Giuseppe Novelli

List of Publications by Citations

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	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
599	Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a European collaborative study. *Journal of Medical Genetics*, **1997**, 34, 798-804	5.8	854
598	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. *Nature Genetics*, **2010**, 42, 985-90	36.3	773
597	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
596	Mandibuloacral dysplasia is caused by a mutation in LMNA-encoding lamin A/C. *American Journal of Human Genetics*, **2002**, 71, 426-31	11	436
595	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. *Nature Genetics*, **2009**, 41, 211-5	36.3	405
594	MicroRNA 217 modulates endothelial cell senescence via silent information regulator 1. *Circulation*, **2009**, 120, 1524-32	16.7	387
593	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	9.9	349
592	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. *Nature Genetics*, **2010**, 42, 996-9	36.3	294
591	The origin of the major cystic fibrosis mutation (delta F508) in European populations. *Nature Genetics*, **1994**, 7, 169-75	36.3	284
590	Laron dwarfism and mutations of the growth hormone-receptor gene. *New England Journal of Medicine*, **1989**, 321, 989-95	59.2	250
589	Mutations in the hepatocyte nuclear factor-1beta gene are associated with familial hypoplastic glomerulocystic kidney disease. *American Journal of Human Genetics*, **2001**, 68, 219-24	11	227
588	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, 176-9	11	199
587	Prenatal Diagnosis of Myotonic Dystrophy Using Fetal DNA Obtained from Maternal Plasma. *Clinical Chemistry*, **2000**, 46, 301-302	5.5	191
586	Localization of Friedreich ataxia phenotype with selective vitamin E deficiency to chromosome 8q by homozygosity mapping. *Nature Genetics*, **1993**, 5, 195-200	36.3	187
585	Altered pre-lamin A processing is a common mechanism leading to lipodystrophy. *Human Molecular Genetics*, **2005**, 14, 1489-502	5.6	178
584	Survival motor neuron gene transcript analysis in muscles from spinal muscular atrophy patients. *Biochemical and Biophysical Research Communications*, **1995**, 213, 342-8	3.4	169

583	Rescue of heterochromatin organization in Hutchinson-Gilford progeria by drug treatment. *Cellular and Molecular Life Sciences*, **2005**, 62, 2669-78	10.3	129
582	Searching for psoriasis susceptibility genes in Italy: genome scan and evidence for a new locus on chromosome 1. *Journal of Investigative Dermatology*, **1999**, 112, 32-5	4.3	125
581	Isolation of CF cell lines corrected at DeltaF508-CFTR locus by SFHR-mediated targeting. *Gene Therapy*, **2002**, 9, 683-5	4	124
580	Classical galactosemia and mutations at the galactose-1-phosphate uridyl transferase (GALT) gene. *Human Mutation*, **1999**, 13, 417-30	4.7	123
579	Transfer and expression of foreign genes in mammalian cells. *BioTechniques*, **2000**, 29, 314-8, 320-2, 324 passim	2.5	121
578	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. *Nature Genetics*, **2013**, 45, 947-50	36.3	120
577	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, 2613-9	14.6	120
576	CYP4F2 genetic variant (rs2108622) significantly contributes to warfarin dosing variability in the Italian population. *Pharmacogenomics*, **2009**, 10, 261-6	2.6	119
575	Primary laminopathy fibroblasts display altered genome organization and apoptosis. *Aging Cell*, **2007**, 6, 139-53	9.9	118
574	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. *Human Genetics*, **2002**, 111, 401-4	6.3	111
573	The search for south European cystic fibrosis mutations: identification of two new mutations, four variants, and intronic sequences. *Genomics*, **1991**, 10, 193-200	4.3	111
572	Coding haplotype analysis supports HCR as the putative susceptibility gene for psoriasis at the MHC PSORS1 locus. *Human Molecular Genetics*, **2002**, 11, 589-97	5.6	106
571	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, 152-8	15.7	102
570	Alterations of nuclear envelope and chromatin organization in mandibuloacral dysplasia, a rare form of laminopathy. *Physiological Genomics*, **2005**, 23, 150-8	3.6	99
569	22q11 deletions in isolated and syndromic patients with tetralogy of Fallot. *Human Genetics*, **1995**, 95, 479-82	6.3	99
568	Correlation between cardiac involvement and CTG trinucleotide repeat length in myotonic dystrophy. *Journal of the American College of Cardiology*, **1995**, 25, 239-45	15.1	98
567	Atypical progeroid syndrome due to heterozygous missense LMNA mutations. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 4971-83	5.6	95
566	Characterisation of mutations in 77 patients with X-linked myotubular myopathy, including a family with a very mild phenotype. *Human Genetics*, **2003**, 112, 135-42	6.3	95

