Adolfo LÃ³pez de Munain

List of Publications by Year
in descending order

198 papers	15,227 citations	44069 48 h-index	21540 114 g-index
203 all docs	203 docs citations	203 times ranked	23178 citing authors

#	Article	IF	Citations
1	The parkinsonian LRRK2 R1441G mutation shows macroautophagy-mitophagy dysregulation concomitant with endoplasmic reticulum stress. Cell Biology and Toxicology, 2022, 38, 889-911.	5.3	9
2	White matter integrity changes and neurocognitive functioning in adult-late onset DM1: a follow-up DTI study. Scientific Reports, 2022, 12, 3988.	3.3	6
3	Targeting the Ubiquitin-Proteasome System in Limb-Girdle Muscular Dystrophy With CAPN3 Mutations. Frontiers in Cell and Developmental Biology, 2022, 10, 822563.	3.7	4
4	Targeting Myotonic Dystrophy Type 1 with Metformin. International Journal of Molecular Sciences, 2022, 23, 2901.	4.1	13
5	Smoking is associated with age at disease onset in Parkinson's disease. Parkinsonism and Related Disorders, 2022, 97, 79-83.	2.2	2
6	New insights into the genetic etiology of Alzheimerâ€™s disease and related dementias. Nature Genetics, 2022, 54, 412-436.	21.4	700
7	A Ca²⁺-Dependent Mechanism Boosting Glycolysis and OXPHOS by Activating Aralar-Malate-Aspartate Shuttle, upon Neuronal Stimulation. Journal of Neuroscience, 2022, 42, 3879-3895.	3.6	18
8	Generation of Calpain-3 knock-out porcine embryos by CRISPR-Cas9 electroporation and intracytoplasmic microinjection of oocytes before insemination. Theriogenology, 2022, 186, 175-184.	2.1	3
9	A validated WAIS-IV short-form to estimate intellectual functioning in myotonic dystrophy type 1. Neuromuscular Disorders, 2022, 32, 749-753.	0.6	3
10	Amyotrophic lateral sclerosis (ALS), cancer, autoimmunity and metabolic disorders: An unsolved tantalizing challenge. British Journal of Pharmacology, 2021, 178, 1269-1278.	5.4	9
11	Clinical and preclinical evidence of somatosensory involvement in amyotrophic lateral sclerosis. British Journal of Pharmacology, 2021, 178, 1257-1268.	5.4	15
12	Clinical characteristics and outcomes of thymomaâ€associated myasthenia gravis. European Journal of Neurology, 2021, 28, 2083-2091.	3.3	39
13	Discovery of a novel family of FKBP12 â€œreshapersâ€•and their use as calcium modulators in skeletal muscle under nitro-oxidative stress. European Journal of Medicinal Chemistry, 2021, 213, 113160.	5.5	5
14	Transcriptional signatures of synaptic vesicle genes define myotonic dystrophy type I neurodegeneration. Neuropathology and Applied Neurobiology, 2021, 47, 1092-1108.	3.2	14
15	Common variants in Alzheimerâ€™s disease and risk stratification by polygenic risk scores. Nature Communications, 2021, 12, 3417.	12.8	140
16	Management of an outbreak of botulism with benign clinical presentation. Journal of Clinical Neuroscience, 2021, 88, 159-162.	1.5	1
17	The Skeletal Muscle Emerges as a New Disease Target in Amyotrophic Lateral Sclerosis. Journal of Personalized Medicine, 2021, 11, 671.	2.5	20
18	Allosteric Modulation of GSK-3Î² as a New Therapeutic Approach in Limb Girdle Muscular Dystrophy R1 Calpain 3-Related. International Journal of Molecular Sciences, 2021, 22, 7367.	4.1	5

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19	Description of Two Families with New Mutations in Familial Cerebral Cavernous Malformations Genes. Journal of Stroke and Cerebrovascular Diseases, 2021, 30, 106130.	1.6	0
20	Preclinical characterization of antagomiR-218 as a potential treatment for myotonic dystrophy. Molecular Therapy - Nucleic Acids, 2021, 26, 174-191.	5.1	9
21	Ageâ€related cognitive decline in myotonic dystrophy type 1: An 11â€year longitudinal followâ€up study. Journal of Neuropsychology, 2020, 14, 121-134.	1.4	16
22	A deep intronic splice variant advises reexamination of presumably dominant SPG7 Cases. Annals of Clinical and Translational Neurology, 2020, 7, 105-111.	3.7	17
23	Leukocyte telomere length in patients with myotonic dystrophy type I: a pilot study. Annals of Clinical and Translational Neurology, 2020, 7, 126-131.	3.7	4
