Giuseppe Novelli

List of Publications by Year in Descending Order

600 papers	19,453 citations	60 h-index	115 g-index
648 ext. papers	22,496 ext. citations	5.4 avg, IF	6.16 L-index

#	Paper	IF	Citations
600	Human genetic and immunological determinants of critical COVID-19 pneumonia.. *Nature*, **2022**,	50.4	23
599	Mitochondrial dysfunction in mandibular hypoplasia, deafness and progeroid features with concomitant lipodystrophy (MDPL) patients.. *Aging*, **2022**, 14, 1651-1664	5.6	0
598	Studying severe long COVID to understand post-infectious disorders beyond COVID-19.. *Nature Medicine*, **2022**,	50.5	6
597	Ultrapotent and Broad Neutralization of SARS-CoV-2 Variants by Modular, Tetravalent, Bi-paratopic Antibodies. *Cell Reports*, **2022**, 110905	10.6	0
596	Epigenetics of Myotonic Dystrophies: A Minireview. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	4
595	Variants in cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy.. *Brain Communications*, **2021**, 3, fcab245	4.5	0
594	Will GWAS eventually allow the identification of genomic biomarkers for COVID-19 severity and mortality?. *Journal of Clinical Investigation*, **2021**, 131,	15.9	3
593	A global effort to dissect the human genetic basis of resistance to SARS-CoV-2 infection. *Nature Immunology*, **2021**,	19.1	7
592	Clinical Features of LMNA-Related Cardiomyopathy in 18 Patients and Characterization of Two Novel Variants. *Journal of Clinical Medicine*, **2021**, 10,	5.1	1
591	Breast cancer in West Africa: molecular analysis of BRCA genes in early-onset breast cancer patients in Burkina Faso. *Human Genomics*, **2021**, 15, 65	6.8	1
590	Inhibition of HECT E3 ligases as potential therapy for COVID-19. *Cell Death and Disease*, **2021**, 12, 310	9.8	13
589	SARS-CoV-2-related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. *Journal of Experimental Medicine*, **2021**, 218,	16.6	45
588	mRNA expression analysis confirms CD44 splicing impairment in systemic lupus erythematosus patients. *Lupus*, **2021**, 30, 1086-1093	2.6	1
587	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	5.7	0
586	Variants in Gene Cause Arrhythmogenic Cardiomyopathy. *Genes*, **2021**, 12,	4.2	2
585	COVID-19 one year into the pandemic: from genetics and genomics to therapy, vaccination, and policy. *Human Genomics*, **2021**, 15, 27	6.8	23
584	Emerging Role of microRNAs and Long Non-Coding RNAs in Sj□renIs Syndrome. *Genes*, **2021**, 12,	4.2	4

