Stylianos Antonarakis

List of Publications by Year in Descending Order

585 papers	82,262 citations	123 h-index	278 g-index
625 ext. papers	93,711 ext. citations	11.2 avg, IF	8.54 L-index

#	Paper	IF	Citations
585	The complete sequence of a human genome.. *Science*, **2022**, 376, 44-53	33.3	107
584	Biallelic truncation variants in ATP9A are associated with a novel autosomal recessive neurodevelopmental disorder. *Npj Genomic Medicine*, **2021**, 6, 94	6.2	2
583	Specific Susceptibility to COVID-19 in Adults with Down Syndrome. *NeuroMolecular Medicine*, **2021**, 23, 561-571	4.6	7
582	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 1246-1254	8.1	0
581	Immune Dysregulation and the Increased Risk of Complications and Mortality Following Respiratory Tract Infections in Adults With Down Syndrome. *Frontiers in Immunology*, **2021**, 12, 621440	8.4	6
580	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. *Genetics in Medicine*, **2021**, 23, 1922-1932	8.1	2
579	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. *Genetics in Medicine*, **2021**, 23, 1873-1881	8.1	1
578	History of the methodology of disease gene identification. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3266-3275	2.5	3
577	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. *Human Mutation*, **2021**, 42, 66-76	4.7	4
576	Perturbations of genes essential for M□llerian duct and W□ffian duct development in Mayer-Rokitansky-K□ster-Hauser syndrome. *American Journal of Human Genetics*, **2021**, 108, 337-345	11	15
575	Three decades of the Human Genome Organization. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3314-3321	2.5	2
574	Kirrel3-Mediated Synapse Formation Is Attenuated by Disease-Associated Missense Variants. *Journal of Neuroscience*, **2020**, 40, 5376-5388	6.6	3
573	Karyotypic Flexibility of the Complex Cancer Genome and the Role of Polyploidization in Maintenance of Structural Integrity of Cancer Chromosomes. *Cancers*, **2020**, 12,	6.6	2
572	Biallelic variants in PSMB1 encoding the proteasome subunit □6 cause impairment of proteasome function, microcephaly, intellectual disability, developmental delay and short stature. *Human Molecular Genetics*, **2020**, 29, 1132-1143	5.6	9
571	Taurine newborn screening to prevent one form of retinal degeneration and cardiomyopathy. *European Journal of Human Genetics*, **2020**, 28, 1479-1480	5.3	0
570	Down syndrome. *Nature Reviews Disease Primers*, **2020**, 6, 9	51.1	120
569	SCN8A heterozygous variants are associated with anoxic-epileptic seizures. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1209-1216	2.5	4

568	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. *American Journal of Human Genetics*, **2020**, 106, 26-40	11	24
567	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. *Human Molecular Genetics*, **2020**, 29, 618-623	5.6	14
566	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. *American Journal of Human Genetics*, **2020**, 107, 564-574	11	8
565	2019 William Allan Award. *American Journal of Human Genetics*, **2020**, 106, 289-294	11	0
564	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. *American Journal of Human Genetics*, **2020**, 107, 311-324	11	6
563	Single cell transcriptome in aneuploidies reveals mechanisms of gene dosage imbalance. *Nature Communications*, **2019**, 10, 4495	17.4	16
562	Carrier screening for recessive disorders. *Nature Reviews Genetics*, **2019**, 20, 549-561	30.1	49
561	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. *American Journal of Human Genetics*, **2019**, 104, 1073-1087	11	8
560	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. *American Journal of Human Genetics*, **2019**, 105, 907-920	11	8
559	Multi-omic measurements of heterogeneity in HeLa cells across laboratories. *Nature Biotechnology*, **2019**, 37, 314-322	44.5	129
558	Human Genomic Variants and Inherited Disease: Molecular Mechanisms and Clinical Consequences **2019**, 125-200		0
557	Biallelic variants in FBXL3 cause intellectual disability, delayed motor development and short stature. *Human Molecular Genetics*, **2019**, 28, 972-979	5.6	9
556	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. *Human Mutation*, **2019**, 40, 267-280	4.7	9
555	Biallelic variants in KIF14 cause intellectual disability with microcephaly. *European Journal of Human Genetics*, **2018**, 26, 330-339	5.3	37
554	Deciphering Cell Lineage Specification during Male Sex Determination with Single-Cell RNA Sequencing. *Cell Reports*, **2018**, 22, 1589-1599	10.6	80
553	Congenital Neuronal Ceroid Lipofuscinosis with a Novel CTSD Gene Mutation: A Rare Cause of Neonatal-Onset Neurodegenerative Disorder. *Neuropediatrics*, **2018**, 49, 150-153	1.6	13
552	Somatic Activating KRAS Mutations in Arteriovenous Malformations of the Brain. *New England Journal of Medicine*, **2018**, 378, 250-261	59.2	195
551	Novel NEXMIF pathogenic variant in a boy with severe autistic features, intellectual disability, and epilepsy, and his mildly affected mother. *Journal of Human Genetics*, **2018**, 63, 847-850	4.3	7

