Peter NÃ¼rnberg

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5854932/publications.pdf

Version: 2024-02-01

142 papers	12,368 citations	34105 52 h-index	27406 106 g-index
145 all docs	145 docs citations	145 times ranked	23795 citing authors

#	Article	IF	Citations
1	Loss-of-function variants in DNM1 cause a specific form of developmental and epileptic encephalopathy only in biallelic state. Journal of Medical Genetics, 2022, 59, 549-553.	3.2	9
2	Unraveling Structural Rearrangements of the CFH Gene Cluster in Atypical Hemolytic Uremic Syndrome Patients Using Molecular Combing and Long-Fragment Targeted Sequencing. Journal of Molecular Diagnostics, 2022, 24, 619-631.	2.8	5
3	De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 3, 100111.	1.7	7
4	Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034.	7.6	3
5	Transmission ratio distortion of mutations in the master regulator of centriole biogenesis PLK4. Human Genetics, 2022, 141, 1785-1794.	3.8	3
6	Monoallelic and biallelic variants in LEF1 are associated with a new syndrome combining ectodermal dysplasia and limb malformations caused by altered WNT signaling. Genetics in Medicine, 2022, 24, 1708-1721.	2.4	4
7	Deregulation and epigenetic modification of BCL2-family genes cause resistance to venetoclax in hematologic malignancies. Blood, 2022, 140, 2113-2126.	1.4	24
8	Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry. Cancers, 2022, 14, 3363.	3.7	2
9	WARS1 and SARS1 : Two tRNA synthetases implicated in autosomal recessive microcephaly. Human Mutation, 2022, 43, 1454-1471.	2.5	5
10	Prevalence of Cancer Predisposition Germline Variants in Male Breast Cancer Patients: Results of the German Consortium for Hereditary Breast and Ovarian Cancer. Cancers, 2022, 14, 3292.	3.7	11
11	Genotypeâ€“phenotype correlation in seven motor neuron disease families with novel <scp>ALS2</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 344-354.	1.2	14
12	Ultraâ€rapid emergency genomic diagnosis of Donahue syndrome in a preterm infant within 17â€‰hours. American Journal of Medical Genetics, Part A, 2021, 185, 90-96.	1.2	14
13	Consumptive coagulopathy is associated with a disturbed host response in patients with sepsis. Journal of Thrombosis and Haemostasis, 2021, 19, 1049-1063.	3.8	10
14	hiPSC-Derived Epidermal Keratinocytes from Ichthyosis Patients Show Altered Expression of Cornification Markers. International Journal of Molecular Sciences, 2021, 22, 1785.	4.1	4
15	Genomic variants causing mitochondrial dysfunction are common in hereditary lower motor neuron disease. Human Mutation, 2021, 42, 460-472.	2.5	6
16	A novel remitting leukodystrophy associated with a variant in FBP2. Brain Communications, 2021, 3, fcab036.	3.3	2
17	Clinical and genetic characterization of <scp>PYROXD1</scp>â€related myopathy patients from Turkey. American Journal of Medical Genetics, Part A, 2021, 185, 1678-1690.	1.2	5
18	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22

