

Hans R Waterham

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/70714/publications.pdf>

Version: 2024-02-01

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	A mild case of SMVT deficiency illustrating the importance of treatment response in variant classification. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006185.	1.2	7
2	How to proceed after "negative" exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 663-681.	3.6	20
3	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. <i>Genetics in Medicine</i> , 2021, 23, 740-750.	2.4	25
4	The <i>Saccharomyces Cerevisiae</i> ABC subfamily D transporter Pxa1/Pxa2p co-imports CoASH into the peroxisome. <i>FEBS Letters</i> , 2021, 595, 763-772.	2.8	18
5	Autophagy Inhibitors Do Not Restore Peroxisomal Functions in Cells With the Most Common Peroxisome Biogenesis Defect. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 661298.	3.7	10
6	Abnormal VLCADD newborn screening resembling MADD in four neonates with decreased riboflavin levels and VLCAD activity. <i>JIMD Reports</i> , 2021, 61, 12-18.	1.5	2
7	A review of treatment modalities in gyrate atrophy of the choroid and retina (GACR). <i>Molecular Genetics and Metabolism</i> , 2021, 134, 96-116.	1.1	9
8	Compromised Protein Prenylation as Pathogenic Mechanism in Mevalonate Kinase Deficiency. <i>Frontiers in Immunology</i> , 2021, 12, 724991.	4.8	26
9	Peroxisomal Disorders. , 2021, , 683-708.		0
10	Edgetic Perturbations Contribute to Phenotypic Variability in PEX26 Deficiency. <i>Frontiers in Genetics</i> , 2021, 12, 726174.	2.3	2
11	Peroxisomal ATP Uptake Is Provided by Two Adenine Nucleotide Transporters and the ABCD Transporters. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 788921.	3.7	4
12	Genome sequencing in persistently unsolved white matter disorders. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 144-152.	3.7	26
13	A Functional SMAD2/3 Binding Site in the PEX11 ² Promoter Identifies a Role for TGF β 2 in Peroxisome Proliferation in Humans. <i>Frontiers in Cell and Developmental Biology</i> , 2020, 8, 577637.	3.7	9
14	Cerebellar and hepatic alterations in ACBD5-deficient mice are associated with unexpected, distinct alterations in cellular lipid homeostasis. <i>Communications Biology</i> , 2020, 3, 713.	4.4	18
15	Functional analysis of thirty-four suspected pathogenic missense variants in ALDH5A1 gene associated with succinic semialdehyde dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 172-178.	1.1	8
16	Adrenoleukodystrophy Newborn Screening in the Netherlands (SCAN Study): The X-Factor. <i>Frontiers in Cell and Developmental Biology</i> , 2020, 8, 499.	3.7	47
17	Fibroblast-specific genome-scale modelling predicts an imbalance in amino acid metabolism in Refsum disease. <i>FEBS Journal</i> , 2020, 287, 5096-5113.	4.7	8
18	Peroxisomal Metabolite and Cofactor Transport in Humans. <i>Frontiers in Cell and Developmental Biology</i> , 2020, 8, 613892.	3.7	30

