Jacques S Beckmann

List of Publications by Year in Descending Order

334 papers	48,260 citations	104 h-index	217 g-index
348 ext. papers	54,273 ext. citations	12.6 avg, IF	6.12 L-index

#	Paper	IF	Citations
334	GA4GH: International policies and standards for data sharing across genomic research and healthcare.. *Cell Genomics*, **2021**, 1, 100029-100029		20
333	COVID19 Disease Map, a computational knowledge repository of virus-host interaction mechanisms. *Molecular Systems Biology*, **2021**, 17, e10387	12.2	9
332	Genome-wide association study identifies 48 common genetic variants associated with handedness. *Nature Human Behaviour*, **2021**, 5, 59-70	12.8	33
331	Great future or greedy venture: Precision medicine needs philosophy. *Health Science Reports*, **2021**, 4, e376	2.2	1
330	A harmonized meta-knowledgebase of clinical interpretations of somatic genomic variants in cancer. *Nature Genetics*, **2020**, 52, 448-457	36.3	58
329	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. *NeuroImage*, **2019**, 203, 116155	7.9	6
328	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. *Biological Psychiatry*, **2018**, 84, 253-264	7.9	33
327	Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. *Molecular Psychiatry*, **2017**, 22, 836-849	15.1	43
326	CNV-association meta-analysis in 191,161 European adults reveals new loci associated with anthropometric traits. *Nature Communications*, **2017**, 8, 744	17.4	37
325	The Immune Signaling Adaptor LAT Contributes to the Neuroanatomical Phenotype of 16p11.2 BP2-BP3 CNVs. *American Journal of Human Genetics*, **2017**, 101, 564-577	11	17
324	RNAseq analysis of heart tissue from mice treated with atenolol and isoproterenol reveals a reciprocal transcriptional response. *BMC Genomics*, **2016**, 17, 717	4.5	4
323	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. *Biological Psychiatry*, **2016**, 80, 129-139	7.9	57
322	Genome-wide Association Studies Identify Genetic Loci Associated With Albuminuria in Diabetes. *Diabetes*, **2016**, 65, 803-17	0.9	96
321	16p11.2 Locus modulates response to satiety before the onset of obesity. *International Journal of Obesity*, **2016**, 40, 870-6	5.5	16
320	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
319	Orexin and sleep quality in anorexia nervosa: Clinical relevance and influence on treatment outcome. *Psychoneuroendocrinology*, **2016**, 65, 102-8	5	26
318	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120

317	IL-17 receptor A and adenosine deaminase 2 deficiency in siblings with recurrent infections and chronic inflammation. *Journal of Allergy and Clinical Immunology*, **2016**, 137, 1189-1196.e2	11.5	44
316	Reconciling evidence-based medicine and precision medicine in the era of big data: challenges and opportunities. *Genome Medicine*, **2016**, 8, 134	14.4	103
315	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
314	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. *PLoS Genetics*, **2015**, 11, e1005378	6	220
313	Copy number variations and cognitive phenotypes in unselected populations. *JAMA - Journal of the American Medical Association*, **2015**, 313, 2044-54	27.4	96
312	Can we afford to sequence every newborn baby? genome?. *Human Mutation*, **2015**, 36, 283-6	4.7	17
311	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
310	The 16p11.2 locus modulates brain structures common to autism, schizophrenia and obesity. *Molecular Psychiatry*, **2015**, 20, 140-7	15.1	112
309	A higher mutational burden in females supports a "female protective model" in neurodevelopmental disorders. *American Journal of Human Genetics*, **2014**, 94, 415-25	11	319
308	MMP13 mutations are the cause of recessive metaphyseal dysplasia, Spahr type. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1175-9	2.5	10
307	Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, **2014**, 46, 1173-86	36.3	1339
306	Investigation of memory, executive functions, and anatomic correlates in asymptomatic FMR1 premutation carriers. *Neurobiology of Aging*, **2014**, 35, 1939-46	5.6	16
305	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
304	Novel approach identifies SNPs in SLC2A10 and KCNK9 with evidence for parent-of-origin effect on body mass index. *PLoS Genetics*, **2014**, 10, e1004508	6	45
303	Genome-wide association study of metabolic traits reveals novel gene-metabolite-disease links. *PLoS Genetics*, **2014**, 10, e1004132	6	70
302	Potocki-Shaffer deletion encompassing ALX4 in a patient with frontonasal dysplasia phenotype. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 346-52	2.5	11
301	GWAS of human bitter taste perception identifies new loci and reveals additional complexity of bitter taste genetics. *Human Molecular Genetics*, **2014**, 23, 259-67	5.6	39
300	Influence of CRTC1 polymorphisms on body mass index and fat mass in psychiatric patients and the general adult population. *JAMA Psychiatry*, **2013**, 70, 1011-9	14.5	37

