Nicolas Levy

List of Publications by Citations

243 papers	9,656 citations	51 h-index	90 g-index
276 ext. papers	11,261 ext. citations	5.8 avg, IF	5.42 L-index

#	Paper	IF	Citations
243	Lamin a truncation in Hutchinson-Gilford progeria. *Science*, **2003**, 300, 2055	33.3	997
242	Homozygous defects in LMNA, encoding lamin A/C nuclear-envelope proteins, cause autosomal recessive axonal neuropathy in human (Charcot-Marie-Tooth disorder type 2) and mouse. *American Journal of Human Genetics*, **2002**, 70, 726-36	11	449
241	Combined treatment with statins and aminobisphosphonates extends longevity in a mouse model of human premature aging. *Nature Medicine*, **2008**, 14, 767-72	50.5	300
240	Lamin A and ZMPSTE24 (FACE-1) defects cause nuclear disorganization and identify restrictive dermopathy as a lethal neonatal laminopathy. *Human Molecular Genetics*, **2004**, 13, 2493-503	5.6	290
239	Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle and Nerve*, **2014**, 50, 477-87	3.4	281
238	Splicing-directed therapy in a new mouse model of human accelerated aging. *Science Translational Medicine*, **2011**, 3, 106ra107	17.5	240
237	Loss of ZMPSTE24 (FACE-1) causes autosomal recessive restrictive dermopathy and accumulation of Lamin A precursors. *Human Molecular Genetics*, **2005**, 14, 1503-13	5.6	237
236	SRPX2 mutations in disorders of language cortex and cognition. *Human Molecular Genetics*, **2006**, 15, 1195-207	5.6	213
235	Phenotypic study in 40 patients with dysferlin gene mutations: high frequency of atypical phenotypes. *Archives of Neurology*, **2007**, 64, 1176-82		196
234	Genotype-phenotype correlation in Costello syndrome: HRAS mutation analysis in 43 cases. *Journal of Medical Genetics*, **2006**, 43, 401-5	5.8	174
233	Immortalized pathological human myoblasts: towards a universal tool for the study of neuromuscular disorders. *Skeletal Muscle*, **2011**, 1, 34	5.1	160
232	Exome sequencing and functional analysis identifies BANF1 mutation as the cause of a hereditary progeroid syndrome. *American Journal of Human Genetics*, **2011**, 88, 650-6	11	148
231	Molecular bases of progeroid syndromes. *Human Molecular Genetics*, **2006**, 15 Spec No 2, R151-61	5.6	139
230	Constitutive activation of the calcium sensor STIM1 causes tubular-aggregate myopathy. *American Journal of Human Genetics*, **2013**, 92, 271-8	11	133
229	Unique preservation of neural cells in Hutchinson- Gilford progeria syndrome is due to the expression of the neural-specific miR-9 microRNA. *Cell Reports*, **2012**, 2, 1-9	10.6	129
228	Mutations in FGD4 encoding the Rho GDP/GTP exchange factor FRABIN cause autosomal recessive Charcot-Marie-Tooth type 4H. *American Journal of Human Genetics*, **2007**, 81, 1-16	11	127
227	Genes for hereditary sensory and autonomic neuropathies: a genotype-phenotype correlation. *Brain*, **2009**, 132, 2699-711	11.2	118

226	Primary laminopathy fibroblasts display altered genome organization and apoptosis. *Aging Cell*, **2007**, 6, 139-53	9.9	118
225	SKIV2L mutations cause syndromic diarrhea, or trichohepatoenteric syndrome. *American Journal of Human Genetics*, **2012**, 90, 689-92	11	108
224	Serotonin transporter (5-HTT) gene polymorphisms are not associated with susceptibility to mood disorders. *American Journal of Medical Genetics Part A*, **1998**, 81, 1-3		103
223	VMA21 deficiency prevents vacuolar ATPase assembly and causes autophagic vacuolar myopathy. *Acta Neuropathologica*, **2013**, 125, 439-57	14.3	101
222	Sequence family variant loss from the AZFc interval of the human Y chromosome, but not gene copy loss, is strongly associated with male infertility. *Journal of Medical Genetics*, **2004**, 41, 814-25	5.8	101
