# Hanns Lochmller

## List of Publications by Citations



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| 456 papers | 18,911 citations | 73 h-index | 111 g-index |
| 485 ext. papers | 22,324 ext. citations | 5.9 avg, IF | 6.17 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 456 | The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, **2017**, 45, D865-D876 | 20.1 | 507 |
| 455 | Phenotypic spectrum associated with mutations of the mitochondrial polymerase gamma gene. *Brain*, **2006**, 129, 1674-84 | 11.2 | 363 |
| 454 | The TREAT-NMD DMD Global Database: analysis of more than 7,000 Duchenne muscular dystrophy mutations. *Human Mutation*, **2015**, 36, 395-402 | 4.7 | 338 |
| 453 | Mutations in dynamin 2 cause dominant centronuclear myopathy. *Nature Genetics*, **2005**, 37, 1207-9 | 36.3 | 335 |
| 452 | Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic Acids Research*, **2019**, 47, D1018-D1027 | 20.1 | 333 |
| 451 | The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. *Brain*, **2007**, 130, 2037-44 | 11.2 | 247 |
| 450 | A mutation in the dimerization domain of filamin c causes a novel type of autosomal dominant myofibrillar myopathy. *American Journal of Human Genetics*, **2005**, 77, 297-304 | 11 | 227 |
| 449 | Mildly affected patients with spinal muscular atrophy are partially protected by an increased SMN2 copy number. *Human Genetics*, **2006**, 119, 422-8 | 6.3 | 222 |
| 448 | Mutation in TACO1, encoding a translational activator of COX I, results in cytochrome c oxidase deficiency and late-onset Leigh syndrome. *Nature Genetics*, **2009**, 41, 833-7 | 36.3 | 218 |
| 447 | Dok-7 mutations underlie a neuromuscular junction synaptopathy. *Science*, **2006**, 313, 1975-8 | 33.3 | 217 |
| 446 | SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. *Nature Genetics*, **2009**, 41, 654-6 | 36.3 | 209 |
| 445 | Prevalence, incidence and carrier frequency of 5q-linked spinal muscular atrophy - a literature review. *Orphanet Journal of Rare Diseases*, **2017**, 12, 124 | 4.2 | 201 |
| 444 | International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705 | 11 | 200 |
| 443 | Mutations in SIL1 cause Marinesco-Sjögren syndrome, a cerebellar ataxia with cataract and myopathy. *Nature Genetics*, **2005**, 37, 1312-4 | 36.3 | 198 |
| 442 | Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. *Nature Genetics*, **2010**, 42, 160-4 | 36.3 | 191 |
| 441 | An agrin minigene rescues dystrophic symptoms in a mouse model for congenital muscular dystrophy. *Nature*, **2001**, 413, 302-7 | 50.4 | 188 |
| 440 | Late onset Pompe disease: clinical and neurophysiological spectrum of 38 patients including long-term follow-up in 18 patients. *Neuromuscular Disorders*, **2007**, 17, 698-706 | 2.9 | 175 |

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| 439 | Risk of developing a mitochondrial DNA deletion disorder. *Lancet, The*, **2004**, 364, 592-6 | 40 | 161 |
| 438 | A founder mutation in Anoctamin 5 is a major cause of limb-girdle muscular dystrophy. *Brain*, **2011**, 134, 171-182 | 11.2 | 157 |
| 437 | Mutations and polymorphisms of the skeletal muscle alpha-actin gene (ACTA1). *Human Mutation*, **2009**, 30, 1267-77 | 4.7 | 150 |
| 436 | Dystrophin expression in muscles of mdx mice after adenovirus-mediated in vivo gene transfer. *Human Gene Therapy*, **1996**, 7, 129-40 | 4.8 | 148 |
| 435 | Autosomal-dominant distal myopathy associated with a recurrent missense mutation in the gene encoding the nuclear matrix protein, matrin 3. *American Journal of Human Genetics*, **2009**, 84, 511-8 | 11 | 136 |
| 434 | Phenotypical spectrum of DOK7 mutations in congenital myasthenic syndromes. *Brain*, **2007**, 130, 1497-506 | 11.2 | 129 |