565	Randomized comparison of awake nonresectional versus nonawake resectional lung volume reduction surgery. *Journal of Thoracic and Cardiovascular Surgery*, **2012**, 143, 47-54, 54.e1	1.5	86
564	Overexpression of microRNA-206 in the skeletal muscle from myotonic dystrophy type 1 patients. *Journal of Translational Medicine*, **2010**, 8, 48	8.5	86
563	Prospective Observational Study on acute Appendicitis Worldwide (POSAW). *World Journal of Emergency Surgery*, **2018**, 13, 19	9.2	84
562	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, 1105-9	4.3	79
561	Oxidized LDL receptor 1 (OLR1) as a possible link between obesity, dyslipidemia and cancer. *PLoS ONE*, **2011**, 6, e20277	3.7	78
560	Brain involvement in myotonic dystrophies: neuroimaging and neuropsychological comparative study in DM1 and DM2. *Journal of Neurology*, **2010**, 257, 1246-55	5.5	78
559	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, 933-6	5.8	77
558	HLA allele frequencies and susceptibility to COVID-19 in a group of 99 Italian patients. *Hla*, **2020**, 96, 610-614	1.9	76
557	UFD1L, a developmentally expressed ubiquitination gene, is deleted in CATCH 22 syndrome. *Human Molecular Genetics*, **1997**, 6, 259-65	5.6	75
556	Molecular basis of disorders of human galactose metabolism: past, present, and future. *Molecular Genetics and Metabolism*, **2000**, 71, 62-5	3.7	75
555	Atypical deletions suggest five 22q11.2 critical regions related to the DiGeorge/velo-cardio-facial syndrome. *European Journal of Human Genetics*, **1999**, 7, 903-9	5.3	74
554	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, 1459-62	11	72
553	Expression of DeltaF508 CFTR in normal mouse lung after site-specific modification of CFTR sequences by SFHR. *Gene Therapy*, **2001**, 8, 961-5	4	71
552	Awake thoracoscopic biopsy of interstitial lung disease. *Annals of Thoracic Surgery*, **2013**, 95, 445-52	2.7	69
551	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
550	Autophagic degradation of farnesylated prelamin A as a therapeutic approach to lamin-linked progeria. *European Journal of Histochemistry*, **2011**, 55, e36	2.1	66
549	Genetic studies of African populations: an overview on disease susceptibility and response to vaccines and therapeutics. *Human Genetics*, **2008**, 123, 557-98	6.3	66
548	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	2.1	66

