# Vincent Timmerman

List of Publications by Year
in descending order

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|---|---|---|---|
| 188 papers | 16,439 citations | 16411 64 h-index | 17546 121 g-index |
| 194 all docs | 194 docs citations | 194 times ranked | 15625 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nature Genetics, 2004, 36, 449-451. | 9.4 | 1,391 |
| 2 | The hnRNP family: insights into their role in health and disease. Human Genetics, 2016, 135, 851-867. | 1.8 | 720 |
| 3 | DNA/RNA Helicase Gene Mutations in a Form of Juvenile Amyotrophic Lateral Sclerosis (ALS4). American Journal of Human Genetics, 2004, 74, 1128-1135. | 2.6 | 717 |
| 4 | Mutant small heat-shock protein 27 causes axonal Charcot-Marie-Tooth disease and distal hereditary motor neuropathy. Nature Genetics, 2004, 36, 602-606. | 9.4 | 541 |
| 5 | Mutations in the Small GTP-ase Late Endosomal Protein RAB7 Cause Charcot-Marie-Tooth Type 2B Neuropathy. American Journal of Human Genetics, 2003, 72, 722-727. | 2.6 | 415 |
| 6 | HDAC6 inhibitors reverse axonal loss in a mouse model of mutant HSPB1â€“induced Charcot-Marie-Tooth disease. Nature Medicine, 2011, 17, 968-974. | 15.2 | 405 |
| 7 | The peripheral myelin protein gene PMPâ€“22 is contained within the Charcotâ€“Marieâ€“Tooth disease type 1A duplication. Nature Genetics, 1992, 1, 171-175. | 9.4 | 404 |
| 8 | Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. Nature Genetics, 2004, 36, 597-601. | 9.4 | 395 |
| 9 | Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. Annals of Neurology, 2006, 59, 276-281. | 2.8 | 380 |
| 10 | MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. Brain, 2006, 129, 2093-2102. | 3.7 | 351 |
| 11 | Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276. | 9.4 | 349 |
| 12 | Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. Nature Genetics, 2005, 37, 289-294. | 9.4 | 324 |
| 13 | Disrupted function and axonal distribution of mutant tyrosyl-tRNA synthetase in dominant intermediate Charcot-Marie-Tooth neuropathy. Nature Genetics, 2006, 38, 197-202. | 9.4 | 323 |
| 14 | A de novo gain-of-function mutation in SCN11A causes loss of pain perception. Nature Genetics, 2013, 45, 1399-1404. | 9.4 | 264 |
| 15 | Mutations in the neurofilament light chain gene (NEFL) cause early onset severe Charcot-Marie-Tooth disease. Brain, 2003, 126, 590-597. | 3.7 | 259 |
| 16 | Mutations in SEPT9 cause hereditary neuralgic amyotrophy. Nature Genetics, 2005, 37, 1044-1046. | 9.4 | 222 |
| 17 | Mutations in FAM134B, encoding a newly identified Golgi protein, cause severe sensory and autonomic neuropathy. Nature Genetics, 2009, 41, 1179-1181. | 9.4 | 205 |
| 18 | Genetics of Charcot-Marie-Tooth (CMT) Disease within the Frame of the Human Genome Project Success. Genes, 2014, 5, 13-32. | 1.0 | 203 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Genes for hereditary sensory and autonomic neuropathies: a genotype-phenotype correlation. Brain, 2009, 132, 2699-2711. | 3.7 | 202 |
| 20 | Astrocytes regulate GluR2 expression in motor neurons and their vulnerability to excitotoxicity. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 14825-14830. | 3.3 | 193 |
