# James R Lupski

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2350399/publications.pdf

Version: 2024-02-01

| | | | |
|---|---|---|---|
| 457 papers | 46,758 citations | 1893 102 h-index | 2509 196 g-index |
| 473 all docs | 473 docs citations | 473 times ranked | 41758 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511. | 27.0 | 1,717 |
| 2 | The complete genome of an individual by massively parallel DNA sequencing. Nature, 2008, 452, 872-876. | 27.8 | 1,635 |
| 3 | DNA duplication associated with Charcot-Marie-Tooth disease type 1A. Cell, 1991, 66, 219-232. | 28.9 | 1,313 |
| 4 | A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Starqardt macular dystrophy. Nature Genetics, 1997, 15, 236-246. | 21.4 | 1,277 |
| 5 | Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870. | 7.4 | 1,171 |
| 6 | Copy Number Variation in Human Health, Disease, and Evolution. Annual Review of Genomics and Human Genetics, 2009, 10, 451-481. | 6.2 | 1,026 |
| 7 | Structural Variation in the Human Genome and its Role in Disease. Annual Review of Medicine, 2010, 61, 437-455. | 12.2 | 1,015 |
| 8 | Mutation of the Stargardt Disease Gene ( ABCR ) in Age-Related Macular Degeneration. Science, 1997, 277, 1805-1807. | 12.6 | 844 |
| 9 | Genomic disorders: structural features of the genome can lead to DNA rearrangements and human disease traits. Trends in Genetics, 1998, 14, 417-422. | 6.7 | 817 |
| 10 | A DNA Replication Mechanism for Generating Nonrecurrent Rearrangements Associated with Genomic Disorders. Cell, 2007, 131, 1235-1247. | 28.9 | 756 |
| 11 | A Microhomology-Mediated Break-Induced Replication Model for the Origin of Human Copy Number Variation. PLoS Genetics, 2009, 5, e1000327. | 3.5 | 700 |
| 12 | Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191. | 27.0 | 698 |
| 13 | Triallelic Inheritance in Bardet-Biedl Syndrome, a Mendelian Recessive Disorder. Science, 2001, 293, 2256-2259. | 12.6 | 599 |
| 14 | The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215. | 6.2 | 574 |
| 15 | Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31. | 27.0 | 565 |
| 16 | The gene for the peripheral myelin protein PMPâ€“22 is a candidate for Charcotâ€“Marieâ€“Tooth disease type 1A. Nature Genetics, 1992, 1, 159-165. | 21.4 | 529 |
| 17 | Mechanisms underlying structural variant formation in genomic disorders. Nature Reviews Genetics, 2016, 17, 224-238. | 16.3 | 526 |
| 18 | Genomic Disorders: Molecular Mechanisms for Rearrangements and Conveyed Phenotypes. PLoS Genetics, 2005, 1, e49. | 3.5 | 496 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Mutations in the early growth response 2 (EGR2) gene are associated with hereditary myelinopathies. Nature Genetics, 1998, 18, 382-384. | 21.4 | 475 |
| 20 | Non-coding genetic variants in human disease: Figure 1.. Human Molecular Genetics, 2015, 24, R102-R110. | 2.9 | 466 |
| 21 | Human Genome Sequencing in Health and Disease. Annual Review of Medicine, 2012, 63, 35-61. | 12.2 | 404 |
| 22 | Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903. | 28.9 | 391 |
| 23 | Charcotâ€“Marieâ€“Tooth type 1A duplication appears to arise from recombination at repeat sequences flanking the 1.5 Mb monomer unit. Nature Genetics, 1992, 2, 292-300. | 21.4 | 385 |
| 24 | Molecular mechanism for distinct neurological phenotypes conveyed by allelic truncating mutations. Nature Genetics, 2004, 36, 361-369. | 21.4 | 383 |
| 25 | The DNA replication FoSTeS/MMBIR mechanism can generate genomic, genic and exonic complex rearrangements in humans. Nature Genetics, 2009, 41, 849-853. | 21.4 | 382 |
| 26 | Charcot-Marie-Tooth Disease Type 1A -- Association with a Spontaneous Point Mutation in the PMP22 Gene. New England Journal of Medicine, 1993, 329, 96-101. | 27.0 | 375 |
| 27 | Genomic rearrangements and sporadic disease. Nature Genetics, 2007, 39, S43-S47. | 21.4 | 373 |
| 28 | Homologous recombination of a flanking repeat gene cluster is a mechanism for a common contiguous gene deletion syndrome. Nature Genetics, 1997, 17, 154-163. | 21.4 | 364 |
| 29 | Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438. | 6.2 | 348 |
| 30 | Clan Genomics and the Complex Architecture of Human Disease. Cell, 2011, 147, 32-43. | 28.9 | 330 |
| 31 | A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214. | 28.9 | 322 |
| 32 | Mutations in MKKS cause obesity, retinal dystrophy and renal malformations associated with Bardet-Biedl syndrome. Nature Genetics, 2000, 26, 67-70. | 21.4 | 311 |
| 33 | A recombination hotspot responsible for two inherited peripheral neuropathies is located near a mariner transposon-like element. Nature Genetics, 1996, 12, 288-297. | 21.4 | 304 |
| 34 | COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660. | 21.4 | 302 |
| 35 | Molecular mechanism for duplication 17p11.2â€” the homologous recombination reciprocal of the Smith-Magenis microdeletion. Nature Genetics, 2000, 24, 84-87. | 21.4 | 297 |
