Jacques S Beckmann

List of Publications by Citations

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	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
333	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48	36.3	2267
332	Discovery and refinement of loci associated with lipid levels. *Nature Genetics*, **2013**, 45, 1274-1283	36.3	1904
331	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. *Nature* , **2011**, 478, 103-9	50.4	1564
330	Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature*, **2010**, 467, 832-8	50.4	1514
329	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
328	Targeted disruption of the mouse Caspase 8 gene ablates cell death induction by the TNF receptors, Fas/Apo1, and DR3 and is lethal prenatally. *Immunity*, **1998**, 9, 267-76	32.3	1048
327	Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nature Genetics*, **2008**, 40, 768-75	36.3	1048
326	Genome-wide association study identifies eight loci associated with blood pressure. *Nature Genetics*, **2009**, 41, 666-76	36.3	970
325	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
324	A Gene Map of the Human Genome. *Science*, **1996**, 274, 540-546	33.3	924
323	Mutations in the proteolytic enzyme calpain 3 cause limb-girdle muscular dystrophy type 2A. *Cell*, **1995**, 81, 27-40	56.2	835
322	FoldIndex: a simple tool to predict whether a given protein sequence is intrinsically unfolded. *Bioinformatics*, **2005**, 21, 3435-8	7.2	776
321	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
320	Genome-wide association analysis identifies 20 loci that influence adult height. *Nature Genetics*, **2008**, 40, 575-83	36.3	654
319	Familial hyperglycemia due to mutations in glucokinase. Definition of a subtype of diabetes mellitus. *New England Journal of Medicine*, **1993**, 328, 697-702	59.2	642
318	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

317	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
316	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
315	A highly significant association between a COMT haplotype and schizophrenia. *American Journal of Human Genetics*, **2002**, 71, 1296-302	11	589
314	Variants in MTNR1B influence fasting glucose levels. *Nature Genetics*, **2009**, 41, 77-81	36.3	584
313	A gene related to Caenorhabditis elegans spermatogenesis factor fer-1 is mutated in limb-girdle muscular dystrophy type 2B. *Nature Genetics*, **1998**, 20, 37-42	36.3	545
312	Dynamic molecular combing: stretching the whole human genome for high-resolution studies. *Science*, **1997**, 277, 1518-23	33.3	506
311	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
310	A physical map of 30,000 human genes. *Science*, **1998**, 282, 744-6	33.3	472
309	Survey of human and rat microsatellites. *Genomics*, **1992**, 12, 627-31	4.3	469
308	Beta-sarcoglycan: characterization and role in limb-girdle muscular dystrophy linked to 4q12. *Nature Genetics*, **1995**, 11, 257-65	36.3	433
307	Mutations in the gene encoding mevalonate kinase cause hyper-IgD and periodic fever syndrome. International Hyper-IgD Study Group. *Nature Genetics*, **1999**, 22, 178-81	36.3	427
306	Missense mutations in the adhalin gene linked to autosomal recessive muscular dystrophy. *Cell*, **1994**, 78, 625-33	56.2	415
305	The UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase gene is mutated in recessive hereditary inclusion body myopathy. *Nature Genetics*, **2001**, 29, 83-7	36.3	407
304	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. *Nature*, **2010**, 463, 671-5	50.4	403
303	Cardiac myosin binding protein-C gene splice acceptor site mutation is associated with familial hypertrophic cardiomyopathy. *Nature Genetics*, **1995**, 11, 438-40	36.3	363
302	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
301	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
300	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322

