# Christiane Zweier

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1438122/publications.pdf

Version: 2024-02-01

| 129 | 9,496 | 53939 47 | 48101 92 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 138 | 138 | 138 | 16721 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | De novo missense variants in FBXO11 alter its protein expression and subcellular localization. Human Molecular Genetics, 2022, 31, 440-454. | 1.4 | 7 |
| 2 | Manifestation of epilepsy in a patient with <scp>EED</scp>â€related overgrowth (<scp>Cohenâ€“Gibson</scp> syndrome). American Journal of Medical Genetics, Part A, 2022, 188, 292-297. | 0.7 | 3 |
| 3 | De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975. | 1.5 | 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. | 1.1 | 14 |
| 5 | Further characterization of <scp>Borjesonâ€Forssmanâ€Lehmann</scp> syndrome in females due to de novo variants in <scp>PHF6</scp>. Clinical Genetics, 2022, 102, 182-190. | 1.0 | 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. | 1.1 | 32 |
| 7 | <scp>QRICH1</scp> variants in <scp>Ververiâ€Brady</scp> syndromeâ€”delineation of the genotypic and phenotypic spectrum. Clinical Genetics, 2021, 99, 199-207. | 1.0 | 5 |
| 8 | Early-onset parkinsonism in PPP2R5D-related neurodevelopmental disorder. European Journal of Medical Genetics, 2021, 64, 104123. | 0.7 | 16 |
| 9 | DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899. | 1.1 | 16 |
| 10 | EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136. | 1.2 | 5 |
| 11 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63. | 3.6 | 50 |
| 12 | Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245. | 1.1 | 6 |
| 13 | Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90. | 3.6 | 16 |
| 14 | PRICKLE2 revisitedâ€”further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244. | 1.4 | 5 |
| 15 | <scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429. | 1.0 | 5 |
| 16 | Phoniatric, 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. | 1.4 | 2 |
| 17 | NRF1 association with AUTS2-Polycomb mediates specific gene activation in the brain. Molecular Cell, 2021, 81, 4663-4676.e8. | 4.5 | 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. | 1.1 | 24 |

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|---|---|---|---|
| 19 | Skeletal abnormalities are common features in AymÃ©â€Gripp syndrome. Clinical Genetics, 2020, 97, 362-369. | 1.0 | 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. | 1.1 | 15 |
| 21 | Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849. | 1.1 | 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. | 2.6 | 13 |
| 23 | Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004. | 0.7 | 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. | 0.7 | 2 |
| 25 | 7q31.2q31.31 deletion downstream of <scp>FOXP2</scp> segregating in a family with speech and language disorder. American Journal of Medical Genetics, Part A, 2020, 182, 2737-2741. | 0.7 | 5 |
| 26 | Loss of PHF6 leads to aberrant development of human neuron-like cells. Scientific Reports, 2020, 10, 19030. | 1.6 | 3 |
| 27 | Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226. | 1.1 | 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. | 0.7 | 7 |
| 29 | Further delineation of the female phenotype with <scp>KDM5C</scp> disease causing variants: 19 new individuals and review of the literature. Clinical Genetics, 2020, 98, 43-55. | 1.0 | 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. | 1.6 | 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. | 0.3 | 0 |
| 32 | Encephalopathies with KCNC1 variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272. | 1.7 | 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. | 1.1 | 48 |
| 34 | Diagnostik seltener Erkrankungen mit â€žnext generation sequencingâ€œ â€“ angekommen oder abgewehrt?. Medizinische Genetik, 2019, 31, 335-343. | 0.1 | 1 |
| 35 | Prenatal diagnosis of HNF1B 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 |
| 36 | Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478. | 1.0 | 63 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Intellectual disability and autism spectrum disorders â€˜on the flyâ€™: insights from Drosophila. DMM Disease Models and Mechanisms, 2019, 12, . | 1.2 | 38 |
| 38 | Habituation Learning Is a Widely Affected Mechanism in Drosophila Models of Intellectual Disability and Autism Spectrum Disorders. Biological Psychiatry, 2019, 86, 294-305. | 0.7 | 39 |
| 39 | Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. BMC Cancer, 2019, 19, 435. | 1.1 | 1 |
| 40 | TCF4 (E2-2) harbors tumor suppressive functions in SHH medulloblastoma. Acta Neuropathologica, 2019, 137, 657-673. | 3.9 | 20 |
| 41 | Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071. | 1.4 | 11 |
| 42 | Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710. | 1.5 | 43 |
| 43 | Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185. | 1.1 | 133 |
| 44 | Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. European Journal of Medical Genetics, 2018, 61, 363-368. | 0.7 | 17 |
| 45 | 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 |
| 46 | Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975. | 1.1 | 67 |
| 47 | Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638. | 1.1 | 101 |
| 48 | Microphthalmia is not a mandatory finding in Xâ€linked recessive syndromic microphthalmia caused by the recurrent BCOR variant p.Pro85Leu. American Journal of Medical Genetics, Part A, 2018, 176, 2872-2876. | 0.7 | 3 |
| 49 | CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619. | 5.8 | 70 |
| 50 | X-chromosomale EntwicklungsstÃ¶rungen im weiblichen Geschlecht. Medizinische Genetik, 2018, 30, 334-341. | 0.1 | 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. | 2.6 | 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. | 2.6 | 45 |
| 53 | Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293. | 6.0 | 186 |
| 54 | FOXP2variants 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 |