| 433 | Pathological consequences of VCP mutations on human striated muscle. *Brain*, **2007**, 130, 381-93 | 11.2 | 128 |
| 432 | An X-linked myopathy with postural muscle atrophy and generalized hypertrophy, termed XMPMA, is caused by mutations in FHL1. *American Journal of Human Genetics*, **2008**, 82, 88-99 | 11 | 127 |
| 431 | RD-Connect: an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. *Journal of General Internal Medicine*, **2014**, 29 Suppl 3, S780-7 | 4 | 124 |
| 430 | Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. *American Journal of Human Genetics*, **2011**, 88, 162-72 | 11 | 124 |
| 429 | The ubiquitin-selective chaperone CDC-48/p97 links myosin assembly to human myopathy. *Nature Cell Biology*, **2007**, 9, 379-90 | 23.4 | 122 |
| 428 | Escobar syndrome is a prenatal myasthenia caused by disruption of the acetylcholine receptor fetal gamma subunit. *American Journal of Human Genetics*, **2006**, 79, 303-12 | 11 | 121 |
| 427 | Clinical and morphological phenotype of the filamin myopathy: a study of 31 German patients. *Brain*, **2007**, 130, 3250-64 | 11.2 | 120 |
| 426 | Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. *Human Mutation*, **2012**, 33, 981-8 | 4.7 | 119 |
| 425 | Quantitative muscle MRI as an assessment tool for monitoring disease progression in LGMD2I: a multicentre longitudinal study. *PLoS ONE*, **2013**, 8, e70993 | 3.7 | 116 |
| 424 | Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. *Nature Genetics*, **2003**, 35, 185-9 | 36.3 | 116 |
| 423 | Mitochondrial phosphate-carrier deficiency: a novel disorder of oxidative phosphorylation. *American Journal of Human Genetics*, **2007**, 80, 478-84 | 11 | 113 |
| 422 | Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. *Brain*, **2008**, 131, 747-59 | 11.2 | 112 |

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| 421 | Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). *Annals of Neurology*, **2003**, 54, 719-24 | 9.4 | 112 |
| 420 | Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). *Neuromuscular Disorders*, **2011**, 21, 569-78 | 2.9 | 110 |
| 419 | The burden of Duchenne muscular dystrophy: an international, cross-sectional study. *Neurology*, **2014**, 83, 529-36 | 6.5 | 108 |
| 418 | Diagnostic value of muscle MRI in differentiating LGMD2I from other LGMDs. *Journal of Neurology*, **2005**, 252, 538-47 | 5.5 | 108 |
| 417 | Mutation history of the roma/gypsies. *American Journal of Human Genetics*, **2004**, 75, 596-609 | 11 | 106 |
| 416 | Congenital myasthenic syndromes due to mutations in ALG2 and ALG14. *Brain*, **2013**, 136, 944-56 | 11.2 | 103 |
| 415 | Factors influencing the efficacy, longevity, and safety of electroporation-assisted plasmid-based gene transfer into mouse muscles. *Molecular Therapy*, **2004**, 10, 447-55 | 11.7 | 102 |
| 414 | Muscle fibres and cultured muscle cells express the B7.1/2-related inducible co-stimulatory molecule, ICOSL: implications for the pathogenesis of inflammatory myopathies. *Brain*, **2003**, 126, 1026-35 | 11.2 | 101 |
| 413 | International Charter of principles for sharing bio-specimens and data. *European Journal of Human Genetics*, **2015**, 23, 721-8 | 5.3 | 98 |
| 412 | Targeted next-generation sequencing of a 12.5 Mb homozygous region reveals ANO10 mutations in patients with autosomal-recessive cerebellar ataxia. *American Journal of Human Genetics*, **2010**, 87, 813-9 | 11 | 98 |
| 411 | Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. *Brain*, **2009**, 132, 3165-74 | 11.2 | 96 |
| 410 | Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. *Human Molecular Genetics*, **2013**, 22, 4368-82 | 5.6 | 94 |
