

Rolph Pfundt

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2674475/publications.pdf>

Version: 2024-02-01

127
papers

7,706
citations

66315

42
h-index

69214

77
g-index

135
all docs

135
docs citations

135
times ranked

14446
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	13.7	996
2	Refining analyses of copy number variation identifies specific genes associated with developmental delay. <i>Nature Genetics</i> , 2014, 46, 1063-1071.	9.4	583
3	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. <i>Nature Neuroscience</i> , 2016, 19, 1194-1196.	7.1	407
4	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	13.7	343
5	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
6	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. <i>Genetics in Medicine</i> , 2017, 19, 1055-1063.	1.1	220
7	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. <i>American Journal of Human Genetics</i> , 2014, 95, 173-182.	2.6	219
8	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. <i>Genetics in Medicine</i> , 2017, 19, 667-675.	1.1	143
9	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2016, 98, 541-552.	2.6	132
10	Novel genetic causes for cerebral visual impairment. <i>European Journal of Human Genetics</i> , 2016, 24, 660-665.	1.4	127
11	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 303-309.	2.6	125
12	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 100, 907-925.	2.6	125
13	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. <i>American Journal of Human Genetics</i> , 2017, 101, 466-477.	2.6	119
14	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. <i>PLoS Genetics</i> , 2017, 13, e1006864.	1.5	116
15	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. <i>American Journal of Human Genetics</i> , 2014, 95, 285-293.	2.6	110
16	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016, 24, 652-659.	1.4	108
17	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. <i>European Journal of Human Genetics</i> , 2017, 25, 591-599.	1.4	104
18	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. <i>American Journal of Human Genetics</i> , 2016, 98, 373-381.	2.6	95

#	ARTICLE	IF	CITATIONS
19	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. <i>European Journal of Human Genetics</i> , 2017, 25, 308-314.	1.4	90
20	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. <i>American Journal of Human Genetics</i> , 2018, 103, 1045-1052.	2.6	89
21	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. <i>American Journal of Human Genetics</i> , 2017, 101, 478-484.	2.6	84
22	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	1.1	80
23	Mutations in N-acetylglucosamine (O-GlcNAc) transferase in patients with X-linked intellectual disability. <i>Journal of Biological Chemistry</i> , 2017, 292, 12621-12631.	1.6	72
24	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. <i>European Journal of Human Genetics</i> , 2018, 26, 64-74.	1.4	72
25	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. <i>American Journal of Human Genetics</i> , 2015, 97, 493-500.	2.6	71
26	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
27	Phenotypic and molecular insights into CASK-related disorders in males. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 44.	1.2	68
28	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. <i>Nature Genetics</i> , 2016, 48, 877-887.	9.4	67
29	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. <i>Epilepsia</i> , 2019, 60, 155-164.	2.6	65
30	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. <i>American Journal of Human Genetics</i> , 2017, 100, 592-604.	2.6	61
31	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 101, 503-515.	2.6	61
32	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	2.6	59
33	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. <i>Journal of Medical Genetics</i> , 2018, 55, 104-113.	1.5	59
34	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	2.6	59
35	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. <i>Genetics in Medicine</i> , 2016, 18, 1158-1162.	1.1	58
36	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	2.6	56

#	ARTICLE	IF	CITATIONS
37	Loss-of-Function Mutations in YY1AP1 Lead to Grange Syndrome and a Fibromuscular Dysplasia-Like Vascular Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 21-30.	2.6	54
38	Further delineation of an entity caused by <i>CREBBP</i> and <i>EP300</i> mutations but not resembling Rubinstein-Taybi syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 862-876.	0.7	52
39	ACAN Gene Mutations in Short Children Born SGA and Response to Growth Hormone Treatment. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 1458-1467.	1.8	50
40	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. <i>Prenatal Diagnosis</i> , 2020, 40, 972-983.	1.1	49
41	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	2.6	48
42	The molecular and phenotypic spectrum of <i>IQSEC2</i> -related epilepsy. <i>Epilepsia</i> , 2016, 57, 1858-1869.	2.6	46
43	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. <i>Human Genetics</i> , 2018, 137, 375-388.	1.8	46
44	Next generation sequencing in synovial sarcoma reveals novel gene mutations. <i>Oncotarget</i> , 2015, 6, 34680-34690.	0.8	45
45	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. <i>American Journal of Human Genetics</i> , 2015, 96, 555-564.	2.6	45
46	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. <i>American Journal of Human Genetics</i> , 2017, 101, 139-148.	2.6	45
47	Mutations in PIK3C2A cause syndromic short stature, skeletal abnormalities, and cataracts associated with ciliary dysfunction. <i>PLoS Genetics</i> , 2019, 15, e1008088.	1.5	45
48	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. <i>European Journal of Human Genetics</i> , 2017, 25, 43-51.	1.4	44
49	<i>NBEA</i> : Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	2.8	44
50	<i>CREBBP</i> mutations in individuals without Rubinstein-Taybi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2681-2693.	0.7	43
51	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	5.8	43
52	Characterization of SETD1A haploinsufficiency in humans and <i>Drosophila</i> defines a novel neurodevelopmental syndrome. <i>Molecular Psychiatry</i> , 2021, 26, 2013-2024.	4.1	43
53	Association of AADAC Deletion and Gilles de la Tourette Syndrome in a Large European Cohort. <i>Biological Psychiatry</i> , 2016, 79, 383-391.	0.7	41
54	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 1195-1203.	2.6	37

