

# AndrÃ© Reis

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5411569/publications.pdf>

Version: 2024-02-01

447  
papers

33,138  
citations

5126

86  
h-index

7043

159  
g-index

474  
all docs

474  
docs citations

474  
times ranked

42291  
citing authors

#	ARTICLE	IF	CITATIONS
1	Nibrin, a Novel DNA Double-Strand Break Repair Protein, Is Mutated in Nijmegen Breakage Syndrome. <i>Cell</i> , 1998, 93, 467-476.	13.5	989
2	Febrile seizures and generalized epilepsy associated with a mutation in the Na <sup>+</sup> -channel $\alpha$ 1 subunit gene SCN1B. <i>Nature Genetics</i> , 1998, 19, 366-370.	9.4	965
3	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. <i>Lancet</i> , The, 2012, 380, 1674-1682.	6.3	940
4	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. <i>Nature Genetics</i> , 2010, 42, 985-990.	9.4	918
5	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. <i>Nature Genetics</i> , 2012, 44, 1341-1348.	9.4	848
6	Genome-wide, large-scale production of mutant mice by ENU mutagenesis. <i>Nature Genetics</i> , 2000, 25, 444-447.	9.4	658
7	Psoriasis is associated with increased $\beta$ 2-defensin genomic copy number. <i>Nature Genetics</i> , 2008, 40, 23-25.	9.4	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. <i>American Journal of Human Genetics</i> , 2009, 85, 655-666.	2.6	573
9	Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. <i>Cell</i> , 2008, 135, 37-48.	13.5	567
10	Human laminin $\beta$ 2 deficiency causes congenital nephrosis with mesangial sclerosis and distinct eye abnormalities. <i>Human Molecular Genetics</i> , 2004, 13, 2625-2632.	1.4	443
11	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2010, 42, 1021-1026.	9.4	431
12	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. <i>Nature</i> , 2011, 478, 97-102.	13.7	394
13	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. <i>JAMA Oncology</i> , 2017, 3, 636.	3.4	376
14	Mutations in the Pericentrin ( <i>PCNT</i> ) Gene Cause Primordial Dwarfism. <i>Science</i> , 2008, 319, 816-819.	6.0	370
15	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2063-2074.	0.7	343
16	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. <i>Nature Genetics</i> , 2010, 42, 996-999.	9.4	334
17	A Genome-wide Search for Linkage to Asthma <sup>22</sup> See the Appendix.. <i>Genomics</i> , 1999, 58, 1-8.	1.3	332
18	The origin of the major cystic fibrosis mutation ( $\Delta$ F508) in European populations. <i>Nature Genetics</i> , 1994, 7, 169-175.	9.4	323

#	ARTICLE	IF	CITATIONS
19	Mutations in the gene encoding the lamin B receptor produce an altered nuclear morphology in granulocytes (Pelger-Huët anomaly). <i>Nature Genetics</i> , 2002, 31, 410-414.	9.4	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. <i>American Journal of Human Genetics</i> , 2007, 80, 550-560.	2.6	316
21	A comprehensive linkage analysis for myocardial infarction and its related risk factors. <i>Nature Genetics</i> , 2002, 30, 210-214.	9.4	313
22	Mutations in Microcephalin Cause Aberrant Regulation of Chromosome Condensation. <i>American Journal of Human Genetics</i> , 2004, 75, 261-266.	2.6	292
23	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. <i>American Journal of Human Genetics</i> , 2016, 98, 149-164.	2.6	270
24	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). <i>American Journal of Human Genetics</i> , 2007, 80, 994-1001.	2.6	261
25	Keratin 9 gene mutations in epidermolytic palmoplantar keratoderma (EPPK). <i>Nature Genetics</i> , 1994, 6, 174-179.	9.4	255
26	Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). <i>Nature Genetics</i> , 2005, 37, 1345-1350.	9.4	252
27	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. <i>Nature Communications</i> , 2017, 8, 15382.	5.8	251
28	A major susceptibility locus for atopic dermatitis maps to chromosome 3q21. <i>Nature Genetics</i> , 2000, 26, 470-473.	9.4	249
29	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. <i>American Journal of Human Genetics</i> , 2015, 97, 816-836.	2.6	245
30	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. <i>Cell</i> , 2014, 157, 651-663.	13.5	228
31	Genotype-phenotype correlations in Noonan syndrome. <i>Journal of Pediatrics</i> , 2004, 144, 368-374.	0.9	227
32	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 90, 565-572.	2.6	225
33	Mutations in CAV3 cause mechanical hyperirritability of skeletal muscle in rippling muscle disease. <i>Nature Genetics</i> , 2001, 28, 218-219.	9.4	206
34	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. <i>American Journal of Human Genetics</i> , 2011, 88, 788-795.	2.6	206
35	Human TBX1 Missense Mutations Cause Gain of Function Resulting in the Same Phenotype as 22q11.2 Deletions. <i>American Journal of Human Genetics</i> , 2007, 80, 510-517.	2.6	195
36	Splitting Schizophrenia: Periodic Catatonia—Susceptibility Locus on Chromosome 15q15. <i>American Journal of Human Genetics</i> , 2000, 67, 1201-1207.	2.6	192

#	ARTICLE	IF	CITATIONS
37	Dysferlin deletion in SJL mice (SJL-Dysf) defines a natural model for limb girdle muscular dystrophy 2B. <i>Nature Genetics</i> , 1999, 23, 141-142.	9.4	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. <i>Human Molecular Genetics</i> , 2013, 22, 5121-5135.	1.4	190
39	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. <i>JAMA Psychiatry</i> , 2017, 74, 293.	6.0	186
40	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017, 49, 223-237.	9.4	186
41	Imprinting mutations suggested by abnormal DNA methylation patterns in familial Angelman and Prader-Willi syndromes. <i>American Journal of Human Genetics</i> , 1994, 54, 741-7.	2.6	179
42	Identification of ZNF313 / RNF114 as a novel psoriasis susceptibility gene. <i>Human Molecular Genetics</i> , 2008, 17, 1938-1945.	1.4	176
43	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. <i>American Journal of Human Genetics</i> , 2015, 97, 886-893.	2.6	171
44	Î±-Synuclein oligomers induce early axonal dysfunction in human iPSC-based models of synucleinopathies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 7813-7818.	3.3	168
45	Common Genetic Determinants of Intraocular Pressure and Primary Open-Angle Glaucoma. <i>PLoS Genetics</i> , 2012, 8, e1002611.	1.5	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. <i>Human Mutation</i> , 2010, 31, 722-733.	1.1	163
47	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. <i>Nature Communications</i> , 2015, 6, 7001.	5.8	156
48	Common genetic variants associated with open-angle glaucoma. <i>Human Molecular Genetics</i> , 2011, 20, 2464-2471.	1.4	152
49	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. <i>American Journal of Human Genetics</i> , 2011, 88, 106-114.	2.6	151
50	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2013, 93, 124-131.	2.6	151
51	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. <i>Nature Communications</i> , 2015, 6, 6046.	5.8	149
52	Genome search for susceptibility loci of common idiopathic generalised epilepsies. <i>Human Molecular Genetics</i> , 2000, 9, 1465-1472.	1.4	147
53	Sporadic Imprinting Defects in Prader-Willi Syndrome and Angelman Syndrome: Implications for Imprint-Switch Models, Genetic Counseling, and Prenatal Diagnosis. <i>American Journal of Human Genetics</i> , 1998, 63, 170-180.	2.6	142
54	Genetic Regulation of Serum Phytosterol Levels and Risk of Coronary Artery Disease. <i>Circulation: Cardiovascular Genetics</i> , 2010, 3, 331-339.	5.1	141

#	ARTICLE	IF	CITATIONS
55	Homozygosity Mapping in Families with Joubert Syndrome Identifies a Locus on Chromosome 9q34.3 and Evidence for Genetic Heterogeneity. <i>American Journal of Human Genetics</i> , 1999, 65, 1666-1671.	2.6	135
56	Genetic and neurodevelopmental spectrum of <i>SYNGAP1</i> -associated intellectual disability and epilepsy. <i>Journal of Medical Genetics</i> , 2016, 53, 511-522.	1.5	135
57	Heterozygous NTF4 Mutations Impairing Neurotrophin-4 Signaling in Patients with Primary Open-Angle Glaucoma. <i>American Journal of Human Genetics</i> , 2009, 85, 447-456.	2.6	134
58	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. <i>Genetics in Medicine</i> , 2018, 20, 1175-1185.	1.1	133
59	Linkage of the gene for the triple A syndrome to chromosome 12q13 near the type II keratin gene cluster. <i>Human Molecular Genetics</i> , 1996, 5, 2061-2066.	1.4	132
60	Disease burden and risk profile in referred patients with moderate chronic kidney disease: composition of the German Chronic Kidney Disease (GCKD) cohort. <i>Nephrology Dialysis Transplantation</i> , 2015, 30, 441-451.	0.4	132
61	Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations. <i>European Journal of Human Genetics</i> , 2000, 8, 900-902.	1.4	130
62	Genomewide Scan in German Families Reveals Evidence for a Novel Psoriasis-Susceptibility Locus on Chromosome 19p13. <i>American Journal of Human Genetics</i> , 2000, 67, 1020-1024.	2.6	129
63	First known microdeletion within the Wolf-Hirschhorn syndrome critical region refines genotype-phenotype correlation. <i>American Journal of Medical Genetics Part A</i> , 2001, 99, 338-342.	2.4	128
64	The German Chronic Kidney Disease (GCKD) study: design and methods. <i>Nephrology Dialysis Transplantation</i> , 2012, 27, 1454-1460.	0.4	127
65	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. <i>Journal of Medical Genetics</i> , 2010, 47, 321-331.	1.5	126
66	Genome-wide association study identifies a new locus for coronary artery disease on chromosome 10p11.23. <i>European Heart Journal</i> , 2011, 32, 158-168.	1.0	124
67	?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. <i>American Journal of Medical Genetics Part A</i> , 2002, 108, 177-181.	2.4	122
68	Clinical and Mutational Spectrum of Mowat-Wilson Syndrome. <i>European Journal of Medical Genetics</i> , 2005, 48, 97-111.	0.7	121
69	Genotype-Related Expression of Lysyl Oxidase-Like 1 in Ocular Tissues of Patients with Pseudoexfoliation Syndrome/Glaucoma and Normal Patients. <i>American Journal of Pathology</i> , 2008, 173, 1724-1735.	1.9	118
70	A Genome Wide Search for Susceptibility Loci in Three European Malignant Hyperthermia Pedigrees. <i>Human Molecular Genetics</i> , 1997, 6, 953-961.	1.4	117
71	Severely Incapacitating Mutations in Patients with Extreme Short Stature Identify RNA-Processing Endoribonuclease RMRP as an Essential Cell Growth Regulator. <i>American Journal of Human Genetics</i> , 2005, 77, 795-806.	2.6	117
72	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. <i>Journal of Medical Genetics</i> , 2007, 44, 651-656.	1.5	114

