Nicolas Levy

List of Publications by Year in Descending Order

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	miR-376a-3p and miR-376b-3p overexpression in Hutchinson-Gilford progeria fibroblasts inhibits cell proliferation and induces premature senescence.. *IScience*, **2022**, 25, 103757	6.1	1
242	Convergence of patient- and physician-reported outcomes in the French National Registry of Facioscapulohumeral Dystrophy.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 96	4.2	0
241	The Dysferlin Transcript Containing the Alternative Exon 40a is Essential for Myocyte Functions. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 754555	5.7	1
240	A novel bi-allelic loss-of-function mutation in STIM1 expands the phenotype of STIM1-related diseases. *Clinical Genetics*, **2021**, 100, 84-89	4	1
239	Commentary: Long-Term Exercise Reduces Formation of Tubular Aggregates and Promotes Maintenance of Ca Entry Units in Aged Muscle. *Frontiers in Physiology*, **2021**, 12, 663677	4.6	
238	Retrospective analysis and reclassification of DYSF variants in a large French series of dysferlinopathy patients. *Genetics in Medicine*, **2021**, 23, 1574-1577	8.1	2
237	Case Report: Identification of Novel Variants in and Genes in Two Tunisian Patients With Atypical Xeroderma Pigmentosum Phenotype. *Frontiers in Genetics*, **2021**, 12, 650639	4.5	1
236	Genotype-phenotype correlation in French patients with myelin protein zero gene-related inherited neuropathy. *European Journal of Neurology*, **2021**, 28, 2913-2921	6	1
235	Hutchinson-Gilford progeria syndrome: Rejuvenating old drugs to fight accelerated ageing. *Methods*, **2021**, 190, 3-12	4.6	9
234	Multiallelic rare variants support an oligogenic origin of sudden cardiac death in the young. *Herz*, **2021**, 46, 94-102	2.6	4
233	Refining NGS diagnosis of muscular disorders. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**, 92, 223-225	5.5	1
232	Evidence of SARS-CoV-2 re-infection with a different genotype. *Journal of Infection*, **2021**, 82, 84-123	18.9	27
231	The Brain Connectome after Gamma Knife Radiosurgery of the Ventro-Intermediate Nucleus for Tremor: Marseille-Lausanne Radiobiology Study Protocol. *Stereotactic and Functional Neurosurgery*, **2021**, 99, 387-392	1.6	
230	Gene Mutations in North African Fanconi Anemia Patients. *Frontiers in Genetics*, **2021**, 12, 610050	4.5	0
229	The lncRNA 44s2 Study Applicability to the Design of 45-55 Exon Skipping Therapeutic Strategy for DMD. *Biomedicines*, **2021**, 9,	4.8	1
228	Novel partial loss-of-function variants in the tyrosyl-tRNA synthetase 1 (YARS1) gene involved in multisystem disease. *European Journal of Medical Genetics*, **2021**, 64, 104294	2.6	0
227	Extension of the phenotypic spectrum of GLE1-related disorders to a mild congenital form resembling congenital myopathy. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1277	2.3	2

