

Aurora Pujol

List of Publications by Year in descending order

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Version: 2024-02-01

141
papers

15,991
citations

41344

49
h-index

18130

120
g-index

144
all docs

144
docs citations

144
times ranked

29386
citing authors

#	ARTICLE	IF	CITATIONS
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	9.1	4,701
2	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	12.6	1,983
3	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , 2020, 370, .	12.6	1,749
4	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. <i>Science Immunology</i> , 2021, 6, .	11.9	357
5	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , 2021, 6, .	11.9	267
6	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. <i>Human Mutation</i> , 2001, 18, 499-515.	2.5	261
7	Human genetic and immunological determinants of critical COVID-19 pneumonia. <i>Nature</i> , 2022, 603, 587-598.	27.8	216
8	Pharmacological Inhibition of Poly(ADP-Ribose) Polymerases Improves Fitness and Mitochondrial Function in Skeletal Muscle. <i>Cell Metabolism</i> , 2014, 19, 1034-1041.	16.2	211
9	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. <i>Cell</i> , 2020, 181, 1194-1199.	28.9	185
10	Early oxidative damage underlying neurodegeneration in X-adrenoleukodystrophy. <i>Human Molecular Genetics</i> , 2008, 17, 1762-1773.	2.9	181
11	Late onset neurological phenotype of the X-ALD gene inactivation in mice: a mouse model for adrenomyeloneuropathy. <i>Human Molecular Genetics</i> , 2002, 11, 499-505.	2.9	176
12	Functional overlap between ABCD1 (ALD) and ABCD2 (ALDR) transporters: a therapeutic target for X-adrenoleukodystrophy. <i>Human Molecular Genetics</i> , 2004, 13, 2997-3006.	2.9	170
13	The Evolutionary Origin of Peroxisomes: An ER-Peroxisome Connection. <i>Molecular Biology and Evolution</i> , 2006, 23, 838-845.	8.9	152
14	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	6.2	136
15	Antioxidants halt axonal degeneration in a mouse model of X-adrenoleukodystrophy. <i>Annals of Neurology</i> , 2011, 70, 84-92.	5.3	122
16	Pathomechanisms Underlying X-adrenoleukodystrophy: A Three-Hit Hypothesis. <i>Brain Pathology</i> , 2010, 20, 838-844.	4.1	116
17	Methylene blue upregulates Nrf2/ARE genes and prevents tau-related neurotoxicity. <i>Human Molecular Genetics</i> , 2014, 23, 3716-3732.	2.9	115
18	General Aspects and Neuropathology of X-linked Adrenoleukodystrophy. <i>Brain Pathology</i> , 2010, 20, 817-830.	4.1	112