#	ARTICLE	IF	CITATIONS
73	Association of <i>LOXL1</i> Common 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 <i>LOXL1</i> and five new susceptibility loci. <i>Nature Genetics</i> , 2017, 49, 993-1004.	9.4	114
75	Splitting Schizophrenia: Periodic Catatoniaâ€“Susceptibility Locus on Chromosome 15q15. <i>American Journal of Human Genetics</i> , 2000, 67, 1201-1207.	2.6	112
76	Congenital nephrosis, mesangial sclerosis, and distinct eye abnormalities with microcoria: An autosomal recessive syndrome. , 2004, 130A, 138-145.		110
77	Expanding the clinical spectrum associated with defects in <i>CNTNAP2</i> and <i>NRXN1</i> . <i>BMC Medical Genetics</i> , 2011, 12, 106.	2.1	109
78	Molecular karyotyping using an SNP array for genome-wide genotyping. <i>Journal of Medical Genetics</i> , 2004, 41, 916-922.	1.5	106
79	Identification of a New Gene Locus for Adolescent Nephronophthisis, on Chromosome 3q22 in a Large Venezuelan Pedigree. <i>American Journal of Human Genetics</i> , 2000, 66, 118-127.	2.6	105
80	Self-Healing Collodion Baby: a Dynamic Phenotype Explained by a Particular Transglutaminase-1 Mutation. <i>Journal of Investigative Dermatology</i> , 2003, 120, 224-228.	0.3	101
81	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. <i>Genetics in Medicine</i> , 2018, 20, 630-638.	1.1	101
82	A unique form of autosomal dominant cataract explained by gene conversion between $\beta$ 2-crystallin B2 and its pseudogene. <i>Journal of Medical Genetics</i> , 2001, 38, 392-396.	1.5	101
83	Spectrum of mutations and genotypeâ€“phenotype analysis in Currarino syndrome. <i>European Journal of Human Genetics</i> , 2001, 9, 599-605.	1.4	100
84	Gene panel sequencing in familial breast/ovarian cancer patients identifies multiple novel mutations also in genes others than <i>BRCA1/2</i> . <i>International Journal of Cancer</i> , 2017, 140, 95-102.	2.3	99
85	Genetic Variants of the IL-23R Pathway: Association with Psoriatic Arthritis and Psoriasis Vulgaris, but No Specific Risk Factor for Arthritis. <i>Journal of Investigative Dermatology</i> , 2009, 129, 355-358.	0.3	97
86	A common variant mapping to <i>CACNA1A</i> is associated with susceptibility to exfoliation syndrome. <i>Nature Genetics</i> , 2015, 47, 387-392.	9.4	97
87	Mutations in the Gene Encoding the Wnt-Signaling Component R-Spondin 4 ( <i>RSPO4</i> ) Cause Autosomal Recessive Anonychia. <i>American Journal of Human Genetics</i> , 2006, 79, 1105-1109.	2.6	94
88	De novo mutations in beta-catenin ( <i>CTNNB1</i> ) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. <i>Human Genetics</i> , 2015, 134, 97-109.	1.8	93
89	Environmental trichlorfon and cluster of congenital abnormalities. <i>Lancet, The</i> , 1993, 341, 539-542.	6.3	92
90	Gene Localization for an Autosomal Dominant Familial Periodic Fever to 12p13. <i>American Journal of Human Genetics</i> , 1998, 62, 884-889.	2.6	92

#	ARTICLE	IF	CITATIONS
91	Meta-Analysis Confirms the LCE3C_LCE3B Deletion as a Risk Factor for Psoriasis in Several Ethnic Groups and Finds Interaction with HLA-Cw6. <i>Journal of Investigative Dermatology</i> , 2011, 131, 1105-1109.	0.3	89
92	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. <i>American Journal of Human Genetics</i> , 2018, 103, 1045-1052.	2.6	89
93	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndrome-like phenotype and hyperactivated MAPK signaling in humans and mice. <i>Journal of Clinical Investigation</i> , 2011, 121, 3479-3491.	3.9	89
94	Limb Mammary Syndrome: A New Genetic Disorder with Mammary Hypoplasia, Ectrodactyly, and Other Hand/Foot Anomalies Maps to Human Chromosome 3q27. <i>American Journal of Human Genetics</i> , 1999, 64, 538-546.	2.6	88
95	TNF polymorphisms in psoriasis: Association of psoriatic arthritis with the promoter polymorphism TNF*-857 independent of the PSORS1 risk allele. <i>Arthritis and Rheumatism</i> , 2007, 56, 2056-2064.	6.7	88
96	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. <i>American Journal of Human Genetics</i> , 2013, 92, 575-583.	2.6	87
97	Renal fibrosis is the common feature of autosomal dominant tubulointerstitial kidney diseases caused by mutations in mucin 1 or uromodulin. <i>Kidney International</i> , 2014, 86, 589-599.	2.6	86
98	Prevalence and correlates of gout in a large cohort of patients with chronic kidney disease: the German Chronic Kidney Disease (GCKD) study. <i>Nephrology Dialysis Transplantation</i> , 2015, 30, 613-621.	0.4	85
99	Localisation of gene for the naevoid basal-cell carcinoma syndrome. <i>Lancet, The</i> , 1992, 339, 617.	6.3	84
100	Familial hypomagnesaemia with hypercalciuria and nephrocalcinosis maps to chromosome 3q27 and is associated with mutations in the PCLN-1 gene. <i>European Journal of Human Genetics</i> , 2000, 8, 414-422.	1.4	84
101	Homozygosity mapping in 64 Syrian consanguineous families with non-specific intellectual disability reveals 11 novel loci and high heterogeneity. <i>European Journal of Human Genetics</i> , 2011, 19, 1161-1166.	1.4	84
102	Nijmegen breakage syndrome: consequences of defective DNA double strand break repair. <i>BioEssays</i> , 1999, 21, 649-656.	1.2	83
103	Mutations in the Nijmegen Breakage Syndrome gene (NBS1) in childhood acute lymphoblastic leukemia (ALL). <i>Cancer Research</i> , 2001, 61, 3570-2.	0.4	83
104	Mapping of a gene for epidermolytic palmoplantar keratoderma to the region of the acidic keratin gene cluster at 17q12-q21. <i>Human Genetics</i> , 1992, 90, 113-6.	1.8	82
105	Disturbed Wnt Signalling due to a Mutation in CCDC88C Causes an Autosomal Recessive Non-Syndromic Hydrocephalus with Medial Diverticulum. <i>Molecular Syndromology</i> , 2010, 1, 99-112.	0.3	82
106	The gene for the ataxia-telangiectasia variant, Nijmegen breakage syndrome, maps to a 1-cM interval on chromosome 8q21. <i>American Journal of Human Genetics</i> , 1997, 60, 605-10.	2.6	81
107	Diaphragmatic Spinal Muscular Atrophy with Respiratory Distress Is Heterogeneous, and One Form Is Linked to Chromosome 11q13-q21. <i>American Journal of Human Genetics</i> , 1999, 65, 1459-1462.	2.6	80
108	Type and Level of RMRP Functional Impairment Predicts Phenotype in the Cartilage Hair Hypoplasia-Anauxetic Dysplasia Spectrum. <i>American Journal of Human Genetics</i> , 2007, 81, 519-529.	2.6	78

#	ARTICLE	IF	CITATIONS
109	Genotype/Phenotype Correlation in Autosomal Recessive Lamellar Ichthyosis. <i>American Journal of Human Genetics</i> , 1998, 62, 1052-1061.	2.6	77
110	Hereditary spastic paraplegia caused by mutations in the SPG4 gene. <i>European Journal of Human Genetics</i> , 2000, 8, 771-776.	1.4	77
111	Exome Pool-Seq in neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2017, 25, 1364-1376.	1.4	77
112	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. <i>Nature Communications</i> , 2017, 8, 15910.	5.8	77
113	Variants in ASB10 are associated with open-angle glaucoma. <i>Human Molecular Genetics</i> , 2012, 21, 1336-1349.	1.4	76
114	Genetic variants and cellular stressors associated with exfoliation syndrome modulate promoter activity of a lncRNA within the <i>LOXL1</i> locus. <i>Human Molecular Genetics</i> , 2015, 24, 6552-6563.	1.4	76
115	Genome Scan for Childhood and Adolescent Obesity in German Families. <i>Pediatrics</i> , 2003, 111, 321-327.	1.0	74
116	A novel in situ method for the detection of deficient transglutaminase activity in the skin. <i>Archives of Dermatological Research</i> , 1998, 290, 621-627.	1.1	73
117	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 753-760.	1.4	73
118	Allelic Heterogeneity in the COH1 Gene Explains Clinical Variability in Cohen Syndrome. <i>American Journal of Human Genetics</i> , 2004, 75, 138-145.	2.6	72
119	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. <i>Journal of Medical Genetics</i> , 2007, 44, 629-636.	1.5	72
120	Null Mutation in PGAP1 Impairing Gpi-Anchor Maturation in Patients with Intellectual Disability and Encephalopathy. <i>PLoS Genetics</i> , 2014, 10, e1004320.	1.5	72
121	The clinical significance of small copy number variants in neurodevelopmental disorders. <i>Journal of Medical Genetics</i> , 2014, 51, 677-688.	1.5	72
122	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. <i>European Journal of Human Genetics</i> , 2015, 23, 602-609.	1.4	72
123	A Second Locus for an Axonal Form of Autosomal Recessive Charcot-Marie-Tooth Disease Maps to Chromosome 19q13.3. <i>American Journal of Human Genetics</i> , 2001, 68, 269-274.	2.6	71
124	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. <i>Human Mutation</i> , 2016, 37, 755-764.	1.1	70
125	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
126	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. <i>Scientific Reports</i> , 2018, 8, 17201.	1.6	70



