Paola Carrera

List of Publications by Year in Descending Order

109 papers	2,889 citations	29 h-index	50 g-index
115 ext. papers	3,434 ext. citations	4.8 avg, IF	4.12 L-index

#	Paper	IF	Citations
109	Variants in a Cohort of Italian Patients With Amyotrophic Lateral Sclerosis.. *Frontiers in Neuroscience*, **2022**, 16, 833051	5.1	1
108	Implementation of preventive and predictive BRCA testing in patients with breast, ovarian, pancreatic, and prostate cancer: a position paper of Italian Scientific Societies. *ESMO Open*, **2022**, 7, 100459	6	1
107	Case report and ten-year follow-up of episodic ataxia type 2 due to a novel variant in CACNA1A. *ENeurologicalSci*, **2021**, 23, 100334	2.1	
106	Generation of Cells from iPSC of a MODY8 Patient with a Novel Mutation in the Carboxyl Ester Lipase (CEL) Gene. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e2322-e2333	5.6	2
105	Current scenario of the genetic testing for rare neurological disorders exploiting next generation sequencing. *Neural Regeneration Research*, **2021**, 16, 475-481	4.5	2
104	Germinal BRCA1-2 pathogenic variants (gBRCA1-2pv) and pancreatic cancer: epidemiology of an Italian patient cohort. *ESMO Open*, **2021**, 6, 100032	6	7
103	Structural MRI Signatures in Genetic Presentations of the Frontotemporal Dementia/Motor Neuron Disease Spectrum. *Neurology*, **2021**, 97, e1594-e1607	6.5	3
102	Burden of Rare Variants in ALS and Axonal Hereditary Neuropathy Genes Influence Survival in ALS: Insights from a Next Generation Sequencing Study of an Italian ALS Cohort. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	5
101	MSH6 gene pathogenic variant identified in familial pancreatic cancer in the absence of colon cancer. *European Journal of Gastroenterology and Hepatology*, **2020**, 32, 345-349	2.2	2
100	Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2020**, 91, 1001-1003	5.5	6
99	Serum phosphorylated neurofilament heavy-chain levels reflect phenotypic heterogeneity and are an independent predictor of survival in motor neuron disease. *Journal of Neurology*, **2020**, 267, 2272-2280	5.5	15
98	ADCY10 frameshift variant leading to severe recessive asthenozoospermia and segregating with absorptive hypercalciuria. *Human Reproduction*, **2019**, 34, 1155-1164	5.7	23
97	Recommendations for the implementation of BRCA testing in ovarian cancer patients and their relatives. *Critical Reviews in Oncology/Hematology*, **2019**, 140, 67-72	7	40
96	Expanding the spectrum of genes responsible for hereditary motor neuropathies. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 1171-1179	5.5	19
95	Concurrence of NMOSD and ALS in a patient with hexanucleotide repeat expansions of C9orf72. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2019**, 20, 449-452	3.6	1
94	An Open-Source Tool for Managing Time-Evolving Variant Annotation. *Lecture Notes in Computer Science*, **2019**, 1-8	0.9	
93	Survival prediction models in motor neuron disease. *European Journal of Neurology*, **2019**, 26, 1143-1152	6	11

