

# Rachele Cagliani

## List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

126 papers	3,355 citations	31 h-index	53 g-index
138 ext. papers	4,114 ext. citations	6.3 avg, IF	5.38 L-index

#	Paper	IF	Citations
126	Molecular Evolution of Human Coronavirus Genomes. <i>Trends in Microbiology</i> , <b>2017</b> , 25, 35-48	12.4	405
125	Parasites represent a major selective force for interleukin genes and shape the genetic predisposition to autoimmune conditions. <i>Journal of Experimental Medicine</i> , <b>2009</b> , 206, 1395-408	16.6	189
124	Evolutionary insights into host-pathogen interactions from mammalian sequence data. <i>Nature Reviews Genetics</i> , <b>2015</b> , 16, 224-36	30.1	139
123	Clinical, molecular, and protein correlations in a large sample of genetically diagnosed Italian limb girdle muscular dystrophy patients. <i>Human Mutation</i> , <b>2008</b> , 29, 258-66	4.7	130
122	Widespread balancing selection and pathogen-driven selection at blood group antigen genes. <i>Genome Research</i> , <b>2009</b> , 19, 199-212	9.7	122
121	Silencer elements as possible inhibitors of pseudoexon splicing. <i>Nucleic Acids Research</i> , <b>2004</b> , 32, 1783-91	10.1	102
120	Computational Inference of Selection Underlying the Evolution of the Novel Coronavirus, Severe Acute Respiratory Syndrome Coronavirus 2. <i>Journal of Virology</i> , <b>2020</b> , 94,	6.6	89
119	A common polymorphism in TLR3 confers natural resistance to HIV-1 infection. <i>Journal of Immunology</i> , <b>2012</b> , 188, 818-23	5.3	87
118	Response to Wilson et al.. <i>American Journal of Human Genetics</i> , <b>2010</b> , 86, 493-495	11	78
117	Genetic diversity at endoplasmic reticulum aminopeptidases is maintained by balancing selection and is associated with natural resistance to HIV-1 infection. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 4705-14	5.6	67
116	Analysis of intronic conserved elements indicates that functional complexity might represent a major source of negative selection on non-coding sequences. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 2533-46	5.6	63
115	The signature of long-standing balancing selection at the human defensin beta-1 promoter. <i>Genome Biology</i> , <b>2008</b> , 9, R143	18.3	53
114	Coding potential and sequence conservation of SARS-CoV-2 and related animal viruses. <i>Infection, Genetics and Evolution</i> , <b>2020</b> , 83, 104353	4.5	51
113	Genome-wide identification of susceptibility alleles for viral infections through a population genetics approach. <i>PLoS Genetics</i> , <b>2010</b> , 6, e1000849	6	50
112	Mutation finding in patients with dysferlin deficiency and role of the dysferlin interacting proteins annexin A1 and A2 in muscular dystrophies. <i>Human Mutation</i> , <b>2005</b> , 26, 283	4.7	50
111	Extensive Positive Selection Drives the Evolution of Nonstructural Proteins in Lineage C Betacoronaviruses. <i>Journal of Virology</i> , <b>2016</b> , 90, 3627-39	6.6	47
110	The landscape of human genes involved in the immune response to parasitic worms. <i>BMC Evolutionary Biology</i> , <b>2010</b> , 10, 264	3	46

