

# Jutta GÄrtner

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9148439/publications.pdf>

Version: 2024-02-01

132  
papers

6,672  
citations

71102

41  
h-index

74163

75  
g-index

137  
all docs

137  
docs citations

137  
times ranked

11192  
citing authors

#	ARTICLE	IF	CITATIONS
1	Deep breathing couples CSF and venous flow dynamics. <i>Scientific Reports</i> , 2022, 12, 2568.	3.3	22
2	Cln5 represents a new type of cysteine-based <i>S</i> -depalmitoylase linked to neurodegeneration. <i>Science Advances</i> , 2022, 8, eabj8633.	10.3	12
3	Improving pediatric multiple sclerosis interventional phase III study design: a meta-analysis. <i>Therapeutic Advances in Neurological Disorders</i> , 2022, 15, 175628642110704.	3.5	0
4	<i>TTC5</i> syndrome: Clinical and molecular spectrum of a severe and recognizable condition. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2652-2665.	1.2	4
5	Temporal profile of lymphocyte counts and relationship with infections with fingolimod therapy in paediatric patients with multiple sclerosis: Results from the PARADIGMS study. <i>Multiple Sclerosis Journal</i> , 2021, 27, 922-932.	3.0	5
6	Tumefactive inflammatory lesions in juvenile metachromatic leukodystrophy. <i>Neurology: Neuroimmunology and Neuroinflammation</i> , 2021, 8, .	6.0	4
7	Follow-Up of a Case of Dopamine-Mediated Yawning-Fatigue-Syndrome Responsive to Opioids, Successful Desensitization via Graded Activity Treatment. <i>Neurology International</i> , 2021, 13, 79-84.	2.8	0
8	A novel remitting leukodystrophy associated with a variant in FBP2. <i>Brain Communications</i> , 2021, 3, fcab036.	3.3	2
9	Biallelic VPS16 variants limit HOPS/CORVET levels and cause a mucopolysaccharidosis-like disease. <i>EMBO Molecular Medicine</i> , 2021, 13, e13376.	6.9	16
10	Targeted metabolomics revealed changes in phospholipids during the development of neuroinflammation in <i>Abcd1</i> <sup>tm1Kds</sup> mice and <i>X</i> -linked adrenoleukodystrophy patients. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1174-1185.	3.6	7
11	Hydrocephalus Revisited: New Insights into Dynamics of Neurofluids on Macro- and Microscales. <i>Neuropediatrics</i> , 2021, 52, 233-241.	0.6	4
12	Concurrent axon and myelin destruction differentiates <i>X</i> -linked adrenoleukodystrophy from multiple sclerosis. <i>Glia</i> , 2021, 69, 2362-2377.	4.9	7
13	Interferon-driven brain phenotype in a mouse model of RNaseT2 deficient leukoencephalopathy. <i>Nature Communications</i> , 2021, 12, 6530.	12.8	16
14	Dopamine-Mediated Yawning-Fatigue Syndrome With Specific Recurrent Initiation and Responsiveness to Opioids. <i>JAMA Neurology</i> , 2020, 77, 254.	9.0	1
15	Inborn errors of metabolism leading to neuronal migration defects. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 145-155.	3.6	16
16	Comparative analysis of alternating hemiplegia of childhood and rapid-onset dystonia-parkinsonism ATP1A3 mutations reveals functional deficits, which do not correlate with disease severity. <i>Neurobiology of Disease</i> , 2020, 143, 105012.	4.4	8
17	Natural history of multiple sulfatase deficiency: Retrospective phenotyping and functional variant analysis to characterize an ultra-rare disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1298-1309.	3.6	23
18	Mannose phosphate isomerase deficiency—a congenital disorder of glycosylation (MPI-CDG) with cerebral venous sinus thrombosis as first and only presenting symptom: A rare but treatable cause of thrombophilia. <i>JIMD Reports</i> , 2020, 55, 38-43.	1.5	8

