Nobuyuki Shimozawa

List of Publications by Year
in descending order

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163 papers	4,778 citations	94433 37 h-index	128289 60 g-index
165 all docs	165 docs citations	165 times ranked	3390 citing authors

#	Article	IF	Citations
1	A human gene responsible for Zellweger syndrome that affects peroxisome assembly. Science, 1992, 255, 1132-1134.	12.6	363
2	Proteomic Analysis of Rat Liver Peroxisome. Journal of Biological Chemistry, 2004, 279, 421-428.	3.4	243
3	Mutation in PEX16 Is Causal in the Peroxisome-Deficient Zellweger Syndrome of Complementation Group D. American Journal of Human Genetics, 1998, 63, 1622-1630.	6.2	156
4	Peroxisome assembly factorâ€“2, a putative ATPase cloned by functional complementation on a peroxisomeâ€“deficient mammalian cell mutant. Nature Genetics, 1995, 11, 395-401.	21.4	117
5	d-3-Hydroxyacyl-CoA Dehydratase/d-3-Hydroxyacyl-CoA Dehydrogenase Bifunctional Protein Deficiency: A Newly Identified Peroxisomal Disorder. American Journal of Human Genetics, 1997, 61, 1153-1162.	6.2	107
6	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. Neurogenetics, 2013, 14, 225-232.	1.4	104
7	Complementation study of peroxisome-deficient disorders by immunofluorescence staining and characterization of fused cells. Human Genetics, 1992, 88, 491-499.	3.8	100
8	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.	2.3	99
9	Gas chromatography/mass spectrometry analysis of very long chain fatty acids, docosahexaenoic acid, phytanic acid and plasmalogen for the screening of peroxisomal disorders. Brain and Development, 2003, 25, 481-487.	1.1	88
10	Mucopolysaccharidosis IV A: Molecular Cloning of the Human N-Acetylgalactosamine-6-sulfatase Gene (GALNS) and Analysis of the 5â€²-Flanking Region. Genomics, 1994, 20, 99-104.	2.9	84
11	Mutations in Novel Peroxin Gene PEX26 That Cause Peroxisome-Biogenesis Disorders of Complementation Group 8 Provide a Genotype-Phenotype Correlation. American Journal of Human Genetics, 2003, 73, 233-246.	6.2	71
12	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. Neurology, 2014, 82, 705-712.	1.1	71
13	PEX3 Is the Causal Gene Responsible for Peroxisome Membrane Assemblyâ€“Defective Zellweger Syndrome of Complementation Group G. American Journal of Human Genetics, 2000, 67, 976-981.	6.2	69
14	Mucopolysaccharidosis IVA: screening and identification of mutations of the N-acetylgalactosamine-6-sulfate sulfatase gene. Human Molecular Genetics, 1995, 4, 341-349.	2.9	68
15	A Cytoplasmic AAA Family Peroxin, Pex1p, Interacts with Pex6p. Biochemical and Biophysical Research Communications, 1998, 245, 883-886.	2.1	67
16	Identification of a new complementation group of the peroxisome biogenesis disorders andPEX14 as the mutated gene. Human Mutation, 2004, 23, 552-558.	2.5	67
17	Genetic Basis of Peroxisome-Assembly Mutants of Humans, Chinese Hamster Ovary Cells, and Yeast: Identification of a New Complementation Group of Peroxisome-Biogenesis Disorders Apparently Lacking Peroxisomal-Membrane Ghosts. American Journal of Human Genetics, 1998, 63, 1898-1906.	6.2	65
18	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

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19	Effects of sodium 2-[5-(4-chlorophenyl)pentyl]-oxirane-2-carboxylate (POCA) on fatty acid oxidation in fibroblasts from patients with peroxisomal diseases. Biochemical Pharmacology, 1991, 41, 453-456.	4.4	64