92	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. *Annals of Neurology*, **2019**, 85, 470-481	9.4	72
91	Molecular Assay for Ovarian Cancer Patients: A Survey through Italian Departments of Oncology and Molecular and Genomic Diagnostic Laboratories. *Diagnostics*, **2019**, 9,	3.8	3
90	Efficacy of lentivirus-mediated gene therapy in an Omenn syndrome recombination-activating gene 2 mouse model is not hindered by inflammation and immune dysregulation. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 928-941.e8	11.5	16
89	ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2018**, 64, 157.e1-157.e5	5.6	24
88	Surfactant proteins gene variants in premature newborn infants with severe respiratory distress syndrome. *Journal of Perinatology*, **2018**, 38, 337-344	3.1	16
87	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
86	The prevalence of autosomal dominant polycystic kidney disease (ADPKD): A meta-analysis of European literature and prevalence evaluation in the Italian province of Modena suggest that ADPKD is a rare and underdiagnosed condition. *PLoS ONE*, **2018**, 13, e0190430	3.7	36
85	Next-generation sequencing approach for the diagnosis of human diseases: open challenges and new opportunities. *Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine*, **2018**, 29, 4-14	2.4	52
84	Integration of multigene panels for the diagnosis of hereditary retinal disorders using Next Generation Sequencing and bioinformatics approaches. *Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine*, **2018**, 29, 15-25	2.4	7
83	The role of clinical and neuroimaging features in the diagnosis of CADASIL. *Journal of Neurology*, **2018**, 265, 2934-2943	5.5	15
82	Structural and functional brain signatures of C9orf72 in motor neuron disease. *Neurobiology of Aging*, **2017**, 57, 206-219	5.6	35
81	mutations in Italian patients with amyotrophic lateral sclerosis: genetic and functional characterisation. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, 869-875	5.5	26
80	Combined early treatment in hemiplegic attacks related to CACNA1A encephalopathy with brain oedema: Blocking the cascade?. *Cephalalgia*, **2017**, 37, 1202-1206	6.1	13
79	X-linked Parkinsonism with Intellectual Disability caused by novel mutations and somatic mosaicism in RAB39B gene. *Parkinsonism and Related Disorders*, **2017**, 44, 142-146	3.6	18
78	Deciphering Variability of PKD1 and PKD2 in an Italian Cohort of 643 Patients with Autosomal Dominant Polycystic Kidney Disease (ADPKD). *Scientific Reports*, **2016**, 6, 30850	4.9	17
77	Clinical Pregenetic Screening for Stroke Monogenic Diseases: Results From Lombardia GENS Registry. *Stroke*, **2016**, 47, 1702-9	6.7	27
76	ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. *Neurobiology of Aging*, **2016**, 39, 218.e5-8	5.6	3
75	Recommendations for the implementation of BRCA testing in the care and treatment pathways of ovarian cancer patients. *Future Oncology*, **2016**, 12, 2071-5	3.6	18

74	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. *Neurobiology of Aging*, **2016**, 43, 180.e1-5	5.6	32
73	Possible role of fructosamine 3-kinase genotyping for the management of diabetic patients. *Clinical Chemistry and Laboratory Medicine*, **2015**, 53, 1315-20	5.9	10
72	A disorder of surfactant metabolism without identified genetic mutations. *Italian Journal of Pediatrics*, **2015**, 41, 93	3.2	3
71	A novel heat shock protein 27 homozygous mutation: widening of the continuum between MND/dHMN/CMT2. *Journal of the Peripheral Nervous System*, **2015**, 20, 419-21	4.7	9
70	New trend in non-invasive prenatal diagnosis. *Clinica Chimica Acta*, **2015**, 451, 9-13	6.2	12
69	Exome sequencing and pathway analysis for identification of genetic variability relevant for bronchopulmonary dysplasia (BPD) in preterm newborns: A pilot study. *Clinica Chimica Acta*, **2015**, 451, 39-45	6.2	38
68	Null ABCA3 in humans: large homozygous ABCA3 deletion, correlation to clinical-pathological findings. *Pediatric Pulmonology*, **2014**, 49, E116-20	3.5	8
67	The E1015K variant in the synprint region of the CaV2.1 channel alters channel function and is associated with different migraine phenotypes. *Journal of Biological Chemistry*, **2013**, 288, 33873-33883	5.4	7
66	Unusual early recurrence of granular dystrophy after deep anterior lamellar keratoplasty: case report. *Arquivos Brasileiros De Oftalmologia*, **2013**, 76, 126-8	1.1	4
65	Genetic predisposing factors to bronchopulmonary dysplasia: preliminary data from a multicentre study. *Journal of Maternal-Fetal and Neonatal Medicine*, **2012**, 25 Suppl 4, 127-30	2	10
64	Recurrent G41S mutation in Cu/Zn superoxide dismutase gene (SOD1) causing familial amyotrophic lateral sclerosis in a large Polish family. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 132-6		3
63	A child with severe pneumomediastinum and ABCA3 gene mutation: a puzzling connection. *Archivos De Bronconeumologia*, **2012**, 48, 139-40	0.7	4
62	Call for participation in the neurogenetics consortium within the Human Variome Project. *Neurogenetics*, **2011**, 12, 169-73	3	4
61	Cystogenic potential of CD133+ progenitor cells of human polycystic kidneys. *Journal of Pathology*, **2011**, 225, 129-41	9.4	7
60	Fatal respiratory failure in a full-term newborn with two ABCA3 gene mutations: a case report. *Journal of Perinatology*, **2011**, 31, 70-2	3.1	10
59	G41S SOD1 mutation: A common ancestor for six ALS Italian families with an aggressive phenotype. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2010**, 11, 210-5		10
58	CACNA1A gene non-synonymous single nucleotide polymorphisms and common migraine in Italy: a case-control association study with a micro-array technology. *Clinical Chemistry and Laboratory Medicine*, **2009**, 47, 783-5	5.9	5
57	Capturing all disease-causing mutations for clinical and research use: toward an effortless system for the Human Variome Project. *Genetics in Medicine*, **2009**, 11, 843-9	8.1	37

