Xavier Estivill

List of Publications by Citations

559 papers	47,531 citations	101 h-index	203 g-index
586 ext. papers	53,180 ext. citations	8.9 avg, IF	6.4 L-index

#	Paper	IF	Citations
559	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature*, **2007**, 447, 799-816	50.4	4121
558	Global variation in copy number in the human genome. *Nature*, **2006**, 444, 444-54	50.4	3306
557	International network of cancer genome projects. *Nature*, **2010**, 464, 993-8	50.4	1613
556	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
555	Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia. *Nature* , **2011**, 475, 101-5	50.4	1206
554	Mutations in the cystic fibrosis gene in patients with congenital absence of the vas deferens. *New England Journal of Medicine*, **1995**, 332, 1475-80	59.2	813
553	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
552	Exome sequencing identifies recurrent mutations of the splicing factor SF3B1 gene in chronic lymphocytic leukemia. *Nature Genetics*, **2011**, 44, 47-52	36.3	752
551	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
550	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99	59.2	587
549	Non-coding recurrent mutations in chronic lymphocytic leukaemia. *Nature*, **2015**, 526, 519-24	50.4	565
548	Connexin-26 mutations in sporadic and inherited sensorineural deafness. *Lancet, The*, **1998**, 351, 394-8	40	542
547	SNPassoc: an R package to perform whole genome association studies. *Bioinformatics*, **2007**, 23, 644-5	7.2	513
546	Connexin26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans. *Human Molecular Genetics*, **1997**, 6, 1605-9	5.6	442
545	Familial progressive sensorineural deafness is mainly due to the mtDNA A1555G mutation and is enhanced by treatment of aminoglycosides. *American Journal of Human Genetics*, **1998**, 62, 27-35	11	436
544	Dating the origin of the CCR5-Delta32 AIDS-resistance allele by the coalescence of haplotypes. *American Journal of Human Genetics*, **1998**, 62, 1507-15	11	428
543	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

542	Continuum of overlapping clones spanning the entire human chromosome 21q. *Nature*, **1992**, 359, 380-7	50.4	394
541	MicroRNA profiling of Parkinson's disease brains identifies early downregulation of miR-34b/c which modulate mitochondrial function. *Human Molecular Genetics*, **2011**, 20, 3067-78	5.6	371
540	DSCR1, overexpressed in Down syndrome, is an inhibitor of calcineurin-mediated signaling pathways. *Human Molecular Genetics*, **2000**, 9, 1681-90	5.6	366
539	A candidate for the cystic fibrosis locus isolated by selection for methylation-free islands. *Nature*, **1987**, 326, 840-5	50.4	341
538	Brain-derived neurotrophic factor Val66Met and psychiatric disorders: meta-analysis of case-control studies confirm association to substance-related disorders, eating disorders, and schizophrenia. *Biological Psychiatry*, **2007**, 61, 911-22	7.9	338
537	Copy number variants and genetic traits: closer to the resolution of phenotypic to genotypic variability. *Nature Reviews Genetics*, **2007**, 8, 639-46	30.1	335
536	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
535	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. *Nature Genetics*, **1994**, 6, 420-5	36.3	322
534	High carrier frequency of the 35delG deafness mutation in European populations. Genetic Analysis Consortium of GJB2 35delG. *European Journal of Human Genetics*, **2000**, 8, 19-23	5.3	318
533	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. *Nature Genetics*, **2019**, 51, 1207-1214	36.3	303
532	Neurodevelopmental delay, motor abnormalities and cognitive deficits in transgenic mice overexpressing Dyrk1A (minibrain), a murine model of Down's syndrome. *Human Molecular Genetics* , **2001**, 10, 1915-23	5.6	299