583	A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated Alleles. *Frontiers in Genetics*, **2021**, 12, 668094	4.5	1
582	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. *Neurology*, **2021**, 97, e577-e586	6.5	2
581	Effects of Simulated Microgravity on Wild Type and Marfan hiPSCs-Derived Embryoid Bodies.. *Cellular and Molecular Bioengineering*, **2021**, 14, 613-626	3.9	1
580	Genetics and Genomics of Breast Cancer: update and translational perspectives. *Seminars in Cancer Biology*, **2021**, 72, 27-35	12.7	7
579	Altered expression of miR-142, miR-155, miR-499a and of their putative common target in systemic lupus erythematosus. *Epigenomics*, **2021**, 13, 5-13	4.4	4
578	LOX-1 and cancer: an indissoluble liaison. *Cancer Gene Therapy*, **2021**, 28, 1088-1098	5.4	19
577	A focus on the spread of the delta variant of SARS-CoV-2 in India. *Indian Journal of Medical Research* , **2021**, 153, 537-541	2.9	7
576	Functional analysis of p.ser605del variant: the aging phenotype of MDPL syndrome is associated with an impaired DNA repair capacity. *Aging*, **2021**, 13, 4926-4945	5.6	4
575	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	4.5	0
574	From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemic-Associated Pernio. *Journal of Investigative Dermatology*, **2021**, 141, 2791-2796	4.3	6
573	Thromboembolism after COVID-19 vaccine in patients with preexisting thrombocytopenia. *Cell Death and Disease*, **2021**, 12, 762	9.8	3
572	Urine LOX-1 and Volatilome as Promising Tools towards the Early Detection of Renal Cancer. *Cancers*, **2021**, 13,	6.6	3
571	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
570	Pharmacogenomics: An Update on Biologics and Small-Molecule Drugs in the Treatment of Psoriasis. *Genes*, **2021**, 12,	4.2	8
569	Tetravalent SARS-CoV-2 Neutralizing Antibodies Show Enhanced Potency and Resistance to Escape Mutations. *Journal of Molecular Biology*, **2021**, 433, 167177	6.5	10
568	Characterization of Repeat Expansion and Intragenic Variants by Indirect Sequence Capture. *Frontiers in Genetics*, **2021**, 12, 743230	4.5	0
567	Neurovascular manifestations in connective tissue diseases: The case of Marfan Syndrome. *Mechanisms of Ageing and Development*, **2020**, 191, 111346	5.6	2
566	Mitochondrial DNA Copy Number in Peripheral Blood Is Reduced in Type 2 Diabetes Patients with Polyneuropathy and Associated with a Gene Polymorphism. *DNA and Cell Biology*, **2020**, 39, 1467-1472	3.6	8

565	Expression study of candidate miRNAs and evaluation of their potential use as biomarkers of diabetic neuropathy. *Epigenomics*, **2020**, 12, 575-585	4.4	11
564	Carrier frequency of CFTR variants in the non-Caucasian populations by genome aggregation database (gnomAD)-based analysis. *Annals of Human Genetics*, **2020**, 84, 463-468	2.2	2
563	The pursuit of good microbiological conditions in domestic softeners: a new improvement. *Journal of Water and Health*, **2020**, 18, 200-206	2.2	
562	Identification of Aberrantly-Expressed Long Non-Coding RNAs in Osteoblastic Cells from Osteoporotic Patients. *Biomedicines*, **2020**, 8,	4.8	11
561	Application of CRISPR/Cas9 to human-induced pluripotent stem cells: from gene editing to drug discovery. *Human Genomics*, **2020**, 14, 25	6.8	21
560	Analysis of Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. *Genes*, **2020**, 11,	4.2	36
559	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. *Frontiers in Genetics*, **2020**, 11, 605	4.5	3
558	Precision Medicine in Non-Communicable Diseases. *High-Throughput*, **2020**, 9,	4.3	5
557	European lipodystrophy registry: background and structure. *Orphanet Journal of Rare Diseases*, **2020**, 15, 17	4.2	9
556	Frataxin deficiency in FriedreichIs ataxia is associated with reduced levels of HAX-1, a regulator of cardiomyocyte death and survival. *Human Molecular Genetics*, **2020**, 29, 471-482	5.6	4
555	RNAseq-Based Prioritization Revealed , , and 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,	6.3	8
554	COVID-19 update: the first 6 months of the pandemic. *Human Genomics*, **2020**, 14, 48	6.8	15
553	Identification, molecular characterization and segregation analysis of a variant pre-mutation allele in a three-generation Italian family. *Acta Myologica*, **2020**, 39, 13-18	1.6	3
552	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	1.6	2
551	Tetravalent SARS-CoV-2 Neutralizing Antibodies Show Enhanced Potency and Resistance to Escape Mutations **2020**,		8
550	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	6.2	5
549	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,	6.3	4
548	Expression profiles of the SARS-CoV-2 host invasion genes in nasopharyngeal and oropharyngeal swabs of COVID-19 patients. *Heliyon*, **2020**, 6, e05143	3.6	8

