

# Aurora Pujol

## List of Publications by Citations

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135  
papers

10,743  
citations

42  
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144  
ext. papers

13,797  
ext. citations

9.4  
avg, IF

5.17  
L-index

#	Paper	IF	Citations
135	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , <b>2016</b> , 12, 1-222	10.2	3838
134	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. <i>Science</i> , <b>2020</b> , 370,	33.3	1090
133	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. <i>Science</i> , <b>2020</b> , 370,	33.3	994
132	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: role in diagnosis and clinical correlations. <i>Human Mutation</i> , <b>2001</b> , 18, 499-515	4.7	210
131	Pharmacological Inhibition of poly(ADP-ribose) polymerases improves fitness and mitochondrial function in skeletal muscle. <i>Cell Metabolism</i> , <b>2014</b> , 19, 1034-41	24.6	175
130	Early oxidative damage underlying neurodegeneration in X-adrenoleukodystrophy. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, 1762-73	5.6	158
129	Late onset neurological phenotype of the X-ALD gene inactivation in mice: a mouse model for adrenomyeloneuropathy. <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 499-505	5.6	151
128	Functional overlap between ABCD1 (ALD) and ABCD2 (ALDR) transporters: a therapeutic target for X-adrenoleukodystrophy. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 2997-3006	5.6	149
127	The evolutionary origin of peroxisomes: an ER-peroxisome connection. <i>Molecular Biology and Evolution</i> , <b>2006</b> , 23, 838-45	8.3	138
126	A Global Effort to Define the Human Genetics of Protective Immunity to SARS-CoV-2 Infection. <i>Cell</i> , <b>2020</b> , 181, 1194-1199	56.2	113
125	Antioxidants halt axonal degeneration in a mouse model of X-adrenoleukodystrophy. <i>Annals of Neurology</i> , <b>2011</b> , 70, 84-92	9.4	107
124	Methylene blue upregulates Nrf2/ARE genes and prevents tau-related neurotoxicity. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 3716-32	5.6	96
123	Pathomechanisms underlying X-adrenoleukodystrophy: a three-hit hypothesis. <i>Brain Pathology</i> , <b>2010</b> , 20, 838-44	6	94
122	General aspects and neuropathology of X-linked adrenoleukodystrophy. <i>Brain Pathology</i> , <b>2010</b> , 20, 817-30		92
121	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> , <b>2021</b> , 6,	28	91
120	Neuroinflammatory signals in Alzheimer disease and APP/PS1 transgenic mice: correlations with plaques, tangles, and oligomeric species. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2015</b> , 74, 319-44	3.1	86
119	Impaired mitochondrial oxidative phosphorylation in the peroxisomal disease X-linked adrenoleukodystrophy. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 3296-305	5.6	83

118	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> , <b>2013</b> , 72, 524-39	3.1	83
117	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 768-788	11	81
116	PeroxisomeDB 2.0: an integrative view of the global peroxisomal metabolome. <i>Nucleic Acids Research</i> , <b>2010</b> , 38, D800-5	20.1	81
115	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> , <b>2005</b> , 14, 3565-77	5.6	81
114	Deregulation of purine metabolism in Alzheimer's disease. <i>Neurobiology of Aging</i> , <b>2015</b> , 36, 68-80	5.6	78
113	Valproic acid induces antioxidant effects in X-linked adrenoleukodystrophy. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 2005-14	5.6	77
112	Current and future pharmacological treatment strategies in X-linked adrenoleukodystrophy. <i>Brain Pathology</i> , <b>2010</b> , 20, 845-56	6	75
111	A key role for the peroxisomal ABCD2 transporter in fatty acid homeostasis. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , <b>2009</b> , 296, E211-21	6	72
110	Amyloid generation and dysfunctional immunoproteasome activation with disease progression in animal model of familial Alzheimer's disease. <i>Brain Pathology</i> , <b>2012</b> , 22, 636-53	6	71
109	Oxidative stress underlying axonal degeneration in adrenoleukodystrophy: a paradigm for multifactorial neurodegenerative diseases?. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2012</b> , 1822, 1475-88	6.9	71
108	Oxidative damage compromises energy metabolism in the axonal degeneration mouse model of X-adrenoleukodystrophy. <i>Antioxidants and Redox Signaling</i> , <b>2011</b> , 15, 2095-107	8.4	68
107	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. <i>Science Immunology</i> , <b>2021</b> , 6,	28	67
106	Neurochemistry and the non-motor aspects of PD. <i>Neurobiology of Disease</i> , <b>2012</b> , 46, 508-26	7.5	66
105	Complete nucleotide sequence of <i>Saccharomyces cerevisiae</i> chromosome X.. <i>EMBO Journal</i> , <b>1996</b> , 15, 2031-2049	13	65
104	PeroxisomeDB: a database for the peroxisomal proteome, functional genomics and disease. <i>Nucleic Acids Research</i> , <b>2007</b> , 35, D815-22	20.1	63
103	Oxidative stress modulates mitochondrial failure and cyclophilin D function in X-linked adrenoleukodystrophy. <i>Brain</i> , <b>2012</b> , 135, 3584-98	11.2	62
102	Pioglitazone halts axonal degeneration in a mouse model of X-linked adrenoleukodystrophy. <i>Brain</i> , <b>2013</b> , 136, 2432-43	11.2	57
101	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> , <b>2015</b> , 114, 388-96	3.7	56