550	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. *Genetics in Medicine*, **2018**, 20, 778-784	8.1	16
549	Excess Synaptojanin 1 Contributes to Place Cell Dysfunction and Memory Deficits in the Aging Hippocampus in Three Types of Alzheimer's Disease. *Cell Reports*, **2018**, 23, 2967-2975	10.6	25
548	Translating molecular advances in Down syndrome and Fragile X syndrome into therapies. *European Neuropsychopharmacology*, **2018**, 28, 675-690	1.2	7
547	Slightly deleterious genomic variants and transcriptome perturbations in Down syndrome embryonic selection. *Genome Research*, **2018**, 28, 1-10	9.7	27
546	Extensive cellular heterogeneity of X inactivation revealed by single-cell allele-specific expression in human fibroblasts. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 13015-13020	11.5	38
545	Bi-allelic Loss-of-Function Variants in DNMBP Cause Infantile Cataracts. *American Journal of Human Genetics*, **2018**, 103, 568-578	11	20
544	Visual impairment and progressive phthisis bulbi caused by recessive pathogenic variant in MARK3. *Human Molecular Genetics*, **2018**, 27, 2703-2711	5.6	15
543	No evidence for the presence of genetic variants predisposing to psychotic disorders on the non-deleted 22q11.2 allele of VCFS patients. *Translational Psychiatry*, **2017**, 7, e1039	8.6	9
542	Detection of Imprinted Genes by Single-Cell Allele-Specific Gene Expression. *American Journal of Human Genetics*, **2017**, 100, 444-453	11	34
541	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. *Nature Communications*, **2017**, 8, 14279	17.4	87
540	MBV: a method to solve sample mislabeling and detect technical bias in large combined genotype and sequencing assay datasets. *Bioinformatics*, **2017**, 33, 1895-1897	7.2	15
539	Genomic databases: A WHO affair. *Science*, **2017**, 356, 812-813	33.3	3
538	Down syndrome and the complexity of genome dosage imbalance. *Nature Reviews Genetics*, **2017**, 18, 147-163	30.1	143
537	Systematic proteome and proteostasis profiling in human Trisomy 21 fibroblast cells. *Nature Communications*, **2017**, 8, 1212	17.4	59
536	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. *Circulation: Cardiovascular Genetics*, **2017**, 10,		14
535	Germline PMS2 and somatic POLE exonuclease mutations cause hypermutability of the leading DNA strand in biallelic mismatch repair deficiency syndrome brain tumours. *Journal of Pathology*, **2017**, 243, 331-341	9.4	9
534	The effect of genetic variation on promoter usage and enhancer activity. *Nature Communications*, **2017**, 8, 1358	17.4	26
533	SERPINI1 pathogenic variants: An emerging cause of childhood-onset progressive myoclonic epilepsy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2456-2460	2.5	7