56	A new mutation in the MEN1 gene. *Cancer Genetics and Cytogenetics*, **2009**, 192, 99-101		1
55	Newly characterised 5Sand 3Sregions of CACNA1A gene harbour mutations associated with Familial Hemiplegic Migraine and Episodic Ataxia. *Journal of the Neurological Sciences*, **2009**, 276, 31-7	3.2	13
54	Genomic typing for patient-specific human leukocyte antigen-alleles is an efficient tool for relapse detection of high-risk hematopoietic malignancies after stem cell transplantation from alternative donors. *Leukemia*, **2008**, 22, 2119-22	10.7	9
53	Evaluation of human gene variant detection in amplicon pools by the GS-FLX parallel Pyrosequencer. *BMC Genomics*, **2008**, 9, 464	4.5	18
52	A novel MEN1 gene mutation. *Cancer Genetics and Cytogenetics*, **2008**, 180, 165-7		3
51	Twins with severe recurrent chest infections. *Thorax*, **2008**, 63, 1082, 1090	7.3	
50	Recommendations for locus-specific databases and their curation. *Human Mutation*, **2008**, 29, 2-5	4.7	52
49	Familial clustering of unexplained transient respiratory distress in 12 newborns from three unrelated families suggests an autosomal-recessive inheritance. *Scientific World Journal, The*, **2007**, 7, 1611-6	2.2	4
48	A structured simple form for ordering genetic tests is needed to ensure coupling of clinical detail (phenotype) with DNA variants (genotype) to ensure utility in publication and databases. *Human Mutation*, **2007**, 28, 931-2	4.7	8
47	Unexplained neonatal respiratory distress due to congenital surfactant deficiency. *Journal of Pediatrics*, **2007**, 150, 649-53, 653.e1	3.6	68
46	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. *Neurology*, **2007**, 69, 1285-92	6.5	99
45	A common mutation in the surfactant protein C gene associated with lung disease. *Journal of Pediatrics*, **2005**, 146, 370-5	3.6	150
44	SOD1 mutations in amyotrophic lateral sclerosis. Results from a multicenter Italian study. *Journal of Neurology*, **2005**, 252, 782-8	5.5	75
43	Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2005**, 76, 1019-21	5.5	55
42	Long-term virological effect of highly active antiretroviral therapy on cerebrospinal fluid and relationship with genotypic resistance. *Journal of NeuroVirology*, **2004**, 10, 52-57	3.9	
41	Long-term virological effect of highly active antiretroviral therapy on cerebrospinal fluid and relationship with genotypic resistance. *Journal of NeuroVirology*, **2004**, 10, 52-57	3.9	
40	Long-term virological effect of highly active antiretroviral therapy on cerebrospinal fluid and relationship with genotypic resistance. *Journal of NeuroVirology*, **2004**, 10 Suppl 1, 52-7	3.9	18
39	Early visual function impairment in CADASIL. *Neurology*, **2003**, 60, 2008-10	6.5	30