299	Discovery and refinement of loci associated with lipid levels. *Nature Genetics*, **2013**, 45, 1274-1283	36.3	1904
298	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
297	SCRIB and PUF60 are primary drivers of the multisystemic phenotypes of the 8q24.3 copy-number variant. *American Journal of Human Genetics*, **2013**, 93, 798-811	11	58
296	Identification of heart rate-associated loci and their effects on cardiac conduction and rhythm disorders. *Nature Genetics*, **2013**, 45, 621-31	36.3	219
295	Target sequencing, cell experiments, and a population study establish endothelial nitric oxide synthase (eNOS) gene as hypertension susceptibility gene. *Hypertension*, **2013**, 62, 844-52	8.5	39
294	Sex-stratified genome-wide association studies including 270,000 individuals show sexual dimorphism in genetic loci for anthropometric traits. *PLoS Genetics*, **2013**, 9, e1003500	6	277
293	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 16139-44	11.5	104
292	Sh3tc2 deficiency affects neuregulin-1/ErbB signaling. *Glia*, **2013**, 61, 1041-51	9	26
291	Novel FOXF1 mutations in sporadic and familial cases of alveolar capillary dysplasia with misaligned pulmonary veins imply a role for its DNA binding domain. *Human Mutation*, **2013**, 34, 801-11	4.7	80
290	The Growing Importance of CNVs: New Insights for Detection and Clinical Interpretation. *Frontiers in Genetics*, **2013**, 4, 92	4.5	38
289	Rare genomic structural variants in complex disease: lessons from the replication of associations with obesity. *PLoS ONE*, **2013**, 8, e58048	3.7	27
288	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. *Human Mutation*, **2012**, 33, 165-79	4.7	36
287	A multi-SNP locus-association method reveals a substantial fraction of the missing heritability. *American Journal of Human Genetics*, **2012**, 91, 863-71	11	37
286	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
285	Genome-wide meta-analysis of common variant differences between men and women. *Human Molecular Genetics*, **2012**, 21, 4805-15	5.6	24
284	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. *Nature*, **2012**, 485, 363-7	50.4	281
283	Genetic testing in patients with obesity. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2012**, 26, 133-43	6.5	16
282	Identification and validation of copy number variants using SNP genotyping arrays from a large clinical cohort. *BMC Genomics*, **2012**, 13, 241	4.5	14

