a peroxisomal biogenesis disorder with mild clinical phenotype, mild biochemical abnormalities in fibroblasts and a mosaic catalase immunofluorescence pattern, even at 40ÂÂ°C. Journal of Human Genetics, 2007, 52, 599-606.	2.3	37
85	Cholic acid therapy in Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2016, 39, 859-868.	3.6	37
86	Evaluation of C26:0â€lysophosphatidylcholine and C26:0â€carnitine as diagnostic markers for Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2017, 40, 875-881.	3.6	37
87	Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127.	1.2	36
88	Impact of newborn screening for veryâ€longâ€chain acylâ€CoA dehydrogenase deficiency on genetic, enzymatic, and clinical outcomes. Journal of Inherited Metabolic Disease, 2019, 42, 414-423.	3.6	36
89	Adult peroxisomal acyl-coenzyme A oxidase deficiency with cerebellar and brainstem atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 310-312.	1.9	35
90	Manipulation of isoprenoid biosynthesis as a possible therapeutic option in mevalonate kinase deficiency. Arthritis and Rheumatism, 2006, 54, 2306-2313.	6.7	33

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91	Absence of functional peroxisomes does not lead to deficiency of enzymes involved in cholesterol biosynthesis. Journal of Lipid Research, 2002, 43, 90-8.	4.2	33
92	Inhibition of the isoprenoid biosynthesis pathway; detection of intermediates by UPLCâ€“MS/MS. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2011, 1811, 227-233.	2.4	31
93	Zellweger Spectrum Disorder with Mild Phenotype Caused by PEX2 Gene Mutations. JIMD Reports, 2012, 6, 43-46.	1.5	31
94	Arginine improves peroxisome functioning in cells from patients with a mild peroxisome biogenesis disorder. Orphanet Journal of Rare Diseases, 2013, 8, 138.	2.7	30
95	Peroxisomal Metabolite and Cofactor Transport in Humans. Frontiers in Cell and Developmental Biology, 2020, 8, 613892.	3.7	30
96	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
97	Spectrum ofPEX6mutations in Zellweger syndrome spectrum patients. Human Mutation, 2010, 31, E1058-E1070.	2.5	28
98	Identification of three patients with a very mild form of Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2003, 122A, 24-29.	2.4	27
99	Human mevalonate pyrophosphate decarboxylase is localized in the cytosol. Molecular Genetics and Metabolism, 2004, 81, 216-224.	1.1	27
100	Pelgerâ€“huet anomaly and a mild skeletal phenotype secondary to mutations in LBR. American Journal of Medical Genetics, Part A, 2013, 161, 2066-2073.	1.2	27
101	NTCP deficiency and persistently raised bile salts: an adult case. Journal of Inherited Metabolic Disease, 2017, 40, 313-315.	3.6	27
102	Compromized geranylgeranylation of RhoA and Rac1 in mevalonate kinase deficiency. Journal of Inherited Metabolic Disease, 2010, 33, 625-632.	3.6	26
103	Barley has two peroxisomal ABC transporters with multiple functions in Î²-oxidation. Journal of Experimental Botany, 2014, 65, 4833-4847.	4.8	26
104	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	3.7	26
105	Compromised Protein Prenylation as Pathogenic Mechanism in Mevalonate Kinase Deficiency. Frontiers in Immunology, 2021, 12, 724991.	4.8	26
106	Fatty Acid Oxidation in Peroxisomes: Enzymology, Metabolic Crosstalk with Other Organelles and Peroxisomal Disorders. Advances in Experimental Medicine and Biology, 2020, 1299, 55-70.	1.6	26
107	Reduced muscle strength in ether lipidâ€deficient mice is accompanied by altered development and function of the neuromuscular junction. Journal of Neurochemistry, 2017, 143, 569-583.	3.9	25
108	Deciphering the potential involvement of PXMP2 and PEX11B in hydrogen peroxide permeation across the peroxisomal membrane reveals a role for PEX11B in protein sorting. Biochimica Et Biophysica Acta - Biomembranes, 2019, 1861, 182991.	2.6	25

