Jutta GÃ¤rtner

List of Publications by Year
in descending order

Version: 2024-02-01

132 papers	6,672 citations	70961 41 h-index	74018 75 g-index
137 all docs	137 docs citations	137 times ranked	11192 citing authors

#	Article	IF	Citations
1	Inspiration Is the Major Regulator of Human CSF Flow. Journal of Neuroscience, 2015, 35, 2485-2491.	1.7	261
2	Mutations in the Gene Encoding Gap Junction Protein Î±12 (Connexin 46.6) Cause Pelizaeus-Merzbacherâ€“Like Disease. American Journal of Human Genetics, 2004, 75, 251-260.	2.6	257
3	Cathepsin D Deficiency Is Associated with a Human Neurodegenerative Disorder. American Journal of Human Genetics, 2006, 78, 988-998.	2.6	255
4	Mutations in the 70K peroxisomal membrane protein gene in Zellweger syndrome. Nature Genetics, 1992, 1, 16-23.	9.4	238
5	Trial of Fingolimod versus Interferon Beta-1a in Pediatric Multiple Sclerosis. New England Journal of Medicine, 2018, 379, 1017-1027.	13.9	237
6	Folate Receptor Alpha Defect Causes Cerebral Folate Transport Deficiency: A Treatable Neurodegenerative Disorder Associated with Disturbed Myelin Metabolism. American Journal of Human Genetics, 2009, 85, 354-363.	2.6	228
7	Heterozygous de-novo mutations in ATP1A3 in patients with alternating hemiplegia of childhood: a whole-exome sequencing gene-identification study. Lancet Neurology, The, 2012, 11, 764-773.	4.9	223
8	Antiâ€“Myelin Oligodendrocyte Glycoprotein Antibodies in Pediatric Patients With Optic Neuritis. Archives of Neurology, 2012, 69, 752-6.	4.9	181
9	Genetic and clinical aspects of X-linked hydrocephalus (L1 disease): Mutations in theL1CAMgene. Human Mutation, 2001, 18, 1-12.	1.1	175
10	Clinical and biochemical spectrum of D-bifunctional protein deficiency. Annals of Neurology, 2006, 59, 92-104.	2.8	175
11	Wild-type microglia do not reverse pathology in mouse models of Rett syndrome. Nature, 2015, 521, E1-E4.	13.7	159
12	Peroxisomal lactate dehydrogenase is generated by translational readthrough in mammals. ELife, 2014, 3, e03640.	2.8	155
13	Breathing dysfunctions associated with impaired control of postinspiratory activity inMecp2â^'/yknockout mice. Journal of Physiology, 2007, 579, 863-876.	1.3	143
14	Acute disseminated encephalomyelitis followed by recurrent or monophasic optic neuritis in pediatric patients. Multiple Sclerosis Journal, 2013, 19, 941-946.	1.4	135
15	Identification of the Upward Movement of Human CSFIn Vivoand its Relation to the Brain Venous System. Journal of Neuroscience, 2017, 37, 2395-2402.	1.7	133
16	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	4.1	131
17	RNASET2-deficient cystic leukoencephalopathy resembles congenital cytomegalovirus brain infection. Nature Genetics, 2009, 41, 773-775.	9.4	124
18	Molecularly defined diffuse leptomeningeal glioneuronal tumor (DLGNT) comprises two subgroups with distinct clinical and genetic features. Acta Neuropathologica, 2018, 136, 239-253.	3.9	118

