Andr Reis

List of Publications by Citations

440 papers	26,390 citations	81 h-index	146 g-index
474 ext. papers	30,226 ext. citations	7.9 avg, IF	5.94 L-index

#	Paper	IF	Citations
440	Nibrin, a novel DNA double-strand break repair protein, is mutated in Nijmegen breakage syndrome. *Cell*, **1998**, 93, 467-76	56.2	880
439	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel beta1 subunit gene SCN1B. *Nature Genetics*, **1998**, 19, 366-70	36.3	851
438	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
437	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. *Lancet, The*, **2012**, 380, 1674-82	40	765
436	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
435	Genome-wide, large-scale production of mutant mice by ENU mutagenesis. *Nature Genetics*, **2000**, 25, 444-7	36.3	578
434	Psoriasis is associated with increased beta-defensin genomic copy number. *Nature Genetics*, **2008**, 40, 23-5	36.3	519
433	CNTNAP2 and NRXN1 are mutated in autosomal-recessive Pitt-Hopkins-like mental retardation and determine the level of a common synaptic protein in Drosophila. *American Journal of Human Genetics*, **2009**, 85, 655-66	11	485
432	Transcription factor E2-2 is an essential and specific regulator of plasmacytoid dendritic cell development. *Cell*, **2008**, 135, 37-48	56.2	477
431	Human laminin beta2 deficiency causes congenital nephrosis with mesangial sclerosis and distinct eye abnormalities. *Human Molecular Genetics*, **2004**, 13, 2625-32	5.6	395
430	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. *Nature Genetics*, **2010**, 42, 1021-6	36.3	347
429	Mutations in the pericentrin (PCNT) gene cause primordial dwarfism. *Science*, **2008**, 319, 816-9	33.3	325
428	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
427	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. *Nature Genetics*, **2010**, 42, 996-9	36.3	294
426	A genome-wide search for linkage to asthma. German Asthma Genetics Group. *Genomics*, **1999**, 58, 1-8	4.3	291
425	A comprehensive linkage analysis for myocardial infarction and its related risk factors. *Nature Genetics*, **2002**, 30, 210-4	36.3	284
424	The origin of the major cystic fibrosis mutation (delta F508) in European populations. *Nature Genetics*, **1994**, 7, 169-75	36.3	284

423	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2063-74	2.5	281
422	Mutations in STRA6 cause a broad spectrum of malformations including anophthalmia, congenital heart defects, diaphragmatic hernia, alveolar capillary dysplasia, lung hypoplasia, and mental retardation. *American Journal of Human Genetics*, **2007**, 80, 550-60	11	281
421	Mutations in microcephalin cause aberrant regulation of chromosome condensation. *American Journal of Human Genetics*, **2004**, 75, 261-6	11	274
420	Mutations in the gene encoding the lamin B receptor produce an altered nuclear morphology in granulocytes (Pelger-Hu anomaly). *Nature Genetics*, **2002**, 31, 410-4	36.3	272
419	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases: A Mendelian Randomization Study. *JAMA Oncology*, **2017**, 3, 636-651	13.4	236
418	Keratin 9 gene mutations in epidermolytic palmoplantar keratoderma (EPPK). *Nature Genetics*, **1994**, 6, 174-9	36.3	230
417	Haploinsufficiency of TCF4 causes syndromal mental retardation with intermittent hyperventilation (Pitt-Hopkins syndrome). *American Journal of Human Genetics*, **2007**, 80, 994-1001	11	225
416	A major susceptibility locus for atopic dermatitis maps to chromosome 3q21. *Nature Genetics*, **2000**, 26, 470-3	36.3	222
415	Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). *Nature Genetics*, **2005**, 37, 1345-50	36.3	211
414	Genotype-phenotype correlations in Noonan syndrome. *Journal of Pediatrics*, **2004**, 144, 368-74	3.6	197
