# Anita Rauch

List of Publications by Year
in descending order

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| 256 | 17,020 | 15504 | 19749 |
|---|---|---|---|
| papers | citations | 65 h-index | 117 g-index |

| 270 | 270 | 270 | 23224 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | 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 |
| 2 | Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336. | 21.4 | 670 |
| 3 | 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 |
| 4 | Transcription Factor E2-2 Is an Essential and Specific Regulator of Plasmacytoid Dendritic Cell Development. Cell, 2008, 135, 37-48. | 28.9 | 567 |
| 5 | Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026. | 21.4 | 431 |
| 6 | Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102. | 27.8 | 394 |
| 7 | Mutations in the Pericentrin ( PCNT ) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819. | 12.6 | 370 |
| 8 | 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. | 1.2 | 343 |
| 9 | Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. American Journal of Human Genetics, 2007, 80, 550-560. | 6.2 | 316 |
| 10 | Contribution of Global Rare Copy-Number Variants to the Risk of Sporadic Congenital Heart Disease. American Journal of Human Genetics, 2012, 91, 489-501. | 6.2 | 272 |
| 11 | Haploinsufficiency of TCF4 Causes Syndromal Mental Retardation with Intermittent Hyperventilation (Pitt-Hopkins Syndrome). American Journal of Human Genetics, 2007, 80, 994-1001. | 6.2 | 261 |
| 12 | De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731. | 21.4 | 236 |
| 13 | Genotype-phenotype correlations in Noonan syndrome. Journal of Pediatrics, 2004, 144, 368-374. | 1.8 | 227 |
| 14 | 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 |
| 15 | The core FOXG1 syndrome phenotype consists of postnatal microcephaly, severe mental retardation, absent language, dyskinesia, and corpus callosum hypogenesis. Journal of Medical Genetics, 2011, 48, 396-406. | 3.2 | 220 |
| 16 | CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26. | 21.4 | 201 |
| 17 | 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. | 6.2 | 195 |
| 18 | Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20. | 11.0 | 195 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. Journal of Medical Genetics, 2009, 46, 598-606. | 3.2 | 194 |
| 20 | Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720. | 3.2 | 191 |
| 21 | 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. | 2.9 | 190 |
| 22 | Identification of a novel loss-of-function calcium channel gene mutation in short QT syndrome (SQTS6). European Heart Journal, 2011, 32, 1077-1088. | 2.2 | 178 |
| 23 | Induction, binding specificity and function of human ICOS. European Journal of Immunology, 2000, 30, 3707-3717. | 2.9 | 166 |
| 24 | 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 |
| 25 | Mutations at the SALL4 locus on chromosome 20 result in a range of clinically overlapping phenotypes, including Okihiro syndrome, Holt-Oram syndrome, acro-renal-ocular syndrome, and patients previously reported to represent thalidomide embryopathy. Journal of Medical Genetics, 2003, 40, 473-478. | 3.2 | 159 |
| 26 | NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114. | 6.2 | 151 |
| 27 | De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131. | 6.2 | 151 |
| 28 | Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. European Journal of Human Genetics, 2004, 12, 879-890. | 2.8 | 149 |
| 29 | Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963. | 2.8 | 147 |
| 30 | PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653. | 21.4 | 146 |
| 31 | Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114. | 2.8 | 144 |
| 32 | Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522. | 3.2 | 135 |
| 33 | Altered TGFÎ² signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 deficiency. European Journal of Human Genetics, 2010, 18, 895-901. | 2.8 | 132 |