221	SHAPE AND TEXTURE INDEXES APPLICATION TO CELL NUCLEI CLASSIFICATION. *International Journal of Pattern Recognition and Artificial Intelligence*, **2013**, 27, 1357002	1.1	98
220	CAPN3 mutations in patients with idiopathic eosinophilic myositis. *Annals of Neurology*, **2006**, 59, 905-11	9.4	98
219	UMD-Predictor: A High-Throughput Sequencing Compliant System for Pathogenicity Prediction of any Human cDNA Substitution. *Human Mutation*, **2016**, 37, 439-46	4.7	86
218	Dysferlin mutations in LGMD2B, Miyoshi myopathy, and atypical dysferlinopathies. *Human Mutation* , **2005**, 26, 165	4.7	85
217	Fine mapping of de novo CMT1A and HNPP rearrangements within CMT1A-REPs evidences two distinct sex-dependent mechanisms and candidate sequences involved in recombination. *Human Molecular Genetics*, **1998**, 7, 141-8	5.6	79
216	Random walk with restart on multiplex and heterogeneous biological networks. *Bioinformatics*, **2019**, 35, 497-505	7.2	78
215	Analysis of the DYSF mutational spectrum in a large cohort of patients. *Human Mutation*, **2009**, 30, E345-75	4.7	78
214	Efficient bypass of mutations in dysferlin deficient patient cells by antisense-induced exon skipping. *Human Mutation*, **2010**, 31, 136-42	4.7	75
213	HGPS and related premature aging disorders: from genomic identification to the first therapeutic approaches. *Mechanisms of Ageing and Development*, **2008**, 129, 449-59	5.6	75
212	Relation of body mass index to high on-treatment platelet reactivity and of failed clopidogrel dose adjustment according to platelet reactivity monitoring in patients undergoing percutaneous coronary intervention. *American Journal of Cardiology*, **2009**, 104, 1511-5	3	73
211	Demyelinating X-linked Charcot-Marie-Tooth disease: unusual electrophysiological findings. *Muscle and Nerve*, **1999**, 22, 1442-7	3.4	72
210	Next generation sequencing for molecular diagnosis of neuromuscular diseases. *Acta Neuropathologica*, **2012**, 124, 273-83	14.3	70
209	Development and Validation of a New Risk Prediction Score for Life-Threatening Ventricular Tachyarrhythmias in Laminopathies. *Circulation*, **2019**, 140, 293-302	16.7	63

208	MG132-induced progerin clearance is mediated by autophagy activation and splicing regulation. *EMBO Molecular Medicine*, **2017**, 9, 1294-1313	12	63
207	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. *Nature Genetics*, **2017**, 49, 249-255	36.3	60
206	The phenotypic manifestations of autosomal recessive axonal Charcot-Marie-Tooth due to a mutation in Lamin A/C gene. *Neuromuscular Disorders*, **2003**, 13, 60-7	2.9	60
205	A naturally occurring human minidysferlin protein repairs sarcolemmal lesions in a mouse model of dysferlinopathy. *Science Translational Medicine*, **2010**, 2, 50ra69	17.5	59
204	The human Y chromosome genes BPY2, CDY1 and DAZ are not essential for sustained fertility. *Molecular Human Reproduction*, **2000**, 6, 789-93	4.4	59
203	Differential DNA methylation of the D4Z4 repeat in patients with FSHD and asymptomatic carriers. *Neurology*, **2014**, 83, 733-42	6.5	58
202	Deregulation of the protocadherin gene FAT1 alters muscle shapes: implications for the pathogenesis of facioscapulohumeral dystrophy. *PLoS Genetics*, **2013**, 9, e1003550	6	58
201	VMA21 deficiency causes an autophagic myopathy by compromising V-ATPase activity and lysosomal acidification. *Cell*, **2009**, 137, 235-46	56.2	54
200	A conserved splicing mechanism of the LMNA gene controls premature aging. *Human Molecular Genetics*, **2011**, 20, 4540-55	5.6	54
199	Sex-dependent rearrangements resulting in CMT1A and HNPP. *Nature Genetics*, **1997**, 17, 136-7	36.3	54
198	PMP22 overexpression causes dysmyelination in mice. *Brain*, **2002**, 125, 2213-21	11.2	54