413	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. *American Journal of Human Genetics*, **2015**, 97, 816-36	11	185
412	Haploinsufficiency of ARID1B, a member of the SWI/SNF-a chromatin-remodeling complex, is a frequent cause of intellectual disability. *American Journal of Human Genetics*, **2012**, 90, 565-72	11	182
411	CLP1 founder mutation links tRNA splicing and maturation to cerebellar development and neurodegeneration. *Cell*, **2014**, 157, 651-63	56.2	174
410	Mutations in CAV3 cause mechanical hyperirritability of skeletal muscle in rippling muscle disease. *Nature Genetics*, **2001**, 28, 218-9	36.3	173
409	Splitting Schizophrenia: Periodic Catatonia Susceptibility Locus on Chromosome 15q15. *American Journal of Human Genetics*, **2000**, 67, 1201-1207	11	172
408	Dysferlin deletion in SJL mice (SJL-Dysf) defines a natural model for limb girdle muscular dystrophy 2B. *Nature Genetics*, **1999**, 23, 141-2	36.3	169
407	Imprinting mutations suggested by abnormal DNA methylation patterns in familial Angelman and Prader-Willi syndromes. *American Journal of Human Genetics*, **1994**, 54, 741-7	11	168
406	Human TBX1 missense mutations cause gain of function resulting in the same phenotype as 22q11.2 deletions. *American Journal of Human Genetics*, **2007**, 80, 510-7	11	166

405	Adaptor protein complex 4 deficiency causes severe autosomal-recessive intellectual disability, progressive spastic paraplegia, shy character, and short stature. *American Journal of Human Genetics*, **2011**, 88, 788-795	11	160
404	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. *American Journal of Human Genetics*, **2016**, 98, 149-64	11	154
403	Identification of ZNF313/RNF114 as a novel psoriasis susceptibility gene. *Human Molecular Genetics* , **2008**, 17, 1938-45	5.6	151
402	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. *Human Molecular Genetics*, **2013**, 22, 5121-35	5.6	138
401	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. *Nature Communications*, **2017**, 8, 15382	17.4	136
400	Common genetic variants associated with open-angle glaucoma. *Human Molecular Genetics*, **2011**, 20, 2464-71	5.6	134
399	Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. *Human Mutation*, **2010**, 31, 722-33	4.7	134
398	NEK1 mutations cause short-rib polydactyly syndrome type majewski. *American Journal of Human Genetics*, **2011**, 88, 106-14	11	131
397	Common genetic determinants of intraocular pressure and primary open-angle glaucoma. *PLoS Genetics*, **2012**, 8, e1002611	6	131
396	Sporadic imprinting defects in Prader-Willi syndrome and Angelman syndrome: implications for imprint-switch models, genetic counseling, and prenatal diagnosis. *American Journal of Human Genetics*, **1998**, 63, 170-80	11	128
395	Genome search for susceptibility loci of common idiopathic generalised epilepsies. *Human Molecular Genetics*, **2000**, 9, 1465-72	5.6	126
394	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. *American Journal of Human Genetics*, **2015**, 97, 886-93	11	125
393	Homozygosity mapping in families with Joubert syndrome identifies a locus on chromosome 9q34.3 and evidence for genetic heterogeneity. *American Journal of Human Genetics*, **1999**, 65, 1666-71	11	124
392	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. *Nature Communications*, **2015**, 6, 7001	17.4	122
391	Heterozygous NTF4 mutations impairing neurotrophin-4 signaling in patients with primary open-angle glaucoma. *American Journal of Human Genetics*, **2009**, 85, 447-56	11	120
390	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. *JAMA Psychiatry*, **2017**, 74, 293-299	14.5	116
389	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. *Nature Genetics*, **2017**, 49, 223-237	36.3	116