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19	Peroxisomes are juxtaposed to strategic sites on mitochondria. Molecular BioSystems, 2014, 10, 1742-1748.	2.9	95
20	rnaset2 mutant zebrafish model familial cystic leukoencephalopathy and reveal a role for RNase T2 in degrading ribosomal RNA. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 1099-1103.	3.3	91
21	Readthrough of nonsense mutations in Rett syndrome: evaluation of novel aminoglycosides and generation of a new mouse model. Journal of Molecular Medicine, 2011, 89, 389-398.	1.7	90
22	Natalizumab Use in Pediatric Multiple Sclerosis. Archives of Neurology, 2008, 65, 1655-8.	4.9	86
23	Mutations in SLC33A1 Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. American Journal of Human Genetics, 2012, 90, 61-68.	2.6	85
24	Immune Sensing of Synthetic, Bacterial, and Protozoan RNA by Toll-like Receptor 8 Requires Coordinated Processing by RNase T2 and RNase 2. Immunity, 2020, 52, 591-605.e6.	6.6	83
25	Molecular analysis ofSUMF1 mutations: stability and residual activity of mutant formylglycine-generating enzyme determine disease severity in multiple sulfatase deficiency. Human Mutation, 2008, 29, 205-205.	1.1	74
26	Penetrating the peroxisome. Nature, 1993, 361, 682-683.	13.7	73
27	Extensive acute axonal damage in pediatric multiple sclerosis lesions. Annals of Neurology, 2015, 77, 655-667.	2.8	69
28	Cerebral involvement in axonal Charcot-Marie-Tooth neuropathy caused by mitofusin2 mutations. Journal of Neurology, 2008, 255, 1049-58.	1.8	66
29	SUMF1 mutations affecting stability and activity of formylglycine generating enzyme predict clinical outcome in multiple sulfatase deficiency. European Journal of Human Genetics, 2011, 19, 253-261.	1.4	63
30	Therapy of highly active pediatric multiple sclerosis. Multiple Sclerosis Journal, 2019, 25, 72-80.	1.4	60
31	PEX1 Mutations in Complementation Group 1 of Zellweger Spectrum Patients Correlate with Severity of Disease. Pediatric Research, 2002, 51, 706-714.	1.1	58
32	Two Different Targeting Signals Direct Human Peroxisomal Membrane Protein 22 to Peroxisomes. Journal of Biological Chemistry, 2002, 277, 774-784.	1.6	57
33	Respiration and the watershed of spinal CSF flow in humans. Scientific Reports, 2018, 8, 5594.	1.6	53
34	From ventriculomegaly to severe muscular atrophy: Expansion of the clinical spectrum related to mutations in AIFM1. Mitochondrion, 2015, 21, 12-18.	1.6	51
35	Live Cell FRET Microscopy. Journal of Biological Chemistry, 2007, 282, 26997-27005.	1.6	50
36	Pediatric multiple sclerosis: Detection of clinically silent lesions by multimodal evoked potentials. Journal of Pediatrics, 2006, 149, 125-127.	0.9	49

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37	Identification of novel mutations inPEX2,PEX6,PEX10,PEX12, andPEX13in Zellweger spectrum patients. Human Mutation, 2006, 27, 1157-1157.	1.1	49
38	Spontaneous central apneas occur in the C57BL/6J mouse strain. Respiratory Physiology and Neurobiology, 2008, 160, 21-27.	0.7	49
39	Ketogenic diet ameliorates axonal defects and promotes myelination in Pelizaeusâ€“Merzbacher disease. Acta Neuropathologica, 2019, 138, 147-161.	3.9	48
40	Clinical and Genetic Aspects of X-Linked Adrenoleukodystrophy. Neuropediatrics, 1998, 29, 3-13.	0.3	47
41	MicroRNA regulation in experimental autoimmune encephalomyelitis in mice and marmosets resembles regulation in human multiple sclerosis lesions. Journal of Neuroimmunology, 2012, 246, 27-33.	1.1	47
42	Common infectious agents in multiple sclerosis: a caseâ€”control study in children. Multiple Sclerosis Journal, 2008, 14, 136-139.	1.4	44
43	Structure of Tripeptidyl-peptidase I Provides Insight into the Molecular Basis of Late Infantile Neuronal Ceroid Lipofuscinosis. Journal of Biological Chemistry, 2009, 284, 3976-3984.	1.6	43
44	Peroxisome Formation Requires the Endoplasmic Reticulum Channel Protein <scp>S</scp>ec61. Traffic, 2012, 13, 599-609.	1.3	42
45	Phenotypic overlap of alternating hemiplegia of childhood and CAPOS syndrome. Neurology, 2014, 83, 861-863.	1.5	42
46	The functional readthrough extension of malate dehydrogenase reveals a modification of the genetic code. Open Biology, 2016, 6, 160246.	1.5	41
47	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	2.6	41
48	Tectonic gene mutations in patients with Joubert syndrome. European Journal of Human Genetics, 2015, 23, 616-620.	1.4	40
49	Tau, Phospho-Tau, and S-100B in the Cerebrospinal Fluid of Children With Multiple Sclerosis. Journal of Child Neurology, 2005, 20, 822-825.	0.7	37
50	Structure and activity of the only human RNase T2. Nucleic Acids Research, 2012, 40, 8733-8742.	6.5	37
51	Microduplication of 3p26.3 in Nonsyndromic Intellectual Disability Indicates an Important Role of CHL1 for Normal Cognitive Function. Neuropediatrics, 2013, 44, 268-271.	0.3	37
52	Suppression of Nonsense Mutations in Rett Syndrome by Aminoglycoside Antibiotics. Pediatric Research, 2009, 65, 520-523.	1.1	36
53	Cerebral metabolic and structural alterations in hereditary spastic paraplegia with thin corpus callosum assessed by MRS and DTI. Neuroradiology, 2006, 48, 893-898.	1.1	35
54	Therapeutic Apheresis in Pediatric Patients with Acute CNS Inflammatory Demyelinating Disease. Blood Purification, 2013, 36, 92-97.	0.9	35