281	Carriers of the fragile X mental retardation 1 (FMR1) premutation allele present with increased levels of cytokine IL-10. *Journal of Neuroinflammation*, **2012**, 9, 238	10.1	17
280	FTO genotype is associated with phenotypic variability of body mass index. *Nature*, **2012**, 490, 267-72	50.4	304
279	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. *Nature Genetics*, **2012**, 44, 260-8	36.3	243
278	Mapping genetic variants associated with beta-adrenergic responses in inbred mice. *PLoS ONE*, **2012**, 7, e41032	3.7	8
277	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. *Nature Genetics*, **2012**, 44, 659-69	36.3	615
276	A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. *Journal of Medical Genetics*, **2012**, 49, 660-8	5.8	182
275	Genomewide association study using a high-density single nucleotide polymorphism array and case-control design identifies a novel essential hypertension susceptibility locus in the promoter region of endothelial NO synthase. *Hypertension*, **2012**, 59, 248-55	8.5	124
274	Caffeine intake and CYP1A2 variants associated with high caffeine intake protect non-smokers from hypertension. *Human Molecular Genetics*, **2012**, 21, 3283-92	5.6	45
273	Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. *Nature Genetics*, **2012**, 44, 991-1005	36.3	621
272	Exome sequencing identifies recurrent somatic MAP2K1 and MAP2K2 mutations in melanoma. *Nature Genetics*, **2011**, 44, 133-9	36.3	313
271	Genetic variation near IRS1 associates with reduced adiposity and an impaired metabolic profile. *Nature Genetics*, **2011**, 43, 753-60	36.3	237
270	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. *Nature* , **2011**, 478, 103-9	50.4	1564
269	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
268	Clarity and claims in variation/mutation databasing. *Nature Biotechnology*, **2011**, 29, 790-2; author reply 792-4	44.5	
267	Genome-wide association identifies nine common variants associated with fasting proinsulin levels and provides new insights into the pathophysiology of type 2 diabetes. *Diabetes*, **2011**, 60, 2624-34	0.9	285
266	Epigenetic modification of the FMR1 gene in fragile X syndrome is associated with differential response to the mGluR5 antagonist AFQ056. *Science Translational Medicine*, **2011**, 3, 64ra1	17.5	287
265	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
264	16q24.1 microdeletion in a premature newborn: usefulness of array-based comparative genomic hybridization in persistent pulmonary hypertension of the newborn. *Pediatric Critical Care Medicine*, **2011**, 12, e427-32	3	13

263	The phenotype of recurrent 10q22q23 deletions and duplications. *European Journal of Human Genetics*, **2011**, 19, 400-8	5.3	52
262	High-level transgene expression by homologous recombination-mediated gene transfer. *Nucleic Acids Research*, **2011**, 39, e104	20.1	38
261	Genome-wide association study identifies two loci strongly affecting transferrin glycosylation. *Human Molecular Genetics*, **2011**, 20, 3710-7	5.6	27
260	Abdominal aortic aneurysm is associated with a variant in low-density lipoprotein receptor-related protein 1. *American Journal of Human Genetics*, **2011**, 89, 619-27	11	145
259	Early occurrence of lung adenocarcinoma and breast cancer after radiotherapy of a chest wall sarcoma in a patient with a de novo germline mutation in TP53. *Familial Cancer*, **2011**, 10, 187-92	3	17
258	Novel method to estimate the phenotypic variation explained by genome-wide association studies reveals large fraction of the missing heritability. *Genetic Epidemiology*, **2011**, 35, 341-9	2.6	22
257	CUBN is a gene locus for albuminuria. *Journal of the American Society of Nephrology: JASN*, **2011**, 22, 555-70	12.7	170
256	Methods for testing association between uncertain genotypes and quantitative traits. *Biostatistics*, **2011**, 12, 1-17	3.7	34
255	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
254	Association of eGFR-Related Loci Identified by GWAS with Incident CKD and ESRD. *PLoS Genetics*, **2011**, 7, e1002292	6	144
253	Genetic male infertility and mutation of CATSPER ion channels. *European Journal of Human Genetics* , **2010**, 18, 1178-84	5.3	110
252	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. *Nature*, **2010**, 463, 671-5	50.4	403
251	Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature*, **2010**, 467, 832-8	50.4	1514
250	Genetic loci influencing kidney function and chronic kidney disease. *Nature Genetics*, **2010**, 42, 373-5	36.3	205
249	Genome-wide association study identifies new HLA class II haplotypes strongly protective against narcolepsy. *Nature Genetics*, **2010**, 42, 786-9	36.3	145
248	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nature Genetics*, **2010**, 42, 949-60	36.3	724
247	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48	36.3	2267
246	Global transcriptional programs in peripheral nerve endoneurium and DRG are resistant to the onset of type 1 diabetic neuropathy in Ins2 mice. *PLoS ONE*, **2010**, 5, e10832	3.7	12