#	ARTICLE	IF	CITATIONS
19	The failure of microglia to digest developmental apoptotic cells contributes to the pathology of RNASET2-deficient leukoencephalopathy. <i>Glia</i> , 2020, 68, 1531-1545.	4.9	35
20	A systematic review and meta-analysis of published cases reveals the natural disease history in multiple sulfatase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1288-1297.	3.6	14
21	Immune Sensing of Synthetic, Bacterial, and Protozoan RNA by Toll-like Receptor 8 Requires Coordinated Processing by RNase T2 and RNase 2. <i>Immunity</i> , 2020, 52, 591-605.e6.	14.3	83
22	B cell depletion can be effective in multiple sclerosis but failed in a patient with advanced childhood cerebral X-linked adrenoleukodystrophy. <i>Therapeutic Advances in Neurological Disorders</i> , 2019, 12, 175628641986813.	3.5	1
23	Severe neonatal multiple sulfatase deficiency presenting with hydrops fetalis in a preterm birth patient. <i>JIMD Reports</i> , 2019, 49, 48-52.	1.5	7
24	Opening New Horizons in the Treatment of Childhood Onset Leukodystrophies. <i>Neuropediatrics</i> , 2019, 50, 211-218.	0.6	6
25	Ketogenic diet ameliorates axonal defects and promotes myelination in Pelizaeus-Merzbacher disease. <i>Acta Neuropathologica</i> , 2019, 138, 147-161.	7.7	48
26	Upward movement of cerebrospinal fluid in obstructive hydrocephalus—revision of an old concept. <i>Child's Nervous System</i> , 2019, 35, 833-841.	1.1	8
27	Breathing drives CSF: Impact on spaceflight disease and hydrocephalus. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 20263-20264.	7.1	7
28	Therapy of highly active pediatric multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2019, 25, 72-80.	3.0	60
29	Genetics of intellectual disability in consanguineous families. <i>Molecular Psychiatry</i> , 2019, 24, 1027-1039.	7.9	131
30	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2018, 103, 592-601.	6.2	41
31	Trial of Fingolimod versus Interferon Beta-1a in Pediatric Multiple Sclerosis. <i>New England Journal of Medicine</i> , 2018, 379, 1017-1027.	27.0	237
32	Molecularly defined diffuse leptomeningeal glioneuronal tumor (DLGNT) comprises two subgroups with distinct clinical and genetic features. <i>Acta Neuropathologica</i> , 2018, 136, 239-253.	7.7	118
33	Potential Risks to Stable Long-term Outcome of Allogeneic Hematopoietic Stem Cell Transplantation for Children With Cerebral X-linked Adrenoleukodystrophy. <i>JAMA Network Open</i> , 2018, 1, e180769.	5.9	30
34	Respiration and the watershed of spinal CSF flow in humans. <i>Scientific Reports</i> , 2018, 8, 5594.	3.3	53
35	Cognitive deficits including executive functioning in relation to clinical parameters in paediatric MS patients. <i>PLoS ONE</i> , 2018, 13, e0194873.	2.5	18
36	Dysferlin mediates membrane tubulation and links T-tubule biogenesis to muscular dystrophy. <i>Journal of Cell Science</i> , 2017, 130, 841-852.	2.0	34

#	ARTICLE	IF	CITATIONS
37	Identification of the Upward Movement of Human CSF <i>In Vivo</i> and its Relation to the Brain Venous System. <i>Journal of Neuroscience</i> , 2017, 37, 2395-2402.	3.6	133
38	The functional readthrough extension of malate dehydrogenase reveals a modification of the genetic code. <i>Open Biology</i> , 2016, 6, 160246.	3.6	41
39	Compound heterozygous variants in PGAP1 causing severe psychomotor retardation, brain atrophy, recurrent apneas and delayed myelination: a case report and literature review. <i>BMC Neurology</i> , 2016, 16, 74.	1.8	19
40	STAR syndrome-associated CDK10/Cyclin M regulates actin network architecture and ciliogenesis. <i>Cell Cycle</i> , 2016, 15, 678-688.	2.6	33
41	Leukoencephalopathy and early death associated with an Ashkenazi-Jewish founder mutation in the Hikeshe gene. <i>Journal of Medical Genetics</i> , 2016, 53, 132-137.	3.2	21
42	Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. <i>Nature</i> , 2015, 521, E1-E4.	27.8	159
43	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. <i>Mitochondrion</i> , 2015, 21, 12-18.	3.4	51
44	JC virus antibody status in a pediatric multiple sclerosis cohort: Prevalence, conversion rate and influence on disease severity. <i>Multiple Sclerosis Journal</i> , 2015, 21, 382-387.	3.0	21
45	Extensive acute axonal damage in pediatric multiple sclerosis lesions. <i>Annals of Neurology</i> , 2015, 77, 655-667.	5.3	69
46	Inspiration Is the Major Regulator of Human CSF Flow. <i>Journal of Neuroscience</i> , 2015, 35, 2485-2491.	3.6	261
47	Tectonic gene mutations in patients with Joubert syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 616-620.	2.8	40
48	Clinical utility gene card for: Zellweger syndrome spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1111-1111.	2.8	6
49	MRI-based diagnostic biomarkers for early onset pediatric multiple sclerosis. <i>NeuroImage: Clinical</i> , 2015, 7, 400-408.	2.7	9
50	Characterization of the MeCP2R168X Knockin Mouse Model for Rett Syndrome. <i>PLoS ONE</i> , 2014, 9, e115444.	2.5	32
51	A novel ATP1A3 mutation with unique clinical presentation. <i>Journal of the Neurological Sciences</i> , 2014, 341, 133-135.	0.6	24
52	Phenotypic overlap of alternating hemiplegia of childhood and CAPOS syndrome. <i>Neurology</i> , 2014, 83, 861-863.	1.1	42
53	Peroxisomes are juxtaposed to strategic sites on mitochondria. <i>Molecular BioSystems</i> , 2014, 10, 1742-1748.	2.9	95
54	Structure of sulfamidase provides insight into the molecular pathology of mucopolysaccharidosis IIIA. <i>Acta Crystallographica Section D: Biological Crystallography</i> , 2014, 70, 1321-1335.	2.5	29

