AndrÃ© Reis

List of Publications by Year
in descending order

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447 papers	33,138 citations	4146 87 h-index	6131 159 g-index
474 all docs	474 docs citations	474 times ranked	38927 citing authors

#	Article	IF	Citations
1	Nibrin, a Novel DNA Double-Strand Break Repair Protein, Is Mutated in Nijmegen Breakage Syndrome. Cell, 1998, 93, 467-476.	28.9	989
2	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel ÃŸ1 subunit gene SCN1B. Nature Genetics, 1998, 19, 366-370.	21.4	965
3	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940
4	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	21.4	918
5	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	21.4	848
6	Genome-wide, large-scale production of mutant mice by ENU mutagenesis. Nature Genetics, 2000, 25, 444-447.	21.4	658
7	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	21.4	587
8	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, 655-666.	6.2	573
9	Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. Cell, 2008, 135, 37-48.	28.9	567
10	Human laminin Î²2 deficiency causes congenital nephrosis with mesangial sclerosis and distinct eye abnormalities. Human Molecular Genetics, 2004, 13, 2625-2632.	2.9	443
11	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
12	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
13	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. JAMA Oncology, 2017, 3, 636.	7.1	376
14	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	12.6	370
15	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 2063-2074.	1.2	343
16	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. Nature Genetics, 2010, 42, 996-999.	21.4	334
17	A Genome-wide Search for Linkage to Asthma22See the Appendix.. Genomics, 1999, 58, 1-8.	2.9	332
18	The origin of the major cystic fibrosis mutation (Î”F508) in European populations. Nature Genetics, 1994, 7, 169-175.	21.4	323

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19	Mutations in the gene encoding the lamin B receptor produce an altered nuclear morphology in granulocytes (Pelgerâ€“HuÃ«t anomaly). Nature Genetics, 2002, 31, 410-414.	21.4	316
20	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, 550-560.	6.2	316
21	A comprehensive linkage analysis for myocardial infarction and its related risk factors. Nature Genetics, 2002, 30, 210-214.	21.4	313
22	Mutations in Microcephalin Cause Aberrant Regulation of Chromosome Condensation. American Journal of Human Genetics, 2004, 75, 261-266.	6.2	292
23	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. American Journal of Human Genetics, 2016, 98, 149-164.	6.2	270
24	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001.	6.2	261
25	Keratin 9 gene mutations in epidermolytic palmoplantar keratoderma (EPPK). Nature Genetics, 1994, 6, 174-179.	21.4	255
26	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, 1345-1350.	21.4	252
27	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. Nature Communications, 2017, 8, 15382.	12.8	251
28	A major susceptibility locus for atopic dermatitis maps to chromosome 3q21. Nature Genetics, 2000, 26, 470-473.	21.4	249
29	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. American Journal of Human Genetics, 2015, 97, 816-836.	6.2	245
30	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
31	Genotype-phenotype correlations in Noonan syndrome. Journal of Pediatrics, 2004, 144, 368-374.	1.8	227
32	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, 565-572.	6.2	225
33	Mutations in CAV3 cause mechanical hyperirritability of skeletal muscle in rippling muscle disease. Nature Genetics, 2001, 28, 218-219.	21.4	206
34	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.	6.2	206
35	Human TBX1 Missense Mutations Cause Gain of Function Resulting in the Same Phenotype as 22q11.2 Deletions. American Journal of Human Genetics, 2007, 80, 510-517.	6.2	195
36	Splitting Schizophrenia: Periodic Catatoniaâ€“Susceptibility Locus on Chromosome 15q15. American Journal of Human Genetics, 2000, 67, 1201-1207.	6.2	192