| 409 | Human muscle cells express a B7-related molecule, B7-H1, with strong negative immune regulatory potential: a novel mechanism of counterbalancing the immune attack in idiopathic inflammatory myopathies. *FASEB Journal*, **2003**, 17, 1892-4 | 0.9 | 90 |
| 408 | Mutations in GMPPB cause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. *Brain*, **2015**, 138, 2493-504 | 11.2 | 89 |
| 407 | Missense mutations of ACTA1 cause dominant congenital myopathy with cores. *Journal of Medical Genetics*, **2004**, 41, 842-8 | 5.8 | 88 |
| 406 | Future of Rare Diseases Research 2017-2027: An IRDiRC Perspective. *Clinical and Translational Science*, **2018**, 11, 21-27 | 4.9 | 86 |
| 405 | Expression of the E6 and E7 genes of human papillomavirus (HPV16) extends the life span of human myoblasts. *Experimental Cell Research*, **1999**, 248, 186-93 | 4.2 | 85 |
| 404 | Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. *Journal of Neuromuscular Diseases*, **2017**, 4, 293-306 | 5 | 84 |

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| 403 | Sympathetic innervation controls homeostasis of neuromuscular junctions in health and disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 746-50 | 11.5 | 82 |
| 402 | Adult-onset cerebellar ataxia due to mutations in CABC1/ADCK3. *Journal of Neurology, Neurosurgery and Psychiatry*, **2012**, 83, 174-8 | 5.5 | 82 |
| 401 | Limb-girdle muscular dystrophies. *Current Opinion in Neurology*, **2008**, 21, 576-84 | 7.1 | 82 |
| 400 | Phenotypes of the N88S Berardinelli-Seip congenital lipodystrophy 2 mutation. *Annals of Neurology* , **2005**, 57, 415-24 | 9.4 | 82 |
| 399 | Affinity proteomics within rare diseases: a BIO-NMD study for blood biomarkers of muscular dystrophies. *EMBO Molecular Medicine*, **2014**, 6, 918-36 | 12 | 81 |
| 398 | Attenuated muscle regeneration is a key factor in dysferlin-deficient muscular dystrophy. *Human Molecular Genetics*, **2009**, 18, 1976-89 | 5.6 | 81 |
| 397 | EFNS guideline on diagnosis and management of limb girdle muscular dystrophies. *European Journal of Neurology*, **2007**, 14, 1305-12 | 6 | 81 |
| 396 | EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. *Nature Communications*, **2014**, 5, 4287 | 17.4 | 80 |
| 395 | Congenital myasthenic syndromes: achievements and limitations of phenotype-guided gene-after-gene sequencing in diagnostic practice: a study of 680 patients. *Human Mutation*, **2012**, 33, 1474-84 | 4.7 | 80 |
| 394 | The TREAT-NMD Duchenne muscular dystrophy registries: conception, design, and utilization by industry and academia. *Human Mutation*, **2013**, 34, 1449-57 | 4.7 | 78 |
| 393 | Commonality of TRIM32 mutation in causing sarcotubular myopathy and LGMD2H. *Annals of Neurology*, **2005**, 57, 591-5 | 9.4 | 78 |
| 392 | Synaptotagmin 2 mutations cause an autosomal-dominant form of lambert-eaton myasthenic syndrome and nonprogressive motor neuropathy. *American Journal of Human Genetics*, **2014**, 95, 332-9 | 11 | 77 |
| 391 | The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. *Journal of Neurology*, **2011**, 258, 1987-97 | 5.5 | 77 |
| 390 | Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. *Neuromuscular Disorders*, **2010**, 20, 438-42 | 2.9 | 77 |
| 389 | No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. *Biochemical and Biophysical Research Communications*, **2005**, 328, 221-6 | 3.4 | 76 |
| 388 | The non-classical MHC molecule HLA-G protects human muscle cells from immune-mediated lysis: implications for myoblast transplantation and gene therapy. *Brain*, **2003**, 126, 176-85 | 11.2 | 76 |
| 387 | Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 - A Prospective Observational Study. *Journal of Neuromuscular Diseases*, **2019**, 6, 453-465 | 5 | 75 |