#	ARTICLE	IF	CITATIONS
55	Peroxisomal lactate dehydrogenase is generated by translational readthrough in mammals. <i>ELife</i> , 2014, 3, e03640.	6.0	155
56	Cerebellar ataxia, mental retardation and dysequilibrium syndrome 1 (CAMRQ1) caused by an unusual constellation of VLDLR mutation. <i>Journal of Neurology</i> , 2013, 260, 1678-1680.	3.6	15
57	Pediatric onset multiple sclerosis: McDonald criteria 2010 and the contribution of spinal cord MRI. <i>Multiple Sclerosis Journal</i> , 2013, 19, 1330-1335.	3.0	33
58	West syndrome, microcephaly, grey matter heterotopia and hypoplasia of corpus callosum due to a novel ARFGEF2 mutation. <i>Journal of Medical Genetics</i> , 2013, 50, 772-775.	3.2	24
59	Acute disseminated encephalomyelitis followed by recurrent or monophasic optic neuritis in pediatric patients. <i>Multiple Sclerosis Journal</i> , 2013, 19, 941-946.	3.0	135
60	Microduplication of 3p26.3 in Nonsyndromic Intellectual Disability Indicates an Important Role of CHL1 for Normal Cognitive Function. <i>Neuropediatrics</i> , 2013, 44, 268-271.	0.6	37
61	Rapid degradation of an active formylglycine generating enzyme variant leads to a late infantile severe form of multiple sulfatase deficiency. <i>European Journal of Human Genetics</i> , 2013, 21, 1020-1023.	2.8	19
62	Functional analysis of PEX13 mutation in a Zellweger syndrome spectrum patient reveals novel homooligomerization of PEX13 and its role in human peroxisome biogenesis. <i>Human Molecular Genetics</i> , 2013, 22, 3844-3857.	2.9	26
63	Therapeutic Apheresis in Pediatric Patients with Acute CNS Inflammatory Demyelinating Disease. <i>Blood Purification</i> , 2013, 36, 92-97.	1.8	35
64	First PEX11 <sup>Δ2</sup> patient extends spectrum of peroxisomal biogenesis disorder phenotypes: Table 1. <i>Journal of Medical Genetics</i> , 2012, 49, 314-316.	3.2	33
65	Structure and activity of the only human RNase T2. <i>Nucleic Acids Research</i> , 2012, 40, 8733-8742.	14.5	37
66	Anti-Myelin Oligodendrocyte Glycoprotein Antibodies in Pediatric Patients With Optic Neuritis. <i>Archives of Neurology</i> , 2012, 69, 752-6.	4.5	181
67	Identification of a New Fatty Acid Synthesis-Transport Machinery at the Peroxisomal Membrane*. <i>Journal of Biological Chemistry</i> , 2012, 287, 210-221.	3.4	31
68	Frequent but nonspecific venous narrowing in paediatric multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2012, 18, 1805-1805.	3.0	1
69	Assessment of myelination in hypomyelinating disorders by quantitative MRI. <i>Journal of Magnetic Resonance Imaging</i> , 2012, 36, 1329-1338.	3.4	21
70	Heterozygous de-novo mutations in ATP1A3 in patients with alternating hemiplegia of childhood: a whole-exome sequencing gene-identification study. <i>Lancet Neurology</i> , The, 2012, 11, 764-773.	10.2	223
71	Molecular and biochemical characterization of a unique mutation in CCS, the human copper chaperone to superoxide dismutase. <i>Human Mutation</i> , 2012, 33, 1207-1215.	2.5	34
72	Mutations in SLC33A1 Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. <i>American Journal of Human Genetics</i> , 2012, 90, 61-68.	6.2	85