#	Article	IF	Citations
19	Modifier Genes in Microcephaly: A Report on WDR62, CEP63, RAD50 and PCNT Variants Exacerbating Disease Caused by Biallelic Mutations of ASPM and CENPJ. Genes, 2021, 12, 731.	2.4	8
20	Longâ€lived macrophage reprogramming drives spike proteinâ€mediated inflammasome activation in COVIDâ€19. EMBO Molecular Medicine, 2021, 13, e14150.	6.9	98
21	A 24â€generationâ€old founder mutation impairs splicing of <scp>RBBP8</scp> in Pakistani families affected with Jawad syndrome. Clinical Genetics, 2021, 100, 486-488.	2.0	1
22	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	2.4	11
23	Cystatin M/E Variant Causes Autosomal Dominant Keratosis Follicularis Spinulosa Decalvans by Dysregulating Cathepsins L and V. Frontiers in Genetics, 2021, 12, 689940.	2.3	5
24	A Novel Missense Mutation in TNNI3K Causes Recessively Inherited Cardiac Conduction Disease in a Consanguineous Pakistani Family. Genes, 2021, 12, 1282.	2.4	5
25	Biallelic SYNE2 Missense Mutations Leading to Nesprin-2 Giant Hypo-Expression Are Associated with Intellectual Disability and Autism. Genes, 2021, 12, 1294.	2.4	6
26	A Homozygous AKNA Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. Genes, 2021, 12, 1494.	2.4	3
27	Biallelic variants in YRDC cause a developmental disorder with progeroid features. Human Genetics, 2021, 140, 1679-1693.	3.8	3
28	MFSD2A-associated primary microcephaly - Expanding the clinical and mutational spectrum of this ultra-rare disease. European Journal of Medical Genetics, 2021, 64, 104310.	1.3	2
29	Familial cleft tongue caused by a unique translation initiation codon variant in TP63. European Journal of Human Genetics, 2021, , .	2.8	7
30	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. Nature Genetics, 2021, 53, 1673-1685.	21.4	61
31	Novel Lysosomal Positioning Defects Due to Biallelic Mutations in BORCS7 Causes a Neurodegenerative Disease Presenting as Hereditary-Spastic Paraplegia. Neuropediatrics, 2021, 52, .	0.6	0
32	A novel missense variant of SCN4A coâ€segregates with congenital essential tremor in a consanguineous Kurdish family. American Journal of Medical Genetics, Part A, 2021, , .	1.2	1
33	Reconstruction of rearranged Tâ€cell receptor loci by whole genome and transcriptome sequencing gives insights into the initial steps of Tâ€cell prolymphocytic leukemia. Genes Chromosomes and Cancer, 2020, 59, 261-267.	2.8	16
34	The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523.	2.4	35
35	An update of pathogenic variants in ASPM, WDR62, CDK5RAP2, STIL, CENPJ, and CEP135 underlying autosomal recessive primary microcephaly in 32 consanguineous families from Pakistan. Molecular Genetics & Genomic Medicine, 2020, 8, e1408.	1.2	18
36	Pseudouridylation defect due to DKC1 and NOP10 mutations causes nephrotic syndrome with cataracts, hearing impairment, and enterocolitis. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 15137-15147.	7.1	32

#	Article	IF	Citations
37	Mutation in <scp>CEP135</scp> causing primary microcephaly and subcortical heterotopia. American Journal of Medical Genetics, Part A, 2020, 182, 2450-2453.	1.2	6
38	Comprehensive molecular analysis of 61 Egyptian families with hereditary nonsyndromic hearing loss. Clinical Genetics, 2020, 98, 32-42.	2.0	22
39	Homozygosity for the c.428delG variant in KIAA0586 in a healthy individual: implications for molecular testing in patients with Joubert syndrome. Journal of Medical Genetics, 2019, 56, 261-264.	3.2	15
40	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	5.3	41
41	Novel mutations in SLC6A5 with benign course in hyperekplexia. Journal of Physical Education and Sports Management, 2019, 5, a004465.	1.2	10
42	First confirmatory study on PTPRQ as an autosomal dominant non-syndromic hearing loss gene. Journal of Translational Medicine, 2019, 17, 351.	4.4	10
43	Distinct genetic variation and heterogeneity of the Iranian population. PLoS Genetics, 2019, 15, e1008385.	3.5	34
44	Homozygous NMNAT2 mutation in sisters with polyneuropathy and erythromelalgia. Experimental Neurology, 2019, 320, 112958.	4.1	48
45	Novel mutations in KMT2B offer pathophysiological insights into childhood-onset progressive dystonia. Journal of Human Genetics, 2019, 64, 803-813.	2.3	25
46	Variant Score Rankerâ€”a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	4.1	5
47	Clonal dynamics towards the development of venetoclax resistance in chronic lymphocytic leukemia. Nature Communications, 2018, 9, 727.	12.8	160
48	HMGB2 Loss upon Senescence Entry Disrupts Genomic Organization and Induces CTCF Clustering across Cell Types. Molecular Cell, 2018, 70, 730-744.e6.	9.7	164
49	Integrative genomic profiling of large-cell neuroendocrine carcinomas reveals distinct subtypes of high-grade neuroendocrine lung tumors. Nature Communications, 2018, 9, 1048.	12.8	254
50	A C-terminal nonsense mutation links PTPRQ with autosomal-dominant hearing loss, DFNA73. Genetics in Medicine, 2018, 20, 614-621.	2.4	21
51	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	89
52	Absence of Goniodysgenesis in Patients with Chromosome 13Q Microdeletion-Related Microcoria. Ophthalmology Glaucoma, 2018, 1, 145-147.	1.9	3
53	A mechanistic classification of clinical phenotypes in neuroblastoma. Science, 2018, 362, 1165-1170.	12.6	213
54	IG-MYC+ neoplasms with precursor B-cell phenotype are molecularly distinct from Burkitt lymphomas. Blood, 2018, 132, 2280-2285.	1.4	50