24	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
25	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
26	Neurodegeneration trajectory in pediatric and adult/late DM1: A followâ€up MRI study across a decade. Annals of Clinical and Translational Neurology, 2020, 7, 1802-1815.	3.7	15
27	Targeted Next-Generation Sequencing in a Large Cohort of Genetically Undiagnosed Patients with Neuromuscular Disorders in Spain. Genes, 2020, 11, 539.	2.4	25
28	Frizzled related protein deficiency impairs muscle strength, gait and calpain 3 levels. Orphanet Journal of Rare Diseases, 2020, 15, 119.	2.7	5
29	Toxicity of Necrostatin-1 in Parkinsonâ€™s Disease Models. Antioxidants, 2020, 9, 524.	5.1	13
30	COL4A1 Mutation as a Cause of Familial Recurrent Intracerebral Hemorrhage. Journal of Stroke and Cerebrovascular Diseases, 2020, 29, 104652.	1.6	4
31	A comprehensive serum lipidome profiling of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 252-262.	1.7	20
32	Spanish Pompe registry: Baseline characteristics of first 49 patients with adult onset of Pompe disease. Medicina ClÃ¡nica (English Edition), 2020, 154, 80-85.	0.2	3
33	ALS-derived fibroblasts exhibit reduced proliferation rate, cytoplasmic TDP-43 aggregation and a higher susceptibility to DNA damage. Journal of Neurology, 2020, 267, 1291-1299.	3.6	20
34	Kynurenic Acid Levels are Increased in the CSF of Alzheimerâ€™s Disease Patients. Biomolecules, 2020, 10, 571.	4.0	37
35	Myotonic Dystrophy type 1 cells display impaired metabolism and mitochondrial dysfunction that are reversed by metformin. Aging, 2020, 12, 6260-6275.	3.1	28
36	Metabolic alterations in plasma from patients with familial and idiopathic Parkinsonâ€™s disease. Aging, 2020, 12, 16690-16708.	3.1	32

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37	Dermic-derived fibroblasts for the study of amyotrophic lateral sclerosis. Neural Regeneration Research, 2020, 15, 2043.	3.0	10
38	Impaired Mitophagy and Protein Acetylation Levels in Fibroblasts from Parkinsonâ€™s Disease Patients. Molecular Neurobiology, 2019, 56, 2466-2481.	4.0	50
39	RAB8, RAB10 and RILPL1 contribute to both LRRK2 kinaseâ€“mediated centrosomal cohesion and ciliogenesis deficits. Human Molecular Genetics, 2019, 28, 3552-3568.	2.9	72
40	Study of the effect of anti-rhGAA antibodies at low and intermediate titers in late onset Pompe patients treated with ERT. Molecular Genetics and Metabolism, 2019, 128, 129-136.	1.1	5
41	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. Movement Disorders, 2019, 34, 1547-1561.	3.9	44
42	Gut microbiome and serum metabolome analyses identify molecular biomarkers and altered glutamate metabolism in fibromyalgia. EBioMedicine, 2019, 46, 499-511.	6.1	128
43	Nonsense mutations in alpha-II spectrin in three families with juvenile onset hereditary motor neuropathy. Brain, 2019, 142, 2605-2616.	7.6	29
44	Identification of serum microRNAs as potential biomarkers in Pompe disease. Annals of Clinical and Translational Neurology, 2019, 6, 1214-1224.	3.7	19
45	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	3.9	47
46	Gene Correction of LGMD2A Patient-Specific iPSCs for the Development of Targeted Autologous Cell Therapy. Molecular Therapy, 2019, 27, 2147-2157.	8.2	36
47	Calcium Mechanisms in Limb-Girdle Muscular Dystrophy with CAPN3 Mutations. International Journal of Molecular Sciences, 2019, 20, 4548.	4.1	25
48	A nonsynonymous mutation in PLCG2 reduces the risk of Alzheimerâ€™s disease, dementia with Lewy bodies and frontotemporal dementia, and increases the likelihood of longevity. Acta Neuropathologica, 2019, 138, 237-250.	7.7	87
49	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	7.6	70
50	Isolation and characterization of myogenic precursor cells from human cremaster muscle. Scientific Reports, 2019, 9, 3454.	3.3	10
51	Regional brain atrophy in gray and white matter is associated with cognitive impairment in Myotonic Dystrophy type 1. NeuroImage: Clinical, 2019, 24, 102078.	2.7	24
