Rolph Pfundt

List of Publications by Year
in descending order

127 papers	7,706 citations	66343 42 h-index	69250 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. Nature, 2014, 511, 344-347.	27.8	996
2	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
3	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
4	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
5	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
6	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
7	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
8	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	2.4	143
9	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
10	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	2.8	127
11	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	6.2	125
12	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
13	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
14	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. PLoS Genetics, 2017, 13, e1006864.	3.5	116
15	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	6.2	110
16	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
17	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599.	2.8	104
18	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95

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19	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. European Journal of Human Genetics, 2017, 25, 308-314.	2.8	90
20	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	6.2	89
21	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. American Journal of Human Genetics, 2017, 101, 478-484.	6.2	84
22	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
23	Mutations in N-acetylglucosamine (O-GlcNAc) transferase in patients with X-linked intellectual disability. Journal of Biological Chemistry, 2017, 292, 12621-12631.	3.4	72
24	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. European Journal of Human Genetics, 2018, 26, 64-74.	2.8	72
25	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	6.2	71
26	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
27	Phenotypic and molecular insights into CASK-related disorders in males. Orphanet Journal of Rare Diseases, 2015, 10, 44.	2.7	68
28	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. Nature Genetics, 2016, 48, 877-887.	21.4	67
29	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. Epilepsia, 2019, 60, 155-164.	5.1	65
30	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
31	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.	6.2	61
32	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
33	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	3.2	59
34	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
35	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	2.4	58
36	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56

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37	Loss-of-Function Mutations in YY1AP1 Lead to Grange Syndrome and a Fibromuscular Dysplasia-Like Vascular Disease. American Journal of Human Genetics, 2017, 100, 21-30.	6.2	54
38	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 862-876.	1.2	52
39	ACAN Gene Mutations in Short Children Born SGA and Response to Growth Hormone Treatment. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1458-1467.	3.6	50
40	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	2.3	49
41	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
42	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	5.1	46
43	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	3.8	46
44	Next generation sequencing in synovial sarcoma reveals novel gene mutations. Oncotarget, 2015, 6, 34680-34690.	1.8	45
45	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	6.2	45
46	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. American Journal of Human Genetics, 2017, 101, 139-148.	6.2	45
47	Mutations in PIK3C2A cause syndromic short stature, skeletal abnormalities, and cataracts associated with ciliary dysfunction. PLoS Genetics, 2019, 15, e1008088.	3.5	45
48	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	2.8	44
49	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
50	CREBBP mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2681-2693.	1.2	43
51	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
52	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
53	Association of AADAC Deletion and Gilles de la Tourette Syndrome in a Large European Cohort. Biological Psychiatry, 2016, 79, 383-391.	1.3	41
54	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37

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55	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045.	2.9	37
56	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. American Journal of Human Genetics, 2017, 101, 824-832.	6.2	36
57	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
58	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
59	Accurate detection of clinically relevant uniparental disomy from exome sequencing data. Genetics in Medicine, 2020, 22, 803-808.	2.4	35
60	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
61	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	2.8	34
62	Biallelic B3GALT6 mutations cause spondylodysplastic Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2018, 27, 3475-3487.	2.9	34
63	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	6.2	34
64	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	2.4	34
65	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
66	Dutch genome diagnostic laboratories accelerated and improved variant interpretation and increased accuracy by sharing data. Human Mutation, 2019, 40, 2230-2238.	2.5	32
67	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	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. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
69	The clustering of functionally related genes contributes to CNV-mediated disease. Genome Research, 2015, 25, 802-813.	5.5	31
70	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
71	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
72	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30

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73	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	21.4	28
74	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
75	Neurodegenerative VPS41 variants inhibit HOPS function and mTORC1â€dependent TFEB/TFE3 regulation. EMBO Molecular Medicine, 2021, 13, e13258.	6.9	26
76	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	2.4	24
77	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. PLoS ONE, 2014, 9, e112687.	2.5	23
78	Broadening the phenotypic spectrum of pathogenic LARP7 variants: two cases with intellectual disability, variable growth retardation and distinct facial features. Journal of Human Genetics, 2016, 61, 229-233.	2.3	23
79	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
80	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	6.2	23
81	Chromosomal aberrations in cerebral visual impairment. European Journal of Paediatric Neurology, 2014, 18, 677-684.	1.6	20
82	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
83	Clinical exome sequencingâ€"Mistakes and caveats. Human Mutation, 2022, 43, 1041-1055.	2.5	20
84	Duplications of SLC1A3: Associated with ADHD and autism. European Journal of Medical Genetics, 2016, 59, 373-376.	1.3	19
85	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	6.2	19
86	Holoprosencephaly and preaxial polydactyly associated with a 1.24ÂMb duplication encompassing FBXW11 at 5q35.1. Journal of Human Genetics, 2006, 51, 721-726.	2.3	18
87	Somatic loss of polycystic disease genes contributes to the formation of isolated and polycystic liver cysts: TableÂ1. Gut, 2015, 64, 688-690.	12.1	18
88	Genome-wide investigation of an ID cohort reveals de novo 3â€²UTR variants affecting gene expression. Human Genetics, 2018, 137, 717-721.	3.8	18
89	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1692-1709.	6.2	18
90	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	2.8	17