388	Genetic regulation of serum phytosterol levels and risk of coronary artery disease. *Circulation: Cardiovascular Genetics*, **2010**, 3, 331-9		116

387	Genomewide scan in german families reveals evidence for a novel psoriasis-susceptibility locus on chromosome 19p13. *American Journal of Human Genetics*, **2000**, 67, 1020-4	11	112
386	Synuclein oligomers induce early axonal dysfunction in human iPSC-based models of synucleinopathies. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 7813-7818	11.5	111
385	Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations. *European Journal of Human Genetics*, **2000**, 8, 900-2	5.3	109
384	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. *Nature Communications*, **2015**, 6, 6046	17.4	103
383	Clinical and mutational spectrum of Mowat-Wilson syndrome. *European Journal of Medical Genetics*, **2005**, 48, 97-111	2.6	103
382	Mowat-Wilson syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomalies-mental retardation syndrome caused by mutations in the zinc finger homeo box 1B gene. *American Journal of Medical Genetics Part A*, **2002**, 108, 177-181		103
381	Association of LOXL1 common sequence variants in German and Italian patients with pseudoexfoliation syndrome and pseudoexfoliation glaucoma. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 1459-63		102
380	Severely incapacitating mutations in patients with extreme short stature identify RNA-processing endoribonuclease RMRP as an essential cell growth regulator. *American Journal of Human Genetics*, **2005**, 77, 795-806	11	102
379	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. *Journal of Medical Genetics*, **2010**, 47, 321-31	5.8	101
378	Linkage of the gene for the triple A syndrome to chromosome 12q13 near the type II keratin gene cluster. *Human Molecular Genetics*, **1996**, 5, 2061-6	5.6	101
377	Genotype-correlated expression of lysyl oxidase-like 1 in ocular tissues of patients with pseudoexfoliation syndrome/glaucoma and normal patients. *American Journal of Pathology*, **2008**, 173, 1724-35	5.8	98
376	First known microdeletion within the Wolf-Hirschhorn syndrome critical region refines genotype-phenotype correlation. *American Journal of Medical Genetics Part A*, **2001**, 99, 338-42		98
375	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. *BMC Medical Genetics*, **2011**, 12, 106	2.1	97
374	A genome wide search for susceptibility loci in three European malignant hyperthermia pedigrees. *Human Molecular Genetics*, **1997**, 6, 953-61	5.6	96
373	Congenital nephrosis, mesangial sclerosis, and distinct eye abnormalities with microcoria: an autosomal recessive syndrome **2004**, 130A, 138-45		95
372	Disease burden and risk profile in referred patients with moderate chronic kidney disease: composition of the German Chronic Kidney Disease (GCKD) cohort. *Nephrology Dialysis Transplantation*, **2015**, 30, 441-51	4.3	93
371	Molecular karyotyping using an SNP array for genomewide genotyping. *Journal of Medical Genetics*, **2004**, 41, 916-22	5.8	93
370	Identification of a new gene locus for adolescent nephronophthisis, on chromosome 3q22 in a large Venezuelan pedigree. *American Journal of Human Genetics*, **2000**, 66, 118-27	11	93

369	Genome-wide association study identifies a new locus for coronary artery disease on chromosome 10p11.23. *European Heart Journal*, **2011**, 32, 158-68	9.5	92
368	De novo mutations in the genome organizer CTCF cause intellectual disability. *American Journal of Human Genetics*, **2013**, 93, 124-31	11	89
367	The German Chronic Kidney Disease (GCKD) study: design and methods. *Nephrology Dialysis Transplantation*, **2012**, 27, 1454-60	4.3	89