#	ARTICLE	IF	CITATIONS
55	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. <i>Human Molecular Genetics</i> , 2018, 27, 3029-3045.	1.4	37
56	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. <i>American Journal of Human Genetics</i> , 2017, 101, 824-832.	2.6	36
57	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	2.6	36
58	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 403-412.	2.6	35
59	Accurate detection of clinically relevant uniparental disomy from exome sequencing data. <i>Genetics in Medicine</i> , 2020, 22, 803-808.	1.1	35
60	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. <i>Nature Communications</i> , 2020, 11, 595.	5.8	35
61	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in <i>Drosophila</i> . <i>European Journal of Human Genetics</i> , 2016, 24, 1145-1153.	1.4	34
62	Biallelic B3GALT6 mutations cause spondylodysplastic Ehlers-Danlos syndrome. <i>Human Molecular Genetics</i> , 2018, 27, 3475-3487.	1.4	34
63	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2019, 104, 758-766.	2.6	34
64	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
65	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 54-63.	1.4	32
66	Dutch genome diagnostic laboratories accelerated and improved variant interpretation and increased accuracy by sharing data. <i>Human Mutation</i> , 2019, 40, 2230-2238.	1.1	32
67	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. <i>European Journal of Human Genetics</i> , 2019, 27, 738-746.	1.4	32
68	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. <i>American Journal of Human Genetics</i> , 2020, 107, 311-324.	2.6	32
69	The clustering of functionally related genes contributes to CNV-mediated disease. <i>Genome Research</i> , 2015, 25, 802-813.	2.4	31
70	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	2.6	31
71	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
72	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	2.6	30

#	ARTICLE	IF	CITATIONS
73	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. <i>Nature Genetics</i> , 2018, 50, 1442-1451.	9.4	28
74	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	1.1	26
75	Neurodegenerative <i>VPS41</i> variants inhibit HOPS function and mTORC1-dependent TFEB/TFE3 regulation. <i>EMBO Molecular Medicine</i> , 2021, 13, e13258.	3.3	26
76	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. <i>Genetics in Medicine</i> , 2021, 23, 1474-1483.	1.1	24
77	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. <i>PLoS ONE</i> , 2014, 9, e112687.	1.1	23
78	Broadening the phenotypic spectrum of pathogenic LARP7 variants: two cases with intellectual disability, variable growth retardation and distinct facial features. <i>Journal of Human Genetics</i> , 2016, 61, 229-233.	1.1	23
79	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	2.6	23
80	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 1669-1691.	2.6	23
81	Chromosomal aberrations in cerebral visual impairment. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 677-684.	0.7	20
82	The epilepsy phenotypic spectrum associated with a recurrent <i>CUX2</i> variant. <i>Annals of Neurology</i> , 2018, 83, 926-934.	2.8	20
83	Clinical exome sequencing—Mistakes and caveats. <i>Human Mutation</i> , 2022, 43, 1041-1055.	1.1	20
84	Duplications of SLC1A3: Associated with ADHD and autism. <i>European Journal of Medical Genetics</i> , 2016, 59, 373-376.	0.7	19
85	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 701-708.	2.6	19
86	Holoprosencephaly and preaxial polydactyly associated with a 1.24 Mb duplication encompassing FBXW11 at 5q35.1. <i>Journal of Human Genetics</i> , 2006, 51, 721-726.	1.1	18
87	Somatic loss of polycystic disease genes contributes to the formation of isolated and polycystic liver cysts: Table 1. <i>Gut</i> , 2015, 64, 688-690.	6.1	18
88	Genome-wide investigation of an ID cohort reveals de novo 3' UTR variants affecting gene expression. <i>Human Genetics</i> , 2018, 137, 717-721.	1.8	18
89	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021, 108, 1692-1709.	2.6	18
90	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. <i>European Journal of Human Genetics</i> , 2016, 24, 392-399.	1.4	17

