Gabriel Miltenberger-Miltenyi

List of Publications by Year
in descending order

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41 papers	1,340 citations	471509 17 h-index	345221 36 g-index
44 all docs	44 docs citations	44 times ranked	2801 citing authors

#	Article	IF	Citations
1	A Panâ€<scp>E</scp>uropean Study of theC9orf72Repeat Associated with<scp>FTLD</scp>: Geographic Prevalence, Genomic Instability, and Intermediate Repeats. Human Mutation, 2013, 34, 363-373.	2.5	247
2	Atomic Force Microscopy-Based Molecular Recognition of a Fibrinogen Receptor on Human Erythrocytes. ACS Nano, 2010, 4, 4609-4620.	14.6	136
3	Rare mutations in SQSTM1 modify susceptibility to frontotemporal lobar degeneration. Acta Neuropathologica, 2014, 128, 397-410.	7.7	93
4	TBK1 Mutation Spectrum in an Extended European Patient Cohort with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Human Mutation, 2017, 38, 297-309.	2.5	87
5	Serum lipid alterations in GBA-associated Parkinson's disease. Parkinsonism and Related Disorders, 2017, 44, 58-65.	2.2	73
6	Mutations and polymorphisms in the human methyl CpG-binding protein MECP2. Human Mutation, 2003, 22, 107-115.	2.5	68
7	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, 100-106.	0.6	64
8	Primary Pulmonary Hypertension in Children May Have a Different Genetic Background Than in Adults. Pediatric Research, 2004, 56, 571-578.	2.3	49
9	Sarcomeric hypertrophic cardiomyopathy: Genetic profile in a Portuguese population. Revista Portuguesa De Cardiologia, 2012, 31, 577-587.	0.5	41
10	Fabry Disease and the Heart: A Comprehensive Review. International Journal of Molecular Sciences, 2021, 22, 4434.	4.1	40
11	Clinical and Electrophysiological Features in Charcot-Marie-Tooth Disease With Mutations in the NEFL Gene. Archives of Neurology, 2007, 64, 966.	4.5	37
12	Gene Expression Differences in Peripheral Blood of Parkinsonâ€™s Disease Patients with Distinct Progression Profiles. PLoS ONE, 2016, 11, e0157852.	2.5	36
13	Fabry Disease Therapy: State-of-the-Art and Current Challenges. International Journal of Molecular Sciences, 2021, 22, 206.	4.1	35
14	Primary pulmonary hypertension may be a heterogeneous disease with a second locus on chromosome 2q31. Journal of the American College of Cardiology, 2003, 41, 2237-2244.	2.8	29
15	Founder effect of Fabry disease due to p.F113L mutation: Clinical profile of a late-onset phenotype. Molecular Genetics and Metabolism, 2020, 129, 150-160.	1.1	29
16	Increased monohexosylceramide levels in the serum of established rheumatoid arthritis patients. Rheumatology, 2020, 59, 2085-2089.	1.9	21
17	Genetic association study of UCMA/GRP and OPTN genes (PDB6 locus) with Paget's disease of bone. Bone, 2012, 51, 720-728.	2.9	20
18	Pachydermoperiostosis in an African patient caused by a Chinese/Japanese SLCO2A1 mutationâ€”Case report and review of literature. Seminars in Arthritis and Rheumatism, 2014, 43, 566-569.	3.4	20

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19	Natural history of the late-onset phenotype of Fabry disease due to the p.F113L mutation. Molecular Genetics and Metabolism Reports, 2020, 22, 100565.	1.1	20
20	Rare nonsynonymous variants in SORT1 are associated with increased risk for frontotemporal dementia. Neurobiology of Aging, 2018, 66, 181.e3-181.e10.	3.1	19
21	Linkage Analysis in a Large Family With Primary Pulmonary Hypertension. Chest, 2002, 121, 54S-56S.	0.8	18
22	No supportive evidence for TIA1 gene mutations in a European cohort of ALS-FTD spectrum patients. Neurobiology of Aging, 2018, 69, 293.e9-293.e11.	3.1	15
23	Identification and in silico analysis of 14 novel GJB1, MPZ and PMP22 gene mutations. European Journal of Human Genetics, 2009, 17, 1154-1159.	2.8	14
24	C9orf72 expansion is associated with accelerated decline of respiratory function and decreased survival in amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 118-120.	1.9	14
25	Parkinsonâ€™s Disease and Fabry Disease: Clinical, Biochemical and Neuroimaging Analysis of Three Pedigrees. Journal of Parkinson's Disease, 2020, 10, 141-152.	2.8	14
26	Genetic polymorphisms of proangiogenic factors seem to favor hepatocellular carcinoma development in alcoholic cirrhosis. European Journal of Gastroenterology and Hepatology, 2014, 26, 438-443.	1.6	13
27	Choreaâ€acanthocytosis presenting as motor neuron disease. Muscle and Nerve, 2012, 45, 293-295.	2.2	11
28	Molecular characterization of parathyroid tumors from two patients with hereditary colorectal cancer syndromes. Familial Cancer, 2012, 11, 355-362.	1.9	11
29	Cleidocranial dysplasia with severe parietal bone dysplasia: a new (p.Val124Serfs) RUNX2 mutation. Clinical Dysmorphology, 2010, 19, 150-152.	0.3	10
30	Phenotypic Variability of Familial and Sporadic Progranulin p.Gln257Profs*27 Mutation. Journal of Alzheimer's Disease, 2013, 37, 335-342.	2.6	9
31	Mutation spectrum of type I glycogen storage disease in Hungary. Journal of Inherited Metabolic Disease, 2005, 28, 939-944.	3.6	7
32	Hypertrophic cardiomyopathy or non-compaction? How the first impression can be wrong. International Journal of Cardiology, 2012, 158, e53-e54.	1.7	6
33	Cardiac Anderson-Fabry disease: Lessons from a 25-year-follow up. Revista Portuguesa De Cardiologia, 2014, 33, 247.e1-247.e7.	0.5	6
34	Screening for Fabry disease in patients with left ventricular noncompaction. Revista Portuguesa De Cardiologia, 2019, 38, 709-716.	0.5	5
35	Predictors of Fabry disease in patients with hypertrophic cardiomyopathy: How to guide the diagnostic strategy?. American Heart Journal, 2020, 226, 114-126.	2.7	5
36	Mild Left Ventricular Hypertrophy Unravels a Novel Nonsense Mutation of the GLA Gene Associated with the Classical Phenotype of Fabry Disease. Cardiology, 2017, 137, 67-73.	1.4	4

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37	Novel mutation in the KCNH2 gene associated with long QT syndrome. Revista Portuguesa De Cardiologia, 2013, 32, 163-164.	0.5	2
38	Teste genÃ©tico post mortem, o diagnÃ³stico clÃnico nÃ£o se esgota com a morte do doente. Revista Portuguesa De Cardiologia, 2019, 38, 503-509.	0.5	2
39	Rare Association of two Genetic Causes of Sudden Death in a Young Survivor. Arquivos Brasileiros De Cardiologia, 2017, 108, 184-186.	0.8	2
40	Identification of the Fibrinogen Receptor on Human Erythrocyte by AFM-Based Force Spectroscopy. Biophysical Journal, 2011, 100, 479a.	0.5	1
41	Soft Tissue Metastasis of Parathyroid Carcinoma: Description of a Difficult Case. Journal of Endocrinology and Metabolism, 2015, 5, 184-188.	0.4	0