#	ARTICLE	IF	CITATIONS
127	Mutations in MBOAT7, Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. <i>American Journal of Human Genetics</i> , 2016, 99, 912-916.	2.6	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. <i>Neurogenetics</i> , 2009, 10, 275-287.	0.7	68
129	Mutation in the $\beta$ A3/A1-Crystallin Encoding Gene Cryba1 Causes a Dominant Cataract in the Mouse. <i>Genomics</i> , 1999, 62, 67-73.	1.3	67
130	Chromatin-Remodeling-Factor ARID1B Represses Wnt/ $\beta$ -Catenin Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 445-456.	2.6	67
131	<i>FOXP2</i> variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. <i>Journal of Medical Genetics</i> , 2017, 54, 64-72.	1.5	67
132	Localization of a locus for the striated form of palmoplantar keratoderma to chromosome 18q near the desmosomal cadherin gene cluster. <i>Human Molecular Genetics</i> , 1995, 4, 1015-1020.	1.4	66
133	Localisation of a gene for Papillon-Lefèvre syndrome to chromosome 11q14-q21 by homozygosity mapping. <i>Human Genetics</i> , 1997, 101, 376-382.	1.8	66
134	Investigation of a Family with Autosomal Dominant Dilated Cardiomyopathy Defines a Novel Locus on Chromosome 2q14-q22. <i>American Journal of Human Genetics</i> , 1999, 65, 1068-1077.	2.6	66
135	PTPN22 is associated with susceptibility to psoriatic arthritis but not psoriasis: evidence for a further PsA-specific risk locus. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1882-1885.	0.5	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. <i>Human Genetics</i> , 2003, 112, 50-56.	1.8	63
137	Mutation in the Scyl1 gene encoding amino-terminal kinase-like protein causes a recessive form of spinocerebellar neurodegeneration. <i>EMBO Reports</i> , 2007, 8, 691-697.	2.0	63
138	Variants in <i>RUNX3</i> Contribute to Susceptibility to Psoriatic Arthritis, Exhibiting Further Common Ground With Ankylosing Spondylitis. <i>Arthritis and Rheumatism</i> , 2013, 65, 1224-1231.	6.7	63
139	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 468-479.	2.6	63
140	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. <i>Clinical Genetics</i> , 2019, 95, 462-478.	1.0	63
141	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. <i>Human Mutation</i> , 2020, 41, 837-849.	1.1	63
142	Mutations in CYP1B1 cause primary congenital glaucoma by reduction of either activity or abundance of the enzyme. <i>Human Mutation</i> , 2008, 29, 1147-1153.	1.1	62
143	Replication of LCE3C/LCE3B CNV as a Risk Factor for Psoriasis and Analysis of Interaction with Other Genetic Risk Factors. <i>Journal of Investigative Dermatology</i> , 2010, 130, 979-984.	0.3	61
144	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 101, 503-515.	2.6	61

#	ARTICLE	IF	CITATIONS
145	Different Mechanisms and Recurrence Risks of Imprinting Defects in Angelman Syndrome. <i>American Journal of Human Genetics</i> , 1997, 61, 88-93.	2.6	60
146	Rare Copy Number Variants Are a Common Cause of Short Stature. <i>PLoS Genetics</i> , 2013, 9, e1003365.	1.5	60
147	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2014, 94, 649-661.	2.6	59
148	In-Frame Deletion and Missense Mutations of the C-Terminal Helicase Domain of SMARCA2 in Three Patients with Nicolaides-Baraitser Syndrome. <i>Molecular Syndromology</i> , 2011, 2, 237-244.	0.3	58
149	Î±-Synuclein-induced myelination deficit defines a novel interventional target for multiple system atrophy. <i>Acta Neuropathologica</i> , 2016, 132, 59-75.	3.9	58
150	A new quantitative PCR multiplex assay for rapid analysis of chromosome 17p11.2-12 duplications and deletions leading to HMSN/HNPP. <i>European Journal of Human Genetics</i> , 2003, 11, 170-178.	1.4	57
151	Heterozygous Loss-of-Function Variants in CYP1B1 Predispose to Primary Open-Angle Glaucoma. , 2010, 51, 249.		57
152	Pseudoexfoliation syndrome-associated genetic variants affect transcription factor binding and alternative splicing of LOXL1. <i>Nature Communications</i> , 2017, 8, 15466.	5.8	57
153	Genome-wide association study with DNA pooling identifies variants at CNTNAP2 associated with pseudoexfoliation syndrome. <i>European Journal of Human Genetics</i> , 2011, 19, 186-193.	1.4	56
154	BRCA mutations and their influence on pathological complete response and prognosis in a clinical cohort of neoadjuvantly treated breast cancer patients. <i>Breast Cancer Research and Treatment</i> , 2018, 171, 85-94.	1.1	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. <i>American Journal of Human Genetics</i> , 2004, 74, 731-737.	2.6	55
156	Profiling of WDR36 Missense Variants in German Patients with Glaucoma. , 2008, 49, 270.		55
157	Keratin 9 gene mutational heterogeneity in patients with epidermolytic palmoplantar keratoderma. <i>Human Genetics</i> , 1994, 93, 649-54.	1.8	54
158	Fine mapping and single nucleotide polymorphism association results of candidate genes for asthma and related phenotypes. <i>Human Mutation</i> , 2001, 18, 327-336.	1.1	54
159	A locus on chromosome 15q for a dominantly inherited nemaline myopathy with core-like lesions. <i>Brain</i> , 2003, 126, 1545-1551.	3.7	54
160	Exploring Functional Candidate Genes for Genetic Association in German Patients with Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. , 2009, 50, 2796.		54
161	Inflammation-induced glycolytic switch controls suppressivity of mesenchymal stem cells via STAT1 glycosylation. <i>Leukemia</i> , 2019, 33, 1783-1796.	3.3	54
162	Mutations in the pyruvate kinase L gene in patients with hereditary hemolytic anemia. <i>Blood</i> , 1994, 83, 2817-2822.	0.6	53

#	ARTICLE	IF	CITATIONS
163	Epigenetic targeting in the mouse zygote marks DNA for later methylation: a mechanism for maternal effects in development. <i>Mechanisms of Development</i> , 2001, 103, 35-47.	1.7	53
164	Periodic catatonia: confirmation of linkage to chromosome 15 and further evidence for genetic heterogeneity. <i>Human Genetics</i> , 2002, 111, 323-330.	1.8	53
165	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. <i>Scientific Reports</i> , 2017, 7, 12225.	1.6	53
166	Mitochondrial DNA copy number is associated with mortality and infections in a large cohort of patients with chronic kidney disease. <i>Kidney International</i> , 2019, 96, 480-488.	2.6	53
167	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. <i>American Journal of Human Genetics</i> , 2020, 107, 527-538.	2.6	53
168	Characterization of a Mutation in the Lens-specific MP70 Encoding Gene of the Mouse Leading to a Dominant Cataract. <i>Experimental Eye Research</i> , 2001, 73, 867-876.	1.2	52
169	Cloning of the mouse dysferlin gene and genomic characterization of the SJL-Dysf mutation. <i>NeuroReport</i> , 2001, 12, 625-629.	0.6	52
170	Familial interstitial 570 kbp deletion of the UBE3A gene region causing Angelman syndrome but not Prader-Willi syndrome. <i>American Journal of Medical Genetics Part A</i> , 2002, 111, 233-237.	2.4	52
171	Neurodevelopmental deficits in Pierson (microcoria-congenital nephrosis) syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 311-319.	0.7	52
172	A Gene for an Autosomal Dominant Scleroatrophic Syndrome Predisposing to Skin Cancer (Huriez) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	2.6	51
173	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). <i>ELife</i> , 2015, 4, .	2.8	51
174	Association of Î²-Defensin Copy Number and Psoriasis in Three Cohorts of European Origin. <i>Journal of Investigative Dermatology</i> , 2012, 132, 2407-2413.	0.3	50
175	A new face of Borjesonâ€“Forssmanâ€“Lehmann syndrome? De novo mutations in <i>PHF6</i> in seven females with a distinct phenotype. <i>Journal of Medical Genetics</i> , 2013, 50, 838-847.	1.5	50
176	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	3.6	50
177	Palmoplantar Keratoderma in Association with Carcinoma of the Esophagus Maps to Chromosome 17q Distal to the Keratin Gene Cluster. <i>Genomics</i> , 1995, 29, 537-540.	1.3	49
178	Hereditary Isolated Renal Magnesium Loss Maps to Chromosome 11q23. <i>American Journal of Human Genetics</i> , 1999, 64, 180-188.	2.6	49
179	Genome-wide linkage reveals a locus for human essential (primary) hypertension on chromosome 12p. <i>Human Molecular Genetics</i> , 2003, 12, 1273-1277.	1.4	49
180	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2018, 102, 44-57.	2.6	49