100	Functional genomic analysis unravels a metabolic-inflammatory interplay in adrenoleukodystrophy. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 1062-77	5.6	55
99	The nucleotide sequence of <i>Saccharomyces cerevisiae</i> chromosome XV. <i>Nature</i> , <b>1997</b> , 387, 98-102	50.4	51
98	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> , <b>1999</b> , 73, 3877-85	6.6	49
97	Bezafibrate administration improves behavioral deficits and tau pathology in P301S mice. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 5091-105	5.6	47
96	Mouse liver PMP70 and ALDP: homomeric interactions prevail in vivo. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2004</b> , 1689, 235-43	6.9	45
95	SARS-CoV-2-related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. <i>Journal of Experimental Medicine</i> , <b>2021</b> , 218,	16.6	45
94	Mitochondrial dysfunction in central nervous system white matter disorders. <i>Glia</i> , <b>2014</b> , 62, 1878-94	9	43
93	PGC-1 $\beta$ overexpression exacerbates $\beta$ amyloid and tau deposition in a transgenic mouse model of Alzheimer's disease. <i>FASEB Journal</i> , <b>2014</b> , 28, 1745-55	0.9	41
92	Isolation of a fully infectious variant of parvovirus H-1 supplanting the standard strain in human cells. <i>Journal of Virology</i> , <b>1995</b> , 69, 4538-43	6.6	40
91	Oxidative stress, mitochondrial and proteostasis malfunction in adrenoleukodystrophy: A paradigm for axonal degeneration. <i>Free Radical Biology and Medicine</i> , <b>2015</b> , 88, 18-29	7.8	39
90	Steroid hormones control circadian Elov13 expression in mouse liver. <i>Endocrinology</i> , <b>2008</b> , 149, 3158-66	4.8	39
89	Inhibition of parvovirus minute virus of mice replication by a peptide involved in the oligomerization of nonstructural protein NS1. <i>Journal of Virology</i> , <b>1997</b> , 71, 7393-403	6.6	37
88	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. <i>Journal of Clinical Investigation</i> , <b>2019</b> , 129, 1240-1256	15.9	37
87	Mitochondrial dysfunction and oxidative damage cooperatively fuel axonal degeneration in X-linked adrenoleukodystrophy. <i>Biochimie</i> , <b>2014</b> , 98, 143-9	4.6	36
86	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> , <b>2015</b> , 1852, 925-36	6.9	36
85	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> , <b>2021</b> , 41, 914-922	5.7	35
84	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. <i>Brain</i> , <b>2019</b> , 142, 1561-1572	11.2	34
83	Glutathione imbalance in patients with X-linked adrenoleukodystrophy. <i>Molecular Genetics and Metabolism</i> , <b>2013</b> , 109, 366-70	3.7	34