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37	Dysferlin deletion in SJL mice (SJL-Dysf) defines a natural model for limb girdle muscular dystrophy 2B. Nature Genetics, 1999, 23, 141-142.	21.4	191
38	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	2.9	190
39	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	11.0	186
40	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
41	Imprinting mutations suggested by abnormal DNA methylation patterns in familial Angelman and Prader-Willi syndromes. American Journal of Human Genetics, 1994, 54, 741-7.	6.2	179
42	Identification of ZNF313 / RNF114 as a novel psoriasis susceptibility gene. Human Molecular Genetics, 2008, 17, 1938-1945.	2.9	176
43	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, 886-893.	6.2	171
44	Î±-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.	7.1	168
45	Common Genetic Determinants of Intraocular Pressure and Primary Open-Angle Glaucoma. PLoS Genetics, 2012, 8, e1002611.	3.5	164
46	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, 722-733.	2.5	163
47	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. Nature Communications, 2015, 6, 7001.	12.8	156
48	Common genetic variants associated with open-angle glaucoma. Human Molecular Genetics, 2011, 20, 2464-2471.	2.9	152
49	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	6.2	151
50	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	6.2	151
51	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. Nature Communications, 2015, 6, 6046.	12.8	149
52	Genome search for susceptibility loci of common idiopathic generalised epilepsies. Human Molecular Genetics, 2000, 9, 1465-1472.	2.9	147
53	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, 170-180.	6.2	142
54	Genetic Regulation of Serum Phytosterol Levels and Risk of Coronary Artery Disease. Circulation: Cardiovascular Genetics, 2010, 3, 331-339.	5.1	141

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55	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, 1666-1671.	6.2	135
56	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	3.2	135
57	Heterozygous NTF4 Mutations Impairing Neurotrophin-4 Signaling in Patients with Primary Open-Angle Glaucoma. American Journal of Human Genetics, 2009, 85, 447-456.	6.2	134
58	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
59	Linkage of the gene for the triple A syndrome to chromosome 12q13 near the type II keratin gene cluster. Human Molecular Genetics, 1996, 5, 2061-2066.	2.9	132
60	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, 441-451.	0.7	132
61	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, 900-902.	2.8	130
62	Genomewide Scan in German Families Reveals Evidence for a Novel Psoriasis-Susceptibility Locus on Chromosome 19p13. American Journal of Human Genetics, 2000, 67, 1020-1024.	6.2	129
63	First known microdeletion within the Wolf-Hirschhorn syndrome critical region refines genotype-phenotype correlation. American Journal of Medical Genetics Part A, 2001, 99, 338-342.	2.4	128
64	The German Chronic Kidney Disease (GCKD) study: design and methods. Nephrology Dialysis Transplantation, 2012, 27, 1454-1460.	0.7	127
65	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331.	3.2	126
66	Genome-wide association study identifies a new locus for coronary artery disease on chromosome 10p11.23. European Heart Journal, 2011, 32, 158-168.	2.2	124
67	â€œMowatâ€Wilsonâ€•syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomaliesâ€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.	2.4	122
68	Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111.	1.3	121
69	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, 1724-1735.	3.8	118
70	A Genome Wide Search for Susceptibility Loci in Three European Malignant Hyperthermia Pedigrees. Human Molecular Genetics, 1997, 6, 953-961.	2.9	117
71	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.	6.2	117
72	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. Journal of Medical Genetics, 2007, 44, 651-656.	3.2	114

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73	Association ofLOXL1Common Sequence Variants in German and Italian Patients with Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. , 2008, 49, 1459.		114
74	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. Nature Genetics, 2017, 49, 993-1004.	21.4	114
75	Splitting Schizophrenia: Periodic Catatoniaâ€“Susceptibility Locus on Chromosome 15q15. American Journal of Human Genetics, 2000, 67, 1201-1207.	6.2	112
76	Congenital nephrosis, mesangial sclerosis, and distinct eye abnormalities with microcoria: An autosomal recessive syndrome. American Journal of Medical Genetics, Part A, 2004, 130A, 138-145.	1.2	110
77	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106.	2.1	109
78	Molecular karyotyping using an SNP array for genomewide genotyping. Journal of Medical Genetics, 2004, 41, 916-922.	3.2	106
79	Identification of a New Gene Locus for Adolescent Nephronophthisis, on Chromosome 3q22 in a Large Venezuelan Pedigree. American Journal of Human Genetics, 2000, 66, 118-127.	6.2	105
80	Self-Healing Collodion Baby: a Dynamic Phenotype Explained by a Particular Transglutaminase-1 Mutation. Journal of Investigative Dermatology, 2003, 120, 224-228.	0.7	101
81	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
82	A unique form of autosomal dominant cataract explained by gene conversion between Î²-crystallin B2 and its pseudogene. Journal of Medical Genetics, 2001, 38, 392-396.	3.2	101
83	Spectrum of mutations and genotypeâ€“phenotype analysis in Currarino syndrome. European Journal of Human Genetics, 2001, 9, 599-605.	2.8	100
84	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.	5.1	99
85	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, 355-358.	0.7	97
86	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	21.4	97
87	Mutations in the Gene Encoding the Wnt-Signaling Component R-Spondin 4 (RSPO4) Cause Autosomal Recessive Anonychia. American Journal of Human Genetics, 2006, 79, 1105-1109.	6.2	94
88	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.	3.8	93
89	Environmental trichlorfon and cluster of congenital abnormalities. Lancet, The, 1993, 341, 539-542.	13.7	92
90	Gene Localization for an Autosomal Dominant Familial Periodic Fever to 12p13. American Journal of Human Genetics, 1998, 62, 884-889.	6.2	92