#	ARTICLE	IF	CITATIONS
181	Exclusion of the GABAA-receptor $\gamma$ 23 subunit gene as the Angelman's syndrome gene. <i>Lancet</i> , The, 1993, 341, 122-123.	6.3	48
182	The Fanconi Anemia Group E Gene, FANCE, Maps to Chromosome 6p. <i>American Journal of Human Genetics</i> , 1999, 64, 1400-1405.	2.6	48
183	A Gene for Hypotrichosis Simplex of the Scalp Maps to Chromosome 6p21.3. <i>American Journal of Human Genetics</i> , 2000, 66, 1979-1983.	2.6	48
184	A European study on the genetics of mite sensitization. <i>Journal of Allergy and Clinical Immunology</i> , 2000, 106, 925-932.	1.5	48
185	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 103, 305-316.	2.6	48
186	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. <i>Genetics in Medicine</i> , 2019, 21, 2723-2733.	1.1	48
187	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 38.	1.2	48
188	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. <i>Journal of Investigative Dermatology</i> , 2020, 140, 1451-1455.e13.	0.3	48
189	Close mapping of the focal non-epidermolytic palmoplantar keratoderma (PPK) locus associated with oesophageal cancer (TOC). <i>Human Molecular Genetics</i> , 1996, 5, 857-860.	1.4	47
190	Localization of a Gene for Syndactyly Type 1 to Chromosome 2q34-q36. <i>American Journal of Human Genetics</i> , 2000, 67, 492-497.	2.6	47
191	Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. <i>Behavioral and Brain Functions</i> , 2013, 9, 20.	1.4	47
192	Eight further individuals with intellectual disability and epilepsy carrying bi-allelic <i>CNTNAP2</i> aberrations allow delineation of the mutational and phenotypic spectrum. <i>Journal of Medical Genetics</i> , 2016, 53, 820-827.	1.5	45
193	Analysis of the expression pattern of the schizophrenia-risk and intellectual disability gene TCF4 in the developing and adult brain suggests a role in development and plasticity of cortical and hippocampal neurons. <i>Molecular Autism</i> , 2018, 9, 20.	2.6	45
194	<i>TRIM28</i> haploinsufficiency predisposes to Wilms tumor. <i>International Journal of Cancer</i> , 2019, 145, 941-951.	2.3	45
195	Mutations in <i>PIK3C2A</i> cause syndromic short stature, skeletal abnormalities, and cataracts associated with ciliary dysfunction. <i>PLoS Genetics</i> , 2019, 15, e1008088.	1.5	45
196	Genetic Basis and Pancreatic Biology of Johanson-Blizzard Syndrome. <i>Endocrinology and Metabolism Clinics of North America</i> , 2006, 35, 243-253.	1.2	44
197	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. <i>American Journal of Human Genetics</i> , 2016, 99, 228-235.	2.6	44
198	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. <i>Journal of Medical Genetics</i> , 2019, 56, 701-710.	1.5	43

#	ARTICLE	IF	CITATIONS
199	Linkage of Familial Euthyroid Goiter to the Multinodular Goiter-1 Locus and Exclusion of the Candidate Genes Thyroglobulin, Thyroperoxidase, and Na <sup>+</sup> /I <sup>-</sup> Symporter. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 3750-3756.	1.8	42
200	Mutation in the gene encoding lysosomal acid phosphatase (Acp2) causes cerebellum and skin malformation in mouse. <i>Neurogenetics</i> , 2004, 5, 229-238.	0.7	41
201	<i>DDX3X</i> mutations in two girls with a phenotype overlapping Torielloâ€™Carey syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1369-1373.	0.7	41
202	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. <i>Human Molecular Genetics</i> , 2017, 26, 4301-4313.	1.4	41
203	Mutations in DCPS and EDC3 in autosomal recessive intellectual disability indicate a crucial role for mRNA decapping in neurodevelopment. <i>Human Molecular Genetics</i> , 2015, 24, 3172-3180.	1.4	40
204	GSK3Û-dependent dysregulation of neurodevelopment in SPG11â€™patient induced pluripotent stem cell model. <i>Annals of Neurology</i> , 2016, 79, 826-840.	2.8	40
205	Single molecule real time sequencing in ADTKD-MUC1 allows complete assembly of the VNTR and exact positioning of causative mutations. <i>Scientific Reports</i> , 2018, 8, 4170.	1.6	40
206	The Gene for Autosomal Dominant Craniometaphyseal Dysplasia Maps to Chromosome 5p and Is Distinct from the Growth Hormone-Receptor Gene. <i>American Journal of Human Genetics</i> , 1997, 61, 918-923.	2.6	39
207	Epidermolytic palmoplantar keratoderma of VÃ¶rner: re-evaluation of VÃ¶rner's original family and identification of a novel keratin 9 mutation. <i>Archives of Dermatological Research</i> , 2002, 294, 268-272.	1.1	39
208	Demonstration of two novel LAMB2 mutations in the original Pierson syndrome family reported 42 years ago. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 73-74.	0.7	39
209	Loss-of-Function Variants of the Filaggrin Gene Are Not Major Susceptibility Factors for Psoriasis Vulgaris or Psoriatic Arthritis in German Patients. <i>Journal of Investigative Dermatology</i> , 2007, 127, 1367-1370.	0.3	39
210	Mutations in <i>BRCA1/2</i> and Other Panel Genes in Patients With Metastatic Breast Cancer â€™Association With Patient and Disease Characteristics and Effect on Prognosis. <i>Journal of Clinical Oncology</i> , 2021, 39, 1619-1630.	0.8	39
211	Localization of the Gene Causing Keratolytic Winter Erythema to Chromosome 8p22-p23, and Evidence for a Founder Effect in South African Afrikaans-Speakers. <i>American Journal of Human Genetics</i> , 1997, 61, 370-378.	2.6	38
212	Assignment of PGL3 to chromosome 1 (q21-q23) in a family with autosomal dominant non-chromaffin paraganglioma. <i>American Journal of Medical Genetics Part A</i> , 2001, 98, 32-36.	2.4	38
213	Tumor necrosis factor receptor-associated periodic syndrome characterized by a mutation affecting the cleavage site of the receptor: Implications for pathogenesis. <i>Arthritis and Rheumatism</i> , 2003, 48, 2386-2388.	6.7	38
214	Systematic Linkage Disequilibrium Analysis of SLC12A8 at PSORS5 Confirms a Role in Susceptibility to Psoriasis Vulgaris. <i>Journal of Investigative Dermatology</i> , 2005, 125, 906-912.	0.3	38
215	Risk, Prediction and Prevention of Hereditary Breast Cancer â€™ Large-Scale Genomic Studies in Times of Big and Smart Data. <i>Geburtshilfe Und Frauenheilkunde</i> , 2018, 78, 481-492.	0.8	38
216	Intellectual disability and autism spectrum disorders â€™on the flyâ€™: insights from <i>Drosophila</i>. <i>DMM Disease Models and Mechanisms</i> , 2019, 12, .	1.2	38

#	ARTICLE	IF	CITATIONS
217	Identification, by Homozygosity Mapping, of a Novel Locus for Autosomal Recessive Congenital Ichthyosis on Chromosome 17p, and Evidence for Further Genetic Heterogeneity. <i>American Journal of Human Genetics</i> , 2001, 69, 216-222.	2.6	37
218	Primary congenital glaucoma and Rieger's anomaly: extended haplotypes reveal founder effects for eight distinct CYP1B1 mutations. <i>Molecular Vision</i> , 2006, 12, 523-31.	1.1	37
219	Analysis of 133 Meioses Places the Genes for Nevroid Basal Cell Carcinoma (Gorlin) Syndrome and Fanconi Anemia Group C in a 2.6-cM Interval and Contributes to the Fine Map of 9q22.3. <i>Genomics</i> , 1994, 23, 486-489.	1.3	36
220	Association between protein tyrosine phosphatase 22 variant R620W in conjunction with the HLA-DRB1 shared epitope and humoral autoimmunity to an immunodominant epitope of cartilage-specific type II collagen in early rheumatoid arthritis. <i>Arthritis and Rheumatism</i> , 2006, 54, 82-89.	6.7	36
221	Genetic influences in the formation of nasal polyps. <i>Lancet, The</i> , 1991, 337, 974.	6.3	35
222	Autosomal dominant spastic paraplegia with anticipation maps to a 4-cM interval on chromosome 2p21-p24 in a large German family. <i>Human Genetics</i> , 1996, 98, 371-375.	1.8	35
223	Phenotypic differences in Angelman syndrome patients: Imprinting mutations show less frequently microcephaly and hypopigmentation than deletions. , 1996, 66, 221-226.		35
224	<i>STAG1</i> mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2017, 54, 479-488.	1.5	35
225	Bainbridge-Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. <i>European Journal of Human Genetics</i> , 2017, 25, 183-191.	1.4	35
226	Envoplakin, a Possible Candidate Gene for Focal NEPPK/Esophageal Cancer (TOC): The Integration of Genetic and Physical Maps of the TOC Region on 17q25. <i>Genomics</i> , 1999, 59, 234-242.	1.3	34
227	Towards the genetic basis of periodic catatonia: pedigree sample for genome scan I and II. <i>European Archives of Psychiatry and Clinical Neuroscience</i> , 2001, 251, 125-130.	1.8	34
228	Characterisation of psoriasis susceptibility locus 6 (PSORS6) in patients with early onset psoriasis and evidence for interaction with PSORS1. <i>Journal of Medical Genetics</i> , 2009, 46, 736-744.	1.5	34
229	Deletion of LCE3C and LCE3B genes at PSORS4 does not contribute to susceptibility to psoriatic arthritis in German patients. <i>Annals of the Rheumatic Diseases</i> , 2010, 69, 876-878.	0.5	34
230	<i>NDST1</i> missense mutations in autosomal recessive intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2753-2763.	0.7	34
231	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
232	Frequent LPA KIV-2 Variants Lower Lipoprotein(a) Concentrations and Protect Against Coronary Artery Disease. <i>Journal of the American College of Cardiology</i> , 2021, 78, 437-449.	1.2	34
233	Encephalopathies with <i>KCNC1</i> variants: genotype-phenotype-functional correlations. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1263-1272.	1.7	33
234	Prevalence of <i>FOXC1</i> Variants in Individuals With a Suspected Diagnosis of Primary Congenital Glaucoma. <i>JAMA Ophthalmology</i> , 2019, 137, 348.	1.4	33