109	Long-term balancing selection maintains trans-specific polymorphisms in the human TRIM5 gene. <i>Human Genetics</i> , <b>2010</b> , 128, 577-88	6.3	42
108	A functional variant in ERAP1 predisposes to multiple sclerosis. <i>PLoS ONE</i> , <b>2012</b> , 7, e29931	3.7	41
107	Nonstructural Proteins Are Preferential Positive Selection Targets in Zika Virus and Related Flaviviruses. <i>PLoS Neglected Tropical Diseases</i> , <b>2016</b> , 10, e0004978	4.8	41
106	The heptad repeat region is a major selection target in MERS-CoV and related coronaviruses. <i>Scientific Reports</i> , <b>2015</b> , 5, 14480	4.9	40
105	Population genetics of IFIH1: ancient population structure, local selection, and implications for susceptibility to type 1 diabetes. <i>Molecular Biology and Evolution</i> , <b>2010</b> , 27, 2555-66	8.3	40
104	Both selective and neutral processes drive GC content evolution in the human genome. <i>BMC Evolutionary Biology</i> , <b>2008</b> , 8, 99	3	39
103	A CAV3 microdeletion differentially affects skeletal muscle and myocardium. <i>Neurology</i> , <b>2003</b> , 61, 1513-8	6.5	39
102	The evolutionary history of genes involved in spoken and written language: beyond FOXP2. <i>Scientific Reports</i> , <b>2016</b> , 6, 22157	4.9	39
101	TLR3 Mutations in Adult Patients With Herpes Simplex Virus and Varicella-Zoster Virus Encephalitis. <i>Journal of Infectious Diseases</i> , <b>2017</b> , 215, 1430-1434	7	36
100	Origin and dispersal of Hepatitis E virus. <i>Emerging Microbes and Infections</i> , <b>2018</b> , 7, 11	18.9	34
99	Fine mapping of AHI1 as a schizophrenia susceptibility gene: from association to evolutionary evidence. <i>FASEB Journal</i> , <b>2010</b> , 24, 3066-82	0.9	32
98	Intron size in mammals: complexity comes to terms with economy. <i>Trends in Genetics</i> , <b>2007</b> , 23, 20-4	8.5	32
97	Molecular analysis of LGMD-2B and MM patients: identification of novel DYSF mutations and possible founder effect in the Italian population. <i>Neuromuscular Disorders</i> , <b>2003</b> , 13, 788-95	2.9	32
96	An evolutionary analysis of antigen processing and presentation across different timescales reveals pervasive selection. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004189	6	31
95	Pathogen-driven selection in the human genome. <i>International Journal of Evolutionary Biology</i> , <b>2013</b> , 2013, 204240		30
94	Diverse evolutionary histories for beta-adrenoreceptor genes in humans. <i>American Journal of Human Genetics</i> , <b>2009</b> , 85, 64-75	11	30
93	The dystrophin gene is alternatively spliced throughout its coding sequence. <i>FEBS Letters</i> , <b>2002</b> , 517, 163-6	3.8	30
92	A nonsense polymorphism (R392X) in TLR5 protects from obesity but predisposes to diabetes. <i>Journal of Immunology</i> , <b>2013</b> , 190, 3716-20	5.3	29