197	Clinicopathologic features of histiocytic lesions following ALL, with a review of the literature. *Pediatric and Developmental Pathology*, **2010**, 13, 225-37	2.2	53
196	Polymorphic Short Tandem Repeats for Diagnosis of the Charcot-Marie-Tooth 1A Duplication. *Clinical Chemistry*, **2001**, 47, 829-837	5.5	53
195	Nuclear matrix, nuclear envelope and premature aging syndromes in a translational research perspective. *Seminars in Cell and Developmental Biology*, **2014**, 29, 125-47	7.5	52
194	Dysferlinopathies. *Neurology India*, **2008**, 56, 289-97	0.7	51
193	Lack of correlation between outcomes of membrane repair assay and correction of dystrophic changes in experimental therapeutic strategy in dysferlinopathy. *PLoS ONE*, **2012**, 7, e38036	3.7	49
192	Loss of Calmodulin- and Radial-Spoke-Associated Complex Protein CFAP251 Leads to Immotile Spermatozoa Lacking Mitochondria and Infertility in Men. *American Journal of Human Genetics*, **2018**, 103, 413-420	11	47
191	Mutations in PHD-like domain of the ATRX gene correlate with severe psychomotor impairment and severe urogenital abnormalities in patients with ATRX syndrome. *Clinical Genetics*, **2006**, 70, 57-62	4	45

190	An overview of treatment strategies for Hutchinson-Gilford Progeria syndrome. *Nucleus*, **2018**, 9, 246-257	3.9	44
189	PMPCA mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. *Brain*, **2015**, 138, 1505-17	11.2	43
188	High prevalence of laminopathies among patients with metabolic syndrome. *Human Molecular Genetics*, **2011**, 20, 3779-86	5.6	43
187	A novel MYH7 mutation links congenital fiber type disproportion and myosin storage myopathy. *Neuromuscular Disorders*, **2011**, 21, 254-62	2.9	42
186	Novel mutations in TTC37 associated with tricho-hepato-enteric syndrome. *Human Mutation*, **2011**, 32, 277-81	4.7	42
185	Molecular dissection of the Schwann cell specific promoter of the PMP22 gene. *Gene*, **2000**, 248, 223-31	3.8	42
184	Mutations in ACTRT1 and its enhancer RNA elements lead to aberrant activation of Hedgehog signaling in inherited and sporadic basal cell carcinomas. *Nature Medicine*, **2017**, 23, 1226-1233	50.5	42
183	Mutations in BCAP31 cause a severe X-linked phenotype with deafness, dystonia, and central hypomyelination and disorganize the Golgi apparatus. *American Journal of Human Genetics*, **2013**, 93, 579-86	11	41
182	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
181	Induced pluripotent stem cells reveal functional differences between drugs currently investigated in patients with hutchinson-gilford progeria syndrome. *Stem Cells Translational Medicine*, **2014**, 3, 510-9	6.9	40
180	Prelamin A accumulation in endothelial cells induces premature senescence and functional impairment. *Atherosclerosis*, **2014**, 237, 45-52	3.1	39
179	Dysregulation of 4q35- and muscle-specific genes in fetuses with a short D4Z4 array linked to facio-scapulo-humeral dystrophy. *Human Molecular Genetics*, **2013**, 22, 4206-14	5.6	38
178	Linkage of X-linked myopathy with excessive autophagy (XMEA) to Xq28. *European Journal of Human Genetics*, **2000**, 8, 125-9	5.3	38
177	AP1S2 is mutated in X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (Pettigrew syndrome). *European Journal of Human Genetics*, **2014**, 22, 363-8	5.3	36
176	New ZMPSTE24 (FACE1) mutations in patients affected with restrictive dermopathy or related progeroid syndromes and mutation update. *European Journal of Human Genetics*, **2014**, 22, 1002-11	5.3	36
175	Novel frameshifting mutations of the ZMPSTE24 gene in two siblings affected with restrictive dermopathy and review of the mutations described in the literature. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 447-52	2.5	35