| 386 | Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. *Human Molecular Genetics*, **2011**, 20, 4334-44 | 5.6 | 75 |

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| 385 | Dysferlin associates with the developing T-tubule system in rodent and human skeletal muscle. *Muscle and Nerve*, **2010**, 41, 166-73 | 3.4 | 75 |
| 384 | Expression of toll-like receptors by human muscle cells in vitro and in vivo: TLR3 is highly expressed in inflammatory and HIV myopathies, mediates IL-8 release and up-regulation of NKG2D-ligands. *FASEB Journal*, **2006**, 20, 118-20 | 0.9 | 73 |
| 383 | The principles of gene therapy for the nervous system. *Trends in Neurosciences*, **1996**, 19, 49-54 | 13.3 | 72 |
| 382 | Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. *Brain*, **2014**, 137, 2429-43 | 11.2 | 69 |
| 381 | Scapuloperoneal syndrome type Kaeser and a wide phenotypic spectrum of adult-onset, dominant myopathies are associated with the desmin mutation R350P. *Brain*, **2007**, 130, 1485-96 | 11.2 | 68 |
| 380 | Muscle pathology in 57 patients with myotonic dystrophy type 2. *Muscle and Nerve*, **2004**, 29, 275-81 | 3.4 | 68 |
| 379 | High-level dystrophin expression after adenovirus-mediated dystrophin minigene transfer to skeletal muscle of dystrophic dogs: prolongation of expression with immunosuppression. *Human Gene Therapy*, **1998**, 9, 629-34 | 4.8 | 68 |
| 378 | The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy. *Neuromuscular Disorders*, **2010**, 20, 255-9 | 2.9 | 67 |
| 377 | Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. *Brain*, **2011**, 134, 183-95 | 11.2 | 64 |
| 376 | Differential short-term transduction efficiency of adult versus newborn mouse tissues by adenoviral recombinants. *Experimental and Molecular Pathology*, **1995**, 62, 131-43 | 4.4 | 64 |
| 375 | New aspects on patients affected by dysferlin deficient muscular dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2010**, 81, 946-53 | 5.5 | 63 |
| 374 | Antibody-mediated targeting of an adenovirus vector modified to contain a synthetic immunoglobulin g-binding domain in the capsid. *Journal of Virology*, **2003**, 77, 2093-104 | 6.6 | 63 |
| 373 | Progress in Rare Diseases Research 2010-2016: An IRDiRC Perspective. *Clinical and Translational Science*, **2018**, 11, 11-20 | 4.9 | 63 |
| 372 | Cognitive behavioural therapy with optional graded exercise therapy in patients with severe fatigue with myotonic dystrophy type 1: a multicentre, single-blind, randomised trial. *Lancet Neurology, The*, **2018**, 17, 671-680 | 24.1 | 62 |
| 371 | Parkinson syndrome, neuropathy, and myopathy caused by the mutation A8344G (MERRF) in tRNALys. *Neurology*, **2007**, 68, 56-8 | 6.5 | 62 |
| 370 | Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. *Experimental Cell Research*, **2005**, 304, 365-79 | 4.2 | 62 |
| 369 | Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study. *PLoS ONE*, **2014**, 9, e90377 | 3.7 | 62 |
| 368 | Validation of genetic modifiers for Duchenne muscular dystrophy: a multicentre study assessing SPP1 and LTBP4 variants. *Journal of Neurology, Neurosurgery and Psychiatry*, **2015**, 86, 1060-5 | 5.5 | 61 |

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| 367 | Recessive mutations in the  (VI) collagen gene COL6A3 cause early-onset isolated dystonia. *American Journal of Human Genetics*, **2015**, 96, 883-93 | 11 | 61 |
| 366 | Mapping the differences in care for 5,000 spinal muscular atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. *Journal of Neurology*, **2014**, 261, 152-63 | 5.5 | 60 |
| 365 | Muscle MRI findings in limb girdle muscular dystrophy type 2L. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S122-9 | 2.9 | 60 |
