Federica Sangiuolo

List of Publications by Year
in descending order

289	9,986	53939 47	64407 83
papers	citations	h-index	g-index

300	300	300	14920
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hsa-miR223-3p circulating level is upregulated in Friedreichâ€™s ataxia and inversely associated with HCLS1 associated protein X-1, HAX-1. Human Molecular Genetics, 2022, , .	1.4	1
2	Mitochondrial dysfunction in mandibular hypoplasia, deafness and progeroid features with concomitant lipodystrophy (MDPL) patients. Aging, 2022, 14, 1651-1664.	1.4	3
3	Two Different Therapeutic Approaches for SARS-CoV-2 in hiPSCs-Derived Lung Organoids. Cells, 2022, 11, 1235.	1.8	21
4	Machine learning phenomics (MLP) combining deep learning with time-lapse-microscopy for monitoring colorectal adenocarcinoma cells gene expression and drug-response. Scientific Reports, 2022, 12, .	1.6	10
5	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
6	Genetics and Genomics of Breast Cancer: update and translational perspectives. Seminars in Cancer Biology, 2021, 72, 27-35.	4.3	14
7	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
8	LOX-1 and cancer: an indissoluble liaison. Cancer Gene Therapy, 2021, 28, 1088-1098.	2.2	53
9	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
10	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
11	mRNA expression analysis confirms CD44 splicing impairment in systemic lupus erythematosus patients. Lupus, 2021, 30, 1086-1093.	0.8	5
12	Variants in MHY7 Gene Cause Arrhythmogenic Cardiomyopathy. Genes, 2021, 12, 793.	1.0	4
13	Emerging Role of microRNAs and Long Non-Coding RNAs in SjÃ¶grenâ€™s Syndrome. Genes, 2021, 12, 903.	1.0	9
14	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
15	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
16	Effects of Simulated Microgravity on Wild Type and Marfan hiPSCs-Derived Embryoid Bodies. Cellular and Molecular Bioengineering, 2021, 14, 613-626.	1.0	3
17	Urine LOX-1 and Volatilome as Promising Tools towards the Early Detection of Renal Cancer. Cancers, 2021, 13, 4213.	1.7	15
18	Peptide Platform as a Powerful Tool in the Fight against COVID-19. Viruses, 2021, 13, 1667.	1.5	9

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19	Pharmacogenomics: An Update on Biologics and Small-Molecule Drugs in the Treatment of Psoriasis. Genes, 2021, 12, 1398.	1.0	25
20	Genetic and Epigenetic Factors of Takotsubo Syndrome: A Systematic Review. International Journal of Molecular Sciences, 2021, 22, 9875.	1.8	13
21	Tetravalent SARS-CoV-2 Neutralizing Antibodies Show Enhanced Potency and Resistance to Escape Mutations. Journal of Molecular Biology, 2021, 433, 167177.	2.0	31
22	Characterization of FMR1 Repeat Expansion and Intragenic Variants by Indirect Sequence Capture. Frontiers in Genetics, 2021, 12, 743230.	1.1	12
23	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
24	Natriuretic peptides are neuroprotective on in vitro models of PD and promote dopaminergic differentiation of hiPSCs-derived neurons via the Wnt/Î²-catenin signaling. Cell Death Discovery, 2021, 7, 330.	2.0	7
25	Epigenetics of Myotonic Dystrophies: A Minireview. International Journal of Molecular Sciences, 2021, 22, 12594.	1.8	8
26	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
27	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
28	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
29	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
30	Specific miRNA and Gene Deregulation Characterize the Increased Angiogenic Remodeling of Thoracic Aneurysmatic Aortopathy in Marfan Syndrome. International Journal of Molecular Sciences, 2020, 21, 6886.	1.8	12
31	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. Italian Journal of Pediatrics, 2020, 46, 130.	1.0	14
32	<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
33	Genetic variability in noncoding RNAs: involvement of miRNAs and long noncoding RNAs in osteoporosis pathogenesis. Epigenomics, 2020, 12, 2035-2049.	1.0	4
34	Neurovascular manifestations in connective tissue diseases: The case of Marfan Syndrome. Mechanisms of Ageing and Development, 2020, 191, 111346.	2.2	6
35	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
36	Expression study of candidate miRNAs and evaluation of their potential use as biomarkers of diabetic neuropathy. Epigenomics, 2020, 12, 575-585.	1.0	21