#	Article	IF	Citations
55	Bi-allelic Mutations in LSS, Encoding Lanosterol Synthase, Cause Autosomal-Recessive Hypotrichosis Simplex. American Journal of Human Genetics, 2018, 103, 777-785.	6.2	55
56	Rare gene deletions in genetic generalized and Rolandic epilepsies. PLoS ONE, 2018, 13, e0202022.	2.5	6
57	Homozygous mutation in TXNRD1 is associated with genetic generalized epilepsy. Free Radical Biology and Medicine, 2017, 106, 270-277.	2.9	31
58	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
59	CDK5RAP2 interaction with components of the Hippo signaling pathway may play a role in primary microcephaly. Molecular Genetics and Genomics, 2017, 292, 365-383.	2.1	18
60	Association Between Loss-of-Function Mutations Within the FANCM Gene and Early-Onset Familial Breast Cancer. JAMA Oncology, 2017, 3, 1245.	7.1	74
61	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	6.2	56
62	Mutations of KIF14 cause primary microcephaly by impairing cytokinesis. Annals of Neurology, 2017, 82, 562-577.	5.3	62
63	Classification of patients with sepsis according to blood genomic endotype: a prospective cohort study. Lancet Respiratory Medicine,the, 2017, 5, 816-826.	10.7	381
64	Germline Mutation Status, Pathological Complete Response, and Disease-Free Survival in Triple-Negative Breast Cancer. JAMA Oncology, 2017, 3, 1378.	7.1	300
65	A new CUL4B variant associated with a mild phenotype and an exceptional pattern of leukoencephalopathy. American Journal of Medical Genetics, Part A, 2017, 173, 2803-2807.	1.2	7
66	P385â€...Hypotonic infant with riboflavin transporter deficiency due to slc52a2 mutations. , 2017, , .		0
67	Prevalence of deleterious germline variants in risk genes including BRCA1/2 in consecutive ovarian cancer patients (AGO-TR-1). PLoS ONE, 2017, 12, e0186043.	2.5	105
68	PEX6 is Expressed in Photoreceptor Cilia and Mutated in Deafblindness with Enamel Dysplasia and Microcephaly. Human Mutation, 2016, 37, 170-174.	2.5	36
69	A syndrome of microcephaly, short stature, polysyndactyly, and dental anomalies caused by a homozygous KATNB1 mutation. American Journal of Medical Genetics, Part A, 2016, 170, 728-733.	1.2	13
70	Thrombocytopenia is associated with a dysregulated host response in critically ill sepsis patients. Blood, 2016, 127, 3062-3072.	1.4	224
71	Complex karyotypes and KRAS and POT1 mutations impact outcome in CLL after chlorambucil-based chemotherapy or chemoimmunotherapy. Blood, 2016, 128, 395-404.	1.4	112
72	The progressive ankylosis protein ANK facilitates clathrin- and adaptor-mediated membrane traffic at the trans-Golgi network-to-endosome interface. Human Molecular Genetics, 2016, 25, 3836-3848.	2.9	10

