AndrÃ© Reis

List of Publications by Year
in descending order

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	Uromodulin and its association with urinary metabolites: the German Chronic Kidney Disease Study. Nephrology Dialysis Transplantation, 2023, 38, 70-79.	0.4	3
2	De novo missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454.	1.4	7
3	Manifestation of epilepsy in a patient with <scp>EED</scp>â€related overgrowth (<scp>Cohenâ€“Gibson</scp> syndrome). American Journal of Medical Genetics, Part A, 2022, 188, 292-297.	0.7	3
4	BiallelicANKS6mutations cause late-onset ciliopathy with chronic kidney disease through YAP dysregulation. Human Molecular Genetics, 2022, 31, 1357-1369.	1.4	5
5	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	1.5	13
6	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
7	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
8	SRD5A3-CDG: Twins with an intragenic tandem duplication. European Journal of Medical Genetics, 2022, 65, 104492.	0.7	4
9	Astrogenesis in the murine dentate gyrus is a lifeâ€long and dynamic process. EMBO Journal, 2022, 41, e110409.	3.5	10
10	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. Kidney International, 2022, 102, 405-420.	2.6	10
11	Identification of Two Genetic Loci Associated with Leukopenia after Chemotherapy in Patients with Breast Cancer. Clinical Cancer Research, 2022, 28, 3342-3355.	3.2	3
12	<scp>QRICH1</scp> variants in <scp>Ververiâ€Brady</scp> syndromeâ€”delineation of the genotypic and phenotypic spectrum. Clinical Genetics, 2021, 99, 199-207.	1.0	5
13	De novo variants in MED12 cause X-linked syndromic neurodevelopmental disorders in 18 females. Genetics in Medicine, 2021, 23, 645-652.	1.1	18
14	Early-onset parkinsonism in PPP2R5D-related neurodevelopmental disorder. European Journal of Medical Genetics, 2021, 64, 104123.	0.7	16
15	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
16	Association of Rare CYP39A1 Variants With Exfoliation Syndrome Involving the Anterior Chamber of the Eye. JAMA - Journal of the American Medical Association, 2021, 325, 753.	3.8	16
17	Clinical and molecular delineation of spondylocostal dysostosis type 3. Clinical Genetics, 2021, 99, 851-852.	1.0	2
18	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	1.2	5

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19	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
20	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
21	Urine Metabolite Levels, Adverse Kidney Outcomes, and Mortality in CKD Patients: A Metabolome-wide Association Study. American Journal of Kidney Diseases, 2021, 78, 669-677.e1.	2.1	22
22	Mutations in BRCA1/2 and Other Panel Genes in Patients With Metastatic Breast Cancer â€"Association With Patient and Disease Characteristics and Effect on Prognosis. Journal of Clinical Oncology, 2021, 39, 1619-1630.	0.8	39
23	A noninvasive diagnostic approach to retrospective donor HLA typing in kidney transplant patients using urine. Transplant International, 2021, 34, 1226-1238.	0.8	1
24	Genome sequencing in families with congenital limb malformations. Human Genetics, 2021, 140, 1229-1239.	1.8	13
25	scRNA sequencing uncovers a TCF4-dependent transcription factor network regulating commissure development in mouse. Development (Cambridge), 2021, 148, .	1.2	8
26	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	1.0	5
27	Frequent LPA KIV-2 Variants Lower Lipoprotein(a) Concentrations and Protect Against Coronary Artery Disease. Journal of the American College of Cardiology, 2021, 78, 437-449.	1.2	34
28	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. Journal of Investigative Dermatology, 2021, 141, 2079-2083.	0.3	3
29	BDV Syndrome: an Emerging Syndrome With Profound Obesity and Neurodevelopmental Delay Resembling Prader-Willi Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 3413-3427.	1.8	9
30	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	1.7	3
31	Drugs linked to plasma homoarginine in chronic kidney disease patientsâ€"a cross-sectional analysis of the German Chronic Kidney Disease cohort. Nephrology Dialysis Transplantation, 2020, 35, 1187-1195.	0.4	4
32	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	1.1	24
33	Molecular diagnosis of kidney transplant failure based on urine. American Journal of Transplantation, 2020, 20, 1410-1416.	2.6	2
34	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	1.1	15
35	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	1.1	63
36	CRISPR/Cas9 mediated generation of human ARID1B heterozygous knockout hESC lines to model Coffin-Siris syndrome. Stem Cell Research, 2020, 47, 101889.	0.3	3

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37	Targeted sequencing of FH-deficient uterine leiomyomas reveals biallelic inactivating somatic fumarase variants and allows characterization of missense variants. Modern Pathology, 2020, 33, 2341-2353.	2.9	19
38	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	2.6	13
39	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. American Journal of Human Genetics, 2020, 107, 527-538.	2.6	53
40	Breast MRI texture analysis for prediction of BRCA-associated genetic risk. BMC Medical Imaging, 2020, 20, 86.	1.4	8
41	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	0.7	7
42	A novel splice variant expands the LAMC3 â€ªassociated cortical phenotype to frontal only polymicrogyria and adultâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2020, 182, 2761-2764.	0.7	2
43	7q31.2q31.31 deletion downstream of <scp>FOXP2</scp> segregating in a family with speech and language disorder. American Journal of Medical Genetics, Part A, 2020, 182, 2737-2741.	0.7	5
44	Loss of PHF6 leads to aberrant development of human neuron-like cells. Scientific Reports, 2020, 10, 19030.	1.6	3
45	A case of severe autosomal recessive spinocerebellar ataxia type 18 with a novel nonsense variant in GRID2. European Journal of Medical Genetics, 2020, 63, 103998.	0.7	7
46	Sox11 is an Activity-Regulated Gene with Dentate-Gyrus-Specific Expression Upon General Neural Activation. Cerebral Cortex, 2020, 30, 3731-3743.	1.6	7
47	Role of Endogenous Regulators of Hem- And Lymphangiogenesis in Corneal Transplantation. Journal of Clinical Medicine, 2020, 9, 479.	1.0	10
48	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. Journal of Investigative Dermatology, 2020, 140, 1451-1455.e13.	0.3	48
49	Transcription factor Tcf4 is the preferred heterodimerization partner for Olig2 in oligodendrocytes and required for differentiation. Nucleic Acids Research, 2020, 48, 4839-4857.	6.5	31
50	Further delineation of the female phenotype with <scp>KDM5C</scp> disease causing variants: 19 new individuals and review of the literature. Clinical Genetics, 2020, 98, 43-55.	1.0	28
51	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. Kidney International, 2020, 98, 488-497.	2.6	16
52	Genetic interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in Drosophila melanogaster. Scientific Reports, 2020, 10, 1204.	1.6	8
53	A biallelic truncating AEBP1 variant causes connective tissue disorder in two siblings. American Journal of Medical Genetics, Part A, 2019, 179, 50-56.	0.7	11
54	Encephalopathies with KCNC1 variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272.	1.7	33