174	Homozygosity mapping of autosomal recessive demyelinating Charcot-Marie-Tooth neuropathy (CMT4H) to a novel locus on chromosome 12p11.21-q13.11. *Journal of Medical Genetics*, **2005**, 42, 260-5	5.8	35
173	Drug screening on Hutchinson Gilford progeria pluripotent stem cells reveals aminopyrimidines as new modulators of farnesylation. *Cell Death and Disease*, **2016**, 7, e2105	9.8	34

172	Molecular combing reveals allelic combinations in facioscapulohumeral dystrophy. *Annals of Neurology*, **2011**, 70, 627-33	9.4	33
171	An association of Hutchinson-Gilford progeria and malignancy. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1821-6	2.5	33
170	LMNA mutations in atypical Werner's syndrome. *Lancet, The*, **2003**, 362, 1585-6; author reply 1586	40	33
169	Exome sequencing identifies recurrent alterations and the absence of , and mutations in splenic diffuse red pulp small B-cell lymphoma. *Haematologica*, **2017**, 102, 1758-1766	6.6	32
168	An inherited LMNA gene mutation in atypical Progeria syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2881-7	2.5	32
167	Type B mandibuloacral dysplasia with congenital myopathy due to homozygous ZMPSTE24 missense mutation. *European Journal of Human Genetics*, **2011**, 19, 647-54	5.3	32
166	Exome sequencing reveals a de novo POLD1 mutation causing phenotypic variability in mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL). *Metabolism: Clinical and Experimental*, **2017**, 71, 213-225	12.7	31
165	WDR73 Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. *Human Mutation*, **2015**, 36, 1021-8	4.7	31
164	Homozygous deletion of SUN5 in three men with decapitated spermatozoa. *Human Molecular Genetics*, **2017**, 26, 3167-3171	5.6	30
163	Metformin decreases progerin expression and alleviates pathological defects of Hutchinson-Gilford progeria syndrome cells. *Npj Aging and Mechanisms of Disease*, **2016**, 2, 16026	5.5	29
162	Identification of variants in the 4q35 gene FAT1 in patients with a facioscapulohumeral dystrophy-like phenotype. *Human Mutation*, **2015**, 36, 443-53	4.7	29
161	Translational research and therapeutic perspectives in dysferlinopathies. *Molecular Medicine*, **2011**, 17, 875-82	6.2	29
160	Characterization of two mutations in the SPTLC1 subunit of serine palmitoyltransferase associated with hereditary sensory and autonomic neuropathy type I. *Human Mutation*, **2011**, 32, E2211-25	4.7	29
159	Molecular genetics of autosomal-recessive axonal Charcot-Marie-Tooth neuropathies. *NeuroMolecular Medicine*, **2006**, 8, 87-106	4.6	29
158	The clinical variability of the MECP2 duplication syndrome: description of two families with duplications excluding L1CAM and FLNA. *Clinical Genetics*, **2009**, 75, 301-3	4	27
157	NDRG1-linked Charcot-Marie-Tooth disease (CMT4D) with central nervous system involvement. *Neuromuscular Disorders*, **2007**, 17, 163-8	2.9	27
156	Evidence of SARS-CoV-2 re-infection with a different genotype. *Journal of Infection*, **2021**, 82, 84-123	18.9	27
155	Exome sequencing identifies a novel mutation in PIK3R1 as the cause of SHORT syndrome. *BMC Medical Genetics*, **2014**, 15, 51	2.1	26

154	Novel LMNA mutation in atypical Werner syndrome presenting with ischemic disease. *Stroke*, **2009**, 40, e11-4	6.7	26
153	UMD-DYSF, a novel locus specific database for the compilation and interactive analysis of mutations in the dysferlin gene. *Human Mutation*, **2012**, 33, E2317-31	4.7	25
152	HSFY genes and the P4 palindrome in the AZFb interval of the human Y chromosome are not required for spermatocyte maturation. *Human Reproduction*, **2012**, 27, 615-24	5.7	25
151	Founder effect and estimation of the age of the c.892C>T (p.Arg298Cys) mutation in LMNA associated to Charcot-Marie-Tooth subtype CMT2B1 in families from North Western Africa. *Annals of Human Genetics*, **2008**, 72, 590-7	2.2	23
