Giuseppe Novelli

List of Publications by Year in descending order

619 papers	25,062 citations	14614 66 h-index	14156 128 g-index
649 all docs	649 docs citations	649 times ranked	31292 citing authors

#	Article	IF	Citations
1	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
2	Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a European collaborative study.. Journal of Medical Genetics, 1997, 34, 798-804.	1.5	1,032
3	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	9.4	918
4	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
5	Mandibuloacral Dysplasia Is Caused by a Mutation in LMNA-Encoding Lamin A/C. American Journal of Human Genetics, 2002, 71, 426-431.	2.6	509
6	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	9.4	482
7	MicroRNA 217 Modulates Endothelial Cell Senescence via Silent Information Regulator 1. Circulation, 2009, 120, 1524-1532.	1.6	438
8	Lectin-like, oxidized low-density lipoprotein receptor-1 (LOX-1): A critical player in the development of atherosclerosis and related disorders. Cardiovascular Research, 2006, 69, 36-45.	1.8	395
9	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. Nature Genetics, 2010, 42, 996-999.	9.4	334
10	The origin of the major cystic fibrosis mutation (Î”F508) in European populations. Nature Genetics, 1994, 7, 169-175.	9.4	323
11	Laron Dwarfism and Mutations of the Growth Hormoneâ€“Receptor Gene. New England Journal of Medicine, 1989, 321, 989-995.	13.9	302
12	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	5.6	267
13	Mutations in the Hepatocyte Nuclear Factor-1Î² Gene Are Associated with Familial Hypoplastic Glomerulocystic Kidney Disease. American Journal of Human Genetics, 2001, 68, 219-224.	2.6	263
14	Variation in a Repeat Sequence Determines Whether a Common Variant of the Cystic Fibrosis Transmembrane Conductance Regulator Gene Is Pathogenic or Benign. American Journal of Human Genetics, 2004, 74, 176-179.	2.6	227
15	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
16	Localization of Friedreich ataxia phenotype with selective vitamin E deficiency to chromosome 8q by homozygosity mapping. Nature Genetics, 1993, 5, 195-200.	9.4	215
17	Altered pre-lamin A processing is a common mechanism leading to lipodystrophy. Human Molecular Genetics, 2005, 14, 1489-1502.	1.4	203
18	Prenatal Diagnosis of Myotonic Dystrophy Using Fetal DNA Obtained from Maternal Plasma. Clinical Chemistry, 2000, 46, 301-302.	1.5	201

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19	Survival Motor-Neuron Gene Transcript Analysis in Muscles from Spinal Muscular-Atrophy Patients. Biochemical and Biophysical Research Communications, 1995, 213, 342-348.	1.0	182
20	Searching for Psoriasis Susceptibility Genes in Italy: Genome Scan and Evidence for a New Locus on Chromosome 1. Journal of Investigative Dermatology, 1999, 112, 32-35.	0.3	161
21	Incidence of Type 1 and Type 2 Diabetes in Adults Aged 30-49 Years: The population-based registry in the province of Turin, Italy. Diabetes Care, 2005, 28, 2613-2619.	4.3	158
22	Transfer and Expression of Foreign Genes in Mammalian Cells. BioTechniques, 2000, 29, 314-331.	0.8	153
23	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. Nature Genetics, 2013, 45, 947-950.	9.4	151
24	Prospective Observational Study on acute Appendicitis Worldwide (POSAW). World Journal of Emergency Surgery, 2018, 13, 19.	2.1	147
25	Isolation of CF cell lines corrected at Î”F508-CFTR locus by SFHR-mediated targeting. Gene Therapy, 2002, 9, 683-685.	2.3	146
26	Classical galactosemia and mutations at the galactose-1-phosphate uridyl transferase (GALT) gene. , 1999, 13, 417-430.		145
27	Primary laminopathy fibroblasts display altered genome organization and apoptosis. Aging Cell, 2007, 6, 139-153.	3.0	140
