

Paola Carrera

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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	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018 , 97, 1268-1283.e6	13.9	296
108	Genetic heterogeneity in Italian families with familial hemiplegic migraine. <i>Neurology</i> , 1999 , 53, 26-33	6.5	187
107	A new CACNA1A gene mutation in acetazolamide-responsive familial hemiplegic migraine and ataxia. <i>Neurology</i> , 1999 , 53, 38-43	6.5	163
106	A common mutation in the surfactant protein C gene associated with lung disease. <i>Journal of Pediatrics</i> , 2005 , 146, 370-5	3.6	150
105	Three new familial hemiplegic migraine mutants affect P/Q-type Ca(2+) channel kinetics. <i>Journal of Biological Chemistry</i> , 2000 , 275, 9239-43	5.4	109
104	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. <i>Neurology</i> , 2007 , 69, 1285-92	6.5	99
103	A role for N-myristoylation in protein targeting: NADH-cytochrome b5 reductase requires myristic acid for association with outer mitochondrial but not ER membranes. <i>Journal of Cell Biology</i> , 1996 , 135, 1501-13	7.3	85
102	SOD1 mutations in amyotrophic lateral sclerosis. Results from a multicenter Italian study. <i>Journal of Neurology</i> , 2005 , 252, 782-8	5.5	75
101	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019 , 85, 470-481	9.4	72
100	Unexplained neonatal respiratory distress due to congenital surfactant deficiency. <i>Journal of Pediatrics</i> , 2007 , 150, 649-53, 653.e1	3.6	68
99	Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2005 , 76, 1019-21	5.5	55
98	Point mutations in Italian patients with classic, non-classic, and cryptic forms of steroid 21-hydroxylase deficiency. <i>Human Genetics</i> , 1996 , 98, 662-5	6.3	54
97	Recommendations for locus-specific databases and their curation. <i>Human Mutation</i> , 2008 , 29, 2-5	4.7	52
96	Next-generation sequencing approach for the diagnosis of human diseases: open challenges and new opportunities. <i>Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine</i> , 2018 , 29, 4-14	2.4	52
95	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. <i>Journal of Infectious Diseases</i> , 2001 , 183, 1476-84	7	43
94	A single mRNA, transcribed from an alternative, erythroid-specific, promoter, codes for two non-myristylated forms of NADH-cytochrome b5 reductase. <i>Journal of Cell Biology</i> , 1992 , 117, 975-86	7.3	41
93	Recommendations for the implementation of BRCA testing in ovarian cancer patients and their relatives. <i>Critical Reviews in Oncology/Hematology</i> , 2019 , 140, 67-72	7	40

92	Two transcripts encode rat cytochrome b5 reductase. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1988 , 85, 7246-50	11.5	40
91	Exome sequencing and pathway analysis for identification of genetic variability relevant for bronchopulmonary dysplasia (BPD) in preterm newborns: A pilot study. <i>Clinica Chimica Acta</i> , 2015 , 451, 39-45	6.2	38
90	Capturing all disease-causing mutations for clinical and research use: toward an effortless system for the Human Variome Project. <i>Genetics in Medicine</i> , 2009 , 11, 843-9	8.1	37
89	Effect of genotypic resistance on the virological response to highly active antiretroviral therapy in cerebrospinal fluid. <i>AIDS Research and Human Retroviruses</i> , 2001 , 17, 377-83	1.6	37
88	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. <i>PLoS ONE</i> , 2018 , 13, e0190430	3.7	36
87	Structural and functional brain signatures of C9orf72 in motor neuron disease. <i>Neurobiology of Aging</i> , 2017 , 57, 206-219	5.6	35
86	Comparison of clinical-radiological and molecular findings in hypochondroplasia. <i>American Journal of Medical Genetics Part A</i> , 1998 , 75, 109-12		35
85	Clinicopathological and genetic studies of two further Italian families with cerebral autosomal dominant arteriopathy. <i>Acta Neuropathologica</i> , 1996 , 92, 115-22	14.3	35
84	Migraine with aura and white matter abnormalities: Notch3 mutation. <i>Neurology</i> , 2000 , 54, 1869-71	6.5	32
83	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. <i>Neurobiology of Aging</i> , 2016 , 43, 180.e1-55.6		32
82	Early visual function impairment in CADASIL. <i>Neurology</i> , 2003 , 60, 2008-10	6.5	30
81	Clinical Pregenetic Screening for Stroke Monogenic Diseases: Results From Lombardia GENS Registry. <i>Stroke</i> , 2016 , 47, 1702-9	6.7	27
80	Voltage-operated calcium channel heterogeneity in pancreatic beta cells: physiopathological implications. <i>Journal of Bioenergetics and Biomembranes</i> , 2003 , 35, 687-96	3.7	27
79	mutations in Italian patients with amyotrophic lateral sclerosis: genetic and functional characterisation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017 , 88, 869-875	5.5	26
78	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. <i>European Neurology</i> , 2001 , 46, 198-201	2.1	26
77	Asymptomatic cores and paracrystalline mitochondrial inclusions in CADASIL. <i>Neurology</i> , 2002 , 59, 617-26.5		25
76	ATXN1 intermediate-length polyglutamine expansions are associated with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2018 , 64, 157.e1-157.e5	5.6	24
75	Familial hemiplegic migraine: a ion channel disorder. <i>Brain Research Bulletin</i> , 2001 , 56, 239-41	3.9	24

