Paola Mandich

List of Publications by Year
in descending order

77 papers	2,117 citations	236925 25 h-index	254184 43 g-index
77 all docs	77 docs citations	77 times ranked	3612 citing authors

#	Article	IF	Citations
1	Hereditary transthyretin amyloidosis overview. Neurological Sciences, 2022, 43, 595-604.	1.9	39
2	An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic <scp>hyperCKemia</scp>. Muscle and Nerve, 2022, 65, 96-104.	2.2	10
3	Association of Essential Tremor With Novel Risk Loci. JAMA Neurology, 2022, 79, 185.	9.0	17
4	Genetic Workup for Charcotâ€“Marieâ€“Tooth Neuropathy: A Retrospective Single-Site Experience Covering 15 Years. Life, 2022, 12, 402.	2.4	4
5	A novel mutation in COL3A1 associates to vascular Ehlersâ€“Danlos syndrome with predominant musculoskeletal involvement. Molecular Genetics & Genomic Medicine, 2021, 9, e1753.	1.2	4
6	An eleven-year history of Vanishing White Matter Disease in an adult patient with no cognitive decline and EIF2B5 mutations. A case report. Neurocase, 2021, 27, 452-456.	0.6	2
7	People with Charcot-Marie-Tooth disease and COVID-19: Impaired physical conditions due to the lockdown. An International cross-sectional survey. Annals of Physical and Rehabilitation Medicine, 2020, 63, 557-559.	2.3	5
8	Recommendations for pre-symptomatic genetic testing for hereditary transthyretin amyloidosis in the era of effective therapy: a multicenter Italian consensus. Orphanet Journal of Rare Diseases, 2020, 15, 348.	2.7	22
9	A novel mutation of Twinkle in Perrault syndrome: A not rare diagnosis?. Annals of Human Genetics, 2020, 84, 417-422.	0.8	7
10	Early onset demyelinating Charcotâ€Marieâ€Tooth disease caused by a novel inâ€frame isoleucine deletion in peripheral myelin protein 2. Journal of the Peripheral Nervous System, 2020, 25, 102-106.	3.1	6
11	A novel mutation in the N-terminal acting-binding domain of Filamin C protein causing a distal myofibrillar myopathy. Journal of the Neurological Sciences, 2019, 398, 75-78.	0.6	10
12	Diagnostic Value of Sural Nerve Biopsy: Retrospective Analysis of Clinical Cases From 1981 to 2017. Frontiers in Neurology, 2019, 10, 1218.	2.4	12
13	Twenty years of molecular analyses in amyotrophic lateral sclerosis: genetic landscape of Italian patients. Neurobiology of Aging, 2018, 66, 179.e5-179.e16.	3.1	16
14	Spinocerebellar ataxia 17: full phenotype in a 41 CAG/CAA repeats carrier. Cerebellum and Ataxias, 2018, 5, 7.	1.9	21
15	Role of <i>MAPT</i> in Pure Motor Neuron Disease: Report of a Recurrent Mutation in Italian Patients. Neurodegenerative Diseases, 2018, 18, 310-314.	1.4	12
16	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
17	Autosomal-dominant transthyretin (TTR)-related amyloidosis is not a frequent CMT2 neuropathy â€œin disguiseâ€• Orphanet Journal of Rare Diseases, 2018, 13, 177.	2.7	2
18	Two novel cases of compound heterozygous mutations in mitofusin2: Finding out the inheritance. Neuromuscular Disorders, 2017, 27, 377-381.	0.6	1