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55	Mitochondrial DNA copy number is associated with mortality and infections in a large cohort of patients with chronic kidney disease. Kidney International, 2019, 96, 480-488.	2.6	53
56	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	1.1	48
57	Diagnostik seltener Erkrankungen mit â€žnext generation sequencingâ€œ â€“ angekommen oder abgewehrt?. Medizinische Genetik, 2019, 31, 335-343.	0.1	1
58	Prenatal diagnosis of HNF1Bâ€associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?. Prenatal Diagnosis, 2019, 39, 1136-1147.	1.1	16
59	Cost effectiveness of bilateral risk-reducing mastectomy and salpingo-oophorectomy. European Journal of Medical Research, 2019, 24, 32.	0.9	6
60	TRIM28 haploinsufficiency predisposes to Wilms tumor. International Journal of Cancer, 2019, 145, 941-951.	2.3	45
61	Inflammation-induced glycolytic switch controls suppressivity of mesenchymal stem cells via STAT1 glycosylation. Leukemia, 2019, 33, 1783-1796.	3.3	54
62	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	1.0	63
63	Prevalence ofFOXC1Variants in Individuals With a Suspected Diagnosis of Primary Congenital Glaucoma. JAMA Ophthalmology, 2019, 137, 348.	1.4	33
64	Intellectual disability and autism spectrum disorders â€˜on the flyâ€™: insights from Drosophila. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	38
65	Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. BMC Cancer, 2019, 19, 435.	1.1	1
66	Mutant RAMP2 causes primary open-angle glaucoma via the CRLR-cAMP axis. Genetics in Medicine, 2019, 21, 2345-2354.	1.1	16
67	A novel human stem cell model for Coffinâ€“Siris syndrome-like syndrome reveals the importance of SOX11 dosage for neuronal differentiation and survival. Human Molecular Genetics, 2019, 28, 2589-2599.	1.4	16
68	Mutations in PIK3C2A cause syndromic short stature, skeletal abnormalities, and cataracts associated with ciliary dysfunction. PLoS Genetics, 2019, 15, e1008088.	1.5	45
69	The protective variant rs7173049 at LOXL1 locus impacts on retinoic acid signaling pathway in pseudoexfoliation syndrome. Human Molecular Genetics, 2019, 28, 2531-2548.	1.4	22
70	TCF4 (E2-2) harbors tumor suppressive functions in SHH medulloblastoma. Acta Neuropathologica, 2019, 137, 657-673.	3.9	20
71	Analyses of association of psoriatic arthritis and psoriasis vulgaris with functional NCF1 variants. Rheumatology, 2019, 58, 915-917.	0.9	6
72	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. Orphanet Journal of Rare Diseases, 2019, 14, 38.	1.2	48

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73	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	1.4	11
74	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	1.5	43
75	Loss of function of SVBP leads to autosomal recessive intellectual disability, microcephaly, ataxia, and hypotonia. Genetics in Medicine, 2019, 21, 1790-1796.	1.1	23
76	Tyrosinase Is a Novel Endogenous Regulator of Developmental and Inflammatory Lymphangiogenesis. American Journal of Pathology, 2019, 189, 440-448.	1.9	11
77	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	1.1	133
78	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. European Journal of Medical Genetics, 2018, 61, 363-368.	0.7	17
79	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	2.6	63
80	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in Drosophila. American Journal of Human Genetics, 2018, 102, 44-57.	2.6	49
81	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.	1.1	56
82	SWI/SNF protein expression status in fumarate hydrataseâ€“deficient renal cell carcinoma: immunohistochemical analysis of 32 tumors from 28 patients. Human Pathology, 2018, 77, 139-146.	1.1	18
83	Single molecule real time sequencing in ADTKD-MUC1 allows complete assembly of the VNTR and exact positioning of causative mutations. Scientific Reports, 2018, 8, 4170.	1.6	40
84	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	1.1	101
85	Novel STRA6 null mutations in the original family described with Matthewâ€“Wood syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 134-138.	0.7	10
86	Evidence for genetic overlap between adult onset Stillâ€™s disease and hereditary periodic fever syndromes. Rheumatology International, 2018, 38, 111-120.	1.5	20
87	Genetik der allgemeinen kognitiven FÃhigkeit. Medizinische Genetik, 2018, 30, 306-317.	0.1	0
88	Microphthalmia is not a mandatory finding in Xâ€linked recessive syndromic microphthalmia caused by the recurrent BCOR variant p.Pro85Leu. American Journal of Medical Genetics, Part A, 2018, 176, 2872-2876.	0.7	3
89	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	5.8	70
90	X-chromosomale EntwicklungsstÃ¶rungen im weiblichen Geschlecht. Medizinische Genetik, 2018, 30, 334-341.	0.1	0

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91	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	1.6	70
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.	2.6	89
93	Addition of triple negativity of breast cancer as an indicator for germline mutations in predisposing genes increases sensitivity of clinical selection criteria. BMC Cancer, 2018, 18, 926.	1.1	16
94	Novel truncating mutation in CACNA1F in a young male patient diagnosed with optic atrophy. Ophthalmic Genetics, 2018, 39, 741-748.	0.5	6
95	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	9.4	28
96	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	2.6	48
97	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. Journal of the American Society of Nephrology: JASN, 2018, 29, 2298-2309.	3.0	25
98	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.	3.3	168
99	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. Molecular Autism, 2018, 9, 20.	2.6	45
100	Integrative bioinformatics analysis characterizing the role of EDC3 in mRNA decay and its association to intellectual disability. BMC Medical Genomics, 2018, 11, 41.	0.7	5
101	The polynucleotide kinase 3â€²-phosphatase gene (PNKP) is involved in Charcot-Marie-Tooth disease (CMT2B2) previously related to MED25. Neurogenetics, 2018, 19, 215-225.	0.7	31
102	Risk, Prediction and Prevention of Hereditary Breast Cancer â€“ Large-Scale Genomic Studies in Times of Big and Smart Data. Geburtshilfe Und Frauenheilkunde, 2018, 78, 481-492.	0.8	38
103	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	6.0	186
104	A Homozygous Mutation in GPT2 Associated with Nonsyndromic Intellectual Disability in a Consanguineous Family from Costa Rica. JIMD Reports, 2017, 36, 59-66.	0.7	6
105	Cost-effectiveness of risk-reducing surgeries in preventing hereditary breast and ovarian cancer. Breast, 2017, 32, 186-191.	0.9	24
106	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases. JAMA Oncology, 2017, 3, 636.	3.4	376
107	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	1.5	67
108	Hypomorphic Pathogenic Variants in TAF13 Are Associated with Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2017, 100, 555-561.	2.6	26

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109	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	1.5	35
110	Pseudoexfoliation syndrome-associated genetic variants affect transcription factor binding and alternative splicing of LOXL1. Nature Communications, 2017, 8, 15466.	5.8	57
111	Haploinsufficiency of NR4A2 is associated with a neurodevelopmental phenotype with prominent language impairment. American Journal of Medical Genetics, Part A, 2017, 173, 2231-2234.	0.7	25
112	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. Nature Communications, 2017, 8, 15382.	5.8	251
113	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. Nature Genetics, 2017, 49, 993-1004.	9.4	114
114	DDX3X mutations in two girls with a phenotype overlapping Torielloâ€“Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	0.7	41
115	Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. Epilepsy and Behavior, 2017, 69, 104-109.	0.9	7
116	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. European Journal of Human Genetics, 2017, 25, 183-191.	1.4	35
117	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	9.4	186
118	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.	2.6	61
119	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	1.6	53
120	Genetic Breast Cancer Susceptibility Variants and Prognosis in the Prospectively Randomized SUCCESS A Study. Geburtshilfe Und Frauenheilkunde, 2017, 77, 651-659.	0.8	14
121	Exome Pool-Seq in neurodevelopmental disorders. European Journal of Human Genetics, 2017, 25, 1364-1376.	1.4	77
122	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. Nature Communications, 2017, 8, 15910.	5.8	77
123	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.	2.3	99
124	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. Human Molecular Genetics, 2017, 26, 4301-4313.	1.4	41
125	Genome-wide association and targeted analysis of copy number variants with psoriatic arthritis in German patients. BMC Medical Genetics, 2017, 18, 92.	2.1	8
126	Posttranscriptional Regulation of LOXL1 Expression Via Alternative Splicing and Nonsense-Mediated mRNA Decay as an Adaptive Stress Response. , 2017, 58, 5930.		20