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55	The failure of microglia to digest developmental apoptotic cells contributes to the pathology of RNASET2â€deficient leukoencephalopathy. Glia, 2020, 68, 1531-1545.	2.5	35
56	Characterization and functional analysis of the nucleotide binding fold in human peroxisomal ATP binding cassette transporters. FEBS Letters, 2001, 492, 66-72.	1.3	34
57	Molecular and biochemical characterization of a unique mutation in CCS, the human copper chaperone to superoxide dismutase. Human Mutation, 2012, 33, 1207-1215.	1.1	34
58	Dysferlin mediates membrane tubulation and links T-tubule biogenesis to muscular dystrophy. Journal of Cell Science, 2017, 130, 841-852.	1.2	34
59	Rational diagnostic strategy for Zellweger syndrome spectrum patients. European Journal of Human Genetics, 2009, 17, 741-748.	1.4	33
60	First PEX11Î² patient extends spectrum of peroxisomal biogenesis disorder phenotypes: Table 1. Journal of Medical Genetics, 2012, 49, 314-316.	1.5	33
61	Pediatric onset multiple sclerosis: McDonald criteria 2010 and the contribution of spinal cord MRI. Multiple Sclerosis Journal, 2013, 19, 1330-1335.	1.4	33
62	STAR syndrome-associated CDK10/Cyclin M regulates actin network architecture and ciliogenesis. Cell Cycle, 2016, 15, 678-688.	1.3	33
63	Multiple symmetric lipomatosis: an unusual cause of childhood obesity and mental retardation. European Journal of Paediatric Neurology, 2000, 4, 63-67.	0.7	32
64	Characterization of the MeCP2R168X Knockin Mouse Model for Rett Syndrome. PLoS ONE, 2014, 9, e115444.	1.1	32
65	Colorectal cancer in two pre-teenage siblings with familial adenomatous polyposis. European Journal of Pediatrics, 2005, 164, 306-310.	1.3	31
66	Identification of a New Fatty Acid Synthesis-Transport Machinery at the Peroxisomal Membrane*. Journal of Biological Chemistry, 2012, 287, 210-221.	1.6	31
67	Potential Risks to Stable Long-term Outcome of Allogeneic Hematopoietic Stem Cell Transplantation for Children With Cerebral X-linked Adrenoleukodystrophy. JAMA Network Open, 2018, 1, e180769.	2.8	30
68	The 70 kDa peroxisomal membrane protein: an ATP-binding cassette transporter protein involved in peroxisome biogenesis. Seminars in Cell Biology, 1993, 4, 45-52.	3.5	29
69	Genotypeâ€“phenotype analysis in patients with giant axonal neuropathy (GAN). Neuromuscular Disorders, 2007, 17, 624-630.	0.3	29
70	Structure of sulfamidase provides insight into the molecular pathology of mucopolysaccharidosis IIIA. Acta Crystallographica Section D: Biological Crystallography, 2014, 70, 1321-1335.	2.5	29
71	Pelizaeusâ€“Merzbacher-like disease is caused not only by a loss of connexin47 function but also by a hemichannel dysfunction. European Journal of Human Genetics, 2010, 18, 985-992.	1.4	27
72	The 22-kD Peroxisomal Integral Membrane Protein in Zellweger Syndromeâ€”Presence, Abundance, and Association with a Peroxisomal Thiolase Precursor Protein. Pediatric Research, 1991, 29, 141-146.	1.1	26