| 21 | Further evidence that neurofilament light chain gene mutations can cause Charcot-Marie-Tooth disease type 2E. Annals of Neurology, 2001, 49, 245-249. | 2.8 | 188 |
| 22 | Mutations in a Gene Encoding a Novel SH3/TPR Domain Protein Cause Autosomal Recessive Charcot-Marie-Tooth Type 4C Neuropathy. American Journal of Human Genetics, 2003, 73, 1106-1119. | 2.6 | 185 |
| 23 | KIF1A, an Axonal Transporter of Synaptic Vesicles, Is Mutated in Hereditary Sensory and Autonomic Neuropathy Type 2. American Journal of Human Genetics, 2011, 89, 219-230. | 2.6 | 172 |
| 24 | Mitochondria-associated membranes as hubs for neurodegeneration. Acta Neuropathologica, 2016, 131, 505-523. | 3.9 | 172 |
| 25 | Missense Mutations in the Copper Transporter Gene ATP7A Cause X-Linked Distal Hereditary Motor Neuropathy. American Journal of Human Genetics, 2010, 86, 343-352. | 2.6 | 170 |
| 26 | Mutations in the SPTLC2 Subunit of Serine Palmitoyltransferase Cause Hereditary Sensory and Autonomic Neuropathy Type I. American Journal of Human Genetics, 2010, 87, 513-522. | 2.6 | 159 |
| 27 | Profiling peripheral nerve macrophages reveals two macrophage subsets with distinct localization, transcriptome and response to injury. Nature Neuroscience, 2020, 23, 676-689. | 7.1 | 148 |
| 28 | The phenotype of motor neuropathies associated with BSCL2 mutations is broader than Silver syndrome and distal HMN type V. Brain, 2004, 127, 2124-2130. | 3.7 | 146 |
| 29 | Mechanisms of disease in hereditary sensory and autonomic neuropathies. Nature Reviews Neurology, 2012, 8, 73-85. | 4.9 | 140 |
| 30 | Mutational analysis of the MPZ, PMP22 and Cx32 genes in patients of Spanish ancestry with Charcot-Marie-Tooth disease and hereditary neuropathy with liability to pressure palsies. Human Genetics, 1997, 99, 746-754. | 1.8 | 137 |
| 31 | Charcotâ€Marieâ€Tooth Disease: A Clinicoâ€genetic Confrontation. Annals of Human Genetics, 2008, 72, 416-441. | 0.3 | 136 |
| 32 | Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. Brain, 2009, 132, 1741-1752. | 3.7 | 134 |
| 33 | Acute injury in the peripheral nervous system triggers an alternative macrophage response. Journal of Neuroinflammation, 2012, 9, 176. | 3.1 | 134 |
| 34 | Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. American Journal of Human Genetics, 2007, 81, 158-164. | 2.6 | 128 |
| 35 | Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability. American Journal of Human Genetics, 2010, 86, 892-903. | 2.6 | 125 |
| 36 | Estimation of the size of the chromosome 17p11.2 duplication in Charcot-Marie-Tooth neuropathy type 1a (CMT1a). HMSN Collaborative Research Group.. Journal of Medical Genetics, 1992, 29, 5-11. | 1.5 | 123 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 37 | Tollâ€like receptor expression in the peripheral nerve. Glia, 2010, 58, 1701-1709. | 2.5 | 121 |
| 38 | Relative contribution of mutations in genes for autosomal dominant distal hereditary motor neuropathies: a genotype-phenotype correlation study. Brain, 2007, 131, 1217-1227. | 3.7 | 113 |
| 39 | Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. Brain, 2010, 133, 1798-1809. | 3.7 | 113 |
| 40 | Genetic spectrum of hereditary neuropathies with onset in the first year of life. Brain, 2011, 134, 2664-2676. | 3.7 | 112 |