547	An enormous Italian pedigree of Marfan syndrome with a novel mutation in the FBN1 gene. *Clinical Case Reports (discontinued)*, **2020**, 8, 1445-1451	0.7	0
546	WWP1 germline variants are associated with normocephalic autism spectrum disorder. *Cell Death and Disease*, **2020**, 11, 529	9.8	2
545	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	5.6	19
544	COVID-19 and Genetic Variants of Protein Involved in the SARS-CoV-2 Entry into the Host Cells. *Genes*, **2020**, 11,	4.2	48
543	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
542	Open Abdomen and Fluid Instillation in the Septic Abdomen: Results from the IROA Study. *World Journal of Surgery*, **2020**, 44, 4032-4040	3.3	1
541	Analysis of ACE2 genetic variants in 131 Italian SARS-CoV-2-positive patients. *Human Genomics*, **2020**, 14, 29	6.8	27
540	Genetic variants of the human host influencing the coronavirus-associated phenotypes (SARS, MERS and COVID-19): rapid systematic review and field synopsis. *Human Genomics*, **2020**, 14, 30	6.8	40
539	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. *Italian Journal of Pediatrics*, **2020**, 46, 130	3.2	3
538	HLA allele frequencies and susceptibility to COVID-19 in a group of 99 Italian patients. *Hla*, **2020**, 96, 610-614	1.9	76
537	Genetic variability in noncoding RNAs: involvement of miRNAs and long noncoding RNAs in osteoporosis pathogenesis. *Epigenomics*, **2020**, 12, 2035-2049	4.4	3
536	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	3.3	2
535	The differential response to anti IL-6 treatment in COVID-19: the genetic counterpart. *Clinical and Experimental Rheumatology*, **2020**, 38, 580	2.2	6
534	Epigenetic Modification in Coronary Atherosclerosis: JACC Review Topic of the Week. *Journal of the American College of Cardiology*, **2019**, 74, 1352-1365	15.1	27
533	Genotypic Categorization of Loeys-Dietz Syndrome Based on 24 Novel Families and Literature Data. *Genes*, **2019**, 10,	4.2	4
532	miRNAs in drug response variability: potential utility as biomarkers for personalized medicine. *Pharmacogenomics*, **2019**, 20, 1049-1059	2.6	13
531	Targeting LOX-1 Inhibits Colorectal Cancer Metastasis in an Animal Model. *Frontiers in Oncology*, **2019**, 9, 927	5.3	17
530	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	3.9	3

529	MiR-423 is differentially expressed in patients with stable and unstable coronary artery disease: A pilot study. *PLoS ONE*, **2019**, 14, e0216363	3.7	18
528	Genetics and Autoimmunity **2019**, 93-104		
527	, , , and Polymorphisms in Sjögrenʼs Syndrome: Association with Disease Susceptibility and Clinical Aspects. *Journal of Immunology Research*, **2019**, 2019, 7682827	4.5	15
526	The Interplay between miRNA-Related Variants and Age-Related Macular Degeneration: EVIDENCE of Association of and. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	11
525	Pro-oncogenic action of LOX-1 and its splice variant LOX-1Δ4 in breast cancer phenotypes. *Cell Death and Disease*, **2019**, 10, 53	9.8	16
524	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	9.8	48
523	Atopic Eczema: Genetic Analysis of , , and in Mediterranean Populations. *BioMed Research International*, **2019**, 2019, 3457898	3	9
522	The variability of SMCHD1 gene in FSHD patients: evidence of new mutations. *Human Molecular Genetics*, **2019**, 28, 3912-3920	5.6	4
521	NGS Analysis for Molecular Diagnosis of Retinitis Pigmentosa (RP): Detection of a Novel Variant in Gene. *Genes*, **2019**, 10,	4.2	8
520	Keratoderma-Deafness-Mucocutaneous Syndrome Associated with Phe142Leu in the GJB2 Gene. *Acta Dermato-Venereologica*, **2019**, 99, 1192-1194	2.2	
519	Open abdomen and entero-atmospheric fistulae: An interim analysis from the International Register of Open Abdomen (IROA). *Injury*, **2019**, 50, 160-166	2.5	25
518	Genetics and Treatment Response in Parkinsonʼs Disease: An Update on Pharmacogenetic Studies. *NeuroMolecular Medicine*, **2018**, 20, 1-17	4.6	25
517	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	6.9	7
516	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	5.8	14
515	Assessing individual risk for AMD with genetic counseling, family history, and genetic testing. *Eye*, **2018**, 32, 446-450	4.4	14
514	Heterozygous Mutation Leads to Iron Accumulation Within Basal Ganglia and Parkinsonʼs Disease. *Frontiers in Neurology*, **2018**, 9, 536	4.1	15
513	Volatile compounds emission from teratogenic human pluripotent stem cells observed during their differentiation in vivo. *Scientific Reports*, **2018**, 8, 11056	4.9	7
512	Biomolecular index of therapeutic efficacy in psoriasis treated with anti-TNF-α agents. *Italian Journal of Dermatology and Venereology*, **2018**, 153, 316-325	1.2	3