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73	Targeting Elements in the Amino-Terminal Part Direct the Human 70-kDa Peroxisomal Integral Membrane Protein (PMP70) to Peroxisomes. Biochemical and Biophysical Research Communications, 2001, 285, 649-655.	1.0	26
74	Axonal neuropathy with unusual pattern of amyotrophy and alacrima associated with a novel AAAS mutation p.Leu430Phe. European Journal of Human Genetics, 2008, 16, 1499-1506.	1.4	26
75	Leukodystrophies and other genetic metabolic leukoencephalopathies in children and adults. Brain and Development, 2010, 32, 82-89.	0.6	26
76	Functional analysis of PEX13 mutation in a Zellweger syndrome spectrum patient reveals novel homooligomerization of PEX13 and its role in human peroxisome biogenesis. Human Molecular Genetics, 2013, 22, 3844-3857.	1.4	26
77	Mutations in classical late infantile neuronal ceroid lipofuscinosis disrupt transport of tripeptidyl-peptidase I to lysosomes. Human Molecular Genetics, 2004, 13, 2483-2491.	1.4	25
78	West syndrome, microcephaly, grey matter heterotopia and hypoplasia of corpus callosum due to a novel ARFGEF2 mutation. Journal of Medical Genetics, 2013, 50, 772-775.	1.5	24
79	A novel ATP1A3 mutation with unique clinical presentation. Journal of the Neurological Sciences, 2014, 341, 133-135.	0.3	24
80	Natural history of multiple sulfatase deficiency: Retrospective phenotyping and functional variant analysis to characterize an ultraâ€rare disease. Journal of Inherited Metabolic Disease, 2020, 43, 1298-1309.	1.7	23
81	Sequence diversity ofKIAA0027/MLC1:are megalencephalic leukoencephalopathy and schizophrenia allelic disorders?. Human Mutation, 2003, 21, 45-52.	1.1	22
82	Deep breathing couples CSF and venous flow dynamics. Scientific Reports, 2022, 12, 2568.	1.6	22
83	Assessment of myelination in hypomyelinating disorders by quantitative MRI. Journal of Magnetic Resonance Imaging, 2012, 36, 1329-1338.	1.9	21
84	JC virus antibody status in a pediatric multiple sclerosis cohort: Prevalence, conversion rate and influence on disease severity. Multiple Sclerosis Journal, 2015, 21, 382-387.	1.4	21
85	Leukoencephalopathy and early death associated with an Ashkenazi-Jewish founder mutation in the Hikeshi gene. Journal of Medical Genetics, 2016, 53, 132-137.	1.5	21
86	Immunoglobulin Therapy in Idiopathic Hypothalamic Dysfunction. Pediatric Neurology, 2009, 41, 232-234.	1.0	20
87	Localization of the 70-kDa Peroxisomal Membrane Protein to Human 1p21-p22 and Mouse 3. Genomics, 1993, 15, 412-414.	1.3	19
88	Serial proton MR spectroscopy and diffusion tensor imaging in infantile Baloâ€™s concentric sclerosis. Neuroradiology, 2009, 51, 113-121.	1.1	19
89	Rapid degradation of an active formylglycine generating enzyme variant leads to a late infantile severe form of multiple sulfatase deficiency. European Journal of Human Genetics, 2013, 21, 1020-1023.	1.4	19
90	Compound heterozygous variants in PGAP1 causing severe psychomotor retardation, brain atrophy, recurrent apneas and delayed myelination: a case report and literature review. BMC Neurology, 2016, 16, 74.	0.8	19

