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	Comprehensive genomic profiles of small cell lung cancer. Nature, 2015, 524, 47-53.	27.8	1,634
2	Telomerase activation by genomic rearrangements in high-risk neuroblastoma. Nature, 2015, 526, 700-704.	27.8	478
3	Correlation between Genetic and Geographic Structure in Europe. Current Biology, 2008, 18, 1241-1248.	3.9	449
4	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	21.4	391
5	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
6	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
7	HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599.	14.5	331
8	Germline Mutation Status, Pathological Complete Response, and Disease-Free Survival in Triple-Negative Breast Cancer. JAMA Oncology, 2017, 3, 1378.	7.1	300
9	Loss of Cav1.3 (CACNA1D) function in a human channelopathy with bradycardia and congenital deafness. Nature Neuroscience, 2011, 14, 77-84.	14.8	265
10	HaploPainter: a tool for drawing pedigrees with complex haplotypes. Bioinformatics, 2005, 21, 1730-1732.	4.1	256
11	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
12	Mutational dynamics between primary and relapse neuroblastomas. Nature Genetics, 2015, 47, 872-877.	21.4	253
13	Frequent mutations in chromatin-remodelling genes in pulmonary carcinoids. Nature Communications, 2014, 5, 3518.	12.8	239
14	Thrombocytopenia is associated with a dysregulated host response in critically ill sepsis patients. Blood, 2016, 127, 3062-3072.	1.4	224
15	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nature Genetics, 2008, 40, 1113-1118.	21.4	217
16	A mechanistic classification of clinical phenotypes in neuroblastoma. Science, 2018, 362, 1165-1170.	12.6	213
17	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	21.4	201
18	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

#	Article	IF	Citations
19	A Specific IFIH1 Gain-of-Function Mutation Causes Singleton-Merten Syndrome. American Journal of Human Genetics, 2015, 96, 275-282.	6.2	188
20	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	21.4	178
21	ALOHOMORA: a tool for linkage analysis using 10K SNP array data. Bioinformatics, 2005, 21, 2123-2125.	4.1	170
22	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	21.4	165
23	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
24	Evaluation of a potential epigenetic biomarker by quantitative methyl-single nucleotide polymorphism analysis. Electrophoresis, 2002, 23, 4072-4079.	2.4	160
25	Clonal dynamics towards the development of venetoclax resistance in chronic lymphocytic leukemia. Nature Communications, 2018, 9, 727.	12.8	160
26	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
27	A Truncating Mutation of CEP135 Causes Primary Microcephaly and Disturbed Centrosomal Function. American Journal of Human Genetics, 2012, 90, 871-878.	6.2	153
28	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
29	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
30	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
31	SNP-Based Analysis of Genetic Substructure in the German Population. Human Heredity, 2006, 62, 20-29.	0.8	121
32	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
33	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	6.2	108
34	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
35	DEPDC5 mutations in genetic focal epilepsies of childhood. Annals of Neurology, 2014, 75, 788-792.	5.3	105
36	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

#	Article	IF	Citations
37	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
38	Longâ€lived macrophage reprogramming drives spike proteinâ€mediated inflammasome activation in COVIDâ€19. EMBO Molecular Medicine, 2021, 13, e14150.	6.9	98
39	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
40	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	3.5	91
41	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	89
42	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	5.5	85
43	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. Nature Genetics, 2017, 49, 537-549.	21.4	81
44	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. Nature Genetics, 2016, 48, 36-43.	21.4	74
45	Association Between Loss-of-Function Mutations Within the FANCM Gene and Early-Onset Familial Breast Cancer. JAMA Oncology, 2017, 3, 1245.	7.1	74
46	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
47	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
48	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
49	Mutations of KIF14 cause primary microcephaly by impairing cytokinesis. Annals of Neurology, 2017, 82, 562-577.	5.3	62
50	Chromothripsis followed by circular recombination drives oncogene amplification in human cancer. Nature Genetics, 2021, 53, 1673-1685.	21.4	61
51	Rare exonic deletions of the <scp>RBFOX1</scp> gene increase risk of idiopathic generalized epilepsy. Epilepsia, 2013, 54, 265-271.	5.1	59
52	A homozygous splice-site mutation in CARS2 is associated with progressive myoclonic epilepsy. Neurology, 2014, 83, 2183-2187.	1.1	59
53	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
54	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

#	Article	IF	Citations
55	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
56	Mutations in <scp>CDK</scp> 5 <scp>RAP</scp> 2 cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480.	1.2	55
57	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
58	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
59	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
60	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
61	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
62	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
63	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). ELife, 2015, 4, .	6.0	51
64	IG-MYC+ neoplasms with precursor B-cell phenotype are molecularly distinct from Burkitt lymphomas. Blood, 2018, 132, 2280-2285.	1.4	50
65	Homozygous NMNAT2 mutation in sisters with polyneuropathy and erythromelalgia. Experimental Neurology, 2019, 320, 112958.	4.1	48
66	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	1.6	44
67	Loss of the smallest subunit of cytochrome c oxidase, COX8A, causes Leigh-like syndrome and epilepsy. Brain, 2016, 139, 338-345.	7.6	44
68	SSBP1 mutations in dominant optic atrophy with variable retinal degeneration. Annals of Neurology, 2019, 86, 368-383.	5.3	41
69	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
70	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	6.2	37
71	Neuropathological signs of inflammation correlate with mitochondrial DNA deletions in mesial temporal lobe epilepsy. Acta Neuropathologica, 2016, 132, 277-288.	7.7	37
72	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