#	ARTICLE	IF	CITATIONS
235	A novel 5q35.3 subtelomeric deletion syndrome. , 2003, 121A, 1-8.		32
236	Non-syndromic autosomal dominant progressive non-specific mid-frequency sensorineural hearing impairment with childhood to late adolescence onset (DFNA21). <i>Clinical Otolaryngology</i> , 2000, 25, 45-54.	0.0	31
237	Evidence for susceptibility determinant(s) to psoriasis vulgaris in or near PTPN22 in German patients. <i>Journal of Medical Genetics</i> , 2006, 43, 517-522.	1.5	31
238	Novel missense, insertion and deletion mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with congenital insensitivity to pain with anhidrosis. <i>Neuromuscular Disorders</i> , 2008, 18, 159-166.	0.3	31
239	Evidence for RPGRI1 gene as risk factor for primary open angle glaucoma. <i>European Journal of Human Genetics</i> , 2011, 19, 445-451.	1.4	31
240	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: A study in Spanish and Italian populations and meta-analysis. <i>Arthritis and Rheumatism</i> , 2011, 63, 1860-1865.	6.7	31
241	Altered <i>GPM6A/M6</i> Dosage Impairs Cognition and Causes Phenotypes Responsive to Cholesterol in Human and <i>Drosophila</i> . <i>Human Mutation</i> , 2014, 35, 1495-1505.	1.1	31
242	The polynucleotide kinase 3â€²-phosphatase gene (PNKP) is involved in Charcot-Marie-Tooth disease (CMT2B2) previously related to MED25. <i>Neurogenetics</i> , 2018, 19, 215-225.	0.7	31
243	Transcription factor Tcf4 is the preferred heterodimerization partner for Olig2 in oligodendrocytes and required for differentiation. <i>Nucleic Acids Research</i> , 2020, 48, 4839-4857.	6.5	31
244	Aey2, a new mutation in the betaB2-crystallin-encoding gene of the mouse. <i>Investigative Ophthalmology and Visual Science</i> , 2001, 42, 1574-80.	3.3	31
245	Prenatal findings in four consecutive pregnancies with fetal Pierson syndrome, a newly defined congenital nephrosis syndrome. <i>Prenatal Diagnosis</i> , 2006, 26, 262-266.	1.1	30
246	The ataxia-telangiectasia-variant genes 1 and 2 are distinct from the ataxia-telangiectasia gene on chromosome 11q23.1. <i>American Journal of Human Genetics</i> , 1995, 57, 960-2.	2.6	30
247	Trinucleotide repeat polymorphism at the PKLR locus. <i>Human Molecular Genetics</i> , 1994, 3, 523-523.	1.4	29
248	Primary Congenital Glaucoma: A Novel Single-Nucleotide Deletion and Varying Phenotypic Expression for the 1546???1555dup Mutation in the GLC3A (CYP1B1) Gene in 2 Families of Different Ethnic Origin. <i>Journal of Glaucoma</i> , 2003, 12, 27-30.	0.8	29
249	Divergent genetic and epigenetic post-zygotic isolation mechanisms in <i>Mus</i> and <i>Peromyscus</i> . <i>Journal of Evolutionary Biology</i> , 2004, 17, 453-460.	0.8	29
250	Lack of genetic association of the three more common polymorphisms of CARD15 with psoriatic arthritis and psoriasis in a German cohort. <i>Annals of the Rheumatic Diseases</i> , 2005, 64, 951-954.	0.5	29
251	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. <i>Human Genetics</i> , 2014, 133, 939-949.	1.8	29
252	Genotype analysis of cystic fibrosis patients in relation to pancreatic sufficiency. <i>Lancet</i> , The, 1990, 335, 738-739.	6.3	28

#	ARTICLE	IF	CITATIONS
253	Genetic and immunohistochemical detection of mutations inactivating the keratinocyte transglutaminase in patients with lamellar ichthyosis. <i>Human Genetics</i> , 1998, 102, 314-318.	1.8	28
254	Comparative association analysis reveals that corneodesmosin is more closely associated with psoriasis than HLA-Cw*0602-B*5701 in German families. <i>Tissue Antigens</i> , 2001, 57, 440-446.	1.0	28
255	Novel mutations in the MYOC/GLC1A gene in a large group of glaucoma patients. <i>Human Mutation</i> , 2002, 20, 479-480.	1.1	28
256	The scoliosis <i>(sco)</i> mouse: a new allele of <i>Pax1</i> . <i>Cytogenetic and Genome Research</i> , 2005, 111, 16-26.	0.6	28
257	Male Restricted Genetic Association of Variant R620W in PTPN22 with Psoriatic Arthritis. <i>Journal of Investigative Dermatology</i> , 2006, 126, 936-938.	0.3	28
258	Two novel mutations in the insulin binding subunit of the insulin receptor gene without insulin binding impairment in a patient with Rabson-Mendenhall syndrome. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 356-362.	0.5	28
259	DYNC2L1 mutations broaden the clinical spectrum of dynein-2 defects. <i>Scientific Reports</i> , 2015, 5, 11649.	1.6	28
260	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. <i>Nature Genetics</i> , 2018, 50, 1442-1451.	9.4	28
261	Further delineation of the female phenotype with <i>KDM5C</i> disease causing variants: 19 new individuals and review of the literature. <i>Clinical Genetics</i> , 2020, 98, 43-55.	1.0	28
262	Clinical variability and novel mutations in the NHEJ1 gene in patients with a Nijmegen breakage syndrome-like phenotype. <i>Human Mutation</i> , 2010, 31, 1059-1068.	1.1	27
263	HIBCH deficiency in a patient with phenotypic characteristics of mitochondrial disorders. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3162-3169.	0.7	27
264	Females with de novo aberrations in <i>PHF6</i> : Clinical overlap of Borjeson-Forsman-Lehmann with Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014, 166, 290-301.	0.7	27
265	Homozygosity mapping in a family with microcephaly, mental retardation, and short stature to a Cohen syndrome region on 8q21.3 - 8q22.1: Redefining a clinical entity. , 2000, 92, 285-292.		26
266	Association scan of the novel psoriasis susceptibility region on chromosome 19: evidence for both susceptible and protective loci. <i>Experimental Dermatology</i> , 2003, 12, 490-496.	1.4	26
267	Evaluation of risk loci for schizophrenia derived from genome-wide association studies in a German population. , 2011, 156, 198-203.		26
268	Deletions in the 3' Part of the <i>NFIX</i> Gene Including a Recurrent Alu-Mediated Deletion of Exon 6 and 7 Account for Previously Unexplained Cases of Marshall-Smith Syndrome. <i>Human Mutation</i> , 2014, 35, 1092-1100.	1.1	26
269	Expanding the clinical spectrum of COL1A1 mutations in different forms of glaucoma. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 108.	1.2	26
270	Hypomorphic Pathogenic Variants in TAF13 Are Associated with Autosomal-Recessive Intellectual Disability and Microcephaly. <i>American Journal of Human Genetics</i> , 2017, 100, 555-561.	2.6	26



#	ARTICLE	IF	CITATIONS
271	The Gene for Human Fibronectin Glomerulopathy Maps to 1q32, in the Region of the Regulation of Complement Activation Gene Cluster. <i>American Journal of Human Genetics</i> , 1998, 63, 1724-1731.	2.6	25
272	Interleukin-10 promoter polymorphism IL10.G and familial early onset psoriasis. <i>British Journal of Dermatology</i> , 2003, 149, 381-385.	1.4	25
273	Tumor necrosis factor promoter polymorphism TNF*-857 is a risk allele for psoriatic arthritis independent of the PSORS1 locus. <i>Arthritis and Rheumatism</i> , 2011, 63, 3801-3806.	6.7	25
274	Haploinsufficiency of <i>NR4A2</i> is associated with a neurodevelopmental phenotype with prominent language impairment. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2231-2234.	0.7	25
275	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2298-2309.	3.0	25
276	Exclusion of the neuronal nicotinic acetylcholine receptor $\alpha 7$ subunit gene as a candidate for catatonic schizophrenia in a large family supporting the chromosome 15q13.2 locus. <i>Molecular Psychiatry</i> , 2002, 7, 220-223.	4.1	24
277	<i>FCRL3</i> genotype is associated with acute coronary syndromes as first manifestation of coronary artery disease. <i>Atherosclerosis</i> , 2009, 205, 512-516.	0.4	24
278	Mutations in the mitochondrial gene C12ORF65 lead to syndromic autosomal recessive intellectual disability and show genotype phenotype correlation. <i>European Journal of Medical Genetics</i> , 2013, 56, 599-602.	0.7	24
279	Cost-effectiveness of risk-reducing surgeries in preventing hereditary breast and ovarian cancer. <i>Breast</i> , 2017, 32, 186-191.	0.9	24
280	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. <i>Genetics in Medicine</i> , 2020, 22, 538-546.	1.1	24
281	V76D mutation in a conserved gD-crystallin region leads to dominant cataracts in mice. <i>Mammalian Genome</i> , 2002, 13, 452-455.	1.0	23
282	Apolipoprotein E Genotypes in Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. <i>Journal of Glaucoma</i> , 2010, 19, 561-565.	0.8	23
283	Loss of function of SVBP leads to autosomal recessive intellectual disability, microcephaly, ataxia, and hypotonia. <i>Genetics in Medicine</i> , 2019, 21, 1790-1796.	1.1	23
284	A YAC Contig Spanning the Nevroid Basal Cell Carcinoma Syndrome, Fanconi Anaemia Group C, and Xeroderma Pigmentosum Group A Loci on Chromosome 9q. <i>Genomics</i> , 1994, 23, 23-29.	1.3	22
285	Localisation of a Fanconi anaemia gene to chromosome 9p. <i>European Journal of Human Genetics</i> , 1998, 6, 501-508.	1.4	22
286	Assessment of association between variants and haplotypes of the remaining TBX1 gene and manifestations of congenital heart defects in 22q11.2 deletion patients. <i>Journal of Medical Genetics</i> , 2004, 41, e40-e40.	1.5	22
287	Lack of Evidence for Genetic Association to RUNX1 Binding Site at PSORS2 in Different German Psoriasis Cohorts. <i>Journal of Investigative Dermatology</i> , 2005, 124, 107-110.	0.3	22
288	The protective variant rs7173049 at LOXL1 locus impacts on retinoic acid signaling pathway in pseudoexfoliation syndrome. <i>Human Molecular Genetics</i> , 2019, 28, 2531-2548.	1.4	22