| 36 | Two autosomal dominant neuropathies result from reciprocal DNA duplication/deletion of a region on chromosome 17. Human Molecular Genetics, 1994, 3, 223-228. | 2.9 | 294 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Mechanisms for recurrent and complex human genomic rearrangements. Current Opinion in Genetics and Development, 2012, 22, 211-220. | 3.3 | 289 |
| 38 | Multi-disciplinary clinical study of Smith-Magenis syndrome (deletion 17p11.2). American Journal of Medical Genetics Part A, 1996, 62, 247-254. | 2.4 | 285 |
| 39 | Genomic Rearrangements and Gene Copy-Number Alterations as a Cause of Nervous System Disorders. Neuron, 2006, 52, 103-121. | 8.1 | 284 |
| 40 | Genotype/Phenotype Analysis of a Photoreceptor-Specific ATP-Binding Cassette Transporter Gene, ABCR, in Stargardt Disease. American Journal of Human Genetics, 1999, 64, 422-434. | 6.2 | 277 |
| 41 | Dejerineâ€“Sottas syndrome associated with point mutation in the peripheral myelin protein 22 (PMP22) gene. Nature Genetics, 1993, 5, 269-273. | 21.4 | 274 |
| 42 | Whole-Genome Sequencing for Optimized Patient Management. Science Translational Medicine, 2011, 03, 87re3. | 12.4 | 272 |
| 43 | Gene dosage is a mechanism for Charcot-Marie-Tooth disease type 1A. Nature Genetics, 1992, 1, 29-33. | 21.4 | 270 |
| 44 | Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245. | 2.9 | 261 |
| 45 | Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513. | 8.1 | 258 |
| 46 | Development and validation of a CGH microarray for clinical cytogenetic diagnosis. Genetics in Medicine, 2005, 7, 422-432. | 2.4 | 241 |
| 47 | Complex human chromosomal and genomic rearrangements. Trends in Genetics, 2009, 25, 298-307. | 6.7 | 239 |
| 48 | TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350. | 27.0 | 239 |
| 49 | Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392. | 6.7 | 234 |
| 50 | Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836. | 9.0 | 234 |
| 51 | Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713. | 12.8 | 227 |
| 52 | Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342. | 2.5 | 225 |
| 53 | Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182. | 6.2 | 219 |
| 54 | Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183. | 6.4 | 211 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Evidence for a recessive PMP22 point mutation in Charcotâ€“Marieâ€“Tooth disease type 1A. Nature Genetics, 1993, 5, 189-194. | 21.4 | 208 |
| 56 | TLR7 gain-of-function genetic variation causes human lupus. Nature, 2022, 605, 349-356. | 27.8 | 208 |
| 57 | Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480. | 27.0 | 205 |
| 58 | Increased LIS1 expression affects human and mouse brain development. Nature Genetics, 2009, 41, 168-177. | 21.4 | 199 |
| 59 | Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650. | 28.9 | 189 |
| 60 | Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685. | 2.4 | 186 |
| 61 | Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. Nature Genetics, 2011, 43, 1074-1081. | 21.4 | 184 |
| 62 | Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26. | 8.2 | 184 |
| 63 | Myelin deficiencies in both the central and the peripheral nervous systems associated with aSOX10 mutation. Annals of Neurology, 1999, 46, 313-318. | 5.3 | 181 |
| 64 | Bardetâ€“Biedl syndrome is linked to DNA markers on chromosome 11 q and is genetically heterogeneous. Nature Genetics, 1994, 7, 108-112. | 21.4 | 179 |
| 65 | Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203. | 2.9 | 165 |
| 66 | Mus81 and converging forks limit the mutagenicity of replication fork breakage. Science, 2015, 349, 742-747. | 12.6 | 162 |
| 67 | Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812. | 2.4 | 161 |
| 68 | Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187. | 6.2 | 160 |
| 69 | Microarrayâ€based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. American Journal of Medical Genetics, Part A, 2007, 143A, 1679-1686. | 1.2 | 158 |
| 70 | Assessing structural variation in a personal genomeâ€”towards a human reference diploid genome. BMC Genomics, 2015, 16, 286. | 2.8 | 153 |
| 71 | The allelic spectrum of Charcotâ€“Marieâ€“Tooth disease in over 17,000 individuals with neuropathy. Molecular Genetics & Genomic Medicine, 2014, 2, 522-529. | 1.2 | 151 |
| 72 | ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367. | 6.2 | 150 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107. | 6.2 | 148 |
| 74 | Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845. | 6.2 | 146 |
| 75 | Genomic Rearrangements Resulting in PLP1 Deletion Occur by Nonhomologous End Joining and Cause Different Dysmyelinating Phenotypes in Males and Females. American Journal of Human Genetics, 2002, 71, 838-853. | 6.2 | 144 |
| 76 | Genome Mosaicismâ€"One Human, Multiple Genomes. Science, 2013, 341, 358-359. | 12.6 | 143 |