82	Bezafibrate lowers very long-chain fatty acids in X-linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation. <i>Journal of Inherited Metabolic Disease</i> , <b>2012</b> , 35, 1137-45	5.4	33
81	Epigenome-wide association study of COVID-19 severity with respiratory failure. <i>EBioMedicine</i> , <b>2021</b> , 66, 103339	8.8	33
80	Oxidative stress regulates the ubiquitin-proteasome system and immunoproteasome functioning in a mouse model of X-adrenoleukodystrophy. <i>Brain</i> , <b>2013</b> , 136, 891-904	11.2	32
79	The role of peroxisomal ABC transporters in the mouse adrenal gland: the loss of Abcd2 (ALDR), Not Abcd1 (ALD), causes oxidative damage. <i>Laboratory Investigation</i> , <b>2007</b> , 87, 261-72	5.9	32
78	Tauroursodeoxycholic bile acid arrests axonal degeneration by inhibiting the unfolded protein response in X-linked adrenoleukodystrophy. <i>Acta Neuropathologica</i> , <b>2017</b> , 133, 283-301	14.3	31
77	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 965-976	11	31
76	Autophagy induction halts axonal degeneration in a mouse model of X-adrenoleukodystrophy. <i>Acta Neuropathologica</i> , <b>2015</b> , 129, 399-415	14.3	31
75	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> , <b>2018</b> , 102, 744-759	11	30
74	ABCD2 is abundant in adipose tissue and opposes the accumulation of dietary erucic acid (C22:1) in fat. <i>Journal of Lipid Research</i> , <b>2010</b> , 51, 162-8	6.3	28
73	Loss of SIRT2 leads to axonal degeneration and locomotor disability associated with redox and energy imbalance. <i>Aging Cell</i> , <b>2017</b> , 16, 1404-1413	9.9	27
72	Staging anti-inflammatory therapy in Alzheimer's disease. <i>Frontiers in Aging Neuroscience</i> , <b>2010</b> , 2, 142	5.3	27
71	Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 105-116	11	26
70	Aberrant regulation of the GSK-3 $\beta$ /NRF2 axis unveils a novel therapy for adrenoleukodystrophy. <i>EMBO Molecular Medicine</i> , <b>2018</b> , 10,	12	26
69	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. <i>Movement Disorders</i> , <b>2019</b> , 34, 1547-1561	7	26
68	Altered glycolipid and glycerophospholipid signaling drive inflammatory cascades in adrenomyeloneuropathy. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6861-76	5.6	25
67	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> , <b>2009</b> , 66, 117-22	9.4	25
66	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. <i>EBioMedicine</i> , <b>2021</b> , 65, 103246	8.8	25
65	Activation of sirtuin 1 as therapy for the peroxisomal disease adrenoleukodystrophy. <i>Cell Death and Differentiation</i> , <b>2015</b> , 22, 1742-53	12.7	23

64	Characterization of the adrenoleukodystrophy-related (ALDR, ABCD2) gene promoter: inductibility by retinoic acid and forskolin. <i>Genomics</i> , <b>2000</b> , 70, 131-9	4.3	23
63	Human genetic and immunological determinants of critical COVID-19 pneumonia.. <i>Nature</i> , <b>2022</b> ,	50.4	23
62	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H/SPG35 in 4 families. <i>Neurology</i> , <b>2016</b> , 87, 186-91	6.5	21
61	Targeted activation of CREB in reactive astrocytes is neuroprotective in focal acute cortical injury. <i>Glia</i> , <b>2016</b> , 64, 853-74	9	21
60	Genetic Variants in HSD17B3, SMAD3, and IPO11 Impact Circulating Lipids in Response to Fenofibrate in Individuals With Type 2 Diabetes. <i>Clinical Pharmacology and Therapeutics</i> , <b>2018</b> , 103, 712-721	6.1	20
59	Inhibition of Gsk3b Reduces Nfkb1 Signaling and Rescues Synaptic Activity to Improve the Rett Syndrome Phenotype in Mecp2-Knockout Mice. <i>Cell Reports</i> , <b>2018</b> , 23, 1665-1677	10.6	20
58	Biomarker Identification, Safety, and Efficacy of High-Dose Antioxidants for Adrenomyeloneuropathy: a Phase II Pilot Study. <i>Neurotherapeutics</i> , <b>2019</b> , 16, 1167-1182	6.4	19
57	Improvement of the Rett syndrome phenotype in a MeCP2 mouse model upon treatment with levodopa and a dopa-decarboxylase inhibitor. <i>Neuropsychopharmacology</i> , <b>2014</b> , 39, 2846-56	8.7	17
56	Clinical presentation and proteomic signature of patients with TANGO2 mutations. <i>Journal of Inherited Metabolic Disease</i> , <b>2020</b> , 43, 297-308	5.4	17
55	Complete loss of KCNA1 activity causes neonatal epileptic encephalopathy and dyskinesia. <i>Journal of Medical Genetics</i> , <b>2020</b> , 57, 132-137	5.8	16
54	Epigenomic signature of adrenoleukodystrophy predicts compromised oligodendrocyte differentiation. <i>Brain Pathology</i> , <b>2018</b> , 28, 902-919	6	15
53	The absence of ABCD2 sensitizes mice to disruptions in lipid metabolism by dietary erucic acid. <i>Journal of Lipid Research</i> , <b>2012</b> , 53, 1071-9	6.3	15
52	Histone deacetylase inhibitor upregulates peroxisomal fatty acid oxidation and inhibits apoptotic cell death in abcd1-deficient glial cells. <i>PLoS ONE</i> , <b>2013</b> , 8, e70712	3.7	14
51	Case Report: Benign Infantile Seizures Temporally Associated With COVID-19. <i>Frontiers in Pediatrics</i> , <b>2020</b> , 8, 507	3.4	14
50	POLR3A variants with striatal involvement and extrapyramidal movement disorder. <i>Neurogenetics</i> , <b>2020</b> , 21, 121-133	3	13
49	De novo loss of function mutations in KIAA2022 are associated with epilepsy and neurodevelopmental delay in females. <i>Clinical Genetics</i> , <b>2017</b> , 91, 756-763	4	13
48	JNK/ERK/FAK mediate promigratory actions of basic fibroblast growth factor in astrocytes via CCL2 and COX2. <i>NeuroSignals</i> , <b>2012</b> , 20, 86-102	1.9	13
47	A deep intronic splice variant advises reexamination of presumably dominant SPG7 Cases. <i>Annals of Clinical and Translational Neurology</i> , <b>2020</b> , 7, 105-111	5.3	13

