

# Christiane Zweier

## List of Publications by Year in descending order

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Version: 2024-02-01

129  
papers

9,496  
citations

44069

48  
h-index

42399

92  
g-index

138  
all docs

138  
docs citations

138  
times ranked

15441  
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>De novo</i> missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454.	2.9	7
2	Manifestation of epilepsy in a patient with <scp><i>EED</i></scp>-related overgrowth (<scp>Cohenâ€“Gibson</scp> syndrome). American Journal of Medical Genetics, Part A, 2022, 188, 292-297.	1.2	3
3	<i>De novo</i> coding variants in the <i>AGO1</i> gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
4	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	2.4	14
5	Further characterization of <scp>Borjesonâ€“Forssmanâ€“Lehmann</scp> syndrome in females due to de novo variants in <scp><i>PHF6</i></scp>. Clinical Genetics, 2022, 102, 182-190.	2.0	5
6	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	2.4	32
7	<scp><i>QRICH1</i></scp> variants in <scp>Ververiâ€“Brady</scp> syndromeâ€“delineation of the genotypic and phenotypic spectrum. Clinical Genetics, 2021, 99, 199-207.	2.0	5
8	Early-onset parkinsonism in PPP2R5D-related neurodevelopmental disorder. European Journal of Medical Genetics, 2021, 64, 104123.	1.3	16
9	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
10	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
11	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
12	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	2.4	6
13	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	8.2	16
14	PRICKLE2 revisitedâ€“further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	2.8	5
15	<scp><i>ZMYND11</i></scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	2.0	5
16	Phoniatic, Audiological, Orodental 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.	3.0	2
17	NRF1 association with AUTS2-Polycomb mediates specific gene activation in the brain. Molecular Cell, 2021, 81, 4663-4676.e8.	9.7	23
18	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24

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19	Skeletal abnormalities are common features in Aymã©Gripp syndrome. Clinical Genetics, 2020, 97, 362-369.	2.0	10
20	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	2.4	15
21	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
22	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
23	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7
24	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.	1.2	2
25	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.	1.2	5
26	Loss of PHF6 leads to aberrant development of human neuron-like cells. Scientific Reports, 2020, 10, 19030.	3.3	3
27	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
28	A case of severe autosomal recessive spinocerebellar ataxia type 18 with a novel nonsense variant in GRID2. European Journal of Medical Genetics, 2020, 63, 103998.	1.3	7
29	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.	2.0	28
30	Genetic interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in Drosophila melanogaster. Scientific Reports, 2020, 10, 1204.	3.3	8
31	Identification and characterization of a GLMN splice site variant in a family with glomuvenous malformations. European Journal of Dermatology, 2020, 30, 179-181.	0.6	0
32	Encephalopathies with <i>KCNC1</i> variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272.	3.7	33
33	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
34	Diagnostik seltener Erkrankungen mit â€next generation sequencingâ€ â€ angekommen oder abgewehrt?. Medizinische Genetik, 2019, 31, 335-343.	0.2	1
35	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.	2.3	16
36	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	2.0	63

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37	Intellectual disability and autism spectrum disorders – on the fly™: insights from <i>Drosophila</i> . DMM Disease Models and Mechanisms, 2019, 12, .	2.4	38
38	Habituation Learning Is a Widely Affected Mechanism in <i>Drosophila</i> Models of Intellectual Disability and Autism Spectrum Disorders. Biological Psychiatry, 2019, 86, 294-305.	1.3	39
39	Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. BMC Cancer, 2019, 19, 435.	2.6	1
40	TCF4 (E2-2) harbors tumor suppressive functions in SHH medulloblastoma. Acta Neuropathologica, 2019, 137, 657-673.	7.7	20
41	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	2.8	11
42	Estimating the effect size of the 15Q11.2 BP1–BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	3.2	43
43	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
44	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. European Journal of Medical Genetics, 2018, 61, 363-368.	1.3	17
45	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in <i>Drosophila</i> . American Journal of Human Genetics, 2018, 102, 44-57.	6.2	49
46	Phenotype and genotype of 87 patients with Mowat–Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
47	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
48	Microphthalmia is not a mandatory finding in X-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.	1.2	3
49	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
50	X-chromosomale Entwicklungsstörungen im weiblichen Geschlecht. Medizinische Genetik, 2018, 30, 334-341.	0.2	0
51	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.	6.2	48
52	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. Molecular Autism, 2018, 9, 20.	4.9	45
53	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	11.0	186
54	<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.	3.2	67

