

# Christiane Zweier

## List of Publications by Year in descending order

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129  
papers

9,496  
citations

53939

47  
h-index

48101

92  
g-index

138  
all docs

138  
docs citations

138  
times ranked

16721  
citing authors

#	ARTICLE	IF	CITATIONS
1	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. <i>Lancet, The</i> , 2012, 380, 1674-1682.	6.3	940
2	CNTNAP2 and NRXN1 Are Mutated in Autosomal-Recessive Pitt-Hopkins-like Mental Retardation and Determine the Level of a Common Synaptic Protein in Drosophila. <i>American Journal of Human Genetics</i> , 2009, 85, 655-666.	2.6	573
3	Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. <i>Cell</i> , 2008, 135, 37-48.	13.5	567
4	Mutations in the Pericentrin ( <i>PCNT</i> ) Gene Cause Primordial Dwarfism. <i>Science</i> , 2008, 319, 816-819.	6.0	370
5	Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2063-2074.	0.7	343
6	Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. <i>Nature Genetics</i> , 2011, 43, 20-22.	9.4	308
7	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. <i>American Journal of Human Genetics</i> , 2016, 98, 149-164.	2.6	270
8	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). <i>American Journal of Human Genetics</i> , 2007, 80, 994-1001.	2.6	261
9	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. <i>Cell</i> , 2014, 157, 651-663.	13.5	228
10	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 90, 565-572.	2.6	225
11	Transcriptional repressor ZEB2 promotes terminal differentiation of CD8+ effector and memory T cell populations during infection. <i>Journal of Experimental Medicine</i> , 2015, 212, 2027-2039.	4.2	206
12	Human TBX1 Missense Mutations Cause Gain of Function Resulting in the Same Phenotype as 22q11.2 Deletions. <i>American Journal of Human Genetics</i> , 2007, 80, 510-517.	2.6	195
13	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. <i>Human Molecular Genetics</i> , 2013, 22, 5121-5135.	1.4	190
14	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. <i>JAMA Psychiatry</i> , 2017, 74, 293.	6.0	186
15	Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. <i>Human Mutation</i> , 2010, 31, 722-733.	1.1	163
16	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2013, 93, 124-131.	2.6	151
17	Genetic and neurodevelopmental spectrum of <i>SYNGAP1</i> -associated intellectual disability and epilepsy. <i>Journal of Medical Genetics</i> , 2016, 53, 511-522.	1.5	135
18	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. <i>Genetics in Medicine</i> , 2018, 20, 1175-1185.	1.1	133

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19	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. <i>Journal of Medical Genetics</i> , 2010, 47, 321-331.	1.5	126
20	?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. <i>American Journal of Medical Genetics Part A</i> , 2002, 108, 177-181.	2.4	122
21	Clinical and Mutational Spectrum of Mowat-Wilson Syndrome. <i>European Journal of Medical Genetics</i> , 2005, 48, 97-111.	0.7	121
22	Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. <i>Journal of Medical Genetics</i> , 2005, 42, 871-876.	1.5	118
23	Severely Incapacitating Mutations in Patients with Extreme Short Stature Identify RNA-Processing Endoribonuclease RMRP as an Essential Cell Growth Regulator. <i>American Journal of Human Genetics</i> , 2005, 77, 795-806.	2.6	117
24	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. <i>Journal of Medical Genetics</i> , 2007, 44, 651-656.	1.5	114
25	Zeb2 recruits HDAC-NuRD to inhibit Notch and controls Schwann cell differentiation and remyelination. <i>Nature Neuroscience</i> , 2016, 19, 1060-1072.	7.1	113
26	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. <i>BMC Medical Genetics</i> , 2011, 12, 106.	2.1	109
27	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. <i>Genetics in Medicine</i> , 2018, 20, 630-638.	1.1	101
28	Further delineation of Pitt-Hopkins syndrome: phenotypic and genotypic description of 16 novel patients. <i>Journal of Medical Genetics</i> , 2008, 45, 738-744.	1.5	86
29	<i><sc>MLL2</sc></i> mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. <i>Clinical Genetics</i> , 2013, 84, 539-545.	1.0	85
30	Exome Pool-Seq in neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2017, 25, 1364-1376.	1.4	77
31	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 753-760.	1.4	73
32	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. <i>Journal of Medical Genetics</i> , 2007, 44, 629-636.	1.5	72
33	The phenotypic spectrum in patients with arginine to cysteine mutations in the COL2A1 gene. <i>Journal of Medical Genetics</i> , 2005, 43, 406-413.	1.5	71
34	Dosage-Dependent Severity of the Phenotype in Patients with Mental Retardation Due to a Recurrent Copy-Number Gain at Xq28 Mediated by an Unusual Recombination. <i>American Journal of Human Genetics</i> , 2009, 85, 809-822.	2.6	70
35	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. <i>Human Mutation</i> , 2016, 37, 755-764.	1.1	70
36	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

