Giuseppe Novelli

List of Publications by Year in descending order

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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	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
2	Human genetic and immunological determinants of critical COVID-19 pneumonia. Nature, 2022, 603, 587-598.	13.7	216
3	Mitochondrial dysfunction in mandibular hypoplasia, deafness and progeroid features with concomitant lipodystrophy (MDPL) patients. Aging, 2022, 14, 1651-1664.	1.4	3
4	RIPK4 regulates cellâ€“cell adhesion in epidermal development and homeostasis. Human Molecular Genetics, 2022, , .	1.4	1
5	Two Different Therapeutic Approaches for SARS-CoV-2 in hiPSCs-Derived Lung Organoids. Cells, 2022, 11, 1235.	1.8	21
6	Studying severe long COVID to understand post-infectious disorders beyond COVID-19. Nature Medicine, 2022, 28, 879-882.	15.2	72
7	COVID-19 and Molecular Genetics. Genes, 2022, 13, 676.	1.0	2
8	Ultrapotent and broad neutralization of SARS-CoV-2 variants by modular, tetravalent, bi-paratopic antibodies. Cell Reports, 2022, 39, 110905.	2.9	5
9	COVID-19 2022 update: transition of the pandemic to the endemic phase. Human Genomics, 2022, 16, .	1.4	68
10	Peptideâ€“Antibody Fusions Engineered by Phage Display Exhibit an Ultrapotent and Broad Neutralization of SARS-CoV-2 Variants. ACS Chemical Biology, 2022, 17, 1978-1988.	1.6	7
11	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
12	Recessive inborn errors of type I IFN immunity in children with COVID-19 pneumonia. Journal of Experimental Medicine, 2022, 219, .	4.2	59
13	Genetics and Genomics of Breast Cancer: update and translational perspectives. Seminars in Cancer Biology, 2021, 72, 27-35.	4.3	14
14	Altered expression of miR-142, miR-155, miR-499a and of their putative common target MDM2 in systemic lupus erythematosus. Epigenomics, 2021, 13, 5-13.	1.0	8
15	LOX-1 and cancer: an indissoluble liaison. Cancer Gene Therapy, 2021, 28, 1088-1098.	2.2	53
16	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
17	Functional analysis of POLD1 p.ser605del variant: the aging phenotype of MDPL syndrome is associated with an impaired DNA repair capacity. Aging, 2021, 13, 4926-4945.	1.4	10
18	Inhibition of HECT E3 ligases as potential therapy for COVID-19. Cell Death and Disease, 2021, 12, 310.	2.7	33

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19	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
20	mRNA expression analysis confirms CD44 splicing impairment in systemic lupus erythematosus patients. Lupus, 2021, 30, 1086-1093.	0.8	5
21	Case Report: An Atypical Form of Familial Partial Lipodystrophy Type 2 Due to Mutation in the Rod Domain of Lamin A/C. Frontiers in Endocrinology, 2021, 12, 675096.	1.5	3
22	Variants in MHY7 Gene Cause Arrhythmogenic Cardiomyopathy. Genes, 2021, 12, 793.	1.0	4
23	COVID-19 one year into the pandemic: from genetics and genomics to therapy, vaccination, and policy. Human Genomics, 2021, 15, 27.	1.4	39
24	Emerging Role of microRNAs and Long Non-Coding RNAs in SjÃ¶grenâ€™s Syndrome. Genes, 2021, 12, 903.	1.0	9
25	A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. Frontiers in Genetics, 2021, 12, 668094.	1.1	3
26	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.5	11
27	Two RECK Splice Variants (Long and Short) Are Differentially Expressed in Patients with Stable and Unstable Coronary Artery Disease: A Pilot Study. Genes, 2021, 12, 939.	1.0	3
28	Effects of Simulated Microgravity on Wild Type and Marfan hiPSCs-Derived Embryoid Bodies. Cellular and Molecular Bioengineering, 2021, 14, 613-626.	1.0	3
29	Cohort Analysis of 67 Charcot-Marie-Tooth Italian Patients: Identification of New Mutations and Broadening of Phenotype Expression Produced by Rare Variants. Frontiers in Genetics, 2021, 12, 682050.	1.1	4
30	From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemicâ€'Associated Pernio. Journal of Investigative Dermatology, 2021, 141, 2791-2796.	0.3	21
