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	Low donor chimerism may be sufficient to prevent demyelination in adrenoleukodystrophy. JIMD Reports, 2022, 63, 19-24.	1.5	3
2	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. Brain and Development, 2021, 43, 475-481.	1.1	5
3	Clinical evaluation of childhood cerebral adrenoleukodystrophy with balintâ€™s symptoms. Brain and Development, 2021, 43, 396-401.	1.1	4
4	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
5	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
6	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
7	Glycosphingolipids with Very Long-Chain Fatty Acids Accumulate in Fibroblasts from Adrenoleukodystrophy Patients. International Journal of Molecular Sciences, 2021, 22, 8645.	4.1	7
8	Novel HSD17B4 Variants Cause Progressive Leukodystrophy in Childhood: Case Report and Literature Review. Child Neurology Open, 2021, 8, 2329048X2110486.	1.1	2
9	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
10	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
11	A 29-year-old patient with adrenoleukodystrophy presenting with Addisonâ€™s disease. Endocrine Journal, 2020, 67, 655-658.	1.6	1
12	POLR3A variants in striatal involvement without diffuse hypomyelination. Brain and Development, 2020, 42, 363-368.	1.1	15
13	Biallelic mutation of HSD17B4 induces middle ageâ€“onset spinocerebellar ataxia. Neurology: Genetics, 2020, 6, e396.	1.9	6
14	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
15	Atypical PEX16 peroxisome biogenesis disorder with mild biochemical disruptions and long survival. Brain and Development, 2019, 41, 57-65.	1.1	11
16	Infantile Refsum Disease Associated with Hypobetalipoproteinemia. Journal of Pediatric Neurology, 2019, 17, 210-212.	0.2	0
17	Expanding the concept of peroxisomal diseases and efficient diagnostic system in Japan. Journal of Human Genetics, 2019, 64, 145-152.	2.3	6
18	Allogeneic stem cell transplantation with reduced intensity conditioning for patients with adrenoleukodystrophy. Molecular Genetics and Metabolism Reports, 2019, 18, 1-6.	1.1	12

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19	Peroxisomal Disorders. , 2019, , 107-136.		1
20	Diagnosis of Peroxisomal Disorders. , 2019, , 159-169.		1
21	Model Organisms for Understanding Peroxisomal Disorders. , 2019, , 137-157.		0
22	Profiling and Imaging of Phospholipids in Brains of Abcd1â€Deficient Mice. Lipids, 2018, 53, 85-102.	1.7	19
23	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
24	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
25	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
26	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
27	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
28	A first case of adrenomyeloneuropathy with mutation Y174S of the adrenoleukodystrophy gene. Neuroendocrinology Letters, 2017, 38, 13-18.	0.2	0
29	Serial Monitoring of Plasma Levetiracetam Levels in a Child With Epilepsy Undergoing Cord Blood Transplantation. Pediatric Neurology, 2016, 64, e5-e6.	2.1	0
30	A novel method for determining peroxisomal fatty acid Î²â€oxidation. Journal of Inherited Metabolic Disease, 2016, 39, 725-731.	3.6	2
31	Living-Donor Liver Transplantation From a Heterozygous Parent for Infantile Refsum Disease. Pediatrics, 2016, 137, e20153102-e20153102.	2.1	15
32	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
33	First Japanese case of Zellweger syndrome with a mutation in PEX14. Pediatrics International, 2015, 57, 1189-1192.	0.5	5
34	Retinal Ganglion Cell Loss in X-linked Adrenoleukodystrophy with anABCD1Mutation (Gly266Arg). Neuro-Ophthalmology, 2014, 38, 331-335.	1.0	7
35	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
36	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan. Neurology, 2014, 82, 705-712.	1.1	71

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37	Evaluation of Fourier Transform Infrared Spectroscopy for Diagnosis of Peroxisomal Diseases with Abnormal Very-Long-Chain Fatty Acid Metabolism. American Journal of Analytical Chemistry, 2014, 05, 359-366.	0.9	1
38	Newly identified milder phenotype of peroxisome biogenesis disorder caused by mutated PEX3 gene. Brain and Development, 2013, 35, 842-848.	1.1	26
39	Molecular Species of Phospholipids with Very Long Chain Fatty Acids in Skin Fibroblasts of Zellweger Syndrome. Lipids, 2013, 48, 1253-1267.	1.7	20
40	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. Neurogenetics, 2013, 14, 225-232.	1.4	104
41	Contiguous <scp>ABCD1 DXS1357E</scp> deletion syndrome: Report of an autopsy case. Neuropathology, 2013, 33, 292-298.	1.2	27
42	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
43	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
44	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
45	Mild case of <scp>d</scp>â€bifunctional protein deficiency associated with novel gene mutations. Pediatrics International, 2012, 54, 303-304.	0.5	10
46	X-linked adrenoleukodystrophy: Diagnostic and follow-up system in Japan. Journal of Human Genetics, 2011, 56, 106-109.	2.3	26
47	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
48	Induction of peroxisomal lipid metabolism in mice fed a high-fat diet. Molecular Medicine Reports, 2011, 4, 1157-62.	2.4	18
49	Molecular and clinical findings and diagnostic flowchart of peroxisomal diseases. Brain and Development, 2011, 33, 770-776.	1.1	31
50	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
51	Clinical aspects and adrenal functions in eleven Japanese children with X-linked adrenoleukodystrophy. Endocrine Journal, 2010, 57, 965-972.	1.6	6
52	Parents of childhood X-linked adrenoleukodystrophy: High risk for depression and neurosis. Brain and Development, 2008, 30, 477-482.	1.1	8
53	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
54	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