| 41 | Molecular Defects in the Motor Adaptor BICD2 Cause Proximal Spinal Muscular Atrophy with Autosomal-Dominant Inheritance. American Journal of Human Genetics, 2013, 92, 955-964. | 2.6 | 112 |
| 42 | Rapid screening of myelin genes in CMT1 patients by SSCP analysis: identification of new mutations and polymorphisms in the P0 gene. Human Genetics, 1994, 94, 653-657. | 1.8 | 110 |
| 43 | Slowed Conduction and Thin Myelination of Peripheral Nerves Associated with Mutant Rho Guanine-Nucleotide Exchange Factor 10. American Journal of Human Genetics, 2003, 73, 926-932. | 2.6 | 107 |
| 44 | Periaxin mutations cause a broad spectrum of demyelinating neuropathies. Annals of Neurology, 2002, 51, 709-715. | 2.8 | 106 |
| 45 | Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. Nature Genetics, 2012, 44, 1080-1083. | 9.4 | 102 |
| 46 | The neuroinflammatory role of Schwann cells in disease. Neurobiology of Disease, 2013, 55, 95-103. | 2.1 | 97 |
| 47 | Dominant mutations in the tyrosyl-tRNA synthetase gene recapitulate in Drosophila features of human Charcotâ€“Marieâ€“Tooth neuropathy. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 11782-11787. | 3.3 | 96 |
| 48 | Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250. | 3.7 | 96 |
| 49 | Increased Monomerization of Mutant HSPB1 Leads to Protein Hyperactivity in Charcot-Marie-Tooth Neuropathy. Journal of Biological Chemistry, 2010, 285, 12778-12786. | 1.6 | 95 |
| 50 | Small Heat-Shock Protein HSPB1 Mutants Stabilize Microtubules in Charcot-Marie-Tooth Neuropathy. Journal of Neuroscience, 2011, 31, 15320-15328. | 1.7 | 95 |
| 51 | 2nd Workshop of the European CMT Consortium: 53rd ENMC International Workshop on Classification and Diagnostic Guidelines for Charcot-Marie-Tooth Type 2 (CMT2â€“HMSN II) and Distal Hereditary Motor Neuropathy (Distal HMNâ€“Spinal CMT). Neuromuscular Disorders, 1998, 8, 426-431. | 0.3 | 89 |
| 52 | Mutant HSPB8 causes motor neuron-specific neurite degeneration. Human Molecular Genetics, 2010, 19, 3254-3265. | 1.4 | 83 |
| 53 | Exome Sequencing Identifies a REEP1 Mutation Involved in Distal Hereditary Motor Neuropathy Type V. American Journal of Human Genetics, 2012, 91, 139-145. | 2.6 | 83 |
| 54 | Sensory neuropathy with bone destruction due to a mutation in the membrane-shaping atlastin GTPase 3. Brain, 2014, 137, 683-692. | 3.7 | 80 |

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|---|---|---|---|
| 55 | Screening for mutations in the peripheral myelin genesPMP22,MPZ andCx32 (GJB1) in Russian Charcot-Marie-Tooth neuropathy patients. Human Mutation, 2000, 15, 340-347. | 1.1 | 78 |
| 56 | Clinicopathological and genetic study of early-onset demyelinating neuropathy. Brain, 2004, 127, 2540-2550. | 3.7 | 76 |
| 57 | Charcotâ€Marieâ€Tooth Neuropathy Type 2 and P0 Point Mutations: Two Novel Amino Acid Substitutions (Asp61Gly; Tyr119Cys) and a Possible â€œHotspotâ€•on Thr124Met. Brain Pathology, 2000, 10, 235-248. | 2.1 | 74 |
| 58 | Autosomal dominant juvenile amyotrophic lateral sclerosis and distal hereditary motor neuronopathy with pyramidal tract signs: synonyms for the same disorder?. Brain, 2002, 125, 1320-1325. | 3.7 | 74 |