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55	<i>STAG1</i> mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2017, 54, 479-488.	3.2	35
56	Haploinsufficiency of <i>NR4A2</i> is associated with a neurodevelopmental phenotype with prominent language impairment. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2231-2234.	1.2	25
57	<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.	1.2	41
58	Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. <i>Epilepsy and Behavior</i> , 2017, 69, 104-109.	1.7	7
59	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.	6.2	61
60	Genetic screening confirms heterozygous mutations in <i>ACAN</i> as a major cause of idiopathic short stature. <i>Scientific Reports</i> , 2017, 7, 12225.	3.3	53
61	Exome Pool-Seq in neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2017, 25, 1364-1376.	2.8	77
62	Eight further individuals with intellectual disability and epilepsy carrying bi-allelic <i>CNTNAP2</i> aberrations allow delineation of the mutational and phenotypic spectrum. <i>Journal of Medical Genetics</i> , 2016, 53, 820-827.	3.2	45
63	Genetic and neurodevelopmental spectrum of <i>SYNGAP1</i> -associated intellectual disability and epilepsy. <i>Journal of Medical Genetics</i> , 2016, 53, 511-522.	3.2	135
64	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in <i>GABRA1</i> . <i>European Journal of Medical Genetics</i> , 2016, 59, 549-553.	1.3	11
65	Expanding the Phenotype Associated with <i>NAA10</i> -Related N-Terminal Acetylation Deficiency. <i>Human Mutation</i> , 2016, 37, 755-764.	2.5	70
66	Zeb2 recruits HDAC-NuRD to inhibit Notch and controls Schwann cell differentiation and remyelination. <i>Nature Neuroscience</i> , 2016, 19, 1060-1072.	14.8	113
67	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.	1.2	44
68	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. <i>American Journal of Human Genetics</i> , 2016, 98, 149-164.	6.2	270
69	A de novo microdeletion in <i>NRXN1</i> in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. <i>Genetical Research</i> , 2015, 97, e19.	0.9	0
70	Mikrozephalie bei psychomotorischen Entwicklungsstörungen und geistiger Behinderung. <i>Medizinische Genetik</i> , 2015, 27, 362-368.	0.2	1
71	Loss-of-function variants of <i>SETD5</i> cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 753-760.	2.8	73
72	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.	8.5	206

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73	Chromatin-Remodeling-Factor ARID1B Represses Wnt/ $\beta$ -Catenin Signaling. American Journal of Human Genetics, 2015, 97, 445-456.	6.2	67
74	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.	5.2	0
75	Altered GPM6A/M6 Dosage Impairs Cognition and Causes Phenotypes Responsive to Cholesterol in Human and Drosophila. Human Mutation, 2014, 35, 1495-1505.	2.5	31
76	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
77	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Human Genetics, 2014, 133, 939-949.	3.8	29
78	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. Gene, 2014, 538, 30-35.	2.2	11
79	Phenotype and genotype in Nicolaides-Baraitser syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 302-314.	1.6	66
80	Females with de novo aberrations in PHF6: Clinical overlap of Borjeson-Forssman-Lehmann with Coffin-Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 290-301.	1.6	27
81	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.	6.2	59
82	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.	2.7	26
83	MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85
84	Somatic mosaicism in a mother of two children with Pitt-Hopkins syndrome. Clinical Genetics, 2013, 83, 73-77.	2.0	31
85	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	6.2	151
86	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	2.9	190
87	A new face of Borjeson-Forssman-Lehmann syndrome? De novo mutations in PHF6 in seven females with a distinct phenotype. Journal of Medical Genetics, 2013, 50, 838-847.	3.2	50
88	Human Intellectual Disability Genes Form Conserved Functional Modules in Drosophila. PLoS Genetics, 2013, 9, e1003911.	3.5	39
89	Biallelic SEMA3A defects cause a novel type of syndromic short stature. American Journal of Medical Genetics, Part A, 2013, 161, 2880-2889.	1.2	9
90	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940

