Enrico Bertini

List of Publications by Citations

701 papers	33,159 citations	91 h-index	148 g-index
759 ext. papers	38,496 ext. citations	5.3 avg, IF	6.57 L-index

#	Paper	IF	Citations
701	Analysis of glutathione: implication in redox and detoxification. *Clinica Chimica Acta*, **2003**, 333, 19-39	6.2	809
700	Consensus statement for standard of care in spinal muscular atrophy. *Journal of Child Neurology*, **2007**, 22, 1027-49	2.5	595
699	An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. *Nature*, **1989**, 339, 309-11	50.4	580
698	Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. *Nature Genetics*, **2012**, 44, 1243-8	36.3	521
697	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. *Nature Genetics*, **2009**, 41, 829-32	36.3	507
696	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection. *Nature Genetics*, **2006**, 38, 910-6	36.3	505
695	Mutations of SURF-1 in Leigh disease associated with cytochrome c oxidase deficiency. *American Journal of Human Genetics*, **1998**, 63, 1609-21	11	454
694	PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. *Nature Genetics*, **2006**, 38, 752-4	36.3	419
693	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. *Nature Genetics*, **2006**, 38, 623-5	36.3	320
692	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*, **2018**, 28, 103-115	2.9	319
691	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. *Nature Genetics*, **2009**, 41, 1032-6	36.3	310
690	Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25	11	310
689	Empirical Studies in Information Visualization: Seven Scenarios. *IEEE Transactions on Visualization and Computer Graphics*, **2012**, 18, 1520-36	4	305
688	Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle and Nerve*, **2014**, 50, 477-87	3.4	281
687	Mutations in the gene encoding immunoglobulin mu-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. *Nature Genetics*, **2001**, 29, 75-7	36.3	272
686	Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. *Nature Genetics*, **2007**, 39, 366-72	36.3	268
685	COQ2 nephropathy: a newly described inherited mitochondriopathy with primary renal involvement. *Journal of the American Society of Nephrology: JASN*, **2007**, 18, 2773-80	12.7	265

684	Ullrich scleroatonic muscular dystrophy is caused by recessive mutations in collagen type VI. *Proceedings of the National Academy of Sciences of the United States of America*, **2001**, 98, 7516-21	11.5	258
683	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. *Lancet, The*, **2017**, 390, 1489-1498	49	237
682	Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. *Neuromuscular Disorders*, **2018**, 28, 197-207	2.9	236
681	Spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2011**, 6, 71	4.2	223
680	Diagnostic approach to the congenital muscular dystrophies. *Neuromuscular Disorders*, **2014**, 24, 289-311	2.9	217
679	De novo LMNA mutations cause a new form of congenital muscular dystrophy. *Annals of Neurology*, **2008**, 64, 177-86	9.4	213
678	Phenylbutyrate increases SMN expression in vitro: relevance for treatment of spinal muscular atrophy. *European Journal of Human Genetics*, **2004**, 12, 59-65	5.3	213
677	Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. *Neurology*, **2003**, 60, 1961-7	6.5	210
676	Neurodegeneration associated with genetic defects in phospholipase A(2). *Neurology*, **2008**, 71, 1402-9	6.5	202
675	miRNAs as serum biomarkers for Duchenne muscular dystrophy. *EMBO Molecular Medicine*, **2011**, 3, 258-65	12	201
674	Childhood spinal muscular atrophy: controversies and challenges. *Lancet Neurology, The*, **2012**, 11, 443-52	24.1	200
673	The 6-minute walk test and other endpoints in Duchenne muscular dystrophy: longitudinal natural history observations over 48 weeks from a multicenter study. *Muscle and Nerve*, **2013**, 48, 343-56	3.4	199
672	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. *Nature Genetics*, **2013**, 45, 83-7	36.3	192
671	RYR1 mutations are a common cause of congenital myopathies with central nuclei. *Annals of Neurology*, **2010**, 68, 717-26	9.4	192
670	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. *Neuromuscular Disorders*, **2019**, 29, 842-856	2.9	188
669	Infantile-onset ascending hereditary spastic paralysis is associated with mutations in the alsin gene. *American Journal of Human Genetics*, **2002**, 71, 518-27	11	182
668	The 6-minute walk test and other clinical endpoints in duchenne muscular dystrophy: reliability, concurrent validity, and minimal clinically important differences from a multicenter study. *Muscle and Nerve*, **2013**, 48, 357-68	3.4	179
667	Approach to the diagnosis of congenital myopathies. *Neuromuscular Disorders*, **2014**, 24, 97-116	2.9	178

