

Byung-Ok Choi

List of Publications by Year in descending order

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Version: 2024-02-01

127
papers

2,592
citations

172457

29
h-index

276875

41
g-index

135
all docs

135
docs citations

135
times ranked

3993
citing authors

#	ARTICLE	IF	CITATIONS
1	Charcot-Marie-Tooth disease type 2CC due to <i>NEFH</i> variants causes a progressive, non-length-dependent, motor-predominant phenotype. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 48-56.	1.9	9
2	Variants of <i>aminoacyl-tRNA</i> synthetase genes in Charcot-Marie-Tooth disease: A Korean cohort study. <i>Journal of the Peripheral Nervous System</i> , 2022, 27, 38-49.	3.1	7
3	Ultrasound-mediated triboelectric nanogenerator for powering on-demand transient electronics. <i>Science Advances</i> , 2022, 8, eabl8423.	10.3	71
4	Phenotypic heterogeneity in patients with <i>NEFL</i> -related Charcot-Marie-Tooth disease. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1870.	1.2	8
5	Cytokines secreted by mesenchymal stem cells reduce demyelination in an animal model of Charcot-Marie-Tooth disease. <i>Biochemical and Biophysical Research Communications</i> , 2022, 597, 1-7.	2.1	2
6	Wide Phenotypic Spectrum of PNMHH Patients With p.R941L Mutation in <i>MYH14</i> . <i>Journal of</i>		

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19	Gait parameters as tools for analyzing phenotypic alterations of a mouse model of Charcot-Marie-Tooth disease. <i>Animal Cells and Systems</i> , 2021, 25, 11-18.	2.2	6
20	<i>Morc2a</i> p.S87L mutant mice develop peripheral and central neuropathies associated with neuronal DNA damage and apoptosis. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	2.4	5
21	Cerebellar White Matter Abnormalities in Charcot-Marie-Tooth Disease: A Combined Volumetry and Diffusion Tensor Imaging Analysis. <i>Journal of Clinical Medicine</i> , 2021, 10, 4945.	2.4	2
22	Intraepineurial fat quantification and cross-sectional area analysis of the sciatic nerve using MRI in Charcot-Marie-Tooth disease type 1A patients. <i>Scientific Reports</i> , 2021, 11, 21535.	3.3	4
23	Farnesol Ameliorates Demyelinating Phenotype in a Cellular and Animal Model of Charcot-Marie-Tooth Disease Type 1A. <i>Current Issues in Molecular Biology</i> , 2021, 43, 2011-2021.	2.4	3
24	Identification of rare coding variants associated with Kawasaki disease by whole exome sequencing. <i>Genomics and Informatics</i> , 2021, 19, e38.	0.8	3
25	Analyzing clinical and genetic aspects of axonal Charcot-Marie-Tooth disease. <i>Journal of Genetic Medicine</i> , 2021, 18, 83-93.	0.2	0
26	Targeted PMP22 TATA-box editing by CRISPR/Cas9 reduces demyelinating neuropathy of Charcot-Marie-Tooth disease type 1A in mice. <i>Nucleic Acids Research</i> , 2020, 48, 130-140.	14.5	23
27	Human HSPB1 mutation recapitulates features of distal hereditary motor neuropathy (dHMN) in <i>Drosophila</i> . <i>Biochemical and Biophysical Research Communications</i> , 2020, 521, 220-226.	2.1	6
28	Paternal gender specificity and mild phenotypes in Charcot-Marie-Tooth type 1A patients with de novo 17p12 rearrangements. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1380.	1.2	7
29	Wearable hip-assist robot modulates cortical activation during gait in stroke patients: a functional near-infrared spectroscopy study. <i>Journal of NeuroEngineering and Rehabilitation</i> , 2020, 17, 145.	4.6	15
30	A novel histone deacetylase 6 inhibitor improves myelination of Schwann cells in a model of Charcot-Marie-Tooth disease type 1A. <i>British Journal of Pharmacology</i> , 2020, 177, 5096-5113.	5.4	27
31	Loss of function of EBP50 is a new cause of hereditary peripheral neuropathy: EBP50 functions in peripheral nerve system. <i>Glia</i> , 2020, 68, 1794-1809.	4.9	6
32	CEP41-mediated ciliary tubulin glutamylation drives angiogenesis through AURKA-dependent deciliation. <i>EMBO Reports</i> , 2020, 21, e48290.	4.5	23
33	Alanyl-tRNA synthetase 1 (AARS1) gene mutation in a family with intermediate Charcot-Marie-Tooth neuropathy. <i>Genes and Genomics</i> , 2020, 42, 663-672.	1.4	18
34	Gene Therapy Options as New Treatment for Inherited Peripheral Neuropathy. <i>Experimental Neurobiology</i> , 2020, 29, 177-188.	1.6	7
35	Fibulin 5, a human Wharton's jelly-derived mesenchymal stem cells-secreted paracrine factor, attenuates peripheral nervous system myelination defects through the Integrin-RAC1 signaling axis. <i>Stem Cells</i> , 2020, , .	3.2	4
36	Fibulin 5, a human Wharton's jelly-derived mesenchymal stem cells-secreted paracrine factor, attenuates peripheral nervous system myelination defects through the Integrin-RAC1 signaling axis. <i>Stem Cells</i> , 2020, 38, 1578-1593.	3.2	9