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91	Genomic Organization of the 70-kDa Peroxisomal Membrane Protein Gene (PXMP1). Genomics, 1998, 48, 203-208.	1.3	18
92	The cystathionine beta-synthase variant c.844_845ins68 protects against CNS demyelination in X-linked adrenoleukodystrophy. Human Mutation, 2006, 27, 1063-1064.	1.1	18
93	Genotype and Protein Expression After Bone Marrow Transplantation for Adrenoleukodystrophy. Archives of Neurology, 2007, 64, 651.	4.9	18
94	Cognitive deficits including executive functioning in relation to clinical parameters in paediatric MS patients. PLoS ONE, 2018, 13, e0194873.	1.1	18
95	The value of new MRI techniques in adrenoleukodystrophy. Pediatric Radiology, 1997, 27, 207-215.	1.1	16
96	Polymicrogyria in fetal alcohol syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 128-131.	1.6	16
97	Inborn errors of metabolism leading to neuronal migration defects. Journal of Inherited Metabolic Disease, 2020, 43, 145-155.	1.7	16
98	Biâ€allelic VPS16 variants limit HOPS/CORVET levels and cause a mucopolysaccharidosisâ€like disease. EMBO Molecular Medicine, 2021, 13, e13376.	3.3	16
99	Interferon-driven brain phenotype in a mouse model of RNaseT2 deficient leukoencephalopathy. Nature Communications, 2021, 12, 6530.	5.8	16
100	Cerebellar ataxia, mental retardation and dysequilibrium syndrome 1 (CAMRQ1) caused by an unusual constellation of VLDLR mutation. Journal of Neurology, 2013, 260, 1678-1680.	1.8	15
101	Restoration of PEX2 peroxisome assembly defects by overexpression of PMP70. European Journal of Cell Biology, 1998, 76, 237-245.	1.6	14
102	The peroxisomal membrane targeting elements of human peroxin 2 (PEX2). European Journal of Cell Biology, 2003, 82, 155-162.	1.6	14
103	A systematic review and metaâ€analysis of published cases reveals the natural disease history in multiple sulfatase deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 1288-1297.	1.7	14
104	Early Reduction of Total N-Acetyl-Aspartate-Compounds in Patients With Classical Vanishing White Matter Disease. A Long-Term Follow-Up MRS Study. Pediatric Research, 2008, 63, 444-449.	1.1	12
105	Cln5 represents a new type of cysteine-based S -depalmitoylase linked to neurodegeneration. Science Advances, 2022, 8, eabj8633.	4.7	12
106	MRI-based diagnostic biomarkers for early onset pediatric multiple sclerosis. NeuroImage: Clinical, 2015, 7, 400-408.	1.4	9
107	Membrane Fluidity of Nonmuscle Cells in Duchenne Muscular Dystrophy: Effect on Lymphocyte Membranes of Incubation in Patient and Control Sera. Pediatric Research, 1987, 22, 488-492.	1.1	8
108	The Human PEX3 Gene Encoding a Peroxisomal Assembly Protein: Genomic Organization, Positional Mapping, and Mutation Analysis in Candidate Phenotypes. Biochemical and Biophysical Research Communications, 2000, 268, 704-710.	1.0	8