531	Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus. *Nature Genetics*, **1999**, 23, 16-8	36.3	293
530	The origin of the major cystic fibrosis mutation (delta F508) in European populations. *Nature Genetics*, **1994**, 7, 169-75	36.3	284
529	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. *American Journal of Psychiatry*, **2017**, 174, 850-858	11.9	276
528	Differential association of circadian genes with mood disorders: CRY1 and NPAS2 are associated with unipolar major depression and CLOCK and VIP with bipolar disorder. *Neuropsychopharmacology*, **2010**, 35, 1279-89	8.7	264
527	BLUEPRINT to decode the epigenetic signature written in blood. *Nature Biotechnology*, **2012**, 30, 224-6	44.5	261
526	Dyrk1A haploinsufficiency affects viability and causes developmental delay and abnormal brain morphology in mice. *Molecular and Cellular Biology*, **2002**, 22, 6636-47	4.8	251
525	Mutations affecting mRNA splicing are the most common molecular defects in patients with neurofibromatosis type 1. *Human Molecular Genetics*, **2000**, 9, 237-47	5.6	245

524	Genetic structure of Europeans: a view from the North-East. *PLoS ONE*, **2009**, 4, e5472	3.7	237
523	Prevalence and evolutionary origins of the del(GJB6-D13S1830) mutation in the DFNB1 locus in hearing-impaired subjects: a multicenter study. *American Journal of Human Genetics*, **2003**, 73, 1452-8	11	236
522	New loci associated with birth weight identify genetic links between intrauterine growth and adult height and metabolism. *Nature Genetics*, **2013**, 45, 76-82	36.3	232
521	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. *Nature Genetics*, **1999**, 23, 52-7	36.3	232
520	KLHL3 mutations cause familial hyperkalemic hypertension by impairing ion transport in the distal nephron. *Nature Genetics*, **2012**, 44, 456-60, S1-3	36.3	228
519	A myriad of miRNA variants in control and Huntington's disease brain regions detected by massively parallel sequencing. *Nucleic Acids Research*, **2010**, 38, 7219-35	20.1	228
518	A genome-wide association study of anorexia nervosa. *Molecular Psychiatry*, **2014**, 19, 1085-94	15.1	224
517	The human early-life exposome (HELIX): project rationale and design. *Environmental Health Perspectives*, **2014**, 122, 535-44	8.4	219
516	A new human gene from the Down syndrome critical region encodes a proline-rich protein highly expressed in fetal brain and heart. *Human Molecular Genetics*, **1995**, 4, 1935-44	5.6	212
515	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705	11	200
514	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
513	Geographic distribution and regional origin of 272 cystic fibrosis mutations in European populations. The Biomed CF Mutation Analysis Consortium. *Human Mutation*, **1997**, 10, 135-54	4.7	193
512	Variants at APOE influence risk of deep and lobar intracerebral hemorrhage. *Annals of Neurology*, **2010**, 68, 934-43	9.4	191
511	Mutation in TRMU related to transfer RNA modification modulates the phenotypic expression of the deafness-associated mitochondrial 12S ribosomal RNA mutations. *American Journal of Human Genetics*, **2006**, 79, 291-302	11	190
510	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. *Nature Biotechnology*, **2013**, 31, 1015-22	44.5	187
509	Detection of a cystic fibrosis modifier locus for meconium ileus on human chromosome 19q13. *Nature Genetics*, **1999**, 22, 128-9	36.3	187
508	MYO6, the human homologue of the gene responsible for deafness in Snell's waltzer mice, is mutated in autosomal dominant nonsyndromic hearing loss. *American Journal of Human Genetics*, **2001**, 69, 635-40	11	186
507	Confirmation of a double-hit model for the NF1 gene in benign neurofibromas. *American Journal of Human Genetics*, **1997**, 61, 512-9	11	185

506	A polymorphic genomic duplication on human chromosome 15 is a susceptibility factor for panic and phobic disorders. *Cell*, **2001**, 106, 367-79	56.2	184