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37	Short hairpin RNA treatment improves gait in a mouse model of Charcot-Marie-Tooth disease type 1A. <i>Molecular Medicine Reports</i> , 2020, 22, 4947-4955.	2.4	0

38 A *POLG2* Homozygous Mutation in an Autosomal Recessive Epilepsy Family Without

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55	Association of miR-149 polymorphism with onset age and severity in Charcotâ€“Marieâ€“Tooth disease type 1A. <i>Neuromuscular Disorders</i> , 2018, 28, 502-507.	0.6	19
56	Small heat shock protein B3 (<i>HSPB3</i>) mutation in an axonal Charcotâ€“Marieâ€“Tooth disease family. <i>Journal of the Peripheral Nervous System</i> , 2018, 23, 60-66.	3.1	12
57	Ankle Pathologic Gait Assistance of a Hip Exoskeleton: Simulation and Experiment. <i>IEEE Robotics and Automation Letters</i> , 2018, 3, 2190-2197.	5.1	4
58	Wide phenotypic spectrum in axonal Charcotâ€“Marieâ€“Tooth neuropathy type 2 patients with KIF5A mutations. <i>Genes and Genomics</i> , 2018, 40, 77-84.	1.4	24
59	Application of differentiated human tonsilâ€“derived stem cells to <i>trembler</i> mice. <i>Muscle and Nerve</i> , 2018, 57, 478-486.	2.2	6
60	A Novel Nonsense Mutation in Leucine-Rich, Glioma-Inactivated-1 Gene as the Underlying Cause of		

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73	Gait performance and foot pressure distribution during wearable robot-assisted gait in elderly adults. <i>Journal of NeuroEngineering and Rehabilitation</i> , 2017, 14, 123.	4.6	43
74	Axonal Charcot-Marie-Tooth neuropathy concurrent with distal and proximal weakness by translational elongation of the 3' UTR in <i>NEFH</i> . <i>Journal of the Peripheral Nervous System</i> , 2017, 22, 200-207.	3.1	12
75	A Large Dominant Myotonia Congenita Family with a V1293I Mutation in <i>SCN4A</i> . <i>Journal of</i>		

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91	Patient fibroblasts-derived induced neurons demonstrate autonomous neuronal defects in adult-onset Krabbe disease. <i>Oncotarget</i> , 2016, 7, 74496-74509.	1.8	26
92	Ultrasonography-guided transplantation facilitates perineural delivery of stem cells. <i>Animal Cells and Systems</i> , 2015, 19, 269-273.	2.2	1
93	A family with axonal sensorimotor polyneuropathy with TUBB3 mutation. <i>Molecular Medicine Reports</i> , 2015, 11, 2729-2734.	2.4	13
94	A novel homozygous MPV17 mutation in two families with axonal sensorimotor polyneuropathy. <i>BMC Neurology</i> , 2015, 15, 179.	1.8	17
95	Overexpression of mutant HSP27 causes axonal neuropathy in mice. <i>Journal of Biomedical Science</i> , 2015, 22, 43.	7.0	31
96	Charcot-Marie-Tooth Disease Type 4H Resulting from Compound Heterozygous Mutations in <i>FGD4</i> from Nonconsanguineous Korean Families. <i>Annals of Human Genetics</i> , 2015, 79, 460-469.	0.8	7
97	Novel Compound Heterozygous Nonsense <i>PRX</i> Mutations in a Korean Dejerine-Sottas Neuropathy		