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127	Clinical validation of genetic variants associated with in vitro chemotherapy-related lymphoblastoid cell toxicity. Oncotarget, 2017, 8, 78133-78143.	0.8	6
128	KIF3A and IL-4 are disease-specific biomarkers for psoriatic arthritis susceptibility. Oncotarget, 2017, 8, 95401-95411.	0.8	12
129	Glycaemic control and antidiabetic therapy in patients with diabetes mellitus and chronic kidney disease â€“ cross-sectional data from the German Chronic Kidney Disease (GCKD) cohort. BMC Nephrology, 2016, 17, 59.	0.8	18
130	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. PLoS ONE, 2016, 11, e0167984.	1.1	21
131	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. American Journal of Human Genetics, 2016, 99, 228-235.	2.6	44
132	Eight further individuals with intellectual disability and epilepsy carrying bi-allelicCNTNAP2aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827.	1.5	45
133	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	1.5	135
134	SAT0011â€…Replication of A Distinct Psoriatic Arthritis Risk Variant at IL23R. Annals of the Rheumatic Diseases, 2016, 75, 667.3-668.	0.5	0
135	A new missense mutation in PLA2G6 gene among a family with infantile neuroaxonal dystrophy INAD. The Gazette of the Egyptian Paediatric Association, 2016, 64, 171-176.	0.1	4
136	The Clinical Data Intelligence Project. Informatik-Spektrum, 2016, 39, 290-300.	1.0	14
137	Î±-Synuclein-induced myelination deficit defines a novel interventional target for multiple system atrophy. Acta Neuropathologica, 2016, 132, 59-75.	3.9	58
138	Expanding the clinical spectrum of COL1A1 mutations in different forms of glaucoma. Orphanet Journal of Rare Diseases, 2016, 11, 108.	1.2	26
139	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. European Journal of Medical Genetics, 2016, 59, 549-553.	0.7	11
140	Association analysis of psoriasis vulgaris and psoriatic arthritis with lossâ€ofâ€function mutations in IL 36 RN in German patients. British Journal of Dermatology, 2016, 175, 639-641.	1.4	4
141	GSK3ÃŸâ€dependent dysregulation of neurodevelopment in SPG11â€patient induced pluripotent stem cell model. Annals of Neurology, 2016, 79, 826-840.	2.8	40
142	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.	2.6	69
143	SPATA5 mutations cause a distinct autosomal recessive phenotype of intellectual disability, hypotonia and hearing loss. Orphanet Journal of Rare Diseases, 2016, 11, 130.	1.2	19
144	Replication of a distinct psoriatic arthritis risk variant at theIL23Rlocus. Annals of the Rheumatic Diseases, 2016, 75, 1417-1418.	0.5	9

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145	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	1.1	70
146	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. American Journal of Human Genetics, 2016, 98, 149-164.	2.6	270
147	A recessive form of extreme macrocephaly and mild intellectual disability complements the spectrum of PTEN hamartoma tumour syndrome. European Journal of Human Genetics, 2016, 24, 889-894.	1.4	6
148	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
149	DYNC2LI1 mutations broaden the clinical spectrum of dynein-2 defects. Scientific Reports, 2015, 5, 11649.	1.6	28
150	Mikrozephalie bei psychomotorischen EntwicklungsstÃ¶rungen und geistiger Behinderung. Medizinische Genetik, 2015, 27, 362-368.	0.1	1
151	OP0128â€...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, 116.3-117.	0.5	1
152	O53.â€ƒPTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. Rheumatology, 2015, , .	0.9	1
153	MAN1B1 Mutation Leads to a Recognizable Phenotype: A Case Report and Future Prospects. Molecular Syndromology, 2015, 6, 58-62.	0.3	12
154	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. Nature Communications, 2015, 6, 7001.	5.8	156
155	Whole exome sequencing reveals a novel de novo FOXC1 mutation in a patient with unrecognized Axenfeldâ€“Rieger syndrome and glaucoma. Gene, 2015, 568, 76-80.	1.0	10
156	Mutations in DCPS and EDC3 in autosomal recessive intellectual disability indicate a crucial role for mRNA decapping in neurodevelopment. Human Molecular Genetics, 2015, 24, 3172-3180.	1.4	40
157	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.	2.6	171
158	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.	0.5	64
159	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.	1.4	73
160	Comprehensive screening for mutations associated with colorectal cancer in unselected cases reveals penetrant and nonpenetrant mutations. International Journal of Cancer, 2015, 136, E559-68.	2.3	21
161	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. Nature Genetics, 2015, 47, 387-392.	9.4	97
162	MAP4-Dependent Regulation of Microtubule Formation Affects Centrosome, Cilia, and Golgi Architecture as a Central Mechanism in Growth Regulation. Human Mutation, 2015, 36, 87-97.	1.1	21

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163	Inhibition of RAS Activation Due to a Homozygous Ezrin Variant in Patients with Profound Intellectual Disability. Human Mutation, 2015, 36, 270-278.	1.1	18
164	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.	1.4	72
165	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.	0.4	132
166	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.	0.4	85
167	Do telomeres have a higher plasticity than thought? Results from the German Chronic Kidney Disease (GCKD) study as a high-risk population. Experimental Gerontology, 2015, 72, 162-166.	1.2	17
168	Genetic variants and cellular stressors associated with exfoliation syndrome modulate promoter activity of a lncRNA within theLOXL1locus. Human Molecular Genetics, 2015, 24, 6552-6563.	1.4	76
169	Chromatin-Remodeling-Factor ARID1B Represses Wnt/Î²-Catenin Signaling. American Journal of Human Genetics, 2015, 97, 445-456.	2.6	67
170	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.	2.6	245
171	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. Nature Communications, 2015, 6, 6046.	5.8	149
172	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.	1.8	93
173	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). ELife, 2015, 4, .	2.8	51
174	Null Mutation in PGAP1 Impairing Gpi-Anchor Maturation in Patients with Intellectual Disability and Encephalopathy. PLoS Genetics, 2014, 10, e1004320.	1.5	72
175	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.	2.6	86
176	AlteredGPM6A/M6Dosage Impairs Cognition and Causes Phenotypes Responsive to Cholesterol in Human andDrosophila. Human Mutation, 2014, 35, 1495-1505.	1.1	31
177	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	1.5	72
178	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	13.5	228
179	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Human Genetics, 2014, 133, 939-949.	1.8	29
180	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. Gene, 2014, 538, 30-35.	1.0	11