666	Cerebellar ataxia and coenzyme Q10 deficiency. *Neurology*, **2003**, 60, 1206-8	6.5	174
665	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. *FASEB Journal*, **2007**, 21, 1210-26	0.9	168
664	Leukoencephalopathy with thalamus and brainstem involvement and high lactate ¢LTBL¢caused by EARS2 mutations. *Brain*, **2012**, 135, 1387-94	11.2	165
663	Impaired skin fibroblast carnitine uptake in primary systemic carnitine deficiency manifested by childhood carnitine-responsive cardiomyopathy. *Pediatric Research*, **1990**, 28, 247-55	3.2	164
662	Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of □dystroglycan. *American Journal of Human Genetics*, **2013**, 93, 29-41	11	162
661	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. *Brain*, **2007**, 130, 862-74	11.2	161
660	Consensus statement on standard of care for congenital muscular dystrophies. *Journal of Child Neurology*, **2010**, 25, 1559-81	2.5	157
659	The Children© Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND): test development and reliability. *Neuromuscular Disorders*, **2010**, 20, 155-61	2.9	152
658	Quality metrics in high-dimensional data visualization: an overview and systematization. *IEEE Transactions on Visualization and Computer Graphics*, **2011**, 17, 2203-12	4	147
657	Phosphomannose isomerase deficiency: a carbohydrate-deficient glycoprotein syndrome with hepatic-intestinal presentation. *American Journal of Human Genetics*, **1998**, 62, 1535-9	11	145
656	Mutations in KLHL40 are a frequent cause of severe autosomal-recessive nemaline myopathy. *American Journal of Human Genetics*, **2013**, 93, 6-18	11	142
655	Genotype-phenotype correlation of paroxysmal nonkinesigenic dyskinesia. *Neurology*, **2007**, 68, 1782-9	6.5	141
654	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. *Neurology*, **2007**, 68, 51-5	6.5	141
653	Homozygosity mapping of Hallervorden-Spatz syndrome to chromosome 20p12.3-p13. *Nature Genetics*, **1996**, 14, 479-81	36.3	139
652	EPI-743 reverses the progression of the pediatric mitochondrial disease--genetically defined Leigh Syndrome. *Molecular Genetics and Metabolism*, **2012**, 107, 383-8	3.7	138
651	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. *Brain*, **2007**, 130, 2024-36	11.2	138
650	Glutathione in blood of patients with Friedreich© ataxia. *European Journal of Clinical Investigation*, **2001**, 31, 1007-11	4.6	138
649	Phenylbutyrate increases SMN gene expression in spinal muscular atrophy patients. *European Journal of Human Genetics*, **2005**, 13, 256-9	5.3	137