74	ADCY10 frameshift variant leading to severe recessive asthenozoospermia and segregating with absorptive hypercalciuria. <i>Human Reproduction</i> , 2019 , 34, 1155-1164	5.7	23
73	Visual electrophysiological responses in subjects with cerebral autosomal arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). <i>Clinical Neurophysiology</i> , 2000 , 111, 1582-8	4.3	23
72	Detection of rifampin resistance in Mycobacterium tuberculosis by double gradient-denaturing gradient gel electrophoresis. <i>Antimicrobial Agents and Chemotherapy</i> , 1999 , 43, 2550-4	5.9	23
71	Amplification of 18 dystrophin gene exons in DMD/BMD patients: simultaneous resolution by capillary electrophoresis in sieving liquid polymers. <i>BioTechniques</i> , 1995 , 19, 254-8, 260-3	2.5	23
70	Expanding the spectrum of genes responsible for hereditary motor neuropathies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019 , 90, 1171-1179	5.5	19
69	Unusual clinical features and early brain MRI lesions in a family with cerebral autosomal dominant arteriopathy. <i>Neurology</i> , 1997 , 48, 1200-3	6.5	19
68	Substitution of Leu for Pro-193 in the insulin receptor in a patient with a genetic form of severe insulin resistance. <i>Human Molecular Genetics</i> , 1993 , 2, 1437-41	5.6	19
67	X-linked Parkinsonism with Intellectual Disability caused by novel mutations and somatic mosaicism in RAB39B gene. <i>Parkinsonism and Related Disorders</i> , 2017 , 44, 142-146	3.6	18
66	Evaluation of human gene variant detection in amplicon pools by the GS-FLX parallel Pyrosequencer. <i>BMC Genomics</i> , 2008 , 9, 464	4.5	18
65	Long-term virological effect of highly active antiretroviral therapy on cerebrospinal fluid and relationship with genotypic resistance. <i>Journal of NeuroVirology</i> , 2004 , 10 Suppl 1, 52-7	3.9	18
64	Recommendations for the implementation of BRCA testing in the care and treatment pathways of ovarian cancer patients. <i>Future Oncology</i> , 2016 , 12, 2071-5	3.6	18
63	Deciphering Variability of PKD1 and PKD2 in an Italian Cohort of 643 Patients with Autosomal Dominant Polycystic Kidney Disease (ADPKD). <i>Scientific Reports</i> , 2016 , 6, 30850	4.9	17
62	Detection of Pneumocystis carinii and characterization of mutations associated with sulfa resistance in bronchoalveolar lavage samples from human immunodeficiency virus-infected subjects. <i>Journal of Clinical Microbiology</i> , 2003 , 41, 2709-12	9.7	17
61	Capillary zone electrophoresis in polymer networks of polymerase chain reaction-amplified oligonucleotides: the case of congenital adrenal hyperplasia. <i>Biomedical Applications</i> , 1994 , 657, 201-5		17
60	Efficacy of lentivirus-mediated gene therapy in an Omenn syndrome recombination-activating gene 2 mouse model is not hindered by inflammation and immune dysregulation. <i>Journal of Allergy and Clinical Immunology</i> , 2018 , 142, 928-941.e8	11.5	16
59	Surfactant proteins gene variants in premature newborn infants with severe respiratory distress syndrome. <i>Journal of Perinatology</i> , 2018 , 38, 337-344	3.1	16
58	CAG triplet analysis in families with androgen insensitivity syndrome by capillary electrophoresis in polymer networks. <i>Journal of Chromatography A</i> , 1995 , 706, 463-8	4.5	15
57	Molecular characterization of 21-hydroxylase deficiency in 70 Italian families. <i>Human Heredity</i> , 1993 , 43, 190-6	1.1	15