| 77 | Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. Genome Medicine, 2013, 5, 57. | 8.2 | 143 |
| 78 | Diagnosis of CMT1A duplications and HNPP deletions by interphase FISH: Implications for testing in the cytogenetics laboratory. American Journal of Medical Genetics Part A, 1997, 69, 325-331. | 2.4 | 141 |
| 79 | Phenotypic Consequences of Copy Number Variation: Insights from Smith-Magenis and Potocki-Lupski Syndrome Mouse Models. PLoS Biology, 2010, 8, e1000543. | 5.6 | 139 |
| 80 | Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood, 2018, 132, 89-100. | 1.4 | 139 |
| 81 | Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651. | 8.2 | 136 |
| 82 | Genomic disorders ten years on. Genome Medicine, 2009, 1, 42. | 8.2 | 135 |
| 83 | A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799. | 8.5 | 132 |
| 84 | The 1.4-Mb CMT1A Duplication/HNPP Deletion Genomic Region Reveals Unique Genome Architectural Features and Provides Insights into the Recent Evolution of New Genes. Genome Research, 2001, 11, 1018-1033. | 5.5 | 129 |
| 85 | Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately severe hearing loss in a patient with Smith-Magenis syndrome. Human Genetics, 2001, 109, 535-541. | 3.8 | 128 |
| 86 | De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11. | 8.2 | 128 |
| 87 | Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142. | 6.2 | 128 |
| 88 | Cell cycle arrest in Era GTPase mutants: a potential growth rateâ€regulated checkpoint in Escherichia coli. Molecular Microbiology, 1998, 27, 739-750. | 2.5 | 127 |
| 89 | Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665. | 2.8 | 127 |
| 90 | Proteolipid protein gene duplications causing Pelizaeus-Merzbacher disease: Molecular mechanism and phenotypic manifestations. Annals of Neurology, 1999, 45, 624-632. | 5.3 | 126 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability. American Journal of Human Genetics, 2010, 86, 892-903. | 6.2 | 125 |
| 92 | Replicative mechanisms for CNV formation are error prone. Nature Genetics, 2013, 45, 1319-1326. | 21.4 | 125 |
| 93 | NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309. | 6.2 | 125 |
| 94 | The Breakpoint Region of the Most Common Isochromosome, i(17q), in Human Neoplasia Is Characterized by a Complex Genomic Architecture with Large, Palindromic, Low-Copy Repeats. American Journal of Human Genetics, 2004, 74, 1-10. | 6.2 | 122 |
| 95 | Oral Curcumin Mitigates the Clinical and Neuropathologic Phenotype of the Trembler-J Mouse: A Potential Therapy for Inherited Neuropathy. American Journal of Human Genetics, 2007, 81, 438-453. | 6.2 | 122 |
| 96 | Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258. | 3.5 | 122 |
| 97 | NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409. | 5.5 | 120 |
| 98 | Structural variation mutagenesis of the human genome: Impact on disease and evolution. Environmental and Molecular Mutagenesis, 2015, 56, 419-436. | 2.2 | 119 |
| 99 | Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806. | 2.9 | 117 |
| 100 | Curcumin Treatment Abrogates Endoplasmic Reticulum Retention and Aggregation-Induced Apoptosis Associated with Neuropathy-Causing Myelin Protein Zeroâ€“Truncating Mutants. American Journal of Human Genetics, 2005, 77, 841-850. | 6.2 | 115 |
| 101 | DUF1220-Domain Copy Number Implicated in Human Brain-Size Pathology and Evolution. American Journal of Human Genetics, 2012, 91, 444-454. | 6.2 | 113 |
| 102 | Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87. | 2.8 | 112 |
| 103 | Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336. | 6.2 | 112 |
| 104 | The Centers for Mendelian Genomics: A new largeâ€scale initiative to identify the genes underlying rare Mendelian conditions. American Journal of Medical Genetics, Part A, 2012, 158A, 1523-1525. | 1.2 | 110 |
| 105 | DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622. | 6.2 | 110 |
| 106 | Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. Acta Neuropathologica Communications, 2016, 4, 56. | 5.2 | 110 |
| 107 | The rod photoreceptor ATP-binding cassette transporter gene, ABCR, and retinal disease: from monogenic to multifactorial. Vision Research, 1999, 39, 2537-2544. | 1.4 | 108 |
| 108 | Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448. | 2.5 | 105 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537. | 2.4 | 104 |
| 110 | Parent of Origin, Mosaicism, and Recurrence Risk: Probabilistic Modeling Explains the Broken Symmetry of Transmission Genetics. American Journal of Human Genetics, 2014, 95, 345-359. | 6.2 | 103 |
| 111 | Modeling del(17)(p11.2p11.2) and dup(17)(p11.2p11.2) Contiguous Gene Syndromes by Chromosome Engineering in Mice: Phenotypic Consequences of Gene Dosage Imbalance. Molecular and Cellular Biology, 2003, 23, 3646-3655. | 2.3 | 100 |
| 112 | Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237. | 14.5 | 98 |
| 113 | Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357. | 6.2 | 98 |