#	Article	IF	Citations
73	PEX6 is Expressed in Photoreceptor Cilia and Mutated in Deafblindness with Enamel Dysplasia and Microcephaly. Human Mutation, 2016, 37, 170-174.	2.5	36
74	Transcription factor activating protein 2 beta (TFAP2B) mediates noradrenergic neuronal differentiation in neuroblastoma. Molecular Oncology, 2016, 10, 344-359.	4.6	36
75	The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523.	2.4	35
76	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
77	Distinct genetic variation and heterogeneity of the Iranian population. PLoS Genetics, 2019, 15, e1008385.	3.5	34
78	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
79	STIL mutation causes autosomal recessive microcephalic lobar holoprosencephaly. Human Genetics, 2015, 134, 45-51.	3.8	32
80	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
81	A novel homozygous splicing mutation of CASC5 causes primary microcephaly in a large Pakistani family. Human Genetics, 2016, 135, 157-170.	3.8	31
82	Homozygous mutation in TXNRD1 is associated with genetic generalized epilepsy. Free Radical Biology and Medicine, 2017, 106, 270-277.	2.9	31
83	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
84	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17.	2.9	26
85	Novel mutations in KMT2B offer pathophysiological insights into childhood-onset progressive dystonia. Journal of Human Genetics, 2019, 64, 803-813.	2.3	25
86	Tumor suppression in basal keratinocytes via dual non-cell-autonomous functions of a Na,K-ATPase beta subunit. ELife, 2016, 5, .	6.0	25
87	Deregulation and epigenetic modification of BCL2-family genes cause resistance to venetoclax in hematologic malignancies. Blood, 2022, 140, 2113-2126.	1.4	24
88	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
89	Comprehensive molecular analysis of 61 Egyptian families with hereditary nonsyndromic hearing loss. Clinical Genetics, 2020, 98, 32-42.	2.0	22
90	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22

#	Article	IF	Citations
91	A C-terminal nonsense mutation links PTPRQ with autosomal-dominant hearing loss, DFNA73. Genetics in Medicine, 2018, 20, 614-621.	2.4	21
92	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
93	New universal primers facilitate Pyrosequencingâ„¢. Electrophoresis, 2006, 27, 394-397.	2.4	19
94	A large deletion in RPGR causes XLPRA in Weimaraner dogs. Canine Genetics and Epidemiology, 2016, 3, 7.	2.8	18
95	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
96	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
97	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
98	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
99	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
100	Homozygosity Mapping and Whole Exome Sequencing Reveal a Novel Homozygous COL18A1 Mutation Causing Knobloch Syndrome. PLoS ONE, 2014, 9, e112747.	2.5	15
101	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
102	Detection of novel NF1 mutations and rapid mutation prescreening with Pyrosequencing. Electrophoresis, 2007, 28, 4295-4301.	2.4	14
103	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
104	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
105	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
106	A genomic view on epilepsy and autism candidate genes. Genomics, 2016, 108, 31-36.	2.9	11
107	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
108	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

#	Article	IF	Citations
109	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
110	Novel mutations in SLC6A5 with benign course in hyperekplexia. Journal of Physical Education and Sports Management, 2019, 5, a004465.	1.2	10
111	First confirmatory study on PTPRQ as an autosomal dominant non-syndromic hearing loss gene. Journal of Translational Medicine, 2019, 17, 351.	4.4	10
112	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
113	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
114	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
115	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
116	Familial cleft tongue caused by a unique translation initiation codon variant in TP63. European Journal of Human Genetics, 2021, , .	2.8	7
117	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
118	Rare gene deletions in genetic generalized and Rolandic epilepsies. PLoS ONE, 2018, 13, e0202022.	2.5	6
119	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
120	Genomic variants causing mitochondrial dysfunction are common in hereditary lower motor neuron disease. Human Mutation, 2021, 42, 460-472.	2.5	6
121	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
122	Variant Score Rankerâ€"a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	4.1	5
123	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
124	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
125	A Novel Missense Mutation in TNNI3K Causes Recessively Inherited Cardiac Conduction Disease in a Consanguineous Pakistani Family. Genes, 2021, 12, 1282.	2.4	5
126	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

#	Article	IF	Citations
127	WARS1 and SARS1 : Two tRNA synthetases implicated in autosomal recessive microcephaly. Human Mutation, 2022, 43, 1454-1471.	2.5	5
128	hiPSC-Derived Epidermal Keratinocytes from Ichthyosis Patients Show Altered Expression of Cornification Markers. International Journal of Molecular Sciences, 2021, 22, 1785.	4.1	4
129	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
130	Absence of Goniodysgenesis in Patients with Chromosome 13Q Microdeletion-Related Microcoria. Ophthalmology Glaucoma, 2018, 1, 145-147.	1.9	3
131	A Homozygous AKNA Frameshift Variant Is Associated with Microcephaly in a Pakistani Family. Genes, 2021, 12, 1494.	2.4	3
132	Biallelic variants in YRDC cause a developmental disorder with progeroid features. Human Genetics, 2021, 140, 1679-1693.	3.8	3
133	Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034.	7.6	3
134	Transmission ratio distortion of mutations in the master regulator of centriole biogenesis PLK4. Human Genetics, 2022, 141, 1785-1794.	3.8	3
135	A novel remitting leukodystrophy associated with a variant in FBP2. Brain Communications, 2021, 3, fcab036.	3.3	2
136	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
137	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
138	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
139	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
140	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
141	P385â€…Hypotonic infant with riboflavin transporter deficiency due to slc52a2 mutations. , 2017, , .		0
142	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