#	ARTICLE	IF	CITATIONS
19	Fatty Acid Oxidation in Peroxisomes: Enzymology, Metabolic Crosstalk with Other Organelles and Peroxisomal Disorders. <i>Advances in Experimental Medicine and Biology</i> , 2020, 1299, 55-70.	1.6	26
20	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 105, 534-548.	6.2	46
21	Mutagenesis separates ATPase and thioesterase activities of the peroxisomal ABC transporter, Comatose. <i>Scientific Reports</i> , 2019, 9, 10502.	3.3	14
22	A mutation creating an upstream translation initiation codon in <i>SLC22A5</i> 5'UTR is a frequent cause of primary carnitine deficiency. <i>Human Mutation</i> , 2019, 40, 1899-1904.	2.5	23
23	Liver disease predominates in a mouse model for mild human Zellweger spectrum disorder. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2019, 1865, 2774-2787.	3.8	12
24	Deciphering the potential involvement of PXMP2 and PEX11B in hydrogen peroxide permeation across the peroxisomal membrane reveals a role for PEX11B in protein sorting. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 2019, 1861, 182991.	2.6	25
25	Fluorescent Tools to Analyze Peroxisome-Endoplasmic Reticulum Interactions in Mammalian Cells. <i>Contact (Thousand Oaks (Ventura County, Calif))</i> , 2019, 2, 251525641984864.	1.3	13
26	A nationwide retrospective observational study of population newborn screening for medium-chain acyl-CoA dehydrogenase (MCAD) deficiency in the Netherlands. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 890-897.	3.6	21
27	An UPLC-MS/MS Assay to Measure Glutathione as Marker for Oxidative Stress in Cultured Cells. <i>Metabolites</i> , 2019, 9, 45.	2.9	18
28	The cholic acid extension study in Zellweger spectrum disorders: Results and implications for therapy. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 303-312.	3.6	18
29	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in <i>GLS</i> . <i>New England Journal of Medicine</i> , 2019, 380, 1433-1441.	27.0	71
30	Proposal for an individualized dietary strategy in patients with very long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 159-168.	3.6	21
31	Impact of newborn screening for very-long-chain acyl-CoA dehydrogenase deficiency on genetic, enzymatic, and clinical outcomes. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 414-423.	3.6	36
32	Translational Metabolism: A multidisciplinary approach towards precision diagnosis of inborn errors of metabolism in the omics era. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 197-208.	3.6	20
33	Mutated SUCLG1 causes mislocalization of SUCLG2 protein, morphological alterations of mitochondria and an early-onset severe neurometabolic disorder. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 43-52.	1.1	20
34	Response to Leigh-like syndrome with mild mtDNA depletion due to the SUCLG1 variant c.626C>A. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 18, 10.	1.1	1
35	Peroxisomes can oxidize medium- and long-chain fatty acids through a pathway involving ABCD3 and HSD17B4. <i>FASEB Journal</i> , 2019, 33, 4355-4364.	0.5	82
36	Proposal for an individualized dietary strategy in patients with very long-chain acyl-CoA dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 159.	3.6	2

#	ARTICLE	IF	CITATIONS
37	Clinical, biochemical, and genetic features of four patients with short-chain enoyl-CoA hydratase (ECHS1) deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1115-1127.	1.2	36
38	The role of the clinician in the multi-omics era: are you ready?. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 571-582.	3.6	55
39	A novel case of ACOX2 deficiency leads to recognition of a third human peroxisomal acyl-CoA oxidase. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 952-958.	3.8	58
40	Plasma lipidomics as a diagnostic tool for peroxisomal disorders. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 489-498.	3.6	22
41	Systematic mapping of contact sites reveals tethers and a function for the peroxisome-mitochondria contact. <i>Nature Communications</i> , 2018, 9, 1761.	12.8	222
42	Oral Cholic Acid in Zellweger Spectrum Disorders. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2018, 66, e57.	1.8	3
43	Functional characterisation of peroxisomal β -oxidation disorders in fibroblasts using lipidomics. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 479-487.	3.6	48
44	Laboratory Diagnosis of Peroxisomal Disorders in the -Omics Era and the Continued Importance of Biomarkers and Biochemical Studies. <i>FIRE Forum for International Research in Education</i> , 2018, 6, 232640981881028.	0.7	3
45	Peroxisomes and Their Central Role in Metabolic Interaction Networks in Humans. <i>Sub-Cellular Biochemistry</i> , 2018, 89, 345-365.	2.4	23
46	Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. <i>American Journal of Human Genetics</i> , 2018, 103, 125-130.	6.2	29
47	ACBD5 deficiency causes a defect in peroxisomal very long-chain fatty acid metabolism. <i>Journal of Medical Genetics</i> , 2017, 54, 330-337.	3.2	90
48	Clinical and Laboratory Diagnosis of Peroxisomal Disorders. <i>Methods in Molecular Biology</i> , 2017, 1595, 329-342.	0.9	24
49	Transfection of Primary Human Skin Fibroblasts for Peroxisomal Studies. <i>Methods in Molecular Biology</i> , 2017, 1595, 63-67.	0.9	5
50	A homozygous missense mutation in ERAL1, encoding a mitochondrial rRNA chaperone, causes Perrault syndrome. <i>Human Molecular Genetics</i> , 2017, 26, 2541-2550.	2.9	61
51	Reduced muscle strength in ether lipid-deficient mice is accompanied by altered development and function of the neuromuscular junction. <i>Journal of Neurochemistry</i> , 2017, 143, 569-583.	3.9	25
52	NTCP deficiency and persistently raised bile salts: an adult case. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 313-315.	3.6	27
53	Peroxisomal abnormalities in the immortalized human hepatocyte (IHH) cell line. <i>Histochemistry and Cell Biology</i> , 2017, 147, 537-541.	1.7	2
54	Severe Fat Accumulation in Multiple Organs in Pediatric Autopsies. <i>Pediatric and Developmental Pathology</i> , 2017, 20, 269-276.	1.0	5