511	Prospective Observational Study on acute Appendicitis Worldwide (POSAW). *World Journal of Emergency Surgery*, **2018**, 13, 19	9.2	84
510	AFM nano-mechanical study of the beating profile of hiPSC-derived cardiomyocytes beating bodies WT and DM1. *Journal of Molecular Recognition*, **2018**, 31, e2725	2.6	5
509	Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. *Frontiers in Physiology*, **2018**, 9, 967	4.6	2
508	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. *European Journal of Human Genetics*, **2018**, 26, 1266-1271	5.3	7
507	Uncovering genetic and non-genetic biomarkers specific for exudative age-related macular degeneration: significant association of twelve variants. *Oncotarget*, **2018**, 9, 7812-7821	3.3	29
506	Lamins and bone disorders: current understanding and perspectives. *Oncotarget*, **2018**, 9, 22817-22831	3.3	14
505	Caregiver in carcere: avere cura di s per avere cura dell'altro. *Ricerche Di Psicologia*, **2018**, 423-438	0.2	0
504	Towards the application of precision medicine in Age-Related Macular Degeneration. *Progress in Retinal and Eye Research*, **2018**, 63, 132-146	20.5	44
503	Mandibuloacral dysplasia: A premature ageing disease with aspects of physiological ageing. *Ageing Research Reviews*, **2018**, 42, 1-13	12	41
502	Association between a MIR499A polymorphism and diabetic neuropathy in type 2 diabetes. *Journal of Diabetes and Its Complications*, **2018**, 32, 11-17	3.2	26
501	Characterization of MDPL Fibroblasts Carrying the Recurrent p.Ser605del Mutation in POLD1 Gene. *DNA and Cell Biology*, **2018**,	3.6	13
500	Vitamin D Receptor in Muscle Atrophy of Elderly Patients: A Key Element of Osteoporosis-Sarcopenia Connection **2018**, 9, 952-964		24
499	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	5.1	1
498	A novel in-frame deletion in ZMPSTE24 is associated with autosomal recessive acrogeria (Gottron type) in an extended consanguineous family. *Clinical Dysmorphology*, **2018**, 27, 88-90	0.9	2
497	Evaluation of ATG5 polymorphisms in Italian patients with systemic lupus erythematosus: contribution to disease susceptibility and clinical phenotypes. *Lupus*, **2018**, 27, 1464-1469	2.6	16
496	Genome-wide association study of nevirapine hypersensitivity in a sub-Saharan African HIV-infected population. *Journal of Antimicrobial Chemotherapy*, **2017**, 72, 1152-1162	5.1	33
495	IROA: International Register of Open Abdomen, preliminary results. *World Journal of Emergency Surgery*, **2017**, 12, 10	9.2	27
494	OLR1 and Loxin Expression in PBMCs of Women with a History of Unexplained Recurrent Miscarriage: A Pilot Study. *Genetic Testing and Molecular Biomarkers*, **2017**, 21, 363-372	1.6	6