648	Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation. *Neurology*, **2013**, 80, 2049-54	6.5	135
647	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2010**, 20, 712-6	2.9	134
646	Congenital muscular dystrophies with defective glycosylation of dystroglycan: a population study. *Neurology*, **2009**, 72, 1802-9	6.5	134
645	The effect of genotype on the natural history of eIF2B-related leukodystrophies. *Neurology*, **2004**, 62, 1509-17	6.5	126
644	Infantile Alexander disease: spectrum of GFAP mutations and genotype-phenotype correlation. *American Journal of Human Genetics*, **2001**, 69, 1134-40	11	126
643	Hemimegalencephaly and intractable epilepsy: benefits of hemispherectomy. *Epilepsia*, **1989**, 30, 833-43	6.4	126
642	Functional changes in Duchenne muscular dystrophy: a 12-month longitudinal cohort study. *Neurology*, **2011**, 77, 250-6	6.5	125
641	Reliability of the North Star Ambulatory Assessment in a multicentric setting. *Neuromuscular Disorders*, **2009**, 19, 458-61	2.9	125
640	Actin glutathionylation increases in fibroblasts of patients with Friedreich's ataxia: a potential role in the pathogenesis of the disease. *Journal of Biological Chemistry*, **2003**, 278, 42588-95	5.4	125
639	CEP290 mutations are frequently identified in the oculo-renal form of Joubert syndrome-related disorders. *American Journal of Human Genetics*, **2007**, 81, 104-13	11	118
638	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. *Journal of Clinical Investigation*, **2014**, 124, 4693-708	15.9	118
637	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. *Brain*, **2008**, 131, 747-59	11.2	112
636	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). *Annals of Neurology*, **2003**, 54, 719-24	9.4	112
635	Hypomyelinating leukodystrophies: translational research progress and prospects. *Annals of Neurology*, **2014**, 76, 5-19	9.4	111
634	AHI1 gene mutations cause specific forms of Joubert syndrome-related disorders. *Annals of Neurology*, **2006**, 59, 527-34	9.4	111
633	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA: clinical, morphologic, and biochemical studies. *Neurology*, **1991**, 41, 1053-9	6.5	110
632	Spinal muscular atrophy associated with progressive myoclonic epilepsy is caused by mutations in ASAH1. *American Journal of Human Genetics*, **2012**, 91, 5-14	11	109
631	Pilot trial of phenylbutyrate in spinal muscular atrophy. *Neuromuscular Disorders*, **2004**, 14, 130-5	2.9	109

630	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. *Brain*, **2013**, 136, 872-81	11.2	108
629	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. *Brain*, **2010**, 133, 3194-209	11.2	108
628	Fatal infantile liver failure associated with mitochondrial DNA depletion. *Journal of Pediatrics*, **1992**, 121, 896-901	3.6	108
627	Iron-related MRI images in patients with pantothenate kinase-associated neurodegeneration (PKAN) treated with deferiprone: results of a phase II pilot trial. *Movement Disorders*, **2011**, 26, 1756-9	7	106
626	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. *Neuromuscular Disorders*, **2016**, 26, 126-31	2.9	105
625	Consensus statement on standard of care for congenital myopathies. *Journal of Child Neurology*, **2012**, 27, 363-82	2.5	104
624	CloudLines: compact display of event episodes in multiple time-series. *IEEE Transactions on Visualization and Computer Graphics*, **2011**, 17, 2432-9	4	104
623	DPM2-CDG: a muscular dystrophy-dystroglycanopathy syndrome with severe epilepsy. *Annals of Neurology*, **2012**, 72, 550-8	9.4	103
622	Collagen VI deficiency affects the organization of fibronectin in the extracellular matrix of cultured fibroblasts. *Matrix Biology*, **2001**, 20, 475-86	11.4	101
621	Mechanisms inducing low bone density in Duchenne muscular dystrophy in mice and humans. *Journal of Bone and Mineral Research*, **2011**, 26, 1891-903	6.3	99
620	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. *Neurology*, **2007**, 69, 1285-92	6.5	99
619	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. *Neurology*, **2009**, 72, 784-92	6.5	98
618	Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. *Molecular Genetics and Metabolism*, **2012**, 105, 463-71	3.7	97
617	Histological effects of givinostat in boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2016**, 26, 643-649	2.9	96
616	Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy. *American Journal of Human Genetics*, **2014**, 95, 708-20	11	95
615	GM1 gangliosidosis and Morquio B disease: an update on genetic alterations and clinical findings. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2011**, 1812, 782-90	6.9	95
614	Mutation of plasma membrane Ca2+ ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca2+ homeostasis. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 14514-9	11.5	93
613	Somatic and germline mosaicisms in severe myoclonic epilepsy of infancy. *Biochemical and Biophysical Research Communications*, **2006**, 341, 489-93	3.4	93