20	Molecular analysis of peroxisomal Î²-oxidation enzymes in infants with Zellweger syndrome and Zellweger-like syndrome: Further heterogeneity of the peroxisomal disorder. Clinica Chimica Acta, 1988, 172, 65-76.	1.1	62
21	BLM (the Causative Gene of Bloom Syndrome) Protein Translocation into the Nucleus by a Nuclear Localization Signal. Biochemical and Biophysical Research Communications, 1997, 240, 348-353.	2.1	62
22	Temperature-Sensitive Phenotypes of Peroxisome-Assembly Processes Represent the Milder Forms of Human Peroxisome-Biogenesis Disorders. American Journal of Human Genetics, 1998, 62, 1539-1543.	6.2	61
23	Functional Heterogeneity of C-Terminal Peroxisome Targeting Signal 1 in PEX5-Defective Patients. Biochemical and Biophysical Research Communications, 1999, 262, 504-508.	2.1	58
24	Newly Identified Chinese Hamster Ovary Cell Mutants Are Defective in Biogenesis of Peroxisomal Membrane Vesicles (Peroxisomal Ghosts), Representing a Novel Complementation Group in Mammals. Journal of Biological Chemistry, 1998, 273, 24122-24130.	3.4	56
25	Rapid UPLC-MS/MS method for routine analysis of plasma pristanic, phytanic, and very long chain fatty acid markers of peroxisomal disorders. Journal of Lipid Research, 2008, 49, 1855-1862.	4.2	53
26	Natural history of X-linked adrenoleukodystrophy in Japan. Brain and Development, 2005, 27, 353-357.	1.1	50
27	Peroxisomal assembly defects: Clinical, pathologic, and biochemical findings in two patients in a newly identified complementation group. Journal of Pediatrics, 1995, 127, 596-599.	1.8	48
28	Role of Pex19p in the targeting of PMP70 to peroxisome. Biochimica Et Biophysica Acta - Molecular Cell Research, 2005, 1746, 116-128.	4.1	47
29	Mucopolysaccharidosis type IVA: common double deletion in the N-acetylgalactosamine-6-sulfatase gene (GALNS). Genomics, 1995, 26, 535-542.	2.9	45
30	Phenotypeâ€“genotype relationships in peroxisome biogenesis disorders of PEX1-defective complementation group 1 are defined by Pex1pâ€“Pex6p interaction. Biochemical Journal, 2001, 357, 417-426.	3.7	45
31	Epidemiology of X-linked adrenoleukodystrophy in Japan. Journal of Human Genetics, 2002, 47, 0590-0593.	2.3	45
32	Biochemical and Morphologic Aspects of Peroxisomes in the Human Rectal Mucosa: Diagnosis of Zellweger Syndrome Simplified by Rectal Biopsy. Pediatric Research, 1988, 24, 723-727.	2.3	44
33	Development of a High-Throughput Microarray-Based Resequencing System for Neurological Disorders and Its Application to Molecular Genetics of Amyotrophic Lateral Sclerosis. Archives of Neurology, 2008, 65, 1326-32.	4.5	44
34	Mucopolysaccharidosis type I: Identification of common mutations that cause Hurler and Scheie syndromes in Japanese populations. , 1996, 7, 23-29.		43
35	Mutational and structural analysis of Japanese patients with mucopolysaccharidosis type II. Journal of Human Genetics, 2005, 50, 395-402.	2.3	43
36	Epilepsy in Peroxisomal Diseases. Epilepsia, 1997, 38, 182-188.	5.1	41

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37	Isolation and Characterization of Peroxisome-Deficient Chinese Hamster Ovary Cell Mutants Representing Human Complementation Group III. Experimental Cell Research, 1997, 233, 11-20.	2.6	39
