Kinji Ohno

List of Publications by Year
in descending order

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		18465	30058
241	12,955	62	103
papers	citations	h-index	g-index

259	259	259	10972
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Altered gut microbiota in Parkinson's disease patients with motor complications. Parkinsonism and Related Disorders, 2022, 95, 11-17.	1.1	10
2	Reply to the Letter to the Editor â€œThe Microbiota in Parkinsonâ€™s Disease: Ranking the Risk of Heart Diseaseâ€. Annals of Nutrition and Metabolism, 2022, , 1-2.	1.0	0
3	Meclozine ameliorates skeletal muscle pathology and increases muscle forces in mdx mice. Biochemical and Biophysical Research Communications, 2022, 592, 87-92.	1.0	0
4	Molecular Hydrogen Enhances Proliferation of Cancer Cells That Exhibit Potent Mitochondrial Unfolded Protein Response. International Journal of Molecular Sciences, 2022, 23, 2888.	1.8	12
5	Promethazine Downregulates Wnt/Î²-Catenin Signaling and Increases the Biomechanical Forces of the Injured Achilles Tendon in the Early Stage of Healing. American Journal of Sports Medicine, 2022, 50, 1317-1327.	1.9	2
6	Possible Repositioning of an Oral Anti-Osteoporotic Drug, Ipriflavone, for Treatment of Inflammatory Arthritis via Inhibitory Activity of KIAA1199, a Novel Potent Hyaluronidase. International Journal of Molecular Sciences, 2022, 23, 4089.	1.8	3
7	Extremely low-frequency pulses of faint magnetic field induce mitophagy to rejuvenate mitochondria. Communications Biology, 2022, 5, 453.	2.0	9
8	Short chain fatty acids-producing and mucin-degrading intestinal bacteria predict the progression of early Parkinsonâ€™s disease. Npj Parkinson's Disease, 2022, 8, .	2.5	41
9	Desloratadine inhibits heterotopic ossification by suppression of BMP2â€Smad1/5/8 signaling. Journal of Orthopaedic Research, 2021, 39, 1297-1304.	1.2	9
10	Efficacy of salbutamol monotherapy in slowâ€channel congenital myasthenic syndrome caused by a novel mutation in CHRND. Muscle and Nerve, 2021, 63, E30-E32.	1.0	2
11	Secreted Signaling Molecules at the Neuromuscular Junction in Physiology and Pathology. International Journal of Molecular Sciences, 2021, 22, 2455.	1.8	20
12	Rapidly Growing Protein-Centric Technologies to Extensively Identify Proteinâ€“RNA Interactions: Application to the Analysis of Co-Transcriptional RNA Processing. International Journal of Molecular Sciences, 2021, 22, 5312.	1.8	7
13	Plasmaâ€activated Ringer's lactate solution inhibits the cellular respiratory system in HeLa cells. Plasma Processes and Polymers, 2021, 18, 2100056.	1.6	9
14	IntSplice2: Prediction of the Splicing Effects of Intronic Single-Nucleotide Variants Using LightGBM Modeling. Frontiers in Genetics, 2021, 12, 701076.	1.1	6
15	Parkinsonâ€™s Disease and Gut Microbiota. Annals of Nutrition and Metabolism, 2021, 77, 28-35.	1.0	41
16	Zonisamide upregulates neuregulin-1 expression and enhances acetylcholine receptor clustering at the in vitro neuromuscular junction. Neuropharmacology, 2021, 195, 108637.	2.0	2
17	Regulated splicing of large exons is linked to phaseâ€separation of vertebrate transcription factors. EMBO Journal, 2021, 40, e107485.	3.5	8
18	Intestinal Collinsella may mitigate infection and exacerbation of COVID-19 by producing ursodeoxycholate. PLoS ONE, 2021, 16, e0260451.	1.1	42

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19	Meclozine Attenuates the MARK Pathway in Mammalian Chondrocytes and Ameliorates FGF2-Induced Bone Hyperossification in Larval Zebrafish. Frontiers in Cell and Developmental Biology, 2021, 9, 694018.	1.8	1
20	Hydrogen water alleviates obliterative airway disease in mice. General Thoracic and Cardiovascular Surgery, 2020, 68, 158-163.	0.4	4
21	Freeze-drying enables homogeneous and stable sample preparation for determination of fecal short-chain fatty acids. Analytical Biochemistry, 2020, 589, 113508.	1.1	23
22	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution. NAR Genomics and Bioinformatics, 2020, 2, lqaa038.	1.5	16
23	Gene Expression Profile at the Motor Endplate of the Neuromuscular Junction of Fast-Twitch Muscle. Frontiers in Molecular Neuroscience, 2020, 13, 154.	1.4	12
24	Identification of Qk as a Glial Precursor Cell Marker that Governs the Fate Specification of Neural Stem Cells to a Glial Cell Lineage. Stem Cell Reports, 2020, 15, 883-897.	2.3	18
25	Zonisamide ameliorates neuropathic pain partly by suppressing microglial activation in the spinal cord in a mouse model. Life Sciences, 2020, 263, 118577.	2.0	7
26	Zonisamide ameliorates progression of cervical spondylotic myelopathy in a rat model. Scientific Reports, 2020, 10, 13138.	1.6	10
27	Short-Chain Fatty Acid-Producing Gut Microbiota Is Decreased in Parkinsonâ€™s Disease but Not in Rapid-Eye-Movement Sleep Behavior Disorder. MSystems, 2020, 5, .	1.7	63
28	tRIPâ€seq reveals repression of premature polyadenylation by coâ€transcriptional FUSâ€U1 snRNP assembly. EMBO Reports, 2020, 21, e49890.	2.0	18
29	<scp>Metaâ€Analysis</scp> of Gut Dysbiosis in Parkinson's Disease. Movement Disorders, 2020, 35, 1626-1635.	2.2	208
30	Inhibition of cyclooxygenase-1 by nonsteroidal anti-inflammatory drugs demethylates MeR2 enhancer and promotes Mbnl1 transcription in myogenic cells. Scientific Reports, 2020, 10, 2558.	1.6	12
31	Congenital myasthenic syndromeâ€“associated agrin variants affect clustering of acetylcholine receptors in a domain-specific manner. JCI Insight, 2020, 5, .	2.3	15
32	CTGF/CCN2 facilitates LRP4â€mediated formation of the embryonic neuromuscular junction. EMBO Reports, 2020, 21, e48462.	2.0	15
33	Editorial: RNA Diseases in Humansâ€”From Fundamental Research to Therapeutic Applications. Frontiers in Molecular Biosciences, 2019, 6, 53.	1.6	1
34	Mianserin suppresses R-spondin 2-induced activation of Wnt/Î²-catenin signaling in chondrocytes and prevents cartilage degradation in a rat model of osteoarthritis. Scientific Reports, 2019, 9, 2808.	1.6	19
35	Protein-anchoring therapy to target extracellular matrix proteins to their physiological destinations. Matrix Biology, 2018, 68-69, 628-636.	1.5	8
36	Molecular hydrogen upregulates heat shock response and collagen biosynthesis, and downregulates cell cycles: meta-analyses of gene expression profiles. Free Radical Research, 2018, 52, 434-445.	1.5	16

