# Xavier Estivill

# List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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

#	Paper	IF	Citations
559	Analysis of incidental findings in Qatar genome participants reveals novel functional variants in LMNA and DSP <i>Human Molecular Genetics</i> , <b>2022</b> ,	5.6	1
558	Cell type-specific novel long non-coding RNA and circular RNA in the BLUEPRINT hematopoietic transcriptomes atlas. <i>Haematologica</i> , <b>2021</b> , 106, 2613-2623	6.6	5
557	Genetic evaluation of cardiomyopathies in Qatar identifies enrichment of pathogenic sarcomere gene variants and possible founder disease mutations in the Arabs. <i>Molecular Genetics &amp; Cenomic Medicine</i> , <b>2021</b> , 9, e1709	2.3	1
556	The alternative serotonin transporter promoter P2 impacts gene function in females with irritable bowel syndrome. <i>Journal of Cellular and Molecular Medicine</i> , <b>2021</b> , 25, 8047-8061	5.6	1
555	Variability of multi-omics profiles in a population-based child cohort. <i>BMC Medicine</i> , <b>2021</b> , 19, 166	11.4	7
554	Shared genetic risk between eating disorder- and substance-use-related phenotypes: Evidence from genome-wide association studies. <i>Addiction Biology</i> , <b>2021</b> , 26, e12880	4.6	12
553	Efficient and flexible Integration of variant characteristics in rare variant association studies using integrated nested Laplace approximation. <i>PLoS Computational Biology</i> , <b>2021</b> , 17, e1007784	5	O
552	Common Genetic Variation And Age at Onset Of Anorexia Nervosa. <i>Biological Psychiatry Global Open Science</i> , <b>2021</b> ,		3
551	Genome sequencing data analysis for rare disease gene discovery. Briefings in Bioinformatics, 2021,	13.4	2
550	Actionable genomic variants in 6045 participants from the Qatar Genome Program. <i>Human Mutation</i> , <b>2021</b> , 42, 1584	4.7	5
549	In utero and childhood exposure to tobacco smoke and multi-layer molecular signatures in children. <i>BMC Medicine</i> , <b>2020</b> , 18, 243	11.4	6
548	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. <i>Science Advances</i> , <b>2019</b> , 5, eaaw3095	14.3	39
547	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. <i>Biological Psychiatry</i> , <b>2019</b> , 86, 577-586	7.9	24
546	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. <i>Nature Genetics</i> , <b>2019</b> , 51, 804-814	36.3	181
545	eDiVA-Classification and prioritization of pathogenic variants for clinical diagnostics. <i>Human Mutation</i> , <b>2019</b> , 40, 865-878	4.7	8
544	Biallelic loss-of-function LACC1/FAMIN Mutations Presenting as Rheumatoid Factor-Negative Polyarticular Juvenile Idiopathic Arthritis. <i>Scientific Reports</i> , <b>2019</b> , 9, 4579	4.9	14
543	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. <i>Nature Communications</i> , <b>2019</b> , 10, 1150	17.4	55

