

# Byung-Ok Choi

## List of Publications by Year in descending order

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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
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1	Novel Compound Heterozygous Nonsense <i>PRX</i> Mutations in a Korean Dejerine-Sottas Neuropathy		
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#	ARTICLE	IF	CITATIONS
19	A novel Lys141Thr mutation in small heat shock protein 22 (HSPB8) gene in Charcot-Marie-Tooth disease type 2L. <i>Neuromuscular Disorders</i> , 2013, 23, 656-663.	0.6	39
20	Anti-apoptotic Effects of Human Wharton's Jelly-derived Mesenchymal Stem Cells on Skeletal Muscle Cells Mediated via Secretion of XCL1. <i>Molecular Therapy</i> , 2016, 24, 1550-1560.	8.2	39
21	Proximal Dominant Hereditary Motor and Sensory Neuropathy With Proximal Dominance Association With Mutation in the TRK-Fused Gene. <i>JAMA Neurology</i> , 2013, 70, 607.	9.0	37
22	Clinical and genetic analysis of MAPT, GRN, and C9orf72 genes in Korean patients with frontotemporal dementia. <i>Neurobiology of Aging</i> , 2014, 35, 1213.e13-1213.e17.	3.1	35
23	Identification of FASTKD2 compound heterozygous mutations as the underlying cause of autosomal recessive MELAS-like syndrome. <i>Mitochondrion</i> , 2017, 35, 54-58.	3.4	35
24	A multicenter retrospective study of charcot-Marie-Tooth disease type 4B (CMT4B) associated with mutations in myotubularin-related proteins (MTMRs). <i>Annals of Neurology</i> , 2019, 86, 55-67.	5.3	35
25	NEFL Pro22Arg mutation in Charcot-Marie-Tooth disease type 1. <i>Journal of Human Genetics</i> , 2008, 53, 936-940.	2.3	34
26	Pmp22 mutant allele-specific siRNA alleviates demyelinating neuropathic phenotype in vivo. <i>Neurobiology of Disease</i> , 2017, 100, 99-107.	4.4	33
27	Variation in <i>SIPA1L2</i> is correlated with phenotype modification in Charcot-Marie-Tooth disease type 1A. <i>Annals of Neurology</i> , 2019, 85, 316-330.	5.3	33
28	<i>ADSSL1</i> mutation relevant to autosomal recessive adolescent onset distal myopathy. <i>Annals of Neurology</i> , 2016, 79, 231-243.	5.3	32
29	Distal hereditary motor neuropathy in Korean patients with a small heat shock protein 27 mutation. <i>Experimental and Molecular Medicine</i> , 2008, 40, 304.	7.7	31
30	Overexpression of mutant HSP27 causes axonal neuropathy in mice. <i>Journal of Biomedical Science</i> , 2015, 22, 43.	7.0	31
31	A wearable hip-assist robot reduces the cardiopulmonary metabolic energy expenditure during stair ascent in elderly adults: a pilot cross-sectional study. <i>BMC Geriatrics</i> , 2018, 18, 230.	2.7	29
32	Clinico-genetics in Korean Charcot-Marie-Tooth disease type 2Z with <i>MORC2</i> mutations. <i>Brain</i> , 2016, 139, e40-e40.	7.6	28
33	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
34	Patient fibroblasts-derived induced neurons demonstrate autonomous neuronal defects in adult-onset Krabbe disease. <i>Oncotarget</i> , 2016, 7, 74496-74509.	1.8	26
35	Identification of mutations in Korean patients with amyotrophic lateral sclerosis using multigene panel testing. <i>Neurobiology of Aging</i> , 2016, 37, 209.e9-209.e16.	3.1	25
36	Wide phenotypic spectrum in axonal Charcot-Marie-Tooth neuropathy type 2 patients with KIF5A mutations. <i>Genes and Genomics</i> , 2018, 40, 77-84.	1.4	24