505	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. *Nature Genetics*, **2019**, 51, 804-814	36.3	181
504	Evidence for the biogenesis of more than 1,000 novel human microRNAs. *Genome Biology*, **2014**, 15, R57	18.3	181
503	Molecular genetics of hearing impairment due to mutations in gap junction genes encoding beta connexins. *Human Mutation*, **2000**, 16, 190-202	4.7	180
502	Schwann cells harbor the somatic NF1 mutation in neurofibromas: evidence of two different Schwann cell subpopulations. *Human Molecular Genetics*, **2000**, 9, 3055-64	5.6	178
501	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
500	A human homologue of Drosophila minibrain (MNB) is expressed in the neuronal regions affected in Down syndrome and maps to the critical region. *Human Molecular Genetics*, **1996**, 5, 1305-10	5.6	170
499	Origin of primate orphan genes: a comparative genomics approach. *Molecular Biology and Evolution* , **2009**, 26, 603-12	8.3	169
498	Association of BDNF with anorexia, bulimia and age of onset of weight loss in six European populations. *Human Molecular Genetics*, **2004**, 13, 1205-12	5.6	168
497	Genome-wide detection of segmental duplications and potential assembly errors in the human genome sequence. *Genome Biology*, **2003**, 4, R25	18.3	164
496	Human chromosome 7: DNA sequence and biology. *Science*, **2003**, 300, 767-72	33.3	159
495	Met66 in the brain-derived neurotrophic factor (BDNF) precursor is associated with anorexia nervosa restrictive type. *Molecular Psychiatry*, **2003**, 8, 745-51	15.1	158
494	Copy number variants and common disorders: filling the gaps and exploring complexity in genome-wide association studies. *PLoS Genetics*, **2007**, 3, 1787-99	6	157
493	Genomic organization, alternative splicing, and expression patterns of the DSCR1 (Down syndrome candidate region 1) gene. *Genomics*, **1997**, 44, 358-61	4.3	153
492	Uniparental disomies, homozygous deletions, amplifications, and target genes in mantle cell lymphoma revealed by integrative high-resolution whole-genome profiling. *Blood*, **2009**, 113, 3059-69	2.2	147
491	Human minibrain homologue (MNBH/DYRK1): characterization, alternative splicing, differential tissue expression, and overexpression in Down syndrome. *Genomics*, **1999**, 57, 407-18	4.3	145
490	Human microRNAs miR-22, miR-138-2, miR-148a, and miR-488 are associated with panic disorder and regulate several anxiety candidate genes and related pathways. *Biological Psychiatry*, **2011**, 69, 526-33	7.9	142
489	Renaming the DSCR1/Adapt78 gene family as RCAN: regulators of calcineurin. *FASEB Journal*, **2007**, 21, 3023-8	0.9	138

488	WASP gene mutations in Wiskott-Aldrich syndrome and X-linked thrombocytopenia. *Human Molecular Genetics*, **1995**, 4, 1127-35	5.6	134
487	Genetic variants and abnormal processing of pre-miR-182, a circadian clock modulator, in major depression patients with late insomnia. *Human Molecular Genetics*, **2010**, 19, 4017-25	5.6	133
486	Genome assembly comparison identifies structural variants in the human genome. *Nature Genetics*, **2006**, 38, 1413-8	36.3	133
485	Neurofibromatosis type 1 due to germ-line mosaicism in a clinically normal father. *New England Journal of Medicine*, **1994**, 331, 1403-7	59.2	132
484	SeqBuster, a bioinformatic tool for the processing and analysis of small RNAs datasets, reveals ubiquitous miRNA modifications in human embryonic cells. *Nucleic Acids Research*, **2010**, 38, e34	20.1	131
483	A pathogenic mechanism in Huntington's disease involves small CAG-repeated RNAs with neurotoxic activity. *PLoS Genetics*, **2012**, 8, e1002481	6	130
482	Spinocerebellar ataxias in Spanish patients: genetic analysis of familial and sporadic cases. The Ataxia Study Group. *Human Genetics*, **1999**, 104, 516-22	6.3	128