38	Isolation of a New Peroxisome-Deficient CHO Cell Mutant Defective in Peroxisome Targeting Signal-1 Receptor. Biochemical and Biophysical Research Communications, 1997, 230, 402-406.	2.1	38
39	Mucopolysaccharidosis IVA: Submicroscopic deletion of 16q24.3 and a novel R386C mutation of n-acetylgalactosamine-6-sulfate sulfatase gene in a classical Morquio disease. Human Mutation, 1996, 7, 123-134.	2.5	37
40	Fourteen novel mucopolysaccharidosis IVA producing mutations in GALNS gene. Human Mutation, 1997, 10, 368-375.	2.5	37
41	Isolation and Characterization of Novel Peroxisome Biogenesis-Defective Chinese Hamster Ovary Cell Mutants Using Green Fluorescent Protein. Experimental Cell Research, 1999, 248, 489-497.	2.6	37
42	Zellweger-like syndrome with detectable hepatic peroxisomes: A variant form of peroxisomal disorder. Journal of Pediatrics, 1988, 113, 841-845.	1.8	36
43	Congenital myotonic dystrophy: report of paternal transmission. Brain and Development, 2000, 22, 132-134.	1.1	34
44	ABC Subfamily D Proteins and Very Long Chain Fatty Acid Metabolism as Novel Targets in Adrenoleukodystrophy. Current Drug Targets, 2011, 12, 694-706.	2.1	34
45	Peroxisomal acyl CoA oxidase deficiency. Journal of Pediatrics, 2002, 140, 128-130.	1.8	33
46	Changes of Autonomic Nervous System Function in Patients With Breath-Holding Spells Treated With Iron. Journal of Child Neurology, 2002, 17, 337-340.	1.4	32
47	Tysnd1 Deficiency in Mice Interferes with the Peroxisomal Localization of PTS2 Enzymes, Causing Lipid Metabolic Abnormalities and Male Infertility. PLoS Genetics, 2013, 9, e1003286.	3.5	32
48	Mucopolysaccharidosis type IVA: identification of six novel mutations among non-Japanese patients. Human Molecular Genetics, 1995, 4, 741-743.	2.9	31
49	Temperature-Sensitive Mutation of PEX6 in Peroxisome Biogenesis Disorders in Complementation Group C (CG-C): Comparative Study of PEX6 and PEX1. Pediatric Research, 2000, 48, 541-545.	2.3	31
50	Molecular and clinical findings and diagnostic flowchart of peroxisomal diseases. Brain and Development, 2011, 33, 770-776.	1.1	31
51	Identification of novel SNPs of ABCD1, ABCD2, ABCD3, and ABCD4 genes in patients with X-linked adrenoleukodystrophy (ALD) based on comprehensive resequencing and association studies with ALD phenotypes. Neurogenetics, 2011, 12, 41-50.	1.4	29
52	Clinical and biochemical characterization of 3-hydroxyisobutyryl-CoA hydrolase (HIBCH) deficiency that causes Leigh-like disease and ketoacidosis. Molecular Genetics and Metabolism Reports, 2014, 1, 455-460.	1.1	29
53	A novel splice site mutation intron 1 of the GALNS gene in a Japanese patient with mucopolysaccharidosis IVA. Human Molecular Genetics, 1994, 3, 1427-1428.	2.9	28
54	Restoration of biochemical function of the peroxisome in the temperature-sensitive mild forms of peroxisome biogenesis disorder in humans. Brain and Development, 2000, 22, 8-12.	1.1	27

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55	Phenotypeâ€'genotype relationships in peroxisome biogenesis disorders of PEX1-defective complementation group 1 are defined by Pex1pâ€'Pex6p interaction. Biochemical Journal, 2001, 357, 417.	3.7	27
56	The common phospholipid-binding activity of the N-terminal domains of PEX1 and VCP/p97. FEBS Journal, 2006, 273, 4959-4971.	4.7	27
57	Adrenoleukodystrophy: subcellular localization and degradation of adrenoleukodystrophy protein (ALDP/ABCD1) with naturally occurring missense mutations. Journal of Neurochemistry, 2007, 101, 1632-1643.	3.9	27