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37	Chromatin-Remodeling-Factor ARID1B Represses Wnt/ $\beta$ 2-Catenin Signaling. American Journal of Human Genetics, 2015, 97, 445-456.	2.6	67
38	<i>FOXP2</i> variants 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
39	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	1.1	67
40	Phenotype and genotype in Nicolaides-Baraitser syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 302-314.	0.7	66
41	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	1.0	63
42	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. Human Mutation, 2020, 41, 837-849.	1.1	63
43	Characterisation of deletions of the ZFX1B region and genotype-phenotype analysis in Mowat-Wilson syndrome. Journal of Medical Genetics, 2003, 40, 601-605.	1.5	61
44	Systematic survey of variants in TBX1 in non-syndromic tetralogy of Fallot identifies a novel 57 base pair deletion that reduces transcriptional activity but finds no evidence for association with common variants. Heart, 2010, 96, 1651-1655.	1.2	61
45	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
46	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
47	Hirschsprung disease, mental retardation, characteristic facial features, and mutation in the gene ZFX1B (SIP1): Confirmation of the Mowat-Wilson syndrome. American Journal of Medical Genetics Part A, 2003, 116A, 385-388.	2.4	58
48	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	1.6	53
49	A new face of Borjeson-Forssman-Lehmann syndrome? De novo mutations in <i>PHF6</i> in seven females with a distinct phenotype. Journal of Medical Genetics, 2013, 50, 838-847.	1.5	50
50	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
51	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
52	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
53	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
54	Eight further individuals with intellectual disability and epilepsy carrying bi-allelic <i>CNTNAP2</i> aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827.	1.5	45

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55	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. <i>Molecular Autism</i> , 2018, 9, 20.	2.6	45
56	Clinical delineation of the <i>PACS1</i>-related syndrome—Report on 19 patients. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 670-675.	0.7	44
57	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. <i>Journal of Medical Genetics</i> , 2019, 56, 701-710.	1.5	43
58	<i>DDX3X</i> mutations in two girls with a phenotype overlapping Toriello-Carey syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1369-1373.	0.7	41
59	Human Intellectual Disability Genes Form Conserved Functional Modules in <i>Drosophila</i> . <i>PLoS Genetics</i> , 2013, 9, e1003911.	1.5	39
60	Habituation Learning Is a Widely Affected Mechanism in <i>Drosophila</i> Models of Intellectual Disability and Autism Spectrum Disorders. <i>Biological Psychiatry</i> , 2019, 86, 294-305.	0.7	39
61	Intellectual disability and autism spectrum disorders —on the fly™: insights from <i>Drosophila</i>. <i>DMM Disease Models and Mechanisms</i> , 2019, 12, .	1.2	38
62	Mowat-Wilson syndrome and mutation in the zinc finger homeo box 1B gene: a well defined clinical entity. <i>Journal of Medical Genetics</i> , 2004, 41, 16e-16.	1.5	36
63	<i>STAG1</i> mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2017, 54, 479-488.	1.5	35
64	Encephalopathies with <i>KCNC1</i> variants: genotype-phenotype-functional correlations. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1263-1272.	1.7	33
65	Atypical ZFX1B mutation associated with a mild Mowat-Wilson syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 869-872.	0.7	32
66	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. <i>Genetics in Medicine</i> , 2021, 23, 543-554.	1.1	32
67	"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. <i>American Journal of Medical Genetics Part A</i> , 2002, 108, 177-81.	2.4	32
68	A missense mutation in the ZFX1B gene associated with an atypical Mowat-Wilson syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1223-1227.	0.7	31
69	Novel missense, insertion and deletion mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with congenital insensitivity to pain with anhidrosis. <i>Neuromuscular Disorders</i> , 2008, 18, 159-166.	0.3	31
70	New Mutations of <i>EXT1</i> and <i>EXT2</i> Genes in German Patients with Multiple Osteochondromas. <i>Annals of Human Genetics</i> , 2009, 73, 283-291.	0.3	31
71	Somatic mosaicism in a mother of two children with Pitt-Hopkins syndrome. <i>Clinical Genetics</i> , 2013, 83, 73-77.	1.0	31
72	Altered <i>GPM6A/M6</i> Dosage Impairs Cognition and Causes Phenotypes Responsive to Cholesterol in Human and <i>Drosophila</i>. <i>Human Mutation</i> , 2014, 35, 1495-1505.	1.1	31