| 114 | Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131. | 8.1 | 95 |
| 115 | Delineation of the common critical region in Williams syndrome and clinical correlation of growth, heart defects, ethnicity, and parental origin. American Journal of Medical Genetics Part A, 1998, 78, 82-89. | 2.4 | 93 |
| 116 | Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583. | 6.2 | 92 |
| 117 | Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811. | 2.5 | 92 |
| 118 | Pro-inflammation Associated with a Gain-of-Function Mutation (R284S) in the Innate Immune Sensor STING. Cell Reports, 2018, 23, 1112-1123. | 6.4 | 92 |
| 119 | Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228. | 6.2 | 90 |
| 120 | Unusual electrophysiological findings in X-linked dominant Charcot-Marie-Tooth disease. Muscle and Nerve, 2000, 23, 182-188. | 2.2 | 89 |
| 121 | Is the carboxyl-terminus of dystrophin required for membrane association? A novel, severe case of duchenne muscular dystrophy. Annals of Neurology, 1991, 30, 605-610. | 5.3 | 88 |
| 122 | DVL3 Alleles Resulting in a â^'1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561. | 6.2 | 88 |
| 123 | WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43. | 6.2 | 88 |
| 124 | The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161. | 6.2 | 87 |
| 125 | A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299. | 2.9 | 87 |
| 126 | De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363. | 6.2 | 86 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586. | 3.8 | 85 |
| 128 | Rapid molecular diagnostics of severe primary immunodeficiency determined by using targeted next-generation sequencing. Journal of Allergy and Clinical Immunology, 2016, 138, 1142-1151.e2. | 2.9 | 85 |
| 129 | Spastic paraplegia type 2 associated with axonal neuropathy and apparent PLP1 position effect. Annals of Neurology, 2006, 59, 398-403. | 5.3 | 83 |
| 130 | Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077. | 2.9 | 83 |
| 131 | Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778. | 8.2 | 82 |
| 132 | The human COX10 gene is disrupted during homologous recombination between the 24 kb proximal and distal CMT1A-REPs. Human Molecular Genetics, 1997, 6, 1595-1603. | 2.9 | 81 |
| 133 | Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754. | 6.2 | 80 |
| 134 | Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. BMC Medical Genomics, 2016, 9, 42. | 1.5 | 80 |
| 135 | Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905. | 3.5 | 80 |
| 136 | Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923. | 6.2 | 79 |
| 137 | POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3. | 8.2 | 78 |
| 138 | Discordance of muscular dystrophy in monozygotic female twins: Evidence supporting asymmetric splitting of the inner cell mass in a manifesting carrier of Duchenne dystrophy. American Journal of Medical Genetics Part A, 1991, 40, 354-364. | 2.4 | 76 |
| 139 | Somatic mosaicism underlies X-linked acrogigantism syndrome in sporadic male subjects. Endocrine-Related Cancer, 2016, 23, 221-233. | 3.1 | 75 |
| 140 | Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988. | 2.9 | 74 |
| 141 | Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19. | 2.4 | 74 |
| 142 | Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242. | 5.5 | 74 |
| 143 | The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150. | 6.2 | 74 |
| 144 | Role of genomic architecture in PLP1 duplication causing Pelizaeus-Merzbacher disease. Human Molecular Genetics, 2006, 15, 2250-2265. | 2.9 | 73 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Reporting Genomic Sequencing Results to Ordering Clinicians. JAMA - Journal of the American Medical Association, 2013, 310, 365. | 7.4 | 73 |
| 146 | Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420. | 2.4 | 73 |
| 147 | Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10. | 28.9 | 73 |
| 148 | Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. Genome Research, 2011, 21, 33-46. | 5.5 | 72 |
| 149 | Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68. | 9.0 | 71 |
| 150 | Mutations in ANKLE2, a ZIKA Virus Target, Disrupt an Asymmetric Cell Division Pathway in Drosophila Neuroblasts to Cause Microcephaly. Developmental Cell, 2019, 51, 713-729.e6. | 7.0 | 71 |
| 151 | Perturbations of BMP/TGF-Î² and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). Journal of Medical Genetics, 2018, 55, 675-684. | 3.2 | 70 |
| 152 | Approaches for identifying germ cell mutagens: Report of the 2013 IWGT workshop on germ cell assaysâ˜†. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2015, 783, 36-54. | 1.7 | 69 |
| 153 | DNA Rearrangements on Both Homologues of Chromosome 17 in a Mildly Delayed Individual with a Family History of Autosomal Dominant Carpal Tunnel Syndrome. American Journal of Human Genetics, 1999, 64, 471-478. | 6.2 | 67 |