532	Viva Europa, a Land of Excellence in Research and Innovation for Health and Wellbeing. *Progress in Preventive Medicine (New York, N Y)*, **2017**, 2, e006	0.7	5
531	The genomic landscape of human cellular circadian variation points to a novel role for the signalosome. *ELife*, **2017**, 6,	8.9	6
530	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. *American Journal of Human Genetics*, **2016**, 99, 555-566	11	41
529	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. *American Journal of Human Genetics*, **2016**, 99, 1368-1376	11	32
528	The International Human Epigenome Consortium: A Blueprint for Scientific Collaboration and Discovery. *Cell*, **2016**, 167, 1145-1149	56.2	232
527	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. *Cell*, **2016**, 167, 1398-1414.e24	56.2	339
526	Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. *Human Genomics*, **2016**, 10, 26	6.8	12
525	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. *Human Mutation*, **2016**, 37, 564-9	4.7	792
524	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. *American Journal of Human Genetics*, **2016**, 98, 615-26	11	59
523	Pure Progressive Ataxia and Palatal Tremor (PAPT) Associated with a New Polymerase Gamma (POLG) Mutation. *Cerebellum*, **2016**, 15, 829-831	4.3	13
522	Genomic analysis identifies new drivers and progression pathways in skin basal cell carcinoma. *Nature Genetics*, **2016**, 48, 398-406	36.3	242
521	APOBEC-induced mutations in human cancers are strongly enriched on the lagging DNA strand during replication. *Genome Research*, **2016**, 26, 174-82	9.7	110
520	Brief report: isogenic induced pluripotent stem cell lines from an adult with mosaic down syndrome model accelerated neuronal ageing and neurodegeneration. *Stem Cells*, **2015**, 33, 2077-84	5.8	42
519	Galanin pathogenic mutations in temporal lobe epilepsy. *Human Molecular Genetics*, **2015**, 24, 3082-91	5.6	17
518	Familial epilepsy in Algeria: Clinical features and inheritance profiles. *Seizure: the Journal of the British Epilepsy Association*, **2015**, 31, 12-8	3.2	6
517	Tissue-specific effects of genetic and epigenetic variation on gene regulation and splicing. *PLoS Genetics*, **2015**, 11, e1004958	6	140
516	Opposite phenotypes of muscle strength and locomotor function in mouse models of partial trisomy and monosomy 21 for the proximal Hspa13-App region. *PLoS Genetics*, **2015**, 11, e1005062	6	23
515	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. *Genome Research*, **2015**, 25, 948-57	9.7	38

514	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
513	Exome sequencing in the diagnosis of an atypical phenotype of infantile hyalinosis. *Middle East Journal of Medical Genetics*, **2015**, 4, 18-23		1
512	HIV-1 Nef promotes infection by excluding SERINC5 from virion incorporation. *Nature*, **2015**, 526, 212-7	50.4	269
511	Association between Variants at BCL11A Erythroid-Specific Enhancer and Fetal Hemoglobin Levels among Sickle Cell Disease Patients in Cameroon: Implications for Future Therapeutic Interventions. *OMICS A Journal of Integrative Biology*, **2015**, 19, 627-31	3.8	15
510	A large genomic deletion leads to enhancer adoption by the lamin B1 gene: a second path to autosomal dominant adult-onset demyelinating leukodystrophy (ADLD). *Human Molecular Genetics*, **2015**, 24, 3143-54	5.6	93
509	CATCHing putative causative variants in consanguineous families. *BMC Bioinformatics*, **2015**, 16, 310	3.6	12
508	The effect of heterogeneous Transcription Start Sites (TSS) on the translatome: implications for the mammalian cellular phenotype. *BMC Genomics*, **2015**, 16, 986	4.5	15
507	HSA21 Single-Minded 2 (Sim2) Binding Sites Co-Localize with Super-Enhancers and Pioneer Transcription Factors in Pluripotent Mouse ES Cells. *PLoS ONE*, **2015**, 10, e0126475	3.7	5
506	A Case of Wiedemann-Steiner Syndrome Associated with a 46,XY Disorder of Sexual Development and Gonadal Dysgenesis. *Sexual Development*, **2015**, 9, 289-95	1.6	10
505	Biased allelic expression in human primary fibroblast single cells. *American Journal of Human Genetics*, **2015**, 96, 70-80	11	88
504	Perturbations of heart development and function in cardiomyocytes from human embryonic stem cells with trisomy 21. *Stem Cells*, **2015**, 33, 1434-46	5.8	19
503	DNA-Methylation Patterns in Trisomy 21 Using Cells from Monozygotic Twins. *PLoS ONE*, **2015**, 10, e0135555	3.7	39
502	Next generation diagnostics on cardiomyopathy. *Molecular Cytogenetics*, **2014**, 7, I4	2	
501	Domains of genome-wide gene expression dysregulation in Down's syndrome. *Nature*, **2014**, 508, 345-50	50.4	195
500	Guidelines for investigating causality of sequence variants in human disease. *Nature*, **2014**, 508, 469-76	50.4	910
499	Cardiomyogenesis is controlled by the miR-99a/let-7c cluster and epigenetic modifications. *Stem Cell Research*, **2014**, 12, 323-37	1.6	49
498	Simultaneous identification and prioritization of variants in familial, de novo, and somatic genetic disorders with VariantMaster. *Genome Research*, **2014**, 24, 349-55	9.7	31
497	Data in brief: Transcriptome analysis of induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. *Genomics Data*, **2014**, 2, 226-9		9