#	Article	IF	Citations
73	A large deletion in RPGR causes XLPRA in Weimaraner dogs. Canine Genetics and Epidemiology, 2016, 3, 7.	2.8	18
74	Mutations in Three Genes Encoding Proteins Involved in Hair Shaft Formation Cause Uncombable Hair Syndrome. American Journal of Human Genetics, 2016, 99, 1292-1304.	6.2	127
75	Novel IFT122 mutations in three Argentinian patients with cranioectodermal dysplasia: Expanding the mutational spectrum. American Journal of Medical Genetics, Part A, 2016, 170, 1295-1301.	1.2	17
76	A genomic view on epilepsy and autism candidate genes. Genomics, 2016, 108, 31-36.	2.9	11
77	Neuropathological signs of inflammation correlate with mitochondrial DNA deletions in mesial temporal lobe epilepsy. Acta Neuropathologica, 2016, 132, 277-288.	7.7	37
78	Transcription factor activating protein 2 beta (TFAP2B) mediates noradrenergic neuronal differentiation in neuroblastoma. Molecular Oncology, 2016, 10, 344-359.	4.6	36
79	Incidence, Risk Factors, and Attributable Mortality of Secondary Infections in the Intensive Care Unit After Admission for Sepsis. JAMA - Journal of the American Medical Association, 2016, 315, 1469.	7.4	367
80	Loss of the smallest subunit of cytochrome c oxidase, COX8A, causes Leigh-like syndrome and epilepsy. Brain, 2016, 139, 338-345.	7.6	44
81	Exome sequencing and CRISPR/Cas genome editing identify mutations of ZAK as a cause of limb defects in humans and mice. Genome Research, 2016, 26, 183-191.	5.5	52
82	A novel homozygous splicing mutation of CASC5 causes primary microcephaly in a large Pakistani family. Human Genetics, 2016, 135, 157-170.	3.8	31
83	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43.	21.4	74
84	Tumor suppression in basal keratinocytes via dual non-cell-autonomous functions of a Na,K-ATPase beta subunit. ELife, 2016, 5, .	6.0	25
85	Mutations in <scp>CDK</scp> 5 <scp>RAP</scp> 2 cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480.	1.2	55
86	Microcephaly, ectodermal dysplasia, multiple skeletal anomalies and distinctive facial appearance: Delineation of cerebroâ€dermatoâ€osseousâ€dysplasia. American Journal of Medical Genetics, Part A, 2015, 167, 842-851.	1.2	1
87	Rare variants in Î³â€aminobutyric acid type <scp>A</scp> receptor genes in rolandic epilepsy and related syndromes. Annals of Neurology, 2015, 77, 972-986.	5.3	51
88	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	1.6	44
89	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	6.2	188
90	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. Human Molecular Genetics, 2015, 24, 2594-2603.	2.9	32

#	Article	IF	Citations
91	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	5.5	85
92	OSBPL2 encodes a protein of inner and outer hair cell stereocilia and is mutated in autosomal dominant hearing loss (DFNA67). Orphanet Journal of Rare Diseases, 2015, 10, 15.	2.7	52
93	STIL mutation causes autosomal recessive microcephalic lobar holoprosencephaly. Human Genetics, 2015, 134, 45-51.	3.8	32
94	Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome. Molecular and Cellular Probes, 2015, 29, 330-334.	2.1	22
95	Comprehensive genomic profiles of small cell lung cancer. Nature, 2015, 524, 47-53.	27.8	1,634
96	Mutational dynamics between primary and relapse neuroblastomas. Nature Genetics, 2015, 47, 872-877.	21.4	253
97	Skeletal dysplasia in a consanguineous clan from the island of Nias/Indonesia is caused by a novel mutation in B3GAT3. Human Genetics, 2015, 134, 691-704.	3.8	27
98	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	3.5	91
99	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17.	2.9	26
100	Floral induction in Arabidopsis thaliana by FLOWERING LOCUS T requires direct repression of BLADE-ON-PETIOLE genes by homeodomain protein PENNYWISE. Plant Physiology, 2015, 169, pp.00960.2015.	4.8	51
101	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	7.6	96
102	Telomerase activation by genomic rearrangements in high-risk neuroblastoma. Nature, 2015, 526, 700-704.	27.8	478
103	De novo FUS mutations are the most frequent genetic cause inÂearly-onset German ALS patients. Neurobiology of Aging, 2015, 36, 3117.e1-3117.e6.	3.1	59
104	Leveraging the Power of High Performance Computing for Next Generation Sequencing Data Analysis: Tricks and Twists from a High Throughput Exome Workflow. PLoS ONE, 2015, 10, e0126321.	2.5	37
105	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). ELife, 2015, 4, .	6.0	51
106	Homozygosity Mapping and Whole Exome Sequencing Reveal a Novel Homozygous COL18A1 Mutation Causing Knobloch Syndrome. PLoS ONE, 2014, 9, e112747.	2.5	15
107	Autosomal dominant SCA5 and autosomal recessive infantile SCA are allelic conditions resulting from SPTBN2 mutations. European Journal of Human Genetics, 2014, 22, 286-288.	2.8	37
108	DEPDC5 mutations in genetic focal epilepsies of childhood. Annals of Neurology, 2014, 75, 788-792.	5.3	105