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73	Facial phenotype allows diagnosis of Mowat-Wilson syndrome in the absence of hirschsprung disease. American Journal of Medical Genetics Part A, 2004, 124A, 102-104.	2.4	30
74	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Human Genetics, 2014, 133, 939-949.	1.8	29
75	Further delineation of the female phenotype with <sc><i>KDM5C</i></sc> disease causing variants: 19 new individuals and review of the literature. Clinical Genetics, 2020, 98, 43-55.	1.0	28
76	Females with de novo aberrations in <i>PHF6</i>: 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
77	Clinical spectrum of females with HCCS mutation: from no clinical signs to a neonatal lethal form of the microphthalmia with linear skin defects (MLS) syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 53.	1.2	26
78	Haploinsufficiency of <i>NR4A2</i> is associated with a neurodevelopmental phenotype with prominent language impairment. American Journal of Medical Genetics, Part A, 2017, 173, 2231-2234.	0.7	25
79	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
80	NRF1 association with AUTS2-Polycomb mediates specific gene activation in the brain. Molecular Cell, 2021, 81, 4663-4676.e8.	4.5	23
81	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
82	Clinical, cytogenetic and molecular characterization of a patient with combined succinic semialdehyde dehydrogenase deficiency and incomplete WAGR syndrome with obesity. Molecular Genetics and Metabolism, 2006, 88, 256-260.	0.5	20
83	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
84	TCF4 (E2-2) harbors tumor suppressive functions in SHH medulloblastoma. Acta Neuropathologica, 2019, 137, 657-673.	3.9	20
85	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. European Journal of Medical Genetics, 2018, 61, 363-368.	0.7	17
86	Pulmonary artery sling and congenital tracheal stenosis in another patient with Mowatâ€“Wilson syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1528-1530.	0.7	16
87	Prenatal diagnosis of <i>HNF1B</i>â€“associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?. Prenatal Diagnosis, 2019, 39, 1136-1147.	1.1	16
88	Early-onset parkinsonism in PPP2R5D-related neurodevelopmental disorder. European Journal of Medical Genetics, 2021, 64, 104123.	0.7	16
89	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
90	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	3.6	16

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91	Genitourinary Anomalies in Mowat-Wilson Syndrome with Deletion/Mutation in the Zinc Finger Homeo Box 1B Gene (ZFX1B). <i>Hormone Research in Paediatrics</i> , 2005, 63, 187-192.	0.8	15
92	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. <i>Genetics in Medicine</i> , 2020, 22, 797-802.	1.1	15
93	Severe clinical course of Hirschsprung disease in a Mowat-Wilson syndrome patient. <i>Journal of Applied Genetics</i> , 2010, 51, 111-113.	1.0	14
94	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. <i>Genetics in Medicine</i> , 2022, 24, 1261-1273.	1.1	14
95	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. <i>American Journal of Human Genetics</i> , 2020, 107, 544-554.	2.6	13
96	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. <i>Journal of Medical Genetics</i> , 2022, 59, 965-975.	1.5	13
97	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. <i>Gene</i> , 2014, 538, 30-35.	1.0	11
98	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. <i>European Journal of Medical Genetics</i> , 2016, 59, 549-553.	0.7	11
99	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. <i>European Journal of Human Genetics</i> , 2019, 27, 1061-1071.	1.4	11
100	Skeletal abnormalities are common features in Aymara Gripp syndrome. <i>Clinical Genetics</i> , 2020, 97, 362-369.	1.0	10
101	Microdeletions of chromosome 7p21, including TWIST1, associated with significant microcephaly, facial dysmorphism, and short stature. <i>European Journal of Medical Genetics</i> , 2011, 54, 256-261.	0.7	9
102	Biallelic SEMA3A defects cause a novel type of syndromic short stature. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2880-2889.	0.7	9
103	A 15Mb duplication of 6q24.1-q25.3 associated with typical but milder features of the duplication 6q syndrome. <i>European Journal of Medical Genetics</i> , 2008, 51, 358-361.	0.7	8
104	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. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1765-1770.	0.7	8
105	Genetic interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in <i>Drosophila melanogaster</i> . <i>Scientific Reports</i> , 2020, 10, 1204.	1.6	8
106	Central nervous system anomalies in two females with Borjeson-Forsman-Lehmann syndrome. <i>Epilepsy and Behavior</i> , 2017, 69, 104-109.	0.9	7
107	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). <i>European Journal of Medical Genetics</i> , 2020, 63, 104004.	0.7	7
108	A case of severe autosomal recessive spinocerebellar ataxia type 18 with a novel nonsense variant in GRID2. <i>European Journal of Medical Genetics</i> , 2020, 63, 103998.	0.7	7