#	ARTICLE	IF	CITATIONS
73	Peroxisome Formation Requires the Endoplasmic Reticulum Channel Protein <i>scp1</i> . <i>Traffic</i> , 2012, 13, 599-609.	2.7	42
74	MicroRNA regulation in experimental autoimmune encephalomyelitis in mice and marmosets resembles regulation in human multiple sclerosis lesions. <i>Journal of Neuroimmunology</i> , 2012, 246, 27-33.	2.3	47
75	SUMF1 mutations affecting stability and activity of formylglycine generating enzyme predict clinical outcome in multiple sulfatase deficiency. <i>European Journal of Human Genetics</i> , 2011, 19, 253-261.	2.8	63
76	Readthrough of nonsense mutations in Rett syndrome: evaluation of novel aminoglycosides and generation of a new mouse model. <i>Journal of Molecular Medicine</i> , 2011, 89, 389-398.	3.9	90
77	<i>rnalet2</i> mutant zebrafish model familial cystic leukoencephalopathy and reveal a role for RNase T2 in degrading ribosomal RNA. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 1099-1103.	7.1	91
78	Polymicrogyria in fetal alcohol syndrome. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2010, 88, 128-131.	1.6	16
79	Leukodystrophies and other genetic metabolic leukoencephalopathies in children and adults. <i>Brain and Development</i> , 2010, 32, 82-89.	1.1	26
80	Pelizaeus-Merzbacher-like disease is caused not only by a loss of connexin47 function but also by a hemichannel dysfunction. <i>European Journal of Human Genetics</i> , 2010, 18, 985-992.	2.8	27
81	Structure of Tripeptidyl-peptidase I Provides Insight into the Molecular Basis of Late Infantile Neuronal Ceroid Lipofuscinosis. <i>Journal of Biological Chemistry</i> , 2009, 284, 3976-3984.	3.4	43
82	Serial proton MR spectroscopy and diffusion tensor imaging in infantile Balo's concentric sclerosis. <i>Neuroradiology</i> , 2009, 51, 113-121.	2.2	19
83	Rational diagnostic strategy for Zellweger syndrome spectrum patients. <i>European Journal of Human Genetics</i> , 2009, 17, 741-748.	2.8	33
84	RNASET2-deficient cystic leukoencephalopathy resembles congenital cytomegalovirus brain infection. <i>Nature Genetics</i> , 2009, 41, 773-775.	21.4	124
85	Folate Receptor Alpha Defect Causes Cerebral Folate Transport Deficiency: A Treatable Neurodegenerative Disorder Associated with Disturbed Myelin Metabolism. <i>American Journal of Human Genetics</i> , 2009, 85, 354-363.	6.2	228
86	Immunoglobulin Therapy in Idiopathic Hypothalamic Dysfunction. <i>Pediatric Neurology</i> , 2009, 41, 232-234.	2.1	20
87	Suppression of Nonsense Mutations in Rett Syndrome by Aminoglycoside Antibiotics. <i>Pediatric Research</i> , 2009, 65, 520-523.	2.3	36
88	Cerebral involvement in axonal Charcot-Marie-Tooth neuropathy caused by mitofusin2 mutations. <i>Journal of Neurology</i> , 2008, 255, 1049-58.	3.6	66
89	Molecular analysis of SUMF1 mutations: stability and residual activity of mutant formylglycine-generating enzyme determine disease severity in multiple sulfatase deficiency. <i>Human Mutation</i> , 2008, 29, 205-205.	2.5	74
90	Axonal neuropathy with unusual pattern of amyotrophy and alacrima associated with a novel AAAS mutation p.Leu430Phe. <i>European Journal of Human Genetics</i> , 2008, 16, 1499-1506.	2.8	26