612	Multi-minicore disease: searching for boundaries: Phenotype analysis of 38 cases. *Annals of Neurology*, **2000**, 48, 745-757	9.4	92
611	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. *Journal of Neurology*, **2014**, 261, 504-10	5.5	91
610	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. *Human Mutation*, **2012**, 33, 949-59	4.7	91
609	The inv dup(15) syndrome: a clinically recognizable syndrome with altered behavior, mental retardation, and epilepsy. *Neurology*, **1997**, 48, 1081-6	6.5	91
608	The expanding phenotype of POMT1 mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly, and mental retardation. *Human Mutation*, **2006**, 27, 453-9	4.7	91
607	Mutations in the MTM1 gene implicated in X-linked myotubular myopathy. ENMC International Consortium on Myotubular Myopathy. European Neuro-Muscular Center. *Human Molecular Genetics* , **1997**, 6, 1505-11	5.6	89
606	Distinguishing the four genetic causes of Jouberts syndrome-related disorders. *Annals of Neurology* , **2005**, 57, 513-9	9.4	89
605	Renal involvement in mitochondrial cytopathies. *Pediatric Nephrology*, **2012**, 27, 539-50	3.2	87
604	INFUSE: Interactive Feature Selection for Predictive Modeling of High Dimensional Data. *IEEE Transactions on Visualization and Computer Graphics*, **2014**, 20, 1614-23	4	86
603	Characterization of recessive RYR1 mutations in core myopathies. *Human Molecular Genetics*, **2006**, 15, 2791-803	5.6	86
602	Clinical features, risk factors, and prognosis in transient global amnesia: a follow-up study. *European Journal of Neurology*, **2005**, 12, 350-6	6	85
601	Daily salbutamol in young patients with SMA type II. *Neuromuscular Disorders*, **2008**, 18, 536-40	2.9	83
600	Long term natural history data in ambulant boys with Duchenne muscular dystrophy: 36-month changes. *PLoS ONE*, **2014**, 9, e108205	3.7	82
599	Attention deficit hyperactivity disorder and cognitive function in Duchenne muscular dystrophy: phenotype-genotype correlation. *Journal of Pediatrics*, **2012**, 161, 705-9.e1	3.6	82
598	Affinity proteomics within rare diseases: a BIO-NMD study for blood biomarkers of muscular dystrophies. *EMBO Molecular Medicine*, **2014**, 6, 918-36	12	81
597	NPHP1 gene deletion is a rare cause of Joubert syndrome related disorders. *Journal of Medical Genetics*, **2005**, 42, e9	5.8	81
596	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. *Neurology*, **2004**, 62, 262-8	6.5	80
595	Oligophrenin 1 mutations frequently cause X-linked mental retardation with cerebellar hypoplasia. *Neurology*, **2005**, 65, 1364-9	6.5	79

594	Role of gabapentin in spinal muscular atrophy: results of a multicenter, randomized Italian study. *Journal of Child Neurology*, **2003**, 18, 537-41	2.5	79
593	Description, nomenclature, and mapping of a novel cerebello-renal syndrome with the molar tooth malformation. *American Journal of Human Genetics*, **2003**, 73, 663-70	11	78
592	Agenesis of the corpus callosum, combined immunodeficiency, bilateral cataract, and hypopigmentation in two brothers. *American Journal of Medical Genetics Part A*, **1988**, 29, 1-8		78
591	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. *Neuromuscular Disorders*, **2010**, 20, 438-42	2.9	77
590	Decreased guanine nucleotide exchange factor activity in eIF2B-mutated patients. *European Journal of Human Genetics*, **2004**, 12, 561-6	5.3	75
589	24 month longitudinal data in ambulant boys with Duchenne muscular dystrophy. *PLoS ONE*, **2013**, 8, e52512	3.7	75
588	Safety and efficacy of olesoxime in patients with type 2 or non-ambulatory type 3 spinal muscular atrophy: a randomised, double-blind, placebo-controlled phase 2 trial. *Lancet Neurology, The*, **2017**, 16, 513-522	24.1	74
587	The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome. *Neurology*, **2001**, 56, 687-90	6.5	74
586	Autophagy regulates satellite cell ability to regenerate normal and dystrophic muscles. *Cell Death and Differentiation*, **2016**, 23, 1839-1849	12.7	72
585	MKS3/TMEM67 mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. *Human Mutation*, **2009**, 30, E432-42	4.7	72
584	Subcomplexes of human ATP synthase mark mitochondrial biosynthesis disorders. *Annals of Neurology*, **2006**, 59, 265-75	9.4	71
583	The genetic basis of undiagnosed muscular dystrophies and myopathies: Results from 504 patients. *Neurology*, **2016**, 87, 71-6	6.5	70
582	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 182-90	2.5	70
581	Natural history of pulmonary function in collagen VI-related myopathies. *Brain*, **2013**, 136, 3625-33	11.2	69
580	Feeding problems and malnutrition in spinal muscular atrophy type II. *Neuromuscular Disorders*, **2008**, 18, 389-93	2.9	69
579	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 201-6	2.9	68
578	Congenital muscular dystrophies: a brief review. *Seminars in Pediatric Neurology*, **2011**, 18, 277-88	2.9	68
577	Friedreich© ataxia: oxidative stress and cytoskeletal abnormalities. *Journal of the Neurological Sciences*, **2009**, 287, 111-8	3.2	68