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19	Functional Genomics Reveals Dysregulation of Cortical Olfactory Receptors in Parkinson Disease: Novel Putative Chemoreceptors in the Human Brain. <i>Journal of Neuropathology and Experimental Neurology</i> , 2013, 72, 524-539.	1.7	111
20	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, e2200413119.	7.1	110
21	Deregulation of purine metabolism in Alzheimer's disease. <i>Neurobiology of Aging</i> , 2015, 36, 68-80.	3.1	108
22	Neuroinflammatory Signals in Alzheimer Disease and APP/PS1 Transgenic Mice. <i>Journal of Neuropathology and Experimental Neurology</i> , 2015, 74, 319-344.	1.7	105
23	PeroxisomeDB 2.0: an integrative view of the global peroxisomal metabolome. <i>Nucleic Acids Research</i> , 2010, 38, D800-D805.	14.5	103
24	SARS-CoV-2 related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	100
25	Neutralizing Autoantibodies to Type I IFNs in >10% of Patients with Severe COVID-19 Pneumonia Hospitalized in Madrid, Spain. <i>Journal of Clinical Immunology</i> , 2021, 41, 914-922.	3.8	100
26	Amyloid Generation and Dysfunctional Immunoproteasome Activation with Disease Progression in Animal Model of Familial Alzheimer's Disease. <i>Brain Pathology</i> , 2012, 22, 636-653.	4.1	95
27	Impaired mitochondrial oxidative phosphorylation in the peroxisomal disease X-linked adrenoleukodystrophy. <i>Human Molecular Genetics</i> , 2013, 22, 3296-3305.	2.9	95
28	A key role for the peroxisomal ABCD2 transporter in fatty acid homeostasis. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , 2009, 296, E211-E221.	3.5	91
29	Inactivation of the peroxisomal ABCD2 transporter in the mouse leads to late-onset ataxia involving mitochondria, Golgi and endoplasmic reticulum damage. <i>Human Molecular Genetics</i> , 2005, 14, 3565-3577.	2.9	90
30	Valproic acid induces antioxidant effects in X-linked adrenoleukodystrophy. <i>Human Molecular Genetics</i> , 2010, 19, 2005-2014.	2.9	90
31	Epigenome-wide association study of COVID-19 severity with respiratory failure. <i>EBioMedicine</i> , 2021, 66, 103339.	6.1	90
32	Accelerated biological aging in COVID-19 patients. <i>Nature Communications</i> , 2022, 13, 2135.	12.8	87
33	Oxidative stress underlying axonal degeneration in adrenoleukodystrophy: A paradigm for multifactorial neurodegenerative diseases?. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1475-1488.	3.8	84
34	Current and Future Pharmacological Treatment Strategies in X-Linked Adrenoleukodystrophy. <i>Brain Pathology</i> , 2010, 20, 845-856.	4.1	80
35	Complete nucleotide sequence of <i>Saccharomyces cerevisiae</i> chromosome X.. <i>EMBO Journal</i> , 1996, 15, 2031-2049.	7.8	78
36	Oxidative Damage Compromises Energy Metabolism in the Axonal Degeneration Mouse Model of X-Adrenoleukodystrophy. <i>Antioxidants and Redox Signaling</i> , 2011, 15, 2095-2107.	5.4	78

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37	Oxidative stress modulates mitochondrial failure and cyclophilin D function in X-linked adrenoleukodystrophy. <i>Brain</i> , 2012, 135, 3584-3598.	7.6	78
38	Bezafibrate administration improves behavioral deficits and tau pathology in P301S mice. <i>Human Molecular Genetics</i> , 2012, 21, 5091-5105.	2.9	77
39	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 388-396.	1.1	76
40	Neurochemistry and the non-motor aspects of PD. <i>Neurobiology of Disease</i> , 2012, 46, 508-526.	4.4	73
41	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. <i>Nature Medicine</i> , 2022, 28, 879-882.	30.7	72
42	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. <i>Brain</i> , 2019, 142, 1561-1572.	7.6	70
43	Pioglitazone halts axonal degeneration in a mouse model of X-linked adrenoleukodystrophy. <i>Brain</i> , 2013, 136, 2432-2443.	7.6	69
44	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. <i>Journal of Clinical Investigation</i> , 2019, 129, 1240-1256.	8.2	68
45	PeroxisomeDB: a database for the peroxisomal proteome, functional genomics and disease. <i>Nucleic Acids Research</i> , 2007, 35, D815-D822.	14.5	65
46	Functional genomic analysis unravels a metabolic-inflammatory interplay in adrenoleukodystrophy. <i>Human Molecular Genetics</i> , 2012, 21, 1062-1077.	2.9	62
47	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. <i>Journal of Experimental Medicine</i> , 2022, 219, .	8.5	59
48	Activation of Promoter P4 of the Autonomous Parvovirus Minute Virus of Mice at Early S Phase Is Required for Productive Infection. <i>Journal of Virology</i> , 1999, 73, 3877-3885.	3.4	55
49	The nucleotide sequence of <i>Saccharomyces cerevisiae</i> chromosome XV. <i>Nature</i> , 1997, 387, 98-102.	27.8	54
50	Oxidative stress, mitochondrial and proteostasis malfunction in adrenoleukodystrophy: A paradigm for axonal degeneration. <i>Free Radical Biology and Medicine</i> , 2015, 88, 18-29.	2.9	54
51	Mitochondrial dysfunction in central nervous system white matter disorders. <i>Glia</i> , 2014, 62, 1878-1894.	4.9	52
52	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. <i>EBioMedicine</i> , 2021, 65, 103246.	6.1	52
53	Mouse liver PMP70 and ALDP: homomeric interactions prevail in vivo. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2004, 1689, 235-243.	3.8	51
54	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	6.2	51