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37	MYRF is associated with encephalopathy with reversible myelin vacuolization. Annals of Neurology, 2018, 83, 98-106.	2.8	35
38	Rare loss of function mutations in N-methyl-d-aspartate glutamate receptors and their contributions to schizophrenia susceptibility. Translational Psychiatry, 2018, 8, 12.	2.4	41
39	Lack of Fgf18 causes abnormal clustering of motor nerve terminals at the neuromuscular junction with reduced acetylcholine receptor clusters. Scientific Reports, 2018, 8, 434.	1.6	12
40	Loss of Sfpq Causes Long-Gene Transcriptopathy in the Brain. Cell Reports, 2018, 23, 1326-1341.	2.9	75
41	Rules and tools to predict the splicing effects of exonic and intronic mutations. Wiley Interdisciplinary Reviews RNA, 2018, 9, e1451.	3.2	90
42	Quantification of hydrogen production by intestinal bacteria that are specifically dysregulated in Parkinson's disease. PLoS ONE, 2018, 13, e0208313.	1.1	41
43	Randomized, doubleâ€blind, multicenter trial of hydrogen water for Parkinson's disease. Movement Disorders, 2018, 33, 1505-1507.	2.2	26
44	Differential effects of spinal motor neuron-derived and skeletal muscle-derived Rspo2 on acetylcholine receptor clustering at the neuromuscular junction. Scientific Reports, 2018, 8, 13577.	1.6	11
45	Inhalation of hydrogen gas elevates urinary 8-hydroxy-2â€²-deoxyguanine in Parkinsonâ€™s disease. Medical Gas Research, 2018, 8, 144.	1.2	31
46	Molecular hydrogen alleviates motor deficits and muscle degeneration in mdx mice. Redox Report, 2017, 22, 26-34.	1.4	19
47	Splicing regulation and dysregulation of cholinergic genes expressed at the neuromuscular junction. Journal of Neurochemistry, 2017, 142, 64-72.	2.1	20
48	Six GU-rich (6GUR) FUS-binding motifs detected by normalization of CLIP-seq by Nascent-seq. Gene, 2017, 618, 57-64.	1.0	9
49	Agrin-LRP4-MuSK signaling as a therapeutic target for myasthenia gravis and other neuromuscular disorders. Expert Opinion on Therapeutic Targets, 2017, 21, 949-958.	1.5	44
50	Interactions between genetic polymorphisms of glucose metabolizing genes and smoking and alcohol consumption in the risk of type 2 diabetes mellitus. Applied Physiology, Nutrition and Metabolism, 2017, 42, 1316-1321.	0.9	8
51	An ENU-induced splice site mutation of mouse Col1a1 causing recessive osteogenesis imperfecta and revealing a novel splicing rescue. Scientific Reports, 2017, 7, 11717.	1.6	7
52	Promethazine Hydrochloride Inhibits Ectopic Fat Cell Formation in Skeletal Muscle. American Journal of Pathology, 2017, 187, 2627-2634.	1.9	12
53	SRSF1 suppresses selection of intron-distal 5â€² splice site of DOK7 intron 4 to generate functional full-length Dok-7 protein. Scientific Reports, 2017, 7, 10446.	1.6	4
54	Clinical dosage of meclozine promotes longitudinal bone growth, bone volume, and trabecular bone quality in transgenic mice with achondroplasia. Scientific Reports, 2017, 7, 7371.	1.6	15