46	PDR-1/hParkin negatively regulates the phagocytosis of apoptotic cell corpses in <i>Caenorhabditis elegans</i> . <i>Cell Death and Disease</i> , <b>2014</b> , 5, e1120	9.8	12
45	ATP and noradrenaline activate CREB in astrocytes via noncanonical Ca(2+) and cyclic AMP independent pathways. <i>Glia</i> , <b>2012</b> , 60, 1330-44	9	12
44	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. <i>Acta Neuropathologica</i> , <b>2020</b> , 140, 971-975	14.3	11
43	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> , <b>2020</b> , 152, 797-809	7.8	9
42	HNRNPH1-related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. <i>Clinical Genetics</i> , <b>2020</b> , 98, 91-98	4	9
41	Oxidative stress and mitochondrial dynamics malfunction are linked in Pelizaeus-Merzbacher disease. <i>Brain Pathology</i> , <b>2018</b> , 28, 611-630	6	9
40	Novel Therapeutic Targets and Drug Candidates for Modifying Disease Progression in Adrenoleukodystrophy. <i>Endocrine Development</i> , <b>2016</b> , 30, 147-60		9
39	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. <i>Journal of Molecular Diagnostics</i> , <b>2020</b> , 22, 1205-1215	5.1	8
38	Truncating variants in UBAP1 associated with childhood-onset nonsyndromic hereditary spastic paraplegia. <i>Human Mutation</i> , <b>2020</b> , 41, 632-640	4.7	8
37	CSVS, a crowdsourcing database of the Spanish population genetic variability. <i>Nucleic Acids Research</i> , <b>2021</b> , 49, D1130-D1137	20.1	8
36	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. <i>Nature Immunology</i> , <b>2021</b> ,	19.1	7
35	Ceramide signalling in inherited and multifactorial brain metabolic diseases. <i>Neurobiology of Disease</i> , <b>2020</b> , 143, 105014	7.5	7
34	Expanding the clinical and genetic spectrum of PCYT2-related disorders. <i>Brain</i> , <b>2020</b> , 143, e76	11.2	7
33	Lipid alterations in human frontal cortex in ALS-FTLD-TDP43 proteinopathy spectrum are partly related to peroxisome impairment. <i>Neuropathology and Applied Neurobiology</i> , <b>2021</b> , 47, 544-563	5.2	7
32	Invariant NKT cells in adrenoleukodystrophy patients and mice. <i>Journal of Neuroimmunology</i> , <b>2010</b> , 229, 204-11	3.5	6
31	Sequencing analysis of a 402 kb fragment of yeast chromosome X reveals 19 open reading frames including URA2 (5' end), TRK1, PBS2, SPT10, GCD14, RPE1, PHO86, NCA3, ASF1, CCT7, GZF3, two tRNA genes, three remnant delta elements and a Ty4 transposon. <i>Yeast</i> , <b>1996</b> , 12, 1471-1474	3.4	6
30	The Value of Mouse Models of Rare Diseases: A Spanish Experience. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 583925	3.2	6
29	From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemic-Associated Pernio. <i>Journal of Investigative Dermatology</i> , <b>2021</b> , 141, 2791-2796	4.3	6