481	Exploration of 19 serotoninergic candidate genes in adults and children with attention-deficit/hyperactivity disorder identifies association for 5HT2A, DDC and MAOB. *Molecular Psychiatry*, **2009**, 14, 71-85	15.1	125
480	Recurrent mutations in the NF1 gene are common among neurofibromatosis type 1 patients. *Journal of Medical Genetics*, **2003**, 40, e82	5.8	124
479	DYRK1A-dosage imbalance perturbs NRSF/REST levels, deregulating pluripotency and embryonic stem cell fate in Down syndrome. *American Journal of Human Genetics*, **2008**, 83, 388-400	11	123
478	CA/GT microsatellite alleles within the cystic fibrosis transmembrane conductance regulator (CFTR) gene are not generated by unequal crossingover. *Genomics*, **1991**, 10, 692-8	4.3	122
477	Reduced steady-state levels of mitochondrial RNA and increased mitochondrial DNA amount in human brain with aging. *Molecular Brain Research*, **1997**, 52, 284-9		120
476	Association of irisin with fat mass, resting energy expenditure, and daily activity in conditions of extreme body mass index. *International Journal of Endocrinology*, **2014**, 2014, 857270	2.7	117
475	Nonmuscle myosin heavy-chain gene MYH14 is expressed in cochlea and mutated in patients affected by autosomal dominant hearing impairment (DFNA4). *American Journal of Human Genetics*, **2004**, 74, 770-6	11	117
474	Role of UEV-1, an inactive variant of the E2 ubiquitin-conjugating enzymes, in in vitro differentiation and cell cycle behavior of HT-29-M6 intestinal mucosecretory cells. *Molecular and Cellular Biology*, **1998**, 18, 576-89	4.8	116
473	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility. *Nature Communications*, **2015**, 6, 6916	17.4	115
472	Association of BDNF with restricting anorexia nervosa and minimum body mass index: a family-based association study of eight European populations. *European Journal of Human Genetics*, **2005**, 13, 428-34	5.3	115
471	Functional analysis of mutations in SLC7A9, and genotype-phenotype correlation in non-Type I cystinuria. *Human Molecular Genetics*, **2001**, 10, 305-16	5.6	115

470	Clinical, neuropathologic, and genetic studies of a large spinocerebellar ataxia type 1 (SCA1) kindred: (CAG)n expansion and early premonitory signs and symptoms. *Neurology*, **1995**, 45, 24-30	6.5	115
469	Alterations of neocortical pyramidal cell phenotype in the Ts65Dn mouse model of Down syndrome: effects of environmental enrichment. *Cerebral Cortex*, **2003**, 13, 758-64	5.1	112
468	Predominant occurrence of somatic mutations of the NF2 gene in meningiomas and schwannomas. *Genes Chromosomes and Cancer*, **1995**, 13, 211-6	5	112
467	DYRK1A accumulates in splicing speckles through a novel targeting signal and induces speckle disassembly. *Journal of Cell Science*, **2003**, 116, 3099-107	5.3	111
466	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
465	Mutations in DCHS1 cause mitral valve prolapse. *Nature*, **2015**, 525, 109-13	50.4	107
464	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
463	Enrichment of segmental duplications in regions of breaks of synteny between the human and mouse genomes suggest their involvement in evolutionary rearrangements. *Human Molecular Genetics*, **2003**, 12, 2201-8	5.6	105
462	On dendrites in Down syndrome and DS murine models: a spiny way to learn. *Progress in Neurobiology*, **2004**, 74, 111-26	10.9	104
461	Heterogeneity for mutations in the CFTR gene and clinical correlations in patients with congenital absence of the vas deferens. *Human Reproduction*, **2000**, 15, 1476-83	5.7	104
460	HLA-DRB1*11 and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 15970-5	11.5	103
459	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. *Human Molecular Genetics*, **2003**, 12, 849-58	5.6	101