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55	Activated FGFR3 promotes bone formation via accelerating endochondral ossification in mouse model of distraction osteogenesis. Bone, 2017, 105, 42-49.	1.4	14
56	Maternal administration of meclozine for the treatment of foramen magnum stenosis in transgenic mice with achondroplasia. Journal of Neurosurgery: Pediatrics, 2017, 19, 91-95.	0.8	7
57	Protein-Anchoring Therapy of Biglycan forMdxMouse Model of Duchenne Muscular Dystrophy. Human Gene Therapy, 2017, 28, 428-436.	1.4	19
58	Wnt/Î²-catenin signaling suppresses expressions of Scx, Mkx, and Tnmd in tendon-derived cells. PLoS ONE, 2017, 12, e0182051.	1.1	44
59	Fluoxetine ameliorates cartilage degradation in osteoarthritis by inhibiting Wnt/Î²-catenin signaling. PLoS ONE, 2017, 12, e0184388.	1.1	27
60	Progression of Parkinson's disease is associated with gut dysbiosis: Two-year follow-up study. PLoS ONE, 2017, 12, e0187307.	1.1	195
61	Polymorphisms in Four Genes (KCNQ1 rs151290, KLF14 rs972283, GCKR rs780094 and MTNR1B rs10830963) and Their Correlation with Type 2 Diabetes Mellitus in Han Chinese in Henan Province, China. International Journal of Environmental Research and Public Health, 2016, 13, 260.	1.2	27
62	Global identification of hnRNP A1 binding sites for SSO-based splicing modulation. BMC Biology, 2016, 14, 54.	1.7	62
63	Serum Tyrosine-to-Phenylalanine Ratio isÂLow in Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2016, 6, 423-431.	1.5	17
64	Is the serum creatine kinase level elevated in congenital myasthenic syndrome?. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 801-801.	0.9	3
65	R-spondin 2 facilitates differentiation of proliferating chondrocytes into hypertrophic chondrocytes by enhancing Wnt/Î²-catenin signaling in endochondral ossification. Biochemical and Biophysical Research Communications, 2016, 473, 255-264.	1.0	31
66	Roles of collagen Q in MuSK antibody-positive myasthenia gravis. Chemico-Biological Interactions, 2016, 259, 266-270.	1.7	12
67	Mutations Causing Slow-Channel Myasthenia Reveal That a Valine Ring in the Channel Pore of Muscle AChR is Optimized for Stabilizing Channel Gating. Human Mutation, 2016, 37, 1051-1059.	1.1	19
68	Competitive regulation of alternative splicing and alternative polyadenylation by hnRNP H and CstF64 determines acetylcholinesterase isoforms. Nucleic Acids Research, 2016, 45, gkw823.	6.5	53
69	Recent advances in congenital myasthenic syndromes. Clinical and Experimental Neuroimmunology, 2016, 7, 246-259.	0.5	9
70	Molecular hydrogen ameliorates several characteristics of preeclampsia in the Reduced Uterine Perfusion Pressure (RUPP) rat model. Free Radical Biology and Medicine, 2016, 101, 524-533.	1.3	25
71	Molecular hydrogen suppresses activated Wnt/Î²-catenin signaling. Scientific Reports, 2016, 6, 31986.	1.6	20
72	R-spondin 2 promotes acetylcholine receptor clustering at the neuromuscular junction via Lgr5. Scientific Reports, 2016, 6, 28512.	1.6	24

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73	Phenylbutazone induces expression of MBNL1 and suppresses formation of MBNL1-CUG RNA foci in a mouse model of myotonic dystrophy. Scientific Reports, 2016, 6, 25317.	1.6	29
74	Hydrogen-rich water ameliorates bronchopulmonary dysplasia (BPD) in newborn rats. Pediatric Pulmonology, 2016, 51, 928-935.	1.0	23
75	<scp>FUS</scp>â€mediated regulation of alternative <scp>RNA</scp> processing in neurons: insights from global transcriptome analysis. Wiley Interdisciplinary Reviews RNA, 2016, 7, 330-340.	3.2	34
76	IntSplice: prediction of the splicing consequences of intronic single-nucleotide variations in the human genome. Journal of Human Genetics, 2016, 61, 633-640.	1.1	29
77	Intestinal dysbiosis and lowered serum lipopolysaccharide-binding protein in PD. Parkinsonism and Related Disorders, 2016, 22, e32.	1.1	1
78	Tranilast stimulates endochondral ossification by upregulating SOX9 and RUNX2 promoters. Biochemical and Biophysical Research Communications, 2016, 470, 356-361.	1.0	8
79	Neuroprotective potential of molecular hydrogen against perinatal brain injury via suppression of activated microglia. Free Radical Biology and Medicine, 2016, 91, 154-163.	1.3	41
80	Repositioning again of zonisamide for nerve regeneration. Neural Regeneration Research, 2016, 11, 541.	1.6	5
81	Collagen Q and anti-MuSK autoantibody competitively suppress agrin/LRP4/MuSK signaling. Scientific Reports, 2015, 5, 13928.	1.6	54
82	SRSF1 and hnRNP H antagonistically regulate splicing of COLQ exon 16 in a congenital myasthenic syndrome. Scientific Reports, 2015, 5, 13208.	1.6	30
83	Lansoprazole Upregulates Polyubiquitination of the TNF Receptor-Associated Factor 6 and Facilitates Runx2-mediated Osteoblastogenesis. EBioMedicine, 2015, 2, 2046-2061.	2.7	15
84	Beneficial biological effects and the underlying mechanisms of molecular hydrogen - comprehensive review of 321 original articles -. Medical Gas Research, 2015, 5, 12.	1.2	199
85	Zonisamide Enhances Neurite Elongation of Primary Motor Neurons and Facilitates Peripheral Nerve Regeneration In Vitro and in a Mouse Model. PLoS ONE, 2015, 10, e0142786.	1.1	28
86	Position-specific binding of FUS to nascent RNA regulates mRNA length. Genes and Development, 2015, 29, 1045-1057.	2.7	98
87	A missense mutation in domain III in HSPG2 in Schwartzâ€"Jampel syndrome compromises secretion of perlecan into the extracellular space. Neuromuscular Disorders, 2015, 25, 667-671.	0.3	18
88	Impaired Synaptic Development, Maintenance, and Neuromuscular Transmission in LRP4-Related Myasthenia. JAMA Neurology, 2015, 72, 889.	4.5	41
89	Simultaneous oral and inhalational intake of molecular hydrogen additively suppresses signaling pathways in rodents. Molecular and Cellular Biochemistry, 2015, 403, 231-241.	1.4	43
90	Congenital myasthenic syndrome in Japan: Ethnically unique mutations in muscle nicotinic acetylcholine receptor subunits. Neuromuscular Disorders, 2015, 25, 60-69.	0.3	18