576	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. *Neuromuscular Disorders*, **2006**, 16, 548-52	2.9	68
575	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2018**, 28, 4-15	2.9	68
574	Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. *Cell Metabolism*, **2016** , 23, 292-302	24.6	67
573	Mutation update and genotype-phenotype correlations of novel and previously described mutations in TPM2 and TPM3 causing congenital myopathies. *Human Mutation*, **2014**, 35, 779-90	4.7	67
572	COQ4 mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency. *American Journal of Human Genetics*, **2015**, 96, 309-17	11	66
571	Mutation analysis in 16 patients with mtDNA depletion. *Human Mutation*, **2003**, 21, 453-4	4.7	66
570	Dominant and recessive COL6A1 mutations in Ullrich scleroatonic muscular dystrophy. *Annals of Neurology*, **2005**, 58, 400-10	9.4	66
569	A new syndrome with ethylmalonic aciduria and normal fatty acid oxidation in fibroblasts. *Journal of Pediatrics*, **1994**, 124, 79-86	3.6	66
568	New clinical phenotype of branched-chain acyl-CoA oxidation defect. *Lancet, The*, **1991**, 338, 1522-3	40	66
567	GJA12 mutations are a rare cause of Pelizaeus-Merzbacher-like disease. *Neurology*, **2008**, 70, 748-54	6.5	65
566	Unstable mutants in the peripheral endosomal membrane component ALS2 cause early-onset motor neuron disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 16041-6	11.5	64
565	Progressive neuropathy and recurrent myoglobinuria in a child with long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency. *Journal of Pediatrics*, **1991**, 118, 744-6	3.6	64
564	Cardiomyopathy may be the only clinical manifestation in female carriers of Duchenne muscular dystrophy. *Neurology*, **1993**, 43, 2342-5	6.5	64
563	Motor function-muscle strength relationship in spinal muscular atrophy. *Muscle and Nerve*, **2004**, 29, 548-52	3.4	63
562	Collagen type VI and related disorders: Bethlem myopathy and Ullrich scleroatonic muscular dystrophy. *European Journal of Paediatric Neurology*, **2002**, 6, 193-8	3.8	63
561	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. *Journal of Inherited Metabolic Disease*, **2013**, 36, 43-53	5.4	62
560	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. *Neurology*, **2012**, 79, 159-62	6.5	62
559	A severe variant of childhood ataxia with central hypomyelination/vanishing white matter leukoencephalopathy related to EIF21B5 mutation. *Neurology*, **2002**, 59, 1966-8	6.5	62