299	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
298	Transcription and processing of intervening sequences in yeast tRNA genes. *Cell*, **1978**, 14, 221-36	56.2	319
297	Exome sequencing identifies recurrent somatic MAP2K1 and MAP2K2 mutations in melanoma. *Nature Genetics*, **2011**, 44, 133-9	36.3	313
296	Common genetic variation and the control of HIV-1 in humans. *PLoS Genetics*, **2009**, 5, e1000791	6	310
295	FTO genotype is associated with phenotypic variability of body mass index. *Nature*, **2012**, 490, 267-72	50.4	304
294	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
293	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
292	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
291	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. *Nature*, **2012**, 485, 363-7	50.4	281
290	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
289	Restriction fragment length polymorphisms in genetic improvement: methodologies, mapping and costs. *Theoretical and Applied Genetics*, **1983**, 67, 35-43	6	271
288	Identification of C7orf11 (TTDN1) gene mutations and genetic heterogeneity in nonphotosensitive trichothiodystrophy. *American Journal of Human Genetics*, **2005**, 76, 510-6	11	262
287	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
286	Chromosomal assignment of the second locus for autosomal dominant cerebellar ataxia (SCA2) to chromosome 12q23-24.1. *Nature Genetics*, **1993**, 4, 295-9	36.3	257
285	Genetic polymorphism in varietal identification and genetic improvement. *Theoretical and Applied Genetics*, **1983**, 67, 25-33	6	250
284	Calpain 3 deficiency is associated with myonuclear apoptosis and profound perturbation of the IkappaB alpha/NF-kappaB pathway in limb-girdle muscular dystrophy type 2A. *Nature Medicine*, **1999**, 5, 503-11	50.5	247
283	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
282	Genetic variation near IRS1 associates with reduced adiposity and an impaired metabolic profile. *Nature Genetics*, **2011**, 43, 753-60	36.3	237

281	Dysferlin is a plasma membrane protein and is expressed early in human development. *Human Molecular Genetics*, **1999**, 8, 855-61	5.6	230
280	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
279	Identification of heart rate-associated loci and their effects on cardiac conduction and rhythm disorders. *Nature Genetics*, **2013**, 45, 621-31	36.3	219
278	Genetic loci influencing kidney function and chronic kidney disease. *Nature Genetics*, **2010**, 42, 373-5	36.3	205
277	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
276	A gene for maturity onset diabetes of the young (MODY) maps to chromosome 12q. *Nature Genetics*, **1995**, 9, 418-23	36.3	188
275	Localization of merosin-negative congenital muscular dystrophy to chromosome 6q2 by homozygosity mapping. *Human Molecular Genetics*, **1994**, 3, 1657-61	5.6	188
274	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
273	CUBN is a gene locus for albuminuria. *Journal of the American Society of Nephrology: JASN*, **2011**, 22, 555-70	12.7	170
272	Primary adhalinopathy: a common cause of autosomal recessive muscular dystrophy of variable severity. *Nature Genetics*, **1995**, 10, 243-5	36.3	170
271	Mendelian disorders deserve more attention. *Nature Reviews Genetics*, **2006**, 7, 277-82	30.1	168
270	Calpainopathy-a survey of mutations and polymorphisms. *American Journal of Human Genetics*, **1999**, 64, 1524-40	11	161
269	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
268	Mapping of a novel gene for familial hypertrophic cardiomyopathy to chromosome 11. *Nature Genetics*, **1993**, 4, 311-3	36.3	158
267	Juvenile limb-girdle muscular dystrophy. Clinical, histopathological and genetic data from a small community living in the Reunion Island. *Brain*, **1996**, 119 (Pt 1), 295-308	11.2	151
266	Intracellular stress signaling pathways activated during human islet preparation and following acute cytokine exposure. *Diabetes*, **2004**, 53, 2815-23	0.9	151
265	Loss of calpain 3 proteolytic activity leads to muscular dystrophy and to apoptosis-associated IkappaBalpha/nuclear factor kappaB pathway perturbation in mice. *Journal of Cell Biology*, **2000**, 151, 1583-90	7.3	147
264	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