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91	CHCHD2 mutations in autosomal dominant late-onset Parkinson's disease: a genome-wide linkage and sequencing study. Lancet Neurology, The, 2015, 14, 274-282.	4.9	285
92	Noninvasive monitoring of plasma l-dopa concentrations using sweat samples in Parkinson's disease. Clinica Chimica Acta, 2015, 442, 52-55.	0.5	32
93	Hydrogen ameliorates pulmonary hypertension in rats by anti-inflammatory and antioxidant effects. Journal of Thoracic and Cardiovascular Surgery, 2015, 150, 645-654.e3.	0.4	40
94	FUS regulates AMPA receptor function and FTLD/ALS-associated behaviour via GluA1 mRNA stabilization. Nature Communications, 2015, 6, 7098.	5.8	129
95	Dopaminergic differentiation of stem cells from human deciduous teeth and their therapeutic benefits for Parkinsonian rats. Brain Research, 2015, 1613, 59-72.	1.1	87
96	Meclozine Promotes Longitudinal Skeletal Growth in Transgenic Mice with Achondroplasia Carrying a Gain-of-Function Mutation in the FGFR3 Gene. Endocrinology, 2015, 156, 548-554.	1.4	44
97	LRP4 myasthenia: Investigation of a second kinship reveals impaired development and maintenance of the neuromuscular junction. Neuromuscular Disorders, 2015, 25, S186-S187.	0.3	1
98	Intestinal Dysbiosis and Lowered Serum Lipopolysaccharide-Binding Protein in Parkinsonâ€™s Disease. PLoS ONE, 2015, 10, e0142164.	1.1	381
99	Decoding Abnormal Splicing Code in Human Diseases. Journal of Investigative Genomics, 2015, 2, .	0.2	1
100	Splicing Aberrations in Congenital Myasthenic Syndromes. Journal of Investigative Genomics, 2015, 2, .	0.2	0
101	Verapamil Protects against Cartilage Degradation in Osteoarthritis by Inhibiting Wnt/Î²-Catenin Signaling. PLoS ONE, 2014, 9, e92699.	1.1	67
102	Searching for Genomic Region of High-Fat Diet-Induced Type 2 Diabetes in Mouse Chromosome 2 by Analysis of Congenic Strains. PLoS ONE, 2014, 9, e96271.	1.1	17
103	Clinical and genetic analysis of the first known Asian family with myotonic dystrophy type 2. Journal of Human Genetics, 2014, 59, 129-133.	1.1	4
104	Mutation analysis of a large cohort of GNE myopathy reveals a diverse array of GNE mutations affecting sialic acid biosynthesis. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 831-831.	0.9	2
105	<scp> SIL1 </scp> , a causative cochaperone gene of <scp>M</scp> arinescoâ€•<scp>S</scp> jÃ¶gren syndrome, plays an essential role in establishing the architecture of the developing cerebral cortex. EMBO Molecular Medicine, 2014, 6, 414-429.	3.3	29
106	A Kir3.4 mutation causes Andersenâ€“Tawil syndrome by an inhibitory effect on Kir2.1. Neurology, 2014, 82, 1058-1064.	1.5	59
107	LRP4 third Î²-propeller domain mutations cause novel congenital myasthenia by compromising agrin-mediated MuSK signaling in a position-specific manner. Human Molecular Genetics, 2014, 23, 1856-1868.	1.4	96
108	The ALS/FTLDâ€related RNAâ€binding proteins TDPâ€43 and FUS have common downstream RNA targets in cortical neurons. FEBS Open Bio, 2014, 4, 1-10.	1.0	50

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109	Collagen Q is a Key Player for Developing Rational Therapy for Congenital Myasthenia and for Dissecting the Mechanisms of Anti-MuSK Myasthenia Gravis. Journal of Molecular Neuroscience, 2014, 53, 359-361.	1.1	8
110	LRP4 induces extracellular matrix productions and facilitates chondrocyte differentiation. Biochemical and Biophysical Research Communications, 2014, 451, 302-307.	1.0	25
111	Maternal molecular hydrogen administration ameliorates rat fetal hippocampal damage caused by in utero ischemiaâ€“reperfusion. Free Radical Biology and Medicine, 2014, 69, 324-330.	1.3	29
112	LDB3 splicing abnormalities are specific to skeletal muscles of patients with myotonic dystrophy type 1 and alter its PKC binding affinity. Neurobiology of Disease, 2014, 69, 200-205.	2.1	26
113	HnRNP C, YB-1 and hnRNP L coordinately enhance skipping of human MUSK exon 10 to generate a Wnt-insensitive MuSK isoform. Scientific Reports, 2014, 4, 6841.	1.6	31
114	Molecular hydrogen attenuates fatty acid uptake and lipid accumulation through downregulating CD36 expression in HepG2 cells. Medical Gas Research, 2013, 3, 6.	1.2	20
115	Exome sequencing of senescence-accelerated mice (SAM) reveals deleterious mutations in degenerative disease-causing genes. BMC Genomics, 2013, 14, 248.	1.2	29
116	Glycosylation defects as an emerging novel cause leading to a limb-girdle type of congenital myasthenic syndromes. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 1064-1064.	0.9	4
117	Mutations in the C-Terminal Domain of ColQ in Endplate Acetylcholinesterase Deficiency Compromise ColQ-MuSK Interaction. Human Mutation, 2013, 34, 997-1004.	1.1	32
118	A simple analytical method involving the use of a monolithic silica disk-packed spin column and HPLC-ECD for determination of l-DOPA in plasma of patients with Parkinson's disease. Analytical Methods, 2013, 5, 5161.	1.3	12
119	S100A10 is required for the organization of actin stress fibers and promotion of cell spreading. Molecular and Cellular Biochemistry, 2013, 374, 105-111.	1.4	23
120	P.12.10 Exome sequencing analysis reveals a mutation of Kir3.4 in a patient with Andersenâ€“Tawil syndrome. Neuromuscular Disorders, 2013, 23, 808.	0.3	0
121	Specific binding of collagen Q to the neuromuscular junction is exploited to cure congenital myasthenia and to explore bases of myasthenia gravis. Chemico-Biological Interactions, 2013, 203, 335-340.	1.7	14
122	FUS-regulated region- and cell-type-specific transcriptome is associated with cell selectivity in ALS/FTLD. Scientific Reports, 2013, 3, 2388.	1.6	41
123	GFPT1-myasthenia. Neurology, 2013, 81, 370-378.	1.5	54
124	Perhexiline maleate in the treatment of fibrodysplasia ossificans progressiva: an open-labeled clinical trial. Orphanet Journal of Rare Diseases, 2013, 8, 163.	1.2	14
125	HnRNP L and hnRNP LL antagonistically modulate PTB-mediated splicing suppression of CHRNA1 pre-mRNA. Scientific Reports, 2013, 3, 2931.	1.6	41
126	Meclozine Facilitates Proliferation and Differentiation of Chondrocytes by Attenuating Abnormally Activated FGFR3 Signaling in Achondroplasia. PLoS ONE, 2013, 8, e81569.	1.1	46