#	ARTICLE	IF	CITATIONS
91	Spontaneous central apneas occur in the C57BL/6J mouse strain. <i>Respiratory Physiology and Neurobiology</i> , 2008, 160, 21-27.	1.6	49
92	Natalizumab Use in Pediatric Multiple Sclerosis. <i>Archives of Neurology</i> , 2008, 65, 1655-8.	4.5	86
93	Early Reduction of Total N-Acetyl-Aspartate-Compounds in Patients With Classical Vanishing White Matter Disease. A Long-Term Follow-Up MRS Study. <i>Pediatric Research</i> , 2008, 63, 444-449.	2.3	12
94	Common infectious agents in multiple sclerosis: a case-control study in children. <i>Multiple Sclerosis Journal</i> , 2008, 14, 136-139.	3.0	44
95	Live Cell FRET Microscopy. <i>Journal of Biological Chemistry</i> , 2007, 282, 26997-27005.	3.4	50
96	Genotype-phenotype analysis in patients with giant axonal neuropathy (GAN). <i>Neuromuscular Disorders</i> , 2007, 17, 624-630.	0.6	29
97	Genotype and Protein Expression After Bone Marrow Transplantation for Adrenoleukodystrophy. <i>Archives of Neurology</i> , 2007, 64, 651.	4.5	18
98	Breathing dysfunctions associated with impaired control of postinspiratory activity in <i>Mecp2</i> <sup>fl/yk</sup> knockout mice. <i>Journal of Physiology</i> , 2007, 579, 863-876.	2.9	143
99	Cathepsin D Deficiency Is Associated with a Human Neurodegenerative Disorder. <i>American Journal of Human Genetics</i> , 2006, 78, 988-998.	6.2	255
100	Pediatric multiple sclerosis: Detection of clinically silent lesions by multimodal evoked potentials. <i>Journal of Pediatrics</i> , 2006, 149, 125-127.	1.8	49
101	Cerebral metabolic and structural alterations in hereditary spastic paraplegia with thin corpus callosum assessed by MRS and DTI. <i>Neuroradiology</i> , 2006, 48, 893-898.	2.2	35
102	The cystathionine beta-synthase variant c.844_845ins68 protects against CNS demyelination in X-linked adrenoleukodystrophy. <i>Human Mutation</i> , 2006, 27, 1063-1064.	2.5	18
103	Identification of novel mutations in PEX2, PEX6, PEX10, PEX12, and PEX13 in Zellweger spectrum patients. <i>Human Mutation</i> , 2006, 27, 1157-1157.	2.5	49
104	Clinical and biochemical spectrum of D-bifunctional protein deficiency. <i>Annals of Neurology</i> , 2006, 59, 92-104.	5.3	175
105	Detection of Tripeptidyl Peptidase I Activity in Living Cells by Fluorogenic Substrates. <i>Journal of Histochemistry and Cytochemistry</i> , 2006, 54, 991-996.	2.5	6
106	Visually Self-induced Seizures Sensitive to Round Objects. <i>Epilepsia</i> , 2005, 46, 786-789.	5.1	3
107	Colorectal cancer in two pre-teenage siblings with familial adenomatous polyposis. <i>European Journal of Pediatrics</i> , 2005, 164, 306-310.	2.7	31
108	Tau, Phospho-Tau, and S-100B in the Cerebrospinal Fluid of Children With Multiple Sclerosis. <i>Journal of Child Neurology</i> , 2005, 20, 822-825.	1.4	37