150	A Heterozygous ZMPSTE24 Mutation Associated with Severe Metabolic Syndrome, Ectopic Fat Accumulation, and Dilated Cardiomyopathy. *Cells*, **2016**, 5,	7.9	23
149	Low penetrance in facioscapulohumeral muscular dystrophy type 1 with large pathological D4Z4 alleles: a cross-sectional multicenter study. *Orphanet Journal of Rare Diseases*, **2015**, 10, 2	4.2	22
148	Muscle Cells Fix Breaches by Orchestrating a Membrane Repair Ballet. *Journal of Neuromuscular Diseases*, **2018**, 5, 21-28	5	22
147	Exon 32 Skipping of Dysferlin Rescues Membrane Repair in PatientsNCells. *Journal of Neuromuscular Diseases*, **2015**, 2, 281-290	5	22
146	Molecular combing reveals complex 4q35 rearrangements in Facioscapulohumeral dystrophy. *Human Mutation*, **2017**, 38, 1432-1441	4.7	22
145	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. *European Journal of Human Genetics*, **2015**, 23, 1051-61	5.3	21
144	A High Throughput Phenotypic Screening reveals compounds that counteract premature osteogenic differentiation of HGPS iPS-derived mesenchymal stem cells. *Scientific Reports*, **2016**, 6, 34798	4.9	21
143	Exome sequencing as a second-tier diagnostic approach for clinically suspected dysferlinopathy patients. *Muscle and Nerve*, **2014**, 50, 1007-10	3.4	21
142	CAMOS, a nonprogressive, autosomal recessive, congenital cerebellar ataxia, is caused by a mutant zinc-finger protein, ZNF592. *European Journal of Human Genetics*, **2010**, 18, 1107-13	5.3	21
141	Assignment of a new congenital fibrosis of extraocular muscles type 3 (CFEOM3) locus, FEOM4, based on a balanced translocation t(2;13) (q37.3;q12.11) and identification of candidate genes. *Journal of Medical Genetics*, **2005**, 42, 253-9	5.8	21
140	Numerical aberrations, including trisomy 22 as the sole anomaly, are recurrent in follicular thyroid adenomas. *Genes Chromosomes and Cancer*, **1993**, 8, 63-6	5	21
139	Antisense-Based Progerin Downregulation in HGPS-Like PatientsNCells. *Cells*, **2016**, 5,	7.9	21
138	Tricho-Hepato-Enteric Syndrome mutation update: Mutations spectrum of TTC37 and SKIV2L, clinical analysis and future prospects. *Human Mutation*, **2018**, 39, 774-789	4.7	20
137	Vascular Endothelial Growth Factor A c.*237C>T polymorphism is associated with bevacizumab efficacy and related hypertension in metastatic colorectal cancer. *Digestive and Liver Disease*, **2015**, 47, 331-7	3.3	20

136	Prelamin A processing and functional effects in restrictive dermopathy. *Cell Cycle*, **2010**, 9, 4766-8	4.7	20
135	Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. *BMJ Open*, **2018**, 8, e021632	3	20
134	Neonatal screening for sickle cell disease in France: evaluation of the selective process. *Journal of Clinical Pathology*, **2010**, 63, 548-51	3.9	19
133	Altered splicing in prelamin A-associated premature aging phenotypes. *Progress in Molecular and Subcellular Biology*, **2006**, 44, 199-232	3	19
132	Clinical heterogeneity and a high proportion of novel mutations in a Chinese cohort of patients with dysferlinopathy. *Neurology India*, **2014**, 62, 635-9	0.7	18
131	LBR mutation and nuclear envelope defects in a patient affected with Reynolds syndrome. *Journal of Medical Genetics*, **2010**, 47, 361-70	5.8	18
130	Transcriptional explorations of CAPN3 identify novel splicing mutations, a large-sized genomic deletion and evidence for messenger RNA decay. *Clinical Genetics*, **2007**, 72, 582-92	4	18
129	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. *American Journal of Human Genetics*, **2018**, 102, 364-374	11	17
128	A patient with limb girdle muscular dystrophy carries a TRIM32 deletion, detected by a novel CGH array, in compound heterozygosis with a nonsense mutation. *Neuromuscular Disorders*, **2013**, 23, 478-82	2.9	17
