Identification of PEX7 as the Second Gene Involved in R

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Citation Report

#	Article	IF	Citations
1	Rare forms of autosomal recessive neurodegenerative ataxia. Seminars in Pediatric Neurology, 2003, 10, 183-192.	2.0	44
2	The WD-repeat protein superfamily in Arabidopsis: conservation and divergence in structure and function. BMC Genomics, 2003, 4, 50.	2.8	256
3	Alpha-oxidation of 3-methyl-substituted fatty acids and its thiamine dependence. FEBS Journal, 2003, 270, 1619-1627.	0.2	41
4	The chemical biology of branched-chain lipid metabolism. Progress in Lipid Research, 2003, 42, 359-376.	11.6	71
5	PEROXISOMEBIOGENESISDISORDERS. Annual Review of Genomics and Human Genetics, 2003, 4, 165-211.	6.2	176
6	Peroxisome Proliferator-Activated Receptors, Fatty Acid Oxidation, Steatohepatitis and Hepatocarcinogenesis. Current Molecular Medicine, 2003, 3, 561-572.	1.3	163
7	Pex7p translocates in and out of peroxisomes in Saccharomyces cerevisiae. Journal of Cell Biology, 2004, 167, 599-604.	5.2	112
8	A new locus for a childhood onset, slowly progressive autosomal recessive spinocerebellar ataxia maps to chromosome 11p15. Journal of Medical Genetics, 2004, 41, 858-866.	3.2	29
9	Smell testing: an additional tool for identification of adult Refsum's disease. Journal of Neurology, Neurosurgery and Psychiatry, 2004, 75, 1334-1336.	1.9	26
10	Differential diagnosis of Charcot-Marie-Tooth disease and related neuropathies. Neurological Sciences, 2004, 25, 72-82.	1.9	51
11	Metabolic and molecular basis of peroxisomal disorders: A review. American Journal of Medical Genetics Part A, 2004, 126A, 355-375.	2.4	140
12	Molecular genetics of the ichthyoses. American Journal of Medical Genetics Part A, 2004, 131C, 32-44.	2.4	53
13	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
14	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
15	The cornified envelope: a model of cell death in the skin. Nature Reviews Molecular Cell Biology, 2005, 6, 328-340.	37.0	1,474
17	The Arabidopsis Peroxisomal Targeting Signal Type 2 Receptor PEX7 Is Necessary for Peroxisome Function and Dependent on PEX5. Molecular Biology of the Cell, 2005, 16, 573-583.	2.1	136
18	Structure of Human Phytanoyl-CoA 2-Hydroxylase Identifies Molecular Mechanisms of Refsum Disease*. Journal of Biological Chemistry, 2005, 280, 41101-41110.	3.4	78
19	Characterization of phytanic acid Ï‰-hydroxylation in human liver microsomes. Molecular Genetics and Metabolism, 2005, 85, 190-195.	1.1	13