| 59 | Autosomal Dominant Inherited Neuropathies With Prominent Sensory Loss and Mutilations. Archives of Neurology, 2003, 60, 329. | 4.9 | 74 |
| 60 | Altered interplay between endoplasmic reticulum and mitochondria in Charcotâ€“Marieâ€“Tooth type 2A neuropathy. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 2328-2337. | 3.3 | 73 |
| 61 | Molecular genetics of distal hereditary motor neuropathies. Human Molecular Genetics, 2004, 13, R195-R202. | 1.4 | 71 |
| 62 | Juvenile open angle glaucoma: fine mapping of the TIGR gene to 1q24.3-q25.2 and mutation analysis. Human Genetics, 1998, 102, 103-106. | 1.8 | 67 |
| 63 | SIMPLE mutation in demyelinating neuropathy and distribution in sciatic nerve. Annals of Neurology, 2004, 55, 713-720. | 2.8 | 67 |
| 64 | Distal hereditary motor neuropathy type II (distal HMN II): mapping of a locus to chromosome 12q24. Human Molecular Genetics, 1996, 5, 1065-1069. | 1.4 | 66 |
| 65 | A new locus for autosomal dominant Charcot-Marie-Tooth disease type 2 (CMT2F) maps to chromosome 7q11-q21. European Journal of Human Genetics, 2001, 9, 646-650. | 1.4 | 66 |
| 66 | Overlapping molecular pathological themes link Charcotâ€“Marieâ€“Tooth neuropathies and hereditary spastic paraplegias. Experimental Neurology, 2013, 246, 14-25. | 2.0 | 64 |
| 67 | Challenges in modelling the Charcot-Marie-Tooth neuropathies for therapy development. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 58-67. | 0.9 | 61 |
| 68 | Identification of a 5' splice site mutation in the PMP-22 gene in autosomal dominant Charcotâ€”Marieâ€” Tooth disease type 1. Human Molecular Genetics, 1994, 3, 515-516. | 1.4 | 60 |
| 69 | Localization of the Gene for the Intermediate Form of Charcot-Marie-Tooth to Chromosome 10q24.1-q25.1. American Journal of Human Genetics, 2001, 69, 889-894. | 2.6 | 60 |
| 70 | Recent advances in Charcotâ€“Marieâ€“Tooth disease. Current Opinion in Neurology, 2014, 27, 532-540. | 1.8 | 60 |
| 71 | Dominant Intermediate Charcot-Marie-Tooth Type C Maps to Chromosome 1p34-p35. American Journal of Human Genetics, 2003, 73, 1423-1430. | 2.6 | 58 |
| 72 | A knock-in/knock-out mouse model of HSPB8-associated distal hereditary motor neuropathy and myopathy reveals toxic gain-of-function of mutant Hspb8. Acta Neuropathologica, 2018, 135, 131-148. | 3.9 | 58 |

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|---|---|---|---|
| 73 | Unraveling the genetics of distal hereditary motor neuronopathies. NeuroMolecular Medicine, 2006, 8, 131-146. | 1.8 | 57 |
| 74 | Small heat shock proteins in neurodegenerative diseases. Cell Stress and Chaperones, 2020, 25, 679-699. | 1.2 | 57 |
| 75 | Recent advances in hereditary sensory and autonomic neuropathies. Current Opinion in Neurology, 2006, 19, 474-480. | 1.8 | 56 |
| 76 | Neuropathy-causing mutations in HSPB1 impair autophagy by disturbing the formation of SQSTM1/p62 bodies. Autophagy, 2019, 15, 1051-1068. | 4.3 | 56 |
| 77 | Axonal Neuropathies due to Mutations in Small Heat Shock Proteins: Clinical, Genetic, and Functional Insights into Novel Mutations. Human Mutation, 2017, 38, 556-568. | 1.1 | 54 |
| 78 | Mutation Scanning the GJB1 Gene with High-Resolution Melting Analysis: Implications for Mutation Scanning of Genes for Charcot-Marie-Tooth Disease. Clinical Chemistry, 2007, 53, 349-352. | 1.5 | 53 |