38	Voltage-operated calcium channel heterogeneity in pancreatic beta cells: physiopathological implications. *Journal of Bioenergetics and Biomembranes*, **2003**, 35, 687-96	3.7	27
37	Detection of Pneumocystis carinii and characterization of mutations associated with sulfa resistance in bronchoalveolar lavage samples from human immunodeficiency virus-infected subjects. *Journal of Clinical Microbiology*, **2003**, 41, 2709-12	9.7	17
36	Asymptomatic cores and paracrystalline mitochondrial inclusions in CADASIL. *Neurology*, **2002**, 59, 617-20	6.5	25
35	Direct detection of Helicobacter pylori mutations associated with macrolide resistance in gastric biopsy material taken from human immunodeficiency virus-infected subjects. *Journal of Clinical Microbiology*, **2002**, 40, 2234-7	9.7	11
34	Effect of genotypic resistance on the virological response to highly active antiretroviral therapy in cerebrospinal fluid. *AIDS Research and Human Retroviruses*, **2001**, 17, 377-83	1.6	37
33	Very high prevalence of right-to-left shunt on transcranial Doppler in an Italian family with cerebral autosomal dominant angiopathy with subcortical infarcts and leukoencephalopathy. *European Neurology*, **2001**, 46, 198-201	2.1	26
32	Amplification refractory mutation system analysis of point mutations by capillary electrophoresis. *Methods in Molecular Biology*, **2001**, 163, 95-108	1.4	2
31	Efficacy of low-dose intermittent subcutaneous interleukin (IL)--2 in antiviral drug--experienced human immunodeficiency virus--infected persons with detectable virus load: a controlled study of 3 il-2 regimens with antiviral drug therapy. *Journal of Infectious Diseases*, **2001**, 183, 1476-84	7	43
30	Familial hemiplegic migraine: a ion channel disorder. *Brain Research Bulletin*, **2001**, 56, 239-41	3.9	24
29	Three new familial hemiplegic migraine mutants affect P/Q-type Ca(2+) channel kinetics. *Journal of Biological Chemistry*, **2000**, 275, 9239-43	5.4	109
28	Migraine with aura and white matter abnormalities: Notch3 mutation. *Neurology*, **2000**, 54, 1869-71	6.5	32
27	Visual electrophysiological responses in subjects with cerebral autosomal arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). *Clinical Neurophysiology*, **2000**, 111, 1582-8	4.3	23
26	Detection of rifampin resistance in Mycobacterium tuberculosis by double gradient-denaturing gradient gel electrophoresis. *Antimicrobial Agents and Chemotherapy*, **1999**, 43, 2550-4	5.9	23
25	Genetic heterogeneity in Italian families with familial hemiplegic migraine. *Neurology*, **1999**, 53, 26-33	6.5	187
24	A new CACNA1A gene mutation in acetazolamide-responsive familial hemiplegic migraine and ataxia. *Neurology*, **1999**, 53, 38-43	6.5	163
23	Comparison of clinical-radiological and molecular findings in hypochondroplasia. *American Journal of Medical Genetics Part A*, **1998**, 75, 109-12		35
22	Optimized detection of DNA point mutations by double gradient denaturing gradient gel electrophoresis. *Clinical Chemistry and Laboratory Medicine*, **1998**, 36, 959-61	5.9	4
21	Unusual clinical features and early brain MRI lesions in a family with cerebral autosomal dominant arteriopathy. *Neurology*, **1997**, 48, 1200-3	6.5	19