| 154 | Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. American Journal of Human Genetics, 2017, 101, 123-129. | 6.2 | 67 |
| 155 | Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570. | 6.2 | 66 |
| 156 | An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7. | 28.9 | 66 |
| 157 | De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724. | 6.2 | 66 |
| 158 | Rai1 duplication causes physical and behavioral phenotypes in a mouse model of dup(17)(p11.2p11.2). Journal of Clinical Investigation, 2006, 116, 3035-3041. | 8.2 | 66 |
| 159 | De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913. | 6.2 | 65 |
| 160 | HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207. | 12.6 | 65 |
| 161 | Hotspots of homologous recombination in the human genome: not all homologous sequences are equal. Genome Biology, 2004, 5, 242. | 9.6 | 64 |
| 162 | PacBio-LITS: a large-insert targeted sequencing method for characterization of human disease-associated chromosomal structural variations. BMC Genomics, 2015, 16, 214. | 2.8 | 63 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394. | 5.5 | 62 |
| 164 | Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127. | 6.2 | 62 |
| 165 | PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952. | 7.6 | 62 |
| 166 | EGR2 mutation R359W causes a spectrum of Dejerine-Sottas neuropathy. Neurogenetics, 2001, 3, 153-157. | 1.4 | 60 |
| 167 | Genomic disorders: A window into human gene and genome evolution. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1765-1771. | 7.1 | 60 |
| 168 | TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. Genetics in Medicine, 2019, 21, 1548-1558. | 2.4 | 60 |
| 169 | A potential founder variant inCARMIL2/RLTPRin three Norwegian families with warts, molluscum contagiosum, and T-cell dysfunction. Molecular Genetics & Genomic Medicine, 2016, 4, 604-616. | 1.2 | 59 |
| 170 | Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994. | 6.2 | 59 |
| 171 | T118M PMP22 mutation causes partial loss of function and HNPPâ€like neuropathy. Annals of Neurology, 2006, 59, 358-364. | 5.3 | 58 |
| 172 | Whole-exome sequencing identifies novel homozygous mutation inÂNPAS2 in family with nonobstructive azoospermia. Fertility and Sterility, 2015, 104, 286-291. | 1.0 | 58 |
| 173 | GNB5 Mutations Cause an Autosomal-Recessive Multisystem Syndrome with Sinus Bradycardia and Cognitive Disability. American Journal of Human Genetics, 2016, 99, 704-710. | 6.2 | 58 |
| 174 | Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162. | 2.4 | 58 |
| 175 | Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192. | 2.5 | 58 |
| 176 | A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501. | 2.5 | 58 |
| 177 | De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789. | 6.2 | 57 |
| 178 | Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. PLoS Genetics, 2015, 11, e1005050. | 3.5 | 57 |
| 179 | Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893. | 6.2 | 57 |
| 180 | The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. Human Genetics, 2018, 137, 553-567. | 3.8 | 57 |

| # | Article | IF | Citations |
|---|---|---|---|
| 181 | Conservation and evolution of the rpsU-dnaG-rpoD macromolecular synthesis operon in bacteria. Molecular Microbiology, 1993, 8, 343-355. | 2.5 | 56 |
| 182 | Myelin protein zero (MPZ) gene mutations in nonduplication type 1 Charcot-Marie-Tooth disease. Human Mutation, 1996, 7, 36-45. | 2.5 | 56 |
| 183 | Enriched rearing improves behavioral responses of an animal model for CNV-based autistic-like traits. Human Molecular Genetics, 2012, 21, 3083-3096. | 2.9 | 56 |
| 184 | Phenotypic expansion of TBX4 mutations to include acinar dysplasia of the lungs. American Journal of Medical Genetics, Part A, 2016, 170, 2440-2444. | 1.2 | 56 |
| 185 | Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316. | 6.2 | 56 |
| 186 | Mutational and genotypeâ€“phenotype correlation analyses in 28 Polish patients with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1531-1541. | 1.2 | 55 |
| 187 | Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660. | 6.2 | 55 |
| 188 | Digenic inheritance and Mendelian disease. Nature Genetics, 2012, 44, 1291-1292. | 21.4 | 54 |
| 189 | Mutations inVRK1Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8. | 9.0 | 54 |
| 190 | Longitudinal studies of the duplication form of Charcot-Marie-Tooth polyneuropathy. , 1996, 19, 74-78. | | 53 |
| 191 | Compensating for central nervous system dysmyelination: Females with a proteolipid protein gene duplication and sustained clinical improvement. Annals of Neurology, 2001, 50, 747-754. | 5.3 | 53 |
| 192 | Alu-specific microhomology-mediated deletion of the final exon of SPAST in three unrelated subjects with hereditary spastic paraplegia. Genetics in Medicine, 2011, 13, 582-592. | 2.4 | 53 |