547	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. *PLoS ONE*, **2016**, 11, e0162866	3.7	66
546	Fine mapping of the PSORS4 psoriasis susceptibility region on chromosome 1q21. *Journal of Investigative Dermatology*, **2001**, 116, 728-30	4.3	65
545	Nevirapine-induced hepatotoxicity and pharmacogenetics: a retrospective study in a population from Mozambique. *Pharmacogenomics*, **2010**, 11, 23-31	2.6	64
544	Evidence for differential S100 gene over-expression in psoriatic patients from genetically heterogeneous pedigrees. *Human Genetics*, **2002**, 111, 310-3	6.3	62
543	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. *Human Genetics*, **1992**, 89, 653-8	6.3	62
542	Normal myogenesis and increased apoptosis in myotonic dystrophy type-1 muscle cells. *Cell Death and Differentiation*, **2010**, 17, 1315-24	12.7	61
541	The splice variant LOXIN inhibits LOX-1 receptor function through hetero-oligomerization. *Journal of Molecular and Cellular Cardiology*, **2008**, 44, 561-70	5.8	60
540	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
539	Review of nutrient actions on age-related macular degeneration. *Nutrition Research*, **2014**, 34, 95-105	4	59
538	Evidence for interaction between psoriasis-susceptibility loci on chromosomes 6p21 and 1q21. *American Journal of Human Genetics*, **1999**, 65, 1798-800	11	58
537	Sequence-specific modification of genomic DNA by small DNA fragments. *Journal of Clinical Investigation*, **2003**, 112, 637-41	15.9	58
536	Cholesterol-lowering drugs inhibit lectin-like oxidized low-density lipoprotein-1 receptor function by membrane raft disruption. *Molecular Pharmacology*, **2012**, 82, 246-54	4.3	56
535	Variants in RUNX3 contribute to susceptibility to psoriatic arthritis, exhibiting further common ground with ankylosing spondylitis. *Arthritis and Rheumatism*, **2013**, 65, 1224-31		56
534	The etiology of acute recurrent pancreatitis in children: a challenge for pediatricians. *Pancreas*, **2011**, 40, 517-21	2.6	56
533	Targeted correction of a defective selectable marker gene in human epithelial cells by small DNA fragments. *Molecular Therapy*, **2001**, 3, 178-85	11.7	55
532	Parental origin of chromosome 4p deletion in Wolf-Hirschhorn syndrome. *American Journal of Medical Genetics Part A*, **1993**, 47, 921-4		55
531	Multilocus analysis of the fragile X syndrome. *Human Genetics*, **1988**, 78, 201-5	6.3	55
530	Genetic Factors in Systemic Lupus Erythematosus: Contribution to Disease Phenotype. *Journal of Immunology Research*, **2015**, 2015, 745647	4.5	53

529	Allelic variants in the CYP2C9 and VKORC1 loci and interindividual variability in the anticoagulant dose effect of warfarin in Italians. *Pharmacogenomics*, **2007**, 8, 1545-50	2.6	53
528	Association between CYP2B6 polymorphisms and Nevirapine-induced SJS/TEN: a pharmacogenetics study. *European Journal of Clinical Pharmacology*, **2013**, 69, 1909-16	2.8	51
527	Biomarkers in COPD. *Pulmonary Pharmacology and Therapeutics*, **2010**, 23, 493-500	3.5	51
526	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, 462-5	4	51
525	Transposition of the great arteries associated with deletion of chromosome 22q11. *American Journal of Cardiology*, **1995**, 75, 95-8	3	51
524	Delta F508 gene deletion in cystic fibrosis in southern Europe. *Lancet, The*, **1989**, 2, 1404	40	51
523	TCF7L2 gene polymorphisms and type 2 diabetes: association with diabetic retinopathy and cardiovascular autonomic neuropathy. *Acta Diabetologica*, **2013**, 50, 789-99	3.9	50
522	cDNA characterization and chromosomal mapping of two human homologues of the Drosophila dishevelled polarity gene. *Human Molecular Genetics*, **1996**, 5, 953-8	5.6	50
521	Age-related macular degeneration: insights into inflammatory genes. *Journal of Ophthalmology*, **2014**, 2014, 582842	2	49
520	MicroRNA genetic variations: association with type 2 diabetes. *Acta Diabetologica*, **2013**, 50, 867-72	3.9	49
519	"The Linosa Study": epidemiological and heritability data of the metabolic syndrome in a Caucasian genetic isolate. *Nutrition, Metabolism and Cardiovascular Diseases*, **2009**, 19, 455-61	4.5	49
518	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	6.4	49
517	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
516	COVID-19 and Genetic Variants of Protein Involved in the SARS-CoV-2 Entry into the Host Cells. *Genes*, **2020**, 11,	4.2	48
515	Functional analysis and molecular dynamics simulation of LOX-1 K167N polymorphism reveal alteration of receptor activity. *PLoS ONE*, **2009**, 4, e4648	3.7	47
514	Biased T-cell receptor repertoires in patients with chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Clinical and Experimental Immunology*, **2003**, 132, 323-31	6.2	47
513	The mutation spectrum of the EDA gene in X-linked anhidrotic ectodermal dysplasia. *Human Mutation*, **2001**, 17, 349	4.7	47
512	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	3.7	47