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55	PGC β : overexpression exacerbates β -amyloid and tau deposition in a transgenic mouse model of Alzheimer's disease. <i>FASEB Journal</i> , 2014, 28, 1745-1755.	0.5	47
56	Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis. <i>American Journal of Human Genetics</i> , 2017, 100, 105-116.	6.2	46
57	Isolation of a fully infectious variant of parvovirus H-1 supplanting the standard strain in human cells. <i>Journal of Virology</i> , 1995, 69, 4538-4543.	3.4	45
58	Steroid Hormones Control Circadian Elov3 Expression in Mouse Liver. <i>Endocrinology</i> , 2008, 149, 3158-3166.	2.8	44
59	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. <i>Movement Disorders</i> , 2019, 34, 1547-1561.	3.9	44
60	Inhibition of parvovirus minute virus of mice replication by a peptide involved in the oligomerization of nonstructural protein NS1. <i>Journal of Virology</i> , 1997, 71, 7393-7403.	3.4	44
61	Astrocytes and mitochondria from adrenoleukodystrophy protein (ABCD1)-deficient mice reveal that the adrenoleukodystrophy-associated very long-chain fatty acids target several cellular energy-dependent functions. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 925-936.	3.8	43
62	Tauroursodeoxycholic bile acid arrests axonal degeneration by inhibiting the unfolded protein response in X-linked adrenoleukodystrophy. <i>Acta Neuropathologica</i> , 2017, 133, 283-301.	7.7	43
63	Clinical presentation and proteomic signature of patients with <i>TANGO2</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 297-308.	3.6	43
64	Mitochondrial dysfunction and oxidative damage cooperatively fuel axonal degeneration in X-linked adrenoleukodystrophy. <i>Biochimie</i> , 2014, 98, 143-149.	2.6	42
65	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 965-976.	6.2	41
66	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. <i>Nature Immunology</i> , 2022, 23, 159-164.	14.5	41
67	The role of peroxisomal ABC transporters in the mouse adrenal gland: the loss of <i>Abcd2</i> (ALDR), Not <i>Abcd1</i> (ALD), causes oxidative damage. <i>Laboratory Investigation</i> , 2007, 87, 261-272.	3.7	39
68	Bezafibrate lowers very long-chain fatty acids in X-linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1137-1145.	3.6	39
69	Glutathione imbalance in patients with X-linked adrenoleukodystrophy. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 366-370.	1.1	39
70	Oxidative stress regulates the ubiquitin-proteasome system and immunoproteasome functioning in a mouse model of X-adrenoleukodystrophy. <i>Brain</i> , 2013, 136, 891-904.	7.6	39
71	Autophagy induction halts axonal degeneration in a mouse model of X-adrenoleukodystrophy. <i>Acta Neuropathologica</i> , 2015, 129, 399-415.	7.7	39
72	Harnessing Type I IFN Immunity Against SARS-CoV-2 with Early Administration of IFN- β . <i>Journal of Clinical Immunology</i> , 2021, 41, 1425-1442.	3.8	39