52	Increased Muscleblind levels by chloroquine treatment improve myotonic dystrophy type 1 phenotypes in in vitro and in vivo models. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 25203-25213.	7.1	32
53	Reply to: â€œMitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletionâ€• Movement Disorders, 2019, 34, 1932-1933.	3.9	0
54	Longitudinal Neuropsychological Study of Presymptomatic c.709-1G>A Progranulin Mutation Carriers. Journal of the International Neuropsychological Society, 2019, 25, 39-47.	1.8	7

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55	Treatment challenges in and outside a network setting: Soft tissue sarcomas. European Journal of Surgical Oncology, 2019, 45, 31-39.	1.0	27
56	Epidemiology of rare cancers and inequalities in oncologic outcomes. European Journal of Surgical Oncology, 2019, 45, 3-11.	1.0	47
57	Treatment challenges in and outside a specialist network setting: Pancreatic neuroendocrine tumours. European Journal of Surgical Oncology, 2019, 45, 46-51.	1.0	3
58	Rare ovarian tumours: Epidemiology, treatment challenges in and outside a network setting. European Journal of Surgical Oncology, 2019, 45, 67-74.	1.0	22
59	Mesothelioma and thymic tumors: Treatment challenges in (outside) a network setting. European Journal of Surgical Oncology, 2019, 45, 75-80.	1.0	15
60	Testicular germ-cell tumours and penile squamous cell carcinoma: Appropriate management makes the difference. European Journal of Surgical Oncology, 2019, 45, 60-66.	1.0	4
61	Treatment challenges in and outside a network setting: Head and neck cancers. European Journal of Surgical Oncology, 2019, 45, 40-45.	1.0	27
62	T cells and immune functions of plasma extracellular vesicles are differentially modulated from adults to centenarians. Aging, 2019, 11, 10723-10741.	3.1	12
63	Amyotrophic lateral sclerosis: a complex syndrome that needs an integrated research approach. Neural Regeneration Research, 2019, 14, 193.	3.0	18
64	Global surveillance of trends in cancer survival 2000â€“14 (CONCORD-3): analysis of individual records for 37â€ˆ513â€ˆ025 patients diagnosed with one of 18 cancers from 322 population-based registries in 71 countries. Lancet, The, 2018, 391, 1023-1075.	13.7	3,228
65	The increasing importance of environmental conditions in amyotrophic lateral sclerosis. International Journal of Biometeorology, 2018, 62, 1361-1374.	3.0	41
66	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	10.2	97
67	Association of lifestyle, inflammatory factors, and dietary patterns with the risk of suffering a stroke: A caseâ€“control study. Nutritional Neuroscience, 2018, 21, 70-78.	3.1	9
68	iPS Cell Cultures from a Gerstmann-StrÃussler-Scheinker Patient with the Y218N PRNP Mutation Recapitulate tau Pathology. Molecular Neurobiology, 2018, 55, 3033-3048.	4.0	27
69	Blood Markers in Healthy-Aged Nonagenarians: A Combination of High Telomere Length and Low Amyloidβ Are Strongly Associated With Healthy Aging in the Oldest Old. Frontiers in Aging Neuroscience, 2018, 10, 380.	3.4	2
70	Social cognition in myotonic dystrophy type 1: Specific or secondary impairment?. PLoS ONE, 2018, 13, e0204227.	2.5	17
71	A new approach based on targeted pooled DNA sequencing identifies novel mutations in patients with Inherited Retinal Dystrophies. Scientific Reports, 2018, 8, 15457.	3.3	17
72	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for followâ€up. Muscle and Nerve, 2018, 58, 812-817.	2.2	8

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73	Acetylome in Human Fibroblasts From Parkinson's Disease Patients. Frontiers in Cellular Neuroscience, 2018, 12, 97.	3.7	15
74	Cancer phenotype in myotonic dystrophy patients: Results from a metaâ€analysis. Muscle and Nerve, 2018, 58, 517-522.	2.2	22
75	Parkinson disease-associated mutations in LRRK2 cause centrosomal defects via Rab8a phosphorylation. Molecular Neurodegeneration, 2018, 13, 3.	10.8	77