| 193 | Mutations in COL27A1 cause Steel syndrome and suggest a founder mutation effect in the Puerto Rican population. European Journal of Human Genetics, 2015, 23, 342-346. | 2.8 | 53 |
| 194 | Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067. | 3.6 | 53 |
| 195 | Characterization of molecular and cellular phenotypes associated with a heterozygous CNTNAP2 deletion using patient-derived hiPSC neural cells. NPJ Schizophrenia, 2015, 1, . | 3.6 | 52 |
| 196 | Germline PRKACA amplification causes variable phenotypes that may depend on the extent of the genomic defect: molecular mechanisms and clinical presentations. European Journal of Endocrinology, 2015, 172, 803-811. | 3.7 | 52 |
| 197 | Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339. | 2.8 | 52 |
| 198 | Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675. | 2.4 | 52 |

| # | Article | IF | Citations |
|---|---|---|---|
| 199 | The Smith-Magenis syndrome [del(17)p11.2]: Clinical review and molecular advances. Mental Retardation and Developmental Disabilities Research Reviews, 1996, 2, 122-129. | 3.6 | 51 |
| 200 | Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. Genetics in Medicine, 2012, 14, 868-876. | 2.4 | 51 |
| 201 | Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978. | 2.8 | 51 |
| 202 | Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83. | 8.2 | 50 |
| 203 | Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220. | 2.5 | 48 |
| 204 | Molecular Genetics and Neuropathology of Charcot-Marie-Tooth Disease Type 1A. Brain Pathology, 1992, 2, 337-349. | 4.1 | 47 |
| 205 | Quantification by flow cytometry of chromosome-17 deletions in Smith-Magenis syndrome patients. Human Genetics, 1996, 98, 710-718. | 3.8 | 47 |
| 206 | Genomic Disorders: Recombination-Based Disease Resulting from Genome Architecture**Previously presented at the annual meeting of The American Society of Human Genetics, in Baltimore, on October 19, 2002.. American Journal of Human Genetics, 2003, 72, 246-252. | 6.2 | 47 |
| 207 | Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54. | 8.2 | 47 |
| 208 | Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippelâ€“Feil syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2795-2799. | 1.2 | 47 |
| 209 | NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625. | 12.8 | 47 |
| 210 | RAI1 point mutations, CAG repeat variation, and SNP analysis in non-deletion Smithâ€“Magenis syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2454-2463. | 1.2 | 46 |
| 211 | Incriminating genomic evidence. Nature, 2008, 455, 178-179. | 27.8 | 46 |
| 212 | TBX6 compound inheritance leads to congenital vertebral malformations in humans and mice. Human Molecular Genetics, 2019, 28, 539-547. | 2.9 | 46 |
| 213 | Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564. | 6.2 | 45 |
| 214 | Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446. | 3.5 | 45 |
| 215 | Aggressive tumor growth and clinical evolution in a patient with X-linked acro-gigantism syndrome. Endocrine, 2016, 51, 236-244. | 2.3 | 45 |
| 216 | Biallelic Mutations in UNC80 Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. American Journal of Human Genetics, 2016, 98, 202-209. | 6.2 | 45 |

| # | Article | IF | Citations |
|---|---|---|---|
| 217 | REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156. | 6.2 | 44 |
| 218 | Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573. | 2.8 | 44 |
| 219 | Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797. | 2.4 | 44 |
| 220 | MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106. | 8.2 | 43 |
| 221 | Mechanism, Prevalence, and More Severe Neuropathy Phenotype of the Charcot-Marie-Tooth Type 1A Triplication. American Journal of Human Genetics, 2014, 94, 462-469. | 6.2 | 42 |
| 222 | Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30. | 8.2 | 42 |
| 223 | A syndrome of short stature, microcephaly and speech delay is associated with duplications reciprocal to the common Sotos syndrome deletion. European Journal of Human Genetics, 2010, 18, 258-261. | 2.8 | 41 |
| 224 | A Human in Human Genetics. Cell, 2019, 177, 9-15. | 28.9 | 41 |
| 225 | Perturbations of genes essential for MÃ¼llerian duct and WÃ¶lffian duct development in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345. | 6.2 | 41 |
| 226 | Charcot-Marie-Tooth Polyneuropathy: Duplication, Gene Dosage, and Genetic Heterogeneity. Pediatric Research, 1999, 45, 159-165. | 2.3 | 41 |
| 227 | Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578. | 6.2 | 40 |
| 228 | Passage Number is a Major Contributor to Genomic Structural Variations in Mouse iPSCs. Stem Cells, 2014, 32, 2657-2667. | 3.2 | 40 |
| 229 | De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409. | 3.8 | 40 |
| 230 | Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617. | 21.4 | 40 |
| 231 | Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708. | 12.8 | 40 |
| 232 | Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). Journal of Medical Genetics, 2021, 58, 41-47. | 3.2 | 40 |