366	Spectrum of mutations and genotype-phenotype analysis in Currarino syndrome. *European Journal of Human Genetics*, **2001**, 9, 599-605	5.3	87
365	Genetic variants of the IL-23R pathway: association with psoriatic arthritis and psoriasis vulgaris, but no specific risk factor for arthritis. *Journal of Investigative Dermatology*, **2009**, 129, 355-8	4.3	86
364	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. *Journal of Medical Genetics*, **2007**, 44, 651-6	5.8	85
363	Mutations in the gene encoding the Wnt-signaling component R-spondin 4 (RSPO4) cause autosomal recessive anonychia. *American Journal of Human Genetics*, **2006**, 79, 1105-9	11	82
362	Splitting schizophrenia: periodic catatonia-susceptibility locus on chromosome 15q15. *American Journal of Human Genetics*, **2000**, 67, 1201-7	11	81
361	Environmental trichlorfon and cluster of congenital abnormalities. *Lancet, The*, **1993**, 341, 539-42	40	81
360	A unique form of autosomal dominant cataract explained by gene conversion between beta-crystallin B2 and its pseudogene. *Journal of Medical Genetics*, **2001**, 38, 392-6	5.8	81
359	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
358	Hypomorphic mutations in PGAP2, encoding a GPI-anchor-remodeling protein, cause autosomal-recessive intellectual disability. *American Journal of Human Genetics*, **2013**, 92, 575-83	11	79
357	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
356	Familial hypomagnesaemia with hypercalciuria and nephrocalcinosis maps to chromosome 3q27 and is associated with mutations in the PCLN-1 gene. *European Journal of Human Genetics*, **2000**, 8, 414-22	5.3	77
355	TNF polymorphisms in psoriasis: association of psoriatic arthritis with the promoter polymorphism TNF*-857 independent of the PSORS1 risk allele. *Arthritis and Rheumatism*, **2007**, 56, 2056-64		76
354	Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. *Journal of Medical Genetics*, **2016**, 53, 511-22	5.8	76
353	Limb mammary syndrome: a new genetic disorder with mammary hypoplasia, ectrodactyly, and other Hand/Foot anomalies maps to human chromosome 3q27. *American Journal of Human Genetics*, **1999**, 64, 538-46	11	75
352	Mutations in the Nijmegen Breakage Syndrome gene (NBS1) in childhood acute lymphoblastic leukemia (ALL). *Cancer Research*, **2001**, 61, 3570-2	10.1	74

351	Self-healing collodion baby: a dynamic phenotype explained by a particular transglutaminase-1 mutation. *Journal of Investigative Dermatology*, **2003**, 120, 224-8	4.3	73
350	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. *Nature Genetics*, **2017**, 49, 993-1004	36.3	72
349	Diaphragmatic spinal muscular atrophy with respiratory distress is heterogeneous, and one form Is linked to chromosome 11q13-q21. *American Journal of Human Genetics*, **1999**, 65, 1459-62	11	72
348	The gene for the ataxia-telangiectasia variant, Nijmegen breakage syndrome, maps to a 1-cM interval on chromosome 8q21. *American Journal of Human Genetics*, **1997**, 60, 605-10	11	72
347	Gene panel sequencing in familial breast/ovarian cancer patients identifies multiple novel mutations also in genes others than BRCA1/2. *International Journal of Cancer*, **2017**, 140, 95-102	7.5	71
346	Gene localization for an autosomal dominant familial periodic fever to 12p13. *American Journal of Human Genetics*, **1998**, 62, 884-9	11	71
345	Hereditary spastic paraplegia caused by mutations in the SPG4 gene. *European Journal of Human Genetics*, **2000**, 8, 771-6	5.3	71
344	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. *Nature Genetics*, **2015**, 47, 387-92	36.3	70
343	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. *Genetics in Medicine*, **2018**, 20, 630-638	8.1	68
342	Variants in ASB10 are associated with open-angle glaucoma. *Human Molecular Genetics*, **2012**, 21, 1336-49	5.6	68