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73	Improvement of the Rett Syndrome Phenotype in a Mecp2 Mouse Model Upon Treatment with Levodopa and a Dopa-Decarboxylase Inhibitor. <i>Neuropsychopharmacology</i> , 2014, 39, 2846-2856.	5.4	38
74	Altered glycolipid and glycerophospholipid signaling drive inflammatory cascades in adrenomyeloneuropathy. <i>Human Molecular Genetics</i> , 2015, 24, ddv375.	2.9	37
75	Loss of <i>SIRT2</i> leads to axonal degeneration and locomotor disability associated with redox and energy imbalance. <i>Aging Cell</i> , 2017, 16, 1404-1413.	6.7	36
76	Inhibition of Gsk3b Reduces Nfkb1 Signaling and Rescues Synaptic Activity to Improve the Rett Syndrome Phenotype in Mecp2-Knockout Mice. <i>Cell Reports</i> , 2018, 23, 1665-1677.	6.4	36
77	Aberrant regulation of the <i>GSK3β/NRF2</i> axis unveils a novel therapy for adrenoleukodystrophy. <i>EMBO Molecular Medicine</i> , 2018, 10, .	6.9	35
78	CSVS, a crowdsourcing database of the Spanish population genetic variability. <i>Nucleic Acids Research</i> , 2021, 49, D1130-D1137.	14.5	34
79	Staging anti-inflammatory therapy in Alzheimer's disease. <i>Frontiers in Aging Neuroscience</i> , 2010, 2, 142.	3.4	32
80	ABCD2 is abundant in adipose tissue and opposes the accumulation of dietary erucic acid (C22:1) in fat. <i>Journal of Lipid Research</i> , 2010, 51, 162-168.	4.2	31
81	Biomarker Identification, Safety, and Efficacy of High-Dose Antioxidants for Adrenomyeloneuropathy: a Phase II Pilot Study. <i>Neurotherapeutics</i> , 2019, 16, 1167-1182.	4.4	31
82	Genetic Variants in <i>HSD17B3</i> , <i>SMAD3</i> , and <i>IPO11</i> Impact Circulating Lipids in Response to Fenofibrate in Individuals With Type 2 Diabetes. <i>Clinical Pharmacology and Therapeutics</i> , 2018, 103, 712-721.	4.7	30
83	Insulin-like growth factor-1 and neurotrophin-3 gene therapy prevents motor decline in an X-linked adrenoleukodystrophy mouse model. <i>Annals of Neurology</i> , 2009, 66, 117-122.	5.3	28
84	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	12.8	28
85	The absence of ABCD2 sensitizes mice to disruptions in lipid metabolism by dietary erucic acid. <i>Journal of Lipid Research</i> , 2012, 53, 1071-1079.	4.2	27
86	Activation of sirtuin 1 as therapy for the peroxisomal disease adrenoleukodystrophy. <i>Cell Death and Differentiation</i> , 2015, 22, 1742-1753.	11.2	27
87	Targeted activation of <i>CREB</i> in reactive astrocytes is neuroprotective in focal acute cortical injury. <i>Glia</i> , 2016, 64, 853-874.	4.9	27
88	Uniparental disomy of chromosome 16 unmasks recessive mutations of <i>FA2H/SPG35</i> in 4 families. <i>Neurology</i> , 2016, 87, 186-191.	1.1	27
89	Ceramide signalling in inherited and multifactorial brain metabolic diseases. <i>Neurobiology of Disease</i> , 2020, 143, 105014.	4.4	27
90	Complete loss of KCNA1 activity causes neonatal epileptic encephalopathy and dyskinesia. <i>Journal of Medical Genetics</i> , 2020, 57, 132-137.	3.2	26