31	Six years as university rector changed how I do genetics. Nature, 2021, 595, 494-494.	13.7	0
32	Thromboembolism after COVID-19 vaccine in patients with preexisting thrombocytopenia. Cell Death and Disease, 2021, 12, 762.	2.7	19
33	Update on human genetic susceptibility to COVID-19: susceptibility to virus and response. Human Genomics, 2021, 15, 57.	1.4	15
34	Urine LOX-1 and Volatilome as Promising Tools towards the Early Detection of Renal Cancer. Cancers, 2021, 13, 4213.	1.7	15
35	Peptide Platform as a Powerful Tool in the Fight against COVID-19. Viruses, 2021, 13, 1667.	1.5	9
36	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

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37	Pharmacogenomics: An Update on Biologics and Small-Molecule Drugs in the Treatment of Psoriasis. Genes, 2021, 12, 1398.	1.0	25
38	COVID. A New Journal to Affirm the Role of Science between Communication and Social Responsibility. Covid, 2021, 1, 335-336.	0.7	0
39	Genetic and Epigenetic Factors of Takotsubo Syndrome: A Systematic Review. International Journal of Molecular Sciences, 2021, 22, 9875.	1.8	13
40	Tetravalent SARS-CoV-2 Neutralizing Antibodies Show Enhanced Potency and Resistance to Escape Mutations. Journal of Molecular Biology, 2021, 433, 167177.	2.0	31
41	Characterization of FMR1 Repeat Expansion and Intragenic Variants by Indirect Sequence Capture. Frontiers in Genetics, 2021, 12, 743230.	1.1	12
42	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	1.5	10
43	Will GWAS eventually allow the identification of genomic biomarkers for COVID-19 severity and mortality?. Journal of Clinical Investigation, 2021, 131, .	3.9	9
44	Clinical Features of LMNA-Related Cardiomyopathy in 18 Patients and Characterization of Two Novel Variants. Journal of Clinical Medicine, 2021, 10, 5075.	1.0	6
45	Breast cancer in West Africa: molecular analysis of BRCA genes in early-onset breast cancer patients in Burkina Faso. Human Genomics, 2021, 15, 65.	1.4	9
46	Epigenetics of Myotonic Dystrophies: A Minireview. International Journal of Molecular Sciences, 2021, 22, 12594.	1.8	8
47	Poking COVID-19: Insights on Genomic Constraints among Immune-Related Genes between Qatari and Italian Populations. Genes, 2021, 12, 1842.	1.0	1
48	Medical-health recommendations for the Serie A League. Medicina Dello Sport, 2021, 74, .	0.1	0
49	Open Abdomen in Obese Patients: Pay Attention! New Evidences from IROA, the International Register of Open Abdomen. World Journal of Surgery, 2020, 44, 53-62.	0.8	7
50	Mutation analysis of the FBN1 gene in a cohort of patients with Marfan Syndrome: A 10-year single center experience. Clinica Chimica Acta, 2020, 501, 154-164.	0.5	13
51	Circulating Long Non-Coding RNA GAS5 Is Overexpressed in Serum from Osteoporotic Patients and Is Associated with Increased Risk of Bone Fragility. International Journal of Molecular Sciences, 2020, 21, 6930.	1.8	12
52	Expression profiles of the SARS-CoV-2 host invasion genes in nasopharyngeal and oropharyngeal swabs of COVID-19 patients. Heliyon, 2020, 6, e05143.	1.4	23
53	An enormous Italian pedigree of Marfan syndrome with a novel mutation in the FBN1 gene. Clinical Case Reports (discontinued), 2020, 8, 1445-1451.	0.2	1
54	WWP1 germline variants are associated with normocephalic autism spectrum disorder. Cell Death and Disease, 2020, 11, 529.	2.7	5

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55	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
56	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
57	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
58	Open Abdomen and Fluid Instillation in the Septic Abdomen: Results from the IROA Study. World Journal of Surgery, 2020, 44, 4032-4040.	0.8	8
59	Analysis of ACE2 genetic variants in 131 Italian SARS-CoV-2-positive patients. Human Genomics, 2020, 14, 29.	1.4	60
60	Genetic variants of the human host influencing the coronavirus-associated phenotypes (SARS, MERS) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.4	74
61	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. Italian Journal of Pediatrics, 2020, 46, 130.	1.0	14