#	ARTICLE	IF	CITATIONS
109	Mutations in classical late infantile neuronal ceroid lipofuscinosis disrupt transport of tripeptidyl-peptidase I to lysosomes. <i>Human Molecular Genetics</i> , 2004, 13, 2483-2491.	2.9	25
110	Mutations in the Gene Encoding Gap Junction Protein $\beta$ 12 (Connexin 46.6) Cause Pelizaeus-Merzbacher- $\beta$ -Like Disease. <i>American Journal of Human Genetics</i> , 2004, 75, 251-260.	6.2	257
111	Sequence diversity of KIAA0027/MLC1: are megalencephalic leukoencephalopathy and schizophrenia allelic disorders?. <i>Human Mutation</i> , 2003, 21, 45-52.	2.5	22
112	The peroxisomal membrane targeting elements of human peroxin 2 (PEX2). <i>European Journal of Cell Biology</i> , 2003, 82, 155-162.	3.6	14
113	Two Different Targeting Signals Direct Human Peroxisomal Membrane Protein 22 to Peroxisomes. <i>Journal of Biological Chemistry</i> , 2002, 277, 774-784.	3.4	57
114	PEX1 Mutations in Complementation Group 1 of Zellweger Spectrum Patients Correlate with Severity of Disease. <i>Pediatric Research</i> , 2002, 51, 706-714.	2.3	58
115	Targeting Elements in the Amino-Terminal Part Direct the Human 70-kDa Peroxisomal Integral Membrane Protein (PMP70) to Peroxisomes. <i>Biochemical and Biophysical Research Communications</i> , 2001, 285, 649-655.	2.1	26
116	Characterization and functional analysis of the nucleotide binding fold in human peroxisomal ATP binding cassette transporters. <i>FEBS Letters</i> , 2001, 492, 66-72.	2.8	34
117	Genetic and clinical aspects of X-linked hydrocephalus (L1 disease): Mutations in the L1CAM gene. <i>Human Mutation</i> , 2001, 18, 1-12.	2.5	175
118	Characteristic Clinical Features of Idiopathic Neuralgic Amyotrophy in Childhood. <i>Neuropediatrics</i> , 2001, 32, 110-110.	0.6	4
119	Retrobulbar Abscess in a Neonate. <i>Neuropediatrics</i> , 2001, 32, 219-220.	0.6	4
120	Multiple symmetric lipomatosis: an unusual cause of childhood obesity and mental retardation. <i>European Journal of Paediatric Neurology</i> , 2000, 4, 63-67.	1.6	32
121	The Human PEX3 Gene Encoding a Peroxisomal Assembly Protein: Genomic Organization, Positional Mapping, and Mutation Analysis in Candidate Phenotypes. <i>Biochemical and Biophysical Research Communications</i> , 2000, 268, 704-710.	2.1	8
122	Genomic Organization and Characterization of Human PEX2 Encoding a 35-kDa Peroxisomal Membrane Protein. <i>Biochemical and Biophysical Research Communications</i> , 2000, 273, 985-990.	2.1	3
123	Restoration of PEX2 peroxisome assembly defects by overexpression of PMP70. <i>European Journal of Cell Biology</i> , 1998, 76, 237-245.	3.6	14
124	Genomic Organization of the 70-kDa Peroxisomal Membrane Protein Gene (PXMP1). <i>Genomics</i> , 1998, 48, 203-208.	2.9	18
125	Clinical and Genetic Aspects of X-Linked Adrenoleukodystrophy. <i>Neuropediatrics</i> , 1998, 29, 3-13.	0.6	47
126	The value of new MRI techniques in adrenoleukodystrophy. <i>Pediatric Radiology</i> , 1997, 27, 207-215.	2.0	16



#	ARTICLE	IF	CITATIONS
127	Localization of the 70-kDa Peroxisomal Membrane Protein to Human 1p21-p22 and Mouse 3. <i>Genomics</i> , 1993, 15, 412-414.	2.9	19
128	The 70 kDa peroxisomal membrane protein: an ATP-binding cassette transporter protein involved in peroxisome biogenesis. <i>Seminars in Cell Biology</i> , 1993, 4, 45-52.	3.4	29
129	Penetrating the peroxisome. <i>Nature</i> , 1993, 361, 682-683.	27.8	73
130	Mutations in the 70K peroxisomal membrane protein gene in Zellweger syndrome. <i>Nature Genetics</i> , 1992, 1, 16-23.	21.4	238
131	The 22-kD Peroxisomal Integral Membrane Protein in Zellweger Syndrome—Presence, Abundance, and Association with a Peroxisomal Thiolase Precursor Protein. <i>Pediatric Research</i> , 1991, 29, 141-146.	2.3	26
132	Membrane Fluidity of Nonmuscle Cells in Duchenne Muscular Dystrophy: Effect on Lymphocyte Membranes of Incubation in Patient and Control Sera. <i>Pediatric Research</i> , 1987, 22, 488-492.	2.3	8