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91	Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. <i>American Journal of Human Genetics</i> , 2021, 108, 2195-2204.	6.2	26
92	Characterization of the Adrenoleukodystrophy-Related (ALDR, ABCD2) Gene Promoter: Inductibility by Retinoic Acid and Forskolin. <i>Genomics</i> , 2000, 70, 131-139.	2.9	25
93	<i>HNRNP1</i> -related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. <i>Clinical Genetics</i> , 2020, 98, 91-98.	2.0	25
94	<i>De novo</i> loss of function mutations in <i>KIAA2022</i> are associated with epilepsy and neurodevelopmental delay in females. <i>Clinical Genetics</i> , 2017, 91, 756-763.	2.0	24
95	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. <i>Acta Neuropathologica</i> , 2020, 140, 971-975.	7.7	24
96	POLR3A variants with striatal involvement and extrapyramidal movement disorder. <i>Neurogenetics</i> , 2020, 21, 121-133.	1.4	24
97	ATP and noradrenaline activate CREB in astrocytes via noncanonical Ca^{2+} and cyclic AMP independent pathways. <i>Glia</i> , 2012, 60, 1330-1344.	4.9	23
98	Case Report: Benign Infantile Seizures Temporally Associated With COVID-19. <i>Frontiers in Pediatrics</i> , 2020, 8, 507.	1.9	23
99	Epigenomic signature of adrenoleukodystrophy predicts compromised oligodendrocyte differentiation. <i>Brain Pathology</i> , 2018, 28, 902-919.	4.1	21
100	From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemic-Associated Pernio. <i>Journal of Investigative Dermatology</i> , 2021, 141, 2791-2796.	0.7	21
101	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. <i>Journal of Experimental Medicine</i> , 2022, 219, .	8.5	21
102	The peroxisomal fatty acid transporter ABCD1/PMP-4 is required in the <i>C. elegans</i> hypodermis for axonal maintenance: A worm model for adrenoleukodystrophy. <i>Free Radical Biology and Medicine</i> , 2020, 152, 797-809.	2.9	19
103	Biallelic <i>PI4KA</i> variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 2659-2669.	7.6	19
104	JNK/ERK/FAK Mediate Promigratory Actions of Basic Fibroblast Growth Factor in Astrocytes via CCL2 and COX2. <i>NeuroSignals</i> , 2012, 20, 86-102.	0.9	17
105	A deep intronic splice variant advises reexamination of presumably dominant SPG7 Cases. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 105-111.	3.7	17
106	PDR-1/hParkin negatively regulates the phagocytosis of apoptotic cell corpses in <i>Caenorhabditis elegans</i> . <i>Cell Death and Disease</i> , 2014, 5, e1120-e1120.	6.3	16
107	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021, 23, 888-899.	2.4	16
108	Histone Deacetylase Inhibitor Upregulates Peroxisomal Fatty Acid Oxidation and Inhibits Apoptotic Cell Death in <i>Abcd1</i> -Deficient Glial Cells. <i>PLoS ONE</i> , 2013, 8, e70712.	2.5	15

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109	Oxidative stress and mitochondrial dynamics malfunction are linked in <sc>P</sc>elizaesus<sc>M</sc>erzbacher disease. <i>Brain Pathology</i> , 2018, 28, 611-630.	4.1	15
110	Truncating variants in <i>UBAP1</i> associated with childhood-onset nonsyndromic hereditary spastic paraplegia. <i>Human Mutation</i> , 2020, 41, 632-640.	2.5	15
111	Expanding the clinical and genetic spectrum of PCYT2-related disorders. <i>Brain</i> , 2020, 143, e76-e76.	7.6	14
112	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1205-1215.	2.8	14
113	Lipid alterations in human frontal cortex in ALS&FTLD&TDP43 proteinopathy spectrum are partly related to peroxisome impairment. <i>Neuropathology and Applied Neurobiology</i> , 2021, 47, 544-563.	3.2	14
114	Novel Therapeutic Targets and Drug Candidates for Modifying Disease Progression in Adrenoleukodystrophy. <i>Endocrine Development</i> , 2016, 30, 147-160.	1.3	13
115	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. <i>Neurology</i> , 2022, , 10.1212/WNL.000000000013278.	1.1	13
116	The Value of Mouse Models of Rare Diseases: A Spanish Experience. <i>Frontiers in Genetics</i> , 2020, 11, 583932.	2.3	12
117	High-dose biotin restores redox balance, energy and lipid homeostasis, and axonal health in a model of adrenoleukodystrophy. <i>Brain Pathology</i> , 2020, 30, 945-963.	4.1	11
118	Epigenetic profiling linked to multisystem inflammatory syndrome in children (MIS-C): A multicenter, retrospective study. <i>EClinicalMedicine</i> , 2022, 50, 101515.	7.1	11
119	Sequencing analysis of a 40&2 kb fragment of yeast chromosome X reveals 19 open reading frames including URA2 (5&2 end), TRK1, PBS2, SPT10, GCD14, RPE1, PHO86, NCA3, ASF1, CCT7, GZF3, two tRNA genes, 1.7 three remnant delta elements and a Ty4 transposon. <i>Yeast</i> , 1996, 12, 1471-1474.		9
120	A novel mutation in the <i>GFAP</i> gene expands the phenotype of Alexander disease. <i>Journal of Medical Genetics</i> , 2019, 56, 846-849.	3.2	9
121	A view on clinical genetics and genomics in Spain: of challenges and opportunities. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 376-391.	1.2	8
122	Phenotypic correlations in a large single-center cohort of patients with BSCL2 nerve disorders: a clinical, neurophysiological and muscle magnetic resonance imaging study. <i>European Journal of Neurology</i> , 2020, 27, 1364-1373.	3.3	8
123	Inhibition of transcription-regulating properties of nonstructural protein 1 (NS1) of parvovirus minute virus of mice by a dominant-negative mutant form of NS1. <i>Journal of General Virology</i> , 2001, 82, 1929-1934.	2.9	8
124	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. <i>Genes</i> , 2021, 12, 1590.	2.4	8
125	Invariant NKT cells in adrenoleukodystrophy patients and mice. <i>Journal of Neuroimmunology</i> , 2010, 229, 204-211.	2.3	7
126	Phylogenomic Evidence for a Myxococcal Contribution to the Mitochondrial Fatty Acid Beta-Oxidation. <i>PLoS ONE</i> , 2011, 6, e21989.	2.5	7