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91	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, 1105-1109.	0.7	89
92	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	6.2	89
93	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491.	8.2	89
94	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, 538-546.	6.2	88
95	TNF polymorphisms in psoriasis: Association of psoriatic arthritis with the promoter polymorphismTNF*-857 independent of thePSORS1 risk allele. Arthritis and Rheumatism, 2007, 56, 2056-2064.	6.7	88
96	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2013, 92, 575-583.	6.2	87
97	Renal fibrosis is the common feature of autosomal dominant tubulointerstitial kidney diseases caused by mutations in mucin 1 or uromodulin. Kidney International, 2014, 86, 589-599.	5.2	86
98	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, 613-621.	0.7	85
99	Localisation of gene for the naevoid basal-cell carcinoma syndrome. Lancet, The, 1992, 339, 617.	13.7	84
100	Familial hypomagnesaemia with hypercalciuria and nephrocalcinosis maps to chromosomeÂ3q27 and is associated with mutations in the PCLN-1 gene. European Journal of Human Genetics, 2000, 8, 414-422.	2.8	84
101	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, 1161-1166.	2.8	84
102	Nijmegen breakage syndrome: consequences of defective DNA double strand break repair. BioEssays, 1999, 21, 649-656.	2.5	83
103	Mutations in the Nijmegen Breakage Syndrome gene (NBS1) in childhood acute lymphoblastic leukemia (ALL). Cancer Research, 2001, 61, 3570-2.	0.9	83
104	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.	3.8	82
105	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.	0.8	82
106	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.	6.2	81
107	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, 1459-1462.	6.2	80
108	Type and Level of RMRP Functional Impairment Predicts Phenotype in the Cartilage Hair Hypoplasiaâ€“Anauxetic Dysplasia Spectrum. American Journal of Human Genetics, 2007, 81, 519-529.	6.2	78

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109	Genotype/Phenotype Correlation in Autosomal Recessive Lamellar Ichthyosis. American Journal of Human Genetics, 1998, 62, 1052-1061.	6.2	77
110	Hereditary spastic paraplegia caused by mutations in the SPG4 gene. European Journal of Human Genetics, 2000, 8, 771-776.	2.8	77
111	Exome Pool-Seq in neurodevelopmental disorders. European Journal of Human Genetics, 2017, 25, 1364-1376.	2.8	77
112	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. Nature Communications, 2017, 8, 15910.	12.8	77
113	Variants in ASB10 are associated with open-angle glaucoma. Human Molecular Genetics, 2012, 21, 1336-1349.	2.9	76
114	Genetic variants and cellular stressors associated with exfoliation syndrome modulate promoter activity of a lncRNA within theLOXL1locus. Human Molecular Genetics, 2015, 24, 6552-6563.	2.9	76
115	Genome Scan for Childhood and Adolescent Obesity in German Families. Pediatrics, 2003, 111, 321-327.	2.1	74
116	A novel in situ method for the detection of deficient transglutaminase activity in the skin. Archives of Dermatological Research, 1998, 290, 621-627.	1.9	73
117	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, 753-760.	2.8	73
118	Allelic Heterogeneity in the COH1 Gene Explains Clinical Variabilityin Cohen Syndrome. American Journal of Human Genetics, 2004, 75, 138-145.	6.2	72
119	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636.	3.2	72
120	Null Mutation in PGAP1 Impairing Gpi-Anchor Maturation in Patients with Intellectual Disability and Encephalopathy. PLoS Genetics, 2014, 10, e1004320.	3.5	72
121	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	3.2	72
122	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, 602-609.	2.8	72
123	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, 269-274.	6.2	71
124	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
125	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
126	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	3.3	70