493	Targeted Next Generation Sequencing in patients with Myotonia Congenita. *Clinica Chimica Acta*, **2017**, 470, 1-7	6.2	6
492	A preliminary analysis of volatile metabolites of human induced pluripotent stem cells along the in vitro differentiation. *Scientific Reports*, **2017**, 7, 1621	4.9	11
491	Myotonic dystrophy type 1: role of CCG, CTC and CGG interruptions within DMPK alleles in the pathogenesis and molecular diagnosis. *Clinical Genetics*, **2017**, 92, 355-364	4	32
490	LOX-1 and Its Splice Variants: A New Challenge for Atherosclerosis and Cancer-Targeted Therapies. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	24
489	Pharmacogenetics of inflammatory bowel disease: a focus on Crohnꞌs disease. *Pharmacogenomics*, **2017**, 18, 1095-1114	2.6	9
488	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	2.8	12
487	Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis. *Neuromuscular Disorders*, **2017**, 27, 163-169	2.9	16
486	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	4	12
485	Polymorphisms in MIR122, MIR196A2, and MIR124A Genes are Associated with Clinical Phenotypes in Inflammatory Bowel Diseases. *Molecular Diagnosis and Therapy*, **2017**, 21, 107-114	4.5	14
484	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	2.6	12
483	Identification and characterization of 5LCCG interruptions in complex DMPK expanded alleles. *European Journal of Human Genetics*, **2017**, 25, 257-261	5.3	26
482	Genotype-phenotype correlation of F484L mutation in three Italian families with Thomsen myotonia. *Muscle and Nerve*, **2017**, 55, E24-E25	3.4	1
481	The Monoamine Brainstem Reticular Formation as a Paradigm for Re-Defining Various Phenotypes of Parkinsonꞌs Disease Owing Genetic and Anatomical Specificity. *Frontiers in Cellular Neuroscience*, **2017**, 11, 102	6.1	9
480	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	3.7	18
479	and are disease-specific biomarkers for psoriatic arthritis susceptibility. *Oncotarget*, **2017**, 8, 95401-95413	5.3	8
478	GC/MS-based Analysis of Volatile Metabolic Profile Along Differentiation of Human Induced Pluripotent Stem Cells. *Bio-protocol*, **2017**, 7, e2642	0.9	2
477	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*, **2017**, 155, 118-130	1.1	
476	Polymorphisms in miRNA genes and their involvement in autoimmune diseases susceptibility. *Immunologic Research*, **2017**, 65, 811-827	4.3	16

475	Characterization of endocrine features and genotype-phenotypes correlations in blepharophimosis-ptosis-epicanthus inversus syndrome type 1. *Journal of Endocrinological Investigation*, **2016**, 39, 227-33	5.2	12
474	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, 938-49	2.9	17
473	Pharmacogenomics of multifactorial diseases: a focus on psoriatic arthritis. *Pharmacogenomics*, **2016**, 17, 943-51	2.6	9
472	Massive obesity and hyperphagia in posterior bilateral periventricular heterotopias: case report. *BMC Medical Genetics*, **2016**, 17, 18	2.1	1
471	Three-hour analysis of non-invasive foetal sex determination: application of Plexor chemistry. *Human Genomics*, **2016**, 10, 9	6.8	1
470	Mutation spectrum of the MTM1 gene in XLMTM patients: 10 years of experience in prenatal and postnatal diagnosis. *Clinical Genetics*, **2016**, 89, 93-8	4	8
469	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, 191-7	5.4	26
468	Mutational analysis of mitochondrial DNA in Brugada syndrome. *Cardiovascular Pathology*, **2016**, 25, 47-54	3.8	13
467	Carnitine palmitoyl transferase-1A (CPT1A): a new tumor specific target in human breast cancer. *Oncotarget*, **2016**, 7, 19982-96	3.3	47
466	Human induced pluripotent stem cells for monogenic disease modelling and therapy. *World Journal of Stem Cells*, **2016**, 8, 118-35	5.6	20
465	Vacuolar Protein Sorting Genes in Parkinsonls Disease: A Re-appraisal of Mutations Detection Rate and Neurobiology of Disease. *Frontiers in Neuroscience*, **2016**, 10, 532	5.1	12
464	SMA Human iPSC-Derived Motor Neurons Show Perturbed Differentiation and Reduced miR-335-5p Expression. *International Journal of Molecular Sciences*, **2016**, 17,	6.3	15
463	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. *PLoS ONE*, **2016**, 11, e0162866	3.7	66
462	The lectin-like oxidized LDL receptor-1: a new potential molecular target in colorectal cancer. *Oncotarget*, **2016**, 7, 14765-80	3.3	38
461	Two molecular assays for the rapid and inexpensive detection of GJB2 and GJB6 mutations. *Electrophoresis*, **2016**, 37, 860-4	3.6	2
460	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	10.7	1
459	A New Splicing Mutation in the L1CAM Gene Responsible for X-Linked Hydrocephalus (HSAS). *Journal of Molecular Neuroscience*, **2016**, 59, 376-81	3.3	10
458	Recent advances in exploring the genetic susceptibility to diabetic neuropathy. *Diabetes Research and Clinical Practice*, **2016**, 120, 198-208	7.4	23