#	Article	IF	Citations
181	Deletions in the 3â€² Part of theNFIXGene Including a Recurrent Alu-Mediated Deletion of Exon 6 and 7 Account for Previously Unexplained Cases of Marshall-Smith Syndrome. Human Mutation, 2014, 35, 1092-1100.	1.1	26
182	HIBCH deficiency in a patient with phenotypic characteristics of mitochondrial disorders. American Journal of Medical Genetics, Part A, 2014, 164, 3162-3169.	0.7	27
183	Females with de novo aberrations in PHF6: Clinical overlap of Borjesonâ€“Forssmanâ€“Lehmann with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 290-301.	0.7	27
184	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	0.7	34
185	The proinflammatory effect of C-reactive protein on human endothelial cells depends on the FcÎ³RIIa genotype. Thrombosis Research, 2014, 133, 426-432.	0.8	9
186	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.	2.6	59
187	Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. Behavioral and Brain Functions, 2013, 9, 20.	1.4	47
188	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2013, 92, 575-583.	2.6	87
189	Mutations in the mitochondrial gene C12ORF65 lead to syndromic autosomal recessive intellectual disability and show genotype phenotype correlation. European Journal of Medical Genetics, 2013, 56, 599-602.	0.7	24
190	A defect of CD16-positive monocytes can occur without disease. Immunobiology, 2013, 218, 169-174.	0.8	16
191	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	2.6	151
192	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.	1.4	190
193	Two novel distinct COL1A2 mutations highlight the complexity of genotypeâ€“phenotype correlations in osteogenesis imperfecta and related connective tissue disorders. European Journal of Medical Genetics, 2013, 56, 669-673.	0.7	8
194	A new face of Borjesonâ€“Forssmanâ€“Lehmann syndrome? De novo mutations inPHF6in seven females with a distinct phenotype. Journal of Medical Genetics, 2013, 50, 838-847.	1.5	50
195	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	1.5	60
196	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
197	Biallelic SEMA3A defects cause a novel type of syndromic short stature. American Journal of Medical Genetics, Part A, 2013, 161, 2880-2889.	0.7	9
198	Common Genetic Determinants of Intraocular Pressure and Primary Open-Angle Glaucoma. PLoS Genetics, 2012, 8, e1002611.	1.5	164

#	Article	IF	Citations
199	Association of Î²-Defensin Copy Number and Psoriasis in Three Cohorts of European Origin. Journal of Investigative Dermatology, 2012, 132, 2407-2413.	0.3	50
200	Variants in ASB10 are associated with open-angle glaucoma. Human Molecular Genetics, 2012, 21, 1336-1349.	1.4	76
201	The German Chronic Kidney Disease (GCKD) study: design and methods. Nephrology Dialysis Transplantation, 2012, 27, 1454-1460.	0.4	127
202	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	6.3	940
203	Patients with unstable angina pectoris show an increased frequency of the Fc gamma RIIa R131 allele. Autoimmunity, 2012, 45, 556-564.	1.2	10
204	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
205	Identification of low frequency TRAF3IP2 coding variants in psoriatic arthritis patients and functional characterization. Arthritis Research and Therapy, 2012, 14, R84.	1.6	16
206	Transcription Factor 4 and Myocyte Enhancer Factor 2C mutations are not common causes of Rett syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 713-719.	0.7	20
207	De novo triplication of the MAPT gene from the recurrent 17q21.31 microdeletion region in a patient with moderate intellectual disability and various minor anomalies. American Journal of Medical Genetics, Part A, 2012, 158A, 1765-1770.	0.7	8
208	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.	2.6	225
209	Evaluation of conserved and ultraâ€conserved nonâ€genic sequences in chromosome 15q15â€linked periodic catatonia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 77-86.	1.1	3
210	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
211	Microdeletions of chromosome 7p21, including TWIST1, associated with significant microcephaly, facial dysmorphism, and short stature. European Journal of Medical Genetics, 2011, 54, 256-261.	0.7	9
212	7ÂMb de novo deletion within 8q21 in a patient with distal arthrogryposis type 2B (DA2B). European Journal of Medical Genetics, 2011, 54, e495-e500.	0.7	10
213	Familial short stature due to a 5q22.1â€“q23.2 duplication refines the 5q duplication spectrum. European Journal of Medical Genetics, 2011, 54, e521-e524.	0.7	5
214	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. Journal of the American Academy of Dermatology, 2011, 64, 611-613.	0.6	4
215	Genome-wide association study with DNA pooling identifies variants at CNTNAP2 associated with pseudoexfoliation syndrome. European Journal of Human Genetics, 2011, 19, 186-193.	1.4	56
216	Evidence for RPGRIP1 gene as risk factor for primary open angle glaucoma. European Journal of Human Genetics, 2011, 19, 445-451.	1.4	31

#	Article	IF	Citations
217	Genome-wide association study identifies a new locus for coronary artery disease on chromosome 10p11.23. European Heart Journal, 2011, 32, 158-168.	1.0	124
218	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	2.6	151
219	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.	2.6	206
220	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106.	2.1	109
221	Evaluation of risk loci for schizophrenia derived from genome-wide association studies in a German population. , 2011, 156, 198-203.		26
222	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: A study in Spanish and Italian populations and meta-analysis. Arthritis and Rheumatism, 2011, 63, 1860-1865.	6.7	31
223	Tumor necrosis factor promoter polymorphism TNF*-857 is a risk allele for psoriatic arthritis independent of the PSORS1 locus. Arthritis and Rheumatism, 2011, 63, 3801-3806.	6.7	25
224	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.	1.4	84
225	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.	0.3	89
226	Common genetic variants associated with open-angle glaucoma. Human Molecular Genetics, 2011, 20, 2464-2471.	1.4	152
227	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.	0.3	58
228	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.	3.9	89
229	Apolipoprotein E Genotypes in Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. Journal of Glaucoma, 2010, 19, 561-565.	0.8	23
230	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.	0.3	82
231	Response to Liu etÂal.. American Journal of Human Genetics, 2010, 86, 500.	2.6	1
232	GPFrontend and GPGraphics: graphical analysis tools for genetic association studies. BMC Bioinformatics, 2010, 11, 472.	1.2	3
233	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.	1.1	163
234	Clinical variability and novel mutations in the NHEJ1 gene in patients with a Nijmegen breakage syndrome-like phenotype. Human Mutation, 2010, 31, 1059-1068.	1.1	27

#	Article	IF	Citations
235	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	9.4	431
236	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. Nature Genetics, 2010, 42, 996-999.	9.4	334
237	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	9.4	918
238	Heterozygous Loss-of-Function Variants inCYP1B1Predispose to Primary Open-Angle Glaucoma. , 2010, 51, 249.		57
239	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.	0.3	61
240	Deletion of LCE3C and LCE3B genes at PSORS4 does not contribute to susceptibility to psoriatic arthritis in German patients. Annals of the Rheumatic Diseases, 2010, 69, 876-878.	0.5	34
241	Genetic Regulation of Serum Phytosterol Levels and Risk of Coronary Artery Disease. Circulation: Cardiovascular Genetics, 2010, 3, 331-339.	5.1	141
242	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331.	1.5	126
243	Exploring Functional Candidate Genes for Genetic Association in German Patients with Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. , 2009, 50, 2796.		54
244	Palmoplantar Keratoderma VÃ¶rner-Unna-Thost. , 2009, , 1560-1561.		0
245	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.	0.7	68
246	Chromosomale Ursachen der geistigen Behinderung. Medizinische Genetik, 2009, 21, 237-245.	0.1	0
247	Neue Entwicklungen in der Psoriasisgenetik. Medizinische Genetik, 2009, 21, 498-504.	0.1	0
248	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.	0.3	97
249	High post surgical opioid requirements in Crohn's disease are not due to a general change in pain sensitivity. European Journal of Pain, 2009, 13, 1036-1042.	1.4	19
250	Heterozygous NTF4 Mutations Impairing Neurotrophin-4 Signaling in Patients with Primary Open-Angle Glaucoma. American Journal of Human Genetics, 2009, 85, 447-456.	2.6	134
251	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.	2.6	573
252	FcÎ³RIIa genotype is associated with acute coronary syndromes as first manifestation of coronary artery disease. Atherosclerosis, 2009, 205, 512-516.	0.4	24