58	Contiguous <scp>ABCD1 DXS1357E</scp> deletion syndrome: Report of an autopsy case. Neuropathology, 2013, 33, 292-298.	1.2	27
59	Effects of erucic acid therapy on Japanese patients with X-linked adrenoleukodystrophy. Brain and Development, 1994, 16, 454-458.	1.1	26
60	X-linked adrenoleukodystrophy: Diagnostic and follow-up system in Japan. Journal of Human Genetics, 2011, 56, 106-109.	2.3	26
61	Newly identified milder phenotype of peroxisome biogenesis disorder caused by mutated PEX3 gene. Brain and Development, 2013, 35, 842-848.	1.1	26
62	Hunter disease in a girl caused by R468Q mutation in the iduronate-2-sulfatase gene and skewed inactivation of the X chromosome carrying the normal allele. Human Mutation, 1997, 10, 361-367.	2.5	25
63	Detection of unusual very-long-chain fatty acid and ether lipid derivatives in the fibroblasts and plasma of patients with peroxisomal diseases using liquid chromatography-mass spectrometry. Molecular Genetics and Metabolism, 2017, 120, 255-268.	1.1	25
64	Newly Identified Chinese Hamster Ovary Cell Mutants Defective in Peroxisome Assembly Represent Complementation Group A of Human Peroxisome Biogenesis Disorders and One Novel Group in Mammals. Experimental Cell Research, 1999, 248, 482-488.	2.6	24
65	Urinary organic acids in peroxisomal disorders: a simple screening method. Biomedical Applications, 2001, 758, 81-86.	1.7	23
66	A Novel Aberrant Splicing Mutation of the PEX16 Gene in Two Patients with Zellweger Syndrome. Biochemical and Biophysical Research Communications, 2002, 292, 109-112.	2.1	23
67	Baicalein 5,6,7-trimethyl ether, a flavonoid derivative, stimulates fatty acid Î²-oxidation in skin fibroblasts of X-linked adrenoleukodystrophy. FEBS Letters, 2005, 579, 409-414.	2.8	23
68	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
69	Different Intracellular Localization of Peroxisomal Proteins in Fibroblasts from Patients with Aberrant Peroxisome Assembly.. Cell Structure and Function, 1992, 17, 1-8.	1.1	23
70	Peroxisome Biogenesis Disorders: Identification of a New Complementation Group Distinct from Peroxisome-Deficient CHO Mutants and Not Complemented by Human PEX 13. Biochemical and Biophysical Research Communications, 1998, 243, 368-371.	2.1	22
71	Genomic structure and identification of 11 novel mutations of thePEX6 (peroxisome assembly factor-2) gene in patients with peroxisome biogenesis disorders. Human Mutation, 1999, 13, 487-496.	2.5	22
72	Prevalence of patients with lysosomal storage disorders and peroxisomal disorders: A nationwide survey in Japan. Molecular Genetics and Metabolism, 2021, 133, 277-288.	1.1	22

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73	Aberrant peroxisome morphology in peroxisomal beta-oxidation enzyme deficiencies. Brain and Development, 2006, 28, 287-292.	1.1	21
74	Molecular Mechanism of Detectable Catalase-Containing Particles, Peroxisomes, in Fibroblasts from a PEX2-Defective Patient. Biochemical and Biophysical Research Communications, 2000, 268, 31-35.	2.1	20
75	Genetic heterogeneity of peroxisome biogenesis disorders among Japanese patients: Evidence for a founder haplotype for the most commonPEX10 gene mutation. American Journal of Medical Genetics Part A, 2003, 120A, 40-43.	2.4	20