457	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	6.2	31
456	The human rs1050286 polymorphism alters LOX-1 expression through modifying miR-24 binding. *Journal of Cellular and Molecular Medicine*, **2016**, 20, 181-7	5.6	16
455	Application of Next Generation Sequencing for personalized medicine for sudden cardiac death. *Frontiers in Genetics*, **2015**, 6, 55	4.5	16
454	Could MicroRNA polymorphisms influence warfarin dosing? A pharmacogenetics study on mir133 genes. *Thrombosis Research*, **2015**, 136, 367-70	8.2	16
453	Autophagy and inflammatory bowel disease: Association between variants of the autophagy-related IRGM gene and susceptibility to CrohnIs disease. *Digestive and Liver Disease*, **2015**, 47, 744-50	3.3	25
452	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, 5830-8	6.3	6
451	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
450	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, 2645-52	6.9	22
449	Complete loss of the DNAJB6 G/F domain and novel missense mutations cause distal-onset DNAJB6 myopathy. *Acta Neuropathologica Communications*, **2015**, 3, 44	7.3	28
448	Comparative analysis between saliva and buccal swabs as source of DNA: lesson from HLA-B*57:01 testing. *Pharmacogenomics*, **2015**, 16, 1039-46	2.6	12
447	Generation of Human Induced Pluripotent Stem Cells from Extraembryonic Tissues of Fetuses Affected by Monogenic Diseases. *Cellular Reprogramming*, **2015**, 17, 275-87	2.1	16
446	Cerebral cavernous malformations associated to meningioma: High penetrance in a novel family mutated in the PDCD10 gene. *Neuroradiology Journal*, **2015**, 28, 289-93	2	8
445	FLG (filaggrin) null mutations and sunlight exposure: Evidence of a correlation. *Journal of the American Academy of Dermatology*, **2015**, 73, 528-9	4.5	12
444	Stevens-Johnson syndrome and toxic epidermal necrolysis: an update on pharmacogenetics studies in drug-induced severe skin reaction. *Pharmacogenomics*, **2015**, 16, 1989-2002	2.6	10
443	Characterization of ANKRD11 mutations in humans and mice related to KBG syndrome. *Human Genetics*, **2015**, 134, 181-90	6.3	37
442	Direct PCR: a new pharmacogenetic approach for the inexpensive testing of HLA-B*57:01. *Pharmacogenomics Journal*, **2015**, 15, 196-200	3.5	18
441	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, 7424-37	3.3	24
440	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, 18312-27	6.3	15

439	Four Copies of SNCA Responsible for Autosomal Dominant Parkinsonʼs Disease in Two Italian Siblings. *Parkinsonʼs Disease*, **2015**, 2015, 546462	2.6	31
438	Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype. *Journal of Immunology Research*, **2015**, 2015, 745647	4.5	53
437	The Genetics and the Genomics of Primary Congenital Glaucoma. *BioMed Research International*, **2015**, 2015, 321291	3	28
436	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, 112-8	3.8	17
435	Common polymorphisms in MIR146a, MIR128a and MIR27a genes contribute to neuropathy susceptibility in type 2 diabetes. *Acta Diabetologica*, **2014**, 51, 663-71	3.9	59
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