91	The role of protozoa-driven selection in shaping human genetic variability. <i>Trends in Genetics</i> , <b>2010</b> , 26, 95-9	8.5	29
90	OASes and STING: adaptive evolution in concert. <i>Genome Biology and Evolution</i> , <b>2015</b> , 7, 1016-32	3.9	27
89	Gene function and expression level influence the insertion/fixation dynamics of distinct transposon families in mammalian introns. <i>Genome Biology</i> , <b>2006</b> , 7, R120	18.3	27
88	Primary beta-sarcoglycanopathy manifesting as recurrent exercise-induced myoglobinuria. <i>Neuromuscular Disorders</i> , <b>2001</b> , 11, 389-94	2.9	27
87	A 175 million year history of T cell regulatory molecules reveals widespread selection, with adaptive evolution of disease alleles. <i>Immunity</i> , <b>2013</b> , 38, 1129-41	32.3	26
86	Fixation of conserved sequences shapes human intron size and influences transposon-insertion dynamics. <i>Trends in Genetics</i> , <b>2005</b> , 21, 484-8	8.5	24
85	RIG-I-like receptors evolved adaptively in mammals, with parallel evolution at LGP2 and RIG-I. <i>Journal of Molecular Biology</i> , <b>2014</b> , 426, 1351-65	6.5	23
84	Xotx1 maternal transcripts are vegetally localized in <i>Xenopus laevis</i> oocytes. <i>Mechanisms of Development</i> , <b>2000</b> , 90, 111-4	1.7	23
83	Genetic adaptation of the human circadian clock to day-length latitudinal variations and relevance for affective disorders. <i>Genome Biology</i> , <b>2014</b> , 15, 499	18.3	22
82	Ancient and recent selective pressures shaped genetic diversity at AIM2-like nucleic acid sensors. <i>Genome Biology and Evolution</i> , <b>2014</b> , 6, 830-45	3.9	22
81	Relevance of sequence and structure elements for deletion events in the dystrophin gene major hot-spot. <i>Human Genetics</i> , <b>2003</b> , 112, 272-88	6.3	22
80	Balancing selection is common in the extended MHC region but most alleles with opposite risk profile for autoimmune diseases are neutrally evolving. <i>BMC Evolutionary Biology</i> , <b>2011</b> , 11, 171	3	21
79	Analysis of splicing parameters in the dystrophin gene: relevance for physiological and pathogenetic splicing mechanisms. <i>Human Genetics</i> , <b>2001</b> , 109, 73-84	6.3	21
78	Crohn's disease loci are common targets of protozoa-driven selection. <i>Molecular Biology and Evolution</i> , <b>2013</b> , 30, 1077-87	8.3	19
77	ABO histo-blood group might modulate predisposition to Crohn's disease and affect disease behavior. <i>Journal of Crohn's and Colitis</i> , <b>2014</b> , 8, 489-94	1.5	19
76	New molecular findings in congenital myopathies due to selenoprotein N gene mutations. <i>Journal of the Neurological Sciences</i> , <b>2011</b> , 300, 107-13	3.2	19
75	A trans-specific polymorphism in ZC3HAV1 is maintained by long-standing balancing selection and may confer susceptibility to multiple sclerosis. <i>Molecular Biology and Evolution</i> , <b>2012</b> , 29, 1599-613	8.3	19
74	Evolutionary analysis identifies an MX2 haplotype associated with natural resistance to HIV-1 infection. <i>Molecular Biology and Evolution</i> , <b>2014</b> , 31, 2402-14	8.3	18

73	Endoplasmic reticulum aminopeptidase 2 haplotypes play a role in modulating susceptibility to HIV infection. <i>Aids</i> , <b>2013</b> , 27, 1697-706	3.5	18
72	A positively selected APOBEC3H haplotype is associated with natural resistance to HIV-1 infection. <i>Evolution; International Journal of Organic Evolution</i> , <b>2011</b> , 65, 3311-22	3.8	18
71	Identification of a new susceptibility variant for multiple sclerosis in OAS1 by population genetics analysis. <i>Human Genetics</i> , <b>2012</b> , 131, 87-97	6.3	17
70	Transcriptional activation of the non-muscle, full-length dystrophin isoforms in Duchenne muscular dystrophy skeletal muscle. <i>Journal of the Neurological Sciences</i> , <b>2001</b> , 186, 51-7	3.2	17
69	Evolutionary analysis of the contact system indicates that kininogen evolved adaptively in mammals and in human populations. <i>Molecular Biology and Evolution</i> , <b>2013</b> , 30, 1397-408	8.3	16
68	A population genetics study of the familial Mediterranean fever gene: evidence of balancing selection under an overdominance regime. <i>Genes and Immunity</i> , <b>2009</b> , 10, 678-86	4.4	16
67	An intragenic deletion/inversion event in the DMD gene determines a novel exon creation and results in a BMD phenotype. <i>Human Genetics</i> , <b>2004</b> , 115, 13-8	6.3	16
66	REST, a master regulator of neurogenesis, evolved under strong positive selection in humans and in non human primates. <i>Scientific Reports</i> , <b>2017</b> , 7, 9530	4.9	15
65	An evolutionary analysis of RAC2 identifies haplotypes associated with human autoimmune diseases. <i>Molecular Biology and Evolution</i> , <b>2011</b> , 28, 3319-29	8.3	15
64	Trans-acting factors may cause dystrophin splicing misregulation in BMD skeletal muscles. <i>FEBS Letters</i> , <b>2003</b> , 537, 30-4	3.8	15
63	Comparative analysis of vertebrate dystrophin loci indicate intron gigantism as a common feature. <i>Genome Research</i> , <b>2003</b> , 13, 764-72	9.7	15
62	Ancient Evolution of Mammarenaviruses: Adaptation via Changes in the L Protein and No Evidence for Host-Virus Codivergence. <i>Genome Biology and Evolution</i> , <b>2018</b> , 10, 863-874	3.9	14
61	Mammalian NPC1 genes may undergo positive selection and human polymorphisms associate with type 2 diabetes. <i>BMC Medicine</i> , <b>2012</b> , 10, 140	11.4	14
60	A common genetic variant in FOXP2 is associated with language-based learning (dis)abilities: Evidence from two Italian independent samples. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2017</b> , 174, 578-586	3.5	13
59	Positive Selection Drives Evolution at the Host-Filovirus Interaction Surface. <i>Molecular Biology and Evolution</i> , <b>2016</b> , 33, 2836-2847	8.3	13
58	Comparative analysis of the human dystrophin and utrophin gene structures. <i>Genetics</i> , <b>2002</b> , 160, 793-8	4	13
57	Strategy of Human Cytomegalovirus To Escape Interferon Beta-Induced APOBEC3G Editing Activity. <i>Journal of Virology</i> , <b>2018</b> , 92,	6.6	12
56	Polymorphisms in the CPB2 gene are maintained by balancing selection and result in haplotype-preferential splicing of exon 7. <i>Molecular Biology and Evolution</i> , <b>2010</b> , 27, 1945-54	8.3	12