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37	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
38	Molecular Genetics of Niemannâ€“Pick Type C Disease in Italy: An Update on 105 Patients and Description of 18 NPC1 Novel Variants. Journal of Clinical Medicine, 2020, 9, 679.	1.0	21
39	Identification of Aberrantly-Expressed Long Non-Coding RNAs in Osteoblastic Cells from Osteoporotic Patients. Biomedicines, 2020, 8, 65.	1.4	15
40	SARSâ€COVâ€2 and infectivity: Possible increase in infectivity associated to integrin motif expression. Journal of Medical Virology, 2020, 92, 1741-1742.	2.5	36
41	Application of CRISPR/Cas9 to human-induced pluripotent stem cells: from gene editing to drug discovery. Human Genomics, 2020, 14, 25.	1.4	53
42	Analysis of ACE2 Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. Genes, 2020, 11, 741.	1.0	54
43	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	1.1	9
44	Precision Medicine in Non-Communicable Diseases. High-Throughput, 2020, 9, 3.	4.4	9
45	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
46	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
47	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
48	The differential response to anti IL-6 treatment in COVID-19: the genetic counterpart. Clinical and Experimental Rheumatology, 2020, 38, 580.	0.4	7
49	Atopic Eczema: Genetic Analysis of COL6A5, COL8A1, and COL10A1 in Mediterranean Populations. BioMed Research International, 2019, 2019, 1-7.	0.9	11
50	The variability of SMCHD1 gene in FSHD patients: evidence of new mutations. Human Molecular Genetics, 2019, 28, 3912-3920.	1.4	9
51	NGS Analysis for Molecular Diagnosis of Retinitis Pigmentosa (RP): Detection of a Novel Variant in PRPH2 Gene. Genes, 2019, 10, 792.	1.0	10
52	Epigenetic Modification in Coronary Atherosclerosis. Journal of the American College of Cardiology, 2019, 74, 1352-1365.	1.2	71
53	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. Genes, 2019, 10, 764.	1.0	20
54	miRNAs in drug response variability: potential utility as biomarkers for personalized medicine. Pharmacogenomics, 2019, 20, 1049-1059.	0.6	20

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55	Targeting LOX-1 Inhibits Colorectal Cancer Metastasis in an Animal Model. Frontiers in Oncology, 2019, 9, 927.	1.3	27
56	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
57	Delineation of MidXq28â€duplication syndrome distal to MECP2 and proximal to RAB39B genes. Clinical Genetics, 2019, 96, 246-253.	1.0	6
58	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 316.	2.2	42
59	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
60	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
61	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
62	Periodontal condition in growing subjects with Marfan Syndrome: a case-control study. PeerJ, 2019, 7, e6606.	0.9	4
63	Genetics and Treatment Response in Parkinsonâ€™s Disease: An Update on Pharmacogenetic Studies. NeuroMolecular Medicine, 2018, 20, 1-17.	1.8	43
64	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
65	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
66	Assessing individual risk for AMD with genetic counseling, family history, and genetic testing. Eye, 2018, 32, 446-450.	1.1	20
67	Towards the application of precision medicine in Age-Related Macular Degeneration. Progress in Retinal and Eye Research, 2018, 63, 132-146.	7.3	56
68	Mandibuloacral dysplasia: A premature ageing disease with aspects of physiological ageing. Ageing Research Reviews, 2018, 42, 1-13.	5.0	60
69	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
70	Characterization of MDPL Fibroblasts Carrying the Recurrent p.Ser605del Mutation in POLD1 Gene. DNA and Cell Biology, 2018, 37, 1061-1067.	0.9	20
71	A novel in-frame deletion in ZMPSTE24 is associated with autosomal recessive acrogeria (Gottron) Tj ETQq1 1 0.784314 rgBT /Overlock	0.1	4
72	Volatile compounds emission from teratogenic human pluripotent stem cells observed during their differentiation in vivo. Scientific Reports, 2018, 8, 11056.	1.6	10

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73	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
74	Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. Frontiers in Physiology, 2018, 9, 967.	1.3	3
75	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	1.4	12
76	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
77	Lamins and bone disorders: current understanding and perspectives. Oncotarget, 2018, 9, 22817-22831.	0.8	19
78	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
79	Targeted Next Generation Sequencing in patients with Myotonia Congenita. Clinica Chimica Acta, 2017, 470, 1-7.	0.5	10
80	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
81	Pharmacogenetics of inflammatory bowel disease: a focus on Crohn's disease. Pharmacogenomics, 2017, 18, 1095-1114.	0.6	11
82	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
83	Identification of stem cells differentiation steps. , 2017, , .		0
84	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
85	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
86	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
87	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
88	Genotypeâ€“phenotype correlation of F484L mutation in three Italian families with Thomsen myotonia. Muscle and Nerve, 2017, 55, E24-E25.	1.0	1
89	Early Hippocampal i-LTP and LOX-1 Overexpression Induced by Anoxia: A Potential Role in Neurodegeneration in NPC Mouse Model. International Journal of Molecular Sciences, 2017, 18, 1442.	1.8	9
90	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