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55	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
56	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
57	The common phospholipid-binding activity of the N-terminal domains of PEX1 and VCP/p97. FEBS Journal, 2006, 273, 4959-4971.	4.7	27
58	Aberrant peroxisome morphology in peroxisomal beta-oxidation enzyme deficiencies. Brain and Development, 2006, 28, 287-292.	1.1	21
59	Role of Pex19p in the targeting of PMP70 to peroxisome. Biochimica Et Biophysica Acta - Molecular Cell Research, 2005, 1746, 116-128.	4.1	47
60	Natural history of X-linked adrenoleukodystrophy in Japan. Brain and Development, 2005, 27, 353-357.	1.1	50
61	Mutational and structural analysis of Japanese patients with mucopolysaccharidosis type II. Journal of Human Genetics, 2005, 50, 395-402.	2.3	43
62	Topical Review: Molecular and Neurologic Findings of Peroxisome Biogenesis Disorders. Journal of Child Neurology, 2005, 20, 326-329.	1.4	12
63	Molecular Mechanism of a Temperature-Sensitive Phenotype in Peroxisomal Biogenesis Disorder. Pediatric Research, 2005, 58, 263-269.	2.3	15
64	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
65	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
66	Topical Review: Molecular and Neurologic Findings of Peroxisome Biogenesis Disorders. Journal of Child Neurology, 2004, 19, 326-329.	1.4	1
67	Proteomic Analysis of Rat Liver Peroxisome. Journal of Biological Chemistry, 2004, 279, 421-428.	3.4	243
68	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
69	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
70	Molecular genetic study in Japanese patients with Alexander disease: a novel mutation, R79L. Brain and Development, 2003, 25, 116-121.	1.1	18
71	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
72	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

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73	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
74	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
75	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
76	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
77	Peroxisomal acyl CoA oxidase deficiency. Journal of Pediatrics, 2002, 140, 128-130.	1.8	33
78	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
79	Epidemiology of X-linked adrenoleukodystrophy in Japan. Journal of Human Genetics, 2002, 47, 0590-0593.	2.3	45
80	Temperature-Sensitive Phenotype of Chinese Hamster Ovary Cells Defective in PEX5 Gene. Biochemical and Biophysical Research Communications, 2001, 288, 321-327.	2.1	4
81	D-bifunctional protein deficiency with fetal ascites, polyhydramnios, and contractures of hands and toes. Journal of Pediatrics, 2001, 139, 865-867.	1.8	7
82	The clinical course of childhood and adolescent adrenoleukodystrophy before and after Lorenzo's oil. Brain and Development, 2001, 23, 30-33.	1.1	16
83	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
84	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
85	Urinary organic acids in peroxisomal disorders: a simple screening method. Biomedical Applications, 2001, 758, 81-86.	1.7	23
86	Genetic and molecular bases of peroxisome biogenesis disorders. Genetics in Medicine, 2001, 3, 372-376.	2.4	12
87	Rapid diagnosis of peroxisome biogenesis disorders through immunofluorescence staining of buccal smears. Annals of Neurology, 2000, 47, 836-837.	5.3	0
88	Temperature Sensitivity in Peroxisome Assembly Processes Characterizes Milder Forms of Peroxisome Biogenesis Disorders. Cell Biochemistry and Biophysics, 2000, 32, 165-170.	1.8	12
89	Very-Long-Chain Fatty Acid Metabolism in Adrenoleukodystrophy Protein-Deficient Mice. Cell Biochemistry and Biophysics, 2000, 32, 239-246.	1.8	12
90	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