#	Article	IF	Citations
253	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*. Journal of Hypertension, 2009, 27, 983-990.	0.3	9
254	Characterisation of psoriasis susceptibility locus 6 (PSORS6) in patients with early onset psoriasis and evidence for interaction with PSORS1. Journal of Medical Genetics, 2009, 46, 736-744.	1.5	34
255	Mutations in CYP1B1 cause primary congenital glaucoma by reduction of either activity or abundance of the enzyme. Human Mutation, 2008, 29, 1147-1153.	1.1	62
256	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	9.4	587
257	Loss-of-function mutations in the filaggrin gene: no contribution to disease susceptibility, but to autoantibody formation against citrullinated peptides in early rheumatoid arthritis. Annals of the Rheumatic Diseases, 2008, 67, 131-133.	0.5	21
258	Novel missense, insertion and deletion mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with congenital insensitivity to pain with anhidrosis. Neuromuscular Disorders, 2008, 18, 159-166.	0.3	31
259	Two novel mutations in the insulin binding subunit of the insulin receptor gene without insulin binding impairment in a patient with Rabsonâ€“Mendenhall syndrome. Molecular Genetics and Metabolism, 2008, 94, 356-362.	0.5	28
260	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. European Journal of Medical Genetics, 2008, 51, 362-367.	0.7	17
261	Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. Cell, 2008, 135, 37-48.	13.5	567
262	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.	1.9	118
263	Profiling ofWDR36Missense Variants in German Patients with Glaucoma. , 2008, 49, 270.		55
264	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	6.0	370
265	Identification of ZNF313 / RNF114 as a novel psoriasis susceptibility gene. Human Molecular Genetics, 2008, 17, 1938-1945.	1.4	176
266	Association ofLOXL1Common Sequence Variants in German and Italian Patients with Pseudoexfoliation Syndrome and Pseudoexfoliation Glaucoma. , 2008, 49, 1459.		114
267	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636.	1.5	72
268	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.	1.5	114
269	Functional Characterization of a Novel CFTR Mutation P67S Identified in a Patient with Atypical Cystic Fibrosis. Cellular Physiology and Biochemistry, 2007, 19, 239-248.	1.1	8
270	Mild variable Noonan syndrome in a family with a novel PTPN11 mutation. European Journal of Medical Genetics, 2007, 50, 43-47.	0.7	13

#	Article	IF	Citations
271	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.	2.6	195
272	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.	2.6	316
273	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001.	2.6	261
274	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.	2.6	78
275	Mutation in the Scyl1 gene encoding aminoâ€terminal kinaseâ€like protein causes a recessive form of spinocerebellar neurodegeneration. EMBO Reports, 2007, 8, 691-697.	2.0	63
276	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
277	Neurodevelopmental deficits in Pierson (microcoria-congenital nephrosis) syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 311-319.	0.7	52
278	Loss-of-Function Variants of the Filaggrin Gene Are Not Major Susceptibility Factors for Psoriasis Vulgaris or Psoriatic Arthritis in German Patients. Journal of Investigative Dermatology, 2007, 127, 1367-1370.	0.3	39
279	Genetik der Psoriasis. Medizinische Genetik, 2007, 19, 350-355.	0.1	2
280	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.	2.6	94
281	Genetic Basis and Pancreatic Biology of Johanson-Blizzard Syndrome. Endocrinology and Metabolism Clinics of North America, 2006, 35, 243-253.	1.2	44
282	Prenatal findings in four consecutive pregnancies with fetal Pierson syndrome, a newly defined congenital nephrosis syndrome. Prenatal Diagnosis, 2006, 26, 262-266.	1.1	30
283	Male Restricted Genetic Association of Variant R620W in PTPN22 with Psoriatic Arthritis. Journal of Investigative Dermatology, 2006, 126, 936-938.	0.3	28
284	Association between protein tyrosine phosphatase 22 variant R620W in conjunction with the HLAâ€“DRB1 shared epitope and humoral autoimmunity to an immunodominant epitope of cartilage-specific type II collagen in early rheumatoid arthritis. Arthritis and Rheumatism, 2006, 54, 82-89.	6.7	36
285	Lack of genetic association of the interleukin-4 receptor single-nucleotide polymorphisms I50V and Q551R with erosive disease in psoriatic arthritis. Arthritis and Rheumatism, 2006, 54, 4023-4024.	6.7	4
286	Genotypeâ€“epigenotypeâ€“phenotype correlations in females with frontometaphyseal dysplasia. American Journal of Medical Genetics, Part A, 2006, 140A, 1069-1073.	0.7	14
287	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.	0.7	343
288	Evidence for susceptibility determinant(s) to psoriasis vulgaris in or near PTPN22 in German patients. Journal of Medical Genetics, 2006, 43, 517-522.	1.5	31

#	Article	IF	Citations
289	Primary congenital glaucoma and Rieger's anomaly: extended haplotypes reveal founder effects for eight distinct CYP1B1 mutations. Molecular Vision, 2006, 12, 523-31.	1.1	37
290	Exclusion of TCOF1 mutations in a case of bilateral Goldenhar syndrome and one familial case of microtia with meatal atresia. Clinical Dysmorphology, 2005, 14, 67-71.	0.1	10
291	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.	9.4	252
292	Lack of Evidence for Genetic Association to RUNX1 Binding Site at PSORS2 in Different German Psoriasis Cohorts. Journal of Investigative Dermatology, 2005, 124, 107-110.	0.3	22
293	Systematic Linkage Disequilibrium Analysis of SLC12A8 at PSORS5 Confirms a Role in Susceptibility to Psoriasis Vulgaris. Journal of Investigative Dermatology, 2005, 125, 906-912.	0.3	38
294	Severe, neonatal-onset OTC deficiency in twin sisters with a de novo balanced reciprocal translocation t(X;5)(p21.1;q11). American Journal of Medical Genetics, Part A, 2005, 132A, 185-188.	0.7	3
295	Novel autosomal recessive progressive hyperpigmentation syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 195-199.	0.7	2
296	Fine mapping of autosomal dominant nonsyndromic hearing impairmentDFNA21 to chromosome 6p24.1-22.3. American Journal of Medical Genetics, Part A, 2005, 137A, 41-46.	0.7	5
297	Demonstration of two novelLAMB2 mutations in the original Pierson syndrome family reported 42 years ago. American Journal of Medical Genetics, Part A, 2005, 138A, 73-74.	0.7	39
298	The scoliosis (sco) mouse: a new allele of Pax1. Cytogenetic and Genome Research, 2005, 111, 16-26.	0.6	28
299	Lack of genetic association of the three more common polymorphisms of CARD15 with psoriatic arthritis and psoriasis in a German cohort. Annals of the Rheumatic Diseases, 2005, 64, 951-954.	0.5	29
300	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.	2.6	117
301	Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111.	0.7	121
302	Assessment of association between variants and haplotypes of the remaining TBX1 gene and manifestations of congenital heart defects in 22q11.2 deletion patients. Journal of Medical Genetics, 2004, 41, e40-e40.	1.5	22
303	Amelogenesis Imperfecta in a New Animal Modelâ€”a Mutation in Chromosome 5 (human 4q21). Journal of Dental Research, 2004, 83, 608-612.	2.5	6
304	Molecular karyotyping using an SNP array for genomewide genotyping. Journal of Medical Genetics, 2004, 41, 916-922.	1.5	106
305	Prospective case control study on genetic assocation of apolipoprotein Â2 with intraocular pressure. British Journal of Ophthalmology, 2004, 88, 581-582.	2.1	17
306	Mutation in the gene encoding lysosomal acid phosphatase (Acp2) causes cerebellum and skin malformation in mouse. Neurogenetics, 2004, 5, 229-238.	0.7	41