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91	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793.	6.2	17
92	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
93	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
94	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
95	Germline and somatic mosaicism in a family with multiple endocrine neoplasia type 1 (MEN1) syndrome. European Journal of Endocrinology, 2019, 180, K15-K19.	3.7	16
96	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
97	Copy number variation analysis and methylome profiling of a GNAQ-mutant primary meningeal melanocytic tumor and its liver metastasis. Experimental and Molecular Pathology, 2017, 102, 25-31.	2.1	15
98	Gene Networks Underlying Convergent and Pleiotropic Phenotypes in a Large and Systematically-Phenotyped Cohort with Heterogeneous Developmental Disorders. PLoS Genetics, 2015, 11, e1005012.	3.5	14
99	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. European Journal of Human Genetics, 2016, 24, 1707-1714.	2.8	14
100	Pathogenic variants in TNRC6B cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. Journal of Medical Genetics, 2020, 57, 717-724.	3.2	14
101	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 2014, 2, 144-146.	1.3	13
102	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotypeâ€“phenotype associations in the muscular dystrophy-dystroglycanopathies. Genome Medicine, 2017, 9, 118.	8.2	13
103	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
104	Exome sequencing for paediatric-onset diseases: impact of the extensive involvement of medical geneticists in the diagnostic odyssey. Npj Genomic Medicine, 2018, 3, 19.	3.8	11
105	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
106	Structural Genomic Variation in Intellectual Disability. Methods in Molecular Biology, 2012, 838, 77-95.	0.9	10
107	Haploinsufficiency of the HIRA gene located in the 22q11 deletion syndrome region is associated with abnormal neurodevelopment and impaired dendritic outgrowth. Human Genetics, 2021, 140, 885-896.	3.8	10
108	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. European Journal of Human Genetics, 2017, 25, 1126-1133.	2.8	10

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109	Identification of androgen-responsive genes that are alternatively regulated in androgen-dependent and androgen-independent rat prostate tumors. Genes Chromosomes and Cancer, 2005, 43, 273-283.	2.8	9
110	Clinical performance of the CytoScan Dx Assay in diagnosing developmental delay/intellectual disability. Genetics in Medicine, 2016, 18, 168-173.	2.4	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. PLoS ONE, 2022, 17, e0266493.	2.5	9
112	Identification of a de novo variant in CHUK in a patient with an EEC/AEC syndromeâ€like phenotype and hypogammaglobulinemia. American Journal of Medical Genetics, Part A, 2017, 173, 1813-1820.	1.2	8
113	A novel MBD5 mutation in an intellectually disabled adult female patient with epilepsy: Suggestive of early onset dementia?. Molecular Genetics & Genomic Medicine, 2019, 7, e849.	1.2	8
114	A de novo CTNNB1 Novel Splice Variant in an Adult Female with Severe Intellectual Disability . International Medical Case Reports Journal, 2020, Volume 13, 487-492.	0.8	8
115	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	6.2	8
116	A Rare, Recurrent,De Novo14q32.2q32.31 Microdeletion of 1.1â€‰Mb in a 20-Year-Old Female Patient with a Maternal UPD(14)-Like Phenotype and Intellectual Disability. Case Reports in Genetics, 2014, 2014, 1-5.	0.2	7
117	Molecular analysis of the erythroid phenotype of a patient with BCL11A haploinsufficiency. Blood Advances, 2021, 5, 2339-2349.	5.2	7
118	Phenotypic characterization of an older adult male with late-onset epilepsy and a novel mutation in ASXL3 shows overlap with the associated Bainbridge-Ropers syndrome. Neuropsychiatric Disease and Treatment, 2018, Volume 14, 867-870.	2.2	6
119	Paternal uniparental disomy of chromosome 19 in a pair of monochorionic diamniotic twins with dysmorphic features and developmental delay. Journal of Medical Genetics, 2018, 55, 847-852.	3.2	6
120	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	2.4	6
121	Constraint and conservation of pairedâ€type homeodomains predicts the clinical outcome of missense variants of uncertain significance. Human Mutation, 2020, 41, 1407-1424.	2.5	2
122	MED13L-related intellectual disability due to paternal germinal mosaicism. Journal of Physical Education and Sports Management, 2021, , mcs.a006124.	1.2	2
123	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. European Journal of Medical Genetics, 2022, 65, 104402.	1.3	2
124	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances, 2021, 2, 100024.	1.7	1
125	Quadrupedal gait and cerebellar hypoplasia, the Uner Tan syndrome, caused by ITPR1 gene mutation. Parkinsonism and Related Disorders, 2021, 92, 33-35.	2.2	1
126	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. Genetical Research, 2015, 97, e19.	0.9	0

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127	Biallelicframeshift mutation in RIN2 in a patient with intellectual disability and cataract, without RIN2 syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3238-3240.	1.2	0