### (2016-2019)

542	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. <i>Nature Genetics</i> , <b>2019</b> , 51, 1207-1214	36.3	303
541	Allele balance bias identifies systematic genotyping errors and false disease associations. <i>Human Mutation</i> , <b>2019</b> , 40, 115-126	4.7	6
540	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. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 742-756	5.6	98
539	Geolocalisation of athletes for out-of-competition drug testing: ethical considerations. Position statement by the WADA Ethics Panel. <i>British Journal of Sports Medicine</i> , <b>2018</b> , 52, 456-459	10.3	5
538	Circulating miRNAs, isomiRs and small RNA clusters in human plasma and breast milk. <i>PLoS ONE</i> , <b>2018</b> , 13, e0193527	3.7	32
537	miRTrace reveals the organismal origins of microRNA sequencing data. <i>Genome Biology</i> , <b>2018</b> , 19, 213	18.3	25
536	Human Early Life Exposome (HELIX) study: a European population-based exposome cohort. <i>BMJ Open</i> , <b>2018</b> , 8, e021311	3	88
535	Survey of 800+ data sets from human tissue and body fluid reveals xenomiRs are likely artifacts. <i>Rna</i> , <b>2017</b> , 23, 433-445	5.8	53
534	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. <i>American Journal of Psychiatry</i> , <b>2017</b> , 174, 850-858	11.9	276
533	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 695-705	11	200
532	Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. <i>Annals of the Rheumatic Diseases</i> , <b>2017</b> , 76, 906-913	2.4	89
531	Signatures of positive selection reveal a universal role of chromatin modifiers as cancer driver genes. <i>Scientific Reports</i> , <b>2017</b> , 7, 13124	4.9	7
530	The acute effects of ultraviolet radiation on the blood transcriptome are independent of plasma 25OHD. <i>Environmental Research</i> , <b>2017</b> , 159, 239-248	7.9	9
529	Cribado ampliado de portadores en un programa de donacili de ovocitos: Implementacili de un nuevo test y resultados tras dos alis de experiencia. <i>Medicina Reproductiva Y Embriologí</i> Cílica, <b>2017</b> , 4, 113-121	0.1	
528	Exploration of large, rare copy number variants associated with psychiatric and neurodevelopmental disorders in individuals with anorexia nervosa. <i>Psychiatric Genetics</i> , <b>2017</b> , 27, 152-	138	11
527	Detailed analysis of inversions predicted between two human genomes: errors, real polymorphisms, and their origin and population distribution. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 567-5	58 <sup>5</sup> 16	8
526	Contribution of the TTC21B gene to glomerular and cystic kidney diseases. <i>Nephrology Dialysis Transplantation</i> , <b>2017</b> , 32, 151-156	4.3	19
525	Smell-taste dysfunctions in extreme weight/eating conditions: analysis of hormonal and psychological interactions. <i>Endocrine</i> , <b>2016</b> , 51, 256-67	4	58

524	A genome-wide association meta-analysis of diarrhoeal disease in young children identifies FUT2 locus and provides plausible biological pathways. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 4127-4142	5.6	24
523	Heritability and Genome-Wide Association Analyses of Sleep Duration in Children: The EAGLE Consortium. <i>Sleep</i> , <b>2016</b> , 39, 1859-1869	1.1	22
522	Prenatal exposure to mixtures of xenoestrogens and genome-wide DNA methylation in human placenta. <i>Epigenomics</i> , <b>2016</b> , 8, 43-54	4.4	13
521	Specific small-RNA signatures in the amygdala at premotor and motor stages of ParkinsonMdisease revealed by deep sequencing analysis. <i>Bioinformatics</i> , <b>2016</b> , 32, 673-81	7.2	23
520	Targeting CAG repeat RNAs reduces HuntingtonMdisease phenotype independently of huntingtin levels. <i>Journal of Clinical Investigation</i> , <b>2016</b> , 126, 4319-4330	15.9	43
519	NGS-Based Assay for the Identification of Individuals Carrying Recessive Genetic Mutations in Reproductive Medicine. <i>Human Mutation</i> , <b>2016</b> , 37, 516-23	4.7	33
518	Identification of Gene Mutations and Fusion Genes in Patients with Sary Syndrome. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 1490-1499	4.3	51
517	Tying malaria and microRNAs: from the biology to future diagnostic perspectives. <i>Malaria Journal</i> , <b>2016</b> , 15, 167	3.6	13
516	Genome-wide DNA methylation study in human placenta identifies novel loci associated with maternal smoking during pregnancy. <i>International Journal of Epidemiology</i> , <b>2016</b> , 45, 1644-1655	7.8	55
515	Genetic Characteristics of Rheumatic Patients Developing Inflammatory Skin Lesions Induced by Biologic Therapy. <i>Reumatolog</i> Claica (English Edition), <b>2015</b> , 11, 126-127	0.1	
514	Missense mutations in TENM4, a regulator of axon guidance and central myelination, cause essential tremor. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 5677-86	5.6	83
513	Deregulation of key signaling pathways involved in oocyte maturation in FMR1 premutation carriers with Fragile X-associated primary ovarian insufficiency. <i>Gene</i> , <b>2015</b> , 571, 52-7	3.8	11
512	Non-coding recurrent mutations in chronic lymphocytic leukaemia. <i>Nature</i> , <b>2015</b> , 526, 519-24	50.4	565
511	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , <b>2015</b> , 348, 666-9	33.3	170
510	Switching to zebrafish neurobehavioral models: The obsessive-compulsive disorder paradigm. <i>European Journal of Pharmacology</i> , <b>2015</b> , 759, 142-50	5.3	11
509	Genetic variation and alternative splicing. <i>Nature Biotechnology</i> , <b>2015</b> , 33, 357-9	44.5	5
508	Mutations in DCHS1 cause mitral valve prolapse. <i>Nature</i> , <b>2015</b> , 525, 109-13	50.4	107
507	Circulating Betatrophin Levels Are Increased in Anorexia and Decreased in Morbidly Obese Women. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1188-96	5.6	34