76	Clinical evidences supporting the Src/c-Abl pathway as potential therapeutic target in amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2018, 393, 80-82.	0.6	6
77	Retinoids and Amyotrophic Lateral Sclerosis. JAMA Neurology, 2018, 75, 1153.	9.0	0
78	The panniculus carnosus muscle: an evolutionary enigma at the intersection of distinct research fields. Journal of Anatomy, 2018, 233, 275-288.	1.5	71
79	High prevalence of mutations affecting the splicing process in a Spanish cohort with autosomal dominant retinitis pigmentosa. Scientific Reports, 2017, 7, 39652.	3.3	25
80	Incidence of varicella zoster virus infections of the central nervous system in the elderly: a large tertiary hospital-based series (2007â€“2014). Journal of NeuroVirology, 2017, 23, 451-459.	2.1	23
81	Nontraditional Lipid Variables Predict Recurrent Brain Ischemia in Embolic Stroke of Undetermined Source. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, 1670-1677.	1.6	13
82	Insights into the mechanisms of copper dyshomeostasis in amyotrophic lateral sclerosis. Expert Reviews in Molecular Medicine, 2017, 19, e7.	3.9	34
83	FRZB and melusin, overexpressed in LGMD2A, regulate integrin Î²1D isoform replacement altering myoblast fusion and the integrin-signalling pathway. Expert Reviews in Molecular Medicine, 2017, 19, e2.	3.9	10
84	Distribution and genotype-phenotype correlation of GDAP1 mutations in Spain. Scientific Reports, 2017, 7, 6677.	3.3	23
85	Copy number variation analysis increases the diagnostic yield in muscle diseases. Neurology: Genetics, 2017, 3, e204.	1.9	17
86	Dominant LGMD2A: alternative diagnosis or hidden digenism?. Brain, 2017, 140, e7-e7.	7.6	9
87	Incidence and survival time trends for Spanish children and adolescents with leukaemia from 1983 to 2007. Clinical and Translational Oncology, 2017, 19, 301-316.	2.4	11
88	The unexpected co-occurrence of GRN and MAPT p.A152T in Basque families: Clinical and pathological characteristics. PLoS ONE, 2017, 12, e0178093.	2.5	5
89	Progressive changes in non-coding RNA profile in leucocytes with age. Aging, 2017, 9, 1202-1218.	3.1	13
90	Editorial: Role of Stem Cells in Skeletal Muscle Development, Regeneration, Repair, Aging, and Disease. Frontiers in Aging Neuroscience, 2016, 8, 95.	3.4	3

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91	DAT imaging and clinical biomarkers in relatives at genetic risk for LRRK2â€‰R1441G Parkinson's disease. Movement Disorders, 2016, 31, 335-343.	3.9	33
92	ALS: A bucket of genes, environment, metabolism and unknown ingredients. Progress in Neurobiology, 2016, 142, 104-129.	5.7	158
93	Calpain 3 deficiency affects SERCA expression and function in the skeletal muscle. Expert Reviews in Molecular Medicine, 2016, 18, e7.	3.9	31
94	Identification and Characterization of the Dermal Panniculus Carnosus Muscle Stem Cells. Stem Cell Reports, 2016, 7, 411-424.	4.8	30
95	A Cost-Effective Mutation Screening Strategy for Inherited Retinal Dystrophies. Ophthalmic Research, 2016, 56, 123-131.	1.9	2
96	Natural history of <scp>LGMD</scp>2A for delineating outcome measures in clinical trials. Annals of Clinical and Translational Neurology, 2016, 3, 248-265.	3.7	52
97	Cancer risk in DM1 is sex-related and linked to miRNA-200/141 downregulation. Neurology, 2016, 87, 1250-1257.	1.1	48
98	Targeting TDP-43 phosphorylation by Casein Kinase-1Î´ inhibitors: a novel strategy for the treatment of frontotemporal dementia. Molecular Neurodegeneration, 2016, 11, 36.	10.8	55
99	SncRNA (microRNA & snoRNA) opposite expression pattern found in multiple sclerosis relapse and remission is sex dependent. Scientific Reports, 2016, 6, 20126.	3.3	38
100	Mutations in LRRK2 impair NF-ÎºB pathway in iPSC-derived neurons. Journal of Neuroinflammation, 2016, 13, 295.	7.2	46
101	Splicing misregulation of SCN5A contributes to cardiac-conduction delay and heart arrhythmia in myotonic dystrophy. Nature Communications, 2016, 7, 11067.	12.8	155