245	Fine mapping of AHI1 as a schizophrenia susceptibility gene: from association to evolutionary evidence. *FASEB Journal*, **2010**, 24, 3066-82	0.9	32
244	Peroxisomal and microsomal lipid pathways associated with resistance to hepatic steatosis and reduced pro-inflammatory state. *Journal of Biological Chemistry*, **2010**, 285, 31011-23	5.4	54
243	Genome-wide meta-analysis for serum calcium identifies significantly associated SNPs near the calcium-sensing receptor (CASR) gene. *PLoS Genetics*, **2010**, 6, e1001035	6	74
242	[R74W;R1070W;D1270N]: a new complex allele responsible for cystic fibrosis. *Journal of Cystic Fibrosis*, **2010**, 9, 447-9	4.1	6
241	Genetic variation in IL28B is associated with chronic hepatitis C and treatment failure: a genome-wide association study. *Gastroenterology*, **2010**, 138, 1338-45, 1345.e1-7	13.3	956
240	Lessons from the Genome-Wide Association Studies for Complex Multifactorial Disorders and Traits **2010**, 287-297		3
239	Penetrance of marked cognitive impairment in older male carriers of the FMR1 gene premutation. *Journal of Medical Genetics*, **2009**, 46, 818-24	5.8	54
238	Transcription factor CTF1 acts as a chromatin domain boundary that shields human telomeric genes from silencing. *Molecular and Cellular Biology*, **2009**, 29, 2409-18	4.8	14
237	Common genetic variation and the control of HIV-1 in humans. *PLoS Genetics*, **2009**, 5, e1000791	6	310
236	Meta-analysis of 28,141 individuals identifies common variants within five new loci that influence uric acid concentrations. *PLoS Genetics*, **2009**, 5, e1000504	6	495
235	Familial occurrence of an association of multiple intestinal atresia and choanal atresia: a new syndrome?. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2661-5	2.5	1
234	Sharing data between LSDBs and central repositories. *Human Mutation*, **2009**, 30, 493-5	4.7	17
233	A single-base substitution within an intronic repetitive element causes dominant retinitis pigmentosa with reduced penetrance. *Human Mutation*, **2009**, 30, 1340-7	4.7	38
232	JNK3 is abundant in insulin-secreting cells and protects against cytokine-induced apoptosis. *Diabetologia*, **2009**, 52, 1871-80	10.3	34
231	Variants in MTNR1B influence fasting glucose levels. *Nature Genetics*, **2009**, 41, 77-81	36.3	584
230	Genome-wide association study identifies eight loci associated with blood pressure. *Nature Genetics*, **2009**, 41, 666-76	36.3	970
229	The Wnt receptor FZD1 mediates chemoresistance in neuroblastoma through activation of the Wnt/beta-catenin pathway. *Oncogene*, **2009**, 28, 2245-56	9.2	137
228	Autosomal-dominant distal myopathy associated with a recurrent missense mutation in the gene encoding the nuclear matrix protein, matrin 3. *American Journal of Human Genetics*, **2009**, 84, 511-8	11	136