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37	miR-381 Attenuates Peripheral Neuropathic Phenotype Caused by Overexpression of PMP22. <i>Experimental Neurobiology</i> , 2019, 28, 279-288.	1.6	24
38	Amyloid Beta-Mediated Hypomethylation of Heme Oxygenase 1 Correlates with Cognitive Impairment in Alzheimer's Disease. <i>PLoS ONE</i> , 2016, 11, e0153156.	2.5	23
39	Age-related differences in muscle co-activation during locomotion and their relationship with gait speed: a pilot study. <i>BMC Geriatrics</i> , 2017, 17, 44.	2.7	23
40	Cerebral white matter abnormalities in patients with Charcot-Marie-Tooth disease. <i>Annals of Neurology</i> , 2017, 81, 147-151.	5.3	23
41	Hidden hearing loss in patients with Charcot-Marie-Tooth disease type 1A. <i>Scientific Reports</i> , 2018, 8, 10335.	3.3	23
42	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
43	CEP41-mediated ciliary tubulin glutamylation drives angiogenesis through AURKA-dependent deciliation. <i>EMBO Reports</i> , 2020, 21, e48290.	4.5	23
44	Mutational analysis of whole mitochondrial DNA in patients with MELAS and MERRF diseases. <i>Experimental and Molecular Medicine</i> , 2010, 42, 446.	7.7	22
45	Early-onset severe hereditary sensory and autonomic neuropathy type 1 with S331F SPTLC1 mutation. <i>Molecular Medicine Reports</i> , 2014, 9, 481-486.	2.4	22
46	Haplotype-specific modulation of a SOX10/CREB response element at the Charcot-Marie-Tooth disease type 4C locus SH3TC2. <i>Human Molecular Genetics</i> , 2014, 23, 5171-5187.	2.9	21
47	Comprehensive Analysis to Improve the Validation Rate for Single Nucleotide Variants Detected by Next-Generation Sequencing. <i>PLoS ONE</i> , 2014, 9, e86664.	2.5	20
48	Association of miR-149 polymorphism with onset age and severity in Charcot-Marie-Tooth disease type 1A. <i>Neuromuscular Disorders</i> , 2018, 28, 502-507.	0.6	19
49	Modifier Gene Candidates in Charcot-Marie-Tooth Disease Type 1A: A Case-Only Genome-Wide Association Study. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 201-211.	2.6	19
50	p75 and neural cell adhesion molecule 1 can identify pathologic Schwann cells in peripheral neuropathies. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1292-1301.	3.7	18
51	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
52	Clinical and Genetic Aspects in Twelve Korean Patients with Adrenomyeloneuropathy. <i>Yonsei Medical Journal</i> , 2014, 55, 676.	2.2	17
53	Clinical and Neuropsychological Characteristics of a Nationwide Hospital-Based Registry of Frontotemporal Dementia Patients in Korea: A CREDOS-FTD Study. <i>Dementia and Geriatric Cognitive Disorders Extra</i> , 2014, 4, 242-251.	1.3	17
54	A novel homozygous MPV17 mutation in two families with axonal sensorimotor polyneuropathy. <i>BMC Neurology</i> , 2015, 15, 179.	1.8	17

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55	Autophagy induction in the skeletal myogenic differentiation of human tonsil-derived mesenchymal stem cells. <i>International Journal of Molecular Medicine</i> , 2017, 39, 831-840.	4.0	17
56	BAG3 mutation in a patient with atypical phenotypes of myofibrillar myopathy and Charcot-Marie-Tooth disease. <i>Genes and Genomics</i> , 2018, 40, 1269-1277.	1.4	17
57	Diffusion tensor imaging of the sciatic nerve in Charcot-Marie-Tooth disease type I patients: a prospective case-control study. <i>European Radiology</i> , 2019, 29, 3241-3252.	4.5	17
58	Muscle fat quantification using magnetic resonance imaging: case-control study of Charcot-Marie-Tooth disease patients and volunteers. <i>Journal of Cachexia, Sarcopenia and Muscle</i> , 2019, 10, 574-585.	7.3	16
59	Distal myopathy with ADSSL1 mutations in Korean patients. <i>Neuromuscular Disorders</i> , 2017, 27, 465-472.	0.6	15
60	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
61	Two recessive intermediate Charcot-Marie-Tooth patients with <i>GDAP1</i> mutations. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 143-146.	3.1	14
62	Recessive optic atrophy, sensorimotor neuropathy and cataract associated with novel compound heterozygous mutations in OPA1. <i>Molecular Medicine Reports</i> , 2016, 14, 33-40.	2.4	14
63	Nanotopographical regulation of pancreatic islet-like cluster formation from human pluripotent stem cells using a gradient-pattern chip. <i>Acta Biomaterialia</i> , 2019, 95, 337-347.	8.3	14
64	Inheritance of Charcot-Marie-Tooth disease 1A with rare nonrecurrent genomic rearrangement. <i>Neurogenetics</i> , 2011, 12, 51-58.	1.4	13
65	A family with axonal sensorimotor polyneuropathy with TUBB3 mutation. <i>Molecular Medicine Reports</i> , 2015, 11, 2729-2734.	2.4	13
66	Human Induced Pluripotent Stem Cell-Derived TDP-43 Mutant Neurons Exhibit Consistent Functional Phenotypes Across Multiple Gene Edited Lines Despite Transcriptomic and Splicing Discrepancies. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 728707.	3.7	13
67	Distal hereditary motor neuropathy type 7B with Dynactin 1 mutation. <i>Molecular Medicine Reports</i> , 2016, 14, 3362-3368.	2.4	12
68	X-linked Charcot-Marie-Tooth disease type 6 ( <i>CMTX6</i> ) patients with a p.R158H mutation in the pyruvate dehydrogenase kinase isoenzyme 3 gene. <i>Journal of the Peripheral Nervous System</i> , 2016, 21, 45-51.	3.1	12
69	Small heat shock protein B3 ( <i>HSPB3</i> ) mutation in an axonal Charcot-Marie-Tooth disease family. <i>Journal of the Peripheral Nervous System</i> , 2018, 23, 60-66.	3.1	12
70	Linkage analysis and whole exome sequencing reveals AHNK2 as a novel genetic cause for autosomal recessive CMT in a Malaysian family. <i>Neurogenetics</i> , 2019, 20, 117-127.	1.4	12
71	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
72	HDAC6 Inhibition Corrects Electrophysiological and Axonal Transport Deficits in a Human Stem Cell-Based Model of Charcot-Marie-Tooth Disease (Type 2D). <i>Advanced Biology</i> , 2022, 6, e2101308.	2.5	12