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127	Protein-anchoring Strategy for Delivering Acetylcholinesterase to the Neuromuscular Junction. Molecular Therapy, 2012, 20, 1384-1392.	3.7	28
128	Four parameters increase the sensitivity and specificity of the exon array analysis and disclose 25 novel aberrantly spliced exons in myotonic dystrophy. Journal of Human Genetics, 2012, 57, 368-374.	1.1	15
129	Myotonic dystrophy type 2 is rare in the Japanese population. Journal of Human Genetics, 2012, 57, 219-220.	1.1	12
130	CUGBP1 and MBNL1 preferentially bind to 3â€² UTRs and facilitate mRNA decay. Scientific Reports, 2012, 2, 209.	1.6	150
131	A novel mutation in SCN4A causes severe myotonia and school-age-onset paralytic episodes. Journal of the Neurological Sciences, 2012, 315, 15-19.	0.3	16
132	Clinically applicable antianginal agents suppress osteoblastic transformation of myogenic cells and heterotopic ossifications in mice. Journal of Bone and Mineral Metabolism, 2012, 31, 26-33.	1.3	17
133	Position-dependent FUS-RNA interactions regulate alternative splicing events and transcriptions. Scientific Reports, 2012, 2, 529.	1.6	180
134	Drinking hydrogen water and intermittent hydrogen gas exposure, but not lactulose or continuous hydrogen gas exposure, prevent 6-hydorxydopamine-induced Parkinsonâ€™s disease in rats. Medical Gas Research, 2012, 2, 15.	1.2	86
135	The Unstable CCTG Repeat Responsible for Myotonic Dystrophy Type 2 Originates from an AluSx Element Insertion into an Early Primate Genome. PLoS ONE, 2012, 7, e38379.	1.1	26
136	Molecular Hydrogen as an Emerging Therapeutic Medical Gas for Neurodegenerative and Other Diseases. Oxidative Medicine and Cellular Longevity, 2012, 2012, 1-11.	1.9	130
137	RNA Pathologies in Neurological Disorders. Advances in Neurobiology, 2011, , 399-415.	1.3	0
138	Molecular hydrogen inhibits lipopolysaccharide/interferon Î³-induced nitric oxide production through modulation of signal transduction in macrophages. Biochemical and Biophysical Research Communications, 2011, 411, 143-149.	1.0	88
139	Urinary 8-hydroxydeoxyguanosine correlate with hallucinations rather than motor symptoms in Parkinsonâ€™s disease. Parkinsonism and Related Disorders, 2011, 17, 46-49.	1.1	30
140	Hyperuricemia cosegregating with osteogenesis imperfecta is associated with a mutation in GPATCH8. Human Genetics, 2011, 130, 671-683.	1.8	8
141	The 2011 Medical Molecular Hydrogen Symposium: An inaugural symposium of the journal Medical Gas Research. Medical Gas Research, 2011, 1, 10.	1.2	20
142	Open-label trial and randomized, double-blind, placebo-controlled, crossover trial of hydrogen-enriched water for mitochondrial and inflammatory myopathies. Medical Gas Research, 2011, 1, 24.	1.2	69
143	AG-dependent 3â€²-splice sites are predisposed to aberrant splicing due to a mutation at the first nucleotide of an exon. Nucleic Acids Research, 2011, 39, 4396-4404.	6.5	28
144	Anti-MuSK autoantibodies block binding of collagen Q to MuSK. Neurology, 2011, 77, 1819-1826.	1.5	112