| 364 | Developmental defects in a zebrafish model for muscular dystrophies associated with the loss of fukutin-related protein (FKRP). *Brain*, **2008**, 131, 1551-61 | 11.2 | 60 |
| 363 | Linker molecules between laminins and dystroglycan ameliorate laminin-alpha2-deficient muscular dystrophy at all disease stages. *Journal of Cell Biology*, **2007**, 176, 979-93 | 7.3 | 60 |
| 362 | Homozygous mutations in caveolin-3 cause a severe form of rippling muscle disease. *Annals of Neurology*, **2003**, 53, 512-20 | 9.4 | 60 |
| 361 | Mutations in the collagen XII gene define a new form of extracellular matrix-related myopathy. *Human Molecular Genetics*, **2014**, 23, 2353-63 | 5.6 | 59 |
| 360 | Treatment of dysferlinopathy with deflazacort: a double-blind, placebo-controlled clinical trial. *Orphanet Journal of Rare Diseases*, **2013**, 8, 26 | 4.2 | 58 |
| 359 | Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 331-8 | 5.5 | 58 |
| 358 | Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *Journal of Neurology*, **2012**, 259, 838-50 | 5.5 | 58 |
| 357 | Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. *Journal of Inherited Metabolic Disease*, **2011**, 34, 197-201 | 5.4 | 58 |
| 356 | ANO10 mutations cause ataxia and coenzyme Q deficiency. *Journal of Neurology*, **2014**, 261, 2192-8 | 5.5 | 57 |
| 355 | Human skeletal muscle-derived CD133(+) cells form functional satellite cells after intramuscular transplantation in immunodeficient host mice. *Molecular Therapy*, **2014**, 22, 1008-17 | 11.7 | 57 |
| 354 | Gentamicin fails to increase dystrophin expression in dystrophin-deficient muscle. *Muscle and Nerve* , **2003**, 27, 624-7 | 3.4 | 57 |
| 353 | Quantifying the burden of caregiving in Duchenne muscular dystrophy. *Journal of Neurology*, **2016**, 263, 906-915 | 5.5 | 56 |
| 352 | A heterozygous 21-bp deletion in CAPN3 causes dominantly inherited limb girdle muscular dystrophy. *Brain*, **2016**, 139, 2154-63 | 11.2 | 56 |
| 351 | Recommendations for Improving the Quality of Rare Disease Registries. *International Journal of Environmental Research and Public Health*, **2018**, 15, | 4.6 | 56 |
| 350 | A multi-source approach to determine SMA incidence and research ready population. *Journal of Neurology*, **2017**, 264, 1465-1473 | 5.5 | 56 |

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| 349 | Fibronectin is a serum biomarker for Duchenne muscular dystrophy. *Proteomics - Clinical Applications*, **2014**, 8, 269-78 | 3.1 | 55 |
| 348 | 5Ntrans-splicing repair of the PLEC1 gene. *Journal of Investigative Dermatology*, **2008**, 128, 568-74 | 4.3 | 55 |
| 347 | A novel homozygous missense mutation in the GNE gene of a patient with quadriceps-sparing hereditary inclusion body myopathy associated with muscle inflammation. *Neuromuscular Disorders*, **2003**, 13, 830-4 | 2.9 | 55 |
| 346 | Genetic heterogeneity of motor neuropathies. *Neurology*, **2017**, 88, 1226-1234 | 6.5 | 54 |
| 345 | Further evidence for genetic heterogeneity of distal HMN type V, CMT2 with predominant hand involvement and Silver syndrome. *Journal of the Neurological Sciences*, **2007**, 263, 100-6 | 3.2 | 54 |
| 344 | Health-related quality of life in patients with Duchenne muscular dystrophy: a multinational, cross-sectional study. *Developmental Medicine and Child Neurology*, **2016**, 58, 508-15 | 3.3 | 54 |
| 343 | Novel POMGnT1 mutations define broader phenotypic spectrum of muscle-eye-brain disease. *Neurogenetics*, **2007**, 8, 279-88 | 3 | 53 |
| 342 | A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome. *Journal of Neurology*, **2012**, 259, 474-81 | 5.5 | 52 |