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19	Genetic Counseling Dilemmas for a Patient with Sporadic Amyotrophic Lateral Sclerosis, Frontotemporal Degeneration & Parkinson's Disease. Journal of Genetic Counseling, 2017, 26, 442-446.	1.6	3
20	1993-2014: two decades of predictive testing for Huntington's disease at the Medical Genetics Unit of the University of Genoa. Molecular Genetics & Genomic Medicine, 2017, 5, 473-480.	1.2	10
21	Theory of Mind Is Impaired in Mild to Moderate Huntingtonâ€™s Disease Independently from Global Cognitive Functioning. Frontiers in Psychology, 2017, 8, 80.	2.1	17
22	Clinical epidemiology of amyotrophic lateral sclerosis in Liguria, Italy: An update of LIGALS register. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 535-542.	1.7	29
23	Molecular Chaperones in the Pathogenesis of Amyotrophic Lateral Sclerosis: The Role of HSPB1. Human Mutation, 2016, 37, 1202-1208.	2.5	45
24	Genetic factors and systemic sclerosis. Autoimmunity Reviews, 2016, 15, 427-432.	5.8	59
25	A novel Arg147Trp MATR3 missense mutation in a slowly progressive ALS Italian patient. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 530-531.	1.7	27
26	Sural nerve biopsy and functional studies support the pathogenic role of a novel <scp>MPZ</scp> mutation. Neuropathology, 2015, 35, 254-259.	1.2	4
27	Quiz Page February 2015. American Journal of Kidney Diseases, 2015, 65, A17-A19.	1.9	0
28	HFE p.H63D polymorphism does not influence ALS phenotype and survival. Neurobiology of Aging, 2015, 36, 2906.e7-2906.e11.	3.1	8
29	Neuroimaging features inC9orf72andTARDBPdouble mutation with FTD phenotype. Neurocase, 2015, 21, 529-534.	0.6	8
30	Complexities of Genetic Counseling for ALS: A Case of Two Siblings with Discordant Genetic Test Results. Journal of Genetic Counseling, 2015, 24, 553-557.	1.6	9
31	CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2015, 36, 1767.e3-1767.e6.	3.1	44
32	Diagnostic genetic testing for Huntington's disease. Practical Neurology, 2015, 15, 80-84.	1.1	44
33	Charcotâ€Marieâ€Tooth disease: frequency of genetic subtypes in a Southern Italy population. Journal of the Peripheral Nervous System, 2014, 19, 292-298.	3.1	64
34	TNFÎ± induces the expression of genes associated with endothelial dysfunction through p38MAPK-mediated down-regulation of miR-149. Biochemical and Biophysical Research Communications, 2014, 443, 246-251.	2.1	34
35	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
36	TNF-Î± Gene Polymorphisms: Association with Disease Susceptibility and Response to Anti-TNF-Î± Treatment in Psoriatic Arthritis. Journal of Investigative Dermatology, 2014, 134, 2503-2509.	0.7	89

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37	A novel LITAF/SIMPLE mutation within a family with a demyelinating form of Charcotâ€“Marieâ€“Tooth disease. Journal of the Neurological Sciences, 2014, 343, 183-186.	0.6	15
38	The FIG4 gene does not play a major role in causing ALS in Italian patients. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 228-229.	1.7	8
39	Analysis of <scp> LMNB </scp> 1 Duplications in Autosomal Dominant Leukodystrophy Provides Insights into Duplication Mechanisms and Alleleâ€Specific Expression. Human Mutation, 2013, 34, 1160-1171.	2.5	33
40	Clinical epidemiology of ALS in Liguria, Italy. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 52-57.	1.7	26
41	Enlarging the clinical spectrum associated with C9orf 72 repeat expansions: Findings in an Italian cohort of patients with Parkinsonian syndromes and relevance for genetic counselling. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 479-480.	1.7	12
42	T137A variant is a pathogenetic SOD1 mutation associated with a slowly progressive ALS phenotype. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 398-399.	2.1	4
43	A novel autosomal dominant GDAP1 mutation in an Italian CMT2 family. Journal of the Peripheral Nervous System, 2012, 17, 351-355.	3.1	13
44	Fast course ALS presenting with vocal cord paralysis: Clinical features, bioinformatic and modelling analysis of the novel SOD1 Gly147Ser mutation. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 144-148.	2.1	14
45	C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Neurobiology of Aging, 2012, 33, 1848.e15-1848.e20.	3.1	76
46	Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Brain, 2012, 135, 784-793.	7.6	182
47	Gain of glycosylation: A new pathomechanism of myelin protein zero mutations. Annals of Neurology, 2012, 71, 427-431.	5.3	20
48	HSPB1 and HSPB8 in inherited neuropathies: study of an Italian cohort of dHMN and CMT2 patients. Journal of the Peripheral Nervous System, 2011, 16, 287-294.	3.1	57
49	Currarino syndrome with pelvic neuroendocrine tumor diagnosed by postâ€mortem genetic analysis of tissue specimens. American Journal of Medical Genetics, Part A, 2011, 155, 2750-2753.	1.2	12
50	Severe Neuropathy After Diphtheria-Tetanus-Pertussis Vaccination in a Child Carrying a Novel Frame-Shift Mutation in the Small Heat-Shock Protein 27 Gene. Journal of Child Neurology, 2010, 25, 107-109.	1.4	32
51	The spectrum of GNE mutations: allelic heterogeneity for a common phenotype. Neurological Sciences, 2010, 31, 377-380.	1.9	17
52	Enlarging clinical spectrum of FALS with TARDBP gene mutations: S393L variant in an Italian family showing phenotypic variability and relevance for genetic counselling. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 223-227.	2.1	29
53	Quantitative Fluorescence-Polymerase Chain Reaction Assay for the Detection of the Duplication of the Charcot Marie Tooth Disease Type 1A Critical Region. Genetic Testing and Molecular Biomarkers, 2010, 14, 225-231.	0.7	0
54	Friedreichâ€™s Ataxia: A New Mutation in Two Compound Heterozygous Siblings with Unusual Clinical Onset. European Neurology, 2009, 61, 240-243.	1.4	5