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|---|---|---|---|
| 55 | STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488. | 1.5 | 35 |
| 56 | Haploinsufficiency of NR4A2 is associated with a neurodevelopmental phenotype with prominent language impairment. American Journal of Medical Genetics, Part A, 2017, 173, 2231-2234. | 0.7 | 25 |
| 57 | DDX3X mutations in two girls with a phenotype overlapping Torielloâ€“Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373. | 0.7 | 41 |
| 58 | Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. Epilepsy and Behavior, 2017, 69, 104-109. | 0.9 | 7 |
| 59 | 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 |
| 60 | Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225. | 1.6 | 53 |
| 61 | Exome Pool-Seq in neurodevelopmental disorders. European Journal of Human Genetics, 2017, 25, 1364-1376. | 1.4 | 77 |
| 62 | Eight further individuals with intellectual disability and epilepsy carrying bi-allelicCNTNAP2aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827. | 1.5 | 45 |
| 63 | Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522. | 1.5 | 135 |
| 64 | Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. European Journal of Medical Genetics, 2016, 59, 549-553. | 0.7 | 11 |
| 65 | Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764. | 1.1 | 70 |
| 66 | Zeb2 recruits HDACâ€“NuRD to inhibit Notch and controls Schwann cell differentiation and remyelination. Nature Neuroscience, 2016, 19, 1060-1072. | 7.1 | 113 |
| 67 | Clinical delineation of the PACS1â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675. | 0.7 | 44 |
| 68 | Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. American Journal of Human Genetics, 2016, 98, 149-164. | 2.6 | 270 |
| 69 | 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 |
| 70 | Mikrozephalie bei psychomotorischen EntwicklungsstÃ¶rungen und geistiger Behinderung. Medizinische Genetik, 2015, 27, 362-368. | 0.1 | 1 |
| 71 | Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. European Journal of Human Genetics, 2015, 23, 753-760. | 1.4 | 73 |
| 72 | Transcriptional repressor ZEB2 promotes terminal differentiation of CD8+ effector and memory T cell populations during infection. Journal of Experimental Medicine, 2015, 212, 2027-2039. | 4.2 | 206 |