511	Carnitine palmitoyl transferase-1A (CPT1A): a new tumor specific target in human breast cancer. *Oncotarget*, **2016**, 7, 19982-96	3.3	47
510	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, E192-7	5.6	46
509	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, 4467-71	5.6	46
508	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
507	TRAF3IP2 gene and systemic lupus erythematosus: association with disease susceptibility and pericarditis development. *Immunogenetics*, **2013**, 65, 703-9	3.2	44
506	Towards the application of precision medicine in Age-Related Macular Degeneration. *Progress in Retinal and Eye Research*, **2018**, 63, 132-146	20.5	44
505	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, 982-4	4.3	43
504	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, 639-46	5.8	42
503	Association of the G289S single nucleotide polymorphism in the HSD17B3 gene with prostate cancer in Italian men. *Prostate*, **2002**, 53, 65-8	4.2	42
502	Mapping a dominant form of multinodular goiter to chromosome Xp22. *American Journal of Human Genetics*, **2000**, 67, 1004-7	11	42
501	Mandibuloacral dysplasia: A premature ageing disease with aspects of physiological ageing. *Ageing Research Reviews*, **2018**, 42, 1-13	12	41
500	Preferential central nucleation of type 2 myofibers is an invariable feature of myotonic dystrophy type 2. *Muscle and Nerve*, **2008**, 38, 1405-1411	3.4	40
499	Human UDP-galactose 4Lepimerase (GALE) gene and identification of five missense mutations in patients with epimerase-deficiency galactosemia. *Molecular Genetics and Metabolism*, **1998**, 63, 26-30	3.7	40
498	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
497	TRAF3IP2 gene is associated with cutaneous extraintestinal manifestations in inflammatory bowel disease. *Journal of Crohn's and Colitis*, **2013**, 7, 44-52	1.5	39
496	Protein farnesylation and disease. *Journal of Inherited Metabolic Disease*, **2012**, 35, 917-26	5.4	39
495	R501X and 2282del4 filaggrin mutations do not confer susceptibility to psoriasis and atopic dermatitis in Italian patients. *Dermatology*, **2008**, 216, 83-4	4.4	39
494	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, 347-51	4.1	39

493	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, 491-7	4.3	39
492	Non-invasive early prenatal molecular diagnosis using retrieved transcervical trophoblast cells. *Human Genetics*, **1996**, 97, 150-5	6.3	39
491	Early structural and functional changes in liver of rats treated with a single dose of valproic acid. *Hepatology*, **1984**, 4, 1159-66	11.2	39
490	Identification of eight novel mutations in a collaborative analysis of a part of the second transmembrane domain of the CFTR gene. *Genomics*, **1993**, 16, 296-7	4.3	38
489	The lectin-like oxidized LDL receptor-1: a new potential molecular target in colorectal cancer. *Oncotarget*, **2016**, 7, 14765-80	3.3	38
488	Characterization of ANKRD11 mutations in humans and mice related to KBG syndrome. *Human Genetics*, **2015**, 134, 181-90	6.3	37
487	Risk prediction for clinical phenotype in myotonic dystrophy type 1: data from 2,650 patients. *Genetic Testing and Molecular Biomarkers*, **2007**, 11, 84-90		37
486	One hundred sixteen cases of acute liver failure treated with MARS. *Transplantation Proceedings*, **2005**, 37, 2557-9	1.1	37
485	(CTG)n triplet mutation and phenotype manifestations in myotonic dystrophy patients. *Biochemical Medicine and Metabolic Biology*, **1993**, 50, 85-92		37
484	Analysis of Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. *Genes*, **2020**, 11,	4.2	36
483	Drugs affecting prelamin A processing: effects on heterochromatin organization. *Experimental Cell Research*, **2008**, 314, 453-62	4.2	36
482	Valproic acid induces neuroendocrine differentiation and UGT2B7 up-regulation in human prostate carcinoma cell line. *Drug Metabolism and Disposition*, **2007**, 35, 968-72	4	36
481	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, 329-34	6.9	36
480	Association of dopamine D4 receptor (DRD4) exon III repeat polymorphism with temperament in 3-year-old infants. *Neurogenetics*, **2003**, 4, 207-12	3	36
479	Lamin A precursor induces barrier-to-autointegration factor nuclear localization. *Cell Cycle*, **2010**, 9, 2600-10	4.1	35
478	Typing of ARMS2 and CFH in age-related macular degeneration: case-control study and assessment of frequency in the Italian population. *JAMA Ophthalmology*, **2009**, 127, 1368-72		35
477	Meiotic drive at the myotonic dystrophy locus. *Journal of Medical Genetics*, **1994**, 31, 980	5.8	35
476	Co-segregation of DM2 with a recessive CLCN1 mutation in juvenile onset of myotonic dystrophy type 2. *Journal of Neurology*, **2012**, 259, 2090-9	5.5	34