28	Rescue of heterochromatin organization in Hutchinson-Gilford progeria by drug treatment. Cellular and Molecular Life Sciences, 2005, 62, 2669-2678.	2.4	139
29	Coding haplotype analysis supports HCR as the putative susceptibility gene for psoriasis at the MHC PSORS1 locus. Human Molecular Genetics, 2002, 11, 589-597.	1.4	131
30	<scp>HLA</scp> allele frequencies and susceptibility to <scp>COVID</scp>â€19 in a group of 99 Italian patients. Hla, 2020, 96, 610-614.	0.4	130
31	CYP4F2genetic variant (rs2108622) significantly contributes to warfarin dosing variability in the Italian population. Pharmacogenomics, 2009, 10, 261-266.	0.6	129
32	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. Human Genetics, 2002, 111, 401-404.	1.8	125
33	Correlation between cardiac involvement and CTG trinucleotide repeat length in myotonic dystrophy. Journal of the American College of Cardiology, 1995, 25, 239-245.	1.2	124
34	The search for South European cystic fibrosis mutations: Identification of two new mutations, four variants, and intronic sequences. Genomics, 1991, 10, 193-200.	1.3	117
35	22q11 deletions in isolated and syndromic patients with tetralogy of Fallot. Human Genetics, 1995, 95, 479-82.	1.8	117
36	In Vivo and In Vitro Studies Support That a New Splicing Isoform of OLR1 Gene Is Protective Against Acute Myocardial Infarction. Circulation Research, 2005, 97, 152-158.	2.0	116

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37	Characterisation of mutations in 77 patients with X-linked myotubular myopathy, including a family with a very mild phenotype. Human Genetics, 2003, 112, 135-142.	1.8	113
38	Atypical Progeroid Syndrome due to Heterozygous Missense LMNA Mutations. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4971-4983.	1.8	113
39	Alterations of nuclear envelope and chromatin organization in mandibuloacral dysplasia, a rare form of laminopathy. Physiological Genomics, 2005, 23, 150-158.	1.0	112
40	Randomized comparison of awake nonresectional versus nonawake resectional lung volume reduction surgery. Journal of Thoracic and Cardiovascular Surgery, 2012, 143, 47-54.e1.	0.4	112
41	Brain involvement in myotonic dystrophies: neuroimaging and neuropsychological comparative study in DM1 and DM2. Journal of Neurology, 2010, 257, 1246-1255.	1.8	101
42	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	4.2	100
43	Overexpression of microRNA-206 in the skeletal muscle from myotonic dystrophy type 1 patients. Journal of Translational Medicine, 2010, 8, 48.	1.8	97
44	Oxidized LDL Receptor 1 (OLR1) as a Possible Link between Obesity, Dyslipidemia and Cancer. PLoS ONE, 2011, 6, e20277.	1.1	96
45	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	1.1	96
46	Molecular Basis of Disorders of Human Galactose Metabolism: Past, Present, and Future. Molecular Genetics and Metabolism, 2000, 71, 62-65.	0.5	90
47	Association of single nucleotide polymorphisms in the oxidised LDL receptor 1 (OLR1) gene in patients with acute myocardial infarction. Journal of Medical Genetics, 2003, 40, 933-936.	1.5	90
48	Meta-Analysis Confirms the LCE3C_LCE3B Deletion as a Risk Factor for Psoriasis in Several Ethnic Groups and Finds Interaction with HLA-Cw6. Journal of Investigative Dermatology, 2011, 131, 1105-1109.	0.3	89
49	Awake Thoracoscopic Biopsy of Interstitial Lung Disease. Annals of Thoracic Surgery, 2013, 95, 445-452.	0.7	89
50	COVID-19 and Genetic Variants of Protein Involved in the SARS-CoV-2 Entry into the Host Cells. Genes, 2020, 11, 1010.	1.0	88
51	UFD1L, a Developmentally Expressed Ubiquitination Gene, is Deleted in CATCH 22 Syndrome. Human Molecular Genetics, 1997, 6, 259-265.	1.4	85