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109	Comprehensive Analysis to Improve the Validation Rate for Single Nucleotide Variants Detected by Next-Generation Sequencing. PLoS ONE, 2014, 9, e86664.	2.5	20
110	A novel Lys141Thr mutation in small heat shock protein 22 (HSPB8) gene in Charcot-Marie-Tooth disease type 2L. Neuromuscular Disorders, 2013, 23, 656-663.	0.6	39
111	Proximal Dominant Hereditary Motor and Sensory Neuropathy With Proximal Dominance Association With Mutation in the TRK-Fused Gene. JAMA Neurology, 2013, 70, 607.	9.0	37
112	Two de novo mutations of MFN2 associated with early-onset Charcot-Marie-Tooth disease type 2A neuropathy. Genes and Genomics, 2012, 34, 653-661.	1.4	3
113	Two novel mutations of <i>GARS</i> in Korean families with distal hereditary motor neuropathy type V. Journal of the Peripheral Nervous System, 2012, 17, 418-421.	3.1	39
114	Exome sequencing is an efficient tool for genetic screening of Charcot-Marie-Tooth Disease. Human Mutation, 2012, 33, 1610-1615.	2.5	94
115	MPZ mutation in an early-onset Charcot-Marie-Tooth disease type 1B family by genome-wide linkage analysis. International Journal of Molecular Medicine, 2011, 28, 389-96.	4.0	6
116	Two recessive intermediate Charcot-Marie-Tooth patients with <i>GDAP1</i> mutations. Journal of the Peripheral Nervous System, 2011, 16, 143-146.	3.1	14
117	Inheritance of Charcot-Marie-Tooth disease 1A with rare nonrecurrent genomic rearrangement. Neurogenetics, 2011, 12, 51-58.	1.4	13
118	Compound mutations of PEO1 and TYMP in a progressive external ophthalmoplegia patient with incomplete mitochondrial neurogastrointestinal encephalomyopathy phenotype. Genes and Genomics, 2011, 33, 431-437.	1.4	2
119	A novel Gly137Asp MPZ mutation in a Charcot-Marie-Tooth disease type 1B family. Genes and Genomics, 2011, 33, 659-664.	1.4	1
120	A complex phenotype of peripheral neuropathy, myopathy, hoarseness, and hearing loss is linked to an autosomal dominant mutation in MYH14. Human Mutation, 2011, 32, 669-677.	2.5	48
121	Wide phenotypic variations in Charcot-Marie-Tooth 1A neuropathy with rare copy number variations on 17p12. Animal Cells and Systems, 2011, 15, 301-309.	2.2	2
122	Mutational analysis of whole mitochondrial DNA in patients with MELAS and MERRF diseases. Experimental and Molecular Medicine, 2010, 42, 446.	7.7	22
123	A novel GDAP1 Q218E mutation in autosomal dominant Charcot-Marie-Tooth disease. Journal of Human Genetics, 2008, 53, 360-364.	2.3	52
124	NEFL Pro22Arg mutation in Charcot-Marie-Tooth disease type 1. Journal of Human Genetics, 2008, 53, 936-940.	2.3	34
125	Distal hereditary motor neuropathy in Korean patients with a small heat shock protein 27 mutation. Experimental and Molecular Medicine, 2008, 40, 304.	7.7	31
126	Mutational analysis of PMP22, MPZ, GJB1, EGR2 and NEFL in Korean Charcot-Marie-Tooth neuropathy patients. Human Mutation, 2004, 24, 185-186.	2.5	116

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127	Identification and clinical characterization of Charcot-Marie-Tooth disease type 1C patients with LITAF p.G112S mutation. <i>Genes and Genomics</i> , 0, , .	1.4	2