127	ATRX syndrome in a girl with a heterozygous mutation in the ATRX Zn finger domain and a totally skewed X-inactivation pattern. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2212-5	2.5	17
126	Screening of the CAPN3 gene in patients with possible LGMD2A. *Clinical Genetics*, **2006**, 69, 444-9	4	17
125	Detection of TRIM32 deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. *European Journal of Human Genetics*, **2015**, 23, 929-34	5.3	16
124	Identification of splicing defects caused by mutations in the dysferlin gene. *Human Mutation*, **2014**, 35, 1532-41	4.7	16
123	Novel mutations in the PRX and the MTMR2 genes are responsible for unusual Charcot-Marie-Tooth disease phenotypes. *Neuromuscular Disorders*, **2011**, 21, 543-50	2.9	16
122	Tight skin and limited joint movements as early presentation of Hutchinson-Gilford progeria in a 7-week-old infant. *European Journal of Pediatrics*, **2005**, 164, 283-6	4.1	16
121	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 340-2	5.5	15
120	Nuclear envelope remodelling during human spermiogenesis involves somatic B-type lamins and a spermatid-specific B3 lamin isoform. *Molecular Human Reproduction*, **2015**, 21, 225-36	4.4	15
119	Behavioral and molecular exploration of the AR-CMT2A mouse model Lmna (R298C/R298C). *NeuroMolecular Medicine*, **2012**, 14, 40-52	4.6	15

118	Eosinophilic infiltration related to CAPN3 mutations: a pathophysiological component of primary calpainopathy?. *Clinical Genetics*, **2011**, 80, 398-402	4	15
117	Identification of different genomic deletions and one duplication in the dysferlin gene using multiplex ligation-dependent probe amplification and genomic quantitative PCR. *Genetic Testing and Molecular Biomarkers*, **2009**, 13, 439-42	1.6	15
116	Low lamin A expression in lung adenocarcinoma cells from pleural effusions is a pejorative factor associated with high number of metastatic sites and poor Performance status. *PLoS ONE*, **2017**, 12, e0183136	3.7	15
115	Deciphering the complexity of the 4q and 10q subtelomeres by molecular combing in healthy individuals and patients with facioscapulohumeral dystrophy. *Journal of Medical Genetics*, **2019**, 56, 590-601	5.8	14
114	Multilineage Differentiation for Formation of Innervated Skeletal Muscle Fibers from Healthy and Diseased Human Pluripotent Stem Cells. *Cells*, **2020**, 9,	7.9	14
113	Clinical and allelic heterogeneity in a pediatric cohort of 11 patients carrying MFN2 mutation. *Brain and Development*, **2016**, 38, 498-506	2.2	14
112	Molecular basis of beta-thalassemia in Morocco: possible origins of the molecular heterogeneity. *Genetic Testing and Molecular Biomarkers*, **2008**, 12, 563-8		14
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110	Respiratory and cardiac function in japanese patients with dysferlinopathy. *Muscle and Nerve*, **2016**, 53, 394-401	3.4	14
109	Novel mutations cause an aggressive atypical neonatal progeria without progerin accumulation. *Journal of Medical Genetics*, **2016**, 53, 776-785	5.8	14
108	A multicenter retrospective study of charcot-marie-tooth disease type 4B (CMT4B) associated with mutations in myotubularin-related proteins (MTMRs). *Annals of Neurology*, **2019**, 86, 55-67	9.4	13
107	Proposition of adjustments to the ACMG-AMP framework for the interpretation of MEN1 missense variants. *Human Mutation*, **2019**, 40, 661-674	4.7	13
106	Two novel missense mutations in FGD4/FRABIN cause Charcot-Marie-Tooth type 4H (CMT4H). *Journal of the Peripheral Nervous System*, **2012**, 17, 141-6	4.7	13
105	A whole-genome scan in a large family with leukodystrophy and oligodontia reveals linkage to 10q22. *Neurogenetics*, **2011**, 12, 73-8	3	13