#	Article	IF	Citations
20	Autosomal recessive cerebellar ataxias. Orphanet Journal of Rare Diseases, 2006, 1, 47.	2.7	145
21	Identification of the cytochrome P450 enzymes responsible for the Ï‰-hydroxylation of phytanic acid. FEBS Letters, 2006, 580, 3794-3798.	2.8	22
22	Refsumâ€™s disease may mimic familial Guillain Barre syndrome. Neuromuscular Disorders, 2006, 16, 805-808.	0.6	9
23	Clinical variation in X-linked dominant chondrodysplasia punctata (X-linked dominant ichthyosis). British Journal of Dermatology, 2006, 154, 766-769.	1.5	21
24	Phytanic acid: production from phytol, its breakdown and role in human disease. Cellular and Molecular Life Sciences, 2006, 63, 1752-1765.	5.4	108
25	Paediatric and adult ataxias (update 5). European Journal of Paediatric Neurology, 2006, 10, 249-253.	1.6	3
26	Chapter 3.1.7. The import receptor Pex7p and the PTS2 targeting sequence. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 1599-1604.	4.1	122
27	Peroxisome biogenesis disorders. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 1733-1748.	4.1	433
28	Fatty Acid Metabolism in Skeletal Muscle and Nerve, and in Neuromuscular Disorders. Food Additives, 2007, , 1197-1227.	0.1	0
29	Dr Brian Gibberd (1931â€“2006): a pioneering clinician in Refsum's disease. Biochemical Society Transactions, 2007, 35, 862-864.	3.4	2
30	Structural and mechanistic studies on the peroxisomal oxygenase phytanoyl-CoA 2-hydroxylase (PhyH). Biochemical Society Transactions, 2007, 35, 870-875.	3.4	22
31	Peroxisomal disorders affecting phytanic acid Î±-oxidation: a review. Biochemical Society Transactions, 2007, 35, 881-886.	3.4	61
32	Phenotype of adult Refsum disease due to a defect in peroxin 7. Neurology, 2007, 68, 698-700.	1.1	37
33	Clinical features and molecular genetics of autosomal recessive cerebellar ataxias. Lancet Neurology, The, 2007, 6, 245-257.	10.2	264
34	Phytanic acid impairs mitochondrial respiration through protonophoric action. Cellular and Molecular Life Sciences, 2007, 64, 3271-3281.	5.4	51
35	Non-manifesting Refsum heterozygotes carrying the c.135-2A>G PAHX gene transition. Neurological Sciences, 2008, 29, 173-175.	1.9	27
36	From Human Genetics and Genomics to Pharmacogenetics and Pharmacogenomics: Past Lessons, Future Directions. Drug Metabolism Reviews, 2008, 40, 187-224.	3.6	162
37	Refsum disease due to the splice-site mutation c.135-2A>G before exon 3 of the PHYH gene, diagnosed eight years after detection of retinitis pigmentosa. Journal of the Neurological Sciences, 2008, 266, 182-186.	0.6	7

#	Article	IF	Citations
38	Midlife diagnosis of Refsum Disease in siblings with Retinitis Pigmentosa â€“ the footprint is the clue: a case report. Journal of Medical Case Reports, 2008, 2, 80.	0.8	6
39	Thematic review series: Skin Lipids. Pathogenesis of permeability barrier abnormalities in the ichthyoses: inherited disorders of lipid metabolism. Journal of Lipid Research, 2008, 49, 697-714.	4.2	171
40	A novel Refsum-like disorder that maps to chromosome 20. Neurology, 2009, 72, 20-27.	1.1	38
41	Ichthyoses â€“ Part 1: Differential diagnosis of vulgar ichthyoses and therapeutic options. JDDG - Journal of the German Society of Dermatology, 2009, 7, 511-519.	0.8	13
42	Ichthyosen Ã¢Â€Â“ Teil 1: Differentialdiagnose vulgÃƒÂrer Ichthyosen und therapeutische ErwÃƒÂgungen. JDDG - Journal of the German Society of Dermatology, 2009, 7, 511-520.	0.8	18
43	Peroxisomal Disorders and Neurological Disease. , 2009, , 579-588.		1
44	Second trimester prenatal diagnosis of rhizomelic chondrodysplasia punctata type 1 on ultrasound findings. Prenatal Diagnosis, 2010, 30, 162-164.	2.3	6
45	Autosomal recessive cerebellar ataxias. , 2010, , 189-228.		1
46	Inherited Clinical Disorders of Lipid Metabolism. Current Problems in Dermatology, 2010, 39, 30-88.	0.7	12
47	Adult Refsum Disease: A Form of Tapetoretinal Dystrophy Accessible to Therapy. Survey of Ophthalmology, 2010, 55, 531-538.	4.0	35
48	Biochemistry and genetics of inherited disorders of peroxisomal fatty acid metabolism. Journal of Lipid Research, 2010, 51, 2863-2895.	4.2	274
49	Refsum's Diseaseâ€"Use of the Intestinal Lipase Inhibitor, Orlistat, as a Novel Therapeutic Approach to a Complex Disorder. Journal of Obesity, 2011, 2011, 1-5.	2.7	5
50	Association Between Peroxisomal Biogenesis Factor 7 and Autism Spectrum Disorders in a Korean Population. Journal of Child Neurology, 2012, 27, 1270-1275.	1.4	10
52	Clinical diagnosis, biochemical findings and MRI spectrum of peroxisomal disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1421-1429.	3.8	69
53	Molecular basis of peroxisomal biogenesis disorders caused by defects in peroxisomal matrix protein import. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1326-1336.	3.8	35
54	The importance of ether-phospholipids: A view from the perspective of mouse models. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1501-1508.	3.8	65
55	Longâ€term strategies for the treatment of Refsum's disease using therapeutic apheresis. Journal of Clinical Apheresis, 2012, 27, 99-105.	1.3	25
56	Isolated and syndromic forms of congenital anosmia. Clinical Genetics, 2012, 81, 210-215.	2.0	66