458	Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. *Human Molecular Genetics*, **2018**, 27, 742-756	5.6	98
457	Characterization of a novel 21-kb deletion, CFTRdele2,3(21 kb), in the CFTR gene: a cystic fibrosis mutation of Slavic origin common in Central and East Europe. *Human Genetics*, **2000**, 106, 259-68	6.3	97
456	The A1555G mutation in the 12S rRNA gene of human mtDNA: recurrent origins and founder events in families affected by sensorineural deafness. *American Journal of Human Genetics*, **1999**, 65, 1349-58	11	96
455	Genetic variants of the FADS gene cluster and ELOVL gene family, colostrums LC-PUFA levels, breastfeeding, and child cognition. *PLoS ONE*, **2011**, 6, e17181	3.7	94
454	Common variants at 12q15 and 12q24 are associated with infant head circumference. *Nature Genetics*, **2012**, 44, 532-538	36.3	94
453	Maximizing association statistics over genetic models. *Genetic Epidemiology*, **2008**, 32, 246-54	2.6	93

452	Dyrk1A expression pattern supports specific roles of this kinase in the adult central nervous system. *Brain Research*, **2003**, 964, 250-63	3.7	92
451	Candidate locus for a nuclear modifier gene for maternally inherited deafness. *American Journal of Human Genetics*, **2000**, 66, 1905-10	11	92
450	Connexin 31 (GJB3) is expressed in the peripheral and auditory nerves and causes neuropathy and hearing impairment. *Human Molecular Genetics*, **2001**, 10, 947-52	5.6	91
449	Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. *Annals of the Rheumatic Diseases*, **2017**, 76, 906-913	2.4	89
448	Genetic heterogeneity in cystinuria: the SLC3A1 gene is linked to type I but not to type III cystinuria. *Proceedings of the National Academy of Sciences of the United States of America*, **1995**, 92, 9667-71	11.5	89
447	Human Early Life Exposome (HELIX) study: a European population-based exposome cohort. *BMJ Open*, **2018**, 8, e021311	3	88
446	High heterogeneity for cystic fibrosis in Spanish families: 75 mutations account for 90% of chromosomes. *Human Genetics*, **1997**, 101, 365-70	6.3	87
445	Microsatellite haplotypes for cystic fibrosis: mutation frameworks and evolutionary tracers. *Human Molecular Genetics*, **1993**, 2, 1015-22	5.6	87
444	Aneuploidy: from a physiological mechanism of variance to Down syndrome. *Physiological Reviews*, **2009**, 89, 887-920	47.9	86
443	DNA hypomethylation at ALOX12 is associated with persistent wheezing in childhood. *American Journal of Respiratory and Critical Care Medicine*, **2012**, 185, 937-43	10.2	86
442	Association study of 10 genes encoding neurotrophic factors and their receptors in adult and child attention-deficit/hyperactivity disorder. *Biological Psychiatry*, **2008**, 63, 935-45	7.9	86
441	Cystic fibrosis transmembrane regulator (CFTR) DeltaF508 mutation and 5T allele in patients with chronic pancreatitis and exocrine pancreatic cancer. PANKRAS II Study Group. *Gut*, **2001**, 48, 70-4	19.2	86
440	Sex differences in mutational rate and mutational mechanism in the NF1 gene in neurofibromatosis type 1 patients. *Human Genetics*, **1996**, 98, 696-9	6.3	85
439	A highly informative CA/GT repeat polymorphism in intron 38 of the human neurofibromatosis type 1 (NF1) gene. *Human Genetics*, **1993**, 92, 429-30	6.3	84
438	Missense mutations in TENM4, a regulator of axon guidance and central myelination, cause essential tremor. *Human Molecular Genetics*, **2015**, 24, 5677-86	5.6	83
437	Presymptomatic analysis of spinocerebellar ataxia type 1 (SCA1) via the expansion of the SCA1 CAG-repeat in a large pedigree displaying anticipation and parental male bias. *Human Molecular Genetics*, **1993**, 2, 2123-8	5.6	81
436	Identification of new putative susceptibility genes for several psychiatric disorders by association analysis of regulatory and non-synonymous SNPs of 306 genes involved in neurotransmission and neurodevelopment. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2009**, 150B, 808-16	3.5	79