| 79 | Detection of the CMT1A/HNPP recombination hotspot in unrelated patients of European descent.. Journal of Medical Genetics, 1997, 34, 43-49. | 1.5 | 52 |
| 80 | Hereditary sensory neuropathy is caused by a mutation in the delta subunit of the cytosolic chaperonin-containing t-complex peptide-1 (Cct4 ) gene. Human Molecular Genetics, 2003, 12, 1917-1925. | 1.4 | 51 |
| 81 | Whole-exome sequencing in patients with inherited neuropathies: outcome and challenges. Journal of Neurology, 2014, 261, 970-982. | 1.8 | 50 |
| 82 | Identification of novel GDAP1 mutations causing autosomal recessive Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2003, 13, 720-728. | 0.3 | 49 |
| 83 | Small heat shock proteins in inherited peripheral neuropathies. Annals of Medicine, 2005, 37, 413-422. | 1.5 | 49 |
| 84 | Genetic variant in theHSPB1 promoter region impairs the HSP27 stress response. Human Mutation, 2007, 28, 830-830. | 1.1 | 47 |
| 85 | Refinement of the hereditary neuralgic amyotrophy (HNA) locus to chromosome 17q24-q25. Human Genetics, 1997, 99, 685-687. | 1.8 | 46 |
| 86 | Molecular Chaperones in the Pathogenesis of Amyotrophic Lateral Sclerosis: The Role of HSPB1. Human Mutation, 2016, 37, 1202-1208. | 1.1 | 45 |
| 87 | Charcotâ€“Marieâ€“Tooth causing HSPB1 mutations increase Cdk5-mediated phosphorylation of neurofilaments. Acta Neuropathologica, 2013, 126, 93-108. | 3.9 | 43 |
| 88 | Nlrp6 promotes recovery after peripheral nerve injury independently of inflammasomes. Journal of Neuroinflammation, 2015, 12, 143. | 3.1 | 42 |
| 89 | Linkage analysis of distal hereditary motor neuropathy type II (distal HMN II) in a single pedigree. Journal of the Neurological Sciences, 1992, 109, 41-48. | 0.3 | 41 |
| 90 | Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708. | 5.8 | 40 |

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|---|---|---|---|
| 91 | Experimental Charcotâ€“Marieâ€“Tooth type 1A: A cDNA microarrays analysis. Molecular and Cellular Neurosciences, 2005, 28, 703-714. | 1.0 | 39 |
| 92 | Neurofilament phosphorylation and their prolineâ€directed kinases in health and disease. Journal of the Peripheral Nervous System, 2012, 17, 365-376. | 1.4 | 38 |
| 93 | Clinical and Electrophysiological Features in Charcot-Marie-Tooth Disease With Mutations in the NEFL Gene. Archives of Neurology, 2007, 64, 966. | 4.9 | 37 |
| 94 | Characterization of two mutations in the SPTLC1 subunit of serine palmitoyltransferase associated with hereditary sensory and autonomic neuropathy type I. Human Mutation, 2011, 32, E2211-E2225. | 1.1 | 37 |
| 95 | A novel homozygous missense mutation in the myotubularin-related protein 2 gene associated with recessive Charcotâ€“Marieâ€“Tooth disease with irregularly folded myelin sheaths. Neuromuscular Disorders, 2002, 12, 869-873. | 0.3 | 36 |
| 96 | Novel Mutations in the DYNC1H1 Tail Domain Refine the Genetic and Clinical Spectrum of Dyneinopathies. Human Mutation, 2015, 36, 287-291. | 1.1 | 36 |
| 97 | Disease mechanisms in hereditary sensory and autonomic neuropathies. Neurobiology of Disease, 2006, 21, 247-255. | 2.1 | 35 |