55	Over-representation of exonic splicing enhancers in human intronless genes suggests multiple functions in mRNA processing. <i>Biochemical and Biophysical Research Communications</i> , <b>2004</b> , 322, 470-6	3.4	12
54	Antigenic variation of SARS-CoV-2 in response to immune pressure. <i>Molecular Ecology</i> , <b>2020</b> , 30, 3548	5.7	12
53	The CCR5 $\Delta$ 32 allele is not a major predisposing factor for severe H1N1pdm09 infection. <i>BMC Research Notes</i> , <b>2014</b> , 7, 504	2.3	11
52	Positive selection underlies the species-specific binding of Plasmodium falciparum RH5 to human basigin. <i>Molecular Ecology</i> , <b>2015</b> , 24, 4711-22	5.7	11
51	Natural Selection at the Brush-Border: Adaptations to Carbohydrate Diets in Humans and Other Mammals. <i>Genome Biology and Evolution</i> , <b>2015</b> , 7, 2569-84	3.9	10
50	Recent Out-of-Africa Migration of Human Herpes Simplex Viruses. <i>Molecular Biology and Evolution</i> , <b>2020</b> , 37, 1259-1271	8.3	10
49	The mammalian complement system as an epitome of host-pathogen genetic conflicts. <i>Molecular Ecology</i> , <b>2016</b> , 25, 1324-39	5.7	10
48	Susceptibility to type 2 diabetes may be modulated by haplotypes in G6PC2, a target of positive selection. <i>BMC Evolutionary Biology</i> , <b>2017</b> , 17, 43	3	9
47	Past and ongoing adaptation of human cytomegalovirus to its host. <i>PLoS Pathogens</i> , <b>2020</b> , 16, e1008476	7.6	9
46	A regulatory polymorphism in HAVCR2 modulates susceptibility to HIV-1 infection. <i>PLoS ONE</i> , <b>2014</b> , 9, e106442	3.7	9
45	Variants in SNAP25 are targets of natural selection and influence verbal performances in women. <i>Cellular and Molecular Life Sciences</i> , <b>2012</b> , 69, 1705-15	10.3	9
44	Evolutionary Analysis Provides Insight Into the Origin and Adaptation of HCV. <i>Frontiers in Microbiology</i> , <b>2018</b> , 9, 854	5.7	8
43	Albuminoid genes: evolving at the interface of dispensability and selection. <i>Genome Biology and Evolution</i> , <b>2014</b> , 6, 2983-97	3.9	8
42	A complex selection signature at the human AVPR1B gene. <i>BMC Evolutionary Biology</i> , <b>2009</b> , 9, 123	3	8
41	Recombination and Positive Selection Differentially Shaped the Diversity of Subgenera. <i>Viruses</i> , <b>2020</b> , 12,	6.2	8
40	Diverse selective regimes shape genetic diversity at ADAR genes and at their coding targets. <i>RNA Biology</i> , <b>2015</b> , 12, 149-61	4.8	7
39	Distinct selective forces and Neanderthal introgression shaped genetic diversity at genes involved in neurodevelopmental disorders. <i>Scientific Reports</i> , <b>2017</b> , 7, 6116	4.9	7
38	Evolutionary analysis of Old World arenaviruses reveals a major adaptive contribution of the viral polymerase. <i>Molecular Ecology</i> , <b>2017</b> , 26, 5173-5188	5.7	7