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109	<i>De novo</i> missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454.	1.4	7
110	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	1.1	6
111	9 Mb deletion including chromosome band 3q24 associated with unsuspecting facial gestalt, persistent ductus omphaloentericus, mild mental retardation and tic. European Journal of Medical Genetics, 2005, 48, 360-362.	0.7	5
112	7q31.2q31.31 deletion downstream of <sc><i>FOXP2</i></sc> segregating in a family with speech and language disorder. American Journal of Medical Genetics, Part A, 2020, 182, 2737-2741.	0.7	5
113	<sc><i>QRICH1</i></sc> variants in <sc>Ververiâ€Brady</sc> syndromeâ€”delineation of the genotypic and phenotypic spectrum. Clinical Genetics, 2021, 99, 199-207.	1.0	5
114	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	1.2	5
115	PRICKLE2 revisitedâ€”further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	1.4	5
116	<sc><i>ZMYND11</i></sc> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	1.0	5
117	Further characterization of <sc>Borjesonâ€Forssmanâ€Lehmann</sc> syndrome in females due to de novo variants in <sc><i>PHF6</i></sc>. Clinical Genetics, 2022, 102, 182-190.	1.0	5
118	Microphthalmia is not a mandatory finding in Xâ€linked recessive syndromic microphthalmia caused by the recurrent <i>BCOR</i> variant p.Pro85Leu. American Journal of Medical Genetics, Part A, 2018, 176, 2872-2876.	0.7	3
119	Loss of PHF6 leads to aberrant development of human neuron-like cells. Scientific Reports, 2020, 10, 19030.	1.6	3
120	Manifestation of epilepsy in a patient with <sc><i>EED</i></sc>â€related overgrowth (<sc>Cohenâ€Gibson</sc> syndrome). American Journal of Medical Genetics, Part A, 2022, 188, 292-297.	0.7	3
121	A novel splice variant expands the LAMC3 â€associated cortical phenotype to frontal only polymicrogyria and adultâ€onset epilepsy. American Journal of Medical Genetics, Part A, 2020, 182, 2761-2764.	0.7	2
122	Phoniatric, Audiological, Orodonal and Speech Problems in a Boy with Cardio-Facio-Cutaneous Syndrome Type 3 (CFC 3) Due to a Pathogenic Variant in MAP2K1 â€ Case Study. The Application of Clinical Genetics, 2021, Volume 14, 389-398.	1.4	2
123	Mikrozephalie bei psychomotorischen EntwicklungsstÃrungen und geistiger Behinderung. Medizinische Genetik, 2015, 27, 362-368.	0.1	1
124	Diagnostik seltener Erkrankungen mit â€next generation sequencingâ€ â€ angekommen oder abgewehrt?. Medizinische Genetik, 2019, 31, 335-343.	0.1	1
125	Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. BMC Cancer, 2019, 19, 435.	1.1	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.	0.3	0

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127	X-chromosomale Entwicklungsstörungen im weiblichen Geschlecht. Medizinische Genetik, 2018, 30, 334-341.	0.1	0
128	Transcriptional repressor ZEB2 promotes terminal differentiation of CD8 <sup>+</sup> effector and memory T cell populations during infection. Journal of Cell Biology, 2015, 211, 2113-2125.	2.3	0
129	Identification and characterization of a GLMN splice site variant in a family with glomuvenous malformations. European Journal of Dermatology, 2020, 30, 179-181.	0.3	0