227	Mutations in the heparan-sulfate proteoglycan glypican 6 (GPC6) impair endochondral ossification and cause recessive omodysplasia. *American Journal of Human Genetics*, **2009**, 84, 760-70	11	84
226	Limb-girdle muscular dystrophy type 2A can result from accelerated autoproteolytic inactivation of calpain 3. *Biochemistry*, **2009**, 48, 3457-67	3.2	21
225	Mutation screening of the glutamate cysteine ligase modifier (GCLM) gene in patients with schizophrenia. *Psychiatric Genetics*, **2009**, 19, 201-8	2.9	9
224	Association between C-reactive protein and adiposity in women. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 3969-77	5.6	50
223	Cardiovascular response to beta-adrenergic blockade or activation in 23 inbred mouse strains. *PLoS ONE*, **2009**, 4, e6610	3.7	30
222	A modular approach for integrative analysis of large-scale gene-expression and drug-response data. *Nature Biotechnology*, **2008**, 26, 531-9	44.5	99
221	Genome-wide association analysis identifies 20 loci that influence adult height. *Nature Genetics*, **2008**, 40, 575-83	36.3	654
220	Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nature Genetics*, **2008**, 40, 768-75	36.3	1048
219	Automated four-color interphase fluorescence in situ hybridization approach for the simultaneous detection of specific aneuploidies of diagnostic and prognostic significance in high hyperdiploid acute lymphoblastic leukemia. *Cancer Genetics and Cytogenetics*, **2008**, 186, 69-77		13
218	Calpain 3, the "gatekeeper" of proper sarcomere assembly, turnover and maintenance. *Neuromuscular Disorders*, **2008**, 18, 913-21	2.9	97
217	Computational problems in perfect phylogeny haplotyping: typing without calling the allele. *IEEE/ACM Transactions on Computational Biology and Bioinformatics*, **2008**, 5, 101-9	3	6
216	Two trans-acting eQTLs modulate the penetrance of PRPF31 mutations. *Human Molecular Genetics*, **2008**, 17, 3154-65	5.6	36
215	CNVs and genetic medicine (excitement and consequences of a rediscovery). *Cytogenetic and Genome Research*, **2008**, 123, 7-16	1.9	16
214	Genomic determinants of the efficiency of internal ribosomal entry sites of viral and cellular origin. *Nucleic Acids Research*, **2008**, 36, 6918-25	20.1	13
213	In vitro whole-genome analysis identifies a susceptibility locus for HIV-1. *PLoS Biology*, **2008**, 6, e32	9.7	59
212	Association of ABCB1 genetic variants with renal function in Africans and in Caucasians. *BMC Medical Genomics*, **2008**, 1, 21	3.7	12
211	Recommendations for locus-specific databases and their curation. *Human Mutation*, **2008**, 29, 2-5	4.7	52
210	Calcium phosphate transfection generates mammalian recombinant cell lines with higher specific productivity than polyfection. *Biotechnology and Bioengineering*, **2008**, 101, 937-45	4.9	30