37	A 6-amino acid insertion/deletion polymorphism in the mucin domain of TIM-1 confers protections against HIV-1 infection. <i>Microbes and Infection</i> , <b>2017</b> , 19, 69-74	9.3	7
36	Possible European Origin of Circulating Varicella Zoster Virus Strains. <i>Journal of Infectious Diseases</i> , <b>2020</b> , 221, 1286-1294	7	6
35	Intrinsically disordered regions are abundant in simplexvirus proteomes and display signatures of positive selection. <i>Virus Evolution</i> , <b>2020</b> , 6, veaa028	3.7	6
34	The influence of DCDC2 risk genetic variants on reading: Testing main and haplotypic effects. <i>Neuropsychologia</i> , <b>2019</b> , 130, 52-58	3.2	6
33	Long-standing balancing selection in the THBS4 gene: influence on sex-specific brain expression and gray matter volumes in Alzheimer disease. <i>Human Mutation</i> , <b>2013</b> , 34, 743-53	4.7	6
32	Genetic variability in the ACE gene region surrounding the Alu I/D polymorphism is maintained by balancing selection in human populations. <i>Pharmacogenetics and Genomics</i> , <b>2010</b> , 20, 131-4	1.9	6
31	Congenital muscular dystrophy with muscle inflammation alpha dystroglycan glycosylation defect and no mutation in FKRP gene. <i>Journal of the Neurological Sciences</i> , <b>2006</b> , 243, 47-51	3.2	6
30	Evolutionary rates of mammalian telomere-stability genes correlate with karyotype features and female germline expression. <i>Nucleic Acids Research</i> , <b>2018</b> , 46, 7153-7168	20.1	5
29	Genetic variability at the TREX1 locus is not associated with natural resistance to HIV-1 infection. <i>Aids</i> , <b>2012</b> , 26, 1443-5	3.5	5
28	Mode and tempo of human hepatitis virus evolution. <i>Computational and Structural Biotechnology Journal</i> , <b>2019</b> , 17, 1384-1395	6.8	5
27	The Diversity of Mammalian Hemoproteins and Microbial Heme Scavengers Is Shaped by an Arms Race for Iron Piracy. <i>Frontiers in Immunology</i> , <b>2018</b> , 9, 2086	8.4	5
26	An evolutionary history of the selectin gene cluster in humans. <i>Heredity</i> , <b>2012</b> , 109, 117-26	3.6	4
25	A novel splice site mutation (3157+1G>T) in the dystrophin gene causing total exon skipping and DMD phenotype. <i>Human Mutation</i> , <b>2001</b> , 17, 239	4.7	4
24	Kinetochore proteins and microtubule-destabilizing factors are fast evolving in eutherian mammals. <i>Molecular Ecology</i> , <b>2021</b> , 30, 1505-1515	5.7	4
23	Calpain 3 deficiency in Quail Eater <sup>B</sup> disease. <i>Annals of Neurology</i> , <b>2004</b> , 55, 146-7	9.4	3
22	Two dystrophin proteins and transcripts in a mild dystrophinopathic patient. <i>Neuromuscular Disorders</i> , <b>2003</b> , 13, 13-6	2.9	3
21	You Will Never Walk Alone: Codispersal of JC Polyomavirus with Human Populations. <i>Molecular Biology and Evolution</i> , <b>2020</b> , 37, 442-454	8.3	3
20	Analysis of Reptarenavirus genomes indicates different selective forces acting on the S and L segments and recent expansion of common genotypes. <i>Infection, Genetics and Evolution</i> , <b>2018</b> , 64, 212-218	4.5	3