| 98 | Magnetic resonance imaging findings of leg musculature in Charcot-Marie-Tooth disease type 2 due to dynamin 2 mutation. Journal of Neurology, 2008, 255, 986-992. | 1.8 | 35 |
| 99 | Autosomal-Dominant Striatal Degeneration Is Caused by a Mutation in the Phosphodiesterase 8B Gene. American Journal of Human Genetics, 2010, 86, 83-87. | 2.6 | 35 |
| 100 | Mutations at Ser331 in the HSN type I gene SPTLC1 are associated with a distinct syndromic phenotype. European Journal of Medical Genetics, 2013, 56, 266-269. | 0.7 | 35 |
| 101 | Genetic refinement of the hereditary neuralgic amyotrophy (HNA) locus at chromosome 17q25. European Journal of Human Genetics, 1999, 7, 920-927. | 1.4 | 34 |
| 102 | Molecular genetics and biology of inherited peripheral neuropathies: a fast-moving field. Neurogenetics, 1999, 2, 137-148. | 0.7 | 34 |
| 103 | CMT-associated mutations in glycyl- and tyrosyl-tRNA synthetases exhibit similar pattern of toxicity and share common genetic modifiers in Drosophila. Neurobiology of Disease, 2014, 68, 180-189. | 2.1 | 34 |
| 104 | HSPB1 facilitates ERK-mediated phosphorylation and degradation of BIM to attenuate endoplasmic reticulum stress-induced apoptosis. Cell Death and Disease, 2017, 8, e3026-e3026. | 2.7 | 33 |
| 105 | Mutation analysis of the connexin 32 (Cx32) gene in charcot-marie-tooth neuropathy type 1: Identification of five new mutations. , 1997, 9, 47-52. | | 32 |
| 106 | Microtubule dynamics in the peripheral nervous system. Bioarchitecture, 2011, 1, 267-270. | 1.5 | 32 |
| 107 | Novel insights in the disease biology of mutant small heat shock proteins in neuromuscular diseases. Brain, 2017, 140, 2541-2549. | 3.7 | 32 |
| 108 | BAG3 Pro209 mutants associated with myopathy and neuropathy relocate chaperones of the CASA-complex to aggresomes. Scientific Reports, 2020, 10, 8755. | 1.6 | 32 |

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|---|---|---|---|
| 109 | Autophagy as an Emerging Common Pathomechanism in Inherited Peripheral Neuropathies. Frontiers in Molecular Neuroscience, 2017, 10, 143. | 1.4 | 31 |
| 110 | Sensory neuropathy-causing mutations in ATL3 affect ERâ€“mitochondria contact sites and impair axonal mitochondrial distribution. Human Molecular Genetics, 2019, 28, 615-627. | 1.4 | 31 |
| 111 | Linkage and mutation analysis in an extended family with Charcot-Marie-Tooth disease type 1B.. Journal of Medical Genetics, 1994, 31, 811-815. | 1.5 | 30 |
| 112 | Diagnostic guidelines for hereditary neuralgic amyotrophy or heredofamilial neuritis with brachial plexus predilection. Neuromuscular Disorders, 2000, 10, 515-517. | 0.3 | 30 |
| 113 | Novel frameshift and splice site mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with hereditary sensory neuropathy type IV. Neuromuscular Disorders, 2006, 16, 19-25. | 0.3 | 30 |
| 114 | Mutant HSPB8 causes protein aggregates and a reduced mitochondrial membrane potential in dermal fibroblasts from distal hereditary motor neuropathy patients. Neuromuscular Disorders, 2012, 22, 699-711. | 0.3 | 30 |
| 115 | Animal models and therapeutic prospects for Charcotâ€“Marieâ€“Tooth disease. Annals of Neurology, 2013, 74, 391-396. | 2.8 | 30 |