#	Article	IF	Citations
57	Functional characterization of novel mutations in GNPAT and AGPS, causing rhizomelic chondrodysplasia punctata (RCDP) types 2 and 3. Human Mutation, 2012, 33, 189-197.	2.5	62
58	Peroxisome biogenesis disorders: Biological, clinical and pathophysiological perspectives. Developmental Disabilities Research Reviews, 2013, 17, 187-196.	2.9	122
59	Retinitis Pigmentosa and Allied Disorders. , 2013, , 761-835.		34
60	Maladies peroxysomales. , 2013, , 363-370.		0
61	Refsum Disease Presenting with a Late-Onset Leukodystrophy. JIMD Reports, 2014, 19, 7-10.	1.5	3
62	Refsumâ€™s Disease and Cochlear Implantation. Annals of Otology, Rhinology and Laryngology, 2014, 123, 425-427.	1.1	10
63	Systemic Diseases Associated with Retinal Dystrophies. Seminars in Ophthalmology, 2014, 29, 319-328.	1.6	44
64	Therapies for Ataxias. Current Treatment Options in Neurology, 2014, 16, 300.	1.8	3
65	JIMD Reports, Volume 19. JIMD Reports, 2015, , .	1.5	0
66	Peroxisomal Disorders: A Review on Cerebellar Pathologies. Brain Pathology, 2015, 25, 663-678.	4.1	33
67	Novel ABHD12 Mutations in PHARC Patients. Annals of Otology, Rhinology and Laryngology, 2015, 124, 77S-83S.	1.1	21
68	Peripheral Neuropathy in Inherited Metabolic Disease. , 2015, , 353-378.		0
69	Peroxisome biogenesis disorders. Translational Science of Rare Diseases, 2016, 1, 111-144.	1.5	44
70	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
71	Clinical and Biochemical Pitfalls in the Diagnosis of Peroxisomal Disorders. Neuropediatrics, 2016, 47, 205-220.	0.6	41
72	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
73	CCT2 Mutations Evoke Leber Congenital Amaurosis due to Chaperone Complex Instability. Scientific Reports, 2016, 6, 33742.	3.3	27
74	Safety of long-term restrictive diets for peroxisomal disorders: vitamin and trace element status of patients treated for Adult Refsum Disease. International Journal of Clinical Practice, 2016, 70, 229-235.	1.7	7

#	Article	IF	Citations
75	Peroxisomes in brain development and function. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 934-955.	4.1	135
76	Human disorders of peroxisome metabolism and biogenesis. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 922-933.	4.1	281
77	Peroxisomes. Methods in Molecular Biology, 2017, , .	0.9	3
78	Clinical and Laboratory Diagnosis of Peroxisomal Disorders. Methods in Molecular Biology, 2017, 1595, 329-342.	0.9	24
79	Cochlear Implantation in Siblings With Refsumâ€™s Disease. Annals of Otology, Rhinology and Laryngology, 2017, 126, 611-614.	1.1	4
80	From peroxisomal disorders to common neurodegenerative diseases â€“ the role of ether phospholipids in the nervous system. FEBS Letters, 2017, 591, 2761-2788.	2.8	97
81	Differentiating Familial Neuropathies from Guillain-BarrÃ© Syndrome. Pediatric Clinics of North America, 2017, 64, 231-252.	1.8	3
83	Phytol and Phytyl Fatty Acid Esters: Occurrence, Concentrations, and Relevance. European Journal of Lipid Science and Technology, 2018, 120, 1700387.	1.5	16
84	Syndromic Hearing Loss: A Brief Review of Common Presentations and Genetics. Journal of Pediatric Genetics, 2018, 07, 001-008.	0.7	39
85	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
86	Olfactory Dysfunction in Patients WithCNGB1-Associated Retinitis Pigmentosa. JAMA Ophthalmology, 2018, 136, 761.	2.5	11
87	Current and Promising Therapies in Autosomal Recessive Ataxias. CNS and Neurological Disorders - Drug Targets, 2018, 17, 161-171.	1.4	9
88	22 Retinal Manifestations of Metabolic Disease. , 2018, , .		0
89	The Classification of Autosomal Recessive Cerebellar Ataxias: a Consensus Statement from the Society for Research on the Cerebellum and Ataxias Task Force. Cerebellum, 2019, 18, 1098-1125.	2.5	80
92	Genotype-Phenotype Correlation in Retinal Degenerations. Essentials in Ophthalmology, 2019, , 323-336.	0.1	0
93	Recent Advances in the Treatment of Cerebellar Disorders. Brain Sciences, 2020, 10, 11.	2.3	12
94	Comprehensive Exonic Sequencing of Known Ataxia Genes in Episodic Ataxia. Biomedicines, 2020, 8, 134.	3.2	8
96	Sarcoptic mange severity is associated with reduced genomic variation and evidence of selection in Yellowstone National Park wolves (Canis lupus). Evolutionary Applications, 2021, 14, 429-445.	3.1	13