#	ARTICLE	IF	CITATIONS
289	Urine Metabolite Levels, Adverse Kidney Outcomes, and Mortality in CKD Patients: A Metabolome-wide Association Study. <i>American Journal of Kidney Diseases</i> , 2021, 78, 669-677.e1.	2.1	22
290	Recurrent nasal polyps as a monosymptomatic form of cystic fibrosis associated with a novel in-frame deletion (591del18) in the CFTR gene. <i>Human Molecular Genetics</i> , 1995, 4, 1463-1464.	1.4	21
291	Loss-of-function mutations in the filaggrin gene: no contribution to disease susceptibility, but to autoantibody formation against citrullinated peptides in early rheumatoid arthritis. <i>Annals of the Rheumatic Diseases</i> , 2008, 67, 131-133.	0.5	21
292	Comprehensive screening for mutations associated with colorectal cancer in unselected cases reveals penetrant and nonpenetrant mutations. <i>International Journal of Cancer</i> , 2015, 136, E559-68.	2.3	21
293	MAP4-Dependent Regulation of Microtubule Formation Affects Centrosome, Cilia, and Golgi Architecture as a Central Mechanism in Growth Regulation. <i>Human Mutation</i> , 2015, 36, 87-97.	1.1	21
294	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. <i>PLoS ONE</i> , 2016, 11, e0167984.	1.1	21
295	Transcription Factor 4 and Myocyte Enhancer Factor 2C mutations are not common causes of Rett syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 713-719.	0.7	20
296	Posttranscriptional Regulation of LOXL1 Expression Via Alternative Splicing and Nonsense-Mediated mRNA Decay as an Adaptive Stress Response. , 2017, 58, 5930.		20
297	Evidence for genetic overlap between adult onset Still's disease and hereditary periodic fever syndromes. <i>Rheumatology International</i> , 2018, 38, 111-120.	1.5	20
298	TCF4 (E2-2) harbors tumor suppressive functions in SHH medulloblastoma. <i>Acta Neuropathologica</i> , 2019, 137, 657-673.	3.9	20
299	Localisation of a gene for an autosomal recessive syndrome of macrocephaly, multiple epiphyseal dysplasia, and distinctive facies to chromosome 15q26. <i>Journal of Medical Genetics</i> , 2001, 38, 369-373.	1.5	19
300	High post surgical opioid requirements in Crohn's disease are not due to a general change in pain sensitivity. <i>European Journal of Pain</i> , 2009, 13, 1036-1042.	1.4	19
301	SPATA5 mutations cause a distinct autosomal recessive phenotype of intellectual disability, hypotonia and hearing loss. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 130.	1.2	19
302	Targeted sequencing of FH-deficient uterine leiomyomas reveals biallelic inactivating somatic fumarase variants and allows characterization of missense variants. <i>Modern Pathology</i> , 2020, 33, 2341-2353.	2.9	19
303	A novel mutation and novel features in Nijmegen breakage syndrome. <i>Journal of Medical Genetics</i> , 2001, 38, 113-117.	1.5	19
304	Renal polyamine excretion, tubular amino acid reabsorption and molecular genetics in cystinuria. <i>Pediatric Nephrology</i> , 2000, 14, 376-384.	0.9	18
305	Novel mutations in the Charcot-Marie-Tooth disease genes PMP22, MPZ, and GJB1. <i>Human Mutation</i> , 2003, 21, 100-100.	1.1	18
306	Inhibition of RAS Activation Due to a Homozygous Ezrin Variant in Patients with Profound Intellectual Disability. <i>Human Mutation</i> , 2015, 36, 270-278.	1.1	18

#	ARTICLE	IF	CITATIONS
307	Glycaemic control and antidiabetic therapy in patients with diabetes mellitus and chronic kidney disease – cross-sectional data from the German Chronic Kidney Disease (GCKD) cohort. <i>BMC Nephrology</i> , 2016, 17, 59.	0.8	18
308	SWI/SNF protein expression status in fumarate hydratase-deficient renal cell carcinoma: immunohistochemical analysis of 32 tumors from 28 patients. <i>Human Pathology</i> , 2018, 77, 139-146.	1.1	18
309	De novo variants in MED12 cause X-linked syndromic neurodevelopmental disorders in 18 females. <i>Genetics in Medicine</i> , 2021, 23, 645-652.	1.1	18
310	Distribution patterns of the F508 mutation in the CFTR gene on CF-linked marker haplotypes in the German population. <i>Human Genetics</i> , 1990, 85, 421-422.	1.8	17
311	Assignment of the Gene for a New Hereditary Nail Disorder, Isolated Congenital Nail Dysplasia, to Chromosome 17p13. <i>Journal of Investigative Dermatology</i> , 2000, 115, 664-667.	0.3	17
312	Charcot-Marie-Tooth disease: a novel Tyr145Ser mutation in the myelin protein zero (MPZ, P0) gene causes different phenotypes in homozygous and heterozygous carriers within one family. <i>Neurogenetics</i> , 2003, 4, 191-197.	0.7	17
313	Prospective case control study on genetic association of apolipoprotein A2 with intraocular pressure. <i>British Journal of Ophthalmology</i> , 2004, 88, 581-582.	2.1	17
314	Clinical and electrophysiological characteristics of autosomal recessive axonal Charcot-Marie-Tooth disease (ARCMT2B) that maps to chromosome 19q13.3. <i>Neuromuscular Disorders</i> , 2004, 14, 301-306.	0.3	17
315	A de novo 7.6Mb tandem duplication of 14q32.2-qter associated with primordial short stature with neurosecretory growth hormone dysfunction, distinct facial anomalies and mild developmental delay. <i>European Journal of Medical Genetics</i> , 2008, 51, 362-367.	0.7	17
316	Do telomeres have a higher plasticity than thought? Results from the German Chronic Kidney Disease (GCKD) study as a high-risk population. <i>Experimental Gerontology</i> , 2015, 72, 162-166.	1.2	17
317	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. <i>European Journal of Medical Genetics</i> , 2018, 61, 363-368.	0.7	17
318	Promoter Polymorphism at -238 of the Tumor Necrosis Factor Alpha Gene is Not Associated with Early Onset Psoriasis when Tested by the Transmission Disequilibrium Test. <i>Journal of Investigative Dermatology</i> , 1999, 112, 514-515.	0.3	16
319	Evaluation of a putative major susceptibility locus for juvenile myoclonic epilepsy on chromosome 15q14. <i>Journal of Medical Genetics</i> , 1999, 88, 182-187.		16
320	Atypical clinical picture of the Nijmegen breakage syndrome associated with developmental abnormalities of the brain. <i>Journal of Medical Genetics</i> , 2001, 38, e3-e3.	1.5	16
321	Identification of low frequency TRAF3IP2 coding variants in psoriatic arthritis patients and functional characterization. <i>Arthritis Research and Therapy</i> , 2012, 14, R84.	1.6	16
322	A defect of CD16-positive monocytes can occur without disease. <i>Immunobiology</i> , 2013, 218, 169-174.	0.8	16
323	Addition of triple negativity of breast cancer as an indicator for germline mutations in predisposing genes increases sensitivity of clinical selection criteria. <i>BMC Cancer</i> , 2018, 18, 926.	1.1	16
324	Prenatal diagnosis of HNF1B-associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?. <i>Prenatal Diagnosis</i> , 2019, 39, 1136-1147.	1.1	16

#	ARTICLE	IF	CITATIONS
325	Mutant RAMP2 causes primary open-angle glaucoma via the CRLR-cAMP axis. <i>Genetics in Medicine</i> , 2019, 21, 2345-2354.	1.1	16
326	A novel human stem cell model for Coffinâ€“Siris syndrome-like syndrome reveals the importance of SOX11 dosage for neuronal differentiation and survival. <i>Human Molecular Genetics</i> , 2019, 28, 2589-2599.	1.4	16
327	Results from the German Chronic Kidney Disease (GCKD) study support association of relative telomere length with mortality in a large cohort of patients with moderate chronic kidney disease. <i>Kidney International</i> , 2020, 98, 488-497.	2.6	16
328	Early-onset parkinsonism in PPP2R5D-related neurodevelopmental disorder. <i>European Journal of Medical Genetics</i> , 2021, 64, 104123.	0.7	16
329	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021, 23, 888-899.	1.1	16
330	Association of Rare <i>CYP39A1</i> Variants With Exfoliation Syndrome Involving the Anterior Chamber of the Eye. <i>JAMA - Journal of the American Medical Association</i> , 2021, 325, 753.	3.8	16
331	Possible association of the allele status of the CS.7/ Hha I polymorphism 5â€² of the CFTR gene with postnatal female survival. <i>Human Genetics</i> , 1997, 99, 565-572.	1.8	15
332	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. <i>Genetics in Medicine</i> , 2020, 22, 797-802.	1.1	15
333	EthylNitrosourea-Induced Mutation in Mice Leads to the Expression of a Novel Protein in the Eye and to Dominant Cataracts. <i>Genetics</i> , 2001, 157, 1313-1320.	1.2	15
334	Physical and transcriptional map of the critical region for keratolytic winter erythema (KWE) on chromosome 8p22-p23 between D8S550 and D8S1759. <i>European Journal of Human Genetics</i> , 2002, 10, 17-25.	1.4	14
335	Genotypeâ€“epigenotypeâ€“phenotype correlations in females with frontometaphyseal dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1069-1073.	0.7	14
336	The Clinical Data Intelligence Project. <i>Informatik-Spektrum</i> , 2016, 39, 290-300.	1.0	14
337	Genetic Breast Cancer Susceptibility Variants and Prognosis in the Prospectively Randomized SUCCESS A Study. <i>Geburtshilfe Und Frauenheilkunde</i> , 2017, 77, 651-659.	0.8	14
338	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. <i>Genetics in Medicine</i> , 2022, 24, 1261-1273.	1.1	14
339	The critical region for Angelman syndrome lies between D15S122 and D15S113. <i>American Journal of Medical Genetics Part A</i> , 1994, 53, 396-398.	2.4	13
340	Genetic and Clinical Heterogeneity in Transgressive Palmoplantar Keratoderma. <i>Journal of Investigative Dermatology</i> , 2001, 116, 825-827.	0.3	13
341	Mutation Analysis of the Nijmegen Breakage Syndrome Gene NBS1 in Nineteen Patients with Acute Myeloid Leukemia with Complex Karyotypes. <i>Leukemia and Lymphoma</i> , 2003, 44, 1931-1934.	0.6	13
342	Mild variable Noonan syndrome in a family with a novel PTPN11 mutation. <i>European Journal of Medical Genetics</i> , 2007, 50, 43-47.	0.7	13