56	Serum phosphorylated neurofilament heavy-chain levels reflect phenotypic heterogeneity and are an independent predictor of survival in motor neuron disease. <i>Journal of Neurology</i> , 2020 , 267, 2272-2280	5.5	15
55	The role of clinical and neuroimaging features in the diagnosis of CADASIL. <i>Journal of Neurology</i> , 2018 , 265, 2934-2943	5.5	15
54	Combined early treatment in hemiplegic attacks related to CACNA1A encephalopathy with brain oedema: Blocking the cascade?. <i>Cephalalgia</i> , 2017 , 37, 1202-1206	6.1	13
53	Newly characterised 5S and 3S regions of CACNA1A gene harbour mutations associated with Familial Hemiplegic Migraine and Episodic Ataxia. <i>Journal of the Neurological Sciences</i> , 2009 , 276, 31-7	3.2	13
52	New trend in non-invasive prenatal diagnosis. <i>Clinica Chimica Acta</i> , 2015 , 451, 9-13	6.2	12
51	A genetic linkage study of schizophrenia to chromosome 5 markers in a northern Italian population. <i>Biological Psychiatry</i> , 1992 , 31, 720-8	7.9	12
50	Survival prediction models in motor neuron disease. <i>European Journal of Neurology</i> , 2019 , 26, 1143-1152		11
49	Rapid detection of 21-hydroxylase deficiency mutations by allele-specific in vitro amplification and capillary zone electrophoresis. <i>Clinical Chemistry</i> , 1997 , 43, 2121-2127	5.5	11
48	Direct detection of Helicobacter pylori mutations associated with macrolide resistance in gastric biopsy material taken from human immunodeficiency virus-infected subjects. <i>Journal of Clinical Microbiology</i> , 2002 , 40, 2234-7	9.7	11
47	Possible role of fructosamine 3-kinase genotyping for the management of diabetic patients. <i>Clinical Chemistry and Laboratory Medicine</i> , 2015 , 53, 1315-20	5.9	10
46	Genetic predisposing factors to bronchopulmonary dysplasia: preliminary data from a multicentre study. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2012 , 25 Suppl 4, 127-30	2	10
45	G41S SOD1 mutation: A common ancestor for six ALS Italian families with an aggressive phenotype. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010 , 11, 210-5		10
44	Fatal respiratory failure in a full-term newborn with two ABCA3 gene mutations: a case report. <i>Journal of Perinatology</i> , 2011 , 31, 70-2	3.1	10
43	Sister chromatid exchanges in first-trimester chorionic villi after in vivo and in vitro exposure to diagnostic ultrasound. <i>Prenatal Diagnosis</i> , 1990 , 10, 141-8	3.2	10
42	A novel heat shock protein 27 homozygous mutation: widening of the continuum between MND/dHMN/CMT2. <i>Journal of the Peripheral Nervous System</i> , 2015 , 20, 419-21	4.7	9
41	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. <i>Leukemia</i> , 2008 , 22, 2119-22	10.7	9
40	Null ABCA3 in humans: large homozygous ABCA3 deletion, correlation to clinical-pathological findings. <i>Pediatric Pulmonology</i> , 2014 , 49, E116-20	3.5	8
39	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. <i>Human Mutation</i> , 2007 , 28, 931-2	4.7	8

38	The E1015K variant in the synprint region of the CaV2.1 channel alters channel function and is associated with different migraine phenotypes. <i>Journal of Biological Chemistry</i> , 2013 , 288, 33873-33883	5.4	7
37	Cystogenic potential of CD133+ progenitor cells of human polycystic kidneys. <i>Journal of Pathology</i> , 2011 , 225, 129-41	9.4	7
36	Integration of multigene panels for the diagnosis of hereditary retinal disorders using Next Generation Sequencing and bioinformatics approaches. <i>Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine</i> , 2018 , 29, 15-25	2.4	7
35	Germinal BRCA1-2 pathogenic variants (gBRCA1-2pv) and pancreatic cancer: epidemiology of an Italian patient cohort. <i>ESMO Open</i> , 2021 , 6, 100032	6	7
34	Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020 , 91, 1001-1003	5.5	6
33	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. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	5
32	CACNA1A gene non-synonymous single nucleotide polymorphisms and common migraine in Italy: a case-control association study with a micro-array technology. <i>Clinical Chemistry and Laboratory Medicine</i> , 2009 , 47, 783-5	5.9	5
31	A child with severe pneumomediastinum and ABCA3 gene mutation: a puzzling connection. <i>Archivos De Bronconeumologia</i> , 2012 , 48, 139-40	0.7	4
30	Unusual early recurrence of granular dystrophy after deep anterior lamellar keratoplasty: case report. <i>Arquivos Brasileiros De Oftalmologia</i> , 2013 , 76, 126-8	1.1	4
29	Call for participation in the neurogenetics consortium within the Human Variome Project. <i>Neurogenetics</i> , 2011 , 12, 169-73	3	4
28	Familial clustering of unexplained transient respiratory distress in 12 newborns from three unrelated families suggests an autosomal-recessive inheritance. <i>Scientific World Journal, The</i> , 2007 , 7, 1611-6	2.2	4
27	Optimized detection of DNA point mutations by double gradient denaturing gradient gel electrophoresis. <i>Clinical Chemistry and Laboratory Medicine</i> , 1998 , 36, 959-61	5.9	4
26	Mutational analysis of muscle and brain specific promoter regions of dystrophin gene in DMD/BMD Italian patients by denaturing gradient gel electrophoresis (DGGE). <i>Molecular and Cellular Probes</i> , 1995 , 9, 441-6	3.3	4
25	Different approaches of molecular scanning of point mutations in genetic diseases. <i>Pure and Applied Chemistry</i> , 1996 , 68, 1913-1918	2.1	4
24	ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. <i>Neurobiology of Aging</i> , 2016 , 39, 218.e5-8	5.6	3
23	Molecular Assay for Ovarian Cancer Patients: A Survey through Italian Departments of Oncology and Molecular and Genomic Diagnostic Laboratories. <i>Diagnostics</i> , 2019 , 9,	3.8	3
22	A disorder of surfactant metabolism without identified genetic mutations. <i>Italian Journal of Pediatrics</i> , 2015 , 41, 93	3.2	3
21	Recurrent G41S mutation in Cu/Zn superoxide dismutase gene (SOD1) causing familial amyotrophic lateral sclerosis in a large Polish family. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012 , 13, 132-6		3