496	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. *Human Mutation*, **2014**, 35, 1203-10	4.7	67
495	A novel homozygous mutation in FGFR3 causes tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly, and arachnodactyly. *Human Mutation*, **2014**, 35, 959-63	4.7	51
494	Multiplex targeted high-throughput sequencing for Mendelian cardiac disorders. *Clinical Genetics*, **2014**, 85, 365-70	4	8
493	Exome sequencing in 53 sporadic cases of schizophrenia identifies 18 putative candidate genes. *PLoS ONE*, **2014**, 9, e112745	3.7	59
492	Cognition and hippocampal plasticity in the mouse is altered by monosomy of a genomic region implicated in Down syndrome. *Genetics*, **2014**, 197, 899-912	4	14
491	Recessive thrombocytopenia likely due to a homozygous pathogenic variant in the FYB gene: case report. *BMC Medical Genetics*, **2014**, 15, 135	2.1	19
490	DEPDC5 mutations in families presenting as autosomal dominant nocturnal frontal lobe epilepsy. *Neurology*, **2014**, 82, 2101-6	6.5	98
489	Loss of function mutation in the palmitoyl-transferase HHAT leads to syndromic 46,XY disorder of sex development by impeding Hedgehog protein palmitoylation and signaling. *PLoS Genetics*, **2014**, 10, e1004340	6	54
488	Exome sequencing reveals a mutation in DMP1 in a family with familial sclerosing bone dysplasia. *Bone*, **2014**, 68, 142-5	4.7	12
487	Frequent cases of RAS-mutated Down syndrome acute lymphoblastic leukaemia lack JAK2 mutations. *Nature Communications*, **2014**, 5, 4654	17.4	41
486	Modelling and rescuing neurodevelopmental defect of Down syndrome using induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. *EMBO Molecular Medicine*, **2014**, 6, 259-77	12	117
485	Extrachromosomal driver mutations in glioblastoma and low-grade glioma. *Nature Communications*, **2014**, 5, 5690	17.4	45
484	Gene age predicts the strength of purifying selection acting on gene expression variation in humans. *American Journal of Human Genetics*, **2014**, 95, 660-74	11	26
483	Analysis of the Born in Bradford birth cohort. *Lancet, The*, **2014**, 383, 123	40	1
482	TNPO3 protects HIV-1 replication from CPSF6-mediated capsid stabilization in the host cell cytoplasm. *Retrovirology*, **2013**, 10, 20	3.6	105
481	Human Gene Mutation in Inherited Disease **2013**, 1-48		1
480	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. *European Journal of Human Genetics*, **2013**, 21, 659-65	5.3	50
479	Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2013**, 162B, 388-403	3.5	76

478	Assessment of transcript reconstruction methods for RNA-seq. *Nature Methods*, **2013**, 10, 1177-84	21.6	477
477	Cell-type, allelic, and genetic signatures in the human pancreatic beta cell transcriptome. *Genome Research*, **2013**, 23, 1554-62	9.7	140
476	Genetic and epigenetic regulation of human lincRNA gene expression. *American Journal of Human Genetics*, **2013**, 93, 1015-26	11	55
475	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. *Nature Biotechnology*, **2013**, 31, 1015-22	44.5	187
474	MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. *Clinical Genetics*, **2013**, 84, 539-45	4	66
473	Pathogenic variants in non-protein-coding sequences. *Clinical Genetics*, **2013**, 84, 422-8	4	28
472	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
471	Purifying selection in mammalian mitochondrial protein-coding genes is highly effective and congruent with evolution of nuclear genes. *Molecular Biology and Evolution*, **2013**, 30, 347-55	8.3	41
470	Mutations in ZMYND10, a gene essential for proper axonemal assembly of inner and outer dynein arms in humans and flies, cause primary ciliary dyskinesia. *American Journal of Human Genetics*, **2013**, 93, 346-56	11	126
469	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. *Genome Research*, **2013**, 23, 1410-21	9.7	48
468	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. *Blood*, **2013**, 122, 554-61	2.2	60
467	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. *ELife*, **2013**, 2, e00523	8.9	295
466	An integrated encyclopedia of DNA elements in the human genome. *Nature*, **2012**, 489, 57-74	50.4	11449
465	Duplications of BHLHA9 are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. *Journal of Medical Genetics*, **2012**, 49, 119-25	5.8	68
464	Preliminary structure and predictive value of attenuated negative symptoms in 22q11.2 deletion syndrome. *Psychiatry Research*, **2012**, 196, 277-84	9.9	55
463	Homozygous deletion of a gene-free region of 4p15 in a child with multiple anomalies: could biallelic loss of conserved, non-coding elements lead to a phenotype?. *European Journal of Medical Genetics*, **2012**, 55, 63-6	2.6	3
462	BDNF and DYRK1A are variable and inversely correlated in lymphoblastoid cell lines from Down syndrome patients. *Molecular Neurobiology*, **2012**, 46, 297-303	6.2	13
461	Landscape of transcription in human cells. *Nature*, **2012**, 489, 101-8	50.4	3544