| 341 | ANO5 gene analysis in a large cohort of patients with anoctaminopathy: confirmation of male prevalence and high occurrence of the common exon 5 gene mutation. *Human Mutation*, **2013**, 34, 1111-8 | 4.7 | 51 |
| 340 | Mutation in dystrophin-encoding gene affects energy metabolism in mouse myoblasts. *Biochemical and Biophysical Research Communications*, **2009**, 386, 463-6 | 3.4 | 51 |
| 339 | Muscle-Derived Proteins as Serum Biomarkers for Monitoring Disease Progression in Three Forms of Muscular Dystrophy. *Journal of Neuromuscular Diseases*, **2015**, 2, 241-255 | 5 | 50 |
| 338 | NF-kappaB-dependent expression of the antiapoptotic factor c-FLIP is regulated by calpain 3, the protein involved in limb-girdle muscular dystrophy type 2A. *FASEB Journal*, **2008**, 22, 1521-9 | 0.9 | 50 |
| 337 | Homozygosity for CCTG mutation in myotonic dystrophy type 2. *Brain*, **2004**, 127, 1868-77 | 11.2 | 50 |
| 336 | Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. *American Journal of Human Genetics*, **2016**, 99, 753-761 | 11 | 50 |
| 335 | The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. *European Journal of Human Genetics*, **2015**, 23, 1116-23 | 5.3 | 49 |
| 334 | Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited. *Neuropathology and Applied Neurobiology*, **2007**, 33, 544-59 | 5.2 | 48 |
| 333 | Creatine monohydrate in myotonic dystrophy: a double-blind, placebo-controlled clinical study. *Journal of Neurology*, **2002**, 249, 1717-22 | 5.5 | 48 |
| 332 | Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. *Journal of Neuromuscular Diseases*, **2014**, 1, 75-90 | 5 | 47 |

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| 331 | Increased susceptibility to ATP via alteration of P2X receptor function in dystrophic mdx mouse muscle cells. *FASEB Journal*, **2006**, 20, 610-20 | 0.9 | 47 |
| 330 | Congenital myasthenic syndromes: spotlight on genetic defects of neuromuscular transmission. *Expert Reviews in Molecular Medicine*, **2007**, 9, 1-20 | 6.7 | 47 |
| 329 | Screening for carnitine palmitoyltransferase II deficiency by tandem mass spectrometry. *Journal of Inherited Metabolic Disease*, **2002**, 25, 17-27 | 5.4 | 47 |
| 328 | Characterization of human muscle type cofilin (CFL2) in normal and regenerating muscle. *FEBS Journal*, **2001**, 268, 3473-82 | | 47 |
| 327 | Exon skipping and gene transfer restore dystrophin expression in human induced pluripotent stem cells-cardiomyocytes harboring DMD mutations. *Stem Cells and Development*, **2013**, 22, 2714-24 | 4.4 | 46 |
| 326 | The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. *European Journal of Human Genetics*, **2017**, 25, 1293-1302 | 5.3 | 45 |
| 325 | Ephedrine therapy in eight patients with congenital myasthenic syndrome due to DOK7 mutations. *Neuromuscular Disorders*, **2009**, 19, 828-32 | 2.9 | 45 |
| 324 | Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. *European Journal of Paediatric Neurology*, **2010**, 14, 326-33 | 3.8 | 45 |
| 323 | Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. *European Journal of Epidemiology*, **2020**, 35, 643-653 | 12.1 | 44 |
| 322 | The Clinical Outcome Study for dysferlinopathy: An international multicenter study. *Neurology: Genetics*, **2016**, 2, e89 | 3.8 | 44 |
| 321 | Salbutamol-responsive limb-girdle congenital myasthenic syndrome due to a novel missense mutation and heteroallelic deletion in MUSK. *Neuromuscular Disorders*, **2014**, 24, 31-5 | 2.9 | 44 |
| 320 | Late onset in dysferlinopathy widens the clinical spectrum. *Neuromuscular Disorders*, **2008**, 18, 288-90 | 2.9 | 44 |
| 319 | Therapeutic strategies in congenital myasthenic syndromes. *Neurotherapeutics*, **2008**, 5, 542-7 | 6.4 | 44 |
| 318 | Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1071-1081 | 5.5 | 43 |