209	Subtelomeric 6p deletion: clinical and array-CGH characterization in two patients. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2094-102	2.5	26
208	Premature termination codons in PRPF31 cause retinitis pigmentosa via haploinsufficiency due to nonsense-mediated mRNA decay. *Journal of Clinical Investigation*, **2008**, 118, 1519-31	15.9	72
207	The zinc transporter SLC39A13/ZIP13 is required for connective tissue development; its involvement in BMP/TGF-beta signaling pathways. *PLoS ONE*, **2008**, 3, e3642	3.7	203
206	Genome-wide prediction of matrix attachment regions that increase gene expression in mammalian cells. *Nature Methods*, **2007**, 4, 747-53	21.6	116
205	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
204	The c-Jun N-terminal kinase JNK participates in cytokine- and isolation stress-induced rat pancreatic islet apoptosis. *Diabetologia*, **2007**, 50, 1660-9	10.3	47
203	Contribution of 20 single nucleotide polymorphisms of 13 genes to dyslipidemia associated with antiretroviral therapy. *Pharmacogenetics and Genomics*, **2007**, 17, 755-64	1.9	60
202	Pharmacogenetics of glatiramer acetate therapy for multiple sclerosis reveals drug-response markers. *Pharmacogenetics and Genomics*, **2007**, 17, 657-66	1.9	61
201	Promoter polymorphisms and allelic imbalance in ABCB1 expression. *Pharmacogenetics and Genomics*, **2007**, 17, 951-9	1.9	22
200	Blocking Apoptotic Intracellular Signaling Cascades with Cell-Permeable Peptides. *Current Signal Transduction Therapy*, **2007**, 2, 175-179	0.8	
199	Molecular and cellular basis of calpainopathy (limb girdle muscular dystrophy type 2A). *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2007**, 1772, 128-44	6.9	59
198	SREBP-1c expression in Schwann cells is affected by diabetes and nutritional status. *Molecular and Cellular Neurosciences*, **2007**, 35, 525-34	4.8	29
197	Cell-permeable peptides induce dose- and length-dependent cytotoxic effects. *Biochimica Et Biophysica Acta - Biomembranes*, **2007**, 1768, 2222-34	3.8	70
196	Myotilin is not the causative gene for vocal cord and pharyngeal weakness with distal myopathy (VCPDM). *Annals of Human Genetics*, **2006**, 70, 414-6	2.2	6
195	Confirmation of the origin of NISCH syndrome. *Human Mutation*, **2006**, 27, 408-10	4.7	66
194	Fetus with two identical reciprocal translocations: description of a rare complication of consanguinity. *American Journal of Medical Genetics, Part A*, **2006**, 140, 769-74	2.5	10
193	Homogeneous and nonradioactive high-throughput screening platform for the characterization of kinase inhibitors in cell lysates. *Journal of Biomolecular Screening*, **2006**, 11, 1015-26		25
192	ATM haplotypes and breast cancer risk in Jewish high-risk women. *British Journal of Cancer*, **2006**, 94, 1537-43	8.7	25

191	Genetic characterization of CHO production host DG44 and derivative recombinant cell lines. *Biochemical and Biophysical Research Communications*, **2006**, 340, 1069-77	3.4	117
190	Linkage and mutational analysis of the CDAN1 gene reveals genetic heterogeneity in congenital dyserythropoietic anemia type I. *Blood*, **2006**, 107, 4968-9	2.2	16
189	Identification of putative in vivo substrates of calpain 3 by comparative proteomics of overexpressing transgenic and nontransgenic mice. *Proteomics*, **2006**, 6, 6075-84	4.8	39
188	Mendelian disorders deserve more attention. *Nature Reviews Genetics*, **2006**, 7, 277-82	30.1	168
187	AHI1, a pivotal neurodevelopmental gene, and C6orf217 are associated with susceptibility to schizophrenia. *European Journal of Human Genetics*, **2006**, 14, 1111-9	5.3	64
186	A unique set of SH3-SH3 interactions controls IB1 homodimerization. *EMBO Journal*, **2006**, 25, 785-97	13	35
185	The trace amine receptor 4 gene is not associated with schizophrenia in a sample linked to chromosome 6q23. *Molecular Psychiatry*, **2006**, 11, 119-21	15.1	19
184	The association of DNA sequence variation at the MAOA genetic locus with quantitative behavioural traits in normal males. *Human Genetics*, **2006**, 120, 447-59	6.3	24
183	Identification of C7orf11 (TTDN1) gene mutations and genetic heterogeneity in nonphotosensitive trichothiodystrophy. *American Journal of Human Genetics*, **2005**, 76, 510-6	11	262
182	Trick or treat: the effect of placebo on the power of pharmacogenetic association studies. *Human Genomics*, **2005**, 2, 28-38	6.8	10
181	Fine mapping of a schizophrenia susceptibility locus at chromosome 6q23: increased evidence for linkage and reduced linkage interval. *European Journal of Human Genetics*, **2005**, 13, 763-71	5.3	37
180	Typing without calling the allele: a strategy for inferring SNP haplotypes. *European Journal of Human Genetics*, **2005**, 13, 898-901	5.3	3
179	On ubiquitin ligases and cancer. *Human Mutation*, **2005**, 25, 507-12	4.7	8
178	Analysis of genetic polymorphisms in acetylcholinesterase as reflected in different populations. *Current Alzheimer Research*, **2005**, 2, 207-18	3	12
177	FoldIndex: a simple tool to predict whether a given protein sequence is intrinsically unfolded. *Bioinformatics*, **2005**, 21, 3435-8	7.2	776
176	Association Down syndrome-retinoblastoma: a new observation. *Ophthalmic Genetics*, **2005**, 26, 151-2	1.2	5
175	Use of a combined ex vivo/in vivo population approach for screening of human genes involved in the human immunodeficiency virus type 1 life cycle for variants influencing disease progression. *Journal of Virology*, **2005**, 79, 12674-80	6.6	50
174	Intracellular stress signaling pathways activated during human islet preparation and following acute cytokine exposure. *Diabetes*, **2004**, 53, 2815-23	0.9	151