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127	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.	6.2	69
128	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, 275-287.	1.4	68
129	Mutation in the Î²A3/A1-Crystallin Encoding Gene Cryba1 Causes a Dominant Cataract in the Mouse. Genomics, 1999, 62, 67-73.	2.9	67
130	Chromatin-Remodeling-Factor ARID1B Represses Wnt/Î²-Catenin Signaling. American Journal of Human Genetics, 2015, 97, 445-456.	6.2	67
131	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	3.2	67
132	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, 1015-1020.	2.9	66
133	Localisation of a gene for Papillon-LefÃ¨vre syndrome to chromosome 11q14-q21 by homozygosity mapping. Human Genetics, 1997, 101, 376-382.	3.8	66
134	Investigation of a Family with Autosomal Dominant Dilated Cardiomyopathy Defines a Novel Locus on Chromosome 2q14-q22. American Journal of Human Genetics, 1999, 65, 1068-1077.	6.2	66
135	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, 1882-1885.	0.9	64
136	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, 50-56.	3.8	63
137	Mutation in the Scyl1 gene encoding aminoâ€terminal kinaseâ€like protein causes a recessive form of spinocerebellar neurodegeneration. EMBO Reports, 2007, 8, 691-697.	4.5	63
138	Variants in RUNX3 Contribute to Susceptibility to Psoriatic Arthritis, Exhibiting Further Common Ground With Ankylosing Spondylitis. Arthritis and Rheumatism, 2013, 65, 1224-1231.	6.7	63
139	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
140	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	2.0	63
141	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
142	Mutations in CYP1B1 cause primary congenital glaucoma by reduction of either activity or abundance of the enzyme. Human Mutation, 2008, 29, 1147-1153.	2.5	62
143	Replication of LCE3Câ€“LCE3B CNV as a Risk Factor for Psoriasis and Analysis of Interaction with Other Genetic Risk Factors. Journal of Investigative Dermatology, 2010, 130, 979-984.	0.7	61
144	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61

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145	Different Mechanisms and Recurrence Risks of Imprinting Defects in Angelman Syndrome. American Journal of Human Genetics, 1997, 61, 88-93.	6.2	60
146	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	3.5	60
147	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	6.2	59
148	In-Frame Deletion and Missense Mutations of the C-Terminal Helicase Domain of <i>SMARCA2</i> in Three Patients with Nicolaides-Baraitser Syndrome. Molecular Syndromology, 2011, 2, 237-244.	0.8	58
149	Î±-Synuclein-induced myelination deficit defines a novel interventional target for multiple system atrophy. Acta Neuropathologica, 2016, 132, 59-75.	7.7	58
150	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, 170-178.	2.8	57
151	Heterozygous Loss-of-Function Variants inCYP1B1Predispose to Primary Open-Angle Glaucoma. , 2010, 51, 249.		57
152	Pseudoexfoliation syndrome-associated genetic variants affect transcription factor binding and alternative splicing of LOXL1. Nature Communications, 2017, 8, 15466.	12.8	57
153	Genome-wide association study with DNA pooling identifies variants at CNTNAP2 associated with pseudoexfoliation syndrome. European Journal of Human Genetics, 2011, 19, 186-193.	2.8	56
154	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.	2.5	56
155	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, 731-737.	6.2	55
156	Profiling ofWDR36Missense Variants in German Patients with Glaucoma. , 2008, 49, 270.		55
157	Keratin 9 gene mutational heterogeneity in patients with epidermolytic palmoplantar keratoderma. Human Genetics, 1994, 93, 649-54.	3.8	54
158	Fine mapping and single nucleotide polymorphism association results of candidate genes for asthma and related phenotypes. Human Mutation, 2001, 18, 327-336.	2.5	54
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