### (2014-2015)

506	A novel common variant in DCST2 is associated with length in early life and height in adulthood. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1155-68	5.6	77
505	Targeted next-generation sequencing in steroid-resistant nephrotic syndrome: mutations in multiple glomerular genes may influence disease severity. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 1192-9	5.3	64
504	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility. <i>Nature Communications</i> , <b>2015</b> , 6, 6916	17.4	115
503	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 1601-6	5.3	69
502	HLA-DRB1*11 and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2015</b> , 112, 15970-5	11.5	103
501	Genetic characteristics of rheumatic patients developing inflammatory skin lesions induced by biologic therapy. <i>Reumatologa Claica</i> , <b>2015</b> , 11, 126-7	0.9	
500	Rare variants in EAmyloid precursor protein (APP) and ParkinsonMdisease. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 1328-33	5.3	29
499	Activating mutations cluster in the "molecular brake" regions of protein kinases and do not associate with conserved or catalytic residues. <i>Human Mutation</i> , <b>2014</b> , 35, 318-28	4.7	16
498	Accurate molecular diagnosis of phenylketonuria and tetrahydrobiopterin-deficient hyperphenylalaninemias using high-throughput targeted sequencing. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 528-34	5.3	29
497	Blood expression profiles of fragile X premutation carriers identify candidate genes involved in neurodegenerative and infertility phenotypes. <i>Neurobiology of Disease</i> , <b>2014</b> , 65, 43-54	7.5	18
496	Diagnosis of autosomal dominant polycystic kidney disease using efficient PKD1 and PKD2 targeted next-generation sequencing. <i>Molecular Genetics &amp; Enomic Medicine</i> , <b>2014</b> , 2, 412-21	2.3	57
495	Prenatal exposure to mixtures of xenoestrogens and repetitive element DNA methylation changes in human placenta. <i>Environment International</i> , <b>2014</b> , 71, 81-7	12.9	41
494	Relationship between genome and epigenomechallenges and requirements for future research. <i>BMC Genomics</i> , <b>2014</b> , 15, 487	4.5	21
493	Evidence for the biogenesis of more than 1,000 novel human microRNAs. <i>Genome Biology</i> , <b>2014</b> , 15, R57	718.3	181
492	A genome-wide association study of anorexia nervosa. <i>Molecular Psychiatry</i> , <b>2014</b> , 19, 1085-94	15.1	224
491	Genome-wide analysis of single nucleotide polymorphisms and copy number variants in fibromyalgia suggest a role for the central nervous system. <i>Pain</i> , <b>2014</b> , 155, 1102-1109	8	42
490	ALDH5A1 variability in opioid dependent patients could influence response to methadone treatment. <i>European Neuropsychopharmacology</i> , <b>2014</b> , 24, 420-4	1.2	8
489	Association of irisin with fat mass, resting energy expenditure, and daily activity in conditions of extreme body mass index. <i>International Journal of Endocrinology</i> , <b>2014</b> , 2014, 857270	2.7	117