341	Genotype/phenotype correlation in autosomal recessive lamellar ichthyosis. *American Journal of Human Genetics*, **1998**, 62, 1052-61	11	68
340	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndrome-like phenotype and hyperactivated MAPK signaling in humans and mice. *Journal of Clinical Investigation*, **2011**, 121, 3479-91	15.9	68
339	Genome scan for childhood and adolescent obesity in German families. *Pediatrics*, **2003**, 111, 321-7	7.4	67
338	Localisation of gene for the naevoid basal-cell carcinoma syndrome. *Lancet, The*, **1992**, 339, 617	40	67
337	Homozygosity mapping in 64 Syrian consanguineous families with non-specific intellectual disability reveals 11 novel loci and high heterogeneity. *European Journal of Human Genetics*, **2011**, 19, 1161-6	5.3	66
336	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. *Journal of Medical Genetics*, **2007**, 44, 629-36	5.8	66
335	A novel in situ method for the detection of deficient transglutaminase activity in the skin. *Archives of Dermatological Research*, **1998**, 290, 621-7	3.3	64
334	Type and level of RMRP functional impairment predicts phenotype in the cartilage hair hypoplasia-anauxetic dysplasia spectrum. *American Journal of Human Genetics*, **2007**, 81, 519-29	11	64

333	Nijmegen breakage syndrome: consequences of defective DNA double strand break repair. *BioEssays*, **1999**, 21, 649-56	4.1	64
332	Mapping of a gene for epidermolytic palmoplantar keratoderma to the region of the acidic keratin gene cluster at 17q12-q21. *Human Genetics*, **1992**, 90, 113-6	6.3	64
331	Null mutation in PGAP1 impairing Gpi-anchor maturation in patients with intellectual disability and encephalopathy. *PLoS Genetics*, **2014**, 10, e1004320	6	63
330	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. *Human Genetics*, **2015**, 134, 97-109	6.3	62
329	Genetic variants and cellular stressors associated with exfoliation syndrome modulate promoter activity of a lncRNA within the LOXL1 locus. *Human Molecular Genetics*, **2015**, 24, 6552-63	5.6	61
328	Renal fibrosis is the common feature of autosomal dominant tubulointerstitial kidney diseases caused by mutations in mucin 1 or uromodulin. *Kidney International*, **2014**, 86, 589-99	9.9	60
327	A second locus for an axonal form of autosomal recessive Charcot-Marie-Tooth disease maps to chromosome 19q13.3. *American Journal of Human Genetics*, **2001**, 68, 269-74	11	60
326	The clinical significance of small copy number variants in neurodevelopmental disorders. *Journal of Medical Genetics*, **2014**, 51, 677-88	5.8	59
325	Identification of the variant Ala335Val of MED25 as responsible for CMT2B2: molecular data, functional studies of the SH3 recognition motif and correlation between wild-type MED25 and PMP22 RNA levels in CMT1A animal models. *Neurogenetics*, **2009**, 10, 275-87	3	59
324	Disturbed Wnt Signalling due to a Mutation in CCDC88C Causes an Autosomal Recessive Non-Syndromic Hydrocephalus with Medial Diverticulum. *Molecular Syndromology*, **2010**, 1, 99-112	1.5	59
323	Localisation of a gene for Papillon-Lefvre syndrome to chromosome 11q14-q21 by homozygosity mapping. *Human Genetics*, **1997**, 101, 376-82	6.3	59
322	Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome. *American Journal of Human Genetics*, **2004**, 75, 138-45	11	59
321	Localization of a locus for the striated form of palmoplantar keratoderma to chromosome 18q near the desmosomal cadherin gene cluster. *Human Molecular Genetics*, **1995**, 4, 1015-20	5.6	59
320	Different mechanisms and recurrence risks of imprinting defects in Angelman syndrome. *American Journal of Human Genetics*, **1997**, 61, 88-93	11	58