460	Genomic determinants in the phenotypic variability of Down syndrome. *Progress in Brain Research*, **2012**, 197, 15-28	2.9	19
459	Evidence for transcript networks composed of chimeric RNAs in human cells. *PLoS ONE*, **2012**, 7, e28213	3.7	51
458	Trisomy for synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. *Human Molecular Genetics*, **2012**, 21, 3156-72	5.6	76
457	Tandem repeat sequence variation as causative cis-eQTLs for protein-coding gene expression variation: the case of CSTB. *Human Mutation*, **2012**, 33, 1302-9	4.7	24
456	BLUEPRINT to decode the epigenetic signature written in blood. *Nature Biotechnology*, **2012**, 30, 224-6	44.5	261
455	Novel homozygous, heterozygous and hemizygous FRMD7 gene mutations segregated in the same consanguineous family with congenital X-linked nystagmus. *European Journal of Human Genetics*, **2012**, 20, 1032-6	5.3	14
454	A single-nucleotide substitution mutator phenotype revealed by exome sequencing of human colon adenomas. *Cancer Research*, **2012**, 72, 6279-89	10.1	50
453	Genome-wide linkage and copy number variation analysis reveals 710 kb duplication on chromosome 1p31.3 responsible for autosomal dominant omphalocele. *Journal of Medical Genetics*, **2012**, 49, 270-6	5.8	8
452	High-throughput sequencing and rare genetic diseases. *Molecular Syndromology*, **2012**, 3, 197-203	1.5	5
451	Extensive natural variation for cellular hydrogen peroxide release is genetically controlled. *PLoS ONE*, **2012**, 7, e43566	3.7	4
450	Exome sequencing identifies recurrent somatic MAP2K1 and MAP2K2 mutations in melanoma. *Nature Genetics*, **2011**, 44, 133-9	36.3	313
449	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
448	Mouse models for Down syndrome-associated developmental cognitive disabilities. *Developmental Neuroscience*, **2011**, 33, 404-13	2.2	44
447	A de novo 12q13.11 microdeletion in a patient with severe mental retardation, cleft palate, and high myopia. *European Journal of Medical Genetics*, **2011**, 54, 94-6	2.6	7
446	A user's guide to the encyclopedia of DNA elements (ENCODE). *PLoS Biology*, **2011**, 9, e1001046	9.7	1060
445	Refinement of the X-linked nonsyndromic high-grade myopia locus MYP1 on Xq28 and exclusion of 13 known positional candidate genes by direct sequencing **2011**, 52, 6814-9		19
444	Network-guided analysis of genes with altered somatic copy number and gene expression reveals pathways commonly perturbed in metastatic melanoma. *PLoS ONE*, **2011**, 6, e18369	3.7	40
443	From sequence to functional understanding: the difficult road ahead. *Genome Medicine*, **2011**, 3, 21	14.4	1