475	Rescue of murine silica-induced lung injury and fibrosis by human embryonic stem cells. *European Respiratory Journal*, **2012**, 39, 446-57	13.6	34
474	Gene expression analysis in myotonic dystrophy: indications for a common molecular pathogenic pathway in DM1 and DM2. *Gene Expression*, **2007**, 13, 339-51	3.4	34
473	Biochemical characterization of two GALK1 mutations in patients with galactokinase deficiency. *Human Mutation*, **2004**, 23, 396	4.7	34
472	The strange case of the "lumper" lamin A/C gene and human premature ageing. *Trends in Molecular Medicine*, **2003**, 9, 370-5	11.5	34
471	Analysis of the elastin gene in 60 patients with clinical diagnosis of Williams syndrome. *Human Genetics*, **1995**, 96, 444-8	6.3	34
470	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
469	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
468	Pediatric acute liver failure with molecular adsorbent recirculating system treatment. *Transplantation Proceedings*, **2008**, 40, 1921-4	1.1	32
467	The R527H mutation in LMNA gene causes an increased sensitivity to ionizing radiation. *Cell Cycle*, **2008**, 7, 2030-7	4.7	32
466	Psoriatic arthritis and CARD15 gene polymorphisms: no evidence for association in the Italian population. *Journal of Investigative Dermatology*, **2004**, 122, 1106-7	4.3	32
465	Variations in the NMDA receptor subunit 2B gene (GRIN2B) and schizophrenia: a case-control study. *American Journal of Medical Genetics Part A*, **2004**, 128B, 27-9		32
464	Paternal origin of LMNA mutations in Hutchinson-Gilford progeria. *Clinical Genetics*, **2004**, 65, 52-4	4	32
463	Association study of a promoter polymorphism of UFD1L gene with schizophrenia. *American Journal of Medical Genetics Part A*, **2001**, 105, 529-33		32
462	Male hypogonadism in myotonic dystrophy is related to (CTG)n triplet mutation. *Journal of Endocrinological Investigation*, **1994**, 17, 381-3	5.2	32
461	Critical involvement of the ATM-dependent DNA damage response in the apoptotic demise of HIV-1-elicited syncytia. *PLoS ONE*, **2008**, 3, e2458	3.7	32
460	Genetic tests and genomic biomarkers: regulation, qualification and validation. *Clinical Cases in Mineral and Bone Metabolism*, **2008**, 5, 149-54		32
459	Four Copies of SNCA Responsible for Autosomal Dominant Parkinsonꞌs Disease in Two Italian Siblings. *Parkinsonꞌs Disease*, **2015**, 2015, 546462	2.6	31
458	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, 927-31	2.5	31