319	Mutations in CYP1B1 cause primary congenital glaucoma by reduction of either activity or abundance of the enzyme. *Human Mutation*, **2008**, 29, 1147-53	4.7	58
318	Mutation in the betaA3/A1-crystallin encoding gene Cryba1 causes a dominant cataract in the mouse. *Genomics*, **1999**, 62, 67-73	4.3	58
317	Variants in RUNX3 contribute to susceptibility to psoriatic arthritis, exhibiting further common ground with ankylosing spondylitis. *Arthritis and Rheumatism*, **2013**, 65, 1224-31		56
316	Investigation of a family with autosomal dominant dilated cardiomyopathy defines a novel locus on chromosome 2q14-q22. *American Journal of Human Genetics*, **1999**, 65, 1068-77	11	56

315	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. *European Journal of Human Genetics*, **2015**, 23, 602-9	5.3	55
314	Prevalence and correlates of gout in a large cohort of patients with chronic kidney disease: the German Chronic Kidney Disease (GCKD) study. *Nephrology Dialysis Transplantation*, **2015**, 30, 613-21	4.3	55
313	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. *European Journal of Human Genetics*, **2015**, 23, 753-60	5.3	55
312	A new quantitative PCR multiplex assay for rapid analysis of chromosome 17p11.2-12 duplications and deletions leading to HMSN/HNPP. *European Journal of Human Genetics*, **2003**, 11, 170-8	5.3	55
311	Profiling of WDR36 missense variants in German patients with glaucoma. *Investigative Ophthalmology and Visual Science*, **2008**, 49, 270-4		54
310	Mal de Meleda (MDM) caused by mutations in the gene for SLURP-1 in patients from Germany, Turkey, Palestine, and the United Arab Emirates. *Human Genetics*, **2003**, 112, 50-6	6.3	53
309	A dual phenotype of periventricular nodular heterotopia and frontometaphyseal dysplasia in one patient caused by a single FLNA mutation leading to two functionally different aberrant transcripts. *American Journal of Human Genetics*, **2004**, 74, 731-7	11	53
308	Exome Pool-Seq in neurodevelopmental disorders. *European Journal of Human Genetics*, **2017**, 25, 1364-1376	5.3	52
307	Replication of LCE3C-LCE3B CNV as a risk factor for psoriasis and analysis of interaction with other genetic risk factors. *Journal of Investigative Dermatology*, **2010**, 130, 979-84	4.3	52
306	Mutation in the Scyl1 gene encoding amino-terminal kinase-like protein causes a recessive form of spinocerebellar neurodegeneration. *EMBO Reports*, **2007**, 8, 691-7	6.5	52
305	Chromatin-Remodeling-Factor ARID1B Represses Wnt/β-Catenin Signaling. *American Journal of Human Genetics*, **2015**, 97, 445-56	11	51
304	Mutations affecting the SAND domain of DEAF1 cause intellectual disability with severe speech impairment and behavioral problems. *American Journal of Human Genetics*, **2014**, 94, 649-61	11	51
303	Fine mapping and single nucleotide polymorphism association results of candidate genes for asthma and related phenotypes. *Human Mutation*, **2001**, 18, 327-36	4.7	51
302	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. *American Journal of Human Genetics*, **2018**, 103, 1045-1052	11	51
301	Rare copy number variants are a common cause of short stature. *PLoS Genetics*, **2013**, 9, e1003365	6	50
300	Epigenetic targeting in the mouse zygote marks DNA for later methylation: a mechanism for maternal effects in development. *Mechanisms of Development*, **2001**, 103, 35-47	1.7	50
299	Keratin 9 gene mutational heterogeneity in patients with epidermolytic palmoplantar keratoderma. *Human Genetics*, **1994**, 93, 649-54	6.3	50
298	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. *Human Mutation*, **2016**, 37, 755-64	4.7	49

297	PTPN22 is associated with susceptibility to psoriatic arthritis but not psoriasis: evidence for a further PsA-specific risk locus. *Annals of the Rheumatic Diseases*, **2015**, 74, 1882-5	2.4	49