76	Molecular Species of Phospholipids with Very Long Chain Fatty Acids in Skin Fibroblasts of Zellweger Syndrome. Lipids, 2013, 48, 1253-1267.	1.7	20
77	Profiling and Imaging of Phospholipids in Brains of Abcd1â€Deficient Mice. Lipids, 2018, 53, 85-102.	1.7	19
78	Immunoblot detection of enzyme proteins of peroxisomal Î²-oxidation in fibroblasts, amniocytes, and chorionic villous cells. Possible marker for prenatal diagnosis of Zellweger's syndrome. Prenatal Diagnosis, 1988, 8, 287-290.	2.3	18
79	Major Peroxisomal Membrane Polypeptides Are Synthesized in Cultured Skin Fibroblasts from Patients with Zellweger Syndrome. Pediatric Research, 1989, 26, 150-153.	2.3	18
80	A case of pseudo-Zellweger syndrome with a possible bifunctional enzyme deficiency but detectable enzyme protein. Brain and Development, 1993, 15, 453-456.	1.1	18
81	Assignment of the Human Peroxisome Assembly Factor-1 Gene (PXMP3) Responsible for Zellweger Syndrome to Chromosome 8q21.1 by Fluorescence in Situ Hybridization. Genomics, 1994, 20, 141-142.	2.9	18
82	Mucopolysaccharidosis IVA: polymorphic haplotypes and informative RFLPs in the Japanese population. Human Genetics, 1995, 95, 257-64.	3.8	18
83	Molecular genetic study in Japanese patients with Alexander disease: a novel mutation, R79L. Brain and Development, 2003, 25, 116-121.	1.1	18
84	Induction of peroxisomal lipid metabolism in mice fed a high-fat diet. Molecular Medicine Reports, 2011, 4, 1157-62.	2.4	18
85	Catalase-less Peroxisomes. Journal of Biological Chemistry, 2000, 275, 37271-37277.	3.4	16
86	The clinical course of childhood and adolescent adrenoleukodystrophy before and after Lorenzo's oil. Brain and Development, 2001, 23, 30-33.	1.1	16
87	Two new mutations, Q473X and N487S, in a caucasian patient with mucopolysaccharidosis IVA (Morquio disease). Human Mutation, 1995, 6, 195-196.	2.5	15
88	Peroxisomal localization in the developing mouse cerebellum: implications for neuronal abnormalities related to deficiencies in peroxisomes. Biochimica Et Biophysica Acta - General Subjects, 2004, 1671, 26-33.	2.4	15
89	Molecular Mechanism of a Temperature-Sensitive Phenotype in Peroxisomal Biogenesis Disorder. Pediatric Research, 2005, 58, 263-269.	2.3	15
90	Two novel missense mutations in the ATPâ€binding domain of the adrenoleukodystrophy gene: immunoblotting and immunocytological study of two patients. Clinical Genetics, 1997, 51, 322-325.	2.0	15

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91	Living-Donor Liver Transplantation From a Heterozygous Parent for Infantile Refsum Disease. Pediatrics, 2016, 137, e20153102-e20153102.	2.1	15
92	Positional determination of the carbonâ€“carbon double bonds in unsaturated fatty acids mediated by solvent plasmatization using LCâ€“MS. Scientific Reports, 2020, 10, 12988.	3.3	15
93	POLR3A variants in striatal involvement without diffuse hypomyelination. Brain and Development, 2020, 42, 363-368.	1.1	15
94	Polymerase chain reaction detection of two novel human N-acetylgalactosamine-6-sulfate sulfatase gene polymorphisms by single-strand conformation polymorphism analysis or by StyI and StuI cleavages. Human Genetics, 1995, 95, 243-4.	3.8	14
95	Very Long Chain Fatty Acid Analysis of Dried Blood Spots on Filter Paper to Screen for Adrenoleukodystrophy. Clinical Chemistry, 1997, 43, 2197-2198.	3.2	14
96	Mucopolysaccharidosis IVA: structural gene alterations identified by Southern blot analysis and identification of racial differences. Human Genetics, 1995, 95, 376-81.	3.8	13