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91	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.	1.2	20
92	De novo triplication of the <i>MAPT</i> gene from the recurrent 17q21.31 microdeletion region in a patient with moderate intellectual disability and various minor anomalies. American Journal of Medical Genetics, Part A, 2012, 158A, 1765-1770.	1.2	8
93	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. American Journal of Human Genetics, 2012, 90, 565-572.	6.2	225
94	Microdeletions of chromosome 7p21, including TWIST1, associated with significant microcephaly, facial dysmorphism, and short stature. European Journal of Medical Genetics, 2011, 54, 256-261.	1.3	9
95	Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. Nature Genetics, 2011, 43, 20-22.	21.4	308
96	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106.	2.1	109
97	Severe clinical course of Hirschsprung disease in a Mowat-Wilson syndrome patient. Journal of Applied Genetics, 2010, 51, 111-113.	1.9	14
98	Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. Human Mutation, 2010, 31, 722-733.	2.5	163
99	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.	2.9	61
100	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331.	3.2	126
101	New Mutations of <i>EXT1</i> and <i>EXT2</i> Genes in German Patients with Multiple Osteochondromas. Annals of Human Genetics, 2009, 73, 283-291.	0.8	31
102	CNTNAP2 and NRXN1 Are Mutated in Autosomal-Recessive Pitt-Hopkins-like Mental Retardation and Determine the Level of a Common Synaptic Protein in Drosophila. American Journal of Human Genetics, 2009, 85, 655-666.	6.2	573
103	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. American Journal of Human Genetics, 2009, 85, 809-822.	6.2	70
104	Further delineation of Pitt-Hopkins syndrome: phenotypic and genotypic description of 16 novel patients. Journal of Medical Genetics, 2008, 45, 738-744.	3.2	86
105	Novel missense, insertion and deletion mutations in the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1) associated with congenital insensitivity to pain with anhidrosis. Neuromuscular Disorders, 2008, 18, 159-166.	0.6	31
106	A 15Mb duplication of 6q24.1â€“q25.3 associated with typical but milder features of the duplication 6q syndrome. European Journal of Medical Genetics, 2008, 51, 358-361.	1.3	8
107	Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. Cell, 2008, 135, 37-48.	28.9	567
108	Mutations in the Pericentrin ( <i>PCNT</i> ) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	12.6	370



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109	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. <i>Journal of Medical Genetics</i> , 2007, 44, 629-636.	3.2	72
110	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.	3.2	114
111	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.	6.2	195
112	Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). <i>American Journal of Human Genetics</i> , 2007, 80, 994-1001.	6.2	261
113	Pulmonary artery sling and congenital tracheal stenosis in another patient with Mowat-Wilson syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1528-1530.	1.2	16
114	Clinical, cytogenetic and molecular characterization of a patient with combined succinic semialdehyde dehydrogenase deficiency and incomplete WAGR syndrome with obesity. <i>Molecular Genetics and Metabolism</i> , 2006, 88, 256-260.	1.1	20
115	Atypical ZFX1B mutation associated with a mild Mowat-Wilson syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 869-872.	1.2	32
116	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.	1.2	31
117	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.	1.2	343
118	The phenotypic spectrum in patients with arginine to cysteine mutations in the COL2A1 gene. <i>Journal of Medical Genetics</i> , 2005, 43, 406-413.	3.2	71
119	Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. <i>Journal of Medical Genetics</i> , 2005, 42, 871-876.	3.2	118
120	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.	1.8	15
121	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.	6.2	117
122	Clinical and Mutational Spectrum of Mowat-Wilson Syndrome. <i>European Journal of Medical Genetics</i> , 2005, 48, 97-111.	1.3	121
123	9 Mb deletion including chromosome band 3q24 associated with unsuspecting facial gestalt, persistent ductus omphaloentericus, mild mental retardation and tic. <i>European Journal of Medical Genetics</i> , 2005, 48, 360-362.	1.3	5
124	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.	3.2	36
125	Facial phenotype allows diagnosis of Mowat-Wilson syndrome in the absence of hirschsprung disease. <i>American Journal of Medical Genetics Part A</i> , 2004, 124A, 102-104.	2.4	30
126	Hirschsprung disease, mental retardation, characteristic facial features, and mutation in the gene <i>ZFX1B</i> ( <i>SIP1</i> ): Confirmation of the Mowat-Wilson syndrome. <i>American Journal of Medical Genetics Part A</i> , 2003, 116A, 385-388.	2.4	58



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127	Characterisation of deletions of the ZFX1B region and genotype-phenotype analysis in Mowat-Wilson syndrome. Journal of Medical Genetics, 2003, 40, 601-605.	3.2	61
128	“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. American Journal of Medical Genetics Part A, 2002, 108, 177-181.	2.4	122
129	"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. American Journal of Medical Genetics Part A, 2002, 108, 177-81.	2.4	32