226	Type 1 FSHD with 6-10 Repeated Units: Factors Underlying Severity in Index Cases and Disease Penetrance in Their Relatives Attention. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	2
225	Multilineage Differentiation for Formation of Innervated Skeletal Muscle Fibers from Healthy and Diseased Human Pluripotent Stem Cells. *Cells*, **2020**, 9,	7.9	14
224	Identification of a CDH12 potential candidate genetic variant for an autosomal dominant form of transgrediens and progrediens palmoplantar keratoderma in a Tunisian family. *Journal of Human Genetics*, **2020**, 65, 397-410	4.3	3
223	Unraveling Mutations in Metabolic Syndrome: Cellular Phenotype and Clinical Pitfalls. *Cells*, **2020**, 9,	7.9	6
222	Novel CAPN3 variant associated with an autosomal dominant calpainopathy. *Neuropathology and Applied Neurobiology*, **2020**, 46, 564-578	5.2	6
221	High prevalence of mutations in perilipin 1 in patients with precocious acute coronary syndrome. *Atherosclerosis*, **2020**, 293, 86-91	3.1	0
220	Splicing impact of deep exonic missense variants in CAPN3 explored systematically by minigene functional assay. *Human Mutation*, **2020**, 41, 1797-1810	4.7	3
219	Autosomal dominant segregation of CAPN3 c.598_612del15 associated with a mild form of calpainopathy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2538-2540	5.3	2
218	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. *Nature Communications*, **2020**, 11, 4589	17.4	13
217	Vulnerability of progeroid smooth muscle cells to biomechanical forces is mediated by MMP13. *Nature Communications*, **2020**, 11, 4110	17.4	7
216	A new tool CovReport generates easy-to-understand sequencing coverage summary for diagnostic reports. *Scientific Reports*, **2020**, 10, 6247	4.9	
215	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. *Annals of Clinical and Translational Neurology*, **2019**, 6, 642-654	5.3	12
214	Development and Validation of a New Risk Prediction Score for Life-Threatening Ventricular Tachyarrhythmias in Laminopathies. *Circulation*, **2019**, 140, 293-302	16.7	63
213	Whole Exome Sequencing Reveals a Large Genetic Heterogeneity and Revisits the Causes of Hypertrophic Cardiomyopathy. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002500	5.2	6
212	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
211	A severe clinical phenotype of Noonan syndrome with neonatal hypertrophic cardiomyopathy in the second case worldwide with RAF1 S259Y neomutation. *Genetical Research*, **2019**, 101, e6	1.1	3
210	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
209	Proposition of adjustments to the ACMG-AMP framework for the interpretation of MEN1 missense variants. *Human Mutation*, **2019**, 40, 661-674	4.7	13

208	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. *Human Molecular Genetics*, **2019**, 28, 2378-2394	5.6	10
207	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
206	Random walk with restart on multiplex and heterogeneous biological networks. *Bioinformatics*, **2019**, 35, 497-505	7.2	78
205	Outcomes of 4 years of molecular genetic diagnosis on a panel of genes involved in premature aging syndromes, including laminopathies and related disorders. *Orphanet Journal of Rare Diseases*, **2019**, 14, 288	4.2	1
204	Performance of semiconductor sequencing platform for non-invasive prenatal genetic screening for fetal aneuploidy: results from a multicenter prospective cohort study in a clinical setting. *Ultrasound in Obstetrics and Gynecology*, **2019**, 54, 246-254	5.8	4
203	Substrate Topography Modulates Cell Aging on a Progeria Cell Model. *ACS Biomaterials Science and Engineering*, **2018**, 4, 1498-1504	5.5	6
202	An overview of treatment strategies for Hutchinson-Gilford Progeria syndrome. *Nucleus*, **2018**, 9, 246-257	3.9	44
201	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
200	MicroRNAs in hereditary and sporadic premature aging syndromes and other laminopathies. *Aging Cell*, **2018**, 17, e12766	9.9	9
199	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
198	Inflammatory facioscapulohumeral muscular dystrophy type 2 in 18p deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1760-1763	2.5	2
197	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
196	Muscle Cells Fix Breaches by Orchestrating a Membrane Repair Ballet. *Journal of Neuromuscular Diseases*, **2018**, 5, 21-28	5	22
195	The involvement of the nuclear lamina in human and rodent spermiogenesis: a systematic review. *Basic and Clinical Andrology*, **2018**, 28, 7	2.8	3
194	Lamins in Lung Cancer: Biomarkers and Key Factors for Disease Progression through miR-9 Regulation?. *Cells*, **2018**, 7,	7.9	9
193	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
192	Pathological modelling of pigmentation disorders associated with Hutchinson-Gilford Progeria Syndrome (HGPS) revealed an impaired melanogenesis pathway in iPS-derived melanocytes. *Scientific Reports*, **2018**, 8, 9112	4.9	2
191	The French National Registry of patients with Facioscapulohumeral muscular dystrophy. *Orphanet Journal of Rare Diseases*, **2018**, 13, 218	4.2	6