#	ARTICLE	IF	CITATIONS
55	Evaluation of C26:0-lysophosphatidylcholine and C26:0-carnitine as diagnostic markers for Zellweger spectrum disorders. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 875-881.	3.6	37
56	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 965-976.	6.2	41
57	Identification and diagnostic value of phytanoyl- and pristanoyl-carnitine in plasma from patients with peroxisomal disorders. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 279-282.	1.1	5
58	Biochemical and genetic characterization of an unusual mild PEX3- related Zellweger spectrum disorder. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 325-328.	1.1	13
59	The important role of biochemical and functional studies in the diagnostics of peroxisomal disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 531-543.	3.6	44
60	Clinical and Biochemical Pitfalls in the Diagnosis of Peroxisomal Disorders. <i>Neuropediatrics</i> , 2016, 47, 205-220.	0.6	41
61	Cholic acid therapy in Zellweger spectrum disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 859-868.	3.6	37
62	The Challenges of a Successful Pregnancy in a Patient with Adult Refsum's Disease due to Phytanoyl-CoA Hydroxylase Deficiency. <i>JIMD Reports</i> , 2016, 33, 49-53.	1.5	6
63	Homozygosity for the V377I mutation in mevalonate kinase causes distinct clinical phenotypes in two sibs with hyperimmunoglobulinaemia D and periodic fever syndrome (HIDS). <i>RMD Open</i> , 2016, 2, e000196.	3.8	20
64	Normal cognitive outcome in a PEX6 deficient girl despite neonatal multisystem presentation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1642-1646.	1.2	4
65	Lipidomic analysis of fibroblasts from Zellweger spectrum disorder patients identifies disease-specific phospholipid ratios. <i>Journal of Lipid Research</i> , 2016, 57, 1447-1454.	4.2	65
66	The Peroxisomal NAD Carrier from Arabidopsis Imports NAD in Exchange with AMP. <i>Plant Physiology</i> , 2016, 171, 2127-2139.	4.8	45
67	Absence of biochemical evidence at an early age delays diagnosis in a patient with a clinically severe peroxisomal biogenesis disorder. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 331-335.	1.6	4
68	Human disorders of peroxisome metabolism and biogenesis. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2016, 1863, 922-933.	4.1	281
69	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 93-106.	3.6	73
70	Disorders of Isoprenoid/Cholesterol Synthesis. , 2016, , 455-464.		0
71	The Newborn Screening Paradox: Sensitivity vs. Overdiagnosis in VLCAD Deficiency. <i>JIMD Reports</i> , 2015, 27, 101-106.	1.5	13
72	Peroxisomal ABC transporters: functions and mechanism. <i>Biochemical Society Transactions</i> , 2015, 43, 959-965.	3.4	71