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109	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. Genetics in Medicine, 2021, 23, 740-750.	2.4	25
110	Clinical and Laboratory Diagnosis of Peroxisomal Disorders. Methods in Molecular Biology, 2017, 1595, 329-342.	0.9	24
111	Defective lipid remodeling of GPI anchors in peroxisomal disorders, Zellweger syndrome, and rhizomelic chondrodysplasia punctata. Journal of Lipid Research, 2012, 53, 653-663.	4.2	23
112	Peroxisomes and Their Central Role in Metabolic Interaction Networks in Humans. Sub-Cellular Biochemistry, 2018, 89, 345-365.	2.4	23
113	A mutation creating an upstream translation initiation codon in SLC22A5 5â€²UTR is a frequent cause of primary carnitine deficiency. Human Mutation, 2019, 40, 1899-1904.	2.5	23
114	Plasma lipidomics as a diagnostic tool for peroxisomal disorders. Journal of Inherited Metabolic Disease, 2018, 41, 489-498.	3.6	22
115	Novel Mutations in the PEX2 Gene of Four Unrelated Patients with a Peroxisome Biogenesis Disorder. Pediatric Research, 2004, 55, 431-436.	2.3	21
116	Genotype-phenotype correlation in PEX5-deficient peroxisome biogenesis defective cell lines. Human Mutation, 2009, 30, 93-98.	2.5	21
117	A nationwide retrospective observational study of population newborn screening for mediumâ€chain acylâ€CoA dehydrogenase (MCAD) deficiency in the Netherlands. Journal of Inherited Metabolic Disease, 2019, 42, 890-897.	3.6	21
118	Proposal for an individualized dietary strategy in patients with very longâ€chain acylâ€CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 159-168.	3.6	21
119	Cholesterol biosynthesis is not defective in peroxisome biogenesis defective fibroblasts. Molecular Genetics and Metabolism, 2003, 80, 290-295.	1.1	20
120	Favorable Outcome After Physiologic Dose of Sodium-<scp>d,l</scp>-3-Hydroxybutyrate in Severe MADD. Pediatrics, 2014, 134, e1224-e1228.	2.1	20
121	Homozygosity for the V377I mutation in mevalonate kinase causes distinct clinical phenotypes in two sibs with hyperimmunoglobulinaemia D and periodic fever syndrome (HIDS). RMD Open, 2016, 2, e000196.	3.8	20
122	Translational Metabolism: A multidisciplinary approach towards precision diagnosis of inborn errors of metabolism in the omics era. Journal of Inherited Metabolic Disease, 2019, 42, 197-208.	3.6	20
123	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
124	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
125	An UPLC-MS/MS Assay to Measure Glutathione as Marker for Oxidative Stress in Cultured Cells. Metabolites, 2019, 9, 45.	2.9	18
126	The cholic acid extension study in Zellweger spectrum disorders: Results and implications for therapy. Journal of Inherited Metabolic Disease, 2019, 42, 303-312.	3.6	18

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127	Cerebellar and hepatic alterations in ACBD5-deficient mice are associated with unexpected, distinct alterations in cellular lipid homeostasis. Communications Biology, 2020, 3, 713.	4.4	18
128	The SaccharomycesÂcerevisiae ABC subfamily D transporter Pxa1/Pxa2p coâ€imports CoASH into the peroxisome. FEBS Letters, 2021, 595, 763-772.	2.8	18
129	Novel mutations in the PEX12 gene of patients with a peroxisome biogenesis disorder. European Journal of Human Genetics, 2004, 12, 115-120.	2.8	17
130	Mutagenesis separates ATPase and thioesterase activities of the peroxisomal ABC transporter, Comatose. Scientific Reports, 2019, 9, 10502.	3.3	14
131	The Newborn Screening Paradox: Sensitivity vs. Overdiagnosis in VLCAD Deficiency. JIMD Reports, 2015, 27, 101-106.	1.5	13
132	Biochemical and genetic characterization of an unusual mild PEX3- related Zellweger spectrum disorder. Molecular Genetics and Metabolism, 2017, 121, 325-328.	1.1	13
133	Fluorescent Tools to Analyze Peroxisomeâ€“Endoplasmic Reticulum Interactions in Mammalian Cells. Contact (Thousand Oaks (Ventura County, Calif)), 2019, 2, 251525641984864.	1.3	13
134	Liver disease predominates in a mouse model for mild human Zellweger spectrum disorder. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2774-2787.	3.8	12
135	Metabolic functions and biogenesis of peroxisomes in health and disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1325.	3.8	10
136	Autophagy Inhibitors Do Not Restore Peroxisomal Functions in Cells With the Most Common Peroxisome Biogenesis Defect. Frontiers in Cell and Developmental Biology, 2021, 9, 661298.	3.7	10
137	A Functional SMAD2/3 Binding Site in the PEX11Î² Promoter Identifies a Role for TGFÎ² in Peroxisome Proliferation in Humans. Frontiers in Cell and Developmental Biology, 2020, 8, 577637.	3.7	9
138	A review of treatment modalities in gyrate atrophy of the choroid and retina (GACR). Molecular Genetics and Metabolism, 2021, 134, 96-116.	1.1	9
139	Functional analysis of thirty-four suspected pathogenic missense variants in ALDH5A1 gene associated with succinic semialdehyde dehydrogenase deficiency. Molecular Genetics and Metabolism, 2020, 130, 172-178.	1.1	8
140	Fibroblastâ€specific genomeâ€scale modelling predicts an imbalance in amino acid metabolism in Refsum disease. FEBS Journal, 2020, 287, 5096-5113.	4.7	8
141	A mild case of SMVT deficiency illustrating the importance of treatment response in variant classification. Journal of Physical Education and Sports Management, 2022, , mcs.a006185.	1.2	7
142	Novel mutations causing hyperimmunoglobulin d and periodic fever syndrome. Indian Pediatrics, 2012, 49, 583-585.	0.4	6
143	Clinical utility gene card for: Zellweger syndrome spectrum. European Journal of Human Genetics, 2015, 23, 1111-1111.	2.8	6
144	The Challenges of a Successful Pregnancy in a Patient with Adult Refsumâ€™s Disease due to Phytanoyl-CoA Hydroxylase Deficiency. JIMD Reports, 2016, 33, 49-53.	1.5	6