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145	Myasthenic syndrome caused by plectinopathy. Neurology, 2011, 76, 327-336.	1.5	71
146	Tannic acid facilitates expression of the polypyrimidine tract binding protein and alleviates deleterious inclusion of CHRNA1 exon P3A due to an hnRNP H-disrupting mutation in congenital myasthenic syndrome. Human Molecular Genetics, 2009, 18, 1229-1237.	1.4	28
147	Myasthenic syndrome due to defects in rapsyn. Neurology, 2009, 73, 228-235.	1.5	65
148	Alu-Mediated Acquisition of Unstable ATTCT Pentanucleotide Repeats in the Human ATXN10 Gene. Molecular Biology and Evolution, 2009, 26, 2573-2579.	3.5	11
149	Ancestral Origin of the ATTCT Repeat Expansion in Spinocerebellar Ataxia Type 10 (SCA10). PLoS ONE, 2009, 4, e4553.	1.1	40
150	Molecular hydrogen suppresses FcÎµRI-mediated signal transduction and prevents degranulation of mast cells. Biochemical and Biophysical Research Communications, 2009, 389, 651-656.	1.0	122
151	Molecular hydrogen is protective against 6-hydroxydopamine-induced nigrostriatal degeneration in a rat model of Parkinson's disease. Neuroscience Letters, 2009, 453, 81-85.	1.0	190
152	Myotonic dystrophy type 2 in Japan: ancestral origin distinct from Caucasian families. Neurogenetics, 2008, 9, 61-63.	0.7	21
153	Long-range PCR for the diagnosis of spinocerebellar ataxia type 10. Neurogenetics, 2008, 9, 151-152.	0.7	3
154	Viral vector-medicated expression of human collagen Q in cultured cells. Chemico-Biological Interactions, 2008, 175, 346-348.	1.7	0
155	Human branch point consensus sequence is yUnAy. Nucleic Acids Research, 2008, 36, 2257-2267.	6.5	213
156	hnRNP H enhances skipping of a nonfunctional exon P3A in CHRNA1 and a mutation disrupting its binding causes congenital myasthenic syndrome. Human Molecular Genetics, 2008, 17, 4022-4035.	1.4	54
157	Congenital myastheniaâ€"related AChR Î´ subunit mutation interferes with intersubunit communication essential for channel gating. Journal of Clinical Investigation, 2008, 118, 1867-1876.	3.9	50
158	Thermodynamic instability of siRNA duplex is a prerequisite for dependable prediction of siRNA activities. Nucleic Acids Research, 2007, 35, e123.	6.5	109
159	Essential Role of GATA Transcriptional Factors in the Activation of Mast Cells. Journal of Immunology, 2007, 178, 360-368.	0.4	32
160	In vitro and in silico analysis reveals an efficient algorithm to predict the splicing consequences of mutations at the 5â€² splice sites. Nucleic Acids Research, 2007, 35, 5995-6003.	6.5	60
161	Effects of preceding sialadenitis on the development of autoimmunity against salivary gland. Oral Diseases, 2007, 13, 158-162.	1.5	8
162	Spectrum of splicing errors caused by CHRNE mutations affecting introns and intron/exon boundaries. Journal of Medical Genetics, 2005, 42, e53-e53.	1.5	17

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163	Gene symbol: CHRNE. Disease: Endplate acetylcholine receptor deficiency. Human Genetics, 2005, 117, 301.	1.8	0
164	Splicing abnormalities in congenital myasthenic syndromes. Acta Myologica, 2005, 24, 50-4.	1.5	5
165	Lack of founder haplotype for the rapsyn N88K mutation: N88K is an ancient founder mutation or arises from multiple founders. Journal of Medical Genetics, 2004, 41, 8e-8.	1.5	18
166	Myofibrillar myopathy: clinical, morphological and genetic studies in 63 patients. Brain, 2004, 127, 439-451.	3.7	244
167	C-terminal and Heparin-binding Domains of Collagenic Tail Subunit Are Both Essential for Anchoring Acetylcholinesterase at the Synapse. Journal of Biological Chemistry, 2004, 279, 10997-11005.	1.6	78
168	Subunit-specific contribution to agonist binding and channel gating revealed by inherited mutation in muscle acetylcholine receptor M3-M4 linker. Brain, 2004, 128, 345-355.	3.7	29
169	Choline acetyltransferase structure reveals distribution of mutations that cause motor disorders. EMBO Journal, 2004, 23, 2047-2058.	3.5	57
170	Novel truncating RAPSN mutations causing congenital myasthenic syndrome responsive to 3,4-diaminopyridine. Neuromuscular Disorders, 2004, 14, 202-207.	0.3	39
171	Molecular insights into acetylcholine receptor structure and function revealed by mutations causing congenital myasthenic syndromes. Advances in Molecular and Cell Biology, 2004, 32, 95-119.	0.1	0
172	Congenital myasthenic syndromes. , 2004, , 213-226.		2
173	Congenital myasthenic syndromes: A diverse array of molecular targets. Journal of Neurocytology, 2003, 32, 1017-1037.	1.6	20
174	Mechanistic Diversity Underlying Fast Channel Congenital Myasthenic Syndromes. Annals of the New York Academy of Sciences, 2003, 998, 128-137.	1.8	13
175	Congenital Myasthenic Syndromes: Multiple Molecular Targets at the Neuromuscular Junction. Annals of the New York Academy of Sciences, 2003, 998, 138-160.	1.8	49
176	Congenital myasthenic syndromes. European Journal of Paediatric Neurology, 2003, 7, 227-228.	0.7	2
177	Congenital myasthenic syndromes: Progress over the past decade. Muscle and Nerve, 2003, 27, 4-25.	1.0	130
178	Sleuthing molecular targets for neurological diseases at the neuromuscular junction. Nature Reviews Neuroscience, 2003, 4, 339-352.	4.9	212
179	A frameshifting mutation in CHRNE unmasks skipping of the preceding exon. Human Molecular Genetics, 2003, 12, 3055-3066.	1.4	29
180	Myasthenic syndrome caused by mutation of the SCN4A sodium channel. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 7377-7382.	3.3	176