190	Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. *BMJ Open*, **2018**, 8, e021632	3	20
189	Clinical profile of comorbidity of rare diseases in a Tunisian patient: a case report associating incontinentia pigmenti and Noonan syndrome. *BMC Pediatrics*, **2018**, 18, 286	2.6	2
188	Dysferlin Exon 32 Skipping in Patient Cells. *Methods in Molecular Biology*, **2018**, 1828, 489-496	1.4	3
187	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. *Nature Genetics*, **2017**, 49, 249-255	36.3	60
186	Genetic Characterization of a French Cohort of GNE-mutation negative inclusion body myopathy patients with exome sequencing. *Muscle and Nerve*, **2017**, 56, 993-997	3.4	6
185	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
184	Abnormal retention of nuclear lamina and disorganization of chromatin-related proteins in spermatozoa from DPY19L2-deleted globozoospermic patients. *Reproductive BioMedicine Online*, **2017**, 35, 562-570	4	6
183	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
182	LEM-domain proteins are lost during human spermiogenesis but BAF and BAF-L persist. *Reproduction*, **2017**, 154, 387-401	3.8	9
181	MG132-induced progerin clearance is mediated by autophagy activation and splicing regulation. *EMBO Molecular Medicine*, **2017**, 9, 1294-1313	12	63
180	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
179	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
178	Homozygous deletion of SUN5 in three men with decapitated spermatozoa. *Human Molecular Genetics*, **2017**, 26, 3167-3171	5.6	30
177	Molecular combing reveals complex 4q35 rearrangements in Facioscapulohumeral dystrophy. *Human Mutation*, **2017**, 38, 1432-1441	4.7	22
176	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 340-2	5.5	15
175	Molecular combing: A new tool in diagnosing leukemia. *Cancer Biomarkers*, **2016**, 17, 405-409	3.8	2
174	Segregation between SMCHD1 mutation, D4Z4 hypomethylation and Facio-Scapulo-Humeral Dystrophy: a case report. *BMC Medical Genetics*, **2016**, 17, 66	2.1	9
173	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

172	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
171	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
170	Clinical and allelic heterogeneity in a pediatric cohort of 11 patients carrying MFN2 mutation. *Brain and Development*, **2016**, 38, 498-506	2.2	14
169	A novel overlapping phenotype characterized by lipodystrophy, mandibular dysplasia, and dilated cardiomyopathy associated with a new mutation in the LMNA gene. *International Journal of Cardiology*, **2016**, 209, 317-8	3.2	8
168	Antisense-Based Progerin Downregulation in HGPS-Like PatientsNCells. *Cells*, **2016**, 5,	7.9	21
167	A Heterozygous ZMPSTE24 Mutation Associated with Severe Metabolic Syndrome, Ectopic Fat Accumulation, and Dilated Cardiomyopathy. *Cells*, **2016**, 5,	7.9	23
166	Respiratory and cardiac function in japanese patients with dysferlinopathy. *Muscle and Nerve*, **2016**, 53, 394-401	3.4	14
165	Novel mutations cause an aggressive atypical neonatal progeria without progerin accumulation. *Journal of Medical Genetics*, **2016**, 53, 776-785	5.8	14
164	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
163	Coverage Analysis of Lists of Genes involved in Heterogeneous Genetic Diseases following Benchtop Exome Sequencing using the Ion Proton. *Journal of Genetics*, **2016**, 95, 203-8	1.2	3
162	Toward an objective measure of functional disability in dysferlinopathy. *Muscle and Nerve*, **2016**, 53, 49-57	3.4	13
161	Clinical massively parallel sequencing for the diagnosis of myopathies. *Revue Neurologique*, **2015**, 171, 558-71	3	8
160	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
159	PMPCA mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. *Brain*, **2015**, 138, 1505-17	11.2	43
158	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. *European Journal of Human Genetics*, **2015**, 23, 1051-61	5.3	21
157	Dysferlinopathy in Iran: Clinical and genetic report. *Journal of the Neurological Sciences*, **2015**, 359, 256-9	3.2	7
156	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
155	Comment on: A novel dysferlin-mutant pseudoexon bypassed with antisense oligonucleotides. *Annals of Clinical and Translational Neurology*, **2015**, 2, 783-4	5.3	2