20	A novel MEN1 gene mutation. <i>Cancer Genetics and Cytogenetics</i> , 2008 , 180, 165-7		3
19	HLA-DQ screening for risk assessment of insulin dependent diabetes in northern Italy. <i>Acta Diabetologica</i> , 1995 , 32, 137-42	3.9	3
18	Molecular diagnosis of genetic diseases. <i>Clinical Biochemistry</i> , 1996 , 29, 201-8	3.5	3
17	Structural MRI Signatures in Genetic Presentations of the Frontotemporal Dementia/Motor Neuron Disease Spectrum. <i>Neurology</i> , 2021 , 97, e1594-e1607	6.5	3
16	Amplification refractory mutation system analysis of point mutations by capillary electrophoresis. <i>Methods in Molecular Biology</i> , 2001 , 163, 95-108	1.4	2
15	MSH6 gene pathogenic variant identified in familial pancreatic cancer in the absence of colon cancer. <i>European Journal of Gastroenterology and Hepatology</i> , 2020 , 32, 345-349	2.2	2
14	Generation of iCells from iPSC of a MODY8 Patient with a Novel Mutation in the Carboxyl Ester Lipase (CEL) Gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, e2322-e2333	5.6	2
13	Current scenario of the genetic testing for rare neurological disorders exploiting next generation sequencing. <i>Neural Regeneration Research</i> , 2021 , 16, 475-481	4.5	2
12	Concurrence of NMOSD and ALS in a patient with hexanucleotide repeat expansions of C9orf72. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019 , 20, 449-452	3.6	1
11	A new mutation in the MEN1 gene. <i>Cancer Genetics and Cytogenetics</i> , 2009 , 192, 99-101		1
10	A nonsense mutation (Gln-673-Term) in exon 17 of the human dystrophin gene detected by heteroduplex analysis. <i>Human Genetics</i> , 1995 , 96, 343-4	6.3	1
9	Variants in a Cohort of Italian Patients With Amyotrophic Lateral Sclerosis.. <i>Frontiers in Neuroscience</i> , 2022 , 16, 833051	5.1	1
8	Implementation of preventive and predictive BRCA testing in patients with breast, ovarian, pancreatic, and prostate cancer: a position paper of Italian Scientific Societies. <i>ESMO Open</i> , 2022 , 7, 100459	6	1
7	An Open-Source Tool for Managing Time-Evolving Variant Annotation. <i>Lecture Notes in Computer Science</i> , 2019 , 1-8	0.9	
6	A new polymorphism in exon 14 of the dystrophin gene detected by RT-PCR and DGGE. <i>Molecular and Cellular Probes</i> , 1997 , 11, 161-2	3.3	
5	Twins with severe recurrent chest infections. <i>Thorax</i> , 2008 , 63, 1082, 1090	7.3	
4	Long-term virological effect of highly active antiretroviral therapy on cerebrospinal fluid and relationship with genotypic resistance. <i>Journal of NeuroVirology</i> , 2004 , 10, 52-57	3.9	
3	Long-term virological effect of highly active antiretroviral therapy on cerebrospinal fluid and relationship with genotypic resistance. <i>Journal of NeuroVirology</i> , 2004 , 10, 52-57	3.9	

- 2 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
- 1 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