#	Article	IF	Citations
181	E-box mutations in the RAPSN promoter region in eight cases with congenital myasthenic syndrome. Human Molecular Genetics, 2003, 12, 739-748.	1.4	83
182	Mutation causing severe myasthenia reveals functional asymmetry of AChR signature cystine loops in agonist binding and gating. Journal of Clinical Investigation, 2003, 111, 497-505.	3.9	68
183	Naturally Occurring Mutations at the Acetylcholine Receptor Binding Site Independently Alter ACh Binding and Channel Gating. Journal of General Physiology, 2002, 120, 483-496.	0.9	59
184	Three novel COLQ mutations and variation of phenotypic expressivity due to G240X. Neurology, 2002, 58, 603-609.	1.5	54
185	Rapsyn Mutations in Humans Cause Endplate Acetylcholine-Receptor Deficiency and Myasthenic Syndrome. American Journal of Human Genetics, 2002, 70, 875-885.	2.6	221
186	Congenital myasthenic syndrome associated with episodic apnea and sudden infant death. Neuromuscular Disorders, 2002, 12, 548-553.	0.3	72
187	Congenital myasthenic syndromes: Genetic defects of the neuromuscular junction. Current Neurology and Neuroscience Reports, 2002, 2, 78-88.	2.0	30
188	The Spectrum of Congenital Myasthenic Syndromes. Molecular Neurobiology, 2002, 26, 347-367.	1.9	21
189	Congenital myasthenic syndromes. Advances in Neurology, 2002, 88, 203-15.	0.8	7
190	Choline acetyltransferase mutations cause myasthenic syndrome associated with episodic apnea in humans. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 2017-2022.	3.3	254
191	Functional characterisation of mitochondrial tRNATyr mutation (5877Gright-arrowA) associated with familial chronic progressive external ophthalmoplegia. Journal of Medical Genetics, 2001, 38, 703-705.	1.5	12
192	The spectrum of mutations causing end-plate acetylcholinesterase deficiency. Annals of Neurology, 2000, 47, 162-170.	2.8	123
193	Fundamental Gating Mechanism of Nicotinic Receptor Channel Revealed by Mutation Causing a Congenital Myasthenic Syndrome. Journal of General Physiology, 2000, 116, 449-462.	0.9	77
194	Acetylcholine receptor M3 domain: stereochemical and volume contributions to channel gating. Nature Neuroscience, 1999, 2, 226-233.	7.1	119
195	Congenital End-Plate Acetylcholinesterase Deficiency Caused by a Nonsense Mutation and an Aâ†'G Splice-Donorâ€"Site Mutation at Position +3 of the Collagenlike-Tailâ€"Subunit Gene (COLQ): How Does G at Position +3 Result in Aberrant Splicing?. American Journal of Human Genetics, 1999, 65, 635-644.	2.6	88
196	Congenital myasthenic syndrome caused by a mutation in the Ets-binding site of the promoter region of the acetylcholine receptor Ìµ subunit gene. Neuromuscular Disorders, 1999, 9, 131-135.	0.3	81
197	Congenital Myasthenic Syndromes. Archives of Neurology, 1999, 56, 163.	4.9	106
198	Mutation causing congenital myasthenia reveals acetylcholine receptor Î²/Î´ subunit interaction essential for assembly. Journal of Clinical Investigation, 1999, 104, 1403-1410.	3.9	71

#	Article	IF	Citations
199	Congenital Myasthenic Syndromes: New Insights from Molecular Genetic and Patch-Clamp Studiesa. Annals of the New York Academy of Sciences, 1998, 841, 140-156.	1.8	17
200	Congenital Myasthenic Syndrome Caused by Novel Loss-of-Function Mutations in the Human AChR e Subunit Genea. Annals of the New York Academy of Sciences, 1998, 841, 184-188.	1.8	6
201	Frameshifting and Splice-Site Mutations in the Acetylcholine Receptor e Subunit Gene in Three Turkish Kinships with Congenital Myasthenic Syndromesa. Annals of the New York Academy of Sciences, 1998, 841, 189-194.	1.8	13
202	AChR Channel Blockade by Quinidine Sulfate Reduces Channel Open Duration in the Slow-Channel Congenital Myasthenic Syndromea. Annals of the New York Academy of Sciences, 1998, 841, 199-202.	1.8	15
203	Congenital myasthenic syndromes: Experiments of nature. Journal of Physiology (Paris), 1998, 92, 113-117.	2.1	9
204	Myasthenic syndromes in Turkish kinships due to mutations in the acetylcholine receptor. Annals of Neurology, 1998, 44, 234-241.	2.8	67
205	Mode Switching Kinetics Produced by a Naturally Occurring Mutation in the Cytoplasmic Loop of the Human Acetylcholine Receptor Îµ Subunit. Neuron, 1998, 20, 575-588.	3.8	109
206	REVIEW â– : Molecular Basis of Congenital Myasthenic Syndromes: Mutations in the Acetylcholine Receptor. Neuroscientist, 1998, 4, 185-194.	2.6	27
207	Human endplate acetylcholinesterase deficiency caused by mutations in the collagen-like tail subunit (ColQ) of the asymmetric enzyme. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 9654-9659.	3.3	249
208	Quinidine normalizes the open duration of slow-channel mutants of the acetylcholine receptor. NeuroReport, 1998, 9, 1907-1911.	0.6	55
209	Congenital Myasthenic Syndromes due to Heteroallelic Nonsense/Missense Mutations in the Acetylcholine Receptor Â Subunit Gene: Identification and Functional Characterization of Six New Mutations. Human Molecular Genetics, 1997, 6, 753-766.	1.4	164
210	Mutation in the M1 Domain of the Acetylcholine Receptor Î± Subunit Decreases the Rate of Agonist Dissociation. Journal of General Physiology, 1997, 109, 757-766.	0.9	138
211	Slow-Channel Myasthenic Syndrome Caused By Enhanced Activation, Desensitization, and Agonist Binding Affinity Attributable to Mutation in the M2 Domain of the Acetylcholine Receptor Î± Subunit. Journal of Neuroscience, 1997, 17, 5651-5665.	1.7	147
212	Congenital Myasthenic Syndrome Caused by Decreased Agonist Binding Affinity Due to a Mutation in the Acetylcholine Receptor Îµ Subunit. Neuron, 1996, 17, 157-170.	3.8	240
213	MELAS- and kearns-sayre-type with myopathy and autoimmune polyendocrinopahy. Annals of Neurology, 1996, 39, 761-766.	2.8	57
214	End-plate acetylcholine receptor deficiency due to nonsense mutations in the ? subunit. Annals of Neurology, 1996, 40, 810-817.	2.8	159
215	New mutations in acetylcholine receptor subunit genes reveal heterogeneity in the slow-channel congenital myasthenic syndrome. Human Molecular Genetics, 1996, 5, 1217-1227.	1.4	176
216	Congenital myasthenic syndrome caused by prolonged acetylcholine receptor channel openings due to a mutation in the M2 domain of the epsilon subunit.. Proceedings of the National Academy of Sciences of the United States of America, 1995, 92, 758-762.	3.3	256