154	WDR73 Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. *Human Mutation*, **2015**, 36, 1021-8	4.7	31
153	Exon 32 Skipping of Dysferlin Rescues Membrane Repair in PatientsNCells. *Journal of Neuromuscular Diseases*, **2015**, 2, 281-290	5	22
152	Novel Pathogenic Variants in a French Cohort Widen the Mutational Spectrum of´GNE Myopathy. *Journal of Neuromuscular Diseases*, **2015**, 2, 131-136	5	6
151	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
150	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
149	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
148	Organism Models: Choosing the Right Model. *Neuromethods*, **2015**, 3-27	0.4	2
147	Differential DNA methylation of the D4Z4 repeat in patients with FSHD and asymptomatic carriers. *Neurology*, **2014**, 83, 733-42	6.5	58
146	Prelamin A accumulation in endothelial cells induces premature senescence and functional impairment. *Atherosclerosis*, **2014**, 237, 45-52	3.1	39
145	Exome sequencing as a second-tier diagnostic approach for clinically suspected dysferlinopathy patients. *Muscle and Nerve*, **2014**, 50, 1007-10	3.4	21
144	Exome sequencing identifies a novel mutation in PIK3R1 as the cause of SHORT syndrome. *BMC Medical Genetics*, **2014**, 15, 51	2.1	26
143	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
142	A new lamin a mutation associated with acrogeria syndrome. *Journal of Investigative Dermatology*, **2014**, 134, 2274-2277	4.3	5
141	Identification of splicing defects caused by mutations in the dysferlin gene. *Human Mutation*, **2014**, 35, 1532-41	4.7	16
140	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
139	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
138	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
137	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

136	Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle and Nerve*, **2014**, 50, 477-87	3.4	281
135	Entire CAPN3 gene deletion in a patient with limb-girdle muscular dystrophy type 2A. *Muscle and Nerve*, **2014**, 50, 448-53	3.4	5
134	Long-term follow-up study on patients with Miyoshi phenotype of distal muscular dystrophy. *European Journal of Neurology*, **2013**, 20, 968-74	6	11
133	VMA21 deficiency prevents vacuolar ATPase assembly and causes autophagic vacuolar myopathy. *Acta Neuropathologica*, **2013**, 125, 439-57	14.3	101
132	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
131	Constitutive activation of the calcium sensor STIM1 causes tubular-aggregate myopathy. *American Journal of Human Genetics*, **2013**, 92, 271-8	11	133
130	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
129	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
128	SHAPE AND TEXTURE INDEXES APPLICATION TO CELL NUCLEI CLASSIFICATION. *International Journal of Pattern Recognition and Artificial Intelligence*, **2013**, 27, 1357002	1.1	98
127	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
126	Deregulation of the protocadherin gene FAT1 alters muscle shapes: implications for the pathogenesis of facioscapulohumeral dystrophy. *PLoS Genetics*, **2013**, 9, e1003550	6	58
125	A collodion baby with facial dysmorphism, limb anomalies, pachygyria and genital hypoplasia: a mild form of Neu-laxova syndrome or a new entity?. *Annals of Dermatology*, **2013**, 25, 483-8	0.4	3
124	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
123	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
122	Further heterogeneity in myopathy with tubular aggregates?. *Muscle and Nerve*, **2012**, 46, 984-5	3.4	2
121	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
120	An inherited LMNA gene mutation in atypical Progeria syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2881-7	2.5	32
119	Diagnostic strategy for limb-girdle muscular dystrophies. *Revue Neurologique*, **2012**, 168, 919-26	3	4

118	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
117	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
116	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
115	Behavioral and molecular exploration of the AR-CMT2A mouse model Lmna (R298C/R298C). *NeuroMolecular Medicine*, **2012**, 14, 40-52	4.6	15
114	Next generation sequencing for molecular diagnosis of neuromuscular diseases. *Acta Neuropathologica*, **2012**, 124, 273-83	14.3	70
113	SKIV2L mutations cause syndromic diarrhea, or trichohepatoenteric syndrome. *American Journal of Human Genetics*, **2012**, 90, 689-92	11	108
112	Validation of comparative genomic hybridization arrays for the detection of genomic rearrangements of the calpain-3 and dysferlin genes. *Clinical Genetics*, **2012**, 81, 99-101	4	8
111	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
110	A 10´Mb duplication in chromosome band 5q31.3-5q33.1 associated with late-onset lipodystrophy, ichthyosis, epilepsy and glomerulonephritis. *European Journal of Medical Genetics*, **2011**, 54, 310-3	2.6	5
109	A novel MYH7 mutation links congenital fiber type disproportion and myosin storage myopathy. *Neuromuscular Disorders*, **2011**, 21, 254-62	2.9	42
108	Novel ancestral Dysferlin splicing mutation which migrated from the Iberian peninsula to South America. *Neuromuscular Disorders*, **2011**, 21, 328-37	2.9	6
107	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
106	Translational research and therapeutic perspectives in dysferlinopathies. *Molecular Medicine*, **2011**, 17, 875-82	6.2	29
105	Eosinophilic infiltration related to CAPN3 mutations: a pathophysiological component of primary calpainopathy?. *Clinical Genetics*, **2011**, 80, 398-402	4	15
104	Restrictive dermopathy in a Turkish newborn. *Pediatric Dermatology*, **2011**, 28, 408-11	1.9	6
103	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
102	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
101	A whole-genome scan in a large family with leukodystrophy and oligodontia reveals linkage to 10q22. *Neurogenetics*, **2011**, 12, 73-8	3	13