52	Atypical deletions suggest five 22q11.2 critical regions related to the DiGeorge/velo-cardio-facial syndrome. European Journal of Human Genetics, 1999, 7, 903-909.	1.4	82
53	Expression of Î”F508 CFTR in normal mouse lung after site-specific modification of CFTR sequences by SFHR. Gene Therapy, 2001, 8, 961-965.	2.3	81
54	Diaphragmatic Spinal Muscular Atrophy with Respiratory Distress Is Heterogeneous, and One Form Is Linked to Chromosome 11q13-q21. American Journal of Human Genetics, 1999, 65, 1459-1462.	2.6	80

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55	Fine Mapping of the PSORS4 Psoriasis Susceptibility Region on Chromosome 1q21. Journal of Investigative Dermatology, 2001, 116, 728-730.	0.3	80
56	Autophagic degradation of farnesylated prelamin A as a therapeutic approach to lamin-linked progeria. European Journal of Histochemistry, 2011, 55, e36.	0.6	80
57	Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. Human Genetics, 2008, 123, 557-598.	1.8	79
58	Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype. Journal of Immunology Research, 2015, 2015, 1-11.	0.9	79
59	Evidence for differential S100 gene over-expression in psoriatic patients from genetically heterogeneous pedigrees. Human Genetics, 2002, 111, 310-313.	1.8	78
60	Cell-free DNA analysis in healthy individuals by next-generation sequencing: a proof of concept and technical validation study. Cell Death and Disease, 2019, 10, 534.	2.7	78
61	Low doses of dexamethasone constantly delivered by autologous erythrocytes slow the progression of lung disease in cystic fibrosis patients. Blood Cells, Molecules, and Diseases, 2004, 33, 57-63.	0.6	76
62	Review of nutrient actions on age-related macular degeneration. Nutrition Research, 2014, 34, 95-105.	1.3	76
63	Normal myogenesis and increased apoptosis in myotonic dystrophy type-1 muscle cells. Cell Death and Differentiation, 2010, 17, 1315-1324.	5.0	74
64	Genetic variants of the human host influencing the coronavirus-associated phenotypes (SARS, MERS) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.4	74
65	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. Nature Medicine, 2022, 28, 879-882.	15.2	72
66	Epigenetic Modification in Coronary Atherosclerosis. Journal of the American College of Cardiology, 2019, 74, 1352-1365.	1.2	71
67	Common polymorphisms in MIR146a, MIR128a and MIR27a genes contribute to neuropathy susceptibility in type 2 diabetes. Acta Diabetologica, 2014, 51, 663-671.	1.2	70
68	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	1.8	69
69	Age-related clinical severity at diagnosis in 1705 patients with ulcerative colitis: a study by GISC (Italian Colon-Rectum Study Group). Digestive Diseases and Sciences, 2000, 45, 462-465.	1.1	69
70	Carnitine palmitoyl transferase-1A (CPT1A): a new tumor specific target in human breast cancer. Oncotarget, 2016, 7, 19982-19996.	0.8	69
71	Sequence-specific modification of genomic DNA by small DNA fragments. Journal of Clinical Investigation, 2003, 112, 637-641.	3.9	68
72	COVID-19 2022 update: transition of the pandemic to the endemic phase. Human Genomics, 2022, 16, .	1.4	68

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73	â€œThe Linosa Studyâ€: Epidemiological and heritability data of the metabolic syndrome in a Caucasian genetic isolate. Nutrition, Metabolism and Cardiovascular Diseases, 2009, 19, 455-461.	1.1	67
74	Nevirapine-induced hepatotoxicity and pharmacogenetics: a retrospective study in a population from Mozambique. Pharmacogenomics, 2010, 11, 23-31.	0.6	67
75	The splice variant LOXIN inhibits LOX-1 receptor function through hetero-oligomerization. Journal of Molecular and Cellular Cardiology, 2008, 44, 561-570.	0.9	66