62	<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
63	Genetic variability in noncoding RNAs: involvement of miRNAs and long noncoding RNAs in osteoporosis pathogenesis. Epigenomics, 2020, 12, 2035-2049.	1.0	4
64	Neurovascular manifestations in connective tissue diseases: The case of Marfan Syndrome. Mechanisms of Ageing and Development, 2020, 191, 111346.	2.2	6
65	Mitochondrial DNA Copy Number in Peripheral Blood Is Reduced in Type 2 Diabetes Patients with Polyneuropathy and Associated with a MIR499A Gene Polymorphism. DNA and Cell Biology, 2020, 39, 1467-1472.	0.9	18
66	Expression study of candidate miRNAs and evaluation of their potential use as biomarkers of diabetic neuropathy. Epigenomics, 2020, 12, 575-585.	1.0	21
67	Genomics of COVID-19: molecular mechanisms going from susceptibility to severity of the disease. Human Genomics, 2020, 14, 22.	1.4	4
68	Carrier frequency of CFTR variants in the nonâ€Caucasian populations by genome aggregation database (gnomAD)â€based analysis. Annals of Human Genetics, 2020, 84, 463-468.	0.3	7
69	The pursuit of good microbiological conditions in domestic softeners: a new improvement. Journal of Water and Health, 2020, 18, 200-206.	1.1	1
70	Identification of Aberrantly-Expressed Long Non-Coding RNAs in Osteoblastic Cells from Osteoporotic Patients. Biomedicines, 2020, 8, 65.	1.4	15
71	Application of CRISPR/Cas9 to human-induced pluripotent stem cells: from gene editing to drug discovery. Human Genomics, 2020, 14, 25.	1.4	53
72	Analysis of ACE2 Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. Genes, 2020, 11, 741.	1.0	54

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73	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	1.1	9
74	Precision Medicine in Non-Communicable Diseases. High-Throughput, 2020, 9, 3.	4.4	9
75	European lipodystrophy registry: background and structure. Orphanet Journal of Rare Diseases, 2020, 15, 17.	1.2	21
76	Frataxin deficiency in Friedreichâ€™s ataxia is associated with reduced levels of HAX-1, a regulator of cardiomyocyte death and survival. Human Molecular Genetics, 2020, 29, 471-482.	1.4	8
77	RNAseq-Based Prioritization Revealed COL6A5, COL8A1, COL10A1 and MIR146A as Common and Differential Susceptibility Biomarkers for Psoriasis and Psoriatic Arthritis: Confirmation from Genotyping Analysis of 1417 Italian Subjects. International Journal of Molecular Sciences, 2020, 21, 2740.	1.8	12
78	COVID-19 update: the first 6 months of the pandemic. Human Genomics, 2020, 14, 48.	1.4	30
79	Identification, molecular characterization and segregation analysis of a variant pre-mutation allele in a three-generation Italian family. Acta Myologica, 2020, 39, 13-18.	1.5	3
80	Cutaneous and metabolic defects associated with nuclear abnormalities in a transgenic mouse model expressing R527H lamin A mutation causing mandibuloacral dysplasia type A (MADA) syndrome. Acta Myologica, 2020, 39, 320-335.	1.5	2
81	The differential response to anti IL-6 treatment in COVID-19: the genetic counterpart. Clinical and Experimental Rheumatology, 2020, 38, 580.	0.4	7
82	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
83	Atopic Eczema: Genetic Analysis of COL6A5, COL8A1, and COL10A1 in Mediterranean Populations. BioMed Research International, 2019, 2019, 1-7.	0.9	11
84	The variability of SMCHD1 gene in FSHD patients: evidence of new mutations. Human Molecular Genetics, 2019, 28, 3912-3920.	1.4	9
85	NGS Analysis for Molecular Diagnosis of Retinitis Pigmentosa (RP): Detection of a Novel Variant in PRPH2 Gene. Genes, 2019, 10, 792.	1.0	10
86	Epigenetic Modification in Coronary Atherosclerosis. Journal of the American College of Cardiology, 2019, 74, 1352-1365.	1.2	71
87	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. Genes, 2019, 10, 764.	1.0	20
88	miRNAs in drug response variability: potential utility as biomarkers for personalized medicine. Pharmacogenomics, 2019, 20, 1049-1059.	0.6	20