488	The human early-life exposome (HELIX): project rationale and design. <i>Environmental Health Perspectives</i> , <b>2014</b> , 122, 535-44	8.4	219
487	Validation and genotyping of multiple human polymorphic inversions mediated by inverted repeats reveals a high degree of recurrence. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004208	6	23
486	Extensive sequence analysis of CFTR, SCNN1A, SCNN1B, SCNN1G and SERPINA1 suggests an oligogenic basis for cystic fibrosis-like phenotypes. <i>Clinical Genetics</i> , <b>2014</b> , 86, 91-5	4	14
485	MicroRNA expression profiling in blood from fragile X-associated tremor/ataxia syndrome patients. <i>Genes, Brain and Behavior</i> , <b>2013</b> , 12, 595-603	3.6	25
484	Worldwide population distribution of the common LCE3C-LCE3B deletion associated with psoriasis and other autoimmune disorders. <i>BMC Genomics</i> , <b>2013</b> , 14, 261	4.5	8
483	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 659-65	5.3	50
482	A highly expressed miR-101 isomiR is a functional silencing small RNA. <i>BMC Genomics</i> , <b>2013</b> , 14, 104	4.5	75
481	A common 56-kilobase deletion in a primate-specific segmental duplication creates a novel butyrophilin-like protein. <i>BMC Genetics</i> , <b>2013</b> , 14, 61	2.6	16
480	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. <i>Nature Biotechnology</i> , <b>2013</b> , 31, 1015-22	44.5	187
479	Storage conditions and stability of global DNA methylation in placental tissue. <i>Epigenomics</i> , <b>2013</b> , 5, 341-8	4.4	29
478	Transcriptome and genome sequencing uncovers functional variation in humans. <i>Nature</i> , <b>2013</b> , 501, 506	5 <b>-5</b> 1.4	1323
477	Interaction of SLC1A1 gene variants and life stress on pharmacological resistance in obsessive-compulsive disorder. <i>Pharmacogenomics Journal</i> , <b>2013</b> , 13, 470-5	3.5	11
476	The interaction between Comt and Bdnf variants influences obsessive-compulsive-related dysfunctional beliefs. <i>Journal of Anxiety Disorders</i> , <b>2013</b> , 27, 321-7	10.9	16
475	Screening for the presence of FMR1 premutation alleles in women with fibromyalgia. <i>Gene</i> , <b>2013</b> , 512, 305-8	3.8	11
474	Val66Met BDNF genotypes in melancholic depression: effects on brain structure and treatment outcome. <i>Depression and Anxiety</i> , <b>2013</b> , 30, 225-33	8.4	36
473	Upregulation of a small vault RNA (svtRNA2-1a) is an early event in Parkinson disease and induces neuronal dysfunction. <i>RNA Biology</i> , <b>2013</b> , 10, 1093-106	4.8	32
472	Next generation diagnostics of cystic fibrosis and CFTR-related disorders by targeted multiplex high-coverage resequencing of CFTR. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 455-62	5.8	35
47 <sup>1</sup>	Sporadic and reversible chromothripsis in chronic lymphocytic leukemia revealed by longitudinal genomic analysis. <i>Leukemia</i> , <b>2013</b> , 27, 2376-9	10.7	26

### (2012-2013)

470	New loci associated with birth weight identify genetic links between intrauterine growth and adult height and metabolism. <i>Nature Genetics</i> , <b>2013</b> , 45, 76-82	36.3	232
469	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. <i>Genome Research</i> , <b>2013</b> , 23, 1410-21	9.7	48
468	PeSV-Fisher: identification of somatic and non-somatic structural variants using next generation sequencing data. <i>PLoS ONE</i> , <b>2013</b> , 8, e63377	3.7	16
467	Cluster analysis of clinical data identifies fibromyalgia subgroups. <i>PLoS ONE</i> , <b>2013</b> , 8, e74873	3.7	41
466	Small non-coding RNAs add complexity to the RNA pathogenic mechanisms in trinucleotide repeat expansion diseases. <i>Frontiers in Molecular Neuroscience</i> , <b>2013</b> , 6, 45	6.1	9
465	Association of neurexin 3 polymorphisms with smoking behavior. <i>Genes, Brain and Behavior</i> , <b>2012</b> , 11, 704-11	3.6	24
464	KLHL3 mutations cause familial hyperkalemic hypertension by impairing ion transport in the distal nephron. <i>Nature Genetics</i> , <b>2012</b> , 44, 456-60, S1-3	36.3	228
463	Variation in the BDNF Val66Met polymorphism and response to cognitive-behavior therapy in obsessive-compulsive disorder. <i>European Psychiatry</i> , <b>2012</b> , 27, 386-90	6	32
462	Genetic epistasis in female suicide attempters. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , <b>2012</b> , 38, 294-301	5.5	8
461	Influence of fetal glutathione S-transferase copy number variants on adverse reproductive outcomes. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , <b>2012</b> , 119, 1141-6	3.7	10
460	Aberrant brain microRNA target and miRISC gene expression in the anx/anx anorexia mouse model. <i>Gene</i> , <b>2012</b> , 497, 181-90	3.8	11
459	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. <i>Nature Genetics</i> , <b>2012</b> , 44, 1341-8	36.3	681
458	Fat mass and obesity-associated gene (FTO) in eating disorders: evidence for association of the rs9939609 obesity risk allele with bulimia nervosa and anorexia nervosa. <i>Obesity Facts</i> , <b>2012</b> , 5, 408-19	5.1	37
457	BLUEPRINT to decode the epigenetic signature written in blood. <i>Nature Biotechnology</i> , <b>2012</b> , 30, 224-6	44.5	261
456	Overexpression of the CHRNA5/A3/B4 genomic cluster in mice increases the sensitivity to nicotine and modifies its reinforcing effects. <i>Amino Acids</i> , <b>2012</b> , 43, 897-909	3.5	32
455	A pathogenic mechanism in HuntingtonMdisease involves small CAG-repeated RNAs with neurotoxic activity. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002481	6	130
454	Association between the NMDA glutamate receptor GRIN2B gene and obsessive-compulsive disorder. <i>Journal of Psychiatry and Neuroscience</i> , <b>2012</b> , 37, 273-81	4.5	41
453	DNA hypomethylation at ALOX12 is associated with persistent wheezing in childhood. <i>American Journal of Respiratory and Critical Care Medicine</i> , <b>2012</b> , 185, 937-43	10.2	86