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73	Differentiation of Human Tonsil-Derived Mesenchymal Stem Cells into Schwann-Like Cells Improves Neuromuscular Function in a Mouse Model of Charcot-Marie-Tooth Disease Type 1A. <i>International Journal of Molecular Sciences</i> , 2018, 19, 2393.	4.1	11
74	Clinical characterization and genetic analysis of Korean patients with X-linked Charcot-Marie-Tooth disease type 1. <i>Journal of the Peripheral Nervous System</i> , 2017, 22, 172-181.	3.1	9
75	Zebrafish is a central model to dissect the peripheral neuropathy. <i>Genes and Genomics</i> , 2019, 41, 993-1000.	1.4	9
76	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
77	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
78	Virus blocking textile for SARS-CoV-2 using human body triboelectric energy harvesting. <i>Cell Reports Physical Science</i> , 2022, 3, 100813.	5.6	9
79	Severe phenotypes in a Charcot-Marie-Tooth 1A patient with PMP22 triplication. <i>Journal of Human Genetics</i> , 2015, 60, 103-106.	2.3	8
80	Compound heterozygous mutations of SH3TC2 in Charcot-Marie-Tooth disease type 4C patients. <i>Journal of Human Genetics</i> , 2019, 64, 961-965.	2.3	8
81	Clinical and Neuroimaging Features in Charcot-Marie-Tooth Patients with <i>GDAP1</i> Mutations.		

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91	Texture analysis using T1-weighted images for muscles in Charcot-Marie-Tooth disease patients and volunteers. <i>European Radiology</i> , 2021, 31, 3508-3517.	4.5	6
92	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
93	Peripheral Myelin Protein 22 Gene Mutations in Charcot-Marie-Tooth Disease Type 1E Patients. <i>Genes</i> , 2022, 13, 1219.	2.4	6
94	Ser135Phe mutation in HSPB1 (HSP27) from Charcot-Marie-Tooth disease type 2F families. <i>Genes and Genomics</i> , 2015, 37, 295-303.	1.4	5
95	Serum CXCL13 reflects local B-cell mediated inflammatory demyelinating peripheral neuropathy. <i>Scientific Reports</i> , 2019, 9, 16535.	3.3	5
96	Genetic and clinical spectrums in Korean Charcot-Marie-Tooth disease patients with myelin protein zero mutations. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1678.	1.2	5
97	<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
98	Genetic and Clinical Studies of Peripheral Neuropathies with Three Small Heat Shock Protein Gene Variants in Korea. <i>Genes</i> , 2022, 13, 462.	2.4	5
99	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
100	A <i>POLG2</i> Homozygous Mutation in an Autosomal Recessive Epilepsy Family Without		

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109	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
110	Identification of rare coding variants associated with Kawasaki disease by whole exome sequencing. <i>Genomics and Informatics</i> , 2021, 19, e38.	0.8	3
111	Compound mutations of PEO1 and TYMP in a progressive external ophthalmoplegia patient with incomplete mitochondrial neurogastrointestinal encephalomyopathy phenotype. <i>Genes and Genomics</i> , 2011, 33, 431-437.	1.4	2
112	Wide phenotypic variations in Charcot-Marie-Tooth 1A neuropathy with rare copy number variations on 17p12. <i>Animal Cells and Systems</i> , 2011, 15, 301-309.	2.2	2
113	Dynamic Transcriptional Events in Distal Sural Nerve Revealed by Transcriptome Analysis. <i>Experimental Neurobiology</i> , 2014, 23, 169-172.	1.6	2
114	Clinical and Neuroimaging Features in Charcot-Marie-Tooth Patients with GNB4 Mutations. <i>Life</i> , 2021, 11, 494.	2.4	2
115	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
116	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
117	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
118	A novel Gly137Asp MPZ mutation in a Charcot-Marie-Tooth disease type 1B family. <i>Genes and Genomics</i> , 2011, 33, 659-664.	1.4	1
119	Ultrasonography-guided transplantation facilitates perineural delivery of stem cells. <i>Animal Cells and Systems</i> , 2015, 19, 269-273.	2.2	1
120	Replication studies of MIR149 association in Charcot-Marie-Tooth disease type 1A in a European population - response. <i>Neuromuscular Disorders</i> , 2019, 29, 160-162.	0.6	1
121	Development of cell models for high-throughput screening system of Charcot-Marie-Tooth disease type 1. <i>Journal of Genetic Medicine</i> , 2015, 12, 25-30.	0.2	1
122	Wide Phenotypic Spectrum of PNMHH Patients With p.R941L Mutation in <i>MYH14</i> . <i>Journal of</i>		



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127	Analyzing clinical and genetic aspects of axonal Charcot-Marie-Tooth disease. Journal of Genetic Medicine, 2021, 18, 83-93.	0.2	0