89	Targeting LOX-1 Inhibits Colorectal Cancer Metastasis in an Animal Model. Frontiers in Oncology, 2019, 9, 927.	1.3	27
90	A common polymorphism in MIR155 gene promoter region is associated with a lower risk to develop type 2 diabetes. Acta Diabetologica, 2019, 56, 717-718.	1.2	7

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91	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
92	Genetics and Autoimmunity. , 2019, , 93-104.		0
93	STAT4, TRAF3IP2, IL10, and HCP5 Polymorphisms in SjÃ¶grenâ€™s Syndrome: Association with Disease Susceptibility and Clinical Aspects. Journal of Immunology Research, 2019, 2019, 1-8.	0.9	25
94	The Interplay between miRNA-Related Variants and Age-Related Macular Degeneration: EVIDENCE of Association of MIR146A and MIR27A. International Journal of Molecular Sciences, 2019, 20, 1578.	1.8	14
95	Pro-oncogenic action of LOX-1 and its splice variant LOX-1Î”4 in breast cancer phenotypes. Cell Death and Disease, 2019, 10, 53.	2.7	24
96	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
97	Keratoderma-Deafness-Mucocutaneous Syndrome Associated with Phe142Leu in the GJB2 Gene. Acta Dermato-Venereologica, 2019, 99, 1192-1194.	0.6	1
98	Genetics and Treatment Response in Parkinsonâ€™s Disease: An Update on Pharmacogenetic Studies. NeuroMolecular Medicine, 2018, 20, 1-17.	1.8	43
99	Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 917-924.	1.8	12
100	Modelling the pathogenesis of Myotonic Dystrophy type 1 cardiac phenotype through human iPSC-derived cardiomyocytes. Journal of Molecular and Cellular Cardiology, 2018, 118, 95-109.	0.9	21
101	Assessing individual risk for AMD with genetic counseling, family history, and genetic testing. Eye, 2018, 32, 446-450.	1.1	20
102	Towards the application of precision medicine in Age-Related Macular Degeneration. Progress in Retinal and Eye Research, 2018, 63, 132-146.	7.3	56
103	Mandibuloacral dysplasia: A premature ageing disease with aspects of physiological ageing. Ageing Research Reviews, 2018, 42, 1-13.	5.0	60
104	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
105	Characterization of MDPL Fibroblasts Carrying the Recurrent p.Ser605del Mutation in POLD1 Gene. DNA and Cell Biology, 2018, 37, 1061-1067.	0.9	20
106	Vitamin D Receptor in Muscle Atrophy of Elderly Patients: A Key Element of Osteoporosis-Sarcopenia Connection. , 2018, 9, 952.		34
107	A multivariate genetic analysis confirms rs5010528 in the human leucocyte antigen-C locus as a significant contributor to Stevens-Johnson syndrome/toxic epidermal necrolysis susceptibility in a Mozambique HIV population treated with nevirapine. Journal of Antimicrobial Chemotherapy, 2018, 73, 2137-2140.	1.3	2
108	A novel in-frame deletion in ZMPSTE24 is associated with autosomal recessive acrogeria (Gottron) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.1	4

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109	Evaluation of ATG5 polymorphisms in Italian patients with systemic lupus erythematosus: contribution to disease susceptibility and clinical phenotypes. Lupus, 2018, 27, 1464-1469.	0.8	25
110	Heterozygous PLA2G6 Mutation Leads to Iron Accumulation Within Basal Ganglia and Parkinson's Disease. Frontiers in Neurology, 2018, 9, 536.	1.1	20
111	Volatile compounds emission from teratogenic human pluripotent stem cells observed during their differentiation in vivo. Scientific Reports, 2018, 8, 11056.	1.6	10
112	Biomolecular index of therapeutic efficacy in psoriasis treated with anti-TNF-Î± agents. Italian Journal of Dermatology and Venereology, 2018, 153, 316-325.	0.1	4
113	Prospective Observational Study on acute Appendicitis Worldwide (POSAW). World Journal of Emergency Surgery, 2018, 13, 19.	2.1	147
114	AFM nanoâ€mechanical study of the beating profile of hiPSCâ€derived cardiomyocytes beating bodies WT and DM1. Journal of Molecular Recognition, 2018, 31, e2725.	1.1	6
115	Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. Frontiers in Physiology, 2018, 9, 967.	1.3	3
116	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	1.4	12