#	Article	IF	Citations
97	Clinical, biochemical, and molecular characterization of mild (nonclassic) rhizomelic chondrodysplasia punctata. Journal of Inherited Metabolic Disease, 2021, 44, 1021-1038.	3.6	19
98	â€œAutopsy Eyeâ€: The Eye in Systemic Disease. , 2021, , 275-304.		0
99	Twins with PEX7 related intellectual disability and cataract: Highlighting phenotypes of peroxisome biogenesis disorder 9B. American Journal of Medical Genetics, Part A, 2021, 185, 1504-1508.	1.2	2
100	Peroxisomal Disorders and Their Mouse Models Point to Essential Roles of Peroxisomes for Retinal Integrity. International Journal of Molecular Sciences, 2021, 22, 4101.	4.1	12
101	Genetic epidemiology approach to estimating birth incidence and current disease prevalence for rhizomelic chondrodysplasia punctata. Orphanet Journal of Rare Diseases, 2021, 16, 300.	2.7	4
102	Peroxisomal Disorders. , 2021, , 683-708.		0
105	Peroxisomal Disorders. , 2009, , 631-670.		2
106	Phenotypic Variability (Heterogeneity) of Peroxisomal Disorders. Advances in Experimental Medicine and Biology, 2003, 544, 9-30.	1.6	10
107	Peroxisomal Fatty Acid Alpha-and Beta-Oxidation in Health and Disease: New insights. Advances in Experimental Medicine and Biology, 2003, 544, 293-302.	1.6	17
108	Clinical Features & Retinal Function In Patients With Adult Refsum Syndrome. Advances in Experimental Medicine and Biology, 2004, 544, 57-58.	1.6	6
109	Identification of PEX7 as the Second Gene Involved in Refsum Disease. Advances in Experimental Medicine and Biology, 2003, 544, 69-70.	1.6	13
110	TRiC/CCT Chaperonin: Structure and Function. Sub-Cellular Biochemistry, 2019, 93, 625-654.	2.4	26
111	Enzymes, Metabolic Pathways, Flux Control Analysis, and the Enzymology of Specific Groups of Inherited Metabolic Diseases. , 2010, , 283-303.		1
112	Peroxisomal Disorders. , 2012, , 591-605.		2
113	Human Disorders of Peroxisome Biogenesis: Zellweger Spectrum and Rhizomelic Chondrodysplasia Punctata. , 2014, , 63-90.		4
114	Peroxisomal Disorders. , 2019, , 107-136.		1
115	Peripheral nervous system plasmalogens regulate Schwann cell differentiation and myelination. Journal of Clinical Investigation, 2014, 124, 2560-2570.	8.2	103
116	Phytyl fatty acid esters in vegetables pose a risk for patients suffering from Refsumâ€™s disease. PLoS ONE, 2017, 12, e0188035.	2.5	8