104	Beta-thalassemia intermedia due to two novel mutations in the promoter region of the beta-globin gene. *European Journal of Haematology*, **2008**, 80, 346-50	3.8	13
103	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. *Nature Communications*, **2020**, 11, 4589	17.4	13
102	Toward an objective measure of functional disability in dysferlinopathy. *Muscle and Nerve*, **2016**, 53, 49-57	3.4	13
101	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. *Annals of Clinical and Translational Neurology*, **2019**, 6, 642-654	5.3	12

100	HIV-1 infection and first line ART induced differential responses in mitochondria from blood lymphocytes and monocytes: the ANRS EP45 "Aging" study. *PLoS ONE*, **2012**, 7, e41129	3.7	12
99	Long-term follow-up study on patients with Miyoshi phenotype of distal muscular dystrophy. *European Journal of Neurology*, **2013**, 20, 968-74	6	11
98	HIV protease inhibitors do not cause the accumulation of prelamin A in PBMCs from patients receiving first line therapy: the ANRS EP45 "aging" study. *PLoS ONE*, **2012**, 7, e53035	3.7	11
97	Immunolabelling and flow cytometry as new tools to explore dysferlinopathies. *Neuromuscular Disorders*, **2010**, 20, 57-60	2.9	11
96	Nuclear localization of a novel human syntaxin 1B isoform. *Gene*, **2008**, 423, 160-71	3.8	11
95	Prenatal detection of the 17p11.2 duplication in Charcot-Marie-Tooth disease type 1A: necessity of a multidisciplinary approach for heterogeneous disorders. *European Journal of Human Genetics*, **2002**, 10, 297-302	5.3	11
94	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. *Human Molecular Genetics*, **2019**, 28, 2378-2394	5.6	10
93	Identification of a ERCC5 c.2333T>C (L778P) Variant in Two Tunisian Siblings With Mild Xeroderma Pigmentosum Phenotype. *Frontiers in Genetics*, **2019**, 10, 111	4.5	9
92	MicroRNAs in hereditary and sporadic premature aging syndromes and other laminopathies. *Aging Cell*, **2018**, 17, e12766	9.9	9
91	Segregation between SMCHD1 mutation, D4Z4 hypomethylation and Facio-Scapulo-Humeral Dystrophy: a case report. *BMC Medical Genetics*, **2016**, 17, 66	2.1	9
90	Novel ALPK3 mutation in a Tunisian patient with pediatric cardiomyopathy and facio-thoraco-skeletal features. *Journal of Human Genetics*, **2018**, 63, 1077-1082	4.3	9
89	Lamins in Lung Cancer: Biomarkers and Key Factors for Disease Progression through miR-9 Regulation?. *Cells*, **2018**, 7,	7.9	9
88	A novel mutation in FGD4/FRABIN causes Charcot Marie Tooth disease type 4H in patients from a consanguineous Tunisian family. *Annals of Human Genetics*, **2013**, 77, 336-43	2.2	9
87	LEM-domain proteins are lost during human spermiogenesis but BAF and BAF-L persist. *Reproduction*, **2017**, 154, 387-401	3.8	9
86	Mutations in the X-linked form of Charcot-Marie-Tooth disease in the French population. *Neurogenetics*, **1997**, 1, 117-23	3	9
85	Fine-mapping the gene for X-linked myopathy with excessive autophagy. *Neurology*, **2008**, 71, 951-3	6.5	9
84	Hutchinson-Gilford progeria syndrome: Rejuvenating old drugs to fight accelerated ageing. *Methods*, **2021**, 190, 3-12	4.6	9
83	Clinical massively parallel sequencing for the diagnosis of myopathies. *Revue Neurologique*, **2015**, 171, 558-71	3	8

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78	Defective prelamin A processing resulting from LMNA or ZMPSTE24 mutations as the cause of restrictive dermopathy. *Archives of Dermatology*, **2005**, 141, 1473-4; author reply 1474		8
77	Isolation of cosmids and fetal brain cDNAs from the proximal long arm of human chromosome 22. *Human Molecular Genetics*, **1993**, 2, 535-40	5.6	8
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