#	ARTICLE	IF	CITATIONS
73	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	2.7	68
74	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
75	A novel bile acid biosynthesis defect due to a deficiency of peroxisomal ABCD3. Human Molecular Genetics, 2015, 24, 361-370.	2.9	115
76	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
77	Fatty acid oxidation flux predicts the clinical severity of VLCAD deficiency. Genetics in Medicine, 2015, 17, 989-994.	2.4	48
78	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
79	Clinical utility gene card for: Zellweger syndrome spectrum. European Journal of Human Genetics, 2015, 23, 1111-1111.	2.8	6
80	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
81	Unprenylated RhoA Contributes to IL-1 β Hypersecretion in Mevalonate Kinase Deficiency Model through Stimulation of Rac1 Activity. Journal of Biological Chemistry, 2014, 289, 27757-27765.	3.4	45
82	Favorable Outcome After Physiologic Dose of Sodium-3-Hydroxybutyrate in Severe MADD. Pediatrics, 2014, 134, e1224-e1228.	2.1	20
83	Barley has two peroxisomal ABC transporters with multiple functions in β -oxidation. Journal of Experimental Botany, 2014, 65, 4833-4847.	4.8	26
84	ECHS1 mutations in Leigh disease: a new inborn error of metabolism affecting valine metabolism. Brain, 2014, 137, 2903-2908.	7.6	111
85	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
86	Chemical chaperones improve peroxisomal biogenesis in fibroblasts of mild Zellweger syndrome spectrum patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 21-21.	0.0	0
87	Oxalate crystals in primary hyperoxaluria type 1 activate the NLRP3 inflammasome. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 37-37.	0.0	0
88	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
89	Rapid prenatal diagnosis of fetal Zellweger syndrome by biochemical tests, complementation studies, and molecular analyses. Prenatal Diagnosis, 2013, 33, 201-203.	2.3	0
90	Pelger's anomaly and a mild skeletal phenotype secondary to mutations in <i>LBR</i> . American Journal of Medical Genetics, Part A, 2013, 161, 2066-2073.	1.2	27

#	ARTICLE	IF	CITATIONS
91	Zellweger Spectrum Disorder with Mild Phenotype Caused by PEX2 Gene Mutations. JIMD Reports, 2012, 6, 43-46.	1.5	31
92	A novel defect of peroxisome division due to a homozygous non-sense mutation in the <i>PEX11</i> gene. Journal of Medical Genetics, 2012, 49, 307-313.	3.2	127
93	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
94	Mutational spectrum of Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 263-284.	1.6	69
95	Genetics and molecular basis of human peroxisome biogenesis disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1430-1441.	3.8	234
96	Metabolic functions and biogenesis of peroxisomes in health and disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1325.	3.8	10
97	Novel mutations causing hyperimmunoglobulin d and periodic fever syndrome. Indian Pediatrics, 2012, 49, 583-585.	0.4	6
98	Disorders of Cholesterol Synthesis. , 2012, , 461-471.		1
99	Differential substrate specificities of human ABCD1 and ABCD2 in peroxisomal fatty acid β^2 -oxidation. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2011, 1811, 148-152.	2.4	113
100	Inhibition of the isoprenoid biosynthesis pathway; detection of intermediates by UPLC-MS/MS. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2011, 1811, 227-233.	2.4	31
101	Brown-Vialetto-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
102	Autosomal recessive cerebellar ataxia caused by mutations in the PEX2 gene. Orphanet Journal of Rare Diseases, 2011, 6, 8.	2.7	58
103	Desmosterolosis 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
104	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
105	Compromized geranylgeranylation of RhoA and Rac1 in mevalonate kinase deficiency. Journal of Inherited Metabolic Disease, 2010, 33, 625-632.	3.6	26
106	Spectrum of PEX6 mutations in Zellweger syndrome spectrum patients. Human Mutation, 2010, 31, E1058-E1070.	2.5	28
107	Mutations in <i>PEX10</i> are a cause of autosomal recessive ataxia. Annals of Neurology, 2010, 68, 259-263.	5.3	74
108	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