#	Article	IF	Citations
117	Pexophagy: Molecular Mechanisms and Implications for Health and Diseases. Molecules and Cells, 2018, 41, 55-64.	2.6	71
118	Ophthalmic Diagnosis and Novel Management of Infantile Refsum Disease with Combination Docosahexaenoic Acid and Cholic Acid. Case Reports in Ophthalmological Medicine, 2021, 2021, 1-5.	0.5	1
119	Inherited Neuropathies. , 2004, , 905-951.		19
120	Peroxisomes and Peroxisomal Disorders. , 2005, , 151-153.		0
121	Refsum Disease. , 2005, , 191-194.		0
122	Peroxisomal Disorders. , 2006, , 509-522.		1
123	Cardiac Involvement in Skeletal Myopathies and Neuromuscular Disorders. , 2007, , 2385-2407.		0
125	Refsum Disease- a Disorder of Peroxisomal Alpha-oxidation. , 2010, , 21-25.		0
126	Genetic Sensorineural Hearing Loss. , 2010, , 2086-2099.		1
127	Molecular diagnosis of autosomal recessive cerebellar ataxia in the whole exome/genome sequencing era. World Journal of Neurology, 2013, 3, 115.	0.6	2
128	Adult Refsum Disease (ARD). , 2014, , 267-270.		1
129	Inborn Errors of Non-Mitochondrial Fatty Acid Metabolism Including Peroxisomal Disorders. , 2016, , 591-606.		1
130	Enzyme Diagnostics in a Changing World of Exome Sequencing and Newborn Screening as Exemplified for Peroxisomal, Mitochondrial, and Lysosomal Disorders. , 2017, , 461-487.		0
131	Papulosquamous Diseases. , 2017, , 23-59.		0
132	Human Nervous System Disorders. Advances in Bioinformatics and Biomedical Engineering Book Series, 2019, , 468-528.	0.4	0
133	Peroxisomal disorders. , 2020, , 931-942.		0
134	Disorders of peroxisomal metabolism in adults. , 2020, , 2157-2173.		0
135	Pex7 selectively imports PTS2 target proteins to peroxisomes and is required for anthracnose disease development in Colletotrichum scovillei. Fungal Genetics and Biology, 2021, 157, 103636.	2.1	3

#	Article	IF	Citations
136	Pregnancy outcome in Refsum disease: Affected fetuses and children born to an affected mother. JIMD Reports, 2019, 46, 11-15.	1.5	0
137	On the differential diagnosis of neuropathy in neurogenetic disorders. Medizinische Genetik, 2020, 32, 243-261.	0.2	0
138	Dynamic lipid turnover in photoreceptors and retinal pigment epithelium throughout life. Progress in Retinal and Eye Research, 2022, 89, 101037.	15.5	31
139	Treatment and Management of Autosomal Recessive Cerebellar Ataxias: Current Advances and Future Perspectives. CNS and Neurological Disorders - Drug Targets, 2023, 22, 678-697.	1.4	1
141	Pregnancy outcome in Refsum disease: Affected fetuses and children born to an affected mother. JIMD Reports, 2019, 46, 11-15.	1.5	2
142	Retinal dystrophies: A look beyond the eyes. American Journal of Ophthalmology Case Reports, 2022, 27, 101613.	0.7	1
143	A Pex7 Deficient Mouse Series Correlates Biochemical and Neurobehavioral Markers to Genotype Severityâ€"Implications for the Disease Spectrum of Rhizomelic Chondrodysplasia Punctata Type 1. Frontiers in Cell and Developmental Biology, 0, 10, .	3.7	6
144	The physiological functions of human peroxisomes. Physiological Reviews, 2023, 103, 957-1024.	28.8	43
145	Antenatal ultrasonographic diagnosis of rhizomelic chondrodysplasia punctata. Journal of Ultrasound, 0, , .	1.3	0
146	Genetics of congenital olfactory dysfunction: a systematic review of the literature. Chemical Senses, 2022, 47, .	2.0	2
147	Ichthyosis. Nature Reviews Disease Primers, 2023, 9, .	30.5	12
148	An Investigation of the Prognostic Role of Genes Related to Lipid Metabolism in Head and Neck Squamous Cell Carcinoma. International Journal of Genomics, 2023, 2023, 1-18.	1.6	1
149	Adult Refsum Disease in Puerto Rico: A Case Report. Cureus, 2023, , .	0.5	0
150	PHYH c.678+5G>T Leads to In-Frame Exon Skipping and Is Associated With Attenuated Refsum Disease. , 2024, 65, 38.		0