| 233 | A clinical and molecular study of mosaicism for trisomy 17. Human Genetics, 1996, 97, 69-72. | 3.8 | 39 |
| 234 | Reduced penetrance of craniofacial anomalies as a function of deletion size and genetic background in a chromosome engineered partial mouse model for Smithâ€“Magenis syndrome. Human Molecular Genetics, 2004, 13, 2613-2624. | 2.9 | 39 |

| # | Article | IF | Citations |
|---|---|---|---|
| 235 | Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. Human Genetics, 2016, 135, 9-19. | 3.8 | 39 |
| 236 | Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73. | 8.2 | 39 |
| 237 | Mutations in the mitochondrial ribosomal protein MRPS22 lead to primary ovarian insufficiency. Human Molecular Genetics, 2018, 27, 1913-1926. | 2.9 | 39 |
| 238 | High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005. | 6.2 | 38 |
| 239 | New Polymorphic Short Tandem Repeats for PCR-based Charcot-Marie-Tooth Disease Type 1A Duplication Diagnosis. Clinical Chemistry, 2001, 47, 838-843. | 3.2 | 37 |
| 240 | Penetrance of Craniofacial Anomalies in Mouse Models of Smith-Magenis Syndrome Is Modified by Genomic Sequence Surrounding Rai1: Not All Null Alleles Are Alike. American Journal of Human Genetics, 2007, 80, 518-525. | 6.2 | 37 |
| 241 | Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54. | 2.4 | 37 |
| 242 | CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076. | 2.8 | 37 |
| 243 | A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. Genome Medicine, 2016, 8, 13. | 8.2 | 37 |
| 244 | Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95. | 8.2 | 37 |
| 245 | Translation of SOX10 3' untranslated region causes a complex severe neurocristopathy by generation of a deleterious functional domain. Human Molecular Genetics, 2007, 16, 3037-3046. | 2.9 | 36 |
| 246 | A Duplication CNV That Conveys Traits Reciprocal to Metabolic Syndrome and Protects against Diet-Induced Obesity in Mice and Men. PLoS Genetics, 2012, 8, e1002713. | 3.5 | 36 |
| 247 | Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386. | 3.8 | 36 |
| 248 | Mutations in PI3K110Î´ cause impaired natural killer cell function partially rescued by rapamycin treatment. Journal of Allergy and Clinical Immunology, 2018, 142, 605-617.e7. | 2.9 | 36 |
| 249 | CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641. | 2.4 | 36 |
| 250 | Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250. | 6.2 | 36 |
| 251 | Stable inheritance of the CMT1A DNA duplication in two patients with CMT1 and NF1. American Journal of Medical Genetics Part A, 1993, 45, 92-96. | 2.4 | 35 |
| 252 | Prenatal interphase FISH diagnosis of PLP1 duplication associated with Pelizaeusâ€“Merzbacher disease. Prenatal Diagnosis, 2001, 21, 1133-1136. | 2.3 | 35 |

| # | Article | IF | Citations |
|---|---|---|---|
| 253 | A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351. | 6.2 | 35 |
| 254 | Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131. | 2.8 | 35 |
| 255 | Phenotypic expansion of POGZâ€related intellectual disability syndrome (Whiteâ€Sutton) Tj ETQq1 1 0.784314 rgBT /Overloc | 1.2 | 35 |
| 256 | Charcot-Marie-Tooth Disease: A Gene-Dosage Effect. Hospital Practice (1995), 1997, 32, 83-122. | 1.0 | 34 |
| 257 | Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252. | 2.8 | 34 |
| 258 | The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326. | 1.2 | 34 |
| 259 | Defining the breakpoints of proximal chromosome 14q rearrangements in nine patients using flow-sorted chromosomes. American Journal of Medical Genetics Part A, 2001, 102, 173-182. | 2.4 | 33 |
| 260 | New mutations and intellectual function. Nature Genetics, 2010, 42, 1036-1038. | 21.4 | 33 |
| 261 | Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707. | 6.2 | 33 |
| 262 | Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130. | 7.6 | 33 |
| 263 | Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279. | 6.2 | 33 |
| 264 | Denaturing high-performance liquid chromatography of the myotubularin-related 2 gene (MTMR2) in unrelated patients with Charcot-Marie-Tooth disease suggests a low frequency of mutation in inherited neuropathy. Neurogenetics, 2001, 3, 107-109. | 1.4 | 32 |
| 265 | An evaluation of the draft human genome sequence. Nature Genetics, 2001, 29, 88-91. | 21.4 | 32 |
| 266 | Human subtelomeric copy number gains suggest a DNA replication mechanism for formation: beyond breakageâ€“fusionâ€“bridge for telomere stabilization. Human Genetics, 2012, 131, 1895-1910. | 3.8 | 32 |
| 267 | From genomic medicine to precision medicine: highlights of 2015. Genome Medicine, 2016, 8, 12. | 8.2 | 32 |
| 268 | Targeted Treatment of Individuals With Psychosis Carrying a Copy Number Variant Containing a Genomic Triplication of the Glycine Decarboxylase Gene. Biological Psychiatry, 2019, 86, 523-535. | 1.3 | 32 |