442	Systems medicine and integrated care to combat chronic noncommunicable diseases. *Genome Medicine*, **2011**, 3, 43	14.4	137
441	Identification of cis- and trans-regulatory variation modulating microRNA expression levels in human fibroblasts. *Genome Research*, **2011**, 21, 68-73	9.7	57
440	Rapid detection of genetic variants in hypertrophic cardiomyopathy by custom DNA resequencing array in clinical practice. *Journal of Medical Genetics*, **2011**, 48, 572-6	5.8	33
439	Tmprss3, a transmembrane serine protease deficient in human DFNB8/10 deafness, is critical for cochlear hair cell survival at the onset of hearing. *Journal of Biological Chemistry*, **2011**, 286, 17383-97	5.4	54
438	DNA methylation profiles of human active and inactive X chromosomes. *Genome Research*, **2011**, 21, 1592-600	9.7	186
437	A high-resolution anatomical atlas of the transcriptome in the mouse embryo. *PLoS Biology*, **2011**, 9, e1000582	9.7	467
436	Chromosome conformation capture uncovers potential genome-wide interactions between human conserved non-coding sequences. *PLoS ONE*, **2011**, 6, e17634	3.7	16
435	NANOG priming before full reprogramming may generate germ cell tumours. *European Cells and Materials*, **2011**, 22, 258-74; discussio 274	4.3	19
434	New class of gene-termini-associated human RNAs suggests a novel RNA copying mechanism. *Nature*, **2010**, 466, 642-6	50.4	84
433	Mendelian disorders and multifactorial traits: the big divide or one for all?. *Nature Reviews Genetics*, **2010**, 11, 380-4	30.1	60
432	De novo duplication of MECP2 in a girl with mental retardation and no obvious dysmorphic features. *Clinical Genetics*, **2010**, 78, 175-80	4	21
431	Methylation profiling in individuals with uniparental disomy identifies novel differentially methylated regions on chromosome 15. *Genome Research*, **2010**, 20, 1271-8	9.7	36
430	Down syndrome: from understanding the neurobiology to therapy. *Journal of Neuroscience*, **2010**, 30, 14943-5	6.6	89
429	A spectrum of LMX1B mutations in Nail-Patella syndrome: new point mutations, deletion, and evidence of mosaicism in unaffected parents. *Genetics in Medicine*, **2010**, 12, 431-9	8.1	21
428	Eye gaze during face processing in children and adolescents with 22q11.2 deletion syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 665-74	7.2	30
427	Human Genome Sequence and Variation **2010**, 31-53		3
426	Lessons from the Genome-Wide Association Studies for Complex Multifactorial Disorders and Traits **2010**, 287-297		3
425	Human Gene Mutation: Mechanisms and Consequences **2010**, 319-363		5

424	Human Hemoglobin **2010**, 365-401		10
423	A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats. *Human Molecular Genetics*, **2010**, 19, 1967-73	5.6	32
422	A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. *Genome Biology*, **2010**, 11, R64	18.3	12
421	Regulation of fibrinogen production by microRNAs. *Blood*, **2010**, 116, 2608-15	2.2	81
420	Eye Gaze During Face Processing in Children and Adolescents With 22q11.2 Deletion Syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 665-674	7.2	1
419	Array-CGH analysis in a patient with WAGR syndrome and a reciprocal translocation t(2;11) inherited from the normal father with double translocation. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2130-3	2.5	4
418	A teratocarcinoma-like human embryonic stem cell (hESC) line and four hESC lines reveal potentially oncogenic genomic changes. *PLoS ONE*, **2010**, 5, e10263	3.7	42
417	A systematic enhancer screen using lentivector transgenesis identifies conserved and non-conserved functional elements at the Olig1 and Olig2 locus. *PLoS ONE*, **2010**, 5, e15741	3.7	23
416	Genetic structure of Europeans: a view from the North-East. *PLoS ONE*, **2009**, 4, e5472	3.7	237
415	Detection of genomic variation by selection of a 9 mb DNA region and high throughput sequencing. *PLoS ONE*, **2009**, 4, e6659	3.7	11
414	The role of biobanking in rare diseases: European consensus expert group report. *Biopreservation and Biobanking*, **2009**, 7, 155-6	2.1	22
413	A new mouse model for the trisomy of the Abcg1-U2af1 region reveals the complexity of the combinatorial genetic code of down syndrome. *Human Molecular Genetics*, **2009**, 18, 4756-69	5.6	86
412	Common regulatory variation impacts gene expression in a cell type-dependent manner. *Science*, **2009**, 325, 1246-50	33.3	607
411	Disease-causing 7.4 kb cis-regulatory deletion disrupting conserved non-coding sequences and their interaction with the FOXL2 promotor: implications for mutation screening. *PLoS Genetics*, **2009**, 5, e1000522	6	76
410	Common genetic variation and the control of HIV-1 in humans. *PLoS Genetics*, **2009**, 5, e1000791	6	310
409	Transcriptional and post-transcriptional profile of human chromosome 21. *Genome Research*, **2009**, 19, 1471-9	9.7	2
408	Aire-deficient C57BL/6 mice mimicking the common human 13-base pair deletion mutation present with only a mild autoimmune phenotype. *Journal of Immunology*, **2009**, 182, 3902-18	5.3	103
407	Three common polymorphisms in the CYBA gene form a haplotype associated with decreased ROS generation. *Human Mutation*, **2009**, 30, 1123-33	4.7	45

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