457	Expression of receptors for native and chemically modified low-density lipoproteins in brain microvessels. *FEBS Letters*, **1997**, 401, 53-8	3.8	31
456	In vitro correction of cystic fibrosis epithelial cell lines by small fragment homologous replacement (SFHR) technique. *BMC Medical Genetics*, **2002**, 3, 8	2.1	31
455	Somatic and gonadal mosaicism in Hutchinson-Gilford progeria. *American Journal of Medical Genetics, Part A*, **2005**, 135, 66-8	2.5	31
454	Isolation and characterization of a novel gene from the DiGeorge chromosomal region that encodes for a mediator subunit. *Genomics*, **2001**, 74, 320-32	4.3	31
453	Simple version of "megaprimer" PCR for site-directed mutagenesis. *BioTechniques*, **1999**, 26, 870-3	2.5	31
452	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
451	Co-localization of susceptibility loci for psoriasis (PSORS4) and atopic dermatitis (ATOD2) on human chromosome 1q21. *Human Heredity*, **2006**, 61, 229-36	1.1	30
450	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, 247-9	11	30
449	Interleukin 12B (IL12B) genetic variation and pulmonary tuberculosis: a study of cohorts from The Gambia, Guinea-Bissau, United States and Argentina. *PLoS ONE*, **2011**, 6, e16656	3.7	29
448	Mandibuloacral dysplasia type A in childhood. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2258-64	2.5	29
447	Predictive factors of outcome after liver transplantation in patients with cirrhosis and hepatocellular carcinoma. *Transplantation Proceedings*, **2005**, 37, 2535-40	1.1	29
446	Hyper-CK-emia as the sole manifestation of myotonic dystrophy type 2. *Muscle and Nerve*, **2005**, 31, 764-7	3.4	29
445	Dopamine D4 receptor (DRD4) polymorphism and adaptability trait during infancy: a longitudinal study in 1- to 5-month-old neonates. *Neurogenetics*, **2001**, 3, 79-82	3	29
444	Evidence for an association between the SRD5A2 (type II steroid 5 alpha-reductase) locus and prostate cancer in Italian patients. *Disease Markers*, **2000**, 16, 147-50	3.2	29
443	Genomic structure, promoter characterisation and mutational analysis of the S100A7 gene: exclusion of a candidate for familial psoriasis susceptibility. *Human Genetics*, **1999**, 104, 130-4	6.3	29
442	Identification of three novel cystic fibrosis mutations in a sample of Italian cystic fibrosis patients. *Human Heredity*, **1993**, 43, 295-300	1.1	29
441	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
440	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

439	The Genetics and the Genomics of Primary Congenital Glaucoma. *BioMed Research International*, **2015**, 2015, 321291	3	28
438	Skeletal phenotype of mandibuloacral dysplasia associated with mutations in ZMPSTE24. *Bone*, **2010**, 47, 591-7	4.7	28
437	Familial partial lipodystrophy, mandibuloacral dysplasia and restrictive dermopathy feature barrier-to-autointegration factor (BAF) nuclear redistribution. *Cell Cycle*, **2012**, 11, 3568-77	4.7	28
436	Characterization of the loricrin (LOR) gene as a positional candidate for the PSORS4 psoriasis susceptibility locus. *Annals of Human Genetics*, **2004**, 68, 639-45	2.2	28
435	Characterization of deletions in the dystrophin gene giving mild phenotypes. *American Journal of Medical Genetics Part A*, **1990**, 37, 136-42		28
434	Exclusion of CARD15/NOD2 as a candidate susceptibility gene to psoriasis in the Italian population. *European Journal of Dermatology*, **2002**, 12, 540-2	0.8	28
433	IROA: International Register of Open Abdomen, preliminary results. *World Journal of Emergency Surgery*, **2017**, 12, 10	9.2	27
432	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
431	IPLEX administration improves motor neuron survival and ameliorates motor functions in a severe mouse model of spinal muscular atrophy. *Molecular Medicine*, **2012**, 18, 1076-85	6.2	27
430	Overexpression of CUGBP1 in skeletal muscle from adult classic myotonic dystrophy type 1 but not from myotonic dystrophy type 2. *PLoS ONE*, **2013**, 8, e83777	3.7	27
429	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: a study in Spanish and Italian populations and meta-analysis. *Arthritis and Rheumatism*, **2011**, 63, 1860-5		27
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