452	CYP2D6 polymorphism in patients with eating disorders. <i>Pharmacogenomics Journal</i> , <b>2012</b> , 12, 173-5	3.5	22
451	Common variants at 12q15 and 12q24 are associated with infant head circumference. <i>Nature Genetics</i> , <b>2012</b> , 44, 532-538	36.3	94
450	Exome sequencing identifies recurrent mutations of the splicing factor SF3B1 gene in chronic lymphocytic leukemia. <i>Nature Genetics</i> , <b>2011</b> , 44, 47-52	36.3	75 <sup>2</sup>
449	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. <i>Biological Psychiatry</i> , <b>2011</b> , 69, 526	5-339	142
448	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. <i>Nature</i> , <b>2011</b> , 478, 97-102	50.4	322
447	D184E mutation in aquaporin-4 gene impairs water permeability and links to deafness. <i>Neuroscience</i> , <b>2011</b> , 197, 80-8	3.9	26
446	Genetic variants of the FADS gene cluster and ELOVL gene family, colostrums LC-PUFA levels, breastfeeding, and child cognition. <i>PLoS ONE</i> , <b>2011</b> , 6, e17181	3.7	94
445	DNA methylation in neurodegenerative disorders: a missing link between genome and environment?. <i>Clinical Genetics</i> , <b>2011</b> , 80, 1-14	4	38
444	Gene-environment interaction in anorexia nervosa: relevance of non-shared environment and the serotonin transporter gene. <i>Molecular Psychiatry</i> , <b>2011</b> , 16, 590-2	15.1	52
443	High risk of lifetime history of suicide attempts among CYP2D6 ultrarapid metabolizers with eating disorders. <i>Molecular Psychiatry</i> , <b>2011</b> , 16, 691-2	15.1	34
442	Variants in estrogen receptor alpha gene are associated with phenotypical expression of obsessive-compulsive disorder. <i>Psychoneuroendocrinology</i> , <b>2011</b> , 36, 473-83	5	35
441	Maternal C-reactive protein levels in pregnancy are associated with wheezing and lower respiratory tract infections in the offspring. <i>American Journal of Obstetrics and Gynecology</i> , <b>2011</b> , 204, 164.e1-9	6.4	22
440	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: a study in Spanish and Italian populations and meta-analysis. <i>Arthritis and Rheumatism</i> , <b>2011</b> , 63, 1860-5		27
439	Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia. <i>Nature</i> , <b>2011</b> , 475, 101-5	50.4	1206
438	MicroRNA profiling of ParkinsonMdisease brains identifies early downregulation of miR-34b/c which modulate mitochondrial function. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 3067-78	5.6	371
437	Meta-analysis confirms the LCE3C_LCE3B deletion as a risk factor for psoriasis in several ethnic groups and finds interaction with HLA-Cw6. <i>Journal of Investigative Dermatology</i> , <b>2011</b> , 131, 1105-9	4.3	79
436	ADRB2 Gly16Arg polymorphism, asthma control and lung function decline. <i>European Respiratory Journal</i> , <b>2011</b> , 38, 1029-35	13.6	18
435	A non-biased framework for the annotation and classification of the non-miRNA small RNA transcriptome. <i>Bioinformatics</i> , <b>2011</b> , 27, 3202-3	7.2	38