76	The Etiology of Acute Recurrent Pancreatitis in Children. Pancreas, 2011, 40, 517-521.	0.5	65
77	Cholesterol-Lowering Drugs Inhibit Lectin-Like Oxidized Low-Density Lipoprotein-1 Receptor Function by Membrane Raft Disruption. Molecular Pharmacology, 2012, 82, 246-254.	1.0	65
78	Evidence for Interaction between Psoriasis-Susceptibility Loci on Chromosomes 6p21 and 1q21. American Journal of Human Genetics, 1999, 65, 1798-1800.	2.6	64
79	Variants in RUNX3 Contribute to Susceptibility to Psoriatic Arthritis, Exhibiting Further Common Ground With Ankylosing Spondylitis. Arthritis and Rheumatism, 2013, 65, 1224-1231.	6.7	63
80	TCF7L2 gene polymorphisms and type 2 diabetes: association with diabetic retinopathy and cardiovascular autonomic neuropathy. Acta Diabetologica, 2013, 50, 789-799.	1.2	62
81	Parental origin of chromosome 4p deletion in Wolf-Hirschhorn syndrome. American Journal of Medical Genetics Part A, 1993, 47, 921-924.	2.4	61
82	Biomarkers in COPD. Pulmonary Pharmacology and Therapeutics, 2010, 23, 493-500.	1.1	61
83	The mutation spectrum of the EDA gene in X-linked anhidrotic ectodermal dysplasia. Human Mutation, 2001, 17, 349-349.	1.1	60
84	Targeted Correction of a Defective Selectable Marker Gene in Human Epithelial Cells by Small DNA Fragments. Molecular Therapy, 2001, 3, 178-185.	3.7	60
85	MicroRNA genetic variations: association with type 2 diabetes. Acta Diabetologica, 2013, 50, 867-872.	1.2	60
86	Mandibuloacral dysplasia: A premature ageing disease with aspects of physiological ageing. Ageing Research Reviews, 2018, 42, 1-13.	5.0	60
87	Analysis of ACE2 genetic variants in 131 Italian SARS-CoV-2-positive patients. Human Genomics, 2020, 14, 29.	1.4	60
88	A Multilocus Genetic Study in a Cohort of Italian SLE Patients Confirms the Association with STAT4 Gene and Describes a New Association with HCP5 Gene. PLoS ONE, 2014, 9, e111991.	1.1	60
89	Allelic variants in theCYP2C9andVKORC1loci and interindividual variability in the anticoagulant dose effect of warfarin in Italians. Pharmacogenomics, 2007, 8, 1545-1550.	0.6	59
90	Compound Heterozygosity for Mutations in LMNA in a Patient with a Myopathic and Lipodystrophic Mandibuloacral Dysplasia Type A Phenotype. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 4467-4471.	1.8	59

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91	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
92	Shared Phenotypes Among Segmental Progeroid Syndromes Suggest Underlying Pathways of Aging. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2005, 60, 10-20.	1.7	58
93	Multilocus analysis of the fragile X syndrome. Human Genetics, 1988, 78, 201-205.	1.8	57
94	cDNA characterization and chromosomal mapping of two human homologues of the Drosophila dishevelled polarity gene. Human Molecular Genetics, 1996, 5, 953-958.	1.4	57
95	Î”F508 GENE DELETION IN CYSTIC FIBROSIS IN SOUTHERN EUROPE. Lancet, The, 1989, 334, 1404.	6.3	56
96	Biased T-cell receptor repertoires in patients with chromosome 22q11.2 deletion syndrome (DiGeorge) Tj ETQq0 0 0 rgBT /Overlock 10 T	1.1	56
97	A Novel Syndrome of Mandibular Hypoplasia, Deafness, and Progeroid Features Associated with Lipodystrophy, Undescended Testes, and Male Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2010, 95, E192-E197.	1.8	56
98	Towards the application of precision medicine in Age-Related Macular Degeneration. Progress in Retinal and Eye Research, 2018, 63, 132-146.	7.3	56
99	Association between CYP2B6 polymorphisms and Nevirapine-induced SJS/TEN: a pharmacogenetics study. European Journal of Clinical Pharmacology, 2013, 69, 1909-1916.	0.8	55