435	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

434	Human chromosome 15q11-q14 regions of rearrangements contain clusters of LCR15 duplicons. *European Journal of Human Genetics*, **2002**, 10, 26-35	5.3	79
433	A nuclear defect in the 4p16 region predisposes to multiple mitochondrial DNA deletions in families with Wolfram syndrome. *Journal of Clinical Investigation*, **1996**, 97, 1570-6	15.9	79
432	Two CA/GT repeat polymorphisms in intron 27 of the human neurofibromatosis (NF1) gene. *Human Genetics*, **1994**, 93, 351-2	6.3	78
431	A novel common variant in DCST2 is associated with length in early life and height in adulthood. *Human Molecular Genetics*, **2015**, 24, 1155-68	5.6	77
430	RCAN1 (DSCR1) increases neuronal susceptibility to oxidative stress: a potential pathogenic process in neurodegeneration. *Human Molecular Genetics*, **2007**, 16, 1039-50	5.6	76
429	A highly expressed miR-101 isomiR is a functional silencing small RNA. *BMC Genomics*, **2013**, 14, 104	4.5	75
428	Phosphorylation of calcipressin 1 increases its ability to inhibit calcineurin and decreases calcipressin half-life. *Biochemical Journal*, **2003**, 374, 567-75	3.8	75
427	Intersectin 2, a new multimodular protein involved in clathrin-mediated endocytosis. *FEBS Letters*, **2000**, 478, 43-51	3.8	75
426	Human connexin26 (GJB2) deafness mutations affect the function of gap junction channels at different levels of protein expression. *Human Genetics*, **2002**, 111, 190-7	6.3	73
425	LRRN6A/LERN1 (leucine-rich repeat neuronal protein 1), a novel gene with enriched expression in limbic system and neocortex. *European Journal of Neuroscience*, **2003**, 18, 3167-82	3.5	73
424	Qualitative and quantitative changes in skeletal muscle mtDNA and expression of mitochondrial-encoded genes in the human aging process. *Biochemical and Molecular Medicine*, **1997**, 62, 165-71		72
423	Chromosomal regions containing high-density and ambiguously mapped putative single nucleotide polymorphisms (SNPs) correlate with segmental duplications in the human genome. *Human Molecular Genetics*, **2002**, 11, 1987-95	5.6	71
422	A new aspartyl protease on 21q22.3, BACE2, is highly similar to Alzheimer's amyloid precursor protein beta-secretase. *Cytogenetic and Genome Research*, **2000**, 89, 177-84	1.9	71
421	Extensive analysis of 40 infertile patients with congenital absence of the vas deferens: in 50% of cases only one CFTR allele could be detected. *Human Genetics*, **1995**, 95, 205-11	6.3	71
420	Assessment of the neuropeptide S system in anxiety disorders. *Biological Psychiatry*, **2010**, 68, 474-83	7.9	70
419	Allele variants in functional MicroRNA target sites of the neurotrophin-3 receptor gene (NTRK3) as susceptibility factors for anxiety disorders. *Human Mutation*, **2009**, 30, 1062-71	4.7	70
418	Towards a European consensus for reporting incidental findings during clinical NGS testing. *European Journal of Human Genetics*, **2015**, 23, 1601-6	5.3	69
417	Human mitochondrial transcription factor B1 as a modifier gene for hearing loss associated with the mitochondrial A1555G mutation. *Molecular Genetics and Metabolism*, **2004**, 82, 27-32	3.7	69

416	Connexin mutations in hearing loss, dermatological and neurological disorders. *Trends in Molecular Medicine*, **2002**, 8, 205-12	11.5	69
415	A brain-derived neurotrophic factor (BDNF) haplotype is associated with antidepressant treatment outcome in mood disorders. *Pharmacogenomics Journal*, **2008**, 8, 101-12	3.5	68
414	Alu-splice cloning of human Intersectin (ITSN), a putative multivalent binding protein expressed in proliferating and differentiating neurons and overexpressed in Down syndrome. *European Journal of Human Genetics*, **1999**, 7, 704-12	5.3	68