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91	Polymorphisms in miRNA genes and their involvement in autoimmune diseases susceptibility. Immunologic Research, 2017, 65, 811-827.	1.3	23
92	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
93	KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. Oncotarget, 2017, 8, 95401-95411.	0.8	12
94	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
95	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
96	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
97	The lectin-like oxidized LDL receptor-1: a new potential molecular target in colorectal cancer. Oncotarget, 2016, 7, 14765-14780.	0.8	45
98	Two molecular assays for the rapid and inexpensive detection of GJB2 and GJB6 mutations. Electrophoresis, 2016, 37, 860-864.	1.3	2
99	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
100	Recent advances in exploring the genetic susceptibility to diabetic neuropathy. Diabetes Research and Clinical Practice, 2016, 120, 198-208.	1.1	28
101	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
102	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
103	Pharmacogenomics of multifactorial diseases: a focus on psoriatic arthritis. Pharmacogenomics, 2016, 17, 943-951.	0.6	14
104	Reliable and versatile immortal muscle cell models from healthy and myotonic dystrophy type 1 primary human myoblasts. Experimental Cell Research, 2016, 342, 39-51.	1.2	32
105	Three-hour analysis of non-invasive foetal sex determination: application of Plexor chemistry. Human Genomics, 2016, 10, 9.	1.4	1
106	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3Î²,5Î±,6Î²-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to NPC1 and SMPD1 Mutations. Clinica Chimica Acta, 2016, 455, 39-45.	0.5	42
107	Rhabdoid tumor predisposition syndrome caused by SMARCB1 constitutional deletion: prenatal detection of new case of recurrence in siblings due to gonadal mosaicism. Familial Cancer, 2016, 15, 123-126.	0.9	13
108	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

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109	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
110	Human induced pluripotent stem cells for monogenic disease modelling and therapy. World Journal of Stem Cells, 2016, 8, 118.	1.3	27
111	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
112	A New Case of Prenatally Diagnosed Pentasomy X: Review of the Literature. Case Reports in Obstetrics and Gynecology, 2015, 2015, 1-5.	0.2	8
113	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
114	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
115	Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype. Journal of Immunology Research, 2015, 2015, 1-11.	0.9	79
116	The Genetics and the Genomics of Primary Congenital Glaucoma. BioMed Research International, 2015, 2015, 1-7.	0.9	31
117	Application of Next Generation Sequencing for personalized medicine for sudden cardiac death. Frontiers in Genetics, 2015, 6, 55.	1.1	17
118	Could MicroRNA polymorphisms influence warfarin dosing? A pharmacogenetics study on mir133 genes. Thrombosis Research, 2015, 136, 367-370.	0.8	20
119	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
120	A Pharmacogenetics Study in Mozambican Patients Treated with Nevirapine: Full Resequencing of TRAF3IP2 Gene Shows a Novel Association with SJS/TEN Susceptibility. International Journal of Molecular Sciences, 2015, 16, 5830-5838.	1.8	7
121	Deletion of REXO1L1 locus in a patient with malabsorption syndrome, growth retardation, and dysmorphic features: a novel recognizable microdeletion syndrome?. BMC Medical Genetics, 2015, 16, 20.	2.1	2
122	Expansion size and presence of CCG/CTC/CGG sequence interruptions in the expanded CTG array are independently associated to hypermethylation at the DMPK locus in myotonic dystrophy type 1 (DM1). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 2645-2652.	1.8	31
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	Comparative analysis between saliva and buccal swabs as source of DNA: lesson from HLA-B*57:01 testing. Pharmacogenomics, 2015, 16, 1039-1046.	0.6	16
125	Generation of Human Induced Pluripotent Stem Cells from Extraembryonic Tissues of Fetuses Affected by Monogenic Diseases. Cellular Reprogramming, 2015, 17, 275-287.	0.5	18
126	Cerebral cavernous malformations associated to meningioma: High penetrance in a novel family mutated in thePDCD10gene. Neuroradiology Journal, 2015, 28, 289-293.	0.6	11