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55	Reliability and Reproducibility of a RNA Preamplification Method for Low-density Array Analysis From Formalin-fixed Paraffin-embedded Breast Cancer Samples. Diagnostic Molecular Pathology, 2009, 18, 112-118.	2.1	19
56	Clinical features and molecular modelling of novel MPZ mutations in demyelinating and axonal neuropathies. European Journal of Human Genetics, 2009, 17, 1129-1134.	2.8	35
57	Two families with novel PMP22 point mutations: genotypeâ€“phenotype correlation. Journal of the Peripheral Nervous System, 2009, 14, 208-212.	3.1	9
58	Germline mutations in the von Hippelâ€“Lindau gene in Italian patients. European Journal of Medical Genetics, 2009, 52, 311-314.	1.3	11
59	Heterozygous D90A-SOD1 mutation in an Italian ALS patient with atypical presentation. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 492-492.	2.1	6
60	Gap junction beta 1 (GJB1) gene mutations in Italian patients with X-linked Charcot-Marie-Tooth disease. Journal of Human Genetics, 2008, 53, 529-533.	2.3	13
61	A putative regulatory subunit (NR3A) of the NMDA receptor complex as candidate gene for susceptibility to schizophrenia: a caseâ€“control study. Psychiatric Genetics, 2007, 17, 355-356.	1.1	5
62	No evidence of association between BDNF gene variants and age-at-onset of Huntington's disease. Neurobiology of Disease, 2006, 24, 274-279.	4.4	18
63	Variations in the NMDA receptor subunit 2B gene (GRIN2B) and schizophrenia: A caseâ€control study. American Journal of Medical Genetics Part A, 2004, 128B, 27-29.	2.4	33
64	Does parkin play a role in the peripheral nervous system? A family report. Movement Disorders, 2004, 19, 978-981.	3.9	33
65	Triplet Repeat Primed PCR (TP PCR) in Molecular Diagnostic Testing for Friedreich Ataxia. Journal of Molecular Diagnostics, 2004, 6, 285-289.	2.8	48
66	Mutational analysis of parkin gene by denaturing high-performance liquid chromatography (DHPLC) in essential tremor. Parkinsonism and Related Disorders, 2004, 10, 357-362.	2.2	20
67	Essential tremor is not associated with Î±â€synuclein gene haplotypes. Movement Disorders, 2003, 18, 823-826.	3.9	19
68	Different consequences of EGR2 mutants on the transactivation of human cx32 promoter. Neurobiology of Disease, 2003, 12, 89-95.	4.4	10
69	A family with autosomal dominant mutilating neuropathy not linked to either Charcotâ€“Marieâ€“Tooth disease type 2B (CMT2B) or hereditary sensory neuropathy type I (HSN I) loci. Neuromuscular Disorders, 2002, 12, 286-291.	0.6	9
70	Search for mutations in the EGR2 corepressor proteins, NAB1 and NAB2, in human peripheral neuropathies. Neurogenetics, 2002, 4, 37-41.	1.4	9
71	The D355V Mutation Decreases EGR2 Binding to an Element within the Cx32 Promoter. Neurobiology of Disease, 2001, 8, 700-706.	4.4	27
72	Corticobasal degeneration shares a common genetic background with progressive supranuclear palsy. Annals of Neurology, 2000, 47, 374-377.	5.3	216

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73	A Novel Mutation (D305V) in the early growth response 2 gene is associated with severe Charcot-Marie-Tooth type 1 disease. Human Mutation, 1999, 14, 353-354.	2.5	63
74	Exclusion of the ninjurin gene as a candidate for hereditary sensory neuropathies type I and type II. , 1999, 83, 409-410.		3
75	Three novel mutations in the von Hippel-Lindau tumour suppressor gene in Italian patients. Human Mutation, 1998, 11, S268-S270.	2.5	9
76	Correlation between PMP-22 messenger mRNA expression and phenotype in hereditary neuropathy with liability to pressure palsies. Annals of Neurology, 1997, 42, 866-872.	5.3	42
77	Identification of a 4 bp deletion (1560de14) in Po gene in a family with severe charcot-Marie-Tooth disease. Human Mutation, 1996, 7, 377-378.	2.5	13