#	ARTICLE	IF	CITATIONS
109	Adult peroxisomal acyl-coenzyme A oxidase deficiency with cerebellar and brainstem atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 310-312.	1.9	35
110	Genotype-phenotype correlation in PEX5-deficient peroxisome biogenesis defective cell lines. <i>Human Mutation</i> , 2009, 30, 93-98.	2.5	21
111	Hyperimmunoglobulinemia D and periodic fever syndrome; treatment with etanercept and follow-up. <i>Clinical Rheumatology</i> , 2008, 27, 1317-1320.	2.2	55
112	Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. <i>Prenatal Diagnosis</i> , 2008, 28, 309-312.	2.3	45
113	Detection of nonsterol isoprenoids by HPLC-MS/MS. <i>Analytical Biochemistry</i> , 2008, 383, 18-24.	2.4	52
114	Statin synergizes with LPS to induce IL-1 β release by THP-1 cells through activation of caspase-1. <i>Molecular Immunology</i> , 2008, 45, 2158-2165.	2.2	77
115	The human peroxisomal ABC half transporter ALDP functions as a homodimer and accepts acyl-CoA esters. <i>FASEB Journal</i> , 2008, 22, 4201-4208.	0.5	200
116	Plasmalogens participate in very-long-chain fatty acid-induced pathology. <i>Brain</i> , 2008, 132, 482-492.	7.6	89
117	HMG-CoA reductase inhibition induces IL-1 β release through Rac1/PI3K/PKB-dependent caspase-1 activation. <i>Blood</i> , 2008, 112, 3563-3573.	1.4	129
118	Diagnosis of Inherited Defects of Cholesterol Biosynthesis. , 2008, , 483-495.		0
119	23 as a Tool for Human Gene Function Discovery. <i>Methods in Microbiology</i> , 2007, 36, 577-595.	0.8	1
120	A Lethal Defect of Mitochondrial and Peroxisomal Fission. <i>New England Journal of Medicine</i> , 2007, 356, 1736-1741.	27.0	665
121	Metabolite transport across the peroxisomal membrane. <i>Biochemical Journal</i> , 2007, 401, 365-375.	3.7	142
122	Clinical, biochemical, and mutational spectrum of peroxisomal acyl-Coenzyme A oxidase deficiency. <i>Human Mutation</i> , 2007, 28, 904-912.	2.5	121
123	The peroxisomal ABC transporter family. <i>Pflugers Archiv European Journal of Physiology</i> , 2007, 453, 719-734.	2.8	95
124	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 40°C. <i>Journal of Human Genetics</i> , 2007, 52, 599-606.	2.3	37
125	Mutational Spectrum of d-Bifunctional Protein Deficiency and Structure-Based Genotype-Phenotype Analysis. <i>American Journal of Human Genetics</i> , 2006, 78, 112-124.	6.2	80
126	Biochemistry of Mammalian Peroxisomes Revisited. <i>Annual Review of Biochemistry</i> , 2006, 75, 295-332.	11.1	853

#	ARTICLE	IF	CITATIONS
127	Defects of cholesterol biosynthesis. FEBS Letters, 2006, 580, 5442-5449.	2.8	92
128	Manipulation of isoprenoid biosynthesis as a possible therapeutic option in mevalonate kinase deficiency. Arthritis and Rheumatism, 2006, 54, 2306-2313.	6.7	33
129	A role for geranylgeranylation in interleukin-1 β secretion. Arthritis and Rheumatism, 2006, 54, 3690-3695.	6.7	169
130	Mutational spectrum and genotype-phenotype correlations in mevalonate kinase deficiency. Human Mutation, 2006, 27, 796-802.	2.5	113
131	Disorders of Cholesterol Synthesis. , 2006, , 411-420.		0
132	Identification of novel mutations in classical galactosemia. Human Mutation, 2005, 25, 502-502.	2.5	44
133	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
134	Novel Mutations in the PEX2 Gene of Four Unrelated Patients with a Peroxisome Biogenesis Disorder. Pediatric Research, 2004, 55, 431-436.	2.3	21
135	Mevalonate kinase is a cytosolic enzyme in humans. Journal of Cell Science, 2004, 117, 631-639.	2.0	57
136	Phosphomevalonate kinase is a cytosolic protein in humans. Journal of Lipid Research, 2004, 45, 697-705.	4.2	46
137	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
138	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
139	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
140	Identification of the molecular defect in patients with peroxisomal mosaicism using a novel method involving culturing of cells at 40 ^\circ C: Implications for other inborn errors of metabolism. Human Mutation, 2004, 24, 130-139.	2.5	48
141	Infefers: An evolving mutation database for auto-inflammatory syndromes. Human Mutation, 2004, 24, 194-198.	2.5	277
142	The mouse as a model to understand peroxisomal biogenesis and its disorders. Drug Discovery Today: Disease Models, 2004, 1, 193-198.	1.2	3
143	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
144	Human mevalonate pyrophosphate decarboxylase is localized in the cytosol. Molecular Genetics and Metabolism, 2004, 81, 216-224.	1.1	27