100	Immortalized pathological human myoblasts: towards a universal tool for the study of neuromuscular disorders. *Skeletal Muscle*, **2011**, 1, 34	5.1	160
99	Ambiguous genitalia, microcephaly, seizures, bone malformations, and early death: a distinct MCA/MR syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1147-51	2.5	1
98	Molecular combing reveals allelic combinations in facioscapulohumeral dystrophy. *Annals of Neurology*, **2011**, 70, 627-33	9.4	33
97	Novel mutations in TTC37 associated with tricho-hepato-enteric syndrome. *Human Mutation*, **2011**, 32, 277-81	4.7	42
96	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
95	Analytical evaluation of the Tosoh HLC-723 G8 automated HPLC analyzer for hemoglobin analysis in beta-thalassemia mode. *Clinical Biochemistry*, **2011**, 44, 441-3	3.5	5
94	A conserved splicing mechanism of the LMNA gene controls premature aging. *Human Molecular Genetics*, **2011**, 20, 4540-55	5.6	54
93	High prevalence of laminopathies among patients with metabolic syndrome. *Human Molecular Genetics*, **2011**, 20, 3779-86	5.6	43
92	Splicing-directed therapy in a new mouse model of human accelerated aging. *Science Translational Medicine*, **2011**, 3, 106ra107	17.5	240
91	New multiplex PCR-based protocol allowing indirect diagnosis of FSHD on single cells: can PGD be offered despite high risk of recombination?. *European Journal of Human Genetics*, **2010**, 18, 533-8	5.3	8
90	Therapeutic exon Ñswitching Ñfor dysferlinopathies?. *European Journal of Human Genetics*, **2010**, 18, 969-70; author reply 971	5.3	7
89	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
88	Neonatal screening for sickle cell disease in France: evaluation of the selective process. *Journal of Clinical Pathology*, **2010**, 63, 548-51	3.9	19
87	Clinicopathologic features of histiocytic lesions following ALL, with a review of the literature. *Pediatric and Developmental Pathology*, **2010**, 13, 225-37	2.2	53
86	Prelamin A processing and functional effects in restrictive dermopathy. *Cell Cycle*, **2010**, 9, 4766-8	4.7	20
85	LBR mutation and nuclear envelope defects in a patient affected with Reynolds syndrome. *Journal of Medical Genetics*, **2010**, 47, 361-70	5.8	18
84	Immunolabelling and flow cytometry as new tools to explore dysferlinopathies. *Neuromuscular Disorders*, **2010**, 20, 57-60	2.9	11
83	A naturally occurring human minidysferlin protein repairs sarcolemmal lesions in a mouse model of dysferlinopathy. *Science Translational Medicine*, **2010**, 2, 50ra69	17.5	59

82	Exclusion of mutations in the dysferlin alternative exons 1 of DYSF-v1, 5a, and 40a in a cohort of 26 patients. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 153-4	1.6	7
81	Polymorphisms of the lamina maturation pathway and their association with the metabolic syndrome: the DESIR prospective study. *Journal of Molecular Medicine*, **2010**, 88, 193-201	5.5	5
80	Efficient bypass of mutations in dysferlin deficient patient cells by antisense-induced exon skipping. *Human Mutation*, **2010**, 31, 136-42	4.7	75
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78	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
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