#	ARTICLE	IF	CITATIONS
91	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. <i>American Journal of Human Genetics</i> , 2018, 103, 786-793.	2.6	17
92	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. <i>European Journal of Human Genetics</i> , 2021, 29, 625-636.	1.4	17
93	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	2.6	17
94	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. <i>Genetics in Medicine</i> , 2021, 23, 2122-2137.	1.1	16
95	Germline and somatic mosaicism in a family with multiple endocrine neoplasia type 1 (MEN1) syndrome. <i>European Journal of Endocrinology</i> , 2019, 180, K15-K19.	1.9	16
96	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 1774-1780.	1.1	16
97	Copy number variation analysis and methylome profiling of a GNAQ-mutant primary meningeal melanocytic tumor and its liver metastasis. <i>Experimental and Molecular Pathology</i> , 2017, 102, 25-31.	0.9	15
98	Gene Networks Underlying Convergent and Pleiotropic Phenotypes in a Large and Systematically-Phenotyped Cohort with Heterogeneous Developmental Disorders. <i>PLoS Genetics</i> , 2015, 11, e1005012.	1.5	14
99	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. <i>European Journal of Human Genetics</i> , 2016, 24, 1707-1714.	1.4	14
100	Pathogenic variants in <i>TNRC6B</i> cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. <i>Journal of Medical Genetics</i> , 2020, 57, 717-724.	1.5	14
101	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. <i>Genomics Data</i> , 2014, 2, 144-146.	1.3	13
102	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies. <i>Genome Medicine</i> , 2017, 9, 118.	3.6	13
103	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 750-758.	2.6	13
104	Exome sequencing for paediatric-onset diseases: impact of the extensive involvement of medical geneticists in the diagnostic odyssey. <i>Npj Genomic Medicine</i> , 2018, 3, 19.	1.7	11
105	<i>BAZ2B</i> haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. <i>Human Mutation</i> , 2020, 41, 921-925.	1.1	11
106	Structural Genomic Variation in Intellectual Disability. <i>Methods in Molecular Biology</i> , 2012, 838, 77-95.	0.4	10
107	Haploinsufficiency of the HIRA gene located in the 22q11 deletion syndrome region is associated with abnormal neurodevelopment and impaired dendritic outgrowth. <i>Human Genetics</i> , 2021, 140, 885-896.	1.8	10
108	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. <i>European Journal of Human Genetics</i> , 2017, 25, 1126-1133.	1.4	10

#	ARTICLE	IF	CITATIONS
109	Identification of androgen-responsive genes that are alternatively regulated in androgen-dependent and androgen-independent rat prostate tumors. <i>Genes Chromosomes and Cancer</i> , 2005, 43, 273-283.	1.5	9
110	Clinical performance of the CytoScan Dx Assay in diagnosing developmental delay/intellectual disability. <i>Genetics in Medicine</i> , 2016, 18, 168-173.	1.1	9
111	Diagnostic yield of patients with undiagnosed intellectual disability, global developmental delay and multiples congenital anomalies using karyotype, microarray analysis, whole exome sequencing from Central Brazil. <i>PLoS ONE</i> , 2022, 17, e0266493.	1.1	9
112	Identification of a de novo variant in <i>CHUK</i> in a patient with an EEC/AEC syndrome-like phenotype and hypogammaglobulinemia. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1813-1820.	0.7	8
113	A novel MBD5 mutation in an intellectually disabled adult female patient with epilepsy: Suggestive of early onset dementia?. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e849.	0.6	8
114	A de novo <i>CTNNB1</i> Novel Splice Variant in an Adult Female with Severe Intellectual Disability. <i>International Medical Case Reports Journal</i> , 2020, Volume 13, 487-492.	0.3	8
115	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 405-411.	2.6	8
116	A Rare, Recurrent, De Novo 14q32.2q32.31 Microdeletion of 1.1 Mb in a 20-Year-Old Female Patient with a Maternal UPD(14)-Like Phenotype and Intellectual Disability. <i>Case Reports in Genetics</i> , 2014, 2014, 1-5.	0.1	7
117	Molecular analysis of the erythroid phenotype of a patient with BCL11A haploinsufficiency. <i>Blood Advances</i> , 2021, 5, 2339-2349.	2.5	7
118	Phenotypic characterization of an older adult male with late-onset epilepsy and a novel mutation in <i>ASXL3</i> shows overlap with the associated Bainbridge-Ropers syndrome. <i>Neuropsychiatric Disease and Treatment</i> , 2018, Volume 14, 867-870.	1.0	6
119	Paternal uniparental disomy of chromosome 19 in a pair of monozygotic diamniotic twins with dysmorphic features and developmental delay. <i>Journal of Medical Genetics</i> , 2018, 55, 847-852.	1.5	6
120	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2022, 24, 645-653.	1.1	6
121	Constraint and conservation of paired-type homeodomains predicts the clinical outcome of missense variants of uncertain significance. <i>Human Mutation</i> , 2020, 41, 1407-1424.	1.1	2
122	MED13L-related intellectual disability due to paternal germinal mosaicism. <i>Journal of Physical Education and Sports Management</i> , 2021, , mcs.a006124.	0.5	2
123	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104402.	0.7	2
124	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100024.	1.0	1
125	Quadrupedal gait and cerebellar hypoplasia, the Uner Tan syndrome, caused by ITPR1 gene mutation. <i>Parkinsonism and Related Disorders</i> , 2021, 92, 33-35.	1.1	1
126	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. <i>Genetical Research</i> , 2015, 97, e19.	0.3	0

#	ARTICLE	IF	CITATIONS
127	Biallelic frameshift mutation in <i>RIN2</i> in a patient with intellectual disability and cataract, without RIN2 syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3238-3240.	0.7	0