#	ARTICLE	IF	CITATIONS
145	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
146	Identification of PEX7 as the Second Gene Involved in Refsum Disease. American Journal of Human Genetics, 2003, 72, 471-477.	6.2	151
147	Autosomal Recessive HEM/Greenberg Skeletal Dysplasia Is Caused by 3 β -Hydroxysterol Δ^24 -Reductase Deficiency Due to Mutations in the Lamin B Receptor Gene. American Journal of Human Genetics, 2003, 72, 1013-1017.	6.2	206
148	Cholesterol biosynthesis is not defective in peroxisome biogenesis defective fibroblasts. Molecular Genetics and Metabolism, 2003, 80, 290-295.	1.1	20
149	Regulation of Isoprenoid/Cholesterol Biosynthesis in Cells from Mevalonate Kinase-deficient Patients. Journal of Biological Chemistry, 2003, 278, 5736-5743.	3.4	62
150	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
151	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
152	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
153	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
154	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
155	Lack of isoprenoid products raises ex vivo interleukin-1 β secretion in hyperimmunoglobulinemia D and periodic fever syndrome. Arthritis and Rheumatism, 2002, 46, 2794-2803.	6.7	165
156	Inherited disorders of cholesterol biosynthesis. Clinical Genetics, 2002, 61, 393-403.	2.0	45
157	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
158	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
159	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
160	Mutations in the 3 β -Hydroxysterol Δ^24 -Reductase Gene Cause Desmosterolosis, an Autosomal Recessive Disorder of Cholesterol Biosynthesis. American Journal of Human Genetics, 2001, 69, 685-694.	6.2	318
161	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. Human Mutation, 2001, 18, 499-515.	2.5	261
162	Organization of the mevalonate kinase (MVK) gene and identification of novel mutations causing mevalonic aciduria and hyperimmunoglobulinemia D and periodic fever syndrome. European Journal of Human Genetics, 2001, 9, 253-259.	2.8	85

#	ARTICLE	IF	CITATIONS
163	Identification of a Peroxisomal ATP Carrier Required for Medium-Chain Fatty Acid β -Oxidation and Normal Peroxisome Proliferation in <i>Saccharomyces cerevisiae</i> . <i>Molecular and Cellular Biology</i> , 2001, 21, 4321-4329.	2.3	101
164	Mutations in the gene encoding peroxisomal β -methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. <i>Nature Genetics</i> , 2000, 24, 188-191.	21.4	241
165	Identification and Characterization of Three Novel Missense Mutations in Mevalonate Kinase cDNA Causing Mevalonic Aciduria, a Disorder of Isoprene Biosynthesis. <i>Human Molecular Genetics</i> , 1999, 8, 1523-1528.	2.9	65
166	Mutations in MVK, encoding mevalonate kinase, cause hyperimmunoglobulinaemia D and periodic fever syndrome. <i>Nature Genetics</i> , 1999, 22, 175-177.	21.4	480
167	Molecular Cloning and Expression of Human Carnitine Octanoyltransferase: Evidence for Its Role in the Peroxisomal β -Oxidation of Branched-Chain Fatty Acids. <i>Biochemical and Biophysical Research Communications</i> , 1999, 263, 213-218.	2.1	59
168	Smith-Lemli-Opitz Syndrome Is Caused by Mutations in the 7-Dehydrocholesterol Reductase Gene. <i>American Journal of Human Genetics</i> , 1998, 63, 329-338.	6.2	271