#	Article	IF	Citations
307	Divergent genetic and epigenetic post-zygotic isolation mechanisms in Mus and Peromyscus. Journal of Evolutionary Biology, 2004, 17, 453-460.	0.8	29
308	Congenital nephrosis, mesangial sclerosis, and distinct eye abnormalities with microcoria: An autosomal recessive syndrome. , 2004, 130A, 138-145.		110
309	Clinical and electrophysiological characteristics of autosomal recessive axonal Charcot-Marie-Tooth disease (ARCMT2B) that maps to chromosome 19q13.3. Neuromuscular Disorders, 2004, 14, 301-306.	0.3	17
310	Genotype-phenotype correlations in Noonan syndrome. Journal of Pediatrics, 2004, 144, 368-374.	0.9	227
311	Human laminin Î²2 deficiency causes congenital nephrosis with mesangial sclerosis and distinct eye abnormalities. Human Molecular Genetics, 2004, 13, 2625-2632.	1.4	443
312	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.	2.6	55
313	Allelic Heterogeneity in the COH1 Gene Explains Clinical Variabilityin Cohen Syndrome. American Journal of Human Genetics, 2004, 75, 138-145.	2.6	72
314	Mutations in Microcephalin Cause Aberrant Regulation of Chromosome Condensation. American Journal of Human Genetics, 2004, 75, 261-266.	2.6	292
315	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.	1.8	63
316	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. Neurogenetics, 2003, 4, 191-197.	0.7	17
317	Novel mutations in the Charcot-Marie-Tooth disease genes PMP22, MPZ, and GJB1. Human Mutation, 2003, 21, 100-100.	1.1	18
318	A novel 5q35.3 subtelomeric deletion syndrome. , 2003, 121A, 1-8.		32
319	Tumor necrosis factor receptor-associated periodic syndrome characterized by a mutation affecting the cleavage site of the receptor: Implications for pathogenesis. Arthritis and Rheumatism, 2003, 48, 2386-2388.	6.7	38
320	Association scan of the novel psoriasis susceptibility region on chromosome 19: evidence for both susceptible and protective loci. Experimental Dermatology, 2003, 12, 490-496.	1.4	26
321	Self-Healing Collodion Baby: a Dynamic Phenotype Explained by a Particular Transglutaminase-1 Mutation. Journal of Investigative Dermatology, 2003, 120, 224-228.	0.3	101
322	Interleukin-10 promoter polymorphism IL10.G and familial early onset psoriasis. British Journal of Dermatology, 2003, 149, 381-385.	1.4	25
323	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.	1.4	57
324	Mutation Analysis of the Nijmegen Breakage Syndrome GeneNBS1in Nineteen Patients with Acute Myeloid Leukemia with Complex Karyotypes. Leukemia and Lymphoma, 2003, 44, 1931-1934.	0.6	13

#	Article	IF	Citations
325	Genome Scan for Childhood and Adolescent Obesity in German Families. Pediatrics, 2003, 111, 321-327.	1.0	74
326	Genome-wide linkage reveals a locus for human essential (primary) hypertension on chromosome 12p. Human Molecular Genetics, 2003, 12, 1273-1277.	1.4	49
327	A locus on chromosome 15q for a dominantly inherited nemaline myopathy with core-like lesions. Brain, 2003, 126, 1545-1551.	3.7	54
328	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. Journal of Glaucoma, 2003, 12, 27-30.	0.8	29
329	Exclusion of the neuronal nicotinic acetylcholine receptor Î±7 subunit gene as a candidate for catatonic schizophrenia in a large family supporting the chromosome 15q13â€“22 locus. Molecular Psychiatry, 2002, 7, 220-223.	4.1	24
330	Physical and transcriptional map of the critical region for keratolytic winter erythema (KWE) on chromosome 8p22-p23 between D8S550 and D8S1759. European Journal of Human Genetics, 2002, 10, 17-25.	1.4	14
331	?Mowat-Wilson? syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomalies-mental retardation syndrome caused by mutations in the zinc finger homeo box 1B gene. American Journal of Medical Genetics Part A, 2002, 108, 177-181.	2.4	122
332	Familial interstitial 570 kbp deletion of theUBE3Agene region causing Angelman syndrome but not Prader-Willi syndrome. American Journal of Medical Genetics Part A, 2002, 111, 233-237.	2.4	52
333	Novel mutations in the MYOC/GLC1A gene in a large group of glaucoma patients. Human Mutation, 2002, 20, 479-480.	1.1	28
334	V76D mutation in a conserved gD-crystallin region leads to dominant cataracts in mice. Mammalian Genome, 2002, 13, 452-455.	1.0	23
335	Epidermolytic palmoplantar keratoderma of VÃ¶rner: re-evaluation of VÃ¶rner's original family and identification of a novel keratin 9 mutation. Archives of Dermatological Research, 2002, 294, 268-272.	1.1	39
336	A comprehensive linkage analysis for myocardial infarction and its related risk factors. Nature Genetics, 2002, 30, 210-214.	9.4	313
337	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.	9.4	316
338	Periodic catatonia: confirmation of linkage to chromosome 15 and further evidence for genetic heterogeneity. Human Genetics, 2002, 111, 323-330.	1.8	53
339	Dissecting the Genetic Component of Complex Diseases in Humans. , 2002, , 1-15.		0
340	Towards the genetic basis of periodic catatonia: pedigree sample for genome scan I and II. European Archives of Psychiatry and Clinical Neuroscience, 2001, 251, I25-I30.	1.8	34
341	Characterization of a Mutation in the Lens-specific MP70 Encoding Gene of the Mouse Leading to a Dominant Cataract. Experimental Eye Research, 2001, 73, 867-876.	1.2	52
342	Identification and Localization of a New Human Myotubularin-Related Protein Gene, MTMR8, on 8p22â€“p23. Genomics, 2001, 75, 6-8.	1.3	12