| 34 | SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248. | 21.4 | 131 |
| 35 | First known microdeletion within the Wolf-Hirschhorn syndrome critical region refines genotype-phenotype correlation. American Journal of Medical Genetics Part A, 2001, 99, 338-342. | 2.4 | 128 |
| 36 | Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331. | 3.2 | 126 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925. | 6.2 | 125 |
| 38 | â€œMowatâ€Wilsonâ€•syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomaliesâ€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 |
| 39 | Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111. | 1.3 | 121 |
| 40 | Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2. Journal of Medical Genetics, 2005, 42, 871-876. | 3.2 | 118 |
| 41 | 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. | 6.2 | 117 |
| 42 | 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. | 3.2 | 114 |
| 43 | Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106. | 2.1 | 109 |
| 44 | Molecular karyotyping using an SNP array for genomewide genotyping. Journal of Medical Genetics, 2004, 41, 916-922. | 3.2 | 106 |
| 45 | Phenotype-specific effect of chromosome 1q21.1 rearrangements and GJA5 duplications in 2436 congenital heart disease patients and 6760 controls. Human Molecular Genetics, 2012, 21, 1513-1520. | 2.9 | 101 |
| 46 | Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638. | 2.4 | 101 |
| 47 | A novel microdeletion syndrome involving 5q14.3-q15: clinical and molecular cytogenetic characterization of three patients. European Journal of Human Genetics, 2009, 17, 1592-1599. | 2.8 | 96 |
| 48 | A Novel 22q11.2 Microdeletion in DiGeorge Syndrome. American Journal of Human Genetics, 1999, 64, 659-667. | 6.2 | 95 |
| 49 | Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491. | 8.2 | 89 |
| 50 | Gene discovery for Mendelian conditions via social networking: de novo variants in KDM1A cause developmental delay and distinctive facial features. Genetics in Medicine, 2016, 18, 788-795. | 2.4 | 88 |
| 51 | 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 |
| 52 | Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6. | 1.2 | 84 |
| 53 | Microcephalin and pericentrin regulate mitotic entry via centrosome-associated Chk1. Journal of Cell Biology, 2009, 185, 1149-1157. | 5.2 | 83 |
| 54 | Disturbed Wnt Signalling due to a Mutation in CCDC88C Causes an Autosomal Recessive Non-Syndromic Hydrocephalus with Medial Diverticulum. Molecular Syndromology, 2010, 1, 99-112. | 0.8 | 82 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Genome-wide association study identifies loci on 12q24 and 13q32 associated with Tetralogy of Fallot. Human Molecular Genetics, 2013, 22, 1473-1481. | 2.9 | 82 |
| 56 | Deletion mapping on chromosome 10p and definition of a critical region for the second DiGeorge syndrome locus (DGS2). European Journal of Human Genetics, 1998, 6, 213-225. | 2.8 | 81 |
| 57 | A study of ten small supernumerary (marker) chromosomes identified by fluorescence in situ hybridization (FISH). Clinical Genetics, 1992, 42, 84-90. | 2.0 | 81 |
| 58 | Type and Level of RMRP Functional Impairment Predicts Phenotype in the Cartilage Hair Hypoplasiaâ€“Anauxetic Dysplasia Spectrum. American Journal of Human Genetics, 2007, 81, 519-529. | 6.2 | 78 |
| 59 | Crisponi Syndrome Is Caused by Mutations in the CRLF1 Gene and Is Allelic to Cold-Induced Sweating Syndrome Type 1. American Journal of Human Genetics, 2007, 80, 971-981. | 6.2 | 76 |
| 60 | Allelic Heterogeneity in the COH1 Gene Explains Clinical Variabilityin Cohen Syndrome. American Journal of Human Genetics, 2004, 75, 138-145. | 6.2 | 72 |
| 61 | Klinefelter syndrome and mediastinal germ cell tumors. American Journal of Medical Genetics, Part A, 2006, 140A, 471-481. | 1.2 | 72 |
| 62 | Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636. | 3.2 | 72 |