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127	FLG (filaggrin) null mutations and sunlight exposure: Evidence of a correlation. Journal of the American Academy of Dermatology, 2015, 73, 528-529.	0.6	15
128	Stevensâ€“Johnson syndrome and toxic epidermal necrolysis: an update on pharmacogenetics studies in drug-induced severe skin reaction. Pharmacogenomics, 2015, 16, 1989-2002.	0.6	10
129	HFE gene variants and iron-induced oxygen radical generation in idiopathic pulmonary fibrosis. European Respiratory Journal, 2015, 45, 483-490.	3.1	33
130	Age-Related Macular Degeneration: Insights into Inflammatory Genes. Journal of Ophthalmology, 2014, 2014, 1-9.	0.6	53
131	Transabdominal coelocentesis as early source of fetal DNA for chromosomal and molecular diagnosis. Journal of Maternal-Fetal and Neonatal Medicine, 2014, 27, 1656-1660.	0.7	2
132	Nectin-4 Mutations Causing Ectodermal Dysplasia with Syndactyly Perturb the Rac1 Pathway and the Kinetics of Adherens Junction Formation. Journal of Investigative Dermatology, 2014, 134, 2146-2153.	0.3	33
133	3â€²-UTR OLR1/LOX-1 gene polymorphism and endothelial dysfunction: molecular and vascular data in never-treated hypertensive patients. Internal and Emergency Medicine, 2014, 9, 273-281.	1.0	4
134	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
135	Review of nutrient actions on age-related macular degeneration. Nutrition Research, 2014, 34, 95-105.	1.3	76
136	De Novo 13q13.3-21.31 deletion involving RB1 gene in a patient with hemangioendothelioma of the liver. Italian Journal of Pediatrics, 2014, 40, 5.	1.0	6
137	Human placenta-derived neurospheres are susceptible to transformation after extensive in vitro expansion. Stem Cell Research and Therapy, 2014, 5, 55.	2.4	5
138	Small Fragment Homologous Replacement (SFHR): Sequence-Specific Modification of Genomic DNA in Eukaryotic Cells by Small DNA Fragments. Methods in Molecular Biology, 2014, 1114, 85-101.	0.4	5
139	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
140	Rapamycin treatment of Mandibuloacral Dysplasia cells rescues localization of chromatin-associated proteins and cell cycle dynamics. Aging, 2014, 6, 755-769.	1.4	30
141	Resequencing of VKORC1, CYP2C9 and CYP4F2 genes in Italian patients requiring extreme low and high warfarin doses. Thrombosis Research, 2013, 132, 123-126.	0.8	9
142	TCF7L2 gene polymorphisms and type 2 diabetes: association with diabetic retinopathy and cardiovascular autonomic neuropathy. Acta Diabetologica, 2013, 50, 789-799.	1.2	62
143	MicroRNA genetic variations: association with type 2 diabetes. Acta Diabetologica, 2013, 50, 867-872.	1.2	60
144	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

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145	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
146	Putting Pleiotropy and Selection Into Context Defines a New Paradigm for Interpreting Genetic Data. Circulation: Cardiovascular Genetics, 2013, 6, 299-307.	5.1	7
147	Haplotypes in IL-8 Gene Are Associated to Age-Related Macular Degeneration: A Case-Control Study. PLoS ONE, 2013, 8, e66978.	1.1	29
148	Human embryonic stem cells recover in vivo acute lung inflammation bleomycin-induced. Sarcoidosis Vasculitis and Diffuse Lung Diseases, 2013, 30, 177-85.	0.2	1
149	The Pharmacogenomic HLA Biomarker Associated to Adverse Abacavir Reactions: Comparative Analysis of Different Genotyping Methods. Current Genomics, 2012, 13, 314-320.	0.7	33
150	Protein farnesylation and disease. Journal of Inherited Metabolic Disease, 2012, 35, 917-926.	1.7	44
151	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
152	Functional characterization and expression analysis of novel alternative splicing isoforms of Olr1 gene during mouse embryogenesis. Gene, 2012, 491, 5-12.	1.0	5
153	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	1.2	7
154	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
155	Rescue of murine silica-induced lung injury and fibrosis by human embryonic stem cells. European Respiratory Journal, 2012, 39, 446-457.	3.1	37
156	Altered chromatin organization and SUN2 localization in mandibuloacral dysplasia are rescued by drug treatment. Histochemistry and Cell Biology, 2012, 138, 643-651.	0.8	27
157	Association between OLR1 K167N SNP and Intima Media Thickness of the Common Carotid Artery in the General Population. PLoS ONE, 2012, 7, e31086.	1.1	21
158	IPLEX Administration Improves Motor Neuron Survival and Ameliorates Motor Functions in a Severe Mouse Model of Spinal Muscular Atrophy. Molecular Medicine, 2012, 18, 1076-1085.	1.9	30
159	Small Fragment Homologous Replacement: Evaluation of Factors Influencing Modification Efficiency in an Eukaryotic Assay System. PLoS ONE, 2012, 7, e30851.	1.1	6
160	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
161	Polymorphisms in ARMS2 (LOC387715) and LOXL1 Genes in the Japanese With Age-Related Macular Degeneration. American Journal of Ophthalmology, 2011, 152, 325-326.	1.7	15
162	Oxidized LDL Receptor 1 (OLR1) as a Possible Link between Obesity, Dyslipidemia and Cancer. PLoS ONE, 2011, 6, e20277.	1.1	96

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