#	Article	IF	Citations
217	Mutation of the acetylcholine receptor Î± subunit causes a slow-channel myasthenic syndrome by enhancing agonist binding affinity. Neuron, 1995, 15, 229-239.	3.8	273
218	Molecular and Genetic Analyses of Two Patients with Pearson's Marrow-Pancreas Syndrome. Pediatric Research, 1993, 34, 105-110.	1.1	31
219	Progressive External Ophthalmoplegia and Myositis.. Internal Medicine, 1993, 32, 319-322.	0.3	7
220	Increased Mitochondrial DNA Deletions in the Skeletal Muscle of Myotonic Dystrophy. Gerontology, 1992, 38, 18-29.	1.4	23
221	Distinct clustering of point mutations in mitochondrial DNA among patients with mitochondrial encephalomyopathies and with Parkinson's disease. Biochemical and Biophysical Research Communications, 1991, 176, 938-946.	1.0	119
222	Mitochondrial leucine tRNA mutation in a mitochondrial encephalomyopathy. Lancet, The, 1991, 337, 234-235.	6.3	33
223	Mitochondrial DNA mutations in mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS). Biochemical and Biophysical Research Communications, 1991, 174, 861-868.	1.0	89
224	Patients with idiopathic cardiomyopathy belong to the same mitochondrial DNA gene family of Parkinson's disease and mitochondrial encephalomyopathy. Biochemical and Biophysical Research Communications, 1991, 177, 518-525.	1.0	111
225	Direct DNA sequencing from colony: analysis of multiple deletions of mitochondrial genome. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1991, 1090, 9-16.	2.4	7
226	Mitochondrial DNA deletions in inherited recurrent myoglobinuria. Annals of Neurology, 1991, 29, 364-369.	2.8	97
227	Mitochondrial DNA Disease: Phylogeny and Expression. , 1991, , 247-272.		0
228	Mitochondrial mutation in fatal infantile cardiomyopathy. Lancet, The, 1990, 336, 1452.	6.3	175
229	Cytoplasmic body and mitochondrial DNA deletion. Journal of the Neurological Sciences, 1990, 99, 291-300.	0.3	5
230	Increase of deleted mitochondrial DNA in the striatum in Parkinson's disease and senescence. Biochemical and Biophysical Research Communications, 1990, 170, 1044-1048.	1.0	363
231	Quantitative determination of deleted mitochondrial DNA relative to normal DNA in parkinsonian striatum by a kinetic PCR analysis. Biochemical and Biophysical Research Communications, 1990, 172, 483-489.	1.0	186
232	Multiple mitochondrial DNA deletions exist in cardiomyocytes of patients with hypertrophic or dilated cardiomyopathy. Biochemical and Biophysical Research Communications, 1990, 170, 830-836.	1.0	184
233	Mitochondrial DNA Mutations as an Etiology of Human Degenerative Diseases. , 1990, , 413-427.		2
234	S1 Nuclease Analysis and Direct Sequencing of Deleted Mitochondrial DNA in Myopathic Patients: Role of Directly Repeated Sequences in Deletion. , 1990, , 441-449.		0

#	Article	IF	Citations
235	Differently deleted mitochondrial genomes in maternally inherited chronic progressive external ophthalmoplegia. Journal of Inherited Metabolic Disease, 1989, 12, 359-362.	1.7	2
236	Specific amplification of deleted mitochondrial DNA from a myopathic patient and analysis of deleted region with S1 nuclease. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1989, 1009, 151-155.	2.4	14
237	Direct sequencing of deleted mitochondrial DNA in myopathic patients. Biochemical and Biophysical Research Communications, 1989, 164, 156-163.	1.0	55
238	Multiple populations of deleted mitochondrial DNA detected by a novel gene amplification method. Biochemical and Biophysical Research Communications, 1989, 162, 664-672.	1.0	64
239	Maternal inheritance of deleted mitochondrial DNA in a family with mitochondrial myopathy. Biochemical and Biophysical Research Communications, 1988, 154, 1240-1247.	1.0	141
240	Congenital Myasthenic Syndromes - Molecular Bases of Congenital Defects of Proteins at the Neuromuscular Junction. , 0, , .		3
241	Meclozine has a potential effects on short stature and foramen magnum stenosis in transgenic mice with achondroplasia. Bone Abstracts, 0, , .	0.0	1