117	Uncovering genetic and non-genetic biomarkers specific for exudative age-related macular degeneration: significant association of twelve variants. Oncotarget, 2018, 9, 7812-7821.	0.8	33
118	Lamins and bone disorders: current understanding and perspectives. Oncotarget, 2018, 9, 22817-22831.	0.8	19
119	Structural modeling of altered CLCN1 conformation following a novel mutation in a patient affected by autosomal dominant myotonia congenita (Thomsen disease). Archives Italiennes De Biologie, 2018, 155, 275-278.	0.1	0
120	Caregiver in carcere: avere cura di sÃ© per avere cura dell'altro. Ricerche Di Psicologia, 2018, , 423-438.	0.2	1
121	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
122	IROA: International Register of Open Abdomen, preliminary results. World Journal of Emergency Surgery, 2017, 12, 10.	2.1	45
123	OLR1andLoxinExpression in PBMCs of Women with a History of Unexplained Recurrent Miscarriage: A Pilot Study. Genetic Testing and Molecular Biomarkers, 2017, 21, 363-372.	0.3	6
124	Targeted Next Generation Sequencing in patients with Myotonia Congenita. Clinica Chimica Acta, 2017, 470, 1-7.	0.5	10
125	A preliminary analysis of volatile metabolites of human induced pluripotent stem cells along the in vitro differentiation. Scientific Reports, 2017, 7, 1621.	1.6	15
126	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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127	Pharmacogenetics of inflammatory bowel disease: a focus on Crohn's disease. Pharmacogenomics, 2017, 18, 1095-1114.	0.6	11
128	Impact of glutathione transferases genes polymorphisms in nevirapine adverse reactions: a possible role for GSTM1 in SJS/TEN susceptibility. European Journal of Clinical Pharmacology, 2017, 73, 1253-1259.	0.8	12
129	Identification of stem cells differentiation steps. , 2017, , .		0
130	Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis. Neuromuscular Disorders, 2017, 27, 163-169.	0.3	18
131	Ku70, Ku80, and sClusterin: A Cluster of Predicting Factors for Response to Neoadjuvant Chemoradiation Therapy in Patients With Locally Advanced Rectal Cancer. International Journal of Radiation Oncology Biology Physics, 2017, 97, 381-388.	0.4	21
132	Polymorphisms in MIR122, MIR196A2, and MIR124A Genes are Associated with Clinical Phenotypes in Inflammatory Bowel Diseases. Molecular Diagnosis and Therapy, 2017, 21, 107-114.	1.6	17
133	A polymorphism upstream MIR1279 gene is associated with pericarditis development in Systemic Lupus Erythematosus and contributes to definition of a genetic risk profile for this complication. Lupus, 2017, 26, 841-848.	0.8	13
134	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
135	Genotypeâ€“phenotype correlation of F484L mutation in three Italian families with Thomsen myotonia. Muscle and Nerve, 2017, 55, E24-E25.	1.0	1
136	The Monoamine Brainstem Reticular Formation as a Paradigm for Re-Defining Various Phenotypes of Parkinsonâ€™s Disease Owing Genetic and Anatomical Specificity. Frontiers in Cellular Neuroscience, 2017, 11, 102.	1.8	9
137	LOX-1 and Its Splice Variants: A New Challenge for Atherosclerosis and Cancer-Targeted Therapies. International Journal of Molecular Sciences, 2017, 18, 290.	1.8	29
138	Polymorphisms in miRNA genes and their involvement in autoimmune diseases susceptibility. Immunologic Research, 2017, 65, 811-827.	1.3	23
139	Polymorphisms in STAT4, PTPN2, PSORS1C1 and TRAF3IP2 Genes Are Associated with the Response to TNF Inhibitors in Patients with Rheumatoid Arthritis. PLoS ONE, 2017, 12, e0169956.	1.1	22
140	KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. Oncotarget, 2017, 8, 95401-95411.	0.8	12
141	GC/MS-based Analysis of Volatile Metabolic Profile Along in vitro Differentiation of Human Induced Pluripotent Stem Cells. Bio-protocol, 2017, 7, e2642.	0.2	3
142	Vacuolar Protein Sorting Genes in Parkinson's Disease: A Re-appraisal of Mutations Detection Rate and Neurobiology of Disease. Frontiers in Neuroscience, 2016, 10, 532.	1.4	15