#	ARTICLE	IF	CITATIONS
343	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. <i>American Journal of Human Genetics</i> , 2020, 107, 544-554.	2.6	13
344	Genome sequencing in families with congenital limb malformations. <i>Human Genetics</i> , 2021, 140, 1229-1239.	1.8	13
345	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. <i>Journal of Medical Genetics</i> , 2022, 59, 965-975.	1.5	13
346	Fructose-1,6-Bisphosphatase: Genetic and Physical Mapping to Human Chromosome 9q22.3 and Evaluation in Non-Insulin-Dependent Diabetes Mellitus. <i>Genomics</i> , 1995, 29, 187-194.	1.3	12
347	Identification and Localization of a New Human Myotubularin-Related Protein Gene, MTMR8, on 8p22-p23. <i>Genomics</i> , 2001, 75, 6-8.	1.3	12
348	MAN1B1 Mutation Leads to a Recognizable Phenotype: A Case Report and Future Prospects. <i>Molecular Syndromology</i> , 2015, 6, 58-62.	0.3	12
349	KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. <i>Oncotarget</i> , 2017, 8, 95401-95411.	0.8	12
350	Pancreatic insufficiency and pulmonary disease in German and Slavic cystic fibrosis patients with the R347P mutation. <i>Human Mutation</i> , 1995, 6, 219-225.	1.1	11
351	Clinical presentation and mutation identification in the NBS1 gene in a boy with Nijmegen breakage syndrome. <i>Clinical Genetics</i> , 2000, 57, 384-387.	1.0	11
352	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. <i>Gene</i> , 2014, 538, 30-35.	1.0	11
353	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. <i>European Journal of Medical Genetics</i> , 2016, 59, 549-553.	0.7	11
354	A biallelic truncating AEBP1 variant causes connective tissue disorder in two siblings. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 50-56.	0.7	11
355	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. <i>European Journal of Human Genetics</i> , 2019, 27, 1061-1071.	1.4	11
356	Tyrosinase Is a Novel Endogenous Regulator of Developmental and Inflammatory Lymphangiogenesis. <i>American Journal of Pathology</i> , 2019, 189, 440-448.	1.9	11
357	Exclusion of TCOF1 mutations in a case of bilateral Goldenhar syndrome and one familial case of microtia with meatal atresia. <i>Clinical Dysmorphology</i> , 2005, 14, 67-71.	0.1	10
358	7 Mb de novo deletion within 8q21 in a patient with distal arthrogyriposis type 2B (DA2B). <i>European Journal of Medical Genetics</i> , 2011, 54, e495-e500.	0.7	10
359	Patients with unstable angina pectoris show an increased frequency of the Fc gamma RIIa R131 allele. <i>Autoimmunity</i> , 2012, 45, 556-564.	1.2	10
360	Whole exome sequencing reveals a novel de novo FOXC1 mutation in a patient with unrecognized Axenfeld-Rieger syndrome and glaucoma. <i>Gene</i> , 2015, 568, 76-80.	1.0	10

#	ARTICLE	IF	CITATIONS
361	Novel <i>STRA6</i> null mutations in the original family described with Matthew-Wood syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 134-138.	0.7	10
362	Role of Endogenous Regulators of Hem- And Lymphangiogenesis in Corneal Transplantation. <i>Journal of Clinical Medicine</i> , 2020, 9, 479.	1.0	10
363	Astrogenesis in the murine dentate gyrus is a life-long and dynamic process. <i>EMBO Journal</i> , 2022, 41, e110409.	3.5	10
364	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. <i>Kidney International</i> , 2022, 102, 405-420.	2.6	10
365	Frequency of the $\Delta F508$ mutation and flanking marker haplotypes at the CF locus from 167 Czech families. <i>Human Genetics</i> , 1990, 85, 417-418.	1.8	9
366	A novel locus for arterial hypertension on chromosome 1p36 maps to a metabolic syndrome trait cluster in the Sorbs, a Slavic population isolate in Germany*. <i>Journal of Hypertension</i> , 2009, 27, 983-990.	0.3	9
367	Microdeletions of chromosome 7p21, including <i>TWIST1</i> , associated with significant microcephaly, facial dysmorphism, and short stature. <i>European Journal of Medical Genetics</i> , 2011, 54, 256-261.	0.7	9
368	Biallelic <i>SEMA3A</i> defects cause a novel type of syndromic short stature. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2880-2889.	0.7	9
369	The proinflammatory effect of C-reactive protein on human endothelial cells depends on the <i>Fc<math>\gamma</math>RIIa</i> genotype. <i>Thrombosis Research</i> , 2014, 133, 426-432.	0.8	9
370	Replication of a distinct psoriatic arthritis risk variant at the <i>IL23R</i> locus. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 1417-1418.	0.5	9
371	BDV Syndrome: an Emerging Syndrome With Profound Obesity and Neurodevelopmental Delay Resembling Prader-Willi Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 3413-3427.	1.8	9
372	Homozygosity mapping in a family with microcephaly, mental retardation, and short stature to a Cohen syndrome region on 8q21.3-8q22.1: redefining a clinical entity. <i>American Journal of Medical Genetics Part A</i> , 2000, 92, 285-92.	2.4	9
373	Functional Characterization of a Novel <i>CFTR</i> Mutation P67S Identified in a Patient with Atypical Cystic Fibrosis. <i>Cellular Physiology and Biochemistry</i> , 2007, 19, 239-248.	1.1	8
374	De novo triplication of the <i>MAPT</i> gene from the recurrent 17q21.31 microdeletion region in a patient with moderate intellectual disability and various minor anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1765-1770.	0.7	8
375	Two novel distinct <i>COL1A2</i> mutations highlight the complexity of genotype-phenotype correlations in osteogenesis imperfecta and related connective tissue disorders. <i>European Journal of Medical Genetics</i> , 2013, 56, 669-673.	0.7	8
376	Genome-wide association and targeted analysis of copy number variants with psoriatic arthritis in German patients. <i>BMC Medical Genetics</i> , 2017, 18, 92.	2.1	8
377	Breast MRI texture analysis for prediction of BRCA-associated genetic risk. <i>BMC Medical Imaging</i> , 2020, 20, 86.	1.4	8
378	scRNA sequencing uncovers a TCF4-dependent transcription factor network regulating commissure development in mouse. <i>Development (Cambridge)</i> , 2021, 148, .	1.2	8

#	ARTICLE	IF	CITATIONS
379	Genetic interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in <i>Drosophila melanogaster</i> . <i>Scientific Reports</i> , 2020, 10, 1204.	1.6	8
380	Possible autosomal recessive inheritance of progressive hearing loss with stapes fixation.. <i>Journal of Medical Genetics</i> , 1996, 33, 597-599.	1.5	7
381	The direct early diagnosis of cystic fibrosis by the detection of the deltaF508 CFTR gene mutation in a prematurely delivered boy. <i>Clinical Genetics</i> , 1991, 39, 219-222.	1.0	7
382	Central nervous system anomalies in two females with Borjeson-Forsman-Lehmann syndrome. <i>Epilepsy and Behavior</i> , 2017, 69, 104-109.	0.9	7
383	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). <i>European Journal of Medical Genetics</i> , 2020, 63, 104004.	0.7	7
384	A case of severe autosomal recessive spinocerebellar ataxia type 18 with a novel nonsense variant in GRID2. <i>European Journal of Medical Genetics</i> , 2020, 63, 103998.	0.7	7
385	Sox11 is an Activity-Regulated Gene with Dentate-Gyrus-Specific Expression Upon General Neural Activation. <i>Cerebral Cortex</i> , 2020, 30, 3731-3743.	1.6	7
386	<i>De novo</i> missense variants in FBXO11 alter its protein expression and subcellular localization. <i>Human Molecular Genetics</i> , 2022, 31, 440-454.	1.4	7
387	Three dinucleotide microsatellite polymorphisms on human chromosome 13. <i>Human Molecular Genetics</i> , 1993, 2, 87-87.	1.4	6
388	Amelogenesis Imperfecta in a New Animal Model—a Mutation in Chromosome 5 (human 4q21). <i>Journal of Dental Research</i> , 2004, 83, 608-612.	2.5	6
389	A recessive form of extreme macrocephaly and mild intellectual disability complements the spectrum of PTEN hamartoma tumour syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 889-894.	1.4	6
390	A Homozygous Mutation in GPT2 Associated with Nonsyndromic Intellectual Disability in a Consanguineous Family from Costa Rica. <i>JIMD Reports</i> , 2017, 36, 59-66.	0.7	6
391	Novel truncating mutation in <i>CACNA1F</i> in a young male patient diagnosed with optic atrophy. <i>Ophthalmic Genetics</i> , 2018, 39, 741-748.	0.5	6
392	Cost effectiveness of bilateral risk-reducing mastectomy and salpingo-oophorectomy. <i>European Journal of Medical Research</i> , 2019, 24, 32.	0.9	6
393	Analyses of association of psoriatic arthritis and psoriasis vulgaris with functional NCF1 variants. <i>Rheumatology</i> , 2019, 58, 915-917.	0.9	6
394	Clinical validation of genetic variants associated with in vitro chemotherapy-related lymphoblastoid cell toxicity. <i>Oncotarget</i> , 2017, 8, 78133-78143.	0.8	6
395	Fine mapping of autosomal dominant nonsyndromic hearing impairment DFNA21 to chromosome 6p24.1-22.3. <i>American Journal of Medical Genetics, Part A</i> , 2005, 137A, 41-46.	0.7	5
396	Linkage studies exclude the AT gene(s) from the translocation breakpoints in an AT patient. <i>Clinical Genetics</i> , 1997, 51, 309-313.	1.0	5