263	Genome-wide association study identifies new HLA class II haplotypes strongly protective against narcolepsy. *Nature Genetics*, **2010**, 42, 786-9	36.3	145
262	Association of eGFR-Related Loci Identified by GWAS with Incident CKD and ESRD. *PLoS Genetics*, **2011**, 7, e1002292	6	144
261	Trait-based analyses for the detection of linkage between marker loci and quantitative trait loci in crosses between inbred lines. *Theoretical and Applied Genetics*, **1987**, 73, 556-62	6	144
260	Characterization of monoclonal antibodies to calpain 3 and protein expression in muscle from patients with limb-girdle muscular dystrophy type 2A. *American Journal of Pathology*, **1998**, 153, 1169-79	5.8	140
259	The Wnt receptor FZD1 mediates chemoresistance in neuroblastoma through activation of the Wnt/beta-catenin pathway. *Oncogene*, **2009**, 28, 2245-56	9.2	137
258	Restriction fragment length polymorphisms and genetic improvement of agricultural species. *Euphytica*, **1986**, 35, 111-124	2.1	137
257	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
256	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
255	A susceptibility locus for early-onset non-insulin dependent (type 2) diabetes mellitus maps to chromosome 20q, proximal to the phosphoenolpyruvate carboxykinase gene. *Human Molecular Genetics*, **1997**, 6, 1401-8	5.6	135
254	CATSPER2, a human autosomal nonsyndromic male infertility gene. *European Journal of Human Genetics*, **2003**, 11, 497-502	5.3	131
253	A homeobox gene, vax2, controls the patterning of the eye dorsoventral axis. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 10729-34	11.5	129
252	An alpha-tectorin gene defect causes a newly identified autosomal recessive form of sensorineural pre-lingual non-syndromic deafness, DFNB21. *Human Molecular Genetics*, **1999**, 8, 409-12	5.6	127
251	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
250	Congenital dyserythropoietic anemia type I is caused by mutations in codanin-1. *American Journal of Human Genetics*, **2002**, 71, 1467-74	11	124
249	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
248	Vocal cord and pharyngeal weakness with autosomal dominant distal myopathy: clinical description and gene localization to 5q31. *American Journal of Human Genetics*, **1998**, 63, 1732-42	11	120
247	Functional defects of a muscle-specific calpain, p94, caused by mutations associated with limb-girdle muscular dystrophy type 2A. *Journal of Biological Chemistry*, **1998**, 273, 17073-8	5.4	119
246	Linguistics of nucleotide sequences: morphology and comparison of vocabularies. *Journal of Biomolecular Structure and Dynamics*, **1986**, 4, 11-21	3.6	119

245	Genetic characterization of CHO production host DG44 and derivative recombinant cell lines. *Biochemical and Biophysical Research Communications*, **2006**, 340, 1069-77	3.4	117
244	Proteomic signatures: amino acid and oligopeptide compositions differentiate among phyla. *Proteins: Structure, Function and Bioinformatics*, **2004**, 54, 20-40	4.2	117
243	Autosomal dominant lateral temporal epilepsy: clinical and genetic study of a large Basque pedigree linked to chromosome 10q. *Annals of Neurology*, **1999**, 45, 182-8	9.4	117
242	Genome-wide prediction of matrix attachment regions that increase gene expression in mammalian cells. *Nature Methods*, **2007**, 4, 747-53	21.6	116
241	Genetic determinants of diastolic and pulse pressure map to different loci in Lyon hypertensive rats. *Nature Genetics*, **1993**, 3, 354-7	36.3	116
240	Toward a unified approach to genetic mapping of eukaryotes based on sequence tagged microsatellite sites. *Nature Biotechnology*, **1990**, 8, 930-2	44.5	114
239	The 16p11.2 locus modulates brain structures common to autism, schizophrenia and obesity. *Molecular Psychiatry*, **2015**, 20, 140-7	15.1	112
238	Genetic male infertility and mutation of CATSPER ion channels. *European Journal of Human Genetics* , **2010**, 18, 1178-84	5.3	110
237	Limb-girdle muscular dystrophy in Guip□coa (Basque Country, Spain). *Brain*, **1998**, 121 (Pt 9), 1735-47	11.2	110
236	Human-mouse differences in the embryonic expression patterns of developmental control genes and disease genes. *Human Molecular Genetics*, **2000**, 9, 165-73	5.6	107
235	Secondary reduction in calpain 3 expression in patients with limb girdle muscular dystrophy type 2B and Miyoshi myopathy (primary dysferlinopathies). *Neuromuscular Disorders*, **2000**, 10, 553-9	2.9	106
234	Expression and functional characteristics of calpain 3 isoforms generated through tissue-specific transcriptional and posttranscriptional events. *Molecular and Cellular Biology*, **1999**, 19, 4047-55	4.8	106
233	A gene for familial juvenile nephronophthisis (recessive medullary cystic kidney disease) maps to chromosome 2p. *Nature Genetics*, **1993**, 3, 342-5	36.3	105
232	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
231	In vitro transcription and processing of a yeast tRNA gene containing an intervening sequence. *Cell*, **1979**, 17, 399-406	56.2	104
230	Reconciling evidence-based medicine and precision medicine in the era of big data: challenges and opportunities. *Genome Medicine*, **2016**, 8, 134	14.4	103
229	Mapping a gene (SRN1) to chromosome 1q25-q31 in idiopathic nephrotic syndrome confirms a distinct entity of autosomal recessive nephrosis. *Human Molecular Genetics*, **1995**, 4, 2155-8	5.6	100
228	A modular approach for integrative analysis of large-scale gene-expression and drug-response data. *Nature Biotechnology*, **2008**, 26, 531-9	44.5	99