100	Transposition of the great arteries associated with deletion of chromosome 22q11. American Journal of Cardiology, 1995, 75, 95-98.	0.7	54
101	Analysis of ACE2 Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. Genes, 2020, 11, 741.	1.0	54
102	Functional Analysis and Molecular Dynamics Simulation of LOX-1 K167N Polymorphism Reveal Alteration of Receptor Activity. PLoS ONE, 2009, 4, e4648.	1.1	53
103	TRAF3IP2 gene and systemic lupus erythematosus: association with disease susceptibility and pericarditis development. Immunogenetics, 2013, 65, 703-709.	1.2	53
104	Age-Related Macular Degeneration: Insights into Inflammatory Genes. Journal of Ophthalmology, 2014, 2014, 1-9.	0.6	53
105	Application of CRISPR/Cas9 to human-induced pluripotent stem cells: from gene editing to drug discovery. Human Genomics, 2020, 14, 25.	1.4	53
106	LOX-1 and cancer: an indissoluble liaison. Cancer Gene Therapy, 2021, 28, 1088-1098.	2.2	53
107	Characterization of ANKRD11 mutations in humans and mice related to KBG syndrome. Human Genetics, 2015, 134, 181-190.	1.8	52
108	Myotonic dystrophy type 1: role of <scp>CCG</scp>, <scp>CTC</scp> and <scp>CGG</scp> interruptions within <scp>DMPK</scp> alleles in the pathogenesis and molecular diagnosis. Clinical Genetics, 2017, 92, 355-364.	1.0	52

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109	Association of the G289S single nucleotide polymorphism in the HSD17B3 gene with prostate cancer in Italian men. Prostate, 2002, 53, 65-68.	1.2	51
110	The CTG repeat expansion size correlates with the splicing defects observed in muscles from myotonic dystrophy type 1 patients. Journal of Medical Genetics, 2008, 45, 639-646.	1.5	51
111	TRAF3IP2 gene is associated with cutaneous extraintestinal manifestations in Inflammatory Bowel Disease. Journal of Crohn's and Colitis, 2013, 7, 44-52.	0.6	51
112	Analysis of the elastin gene in 60 patients with clinical diagnosis of Williams syndrome. Human Genetics, 1995, 96, 444-8.	1.8	50
113	Open abdomen and entero-atmospheric fistulae: An interim analysis from the International Register of Open Abdomen (IROA). Injury, 2019, 50, 160-166.	0.7	50
114	Full Sequencing of the FLG Gene in Italian Patients with Atopic Eczema: Evidence of New Mutations, but Lack of an Association. Journal of Investigative Dermatology, 2011, 131, 982-984.	0.3	49
115	Mapping a Dominant Form of Multinodular Goiter to Chromosome Xp22. American Journal of Human Genetics, 2000, 67, 1004-1007.	2.6	48
116	Preferential central nucleation of type 2 myofibers is an invariable feature of myotonic dystrophy type 2. Muscle and Nerve, 2008, 38, 1405-1411.	1.0	48
117	(CTG)n Triplet Mutation and Phenotype Manifestations in Myotonic Dystrophy Patients. Biochemical Medicine and Metabolic Biology, 1993, 50, 85-92.	0.7	47
118	Human UDP-Galactose 4â€² Epimerase (GALE) Gene and Identification of Five Missense Mutations in Patients with Epimerase-Deficiency Galactosemia. Molecular Genetics and Metabolism, 1998, 63, 26-30.	0.5	47
119	Epidemiology and a novel procedure for large scale analysis of CFTR rearrangements in classic and atypical CF patients: A multicentric Italian study. Journal of Cystic Fibrosis, 2008, 7, 347-351.	0.3	47
120	Co-segregation of DM2 with a recessive CLCN1 mutation in juvenile onset of myotonic dystrophy type 2. Journal of Neurology, 2012, 259, 2090-2099.	1.8	47
121	Risk Prediction for Clinical Phenotype in Myotonic Dystrophy Type 1: Data from 2,650 Patients. Genetic Testing and Molecular Biomarkers, 2007, 11, 84-90.	1.7	46
122	Drugs affecting prelamin A processing: Effects on heterochromatin organization. Experimental Cell Research, 2008, 314, 453-462.	1.2	45
123	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. Acta Neuropathologica Communications, 2015, 3, 44.	2.4	45