434	Tēnicas de Reflexiē Estratējica: Search Conference Momentum. <i>Profesional De La Informacion</i> , <b>2011</b> , 20, 71-77	3.7	
433	Resequencing and association analysis of arylalkylamine N-acetyltransferase (AANAT) gene and its contribution to major depression susceptibility. <i>Journal of Pineal Research</i> , <b>2010</b> , 49, 35-44	10.4	30
432	Characterization of a mouse model overexpressing beta-site APP-cleaving enzyme 2 reveals a new role for BACE2. <i>Genes, Brain and Behavior</i> , <b>2010</b> , 9, 160-72	3.6	19
431	A haplotype of glycogen synthase kinase 3lls associated with early onset of unipolar major depression. <i>Genes, Brain and Behavior</i> , <b>2010</b> , 9, 799-807	3.6	39
430	Design and evaluation of a panel of single-nucleotide polymorphisms in microRNA genomic regions for association studies in human disease. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 218-26	5.3	29
429	Additional support for the association of SLITRK1 var321 and Tourette syndrome. <i>Molecular Psychiatry</i> , <b>2010</b> , 15, 447-50	15.1	53
428	Association of common copy number variants at the glutathione S-transferase genes and rare novel genomic changes with schizophrenia. <i>Molecular Psychiatry</i> , <b>2010</b> , 15, 1023-33	15.1	65
427	International network of cancer genome projects. <i>Nature</i> , <b>2010</b> , 464, 993-8	50.4	1613
426	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. <i>Nature Genetics</i> , <b>2010</b> , 42, 985-90	36.3	773
425	Positionally cloned genes and age-specific effects in asthma and atopy: an international population-based cohort study (ECRHS). <i>Thorax</i> , <b>2010</b> , 65, 124-31	7.3	20
424	Genetic variants and abnormal processing of pre-miR-182, a circadian clock modulator, in major depression patients with late insomnia. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 4017-25	5.6	133
423	SeqBuster, a bioinformatic tool for the processing and analysis of small RNAs datasets, reveals ubiquitous miRNA modifications in human embryonic cells. <i>Nucleic Acids Research</i> , <b>2010</b> , 38, e34	20.1	131
422	A myriad of miRNA variants in control and HuntingtonMdisease brain regions detected by massively parallel sequencing. <i>Nucleic Acids Research</i> , <b>2010</b> , 38, 7219-35	20.1	228
421	Deletion of Late Cornified Envelope 3B and 3C genes is not associated with atopic dermatitis. Journal of Investigative Dermatology, <b>2010</b> , 130, 2057-61	4.3	24
420	Replication of LCE3C-LCE3B CNV as a risk factor for psoriasis and analysis of interaction with other genetic risk factors. <i>Journal of Investigative Dermatology</i> , <b>2010</b> , 130, 979-84	4.3	52
419	Deletion of LCE3C and LCE3B genes at PSORS4 does not contribute to susceptibility to psoriatic arthritis in German patients. <i>Annals of the Rheumatic Diseases</i> , <b>2010</b> , 69, 876-8	2.4	30
418	Assessment of the neuropeptide S system in anxiety disorders. <i>Biological Psychiatry</i> , <b>2010</b> , 68, 474-83	7.9	70
417	Differential association of circadian genes with mood disorders: CRY1 and NPAS2 are associated with unipolar major depression and CLOCK and VIP with bipolar disorder.	8.7	264