97	A novel mutation, R125X in peroxisome assembly factor-1 responsible for zellweger syndrome. Human Mutation, 1998, 11, S134-S136.	2.5	13
98	Accumulation of glycolipids in mutant Chinese hamster ovary cells (Z65) with defective peroxisomal assembly and comparison of the metabolic rate of glycosphingolipids between Z65 cells and wild-type CHO-K1 cells. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 1999, 1438, 55-62.	2.4	13
99	Incidence of peroxisomal disorders in Japan. Japanese Journal of Human Genetics, 1996, 41, 167-175.	0.8	12
100	Magnetic resonance imaging of acute cerebellar ataxia: Report of a case with gadolinium enhancement and review of the literature. Pediatrics International, 1998, 40, 138-142.	0.5	12
101	Fifteen polymorphisms in the N-acetylgalactosamine-6-sulfate sulfatase (GALNS) gene: Diagnostic implications in morquio disease. Human Mutation, 1998, 11, S42-S46.	2.5	12
102	Ataxia Telangiectasia Associated with B-Cell Lymphoma: The Effect of a Half-Dose of the Drugs Administered According to the Acute Lymphoblastic Leukemia Standard Risk Protocol. Pediatric Hematology and Oncology, 1998, 15, 425-429.	0.8	12
103	Glucose metabolism evaluated by positron emission tomography in Lafora disease. Pediatrics International, 1999, 41, 689-692.	0.5	12
104	Temperature Sensitivity in Peroxisome Assembly Processes Characterizes Milder Forms of Peroxisome Biogenesis Disorders. Cell Biochemistry and Biophysics, 2000, 32, 165-170.	1.8	12
105	Very-Long-Chain Fatty Acid Metabolism in Adrenoleukodystrophy Protein-Deficient Mice. Cell Biochemistry and Biophysics, 2000, 32, 239-246.	1.8	12
106	Genetic and molecular bases of peroxisome biogenesis disorders. Genetics in Medicine, 2001, 3, 372-376.	2.4	12
107	Topical Review: Molecular and Neurologic Findings of Peroxisome Biogenesis Disorders. Journal of Child Neurology, 2005, 20, 326-329.	1.4	12
108	A Novel Double Mutation in the ABCD1 Gene in a Patient with X-linked Adrenoleukodystrophy: Analysis of the Stability and Function of the Mutant ABCD1 Protein. JIMD Reports, 2012, 10, 95-102.	1.5	12

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109	Successive MRI Findings of Reversible Cerebral White Matter Lesions in a Patient with Cystathionine <i>Î²</i>-Synthase Deficiency. Tohoku Journal of Experimental Medicine, 2015, 237, 323-327.	1.2	12
110	Ataxic form of autosomal recessive PEX10-related peroxisome biogenesis disorders with a novel compound heterozygous gene mutation and characteristic clinical phenotype. Journal of the Neurological Sciences, 2017, 375, 424-429.	0.6	12
111	Allogeneic stem cell transplantation with reduced intensity conditioning for patients with adrenoleukodystrophy. Molecular Genetics and Metabolism Reports, 2019, 18, 1-6.	1.1	12
112	Atypical PEX16 peroxisome biogenesis disorder with mild biochemical disruptions and long survival. Brain and Development, 2019, 41, 57-65.	1.1	11
113	Advanced Diagnostic System and Introduction of Newborn Screening of Adrenoleukodystrophy and Peroxisomal Disorders in Japan. International Journal of Neonatal Screening, 2021, 7, 58.	3.2	11
114	Chediakâ€Higashi syndrome with cerebellar cortical atrophy detected by MRI. Clinical Genetics, 1994, 46, 439-440.	2.0	10