#	Article	IF	Citations
343	Epigenetic targeting in the mouse zygote marks DNA for later methylation: a mechanism for maternal effects in development. Mechanisms of Development, 2001, 103, 35-47.	1.7	53
344	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.	2.6	71
345	Identification, by Homozygosity Mapping, of a Novel Locus for Autosomal Recessive Congenital Ichthyosis on Chromosome 17p, and Evidence for Further Genetic Heterogeneity. American Journal of Human Genetics, 2001, 69, 216-222.	2.6	37
346	Cloning of the mouse dysferlin gene and genomic characterization of the SJL-Dysf mutation. NeuroReport, 2001, 12, 625-629.	0.6	52
347	Genetic and Clinical Heterogeneity in Transgressive Palmoplantar Keratoderma. Journal of Investigative Dermatology, 2001, 116, 825-827.	0.3	13
348	Comparative association analysis reveals that corneodesmosin is more closely associated with psoriasis than HLA-Cw*0602-B*5701 in German families. Tissue Antigens, 2001, 57, 440-446.	1.0	28
349	Assignment of PGL3 to chromosome 1 (q21-q23) in a family with autosomal dominant non-chromaffin paraganglioma. American Journal of Medical Genetics Part A, 2001, 98, 32-36.	2.4	38
350	Fine mapping and single nucleotide polymorphism association results of candidate genes for asthma and related phenotypes. Human Mutation, 2001, 18, 327-336.	1.1	54
351	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
352	Mutations in CAV3 cause mechanical hyperirritability of skeletal muscle in rippling muscle disease. Nature Genetics, 2001, 28, 218-219.	9.4	206
353	Spectrum of mutations and genotypeâ€“phenotype analysis in Currarino syndrome. European Journal of Human Genetics, 2001, 9, 599-605.	1.4	100
354	Localisation of a gene for an autosomal recessive syndrome of macrocephaly, multiple epiphyseal dysplasia, and distinctive facies to chromosome 15q26. Journal of Medical Genetics, 2001, 38, 369-373.	1.5	19
355	Atypical clinical picture of the Nijmegen breakage syndrome associated with developmental abnormalities of the brain. Journal of Medical Genetics, 2001, 38, e3-e3.	1.5	16
356	Ethylnitrosourea-Induced Mutation in Mice Leads to the Expression of a Novel Protein in the Eye and to Dominant Cataracts. Genetics, 2001, 157, 1313-1320.	1.2	15
357	A novel mutation and novel features in Nijmegen breakage syndrome. Journal of Medical Genetics, 2001, 38, 113-117.	1.5	19
358	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.	1.5	101
359	Mutations in the Nijmegen Breakage Syndrome gene (NBS1) in childhood acute lymphoblastic leukemia (ALL). Cancer Research, 2001, 61, 3570-2.	0.4	83
360	Aey2, a new mutation in the betaB2-crystallin-encoding gene of the mouse. Investigative Ophthalmology and Visual Science, 2001, 42, 1574-80.	3.3	31

#	Article	IF	Citations
361	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
362	Assignment of the Gene for a New Hereditary Nail Disorder, Isolated Congenital Nail Dysplasia, to Chromosome 17p13. Journal of Investigative Dermatology, 2000, 115, 664-667.	0.3	17
363	Non-syndromic autosomal dominant progressive non-specific mid-frequency sensorineural hearing impairment with childhood to late adolescence onset (DFNA21). Clinical Otolaryngology, 2000, 25, 45-54.	0.0	31
364	Clinical presentation and mutation identification in the NBS1 gene in a boy with Nijmegen breakage syndrome. Clinical Genetics, 2000, 57, 384-387.	1.0	11
365	Genome-wide, large-scale production of mutant mice by ENU mutagenesis. Nature Genetics, 2000, 25, 444-447.	9.4	658
366	A major susceptibility locus for atopic dermatitis maps to chromosome 3q21. Nature Genetics, 2000, 26, 470-473.	9.4	249
367	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.	1.4	84
368	Hereditary spastic paraplegia caused by mutations in the SPG4 gene. European Journal of Human Genetics, 2000, 8, 771-776.	1.4	77
369	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.	1.4	130
370	Renal polyamine excretion, tubular amino acid reabsorption and molecular genetics in cystinuria. Pediatric Nephrology, 2000, 14, 376-384.	0.9	18
371	Genome search for susceptibility loci of common idiopathic generalised epilepsies. Human Molecular Genetics, 2000, 9, 1465-1472.	1.4	147
372	Splitting Schizophrenia: Periodic Catatoniaâ€“Susceptibility Locus on Chromosome 15q15. American Journal of Human Genetics, 2000, 67, 1201-1207.	2.6	112
373	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.	2.6	105
374	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
375	A Gene for Hypotrichosis Simplex of the Scalp Maps to Chromosome 6p21.3. American Journal of Human Genetics, 2000, 66, 1979-1983.	2.6	48
376	Localization of a Gene for Syndactyly Type 1 to Chromosome 2q34-q36. American Journal of Human Genetics, 2000, 67, 492-497.	2.6	47
377	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.	2.6	129
378	A European study on the genetics of mite sensitization. Journal of Allergy and Clinical Immunology, 2000, 106, 925-932.	1.5	48

#	Article	IF	Citations
379	Korrelation zwischen Patched-Genmutation und klinischem Bild bei BasalzellnÃvussyndrom (Gorlin-Syndrom). , 2000, , 559-561.		1
380	Splitting Schizophrenia: Periodic Catatoniaâ€“Susceptibility Locus on Chromosome 15q15. American Journal of Human Genetics, 2000, 67, 1201-1207.	2.6	192
381	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. American Journal of Medical Genetics Part A, 2000, 92, 285-92.	2.4	9
382	Promoter Polymorphism at â€“238 of the Tumor Necrosis Factor Alpha Gene is Not Associated with Early Onset Psoriasis when Tested by the Transmission Disequilibrium Test. Journal of Investigative Dermatology, 1999, 112, 514-515.	0.3	16
383	Dysferlin deletion in SJL mice (SJL-Dysf) defines a natural model for limb girdle muscular dystrophy 2B. Nature Genetics, 1999, 23, 141-142.	9.4	191
384	Evaluation of a putative major susceptibility locus for juvenile myoclonic epilepsy on chromosome 15q14. , 1999, 88, 182-187.		16
385	Nijmegen breakage syndrome: consequences of defective DNA double strand break repair. BioEssays, 1999, 21, 649-656.	1.2	83
386	Hereditary Isolated Renal Magnesium Loss Maps to Chromosome 11q23. American Journal of Human Genetics, 1999, 64, 180-188.	2.6	49
387	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.	2.6	88
388	The Fanconi Anemia Group E Gene, FANCE, Maps to Chromosome 6p. American Journal of Human Genetics, 1999, 64, 1400-1405.	2.6	48
389	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.	2.6	66
390	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.	2.6	80
391	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.	2.6	135
392	A Genome-wide Search for Linkage to Asthma22See the Appendix.. Genomics, 1999, 58, 1-8.	1.3	332
393	Envoplakin, a Possible Candidate Gene for Focal NEPPK/Esophageal Cancer (TOC): The Integration of Genetic and Physical Maps of the TOC Region on 17q25. Genomics, 1999, 59, 234-242.	1.3	34
394	Mutation in the Î²A3/A1-Crystallin Encoding Gene Cryba1 Causes a Dominant Cataract in the Mouse. Genomics, 1999, 62, 67-73.	1.3	67
395	Linkage of Familial Euthyroid Goiter to the Multinodular Goiter-1 Locus and Exclusion of the Candidate Genes Thyroglobulin, Thyroperoxidase, and Na+/I- Symporter. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 3750-3756.	1.8	42
396	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel ÃŸ1 subunit gene SCN1B. Nature Genetics, 1998, 19, 366-370.	9.4	965