102	Genetic Mutation Analysis of Parkinsonâ€™s Disease Patients Using Multigene Next-Generation Sequencing Panels. Molecular Diagnosis and Therapy, 2016, 20, 481-491.	3.8	18
103	Assessing the role of TUBA4A gene in frontotemporal degeneration. Neurobiology of Aging, 2016, 38, 215.e13-215.e14.	3.1	9
104	Muscle imaging in muscle dystrophies produced by mutations in the EMD and LMNA genes. Neuromuscular Disorders, 2016, 26, 33-40.	0.6	40
105	Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 variants on D4Z4 hypomethylation and disease penetrance in FSHD2. European Journal of Human Genetics, 2016, 24, 78-85.	2.8	23
106	Six Serum miRNAs Fail to Validate as Myotonic Dystrophy Type 1 Biomarkers. PLoS ONE, 2016, 11, e0150501.	2.5	7
107	Progranulin deficiency induces overactivation of WNT5A expression via TNF-Î±/NF-ÎºB pathway in peripheral cells from frontotemporal dementia-linked granulin mutation carriers. Journal of Psychiatry and Neuroscience, 2016, 41, 225-239.	2.4	32
108	Pompe Disease and Autophagy: Partners in Crime, or Cause and Consequence?. Current Medicinal Chemistry, 2016, 23, 2275-2285.	2.4	6

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109	Costamere proteins and their involvement in myopathic processes. Expert Reviews in Molecular Medicine, 2015, 17, e12.	3.9	37
110	MAPT H1 Haplotype is Associated with Late-Onset Alzheimerâ€™s Disease Risk in APOE É›4 Noncarriers: Results from the Dementia Genetics Spanish Consortium. Journal of Alzheimer's Disease, 2015, 49, 343-352.	2.6	32
111	Rapidly Reversible Winging Scapula. Arthritis and Rheumatology, 2015, 67, 2502-2502.	5.6	0
112	Muscle wasting in myotonic dystrophies: a model of premature aging. Frontiers in Aging Neuroscience, 2015, 7, 125.	3.4	72
113	Large-scale recent expansion of European patrilineages shown by population resequencing. Nature Communications, 2015, 6, 7152.	12.8	69
114	Clinical Correlations With Lewy Body Pathology inLRRK2-Related Parkinson Disease. JAMA Neurology, 2015, 72, 100.	9.0	272
115	Ventricular tachycardia on chronic fingolimod treatment for multiple sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 931-932.	1.9	8
116	SORT1 Mutation Resulting in Sortilin Deficiency and p75^{NTR} Upregulation in a Family With Essential Tremor. ASN Neuro, 2015, 7, 175909141559829.	2.7	28
117	Analysis of theCHCHD10gene in patients with frontotemporal dementia and amyotrophic lateral sclerosis from Spain. Brain, 2015, 138, e400-e400.	7.6	56
118	Increasing progranulin levels and blockade of the ERK1/2 pathway: Upstream and downstream strategies for the treatment of progranulin deficient frontotemporal dementia. European Neuropsychopharmacology, 2015, 25, 386-403.	0.7	20
119	Targeted screening for the detection of Pompe disease in patients with unclassified limb-girdle muscular dystrophy or asymptomatic hyperCKemia using dried blood: A Spanish cohort. Neuromuscular Disorders, 2015, 25, 548-553.	0.6	47
120	Identification of ncRNAs as potential therapeutic targets in multiple sclerosis through differential ncRNA â€“ mRNA network analysis. BMC Genomics, 2015, 16, 250.	2.8	17
121	Neurogenetic Disorders in the Basque Population. Annals of Human Genetics, 2015, 79, 57-75.	0.8	7
122	Age gene expression and coexpression progressive signatures in peripheral blood leukocytes. Experimental Gerontology, 2015, 72, 50-56.	2.8	14
123	The Y-Chromosome Tree Bursts into Leaf: 13,000 High-Confidence SNPs Covering the Majority of Known Clades. Molecular Biology and Evolution, 2015, 32, 661-673.	8.9	137
124	Obesity and ischemic stroke modulate the methylation levels of KCNQ1 in white blood cells. Human Molecular Genetics, 2015, 24, 1432-1440.	2.9	42
125	Association between different obesity measures and the risk of stroke in the EPIC Spanish cohort. European Journal of Nutrition, 2015, 54, 365-375.	3.9	32
126	Epigenetic Changes in the Methylation Patterns of KCNQ1 and WT1 after a Weight Loss Intervention Program in Obese Stroke Patients. Current Neurovascular Research, 2015, 12, 321-333.	1.1	23