115	Mild case of <scp>d</scp>â€bifunctional protein deficiency associated with novel gene mutations. Pediatrics International, 2012, 54, 303-304.	0.5	10
116	Prenatal diagnosis of Zellweger syndrome using DNA analysis. Prenatal Diagnosis, 1993, 13, 149-149.	2.3	9
117	Prenatal diagnosis of peroxisomal disorders Biochemical and immunocytochemical studies on peroxisomes in human amniocytes. Brain and Development, 1994, 16, 27-31.	1.1	9
118	Hexacosenoyl-CoA is the most abundant very long-chain acyl-CoA in ATP binding cassette transporter D1-deficient cells. Journal of Lipid Research, 2020, 61, 523-536.	4.2	9
119	Correction by Gene Expression of Biochemical Abnormalities in Fibroblasts from Zellweger Patients. Pediatric Research, 1996, 39, 812-815.	2.3	9
120	Proliferative responses towards native, heat-denatured and pepsin-treated ovalbumin by peripheral blood mononuclear cells from patients with hen's egg-sensitive atopic dermatitis. Biotherapy (Dordrecht, Netherlands), 1994, 8, 33-40.	0.7	8
121	PRENATAL DIAGNOSIS OF ADRENOLEUKODYSTROPHY BY MEANS OF MUTATION ANALYSIS. , 1996, 16, 259-261.		8
122	Prenatal diagnosis of peroxisomal d-3-hydroxyacyl-CoA dehydratase / d-3-hydroxyacyl-CoA dehydrogenase bifunctional protein deficiency. Journal of Human Genetics, 1999, 44, 143-147.	2.3	8
123	Parents of childhood X-linked adrenoleukodystrophy: High risk for depression and neurosis. Brain and Development, 2008, 30, 477-482.	1.1	8
124	D-bifunctional protein deficiency with fetal ascites, polyhydramnios, and contractures of hands and toes. Journal of Pediatrics, 2001, 139, 865-867.	1.8	7
125	Retinal Ganglion Cell Loss in X-linked Adrenoleukodystrophy with an<i>ABCD1</i>Mutation (Gly266Arg). Neuro-Ophthalmology, 2014, 38, 331-335.	1.0	7
126	Glycosphingolipids with Very Long-Chain Fatty Acids Accumulate in Fibroblasts from Adrenoleukodystrophy Patients. International Journal of Molecular Sciences, 2021, 22, 8645.	4.1	7

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127	Peroxisomal Disorders: Clinical Aspects. Annals of the New York Academy of Sciences, 1996, 804, 442-449.	3.8	6
128	Rapid isolation and characterization of CHO mutants deficient in peroxisome biogenesis using the peroxisomal forms of fluorescent proteins. Biochimica Et Biophysica Acta - Molecular Cell Research, 2000, 1496, 232-242.	4.1	6
129	Clinical aspects and adrenal functions in eleven Japanese children with X-linked adrenoleukodystrophy. Endocrine Journal, 2010, 57, 965-972.	1.6	6
130	Expanding the concept of peroxisomal diseases and efficient diagnostic system in Japan. Journal of Human Genetics, 2019, 64, 145-152.	2.3	6
131	Biallelic mutation of HSD17B4 induces middle ageâ€“onset spinocerebellar ataxia. Neurology: Genetics, 2020, 6, e396.	1.9	6
132	Zebrafish model of human Zellweger syndrome reveals organ-specific accumulation of distinct fatty acid species and widespread gene expression changes. Molecular Genetics and Metabolism, 2021, 133, 307-323.	1.1	6
133	Identification of Pex5pM, and Retarded Maturation of 3-Ketoacyl-CoA Thiolase and Acyl-CoA Oxidase in CHO Cells Expressing Mutant Pex5p Isoforms. Journal of Biochemistry, 2005, 138, 781-790.	1.7	5
134	Changes in the amounts of myelin lipids and molecular species of plasmalogen PE in the brain of an autopsy case with d-bifunctional protein deficiency. Neuroscience Letters, 2008, 442, 4-9.	2.1	5