28	Studying severe long COVID to understand post-infectious disorders beyond COVID-19.. <i>Nature Medicine</i> , <b>2022</b> ,	50.5	6
27	ABCD2 alters peroxisome proliferator-activated receptor $\beta$ signaling in vitro, but does not impair responses to fenofibrate therapy in a mouse model of diet-induced obesity. <i>Molecular Pharmacology</i> , <b>2014</b> , 86, 505-13	4.3	5
26	Phylogenomic evidence for a myxococcal contribution to the mitochondrial fatty acid beta-oxidation. <i>PLoS ONE</i> , <b>2011</b> , 6, e21989	3.7	5
25	SIRT2 in age-related neurodegenerative disorders. <i>Aging</i> , <b>2018</b> , 10, 295-296	5.6	5
24	A novel mutation in the gene expands the phenotype of Alexander disease. <i>Journal of Medical Genetics</i> , <b>2019</b> , 56, 846-849	5.8	4
23	High-dose biotin restores redox balance, energy and lipid homeostasis, and axonal health in a model of adrenoleukodystrophy. <i>Brain Pathology</i> , <b>2020</b> , 30, 945-963	6	4
22	Cyclophilin D as a potential target for antioxidants in neurodegeneration: the X-ALD case. <i>Biological Chemistry</i> , <b>2013</b> , 394, 621-9	4.5	4
21	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> , <b>2001</b> , 82, 1929-1934	4.9	4
20	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. <i>Nature Communications</i> , <b>2021</b> , 12, 2558	17.4	4
19	Accelerated biological aging in COVID-19 patients.. <i>Nature Communications</i> , <b>2022</b> , 13, 2135	17.4	4
18	Evaluation of afferent pain pathways in adrenomyeloneuropathic patients. <i>Clinical Neurophysiology</i> , <b>2018</b> , 129, 507-515	4.3	3
17	A view on clinical genetics and genomics in Spain: of challenges and opportunities. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2016</b> , 4, 376-91	2.3	3
16	Schilder's disease a heterogeneous group of disorders known as X-linked adrenoleukodystrophy. Foreword. <i>Brain Pathology</i> , <b>2010</b> , 20, 815-6	6	3
15	Sequence and analysis of a 36.2 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. <i>Yeast</i> , <b>1997</b> , 13, 479-82	3.4	3
14	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> , <b>2021</b> , 108, 2195-2204	11	3
13	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> , <b>2022</b> , 119, e2200413119 <sup>11.5</sup>		3
12	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization.. <i>Neurology</i> , <b>2022</b> ,	6.5	2
11	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males		2



10	A novel hypomorphic splice variant in EIF2B5 gene is associated with mild ovarioleukodystrophy. <i>Annals of Clinical and Translational Neurology</i> , <b>2020</b> , 7, 1574-1579	5.3	2
9	Modulation of mitochondrial and inflammatory homeostasis through RIP140 is neuroprotective in an adrenoleukodystrophy mouse model. <i>Neuropathology and Applied Neurobiology</i> , <b>2021</b> ,	5.2	2
8	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. <i>Brain</i> , <b>2021</b> , 144, 2659-2669	11.2	2
7	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in males		1
6	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases.. <i>Journal of Molecular Diagnostics</i> , <b>2022</b> , 24, 529-542	5.1	1
5	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> , <b>2020</b> , 27, 1364-1373	6	0
4	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 888-899	8.1	0
3	Low Lymphocytes and IFN-Neutralizing Autoantibodies as Biomarkers of COVID-19 Mortality.. <i>Journal of Clinical Immunology</i> , <b>2022</b> , 1	5.7	0
2	The ABCD Subfamily: Peroxisomal Transporters in Health and Disease <b>2011</b> , 347-373		
1	Reply to: "Mitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletion". <i>Movement Disorders</i> , <b>2019</b> , 34, 1932-1933	7	