173	Genomic profiling of interpopulation diversity guides prioritization of candidate-genes for autoimmunity. *Genes and Immunity*, **2004**, 5, 493-504	4.4	10
172	On the applicability of a haplotype map to un-assayed populations. *Human Genetics*, **2004**, 114, 214-7	6.3	4
171	A paradigm for single nucleotide polymorphism analysis: the case of the acetylcholinesterase gene. *Human Mutation*, **2004**, 24, 408-16	4.7	20
170	Proteomic signatures: amino acid and oligopeptide compositions differentiate among phyla. *Proteins: Structure, Function and Bioinformatics*, **2004**, 54, 20-40	4.2	117
169	Circadian regulation of islet genes involved in insulin production and secretion. *Molecular and Cellular Endocrinology*, **2004**, 226, 59-66	4.4	77
168	Recovering frequencies of known haplotype blocks from single-nucleotide polymorphism allele frequencies. *Genetics*, **2004**, 166, 2001-6	4	2
167	Recovering Frequencies of Known Haplotype Blocks From Single-Nucleotide Polymorphism Allele Frequencies. *Genetics*, **2004**, 166, 2001-2006	4	
166	Ubiquitin ligases as cancer genes. *Nature Reviews Cancer*, **2004**, 4, 654-654	31.3	1
165	Computational Problems in Perfect Phylogeny Haplotyping: Xor-Genotypes and Tag SNPs. *Lecture Notes in Computer Science*, **2004**, 14-31	0.9	15
164	Calpain 3 cleaves filamin C and regulates its ability to interact with gamma- and delta-sarcoglycans. *Muscle and Nerve*, **2003**, 28, 472-83	3.4	79
163	CATSPER2, a human autosomal nonsyndromic male infertility gene. *European Journal of Human Genetics*, **2003**, 11, 497-502	5.3	131
162	Calcium- and proteasome-dependent degradation of the JNK scaffold protein islet-brain 1. *Journal of Biological Chemistry*, **2003**, 278, 48720-6	5.4	18
161	Resolution of haplotypes and haplotype frequencies from SNP genotypes of pooled samples **2003**,		5
160	USH3A transcripts encode clarin-1, a four-transmembrane-domain protein with a possible role in sensory synapses. *European Journal of Human Genetics*, **2002**, 10, 339-50	5.3	136
159	Molecular adaptations of neuromuscular disease-associated proteins in response to eccentric exercise in human skeletal muscle. *Journal of Physiology*, **2002**, 543, 297-306	3.9	159
158	Six and Eya expression during human somitogenesis and MyoD gene family activation. *Journal of Muscle Research and Cell Motility*, **2002**, 23, 255-64	3.5	52
157	Stable expression of calpain 3 from a muscle transgene in vivo: immature muscle in transgenic mice suggests a role for calpain 3 in muscle maturation. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 8874-9	11.5	82
156	A highly significant association between a COMT haplotype and schizophrenia. *American Journal of Human Genetics*, **2002**, 71, 1296-302	11	589

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