296	Heterozygous loss-of-function variants in CYP1B1 predispose to primary open-angle glaucoma **2010**, 51, 249-54		49
295	Mutations in MBOAT7, Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. *American Journal of Human Genetics*, **2016**, 99, 912-916	11	48
294	Genome-wide association study with DNA pooling identifies variants at CNTNAP2 associated with pseudoexfoliation syndrome. *European Journal of Human Genetics*, **2011**, 19, 186-93	5.3	47
293	Mutations in the pyruvate kinase L gene in patients with hereditary hemolytic anemia. *Blood*, **1994**, 83, 2817-2822	2.2	47
292	Periodic catatonia: confirmation of linkage to chromosome 15 and further evidence for genetic heterogeneity. *Human Genetics*, **2002**, 111, 323-30	6.3	46
291	Characterization of a mutation in the lens-specific MP70 encoding gene of the mouse leading to a dominant cataract. *Experimental Eye Research*, **2001**, 73, 867-76	3.7	46
290	Cloning of the mouse dysferlin gene and genomic characterization of the SJL-Dysf mutation. *NeuroReport*, **2001**, 12, 625-9	1.7	46
289	In-Frame Deletion and Missense Mutations of the C-Terminal Helicase Domain of SMARCA2 in Three Patients with Nicolaides-Baraitser Syndrome. *Molecular Syndromology*, **2012**, 2, 237-244	1.5	45
288	Association of □defensin copy number and psoriasis in three cohorts of European origin. *Journal of Investigative Dermatology*, **2012**, 132, 2407-2413	4.3	45
287	Neurodevelopmental deficits in Pierson (microcoria-congenital nephrosis) syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143, 311-9	2.5	45
286	Hereditary isolated renal magnesium loss maps to chromosome 11q23. *American Journal of Human Genetics*, **1999**, 64, 180-8	11	45
285	Exclusion of the GABAA-receptor beta 3 subunit gene as the Angelman's syndrome gene. *Lancet, The*, **1993**, 341, 122-3	40	45
284	Palmoplantar keratoderma in association with carcinoma of the esophagus maps to chromosome 17q distal to the keratin gene cluster. *Genomics*, **1995**, 29, 537-40	4.3	44
283	FOXP2 variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. *Journal of Medical Genetics*, **2017**, 54, 64-72	5.8	43
282	Familial interstitial 570 kbp deletion of the UBE3A gene region causing Angelman syndrome but not Prader-Willi syndrome. *American Journal of Medical Genetics Part A*, **2002**, 111, 233-7		43
281	A European study on the genetics of mite sensitization. *Journal of Allergy and Clinical Immunology*, **2000**, 106, 925-32	11.5	43
280	The Fanconi anemia group E gene, FANCE, maps to chromosome 6p. *American Journal of Human Genetics*, **1999**, 64, 1400-5	11	43

279	A gene for hypotrichosis simplex of the scalp maps to chromosome 6p21.3. *American Journal of Human Genetics*, **2000**, 66, 1979-83	11	42
278	□Synuclein-induced myelination deficit defines a novel interventional target for multiple system atrophy. *Acta Neuropathologica*, **2016**, 132, 59-75	14.3	42
277	Genome-wide linkage reveals a locus for human essential (primary) hypertension on chromosome 12p. *Human Molecular Genetics*, **2003**, 12, 1273-7	5.6	41
276	Localization of a gene for syndactyly type 1 to chromosome 2q34-q36. *American Journal of Human Genetics*, **2000**, 67, 492-7	11	41
275	BRCA mutations and their influence on pathological complete response and prognosis in a clinical cohort of neoadjuvantly treated breast cancer patients. *Breast Cancer Research and Treatment*, **2018**, 171, 85-94	4.4	40
274	Exploring functional candidate genes for genetic association in german patients with pseudoexfoliation syndrome and pseudoexfoliation glaucoma **2009**, 50, 2796-801		40
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