19	The substitution spectra of coronavirus genomes. <i>Briefings in Bioinformatics</i> , <b>2021</b> ,	13.4	3
18	Evolution and Genetic Diversity of Primate Cytomegaloviruses. <i>Microorganisms</i> , <b>2020</b> , 8,	4.9	2
17	Genetic Variability of Human Cytomegalovirus Clinical Isolates Correlates With Altered Expression of Natural Killer Cell-Activating Ligands and IFN- $\gamma$ <i>Frontiers in Immunology</i> , <b>2021</b> , 12, 532484	8.4	2
16	Adaptation of the endemic coronaviruses HCoV-OC43 and HCoV-229E to the human host. <i>Virus Evolution</i> , <b>2021</b> , 7, veab061	3.7	2
15	Multiple Selected Changes May Modulate the Molecular Interaction between RH5 and Primate Basigin. <i>MBio</i> , <b>2018</b> , 9,	7.8	2
14	Dating the Emergence of Human Endemic Coronaviruses. <i>Viruses</i> , <b>2022</b> , 14, 1095	6.2	2
13	Evolutionary analysis of exogenous and integrated HHV-6A/HHV-6B populations. <i>Virus Evolution</i> , <b>2020</b> , 6, veaa035	3.7	1
12	A complex evolutionary relationship between HHV-6A and HHV-6B. <i>Virus Evolution</i> , <b>2019</b> , 5, vez043	3.7	1
11	No association of IFI16 (interferon-inducible protein 16) variants with susceptibility to multiple sclerosis. <i>Journal of Neuroimmunology</i> , <b>2014</b> , 271, 49-52	3.5	1
10	Variants in the CYP7B1 gene region do not affect natural resistance to HIV-1 infection. <i>Retrovirology</i> , <b>2015</b> , 12, 80	3.6	1
9	Antigenic variation of SARS-CoV-2 in response to immune pressure		1
8	Genetic conflicts with Plasmodium parasites and functional constraints shape the evolution of erythrocyte cytoskeletal proteins. <i>Scientific Reports</i> , <b>2018</b> , 8, 14682	4.9	1
7	Muscular dystrophy: central nervous system alpha-dystroglycan glycosylation defects and brain malformation. <i>Journal of Child Neurology</i> , <b>2010</b> , 25, 312-20	2.5	0
6	Evolutionary history of type II transmembrane serine proteases involved in viral priming.. <i>Human Genetics</i> , <b>2022</b> , 1	6.3	0
5	A CAV3 microdeletion differentially affects skeletal muscle and myocardium. <i>Neurology</i> , <b>2004</b> , 63, 195; author reply 195	6.5	
4	Evolution and Origin of Human Viruses <b>2021</b> , 289-340		
3	Chromosomally integrated human herpesvirus 6 (ci-HHV-6) in autologous bone marrow transplant recipients: are we missing a reactivation or is it just mimicking?. <i>Journal of Clinical Virology</i> , <b>2021</b> , 139, 104823	14.5	
2	Retraction to: A complex evolutionary relationship between HHV-6A and HHV-6B. <i>Virus Evolution</i> , <b>2019</b> , 5, vez054	3.7	



- 1 Alternation between taxonomically divergent hosts is not the major determinant of flavivirus evolution. *Virus Evolution*, **2021**, 7, veab040 3·7