413	Multiplex PCR amplification of three microsatellites within the CFTR gene. *Genomics*, **1992**, 13, 1362-4	4.3	68
412	Overexpression of miR-128 specifically inhibits the truncated isoform of NTRK3 and upregulates BCL2 in SH-SY5Y neuroblastoma cells. *BMC Molecular Biology*, **2010**, 11, 95	4.5	67
411	Altered brain-derived neurotrophic factor blood levels and gene variability are associated with anorexia and bulimia. *Genes, Brain and Behavior*, **2007**, 6, 706-16	3.6	67
410	Bronchiectasis in adult patients: an expression of heterozygosity for CFTR gene mutations?. *Clinical Genetics*, **2004**, 65, 490-5	4	67
409	Association of common copy number variants at the glutathione S-transferase genes and rare novel genomic changes with schizophrenia. *Molecular Psychiatry*, **2010**, 15, 1023-33	15.1	65
408	Traffic-related air pollution, oxidative stress genes, and asthma (ECHRS). *Environmental Health Perspectives*, **2009**, 117, 1919-24	8.4	65
407	Targeted next-generation sequencing in steroid-resistant nephrotic syndrome: mutations in multiple glomerular genes may influence disease severity. *European Journal of Human Genetics*, **2015**, 23, 1192-9	5.3	64
406	Mitotic recombination effects homozygosity for NF1 germline mutations in neurofibromas. *Nature Genetics*, **2001**, 28, 294-6	36.3	64
405	Extensive genotyping of the BDNF and NTRK2 genes define protective haplotypes against obsessive-compulsive disorder. *Biological Psychiatry*, **2008**, 63, 619-28	7.9	63
404	Somatic NF1 mutational spectrum in benign neurofibromas: mRNA splice defects are common among point mutations. *Human Genetics*, **2001**, 108, 416-29	6.3	63
403	Molecular cloning, expression, and chromosomal localization of a ubiquitously expressed human 6-phosphofructo-2-kinase/ fructose-2, 6-bisphosphatase gene (PFKFB3). *Cytogenetic and Genome Research*, **1998**, 83, 214-7	1.9	63
402	Complex patterns of copy number variation at sites of segmental duplications: an important category of structural variation in the human genome. *Human Genetics*, **2006**, 120, 270-84	6.3	62
401	Motor phenotypic alterations in TgDyrk1a transgenic mice implicate DYRK1A in Down syndrome motor dysfunction. *Neurobiology of Disease*, **2004**, 15, 132-42	7.5	62
400	The effect of zidovudine on skeletal muscle mtDNA in HIV-1 infected patients with mild or no muscle dysfunction. *Brain*, **1996**, 119 (Pt 4), 1357-64	11.2	62
399	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. *Human Genetics*, **1992**, 89, 653-8	6.3	62

398	Testicular CFTR splice variants in patients with congenital absence of the vas deferens. *Human Molecular Genetics*, **1998**, 7, 1739-43	5.6	61
397	Comprehensive copy number variant (CNV) analysis of neuronal pathways genes in psychiatric disorders identifies rare variants within patients. *Journal of Psychiatric Research*, **2010**, 44, 971-8	5.2	60
396	Multiple deletions of mtDNA in two brothers with sideroblastic anemia and mitochondrial myopathy and in their asymptomatic mother. *Human Molecular Genetics*, **1994**, 3, 1945-9	5.6	60
395	Cloning and characterization of DYRK1B, a novel member of the DYRK family of protein kinases. *Biochemical and Biophysical Research Communications*, **1999**, 254, 474-9	3.4	59
394	Smell-taste dysfunctions in extreme weight/eating conditions: analysis of hormonal and psychological interactions. *Endocrine*, **2016**, 51, 256-67	4	58
393	Diagnosis of autosomal dominant polycystic kidney disease using efficient PKD1 and PKD2 targeted next-generation sequencing. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 412-21	2.3	57
392	Association study of 44 candidate genes with depressive and anxiety symptoms in post-partum women. *Journal of Psychiatric Research*, **2010**, 44, 717-24	5.2	57
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