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143	Immunohistochemical study of alpha, mu and pi class glutathione S transferase expression in malignant melanoma. <i>British Journal of Dermatology</i> , <b>1997</b> , 136, 345-350	4	22	
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20	Linkage disequilibrium for DNA haplotypes near the cystic fibrosis locus in two south European populations. <i>Human Genetics</i> , <b>1989</b> , 83, 175-8	6.3	17
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18	Detection of a rare-cutter RFLP in a CpG-rich island near the cystic fibrosis locus. <i>Human Genetics</i> , <b>1988</b> , 80, 309-10	6.3	3
17	Genetic differences between cystic fibrosis with and without meconium ileus. <i>Lancet, The</i> , <b>1988</b> , 1, 376-	<b>8</b> 40	45
16	Experience with new DNA markers for the diagnosis of cystic fibrosis. <i>New England Journal of Medicine</i> , <b>1988</b> , 318, 50-1	59.2	49
15	Isolation and mapping of a polymorphic DNA sequence pXH3 on chromosome X [DXS235]. <i>Nucleic Acids Research</i> , <b>1988</b> , 16, 2740	20.1	
14	A rapid method to identify cosmids containing rare restriction sites. <i>Nucleic Acids Research</i> , <b>1987</b> , 15, 1415-25	20.1	22
13	Physical and genetic analysis of cosmids from the vicinity of the cystic fibrosis locus. <i>Nucleic Acids Research</i> , <b>1987</b> , 15, 3639-52	20.1	20
12	The cystic fibrosis locus. <i>Enzyme</i> , <b>1987</b> , 38, 8-13		2
11	A candidate for the cystic fibrosis locus isolated by selection for methylation-free islands. <i>Nature</i> , <b>1987</b> , 326, 840-5	50.4	341
10		50.4	341
	1987, 326, 840-5  Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous		
10	1987, 326, 840-5  Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous DNA probe B79a at 7q22 (HMG8 assignment D7S13). <i>Human Genetics</i> , 1986, 74, 320-2  Chronic T-cell lymphocytosis associated with pure red call aplasia, thymoma and	6.3	24
10	Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous DNA probe B79a at 7q22 (HMG8 assignment D7S13). <i>Human Genetics</i> , <b>1986</b> , 74, 320-2  Chronic T-cell lymphocytosis associated with pure red call aplasia, thymoma and hypogammaglobulinemia. <i>British Journal of Haematology</i> , <b>1985</b> , 61, 582-4  Plasmatic and Urinary Protein C Levels in Nephrotic Syndrome. <i>Thrombosis and Haemostasis</i> , <b>1985</b> ,	6.3 4·5	24
10 9 8	Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous DNA probe B79a at 7q22 (HMG8 assignment D7S13). <i>Human Genetics</i> , <b>1986</b> , 74, 320-2  Chronic T-cell lymphocytosis associated with pure red call aplasia, thymoma and hypogammaglobulinemia. <i>British Journal of Haematology</i> , <b>1985</b> , 61, 582-4  Plasmatic and Urinary Protein C Levels in Nephrotic Syndrome. <i>Thrombosis and Haemostasis</i> , <b>1985</b> , 54, 900-900	6.3 4·5	24 14 4
10 9 8	Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous DNA probe B79a at 7q22 (HMG8 assignment D7S13). <i>Human Genetics</i> , <b>1986</b> , 74, 320-2  Chronic T-cell lymphocytosis associated with pure red call aplasia, thymoma and hypogammaglobulinemia. <i>British Journal of Haematology</i> , <b>1985</b> , 61, 582-4  Plasmatic and Urinary Protein C Levels in Nephrotic Syndrome. <i>Thrombosis and Haemostasis</i> , <b>1985</b> , 54, 900-900  Cell type specific novel lincRNAs and circRNAs in the BLUEPRINT haematopoietic transcriptomes atlas  The 5-HT2A 1438G/A polymorphism in anorexia nervosa: a combined analysis of 316 trios from six	6.3 4·5	24 14 4
10 9 8 7 6	Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous DNA probe B79a at 7q22 (HMG8 assignment D7S13). <i>Human Genetics</i> , 1986, 74, 320-2  Chronic T-cell lymphocytosis associated with pure red call aplasia, thymoma and hypogammaglobulinemia. <i>British Journal of Haematology</i> , 1985, 61, 582-4  Plasmatic and Urinary Protein C Levels in Nephrotic Syndrome. <i>Thrombosis and Haemostasis</i> , 1985, 54, 900-900  Cell type specific novel lincRNAs and circRNAs in the BLUEPRINT haematopoietic transcriptomes atlas  The 5-HT2A 1438G/A polymorphism in anorexia nervosa: a combined analysis of 316 trios from six European centres	6.3 4·5	24 14 4 3

2 Identifying tissues implicated in Anorexia Nervosa using Transcriptomic Imputation

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Multi-omics signatures of the human early life exposome

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