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127	Epitope Mapping of Antibodies to Alpha-Synuclein in LRRK2 Mutation Carriers, Idiopathic Parkinson Disease Patients, and Healthy Controls. Frontiers in Aging Neuroscience, 2014, 6, 169.	3.4	9
128	Circulating microparticles reflect treatment effects and clinical status in multiple sclerosis. Biomarkers in Medicine, 2014, 8, 653-661.	1.4	84
129	Dysregulation of calcium homeostasis in muscular dystrophies. Expert Reviews in Molecular Medicine, 2014, 16, e16.	3.9	79
130	Association between total, processed, red and white meat consumption and all-cause, CVD and IHD mortality: a meta-analysis of cohort studies. British Journal of Nutrition, 2014, 112, 762-775.	2.3	347
131	Prevalence of cancer in Parkinson's disease related to R1441G and G2019S mutations in LRRK2. Movement Disorders, 2014, 29, 750-755.	3.9	22
132	Entire CAPN3 gene deletion in a patient with limbâ€girdle muscular dystrophy type 2A. Muscle and Nerve, 2014, 50, 448-453.	2.2	5
133	Assessing the role of the TREM2 p.R47H variant as a risk factor for Alzheimer's disease and frontotemporal dementia. Neurobiology of Aging, 2014, 35, 444.e1-444.e4.	3.1	92
134	Leucine-rich repeat kinase 2 modulates cyclooxygenase 2 and the inflammatory response in idiopathic and genetic Parkinson's disease. Neurobiology of Aging, 2014, 35, 1116-1124.	3.1	34
135	A small noncoding RNA signature found in exosomes of GBM patient serum as a diagnostic tool. Neuro-Oncology, 2014, 16, 520-527.	1.2	298
136	Epigenetic patterns of two gene promoters (TNF-Î± and PON) in stroke considering obesity condition and dietary intake. Journal of Physiology and Biochemistry, 2014, 70, 603-614.	3.0	30
137	Murine Muscle Engineered from Dermal Precursors: An In Vitro Model for Skeletal Muscle Generation, Degeneration, and Fatty Infiltration. Tissue Engineering - Part C: Methods, 2014, 20, 28-41.	2.1	10
138	G2019S LRRK2 mutant fibroblasts from Parkinsonâ€™s disease patients show increased sensitivity to neurotoxin 1-methyl-4-phenylpyridinium dependent of autophagy. Toxicology, 2014, 324, 1-9.	4.2	40
139	Investigation of the role of rare TREM2 variants in frontotemporal dementia subtypes. Neurobiology of Aging, 2014, 35, 2657.e13-2657.e19.	3.1	34
140	LRRK2 delays degradative receptor trafficking by impeding late endosomal budding through decreasing Rab7 activity. Human Molecular Genetics, 2014, 23, 6779-6796.	2.9	139
141	Cognitive dysfunction in Parkinson's disease related to the R1441G mutation in LRRK2. Parkinsonism and Related Disorders, 2014, 20, 1097-1100.	2.2	25
142	PGRN haploinsufficiency increased Wnt5a signaling in peripheral cells from frontotemporal lobar degeneration-progranulin mutation carriers. Neurobiology of Aging, 2014, 35, 886-898.	3.1	24
143	Blood miRNA expression pattern is a possible risk marker for natalizumab-associated progressive multifocal leukoencephalopathy in multiple sclerosis patients. Multiple Sclerosis Journal, 2014, 20, 1851-1859.	3.0	50
144	Advances in gene therapies for limb-girdle muscular dystrophies. Advances in Regenerative Biology, 2014, 1, 25048.	0.2	0

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145	Transcriptomic Profile Reveals Gender-Specific Molecular Mechanisms Driving Multiple Sclerosis Progression. PLoS ONE, 2014, 9, e90482.	2.5	46
146	A neural extracellular matrix-based method for in vitrohippocampal neuron culture and dopaminergic differentiation of neural stem cells. BMC Neuroscience, 2013, 14, 48.	1.9	12
147	Genetic high throughput screening in Retinitis Pigmentosa based on high resolution melting (HRM) analysis. Experimental Eye Research, 2013, 116, 386-394.	2.6	14
148	The LRRK2 G2019S mutant exacerbates basal autophagy through activation of the MEK/ERK pathway. Cellular and Molecular Life Sciences, 2013, 70, 121-136.	5.4	148
149	Analysis of theC9orf72Gene in Patients with Amyotrophic Lateral Sclerosis in Spain and Different Populations Worldwide. Human Mutation, 2013, 34, 79-82.	2.5	85