124	The lectin-like oxidized LDL receptor-1: a new potential molecular target in colorectal cancer. Oncotarget, 2016, 7, 14765-14780.	0.8	45
125	IROA: International Register of Open Abdomen, preliminary results. World Journal of Emergency Surgery, 2017, 12, 10.	2.1	45
126	Effect of the [CCTG]n repeat expansion on ZNF9 expression in myotonic dystrophy type II (DM2). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 329-334.	1.8	44

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127	R501X and 2282del4 Filaggrin Mutations Do Not Confer Susceptibility to Psoriasis and Atopic Dermatitis in Italian Patients. Dermatology, 2008, 216, 83-84.	0.9	44
128	Protein farnesylation and disease. Journal of Inherited Metabolic Disease, 2012, 35, 917-926.	1.7	44
129	Genetics and Treatment Response in Parkinsonâ€™s Disease: An Update on Pharmacogenetic Studies. NeuroMolecular Medicine, 2018, 20, 1-17.	1.8	43
130	Non-invasive early prenatal molecular diagnosis using retrieved transcervical trophoblast cells. Human Genetics, 1996, 97, 150-155.	1.8	42
131	One Hundred Sixteen Cases of Acute Liver Failure Treated With MARS. Transplantation Proceedings, 2005, 37, 2557-2559.	0.3	42
132	Genome-wide association study of nevirapine hypersensitivity in a sub-Saharan African HIV-infected population. Journal of Antimicrobial Chemotherapy, 2017, 72, dkw545.	1.3	42
133	Early Structural and Functional Changes in Liver of Rats Treated with a Single Dose of Valproic Acid. Hepatology, 1984, 4, 1159-1166.	3.6	41
134	Identification of Eight Novel Mutations in a Collaborative Analysis of a Part of the Second Transmembrane Domain of the CFTR Gene. Genomics, 1993, 16, 296-297.	1.3	41
135	Effects of dutasteride on the expression of genes related to androgen metabolism and related pathway in human prostate cancer cell lines. Investigational New Drugs, 2007, 25, 491-497.	1.2	41
136	Four Copies of SNCA Responsible for Autosomal Dominant Parkinsonâ€™s Disease in Two Italian Siblings. Parkinson's Disease, 2015, 2015, 1-6.	0.6	41
137	Overview of the molecular determinants contributing to the expression of Psoriasis and Psoriatic Arthritis phenotypes. Journal of Cellular and Molecular Medicine, 2020, 24, 13554-13563.	1.6	41
138	Critical Involvement of the ATM-Dependent DNA Damage Response in the Apoptotic Demise of HIV-1-Elicited Syncytia. PLoS ONE, 2008, 3, e2458.	1.1	41
139	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. Nature Immunology, 2022, 23, 159-164.	7.0	41
140	Association of dopamine D4 receptor (DRD4) exon III repeat polymorphism with temperament in 3-year-old infants. Neurogenetics, 2003, 4, 207-212.	0.7	40
141	Biochemical characterization of two GALK1 mutations in patients with galactokinase deficiency. Human Mutation, 2004, 23, 396-396.	1.1	40
142	Valproic Acid Induces Neuroendocrine Differentiation and UGT2B7 Up-Regulation in Human Prostate Carcinoma Cell Line. Drug Metabolism and Disposition, 2007, 35, 968-972.	1.7	40
143	Meiotic drive at the myotonic dystrophy locus.. Journal of Medical Genetics, 1994, 31, 980-980.	1.5	39
144	In vitrocorrection of cystic fibrosis epithelial cell lines by small fragment homologous replacement (SFHR) technique. BMC Medical Genetics, 2002, 3, 8.	2.1	39

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145	Gene Expression Analysis in Myotonic Dystrophy: Indications for a Common Molecular Pathogenic Pathway in DM1 and DM2. Gene Expression, 2006, 13, 339-351.	0.5	39
146	Typing of ARMS2 and CFH in Age-Related Macular Degeneration. JAMA Ophthalmology, 2009, 127, 1368.	2.6	39
147	Lamin A precursor induces barrier-to-autointegration factor nuclear localization. Cell Cycle, 2010, 9, 2600-2610.	1.3	39