#	Article	IF	Citations
397	Localisation of a Fanconi anaemia gene to chromosome 9p. European Journal of Human Genetics, 1998, 6, 501-508.	1.4	22
398	A novel in situ method for the detection of deficient transglutaminase activity in the skin. Archives of Dermatological Research, 1998, 290, 621-627.	1.1	73
399	Genetic and immunohistochemical detection of mutations inactivating the keratinocyte transglutaminase in patients with lamellar ichthyosis. Human Genetics, 1998, 102, 314-318.	1.8	28
400	Nibrin, a Novel DNA Double-Strand Break Repair Protein, Is Mutated in Nijmegen Breakage Syndrome. Cell, 1998, 93, 467-476.	13.5	989
401	Gene Localization for an Autosomal Dominant Familial Periodic Fever to 12p13. American Journal of Human Genetics, 1998, 62, 884-889.	2.6	92
402	Genotype/Phenotype Correlation in Autosomal Recessive Lamellar Ichthyosis. American Journal of Human Genetics, 1998, 62, 1052-1061.	2.6	77
403	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.	2.6	142
404	The Gene for Human Fibronectin Glomerulopathy Maps to 1q32, in the Region of the Regulation of Complement Activation Gene Cluster. American Journal of Human Genetics, 1998, 63, 1724-1731.	2.6	25
405	A Genome Wide Search for Susceptibility Loci in Three European Malignant Hyperthermia Pedigrees. Human Molecular Genetics, 1997, 6, 953-961.	1.4	117
406	Different Mechanisms and Recurrence Risks of Imprinting Defects in Angelman Syndrome. American Journal of Human Genetics, 1997, 61, 88-93.	2.6	60
407	Localization of the Gene Causing Keratolytic Winter Erythema to Chromosome 8p22-p23, and Evidence for a Founder Effect in South African Afrikaans-Speakers. American Journal of Human Genetics, 1997, 61, 370-378.	2.6	38
408	The Gene for Autosomal Dominant Craniometaphyseal Dysplasia Maps to Chromosome 5p and Is Distinct from the Growth Hormone-Receptor Gene. American Journal of Human Genetics, 1997, 61, 918-923.	2.6	39
409	Possible association of the allele status of the CS.7/ Hha I polymorphism 5â€² of the CFTR gene with postnatal female survival. Human Genetics, 1997, 99, 565-572.	1.8	15
410	Localisation of a gene for Papillon-LefÃ¨vre syndrome to chromosome 11q14-q21 by homozygosity mapping. Human Genetics, 1997, 101, 376-382.	1.8	66
411	Linkage studies exclude the ATâ€V gene(s) from the translocation breakpoints in an ATâ€V patient. Clinical Genetics, 1997, 51, 309-313.	1.0	5
412	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.	2.6	81
413	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.	1.4	132
414	Microsatellite Haplotypes of Polish Cystic Fibrosis Alleles: ΔF508 Chromosomes Demonstrate a North-South Haplotype Frequency Gradient. Human Heredity, 1996, 46, 310-314.	0.4	4

#	Article	IF	Citations
415	Autosomal dominant spastic paraplegia with anticipation maps to a 4-cM interval on chromosome 2p21-p24 in a large German family. Human Genetics, 1996, 98, 371-375.	1.8	35
416	Phenotypic differences in Angelman syndrome patients: Imprinting mutations show less frequently microcephaly and hypopigmentation than deletions. , 1996, 66, 221-226.		35
417	Possible autosomal recessive inheritance of progressive hearing loss with stapes fixation.. Journal of Medical Genetics, 1996, 33, 597-599.	1.5	7
418	Close mapping of the focal non-epidermolytic palmoplantar keratoderma (PPK) locus associated with oesophageal cancer (TOC). Human Molecular Genetics, 1996, 5, 857-860.	1.4	47
419	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
420	Pancreatic insufficiency and pulmonary disease in German and Slavic cystic fibrosis patients with the R347P mutation. Human Mutation, 1995, 6, 219-225.	1.1	11
421	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.	1.4	66
422	Recurrent nasal polyps as a monosymptomatic form of cystic fibrosis associated with a novel in-frame deletion (591del18) in the CFTR gene. Human Molecular Genetics, 1995, 4, 1463-1464.	1.4	21
423	Fructose-1,6-Bisphosphatase: Genetic and Physical Mapping to Human Chromosome 9q22.3 and Evaluation in Non-Insulin-Dependent Diabetes Mellitus. Genomics, 1995, 29, 187-194.	1.3	12
424	Palmoplantar Keratoderma in Association with Carcinoma of the Esophagus Maps to Chromosome 17q Distal to the Keratin Gene Cluster. Genomics, 1995, 29, 537-540.	1.3	49
425	The ataxia-telangiectasia-variant genes 1 and 2 are distinct from the ataxia-telangiectasia gene on chromosome 11q23.1. American Journal of Human Genetics, 1995, 57, 960-2.	2.6	30
426	Mutations in the pyruvate kinase L gene in patients with hereditary hemolytic anemia. Blood, 1994, 83, 2817-2822.	0.6	53
427	Trinucleotide repeat polymorphism at the PKLR locus. Human Molecular Genetics, 1994, 3, 523-523.	1.4	29
428	Keratin 9 gene mutational heterogeneity in patients with epidermolytic palmoplantar keratoderma. Human Genetics, 1994, 93, 649-54.	1.8	54
429	The critical region for Angelman syndrome lies between D15S122 and D15S113. American Journal of Medical Genetics Part A, 1994, 53, 396-398.	2.4	13
430	Keratin 9 gene mutations in epidermolytic palmoplantar keratoderma (EPPK). Nature Genetics, 1994, 6, 174-179.	9.4	255
431	The origin of the major cystic fibrosis mutation (Î”F508) in European populations. Nature Genetics, 1994, 7, 169-175.	9.4	323
432	A YAC Contig Spanning the Nevoid Basal Cell Carcinoma Syndrome, Fanconi Anaemia Group C, and Xeroderma Pigmentosum Group A Loci on Chromosome 9q. Genomics, 1994, 23, 23-29.	1.3	22

#	Article	IF	Citations
433	Analysis of 133 Meioses Places the Genes for Nevoid 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. Genomics, 1994, 23, 486-489.	1.3	36
434	Imprinting mutations suggested by abnormal DNA methylation patterns in familial Angelman and Prader-Willi syndromes. American Journal of Human Genetics, 1994, 54, 741-7.	2.6	179
435	Environmental trichlorfon and cluster of congenital abnormalities. Lancet, The, 1993, 341, 539-542.	6.3	92
436	Exclusion of the GABAA-receptor Î²3 subunit gene as the Angelman's syndrome gene. Lancet, The, 1993, 341, 122-123.	6.3	48
437	Dinucleotide repeat polymorphism at the locus D13S231. Human Molecular Genetics, 1993, 2, 1082-1082.	1.4	1
438	Three dinucleotide microsatellite polymorphisms on human chromosome 13. Human Molecular Genetics, 1993, 2, 87-87.	1.4	6
439	Ectopic transcription of the parathyroid hormone gene in lymphocytes, lymphoblastoid cells and tumour tissue. Journal of Endocrinology, 1992, 135, 249-256.	1.2	2
440	Localisation of gene for the naevoid basal-cell carcinoma syndrome. Lancet, The, 1992, 339, 617.	6.3	84
441	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.	1.8	82
442	Genetic influences in the formation of nasal polyps. Lancet, The, 1991, 337, 974.	6.3	35
443	The direct early diagnosis of cystic fibrosis by the detection of the deltaF508 CFTR gene mutation in a prematurely delivered boy. Clinical Genetics, 1991, 39, 219-222.	1.0	7
444	Genotype-Phenotype Correlations in Cystic Fibrosis Patients. Advances in Experimental Medicine and Biology, 1991, 290, 97-103.	0.8	5
445	Frequency of the Î”F508 mutation and flanking marker haplotypes at the CF locus from 167 Czech families. Human Genetics, 1990, 85, 417-418.	1.8	9
446	Distribution patterns of the Î”F508 mutation in the CFTR gene on CF-linked marker haplotypes in the German population. Human Genetics, 1990, 85, 421-422.	1.8	17
447	Genotype analysis of cystic fibrosis patients in relation to pancreatic sufficiency. Lancet, The, 1990, 335, 738-739.	6.3	28