143	SMA Human iPSC-Derived Motor Neurons Show Perturbed Differentiation and Reduced miR-335-5p Expression. International Journal of Molecular Sciences, 2016, 17, 1231.	1.8	20
144	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	1.1	96

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145	The lectin-like oxidized LDL receptor-1: a new potential molecular target in colorectal cancer. Oncotarget, 2016, 7, 14765-14780.	0.8	45
146	Two molecular assays for the rapid and inexpensive detection of GJB2 and GJB6 mutations. Electrophoresis, 2016, 37, 860-864.	1.3	2
147	The Gene Targeting Approach of Small Fragment Homologous Replacement (SFHR) Alters the Expression Patterns of DNA Repair and Cell Cycle Control Genes. Molecular Therapy - Nucleic Acids, 2016, 5, e304.	2.3	1
148	A New Splicing Mutation in the L1CAM Gene Responsible for X-Linked Hydrocephalus (HSAS). Journal of Molecular Neuroscience, 2016, 59, 376-381.	1.1	16
149	Recent advances in exploring the genetic susceptibility to diabetic neuropathy. Diabetes Research and Clinical Practice, 2016, 120, 198-208.	1.1	28
150	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
151	The human rs1050286 polymorphism alters <scp>LOX</scp>â€1 expression through modifying miRâ€24 binding. Journal of Cellular and Molecular Medicine, 2016, 20, 181-187.	1.6	19
152	Next Generation Sequencing and Linkage Analysis for the Molecular Diagnosis of a Novel Overlapping Syndrome Characterized by Hypertrophic Cardiomyopathy and Typical Electrical Instability of Brugada Syndrome. Circulation Journal, 2016, 80, 938-949.	0.7	21
153	Pharmacogenomics of multifactorial diseases: a focus on psoriatic arthritis. Pharmacogenomics, 2016, 17, 943-951.	0.6	14
154	Massive obesity and hyperphagia in posterior bilateral periventricular heterotopias: case report. BMC Medical Genetics, 2016, 17, 18.	2.1	3
155	Three-hour analysis of non-invasive foetal sex determination: application of Plexor chemistry. Human Genomics, 2016, 10, 9.	1.4	1
156	Mutation spectrum of the MTM1 gene in XLMTM patients: 10 years of experience in prenatal and postnatal diagnosis. Clinical Genetics, 2016, 89, 93-98.	1.0	10
157	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
158	Mutational analysis of mitochondrial DNA in Brugada syndrome. Cardiovascular Pathology, 2016, 25, 47-54.	0.7	13
159	Characterization of endocrine features and genotypeâ€“phenotypes correlations in blepharophimosisâ€“ptosisâ€“epicanthus inversus syndrome type 1. Journal of Endocrinological Investigation, 2016, 39, 227-233.	1.8	19
160	Carnitine palmitoyl transferase-1A (CPT1A): a new tumor specific target in human breast cancer. Oncotarget, 2016, 7, 19982-19996.	0.8	69
161	Beyond the cardiovascular risk charts. Journal of Cardiovascular Medicine, 2016, 17, 851-854.	0.6	2
162	Human induced pluripotent stem cells for monogenic disease modelling and therapy. World Journal of Stem Cells, 2016, 8, 118.	1.3	27

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163	Modulation of TGFbeta 2 levels by lamin A in U2-OS osteoblast-like cells: understanding the osteolytic process triggered by altered lamins. Oncotarget, 2015, 6, 7424-7437.	0.8	25
164	A Perturbed MicroRNA Expression Pattern Characterizes Embryonic Neural Stem Cells Derived from a Severe Mouse Model of Spinal Muscular Atrophy (SMA). International Journal of Molecular Sciences, 2015, 16, 18312-18327.	1.8	20
165	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
166	Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype. Journal of Immunology Research, 2015, 2015, 1-11.	0.9	79
167	The Genetics and the Genomics of Primary Congenital Glaucoma. BioMed Research International, 2015, 2015, 1-7.	0.9	31
168	Clinical and molecular spectra in galactosemic patients from neonatal screening in northeastern Italy: Structural and functional characterization of new variations in the galactose-1-phosphate uridyltransferase (GALT) gene. Gene, 2015, 559, 112-118.	1.0	23
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