148	COVID-19 one year into the pandemic: from genetics and genomics to therapy, vaccination, and policy. Human Genomics, 2021, 15, 27.	1.4	39
149	Somatic and gonadal mosaicism in Hutchinson-Gilford progeria. American Journal of Medical Genetics, Part A, 2005, 135A, 66-68.	0.7	38
150	Pediatric Acute Liver Failure With Molecular Adsorbent Recirculating System Treatment. Transplantation Proceedings, 2008, 40, 1921-1924.	0.3	38
151	Identification and characterization of 5â€² CCG interruptions in complex DMPK expanded alleles. European Journal of Human Genetics, 2017, 25, 257-261.	1.4	38
152	Male Hypogonadism in Myotonic Dystrophy is Related to (Ctg)N Triplet Mutation. Journal of Endocrinological Investigation, 1994, 17, 381-383.	1.8	37
153	Genomic structure, promoter characterisation and mutational analysis of the S100A7 gene: exclusion of a candidate for familial psoriasis susceptibility. Human Genetics, 1999, 104, 130-134.	1.8	37
154	Association study of a promoter polymorphism of UFD1L gene with schizophrenia. American Journal of Medical Genetics Part A, 2001, 105, 529-533.	2.4	37
155	The R527H mutation in LMNA gene causes an increased sensitivity to ionizing radiation. Cell Cycle, 2008, 7, 2030-2037.	1.3	37
156	Rescue of murine silica-induced lung injury and fibrosis by human embryonic stem cells. European Respiratory Journal, 2012, 39, 446-457.	3.1	37
157	De Barsy Syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. American Journal of Medical Genetics, Part A, 2012, 158A, 927-931.	0.7	37
158	An Age-Standardized Prevalence Estimate and a Sex and Age Distribution of Myotonic Dystrophy Types 1 and 2 in the Rome Province, Italy. Neuroepidemiology, 2016, 46, 191-197.	1.1	37
159	MiR-423 is differentially expressed in patients with stable and unstable coronary artery disease: A pilot study. PLoS ONE, 2019, 14, e0216363.	1.1	37
160	A focus on the spread of the delta variant of SARS-CoV-2 in India. Indian Journal of Medical Research, 2021, 153, 537.	0.4	37
161	Genetic tests and genomic biomarkers: regulation, qualification and validation. Clinical Cases in Mineral and Bone Metabolism, 2008, 5, 149-54.	1.0	37
162	Mutations of UFD1L Are Not Responsible for the Majority of Cases of DiGeorge Syndrome/Velocardiofacial Syndrome without Deletions within Chromosome 22q11. American Journal of Human Genetics, 1999, 65, 247-249.	2.6	36

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163	Psoriatic Arthritis and CARD15 Gene Polymorphisms: No Evidence for Association in the Italian Population. Journal of Investigative Dermatology, 2004, 122, 1106-1107.	0.3	36
164	Polymorphisms in STAT-4, IL-10, PSORS1C1, PTPN2 and MIR146A genes are associated differently with prognostic factors in Italian patients affected by rheumatoid arthritis. Clinical and Experimental Immunology, 2016, 186, 157-163.	1.1	36
165	Expression of receptors for native and chemically modified low-density lipoproteins in brain microvessels. FEBS Letters, 1997, 401, 53-58.	1.3	35
166	The strange case of the â€˜lumperâ€™ lamin A/C gene and human premature ageing. Trends in Molecular Medicine, 2003, 9, 370-375.	3.5	35
167	Autophagy and inflammatory bowel disease: Association between variants of the autophagy-related IRGM gene and susceptibility to Crohn's disease. Digestive and Liver Disease, 2015, 47, 744-750.	0.4	35
168	Association between a MIR499A polymorphism and diabetic neuropathy in type 2 diabetes. Journal of Diabetes and Its Complications, 2018, 32, 11-17.	1.2	35
169	Mandibuloacral dysplasia type A in childhood. American Journal of Medical Genetics, Part A, 2009, 149A, 2258-2264.	0.7	34
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