227	Calpain 3, the "gatekeeper" of proper sarcomere assembly, turnover and maintenance. *Neuromuscular Disorders*, **2008**, 18, 913-21	2.9	97
226	Genome-wide Association Studies Identify Genetic Loci Associated With Albuminuria in Diabetes. *Diabetes*, **2016**, 65, 803-17	0.9	96
225	Copy number variations and cognitive phenotypes in unselected populations. *JAMA - Journal of the American Medical Association*, **2015**, 313, 2044-54	27.4	96
224	Genetic linkage of the Muckle-Wells syndrome to chromosome 1q44. *American Journal of Human Genetics*, **1999**, 65, 1054-9	11	93
223	A founder mutation in the gamma-sarcoglycan gene of gypsies possibly predating their migration out of India. *Human Molecular Genetics*, **1996**, 5, 2019-22	5.6	91
222	Severe childhood autosomal recessive muscular dystrophy with the deficiency of the 50 kDa dystrophin-associated glycoprotein maps to chromosome 13q12. *Human Molecular Genetics*, **1993**, 2, 1423-8	5.6	88
221	Dimeric tRNA precursors in yeast. *Nature*, **1980**, 287, 750-2	50.4	85
220	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
219	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
218	Vacuoliting megalencephalic leukoencephalopathy with subcortical cysts, mapped to chromosome 22qtel. *American Journal of Human Genetics*, **2000**, 66, 733-9	11	82
217	Linkage of the gene for cystinosis to markers on the short arm of chromosome 17. The Cystinosis Collaborative Research Group. *Nature Genetics*, **1995**, 10, 246-8	36.3	82
216	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
215	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
214	Circadian regulation of islet genes involved in insulin production and secretion. *Molecular and Cellular Endocrinology*, **2004**, 226, 59-66	4.4	77
213	The region on 9p associated with 46,XY sex reversal contains several transcripts expressed in the urogenital system and a novel doublesex-related domain. *Genomics*, **2000**, 64, 170-8	4.3	77
212	Marker-based mapping of quantitative trait loci using replicated progenies. *Theoretical and Applied Genetics*, **1990**, 80, 205-8	6	75
211	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
210	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

209	Muscular dystrophy due to dysferlin deficiency in Libyan Jews. Clinical and genetic features. *Brain*, **2000**, 123 (Pt 6), 1229-37	11.2	71
208	Genome-wide association study of metabolic traits reveals novel gene-metabolite-disease links. *PLoS Genetics*, **2014**, 10, e1004132	6	70
207	A biochemical, genetic, and clinical survey of autosomal recessive limb girdle muscular dystrophies in Turkey. *Annals of Neurology*, **1997**, 42, 222-9	9.4	70
206	Cell-permeable peptides induce dose- and length-dependent cytotoxic effects. *Biochimica Et Biophysica Acta - Biomembranes*, **2007**, 1768, 2222-34	3.8	70
205	Hepatitis B virus-related insertional mutagenesis implicates SERCA1 gene in the control of apoptosis. *Oncogene*, **2000**, 19, 2877-86	9.2	70
204	Report of the DNA committee and catalogues of cloned and mapped genes, markers formatted for PCR and DNA polymorphisms. *Cytogenetic and Genome Research*, **1991**, 58, 1190-1832	1.9	69
203	Confirmation of the origin of NISCH syndrome. *Human Mutation*, **2006**, 27, 408-10	4.7	66
202	Chromosome 15-linked limb-girdle muscular dystrophy: clinical phenotypes in Reunion Island and French metropolitan communities. *Neuromuscular Disorders*, **1996**, 6, 447-53	2.9	65
201	DNA-Level Polymorphism as a Tool in Fisheries Science. *Canadian Journal of Fisheries and Aquatic Sciences*, **1988**, 45, 1075-1087	2.4	65
200	Physiology and genetics of carbamoylphosphate synthesis in Escherichia coli K12. *Molecular Genetics and Genomics*, **1974**, 133, 299-316		65
199	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
198	Detection of linkage between marker loci and loci affecting quantitative traits in crosses between segregating populations. *Theoretical and Applied Genetics*, **1988**, 76, 228-36	6	64
197	Familial hypertrophic cardiomyopathy. Microsatellite haplotyping and identification of a hot spot for mutations in the beta-myosin heavy chain gene. *Journal of Clinical Investigation*, **1993**, 92, 2807-13	15.9	63
196	The gene for the TATA binding protein (TBP) that contains a highly polymorphic protein coding CAG repeat maps to 6q27. *Genomics*, **1994**, 21, 667-8	4.3	62
195	Pharmacogenetics of glatiramer acetate therapy for multiple sclerosis reveals drug-response markers. *Pharmacogenetics and Genomics*, **2007**, 17, 657-66	1.9	61
194	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
193	In vitro whole-genome analysis identifies a susceptibility locus for HIV-1. *PLoS Biology*, **2008**, 6, e32	9.7	59
192	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