| 116 | Loss of Neurological Disease HSAN-I-Associated Gene SPTLC2 Impairs CD8+ T Cell Responses to Infection by Inhibiting T Cell Metabolic Fitness. Immunity, 2019, 50, 1218-1231.e5. | 6.6 | 30 |
| 117 | Molecular genetic analysis of the 17p11.2 region in patients with hereditary neuropathy with liability to pressure palsies (HNPP). Human Genetics, 1996, 97, 26-34. | 1.8 | 29 |
| 118 | Genotypeâ€“phenotype analysis in patients with giant axonal neuropathy (GAN). Neuromuscular Disorders, 2007, 17, 624-630. | 0.3 | 29 |
| 119 | Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach. Neurogenetics, 2015, 16, 33-42. | 0.7 | 29 |
| 120 | Mutant HSPB1 causes loss of translational repression by binding to PCBP1, an RNA binding protein with a possible role in neurodegenerative disease. Acta Neuropathologica Communications, 2017, 5, 5. | 2.4 | 29 |
| 121 | Sensory-Neuropathy-Causing Mutations in ATL3 Cause Aberrant ER Membrane Tethering. Cell Reports, 2018, 23, 2026-2038. | 2.9 | 29 |
| 122 | Human Rab7 mutation mimics features of Charcotâ€“Marieâ€“Tooth neuropathy type 2B in Drosophila. Neurobiology of Disease, 2014, 65, 211-219. | 2.1 | 28 |
| 123 | Microglial derived extracellular vesicles activate autophagy and mediate multiâ€target signaling to maintain cellular homeostasis. Journal of Extracellular Vesicles, 2020, 10, e12022. | 5.5 | 28 |
| 124 | A novel 3â€²-splice site mutation in peripheral myelin protein 22 causing hereditary neuropathy with liability to pressure palsies. Neuromuscular Disorders, 2001, 11, 400-403. | 0.3 | 27 |
| 125 | Autosomal dominant axonal Charcot-Marie-Tooth disease type 2 (CMT2G) maps to chromosome 12q12-q13.3. Journal of Medical Genetics, 2004, 41, 193-197. | 1.5 | 27 |
| 126 | Induced pluripotent stem cell-derived motor neurons of CMT type 2 patients reveal progressive mitochondrial dysfunction. Brain, 2021, 144, 2471-2485. | 3.7 | 27 |

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|---|---|---|---|
| 127 | Charcot-Marie-Tooth disease: an intermediate form. Neuromuscular Disorders, 1998, 8, 392-393. | 0.3 | 26 |
| 128 | Hereditary Neuralgic Amyotrophy (HNA) is genetically heterogeneous. Journal of Neurology, 2001, 248, 861-865. | 1.8 | 26 |
| 129 | A systematic comparison of all mutations in hereditary sensory neuropathy type I (HSAN I) reveals that the G387A mutation is not disease associated. Neurogenetics, 2009, 10, 135-143. | 0.7 | 26 |
| 130 | Reduced penetrance in hereditary motor neuropathy caused by TRPV4 Arg269Cys mutation. Journal of Neurology, 2011, 258, 1413-1421. | 1.8 | 24 |
| 131 | Exclusion of serine palmitoyltransferase long chain base subunit 2 (SPTLC2) as a common cause for hereditary sensory neuropathy. Neuromuscular Disorders, 2002, 12, 656-658. | 0.3 | 23 |
| 132 | Humoral immunodeficiency in congenital insensitivity to pain with anhidrosis. Neurogenetics, 2009, 10, 161-165. | 0.7 | 23 |
| 133 | Defects in Axonal Transport in Inherited Neuropathies. Journal of Neuromuscular Diseases, 2019, 6, 401-419. | 1.1 | 23 |
| 134 | PMP22 Thr118Met is not a clinically relevant CMT1 marker. Journal of Neurology, 2000, 247, 696-700. | 1.8 | 22 |
| 135 | Unraveling the Genetics of Distal Hereditary Motor Neuronopathies. NeuroMolecular Medicine, 0, 8, 131-146. | 1.8 | 21 |