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127	ABCD2 Alters Peroxisome Proliferator-Activated Receptor Signaling In Vitro, but Does Not Impair Responses to Fenofibrate Therapy in a Mouse Model of Diet-Induced Obesity. <i>Molecular Pharmacology</i> , 2014, 86, 505-513.	2.3	7
128	Modulation of mitochondrial and inflammatory homeostasis through RIP140 is neuroprotective in an adrenoleukodystrophy mouse model. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	3.2	6
129	SIRT2 in age-related neurodegenerative disorders. <i>Aging</i> , 2018, 10, 295-296.	3.1	6
130	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 529-542.	2.8	6
131	Cyclophilin D as a potential target for antioxidants in neurodegeneration: the X-ALD case. <i>Biological Chemistry</i> , 2013, 394, 621-629.	2.5	5
132	Low Lymphocytes and IFN-Neutralizing Autoantibodies as Biomarkers of COVID-19 Mortality. <i>Journal of Clinical Immunology</i> , 2022, 42, 738-741.	3.8	5
133	Epigenetic activation of antiviral sensors and effectors of interferon response pathways during SARS-CoV-2 infection. <i>Biomedicine and Pharmacotherapy</i> , 2022, 153, 113396.	5.6	5
134	Sequence and Analysis of a 36-kb Fragment from the Right Arm of Yeast Chromosome XV Reveals 19 Open Reading Frames Including SNF2 (5' end), CPA1, SLY41, a Putative Transport ATPase, a Putative Ribosomal Protein and an SNF2 Homologue. , 1997, 13, 479-482.		3
135	Foreword. <i>Brain Pathology</i> , 2010, 20, 815-816.	4.1	3
136	Evaluation of afferent pain pathways in adrenomyeloneuropathic patients. <i>Clinical Neurophysiology</i> , 2018, 129, 507-515.	1.5	3
137	A novel hypomorphic splice variant in EIF2B5 gene is associated with mild ovarioleukodystrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1574-1579.	3.7	3
138	Activating cannabinoid receptor 2 preserves axonal health through GSK-3 ^β /NRF2 axis in adrenoleukodystrophy. <i>Acta Neuropathologica</i> , 2022, 144, 241-258.	7.7	2
139	The ABCD Subfamily: Peroxisomal Transporters in Health and Disease. , 2011, , 347-373.		0
140	Reply to: "Mitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletion". <i>Movement Disorders</i> , 2019, 34, 1932-1933.	3.9	0
141	Anti-oxidant MitoQ rescue of AWB chemosensory neuron impairment in a model of X-linked Adrenoleukodystrophy. <i>MicroPublication Biology</i> , 2021, 2021, .	0.1	0