150	Novel valosin containing protein mutation in a Swiss family with hereditary inclusion body myopathy and dementia. Neuromuscular Disorders, 2013, 23, 149-154.	0.6	11
151	Distinctive age-related temporal cortical thinning in asymptomatic granulin gene mutation carriers. Neurobiology of Aging, 2013, 34, 1462-1468.	3.1	18
152	SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V. Clinical Genetics, 2013, 83, 257-262.	2.0	94
153	The ACMSD gene, involved in tryptophan metabolism, is mutated in a family with cortical myoclonus, epilepsy, and parkinsonism. Journal of Molecular Medicine, 2013, 91, 1399-1406.	3.9	111
154	In Vitro Correction of a Pseudoexon-Generating Deep Intronic Mutation in LGMD2A by Antisense Oligonucleotides and Modified Small Nuclear RNAs. Human Mutation, 2013, 34, 1387-1395.	2.5	22
155	Clinical Response to Thalidomide in the Treatment of Intracranial Tuberculomas. Clinical Neuropharmacology, 2013, 36, 70-72.	0.7	10
156	Neurodegenerative Disease Phenotypes in Carriers of MAPT p.A152T, A Risk Factor for Frontotemporal Dementia Spectrum Disorders and Alzheimer Disease. Alzheimer Disease and Associated Disorders, 2013, 27, 302-309.	1.3	40
157	Expanded CTG repeats trigger miRNA alterations in Drosophila that are conserved in myotonic dystrophy type 1 patients. Human Molecular Genetics, 2013, 22, 704-716.	2.9	62
158	HLA-DRB1*15:01 and multiple sclerosis: a female association?. Multiple Sclerosis Journal, 2012, 18, 569-577.	3.0	59
159	Neuropsychological Features of Asymptomatic c.709-1G>A Progranulin Mutation Carriers. Journal of the International Neuropsychological Society, 2012, 18, 1086-1090.	1.8	20
160	Oncogenicity of the Developmental Transcription Factor Sox9. Cancer Research, 2012, 72, 1301-1315.	0.9	180
161	C3KO mouse expression analysis: downregulation of the muscular dystrophy Ky protein and alterations in muscle aging. Neurogenetics, 2012, 13, 347-357.	1.4	4
162	Alteration in cell cycle-related proteins in lymphoblasts from carriers of the c.709-1G>A PGRN mutation associated with FTLD-TDP dementia. Neurobiology of Aging, 2012, 33, 429.e7-429.e20.	3.1	20

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163	The MAPK1/3 pathway is essential for the deregulation of autophagy observed in G2019S LRRK2 mutant fibroblasts. Autophagy, 2012, 8, 1537-1539.	9.1	23
164	Current mutation discovery approaches in Retinitis Pigmentosa. Vision Research, 2012, 75, 117-129.	1.4	57
165	Coexistence of protease sensitive and resistant prion protein in 129VV homozygous sporadic Creutzfeldtâ€“Jakob disease: a case report. Journal of Medical Case Reports, 2012, 6, 348.	0.8	17
166	Inactivation of CDK/pRb Pathway Normalizes Survival Pattern of Lymphoblasts Expressing the FTLD-Progranulin Mutation c.709-1G>A. PLoS ONE, 2012, 7, e37057.	2.5	11
167	Î±-Synuclein Levels in Blood Plasma from LRRK2 Mutation Carriers. PLoS ONE, 2012, 7, e52312.	2.5	45
168	Modeling neural differentiation on micropatterned substrates coated with neural matrix components. Frontiers in Cellular Neuroscience, 2012, 6, 10.	3.7	19
169	Exome sequencing identifies GCDH (glutaryl-CoA dehydrogenase) mutations as a cause of a progressive form of early-onset generalized dystonia. Human Genetics, 2012, 131, 435-442.	3.8	23
170	Mitochondrial DNA polymorphisms/haplogroups in hereditary spastic paraplegia. Journal of Neurology, 2012, 259, 246-250.	3.6	9
171	Erratum to â€˜Clinical outcome in 19 French and Spanish patients with valosin-containing protein myopathy associated with Pagetâ€™s disease of bone and frontotemporal dementiaâ€™ [Neuromuscular Disorders 19 (2009) 316â€“323]. Neuromuscular Disorders, 2011, 21, e1.	0.6	0
172	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. Nature Medicine, 2011, 17, 720-725.	30.7	299
173	Olfactory deficits and cardiac ¹²³Iâ€MIBG in Parkinson's disease related to the LRRK2 R1441G and G2019S mutations. Movement Disorders, 2011, 26, 2026-2031.	3.9	51