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109	Upward movement of cerebrospinal fluid in obstructive hydrocephalusâ€"revision of an old concept. Child's Nervous System, 2019, 35, 833-841.	0.6	8
110	Comparative analysis of alternating hemiplegia of childhood and rapid-onset dystonia-parkinsonism ATP1A3 mutations reveals functional deficits, which do not correlate with disease severity. Neurobiology of Disease, 2020, 143, 105012.	2.1	8
111	Mannose phosphate isomerase deficiencyâ€congenital disorder of glycosylation (MPIâ€CDG) with cerebral venous sinus thrombosis as first and only presenting symptom: A rare but treatable cause of thrombophilia. JIMD Reports, 2020, 55, 38-43.	0.7	8
112	Severe neonatal multiple sulfatase deficiency presenting with hydrops fetalis in a preterm birth patient. JIMD Reports, 2019, 49, 48-52.	0.7	7
113	Breathing drives CSF: Impact on spaceflight disease and hydrocephalus. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 20263-20264.	3.3	7
114	Targeted metabolomics revealed changes in phospholipids during the development of neuroinflammation in <scp>Abcd1^{tm1Kds}</scp> mice and Xâ€linked adrenoleukodystrophy patients. Journal of Inherited Metabolic Disease, 2021, 44, 1174-1185.	1.7	7
115	Concurrent axon and myelin destruction differentiates Xâ€linked adrenoleukodystrophy from multiple sclerosis. Glia, 2021, 69, 2362-2377.	2.5	7
116	Detection of Tripeptidyl Peptidase I Activity in Living Cells by Fluorogenic Substrates. Journal of Histochemistry and Cytochemistry, 2006, 54, 991-996.	1.3	6
117	Clinical utility gene card for: Zellweger syndrome spectrum. European Journal of Human Genetics, 2015, 23, 1111-1111.	1.4	6
118	Opening New Horizons in the Treatment of Childhood Onset Leukodystrophies. Neuropediatrics, 2019, 50, 211-218.	0.3	6
119	Temporal profile of lymphocyte counts and relationship with infections with fingolimod therapy in paediatric patients with multiple sclerosis: Results from the PARADIGMS study. Multiple Sclerosis Journal, 2021, 27, 922-932.	1.4	5
120	Characteristic Clinical Features of Idiopathic Neuralgic Amyotrophy in Childhood. Neuropediatrics, 2001, 32, 110-110.	0.3	4
121	Retrobulbar Abscess in a Neonate. Neuropediatrics, 2001, 32, 219-220.	0.3	4
122	Tumefactive inflammatory lesions in juvenile metachromatic leukodystrophy. Neurology: Neuroimmunology and NeuroInflammation, 2021, 8, .	3.1	4
123	Hydrocephalus Revisited: New Insights into Dynamics of Neurofluids on Macro- and Microscales. Neuropediatrics, 2021, 52, 233-241.	0.3	4
124	TTC5 syndrome: Clinical and molecular spectrum of a severe and recognizable condition. American Journal of Medical Genetics, Part A, 2022, 188, 2652-2665.	0.7	4
125	Genomic Organization and Characterization of Human PEX2 Encoding a 35-kDa Peroxisomal Membrane Protein. Biochemical and Biophysical Research Communications, 2000, 273, 985-990.	1.0	3
126	Visually Self-induced Seizures Sensitive to Round Objects. Epilepsia, 2005, 46, 786-789.	2.6	3

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127	A novel remitting leukodystrophy associated with a variant in FBP2. Brain Communications, 2021, 3, fcab036.	1.5	2
128	Frequent but nonspecific venous narrowing in paediatric multiple sclerosis. Multiple Sclerosis Journal, 2012, 18, 1805-1805.	1.4	1
129	B cell depletion can be effective in multiple sclerosis but failed in a patient with advanced childhood cerebral X-linked adrenoleukodystrophy. Therapeutic Advances in Neurological Disorders, 2019, 12, 175628641986813.	1.5	1
130	Dopamine-Mediated Yawning-Fatigue Syndrome With Specific Recurrent Initiation and Responsiveness to Opioids. JAMA Neurology, 2020, 77, 254.	4.5	1
131	Follow-Up of a Case of Dopamine-Mediated Yawning-Fatigue-Syndrome Responsive to Opioids, Successful Desensitization via Graded Activity Treatment. Neurology International, 2021, 13, 79-84.	1.3	0
132	Improving pediatric multiple sclerosis interventional phase III study design: a meta-analysis. Therapeutic Advances in Neurological Disorders, 2022, 15, 175628642110704.	1.5	0