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91	Catalase-less Peroxisomes. Journal of Biological Chemistry, 2000, 275, 37271-37277.	3.4	16
92	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
93	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
94	Congenital myotonic dystrophy: report of paternal transmission. Brain and Development, 2000, 22, 132-134.	1.1	34
95	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
96	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
97	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
98	Glucose metabolism evaluated by positron emission tomography in Lafora disease. Pediatrics International, 1999, 41, 689-692.	0.5	12
99	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
100	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
101	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
102	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
103	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
104	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
105	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
106	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
107	A novel mutation, R125X in peroxisome assembly factor-1 responsible for zellweger syndrome. Human Mutation, 1998, 11, S134-S136.	2.5	13
108	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

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109	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
110	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
111	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
112	A Cytoplasmic AAA Family Peroxin, Pex1p, Interacts with Pex6p. Biochemical and Biophysical Research Communications, 1998, 245, 883-886.	2.1	67
113	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
114	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
115	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
116	THEOPHYLLINE-ASSOCIATED STATUS EPILEPTICUS RESULTING IN QUADRIPLEGIA. Annals of Allergy, Asthma and Immunology, 1997, 78, 332.	1.0	2
117	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
118	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
119	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
120	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
121	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
122	Epilepsy in Peroxisomal Diseases. Epilepsia, 1997, 38, 182-188.	5.1	41
123	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
124	Fourteen novel mucopolysaccharidosis IVA producing mutations in GALNS gene. Human Mutation, 1997, 10, 368-375.	2.5	37
125	Marinescoâ€šjÃ¶gren syndrome associated with acute mveloblastic leukemia. Clinical Genetics, 1997, 51, 278-280.	2.0	3
126	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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127	Biochemical and Immunocytochemical Properties of Peroxisomes and Mitochondria in Bovine Chromaffin Cells.. Cell Structure and Function, 1997, 22, 615-619.	1.1	1
128	Incidence of peroxisomal disorders in Japan. Japanese Journal of Human Genetics, 1996, 41, 167-175.	0.8	12
129	Trial of docosahexaenoic acid supplementation on a Japanese patient with a peroxisome biogenesis defect. Pediatrics International, 1996, 38, 520-523.	0.5	3
130	PRENATAL DIAGNOSIS OF ADRENOLEUKODYSTROPHY BY MEANS OF MUTATION ANALYSIS. , 1996, 16, 259-261.		8
131	Mucopolysaccharidosis type I: Identification of common mutations that cause Hurler and Scheie syndromes in Japanese populations. , 1996, 7, 23-29.		43
132	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
133	Peroxisomal Disorders: Clinical Aspects. Annals of the New York Academy of Sciences, 1996, 804, 442-449.	3.8	6
134	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
135	Correction by Gene Expression of Biochemical Abnormalities in Fibroblasts from Zellweger Patients. Pediatric Research, 1996, 39, 812-815.	2.3	9
136	Inborn Errors of Peroxisome Biogenesis and Brain Malformation: Clinical and Biochemical Studies. Congenital Anomalies (discontinued), 1995, 35, 43-53.	0.6	0
137	Two new mutations, Q473X and N487S, in a caucasian patient with mucopolysaccharidosis IVA (Morquio disease). Human Mutation, 1995, 6, 195-196.	2.5	15
138	Mucopolysaccharidosis IVA: structural gene alterations identified by Southern blot analysis and identification of racial differences. Human Genetics, 1995, 95, 376-81.	3.8	13
139	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
140	Mucopolysaccharidosis IVA: polymorphic haplotypes and informative RFLPs in the Japanese population. Human Genetics, 1995, 95, 257-64.	3.8	18
141	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
142	Mucopolysaccharidosis type IVA: identification of six novel mutations among non-Japanese patients. Human Molecular Genetics, 1995, 4, 741-743.	2.9	31
143	Mucopolysaccharidosis type IVA: common double deletion in the N-acetylgalactosamine-6-sulfatase gene (GALNS). Genomics, 1995, 26, 535-542.	2.9	45
144	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

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145	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
146	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
147	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
148	Effects of erucic acid therapy on Japanese patients with X-linked adrenoleukodystrophy. Brain and Development, 1994, 16, 454-458.	1.1	26
149	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
150	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
151	Prenatal diagnosis of peroxisomal disorders Biochemical and immunocytochemical studies on peroxisomes in human amniocytes. Brain and Development, 1994, 16, 27-31.	1.1	9
152	Chediakâ€Higashi syndrome with cerebellar cortical atrophy detected by MRI. Clinical Genetics, 1994, 46, 439-440.	2.0	10
153	Prenatal diagnosis of Zellweger syndrome using DNA analysis. Prenatal Diagnosis, 1993, 13, 149-149.	2.3	9
154	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
155	A human gene responsible for Zellweger syndrome that affects peroxisome assembly. Science, 1992, 255, 1132-1134.	12.6	363
156	Complementation study of peroxisome-deficient disorders by immunofluorescence staining and characterization of fused cells. Human Genetics, 1992, 88, 491-499.	3.8	100
157	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
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#	Article	IF	Citations
163	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