135	First Japanese case of Zellweger syndrome with a mutation in PEX14. Pediatrics International, 2015, 57, 1189-1192.	0.5	5
136	Characteristics of Japanese patients with X-linked adrenoleukodystrophy and concerns of their families from the 1st registry system. Brain and Development, 2019, 41, 50-56.	1.1	5
137	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. Brain and Development, 2021, 43, 475-481.	1.1	5
138	Genetic Heterogeneity in Japanese Patients with Peroxisome Biogenesis Disorders and Evidence for a Founder Haplotype for the Most Common Mutation in PEX10 Gene. Advances in Experimental Medicine and Biology, 2003, 544, 71-71.	1.6	5
139	Temperature-Sensitive Phenotype of Chinese Hamster Ovary Cells Defective in PEX5 Gene. Biochemical and Biophysical Research Communications, 2001, 288, 321-327.	2.1	4
140	Stability of the ABCD1 Protein with a Missense Mutation: A Novel Approach to Finding Therapeutic Compounds for X-Linked Adrenoleukodystrophy. JIMD Reports, 2018, 44, 23-31.	1.5	4
141	Clinical evaluation of childhood cerebral adrenoleukodystrophy with balintâ€™s symptoms. Brain and Development, 2021, 43, 396-401.	1.1	4
142	Trial of docosahexaenoic acid supplementation on a Japanese patient with a peroxisome biogenesis defect. Pediatrics International, 1996, 38, 520-523.	0.5	3
143	Prenatal diagnosis of peroxisome biogenesis disorders by means of immunofluorescence staining of cultured chorionic villous cells. Clinical Genetics, 1999, 56, 467-468.	2.0	3
144	Marinescoâ€“SjÃ¶gren syndrome associated with acute mveloblastic leukemia. Clinical Genetics, 1997, 51, 278-280.	2.0	3

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145	Carrier identification of Xâ€linked adrenoleukodystrophy by measurement of very long chain fatty acids and lignoceric acid oxidation. Clinical Genetics, 1996, 50, 348-352.	2.0	3
146	Effect of Lorenzoâ€™s Oil on Hepatic Gene Expression and the Serum Fatty Acid Level in abcd1-Deficient Mice. JIMD Reports, 2017, 38, 67-74.	1.5	3
147	Low donor chimerism may be sufficient to prevent demyelination in adrenoleukodystrophy. JIMD Reports, 2022, 63, 19-24.	1.5	3
148	THEOPHYLLINE-ASSOCIATED STATUS EPILEPTICUS RESULTING IN QUADRIPLEGIA. Annals of Allergy, Asthma and Immunology, 1997, 78, 332.	1.0	2
149	A novel method for determining peroxisomal fatty acid Î²â€oxidation. Journal of Inherited Metabolic Disease, 2016, 39, 725-731.	3.6	2
150	Highly asymmetric and subacutely progressive motor weakness with unilateral T2-weighted high intensities along the pyramidal tract in the brainstem in adrenomyeloneuropathy. Journal of the Neurological Sciences, 2017, 381, 107-109.	0.6	2
151	Novel HSD17B4 Variants Cause Progressive Leukodystrophy in Childhood: Case Report and Literature Review. Child Neurology Open, 2021, 8, 2329048X2110486.	1.1	2
152	Topical Review: Molecular and Neurologic Findings of Peroxisome Biogenesis Disorders. Journal of Child Neurology, 2004, 19, 326-329.	1.4	1
153	A 29-year-old patient with adrenoleukodystrophy presenting with Addisonâ€™s disease. Endocrine Journal, 2020, 67, 655-658.	1.6	1
154	Peroxisomal Disorders. , 2019, , 107-136.		1
155	Diagnosis of Peroxisomal Disorders. , 2019, , 159-169.		1
156	Biochemical and Immunocytochemical Properties of Peroxisomes and Mitochondria in Bovine Chromaffin Cells.. Cell Structure and Function, 1997, 22, 615-619.	1.1	1
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