#	ARTICLE	IF	CITATIONS
397	Familial short stature due to a 5q22.1â€“q23.2 duplication refines the 5q duplication spectrum. <i>European Journal of Medical Genetics</i> , 2011, 54, e521-e524.	0.7	5
398	Integrative bioinformatics analysis characterizing the role of EDC3 in mRNA decay and its association to intellectual disability. <i>BMC Medical Genomics</i> , 2018, 11, 41.	0.7	5
399	7q31.2q31.31 deletion downstream of <sc><i>FOXP2</i></sc> segregating in a family with speech and language disorder. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2737-2741.	0.7	5
400	<sc><i>QRICH1</i></sc> variants in <sc>Ververiâ€“Brady</sc> syndromeâ€”delineation of the genotypic and phenotypic spectrum. <i>Clinical Genetics</i> , 2021, 99, 199-207.	1.0	5
401	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 136.	1.2	5
402	<sc><i>ZMYND11</i></sc> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. <i>Clinical Genetics</i> , 2021, 100, 412-429.	1.0	5
403	Genotype-Phenotype Correlations in Cystic Fibrosis Patients. <i>Advances in Experimental Medicine and Biology</i> , 1991, 290, 97-103.	0.8	5
404	Biallelic<i>ANKS6</i> mutations cause late-onset ciliopathy with chronic kidney disease through YAP dysregulation. <i>Human Molecular Genetics</i> , 2022, 31, 1357-1369.	1.4	5
405	Microsatellite Haplotypes of Polish Cystic Fibrosis Alleles: &Delta;F508 Chromosomes Demonstrate a North-South Haplotype Frequency Gradient. <i>Human Heredity</i> , 1996, 46, 310-314.	0.4	4
406	Lack of genetic association of the interleukin-4 receptor single-nucleotide polymorphisms I50V and Q551R with erosive disease in psoriatic arthritis. <i>Arthritis and Rheumatism</i> , 2006, 54, 4023-4024.	6.7	4
407	Complete basal cell carcinoma remission with imiquimod in a patient with nevoid basal cell carcinoma syndrome and associated basal cell carcinoma of the scalp and invasive ductal breast cancer. <i>Journal of the American Academy of Dermatology</i> , 2011, 64, 611-613.	0.6	4
408	A new missense mutation in PLA2G6 gene among a family with infantile neuroaxonal dystrophy INAD. <i>The Gazette of the Egyptian Paediatric Association</i> , 2016, 64, 171-176.	0.1	4
409	Association analysis of psoriasis vulgaris and psoriatic arthritis with lossâ€“ofâ€“function mutations in IL36 RN in German patients. <i>British Journal of Dermatology</i> , 2016, 175, 639-641.	1.4	4
410	Drugs linked to plasma homoarginine in chronic kidney disease patientsâ€”a cross-sectional analysis of the German Chronic Kidney Disease cohort. <i>Nephrology Dialysis Transplantation</i> , 2020, 35, 1187-1195.	0.4	4
411	SRD5A3-CDG: Twins with an intragenic tandem duplication. <i>European Journal of Medical Genetics</i> , 2022, 65, 104492.	0.7	4
412	Severe, neonatal-onset OTC deficiency in twin sisters with a de novo balanced reciprocal translocation t(X;5)(p21.1;q11). <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 185-188.	0.7	3
413	GPFrontend and GPGraphics: graphical analysis tools for genetic association studies. <i>BMC Bioinformatics</i> , 2010, 11, 472.	1.2	3
414	Evaluation of conserved and ultraâ€“conserved nonâ€“coding sequences in chromosome 15q15â€“linked periodic catatonia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 77-86.	1.1	3



#	ARTICLE	IF	CITATIONS
415	Microphthalmia is not a mandatory finding in X-linked recessive syndromic microphthalmia caused by the recurrent <i>BCOR</i> variant p.Pro85Leu. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2872-2876.	0.7	3
416	CRISPR/Cas9 mediated generation of human ARID1B heterozygous knockout hESC lines to model Coffin-Siris syndrome. <i>Stem Cell Research</i> , 2020, 47, 101889.	0.3	3
417	Loss of PHF6 leads to aberrant development of human neuron-like cells. <i>Scientific Reports</i> , 2020, 10, 19030.	1.6	3
418	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. <i>Journal of Investigative Dermatology</i> , 2021, 141, 2079-2083.	0.3	3
419	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. <i>Journal of Molecular Medicine</i> , 2021, 99, 1755-1768.	1.7	3
420	Manifestation of epilepsy in a patient with <i>EED</i> -related overgrowth ( <i>Cohen-Gibson</i> syndrome). <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 292-297.	0.7	3
421	Uromodulin and its association with urinary metabolites: the German Chronic Kidney Disease Study. <i>Nephrology Dialysis Transplantation</i> , 2023, 38, 70-79.	0.4	3
422	Identification of Two Genetic Loci Associated with Leukopenia after Chemotherapy in Patients with Breast Cancer. <i>Clinical Cancer Research</i> , 2022, 28, 3342-3355.	3.2	3
423	Ectopic transcription of the parathyroid hormone gene in lymphocytes, lymphoblastoid cells and tumour tissue. <i>Journal of Endocrinology</i> , 1992, 135, 249-256.	1.2	2
424	Novel autosomal recessive progressive hyperpigmentation syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005, 135A, 195-199.	0.7	2
425	Genetik der Psoriasis. <i>Medizinische Genetik</i> , 2007, 19, 350-355.	0.1	2
426	Molecular diagnosis of kidney transplant failure based on urine. <i>American Journal of Transplantation</i> , 2020, 20, 1410-1416.	2.6	2
427	A novel splice variant expands the <i>LAMC3</i> associated cortical phenotype to frontal only polymicrogyria and adult-onset epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2761-2764.	0.7	2
428	Clinical and molecular delineation of spondylocostal dysostosis type 3. <i>Clinical Genetics</i> , 2021, 99, 851-852.	1.0	2
429	Dinucleotide repeat polymorphism at the locus D13S231. <i>Human Molecular Genetics</i> , 1993, 2, 1082-1082.	1.4	1
430	Response to Liu et al.. <i>American Journal of Human Genetics</i> , 2010, 86, 500.	2.6	1
431	Mikrozephalie bei psychomotorischen Entwicklungsstörungen und geistiger Behinderung. <i>Medizinische Genetik</i> , 2015, 27, 362-368.	0.1	1
432	OP0128...PTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 116.3-117.	0.5	1

#	ARTICLE	IF	CITATIONS
433	O53.â€fPTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. Rheumatology, 2015, , .	0.9	1
434	Diagnostik seltener Erkrankungen mit â€žnext generation sequencingâ€œ â€“ angekommen oder abgewehrt?. Medizinische Genetik, 2019, 31, 335-343.	0.1	1
435	Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. BMC Cancer, 2019, 19, 435.	1.1	1
436	A noninvasive diagnostic approach to retrospective donor HLA typing in kidney transplant patients using urine. Transplant International, 2021, 34, 1226-1238.	0.8	1
437	Phenotypic differences in Angelman syndrome patients: Imprinting mutations show less frequently microcephaly and hypopigmentation than deletions. American Journal of Medical Genetics Part A, 1996, 66, 221-226.	2.4	1
438	Korrelation zwischen Patched-Genmutation und klinischem Bild bei BasalzellnÃvussyndrom (Gorlin-Syndrom). , 2000, , 559-561.		1
439	Palmoplantar Keratoderma VÃrner-Unna-Thost. , 2009, , 1560-1561.		0
440	Chromosomale Ursachen der geistigen Behinderung. Medizinische Genetik, 2009, 21, 237-245.	0.1	0
441	Neue Entwicklungen in der Psoriasisgenetik. Medizinische Genetik, 2009, 21, 498-504.	0.1	0
442	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. Genetical Research, 2015, 97, e19.	0.3	0
443	SAT0011â€...Replication of A Distinct Psoriatic Arthritis Risk Variant at IL23R. Annals of the Rheumatic Diseases, 2016, 75, 667.3-668.	0.5	0
444	Genetik der allgemeinen kognitiven FÃhigkeit. Medizinische Genetik, 2018, 30, 306-317.	0.1	0
445	X-chromosomale EntwicklungsstÃrungen im weiblichen Geschlecht. Medizinische Genetik, 2018, 30, 334-341.	0.1	0
446	Dissecting the Genetic Component of Complex Diseases in Humans. , 2002, , 1-15.		0
447	Heart-Type Fatty Acid Binding Protein, Cardiovascular Outcomes, and Death: Findings From the German CKD Cohort Study. American Journal of Kidney Diseases, 2022, , .	2.1	0