| 136 | A de novo duplication in 17p11.2 and a novel mutation in the Po gene in two DÃ©jÃ©rineâ€”Sottas syndrome patients. , 1996, 8, 304-310. | | 20 |
| 137 | Genetics of motor neuron disease. Current Neurology and Neuroscience Reports, 2006, 6, 423-431. | 2.0 | 20 |
| 138 | Fine mapping of the neurally expressed gene SOX14 to human 3q23, relative to three congenital diseases. Human Genetics, 2000, 106, 432-439. | 1.8 | 19 |
| 139 | Genetic pain loss disorders. Nature Reviews Disease Primers, 2022, 8, . | 18.1 | 18 |
| 140 | Absence of mutations in peripheral myelin protein-22, myelin protein zero, and connexin 32 in autosomal recessive Dejerine-Sottas syndrome. Neuroscience Letters, 1998, 240, 1-4. | 1.0 | 17 |
| 141 | A Novel Type of Hereditary Motor and Sensory Neuropathy Characterized by a Mild Phenotype. Archives of Neurology, 1999, 56, 1283. | 4.9 | 16 |
| 142 | PFN2 and GAMT as common molecular determinants of axonal Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 870-878. | 0.9 | 16 |
| 143 | A Sequence-Ready BAC/PAC Contig and Partial Transcript Map of Approximately 1.5 Mb in Human Chromosome 17q25 Comprising Multiple Disease Genes. Genomics, 1999, 62, 242-250. | 1.3 | 15 |
| 144 | L239F founder mutation in GDAP1 is associated with a mild Charcotâ€“Marieâ€“Tooth type 4C4 (CMT4C4) phenotype. Neurogenetics, 2010, 11, 357-366. | 0.7 | 15 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Report of a novel ATP7A mutation causing distal motor neuropathy. Neuromuscular Disorders, 2019, 29, 776-785. | 0.3 | 15 |
| 146 | A weakened interface in the P182L variant of HSP27 associated with severe Charcotâ€Marieâ€Tooth neuropathy causes aberrant binding to interacting proteins. EMBO Journal, 2021, 40, e103811. | 3.5 | 14 |
| 147 | HSPB1 Facilitates the Formation of Non-Centrosomal Microtubules. PLoS ONE, 2013, 8, e66541. | 1.1 | 14 |
| 148 | Mutation analysis of 4 candidate genes for hereditary neuralgic amyotrophy (HNA). Human Genetics, 2001, 108, 390-393. | 1.8 | 13 |
| 149 | Charcotâ€“Marieâ€“Tooth disease type 2G redefined by a novel mutation in LRSAM1. Annals of Neurology, 2016, 80, 823-833. | 2.8 | 13 |
| 150 | NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. Brain, 2022, 145, 3999-4015. | 3.7 | 12 |
| 151 | Of giant axons and curly hair. Nature Genetics, 2000, 26, 254-255. | 9.4 | 11 |
| 152 | A Clone Contig of 12q24.3 Encompassing the Distal Hereditary Motor Neuropathy Type II Gene. Genomics, 2000, 65, 34-43. | 1.3 | 11 |
| 153 | Hereditary neuralgic amyotrophy. Neurogenetics, 2001, 3, 115-118. | 0.7 | 11 |
| 154 | Peripheral neuropathy and 46XY gonadal dysgenesis: A heterogeneous entity. Neuromuscular Disorders, 2009, 19, 172-175. | 0.3 | 11 |
| 155 | Drosophila as a platform to predict the pathogenicity of novel aminoacyl-tRNA synthetase mutations in CMT. Amino Acids, 2012, 42, 1661-1668. | 1.2 | 11 |
| 156 | Oligodendroglia-derived extracellular vesicles activate autophagy via LC3B/BAG3 to protect against oxidative stress with an enhanced effect for HSPB8 enriched vesicles. Cell Communication and Signaling, 2022, 20, 58. | 2.7 | 10 |
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