| 63 | The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688. | 3.2 | 72 |
| 64 | De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. European Journal of Human Genetics, 2015, 23, 602-609. | 2.8 | 72 |
| 65 | 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 |
| 66 | Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201. | 3.3 | 70 |
| 67 | Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945. | 7.6 | 70 |
| 68 | FOXP2variants 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 |
| 69 | Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975. | 2.4 | 67 |
| 70 | Confirmation of mutations inPROSCas a novel cause of vitamin B<sub><sub>6</sub></sub>-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814. | 3.2 | 66 |
| 71 | Incidence and significance of 22q11.2 hemizygosity in patients with interrupted aortic arch. , 1998, 78, 322-331. | | 62 |
| 72 | Characterisation of deletions of the ZFHX1B region and genotype-phenotype analysis in Mowat-Wilson syndrome. Journal of Medical Genetics, 2003, 40, 601-605. | 3.2 | 61 |

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|---|---|---|---|
| 73 | 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 |
| 74 | Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365. | 3.5 | 60 |
| 75 | The face of Noonan syndrome: Does phenotype predict genotype. American Journal of Medical Genetics, Part A, 2010, 152A, 1960-1966. | 1.2 | 59 |
| 76 | 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 |
| 77 | Hirschsprung disease, mental retardation, characteristic facial features, and mutation in the gene ZFHX1B (SIP1): Confirmation of the Mowatâ€Wilson syndrome. American Journal of Medical Genetics Part A, 2003, 116A, 385-388. | 2.4 | 58 |
| 78 | Laterality of the aortic arch and anomalies of the subclavian artery?reliable indicators for 22q11.2 deletion syndromes?. European Journal of Pediatrics, 2004, 163, 642-5. | 2.7 | 58 |
| 79 | In-Frame Deletion and Missense Mutations of the C-Terminal Helicase Domain of <b><i>SMARCA2</i></b> in Three Patients with Nicolaides-Baraitser Syndrome. Molecular Syndromology, 2011, 2, 237-244. | 0.8 | 58 |
| 80 | A new quantitative PCR multiplex assay for rapid analysis of chromosome 17p11.2-12 duplications and deletions leading to HMSN/HNPP. European Journal of Human Genetics, 2003, 11, 170-178. | 2.8 | 57 |
| 81 | Dosage changes of MED13L further delineate its role in congenital heart defects and intellectual disability. European Journal of Human Genetics, 2013, 21, 1100-1104. | 2.8 | 57 |
| 82 | Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058. | 2.4 | 57 |
| 83 | Highly variable cutis laxa resulting from a dominant splicing mutation of the elastin gene. American Journal of Medical Genetics, Part A, 2008, 146A, 977-983. | 1.2 | 56 |
| 84 | Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222. | 6.2 | 56 |
| 85 | A Dual Phenotype of Periventricular Nodular Heterotopia and Frontometaphyseal Dysplasia in One Patient Caused by a Single FLNA Mutation Leading to Two Functionally Different Aberrant Transcripts. American Journal of Human Genetics, 2004, 74, 731-737. | 6.2 | 55 |
| 86 | Independent NF1 and PTPN11 mutations in a family with neurofibromatosisâ€Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1263-1267. | 1.2 | 55 |
| 87 | Mutations in  <scp>CDK</scp> 5 <scp>RAP</scp> 2  cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480. | 1.2 | 55 |
| 88 | Clinical and molecular cytogenetic observations in three cases of â€œtrisomy 12p syndromeâ€. American Journal of Medical Genetics Part A, 1996, 63, 243-249. | 2.4 | 54 |
| 89 | A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796. | 6.2 | 53 |
| 90 | Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332. | 2.5 | 53 |

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|---|---|---|---|