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145	Transfection of Primary Human Skin Fibroblasts for Peroxisomal Studies. Methods in Molecular Biology, 2017, 1595, 63-67.	0.9	5
146	Severe Fat Accumulation in Multiple Organs in Pediatric Autopsies. Pediatric and Developmental Pathology, 2017, 20, 269-276.	1.0	5
147	Identification and diagnostic value of phytanoyl- and pristanoyl-carnitine in plasma from patients with peroxisomal disorders. Molecular Genetics and Metabolism, 2017, 121, 279-282.	1.1	5
148	Normal cognitive outcome in a PEX6 deficient girl despite neonatal multisystem presentation. American Journal of Medical Genetics, Part A, 2016, 170, 1642-1646.	1.2	4
149	Absence of biochemical evidence at an early age delays diagnosis in a patient with a clinically severe peroxisomal biogenesis disorder. European Journal of Paediatric Neurology, 2016, 20, 331-335.	1.6	4
150	Peroxisomal ATP Uptake Is Provided by Two Adenine Nucleotide Transporters and the ABCD Transporters. Frontiers in Cell and Developmental Biology, 2021, 9, 788921.	3.7	4
151	The mouse as a model to understand peroxisomal biogenesis and its disorders. Drug Discovery Today: Disease Models, 2004, 1, 193-198.	1.2	3
152	Oral Cholic Acid in Zellweger Spectrum Disorders. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, e57.	1.8	3
153	Laboratory Diagnosis of Peroxisomal Disorders in the -Omics Era and the Continued Importance of Biomarkers and Biochemical Studies. FIRE Forum for International Research in Education, 2018, 6, 232640981881028.	0.7	3
154	Peroxisomal abnormalities in the immortalized human hepatocyte (IHH) cell line. Histochemistry and Cell Biology, 2017, 147, 537-541.	1.7	2
155	Abnormal <scp>VLCADD</scp> newborn screening resembling <scp>MADD</scp> in four neonates with decreased riboflavin levels and <scp>VLCAD</scp> activity. JIMD Reports, 2021, 61, 12-18.	1.5	2
156	Resolution of the Molecular Defect in a Patient with Peroxisomal Mosaicism in the Liver. Advances in Experimental Medicine and Biology, 2003, 544, 107-111.	1.6	2
157	Proposal for an individualized dietary strategy in patients with very long-chain acyl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 159.	3.6	2
158	Edgetic Perturbations Contribute to Phenotypic Variability in PEX26 Deficiency. Frontiers in Genetics, 2021, 12, 726174.	2.3	2
159	23 as a Tool for Human Gene Function Discovery. Methods in Microbiology, 2007, 36, 577-595.	0.8	1
160	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
161	Disorders of Cholesterol Synthesis. , 2012, , 461-471.		1
162	Chemical chaperones improve peroxisomal biogenesis in fibroblasts of mild Zellweger syndrome spectrum patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 21-21.	0.0	0

#	Article	IF	Citations
163	Oxalate crystals in primary hyperoxaluria type 1 activate the NLRP3 inflammasome. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 37-37.	0.0	0
164	Rapid prenatal diagnosis of fetal Zellweger syndrome by biochemical tests, complementation studies, and molecular analyses. Prenatal Diagnosis, 2013, 33, 201-203.	2.3	0
165	Peroxisomal Disorders. , 2021, , 683-708.		0
166	Disorders of Cholesterol Synthesis. , 2006, , 411-420.		0
167	Disorders of Isoprenoid/Cholesterol Synthesis. , 2016, , 455-464.		0
168	Diagnosis of Inherited Defects of Cholesterol Biosynthesis. , 2008, , 483-495.		0