174	Does the severity of the LGMD2A phenotype in compound heterozygotes depend on the combination of mutations?. Muscle and Nerve, 2011, 44, 710-714.	2.2	13
175	Prion Protein Codon 129 Polymorphism Modifies Age at Onset of Frontotemporal Dementia With the C.709-1G>A Progranulin Mutation. Alzheimer Disease and Associated Disorders, 2011, 25, 93-95.	1.3	11
176	Genetic and Epigenetic Modifications of Sox2 Contribute to the Invasive Phenotype of Malignant Gliomas. PLoS ONE, 2011, 6, e26740.	2.5	187
177	Penetrance in Parkinson's disease related to the LRRK2 R1441G mutation in the Basque country (Spain). Movement Disorders, 2010, 25, 2340-2345.	3.9	52
178	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	21.4	479
179	A NovelPRNP Y218NMutation in Gerstmann-StrÃussler-Scheinker Disease With Neurofibrillary Degeneration. Journal of Neuropathology and Experimental Neurology, 2010, 69, 789-800.	1.7	46
180	Neuropathology of Parkinson's disease with the R1441G mutation in LRRK2. Movement Disorders, 2009, 24, 1998-2001.	3.9	60

#	Article	IF	Citations
181	Negative Evidence for Stachydrine or Galeopsis ladanum L. Seeds as the Causal Agents of Coturnism after Quail Meat Ingestion. Journal of Agricultural and Food Chemistry, 2009, 57, 11055-11059.	5.2	6
182	Clinical outcome in 19 French and Spanish patients with valosin-containing protein myopathy associated with Pagetâ€™s disease of bone and frontotemporal dementia. Neuromuscular Disorders, 2009, 19, 316-323.	0.6	79
183	Differential Micro RNA Expression in PBMC from Multiple Sclerosis Patients. PLoS ONE, 2009, 4, e6309.	2.5	222
184	Mutations in Progranulin Gene: Clinical, Pathological, and Ribonucleic Acid Expression Findings. Biological Psychiatry, 2008, 63, 946-952.	1.3	62
185	Gene Expression Profiling in Limb-Girdle Muscular Dystrophy 2A. PLoS ONE, 2008, 3, e3750.	2.5	41
186	Increased Transcriptional Activity of Milk-Related Genes following the Active Phase of Experimental Autoimmune Encephalomyelitis and Multiple Sclerosis. Journal of Immunology, 2007, 179, 4074-4082.	0.8	19
187	Screening of the CAPN3 gene in patients with possible LGMD2A. Clinical Genetics, 2006, 69, 444-449.	2.0	22
188	Parkinson's disease due to the R1441G mutation in Dardarin: A founder effect in the basques. Movement Disorders, 2006, 21, 1954-1959.	3.9	84
189	CAPN3mutations in patients with idiopathic eosinophilic myositis. Annals of Neurology, 2006, 59, 905-911.	5.3	117
190	Familial Parkinson's disease: Clinical and genetic analysis of four Basque families. Annals of Neurology, 2005, 57, 365-372.	5.3	56
191	A Common Haplotype Associated with the Basque 2362AG-TCATCT Mutation in the Muscular Calpain-3 Gene. Human Biology, 2004, 76, 731-741.	0.2	15
192	Cloning of the Gene Containing Mutations that Cause PARK8-Linked Parkinson's Disease. Neuron, 2004, 44, 595-600.	8.1	2,183
193	A genomic screen of Spanish multiple sclerosis patients reveals multiple loci associated with the disease. Journal of Neuroimmunology, 2003, 143, 124-128.	2.3	35
194	Mutations in the LGI1/Epitempin gene on 10q24 cause autosomal dominant lateral temporal epilepsy. Human Molecular Genetics, 2002, 11, 1119-1128.	2.9	289
195	Calpain 3 deficiency is associated with myonuclear apoptosis and profound perturbation of the IÎºBÎ±/NF-Îº B pathway in limb-girdle muscular dystrophy type 2A. Nature Medicine, 1999, 5, 503-511.	30.7	261
196	Recent Male-Mediated Gene Flow over a Linguistic Barrier in Iberia, Suggested by Analysis of a Y-Chromosomal DNA Polymorphism. American Journal of Human Genetics, 1999, 65, 1437-1448.	6.2	132
197	The A1555G Mutation in the 12S rRNA Gene of Human mtDNA: Recurrent Origins and Founder Events in Families Affected by Sensorineural Deafness. American Journal of Human Genetics, 1999, 65, 1349-1358.	6.2	111
198	Autosomal Dominant Nocturnal Frontal Lobe Epilepsy in a Spanish Family With a Ser252Phe Mutation in the CHRNA4 Gene. Archives of Neurology, 1999, 56, 1004.	4.5	85