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|---|---|---|---|
| 73 | Chromatin-Remodeling-Factor ARID1B Represses Wnt/Î²-Catenin Signaling. American Journal of Human Genetics, 2015, 97, 445-456. | 2.6 | 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, 2113OIA259. | 2.3 | 0 |
| 75 | AlteredGPM6A/M6Dosage Impairs Cognition and Causes Phenotypes Responsive to Cholesterol in Human andDrosophila. Human Mutation, 2014, 35, 1495-1505. | 1.1 | 31 |
| 76 | CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663. | 13.5 | 228 |
| 77 | Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Human Genetics, 2014, 133, 939-949. | 1.8 | 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. | 1.0 | 11 |
| 79 | Phenotype and genotype in Nicolaidesâ€“Baraitser syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 302-314. | 0.7 | 66 |
| 80 | Females with de novo aberrations in PHF6: 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 |
| 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. | 2.6 | 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. | 1.2 | 26 |
| 83 | <scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545. | 1.0 | 85 |
| 84 | Somatic mosaicism in a mother of two children with Pittâ€“Hopkins syndrome. Clinical Genetics, 2013, 83, 73-77. | 1.0 | 31 |
| 85 | De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131. | 2.6 | 151 |
| 86 | A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135. | 1.4 | 190 |
| 87 | A new face of Borjesonâ€“Forssmanâ€“Lehmann syndrome? De novo mutations inPHF6in seven females with a distinct phenotype. Journal of Medical Genetics, 2013, 50, 838-847. | 1.5 | 50 |
| 88 | Human Intellectual Disability Genes Form Conserved Functional Modules in Drosophila. PLoS Genetics, 2013, 9, e1003911. | 1.5 | 39 |
| 89 | Biallelic SEMA3A defects cause a novel type of syndromic short stature. American Journal of Medical Genetics, Part A, 2013, 161, 2880-2889. | 0.7 | 9 |
| 90 | Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682. | 6.3 | 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. | 0.7 | 20 |
| 92 | 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. American Journal of Medical Genetics, Part A, 2012, 158A, 1765-1770. | 0.7 | 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. | 2.6 | 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. | 0.7 | 9 |
| 95 | Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. Nature Genetics, 2011, 43, 20-22. | 9.4 | 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. | 1.0 | 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. | 1.1 | 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. | 1.2 | 61 |
| 100 | Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331. | 1.5 | 126 |
| 101 | New Mutations of EXT1 and EXT2 Genes in German Patients with Multiple Osteochondromas. Annals of Human Genetics, 2009, 73, 283-291. | 0.3 | 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. | 2.6 | 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. | 2.6 | 70 |
| 104 | Further delineation of Pitt-Hopkins syndrome: phenotypic and genotypic description of 16 novel patients. Journal of Medical Genetics, 2008, 45, 738-744. | 1.5 | 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. | 0.3 | 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. | 0.7 | 8 |
| 107 | Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. Cell, 2008, 135, 37-48. | 13.5 | 567 |
| 108 | Mutations in the Pericentrin ( PCNT ) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819. | 6.0 | 370 |

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|---|---|---|---|
| 109 | Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636. | 1.5 | 72 |
| 110 | SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. Journal of Medical Genetics, 2007, 44, 651-656. | 1.5 | 114 |
| 111 | Human TBX1 Missense Mutations Cause Gain of Function Resulting in the Same Phenotype as 22q11.2 Deletions. American Journal of Human Genetics, 2007, 80, 510-517. | 2.6 | 195 |
| 112 | Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001. | 2.6 | 261 |
| 113 | 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 |
| 114 | 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 |
| 115 | AtypicalZFHX1B mutation associated with a mild Mowatâ€“Wilson syndrome phenotype. American Journal of Medical Genetics, Part A, 2006, 140A, 869-872. | 0.7 | 32 |
| 116 | A missense mutation in theZFHX1B gene associated with an atypical Mowatâ€“Wilson syndrome phenotype. American Journal of Medical Genetics, Part A, 2006, 140A, 1223-1227. | 0.7 | 31 |
| 117 | Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 2063-2074. | 0.7 | 343 |
| 118 | The phenotypic spectrum in patients with arginine to cysteine mutations in the COL2A1 gene. Journal of Medical Genetics, 2005, 43, 406-413. | 1.5 | 71 |
| 119 | Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. Journal of Medical Genetics, 2005, 42, 871-876. | 1.5 | 118 |
| 120 | Genitourinary Anomalies in Mowat-Wilson Syndrome with Deletion/Mutation in the Zinc Finger Homeo Box 1B Gene (ZFHX1B). Hormone Research in Paediatrics, 2005, 63, 187-192. | 0.8 | 15 |
| 121 | Severely Incapacitating Mutations in Patients with Extreme Short Stature Identify RNA-Processing Endoribonuclease RMRP as an Essential Cell Growth Regulator. American Journal of Human Genetics, 2005, 77, 795-806. | 2.6 | 117 |
| 122 | Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111. | 0.7 | 121 |
| 123 | 9 Mb deletion including chromosome band 3q24 associated with unsuspicious facial gestalt, persistent ductus omphaloentericus, mild mental retardation and tic. European Journal of Medical Genetics, 2005, 48, 360-362. | 0.7 | 5 |
| 124 | Mowat-Wilson syndrome and mutation in the zinc finger homeo box 1B gene: a well defined clinical entity. Journal of Medical Genetics, 2004, 41, 16e-16. | 1.5 | 36 |
| 125 | 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 |
| 126 | Hirschsprung disease, mental retardation, characteristic facial features, and mutation in the geneZFHX1B (SIP1): Confirmation of the Mowat-Wilson syndrome. American Journal of Medical Genetics Part A, 2003, 116A, 385-388. | 2.4 | 58 |

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|---|---|---|---|
| 127 | Characterisation of deletions of the ZFHX1B region and genotype-phenotype analysis in Mowat-Wilson syndrome. Journal of Medical Genetics, 2003, 40, 601-605. | 1.5 | 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 |