558	Spectrum of pontocerebellar hypoplasia in 13 girls and boys with CASK mutations: confirmation of a recognizable phenotype and first description of a male mosaic patient. *Orphanet Journal of Rare Diseases*, **2012**, 7, 18	4.2	61
557	Glutathione metabolism and antioxidant enzymes in patients affected by nonalcoholic steatohepatitis. *Clinica Chimica Acta*, **2005**, 355, 105-11	6.2	61
556	Joubert syndrome and related disorders. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 113, 1879-88	3	60
555	Evaluation of alternative glyph designs for time series data in a small multiple setting **2013**,		60
554	Clinical and genetic characterization of Chanarin-Dorfman syndrome. *Biochemical and Biophysical Research Communications*, **2008**, 369, 1125-8	3.4	60
553	Novel SACS mutations in autosomal recessive spastic ataxia of Charlevoix-Saguenay type. *Neurology*, **2004**, 62, 103-6	6.5	59
552	Succinate-CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. *Journal of Inherited Metabolic Disease*, **2016**, 39, 243-52	5.4	58
551	Frataxin deficiency leads to reduced expression and impaired translocation of NF-E2-related factor (Nrf2) in cultured motor neurons. *International Journal of Molecular Sciences*, **2013**, 14, 7853-65	6.3	58
550	Assessing upper limb function in nonambulant SMA patients: development of a new module. *Neuromuscular Disorders*, **2011**, 21, 406-12	2.9	58
549	Spectrum of phenotypes in female patients with epilepsy due to protocadherin 19 mutations. *Epilepsia*, **2011**, 52, 1251-7	6.4	58
548	Hypoparathyroidism in mitochondrial trifunctional protein deficiency. *Journal of Pediatrics*, **1996**, 129, 159-62	3.6	58
547	Delineation and diagnostic criteria of Oral-Facial-Digital Syndrome type VI. *Orphanet Journal of Rare Diseases*, **2012**, 7, 4	4.2	57
546	Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies. *Human Mutation*, **2010**, 31, E1319-31	4.7	57
545	6 Minute walk test in Duchenne MD patients with different mutations: 12 month changes. *PLoS ONE*, **2014**, 9, e83400	3.7	56
544	Fatal infantile leukodystrophy: a severe variant of CACH/VWM syndrome, allelic to chromosome 3q27. *Neurology*, **2001**, 57, 265-70	6.5	56
543	A new simple and rapid LC-ESI-MS/MS method for quantification of plasma oxysterols as dimethylaminobutyrate esters. Its successful use for the diagnosis of Niemann-Pick type C disease. *Clinica Chimica Acta*, **2014**, 437, 93-100	6.2	55
542	Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2005**, 76, 1019-21	5.5	55
541	Antioxidant enzymes in blood of patients with Friedreich© ataxia. *Archives of Disease in Childhood*, **2002**, 86, 376-9	2.2	55

540	POPDC1(S201F) causes muscular dystrophy and arrhythmia by affecting protein trafficking. *Journal of Clinical Investigation*, **2016**, 126, 239-53	15.9	55
539	Biallelic Mutations in TBCD, Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 962-973	11	55
538	Salbutamol increases survival motor neuron (SMN) transcript levels in leucocytes of spinal muscular atrophy (SMA) patients: relevance for clinical trial design. *Journal of Medical Genetics*, **2010**, 47, 856-8	5.8	54
537	Clinical and molecular findings in patients with giant axonal neuropathy (GAN). *Neurology*, **2004**, 62, 13-6	6.5	54
536	Redefining phenotypes associated with mitochondrial DNA single deletion. *Journal of Neurology*, **2015**, 262, 1301-9	5.5	53
535	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. *Neuromuscular Disorders*, **2018**, 28, 582-585	2.9	53
534	Infantile ascending hereditary spastic paralysis (IAHSP): clinical features in 11 families. *Neurology*, **2003**, 60, 674-82	6.5	53
533	Schwann cell expression of PLP1 but not DM20 is necessary to prevent neuropathy. *Annals of Neurology*, **2003**, 53, 354-65	9.4	53
532	Genetic heterogeneity of megalencephalic leukoencephalopathy and subcortical cysts. *Neurology*, **2003**, 61, 534-7	6.5	52
531	Two patients with "dropped head syndrome" due to mutations in LMNA or SEPN1 genes. *Neuromuscular Disorders*, **2005**, 15, 521-4	2.9	51
530	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. *Neuromuscular Disorders*, **2006**, 16, 93-8	2.9	51
529	Megalencephalic leukoencephalopathy with subcortical cysts protein 1 functionally cooperates with the TRPV4 cation channel to activate the response of astrocytes to osmotic stress: dysregulation by pathological mutations. *Human Molecular Genetics*, **2012**, 21, 2166-80	5.6	50
528	Expanding the clinical spectrum of POMT1 phenotype. *Neurology*, **2006**, 66, 1564-7; discussion 1461	6.5	50
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