191	A harmonized meta-knowledgebase of clinical interpretations of somatic genomic variants in cancer. *Nature Genetics*, **2020**, 52, 448-457	36.3	58
190	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
189	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
188	A new dimension for the human genome project: towards comprehensive expression maps. *Nature Genetics*, **1997**, 16, 126-32	36.3	57
187	How neutral are synonymous codon mutations?. *Nature Genetics*, **1995**, 10, 259	36.3	57
186	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
185	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
184	DNA segments mapped by reciprocal use of microsatellite primers between mouse and rat. *Mammalian Genome*, **1993**, 4, 571-6	3.2	54
183	Large restriction fragments containing poly-TG are highly polymorphic in a variety of vertebrates. *Nucleic Acids Research*, **1990**, 18, 1129-32	20.1	53
182	The phenotype of recurrent 10q22q23 deletions and duplications. *European Journal of Human Genetics*, **2011**, 19, 400-8	5.3	52
181	Recommendations for locus-specific databases and their curation. *Human Mutation*, **2008**, 29, 2-5	4.7	52
180	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
179	Expression of genes (CAPN3, SGCA, SGCB, and TTN) involved in progressive muscular dystrophies during early human development. *Genomics*, **1998**, 48, 145-56	4.3	52
178	Cardiac myosin binding protein C gene is specifically expressed in heart during murine and human development. *Circulation Research*, **1998**, 82, 130-3	15.7	51
177	A linkage map of human chromosome 15 with an average resolution of 2 cM and containing 55 polymorphic microsatellites. *Human Molecular Genetics*, **1993**, 2, 2019-30	5.6	51
176	Association between C-reactive protein and adiposity in women. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 3969-77	5.6	50
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	Pathophysiology of limb girdle muscular dystrophy type 2A: hypothesis and new insights into the IkappaBalpha/NF-kappaB survival pathway in skeletal muscle. *Journal of Molecular Medicine*, **2001**, 79, 254-61	5.5	50

173	Mutations in calpain 3 associated with limb girdle muscular dystrophy: analysis by molecular modeling and by mutation in m-calpain. *Biophysical Journal*, **2001**, 80, 2590-6	2.9	50
172	Mapping of a chromosome 15 region involved in limb girdle muscular dystrophy. *Human Molecular Genetics*, **1994**, 3, 285-93	5.6	50
171	Physical mapping of plastid DNA variation among eleven Nicotiana species. *Theoretical and Applied Genetics*, **1984**, 69, 1-14	6	50
170	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
169	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. *Human Molecular Genetics*, **2014**, 23, 6069-80	5.6	45
168	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
167	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
166	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
165	Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. *Molecular Psychiatry*, **2017**, 22, 836-849	15.1	43
164	G-418, an elongation inhibitor of 80 S ribosomes. *Biochimica Et Biophysica Acta Gene Regulatory Mechanisms*, **1983**, 741, 123-7		42
163	A new locus for autosomal dominant dilated cardiomyopathy identified on chromosome 6q12-q16. *American Journal of Human Genetics*, **2001**, 68, 241-6	11	41
162	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
161	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
160	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
159	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
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