#	Article	IF	Citations
109	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	6.2	37
110	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
111	A homozygous splice-site mutation in CARS2 is associated with progressive myoclonic epilepsy. Neurology, 2014, 83, 2183-2187.	1.1	59
112	The missing â€œlinkâ€• an autosomal recessive short stature syndrome caused by a hypofunctional XYLT1 mutation. Human Genetics, 2014, 133, 29-39.	3.8	63
113	A missense mutation in the PISA domain of HsSAS-6 causes autosomal recessive primary microcephaly in a large consanguineous Pakistani family. Human Molecular Genetics, 2014, 23, 5940-5949.	2.9	63
114	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	6.2	34
115	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	21.4	178
116	Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. Nature Genetics, 2014, 46, 1283-1292.	21.4	156
117	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	21.4	165
118	Frequent mutations in chromatin-remodelling genes in pulmonary carcinoids. Nature Communications, 2014, 5, 3518.	12.8	239
119	Mutations in POGLUT1, Encoding Protein O-Glucosyltransferase 1, Cause Autosomal-Dominant Dowling-Degos Disease. American Journal of Human Genetics, 2014, 94, 135-143.	6.2	136
120	Exonic microdeletions of the gephyrin gene impair GABAergic synaptic inhibition in patients with idiopathic generalized epilepsy. Neurobiology of Disease, 2014, 67, 88-96.	4.4	51
121	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	21.4	391
122	CDK6 associates with the centrosome during mitosis and is mutated in a large Pakistani family with primary microcephaly. Human Molecular Genetics, 2013, 22, 5199-5214.	2.9	104
123	Impaired Epidermal Ceramide Synthesis Causes Autosomal Recessive Congenital Ichthyosis and Reveals the Importance of Ceramide Acyl Chain Length. Journal of Investigative Dermatology, 2013, 133, 2202-2211.	0.7	138
124	Rare exonic deletions of the <scp>RBFOX1</scp> gene increase risk of idiopathic generalized epilepsy. Epilepsia, 2013, 54, 265-271.	5.1	59
125	A Truncating Mutation of CEP135 Causes Primary Microcephaly and Disturbed Centrosomal Function. American Journal of Human Genetics, 2012, 90, 871-878.	6.2	153
126	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	21.4	201

#	Article	IF	Citations
127	Loss of Cav1.3 (CACNA1D) function in a human channelopathy with bradycardia and congenital deafness. Nature Neuroscience, 2011, 14, 77-84.	14.8	265
128	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	6.2	108
129	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
130	Three novel mutations in the ANK membrane protein cause craniometaphyseal dysplasia with variable conductive hearing loss. American Journal of Medical Genetics, Part A, 2010, 152A, 870-874.	1.2	20
131	HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599.	14.5	331
132	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nature Genetics, 2008, 40, 1113-1118.	21.4	217
133	Correlation between Genetic and Geographic Structure in Europe. Current Biology, 2008, 18, 1241-1248.	3.9	449
134	Detection of novel NF1 mutations and rapid mutation prescreening with Pyrosequencing. Electrophoresis, 2007, 28, 4295-4301.	2.4	14
135	New universal primers facilitate Pyrosequencingâ„¢. Electrophoresis, 2006, 27, 394-397.	2.4	19
136	SNP-Based Analysis of Genetic Substructure in the German Population. Human Heredity, 2006, 62, 20-29.	0.8	121
137	HaploPainter: a tool for drawing pedigrees with complex haplotypes. Bioinformatics, 2005, 21, 1730-1732.	4.1	256
138	ALOHOMORA: a tool for linkage analysis using 10K SNP array data. Bioinformatics, 2005, 21, 2123-2125.	4.1	170
139	A Genotype-Phenotype Correlation with Gender-Effect for Hearing Impairment Caused by <i>TECTA</i> Mutations. Cellular Physiology and Biochemistry, 2004, 14, 369-376.	1.6	56
140	Evaluation of a potential epigenetic biomarker by quantitative methyl-single nucleotide polymorphism analysis. Electrophoresis, 2002, 23, 4072-4079.	2.4	160
141	Heterozygous mutations in ANKH, the human ortholog of the mouse progressive ankylosis gene, result in craniometaphyseal dysplasia. Nature Genetics, 2001, 28, 37-41.	21.4	199
142	Complete Genomic Sequence of the Human PK-L/R-Gene Includes Four Intragenic Polymorphisms Defining Different Haplotype Backgrounds of Normal and Mutant PK-Genes. DNA Sequence, 1997, 8, 45-53.	0.7	17