| 317 | Clinical features of the myasthenic syndrome arising from mutations in GMPPB. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 802-9 | 5.5 | 43 |
| 316 | Genetic determinants of disease severity in the myotonic dystrophy type 1 OPTIMISTIC cohort. *Neurology*, **2019**, 93, e995-e1009 | 6.5 | 43 |
| 315 | Association Study of Exon Variants in the NF-B and TGF Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. *American Journal of Human Genetics*, **2016**, 99, 1163-1171 | 11 | 42 |
| 314 | P2X7 purinoceptor alterations in dystrophic mdx mouse muscles: relationship to pathology and potential target for treatment. *Journal of Cellular and Molecular Medicine*, **2012**, 16, 1026-37 | 5.6 | 42 |

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| 313 | You should at least ask. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. *European Journal of Human Genetics*, **2016**, 24, 1403-8 | 5.3 | 42 |
| 312 | Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. *Neuromuscular Disorders*, **2011**, 21, 556-62 | 2.9 | 41 |
| 311 | Mutation screening of the N-myc downstream-regulated gene 1 (NDRG1) in patients with Charcot-Marie-Tooth Disease. *Human Mutation*, **2003**, 22, 129-35 | 4.7 | 41 |
| 310 | Mutations in INPP5K, Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. *American Journal of Human Genetics*, **2017**, 100, 523-536 | 11 | 40 |
| 309 | A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. *Human Molecular Genetics*, **2018**, 27, 1186-1195 | 5.6 | 40 |
| 308 | Compliance to Care Guidelines for Duchenne Muscular Dystrophy. *Journal of Neuromuscular Diseases*, **2015**, 2, 63-72 | 5 | 40 |
| 307 | Mutations in GFPT1 that underlie limb-girdle congenital myasthenic syndrome result in reduced cell-surface expression of muscle AChR. *Human Molecular Genetics*, **2013**, 22, 2905-13 | 5.6 | 40 |
| 306 | High frequency of co-segregating CLCN1 mutations among myotonic dystrophy type 2 patients from Finland and Germany. *Journal of Neurology*, **2008**, 255, 1731-6 | 5.5 | 40 |
| 305 | Antigen processing and presentation in human muscle: cathepsin S is critical for MHC class II expression and upregulated in inflammatory myopathies. *Journal of Neuroimmunology*, **2003**, 138, 132-43 | 3.5 | 40 |
| 304 | RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. *European Journal of Human Genetics*, **2018**, 26, 778-785 | 5.3 | 39 |
| 303 | Identification of mutations in the MYO9A gene in patients with congenital myasthenic syndrome. *Brain*, **2016**, 139, 2143-53 | 11.2 | 39 |
| 302 | A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. *Movement Disorders*, **2012**, 27, 789-93 | 7 | 39 |
| 301 | In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. *Human Molecular Genetics*, **2009**, 18, 1590-9 | 5.6 | 39 |
| 300 | What is influencing the phenotype of the common homozygous polymerase-γ mutation p.Ala467Thr?. *Brain*, **2012**, 135, 3614-26 | 11.2 | 39 |
| 299 | Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. *Neurology*, **2015**, 85, 1964-71 | 6.5 | 38 |
| 298 | A third of LGMD2A biopsies have normal calpain 3 proteolytic activity as determined by an in vitro assay. *Neuromuscular Disorders*, **2007**, 17, 148-56 | 2.9 | 38 |
| 297 | Long-term improvement of slow-channel congenital myasthenic syndrome with fluoxetine. *Neuromuscular Disorders*, **2006**, 16, 329-33 | 2.9 | 38 |
| 296 | Comparative proteomic analyses of Duchenne muscular dystrophy and Becker muscular dystrophy muscles: changes contributing to preserve muscle function in Becker muscular dystrophy patients. *Journal of Cachexia, Sarcopenia and Muscle*, **2020**, 11, 547-563 | 10.3 | 37 |

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