| 91 | The role of recessive inheritance in early-onset epileptic encephalopathies: a combined whole-exome sequencing and copy number study. European Journal of Human Genetics, 2019, 27, 408-421. | 2.8 | 52 |
| 92 | The MEF2C-Related and 5q14.3q15 Microdeletion Syndrome. Molecular Syndromology, 2011, 2, 164-170. | 0.8 | 51 |
| 93 | Lethal cutis laxa with contractural arachnodactyly, overgrowth and soft tissue bleeding due to a novel homozygous fibulinâ€4 gene mutation. Clinical Genetics, 2009, 76, 276-281. | 2.0 | 50 |
| 94 | 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. | 3.2 | 50 |
| 95 | Mutations in CRADD Result in Reduced Caspase-2-Mediated Neuronal Apoptosis and Cause Megalencephaly with a Rare Lissencephaly Variant. American Journal of Human Genetics, 2016, 99, 1117-1129. | 6.2 | 50 |
| 96 | Partial deletion of the critical 1.5 Mb interval in Williams-Beuren syndrome. Journal of Medical Genetics, 2003, 40, 99e-99. | 3.2 | 49 |
| 97 | 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 |
| 98 | 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 |
| 99 | IntragenicKANSL1mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotypeâ€“phenotype correlations in a large cohort of patients. Journal of Medical Genetics, 2015, 52, 804-814. | 3.2 | 47 |
| 100 | Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759. | 2.8 | 47 |
| 101 | Mosaicism for the Charcot-Marie-Tooth disease type 1A duplication suggests somatic reversion. Human Genetics, 1996, 98, 22-28. | 3.8 | 46 |
| 102 | A variable combination of features of Noonan syndrome and neurofibromatosis type I are caused by mutations in the NF1 gene. American Journal of Medical Genetics, Part A, 2006, 140A, 2749-2756. | 1.2 | 46 |
| 103 | The molecular basis of the cartilage-hair hypoplasiaâ€“anauxetic dysplasia spectrum. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 131-142. | 4.7 | 46 |
| 104 | Monosomy 22q11 in patients with pulmonary atresia, ventricular septal defect, and major aortopulmonary collateral arteries. Heart, 1998, 79, 180-185. | 2.9 | 45 |
| 105 | Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994. | 6.2 | 44 |
| 106 | The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 152 Td (a Report of eight cases including a living child and further evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1268-1281. | 1.2 | 43 |
| 107 | Microarrays in prenatal diagnosis. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 42, 53-63. | 2.8 | 43 |
| 108 | Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024. | 7.9 | 43 |

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|---|---|---|---|
| 109 | Mesomelia-Synostoses Syndrome Results from Deletion of SULF1 and SLCO5A1 Genes at 8q13. American Journal of Human Genetics, 2010, 87, 95-100. | 6.2 | 42 |
| 110 | Highâ€resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. Prenatal Diagnosis, 2014, 34, 525-533. | 2.3 | 42 |
| 111 | Further corroboration of distinct functional features in SCN2A variants causing intellectual disability or epileptic phenotypes. Molecular Medicine, 2019, 25, 6. | 4.4 | 42 |
| 112 | Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112. | 1.3 | 42 |
| 113 | Ichthyosis follicularis, alopecia, and photophobia (IFAP) syndrome: Clinical and neuropathological observations in a 33-year-old man. , 1998, 78, 371-377. | | 40 |
| 114 | The shortest of the short: Pericentrin mutations and beyond. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 125-130. | 4.7 | 40 |
| 115 | Plasma metabolomics reveals a diagnostic metabolic fingerprint for mitochondrial aconitase (ACO2) deficiency. PLoS ONE, 2017, 12, e0176363. | 2.5 | 40 |
| 116 | Neocentric small supernumerary marker chromosomes (sSMC) â€“ three more cases and review of the literature. Cytogenetic and Genome Research, 2007, 118, 31-37. | 1.1 | 37 |
| 117 | Analysis of an interstitial deletion in a patient with Kallmann syndrome, Xâ€linked ichthyosis and mental retardation. Clinical Genetics, 1998, 54, 45-51. | 2.0 | 37 |