20	A new polymorphism in exon 14 of the dystrophin gene detected by RT-PCR and DGGE. *Molecular and Cellular Probes*, **1997**, 11, 161-2	3.3	
19	Rapid detection of 21-hydroxylase deficiency mutations by allele-specific in vitro amplification and capillary zone electrophoresis. *Clinical Chemistry*, **1997**, 43, 2121-2127	5.5	11
18	Clinicopathological and genetic studies of two further Italian families with cerebral autosomal dominant arteriopathy. *Acta Neuropathologica*, **1996**, 92, 115-22	14.3	35
17	Point mutations in Italian patients with classic, non-classic, and cryptic forms of steroid 21-hydroxylase deficiency. *Human Genetics*, **1996**, 98, 662-5	6.3	54
16	Molecular diagnosis of genetic diseases. *Clinical Biochemistry*, **1996**, 29, 201-8	3.5	3
15	A role for N-myristoylation in protein targeting: NADH-cytochrome b5 reductase requires myristic acid for association with outer mitochondrial but not ER membranes. *Journal of Cell Biology*, **1996**, 135, 1501-13	7.3	85
14	Different approaches of molecular scanning of point mutations in genetic diseases. *Pure and Applied Chemistry*, **1996**, 68, 1913-1918	2.1	4
13	HLA-DQ screening for risk assessment of insulin dependent diabetes in northern Italy. *Acta Diabetologica*, **1995**, 32, 137-42	3.9	3
12	A nonsense mutation (Gln-673-Term) in exon 17 of the human dystrophin gene detected by heteroduplex analysis. *Human Genetics*, **1995**, 96, 343-4	6.3	1
11	Failure to detect Glut4-Ile383 and IR-Gln1152 variants in NIDDM (non-insulin dependent diabetes mellitus) and control subjects in an Italian population. *Human Genetics*, **1995**, 95, 115-6	6.3	
10	CAG triplet analysis in families with androgen insensitivity syndrome by capillary electrophoresis in polymer networks. *Journal of Chromatography A*, **1995**, 706, 463-8	4.5	15
9	Mutational analysis of muscle and brain specific promoter regions of dystrophin gene in DMD/BMD Italian patients by denaturing gradient gel electrophoresis (DGGE). *Molecular and Cellular Probes*, **1995**, 9, 441-6	3.3	4
8	Amplification of 18 dystrophin gene exons in DMD/BMD patients: simultaneous resolution by capillary electrophoresis in sieving liquid polymers. *BioTechniques*, **1995**, 19, 254-8, 260-3	2.5	23
7	Capillary zone electrophoresis in polymer networks of polymerase chain reaction-amplified oligonucleotides: the case of congenital adrenal hyperplasia. *Biomedical Applications*, **1994**, 657, 201-5		17
6	Substitution of Leu for Pro-193 in the insulin receptor in a patient with a genetic form of severe insulin resistance. *Human Molecular Genetics*, **1993**, 2, 1437-41	5.6	19
5	Molecular characterization of 21-hydroxylase deficiency in 70 Italian families. *Human Heredity*, **1993**, 43, 190-6	1.1	15
4	A single mRNA, transcribed from an alternative, erythroid-specific, promoter, codes for two non-myristylated forms of NADH-cytochrome b5 reductase. *Journal of Cell Biology*, **1992**, 117, 975-86	7.3	41
3	A genetic linkage study of schizophrenia to chromosome 5 markers in a northern Italian population. *Biological Psychiatry*, **1992**, 31, 720-8	7.9	12

2	Sister chromatid exchanges in first-trimester chorionic villi after in vivo and in vitro exposure to diagnostic ultrasound. *Prenatal Diagnosis*, **1990**, 10, 141-8	3.2	10
1	Two transcripts encode rat cytochrome b5 reductase. *Proceedings of the National Academy of Sciences of the United States of America*, **1988**, 85, 7246-50	11.5	40