| 118 | The smallest teeth in the world are caused by mutations in the PCNT gene. American Journal of Medical Genetics, Part A, 2011, 155, 1398-1403. | 1.2 | 37 |
| 119 | Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117. | 2.5 | 37 |
| 120 | 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. | 3.2 | 36 |
| 121 | Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796. | 2.5 | 36 |
| 122 | Autosomal-Dominant Hypertension With Type E Brachydactyly Is Caused by Rearrangement on the Short Arm of Chromosome 12. Hypertension, 2004, 43, 471-476. | 2.7 | 35 |
| 123 | Pulmonary hypoplasiaâ€“diaphragmatic herniaâ€“anophthalmiaâ€“cardiac defect (PDAC) syndrome due to STRA6 mutationsâ€”What are the minimal criteria?. American Journal of Medical Genetics, Part A, 2009, 149A, 2457-2463. | 1.2 | 35 |
| 124 | Goltzâ€“Gorlin (focal dermal hypoplasia) and the microphthalmia with linear skin defects (MLS) syndrome: no evidence of genetic overlap. European Journal of Human Genetics, 2009, 17, 1207-1215. | 2.8 | 35 |
| 125 | STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488. | 3.2 | 35 |
| 126 | A recurrent germline mutation in the PIGA gene causes Simpsonâ€Golabiâ€Behmel syndrome type 2. American Journal of Medical Genetics, Part A, 2016, 170, 392-402. | 1.2 | 34 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. European Journal of Medical Genetics, 2017, 60, 451-464. | 1.3 | 34 |
| 128 | De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. Genetics in Medicine, 2019, 21, 1008-1014. | 2.4 | 34 |
| 129 | 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 |
| 130 | N<sup>8</sup>â€acetylspermidine as a potential plasma biomarker for Snyderâ€Robinson syndrome identified by clinical metabolomics. Journal of Inherited Metabolic Disease, 2016, 39, 131-137. | 3.6 | 33 |
| 131 | A novel 5q35.3 subtelomeric deletion syndrome. American Journal of Medical Genetics, Part A, 2003, 121A, 1-8. | 1.2 | 32 |
| 132 | AtypicalZFHX1B mutation associated with a mild Mowatâ€“Wilson syndrome phenotype. American Journal of Medical Genetics, Part A, 2006, 140A, 869-872. | 1.2 | 32 |
| 133 | Unmasking of a Recessive SCARF2 Mutation by a 22q11.12 de novo Deletion in a Patient with Van den Ende-Gupta Syndrome. Molecular Syndromology, 2010, 1, 239-245. | 0.8 | 32 |
| 134 | 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 |
| 135 | "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 |
| 136 | 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. | 1.2 | 31 |
| 137 | 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 |
| 138 | Somatic mosaicism in a mother of two children with Pittâ€“Hopkins syndrome. Clinical Genetics, 2013, 83, 73-77. | 2.0 | 31 |
| 139 | Association Between C677T Polymorphism of Methylene Tetrahydrofolate Reductase and Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2013, 6, 347-353. | 5.1 | 31 |
| 140 | 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 |
| 141 | Severe skeletal dysplasia caused by undiagnosed hypothyroidism. European Journal of Medical Genetics, 2007, 50, 209-215. | 1.3 | 30 |
| 142 | Infantile Epileptic Encephalopathy, Transient Choreoathetotic Movements, and Hypersomnia due to a De Novo Missense Mutation in the SCN2A Gene. Neuropediatrics, 2014, 45, 261-264. | 0.6 | 30 |
| 143 | De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868. | 6.2 | 29 |
| 144 | Balanced translocation in a patient with craniosynostosis disrupts the SOX6 gene and an evolutionarily conserved non-transcribed region. Journal of Medical Genetics, 2006, 43, 534-540. | 3.2 | 28 |

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|---|---|---|---|
| 145 | Two novel mutations in the insulin binding subunit of the insulin receptor gene without insulin binding impairment in a patient with Rabsonâ€“Mendenhall syndrome. Molecular Genetics and Metabolism, 2008, 94, 356-362. | 1.1 | 28 |
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