| 269 | Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. Journal of Experimental Medicine, 2021, 218, . | 8.5 | 31 |
| 270 | NIPBL rearrangements in Cornelia de Lange syndrome: evidence for replicative mechanism and genotypeâ€“phenotype correlation. Genetics in Medicine, 2012, 14, 313-322. | 2.4 | 30 |

| # | Article | IF | Citations |
|---|---|---|---|
| 271 | Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D. Genetics in Medicine, 2014, 16, 386-394. | 2.4 | 30 |
| 272 | New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780. | 2.9 | 30 |
| 273 | Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056. | 6.2 | 30 |
| 274 | A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986. | 6.2 | 30 |
| 275 | Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541. | 6.2 | 30 |
| 276 | Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776. | 2.4 | 30 |
| 277 | The Deep Genome Project. Genome Biology, 2020, 21, 18. | 8.8 | 30 |
| 278 | De novo proximal interstitial deletions of 14q: Cytogenetic and molecular investigations. American Journal of Medical Genetics Part A, 1994, 52, 44-50. | 2.4 | 29 |
| 279 | Multiplede novo MPZ (P0) point mutations in a sporadic Dejerine-Sottas case. , 1997, 10, 21-24. | | 29 |
| 280 | Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028. | 2.5 | 29 |
| 281 | New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133. | 2.5 | 28 |
| 282 | A novel NAA10 variant with impaired acetyltransferase activity causes developmental delay, intellectual disability, and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1294-1305. | 2.8 | 28 |
| 283 | Exploring the utility of wholeâ€exome sequencing as a diagnostic tool in a child withÂatypical episodic muscle weakness. Clinical Genetics, 2013, 83, 457-461. | 2.0 | 27 |
| 284 | TBX6 missense variants expand the mutational spectrum in a nonâ€Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195. | 2.5 | 27 |
| 285 | Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654. | 2.5 | 27 |
| 286 | Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. European Journal of Human Genetics, 2020, 28, 1243-1264. | 2.8 | 27 |
| 287 | Absence ofPMP22 coding region mutations in CMT1A duplication patients: Further evidence supporting gene dosage as a mechanism for charcot-marie-tooth disease type 1A. Human Mutation, 1996, 8, 362-365. | 2.5 | 26 |
| 288 | Recessive Charcot-Marie-Tooth disease. Annals of Neurology, 2000, 47, 6-8. | 5.3 | 26 |

| # | Article | IF | Citations |
|---|---|---|---|
| 289 | Clinical utility of whole-exome sequencing in rare diseases: Galactosialidosis. European Journal of Medical Genetics, 2014, 57, 339-344. | 1.3 | 26 |
| 290 | Dual molecular diagnosis contributes to atypical Praderâ€“Willi phenotype in monozygotic twins. American Journal of Medical Genetics, Part A, 2017, 173, 2451-2455. | 1.2 | 26 |
| 291 | Marker chromosome genomic structure and temporal origin implicate a chromoanasynthesis event in a family with pleiotropic psychiatric phenotypes. Human Mutation, 2018, 39, 939-946. | 2.5 | 26 |
| 292 | Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499. | 1.4 | 26 |
| 293 | Harnessing genomics to identify environmental determinants of heritable disease. Mutation Research - Reviews in Mutation Research, 2013, 752, 6-9. | 5.5 | 25 |
| 294 | PEHO Syndrome May Represent Phenotypic Expansion at the Severe End of the Early-Onset Encephalopathies. Pediatric Neurology, 2016, 60, 83-87. | 2.1 | 25 |
| 295 | Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504. | 3.5 | 25 |
| 296 | Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347. | 2.4 | 25 |
| 297 | Clan genomics: From <scp>OMIM</scp> phenotypic traits to genes and biology. American Journal of Medical Genetics, Part A, 2021, 185, 3294-3313. | 1.2 | 25 |
| 298 | Smith-Magenis syndrome deletion: A case with equivocal cytogenetic findings resolved by fluorescence in situ hybridization. American Journal of Medical Genetics Part A, 1995, 58, 286-291. | 2.4 | 24 |
| 299 | Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53. | 3.2 | 24 |
| 300 | Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703. | 3.8 | 24 |
| 301 | Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015. | 6.2 | 24 |
| 302 | Distinct patterns of complex rearrangements and a mutational signature of microhomeology are frequently observed in PLP1 copy number gain structural variants. Genome Medicine, 2019, 11, 80. | 8.2 | 24 |
| 303 | PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365. | 2.7 | 24 |
| 304 | 22q11.2q13 duplication including SOX10 causes sexâ€reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. American Journal of Medical Genetics, Part A, 2017, 173, 1066-1070. | 1.2 | 23 |
| 305 | Novel Combined Immune Deficiency and Radiation Sensitivity Blended Phenotype in an Adult with Biallelic Variations in ZAP70 and RNF168. Frontiers in Immunology, 2017, 8, 576. | 4.8 | 23 |
| 306 | Increased TBX6 gene dosages induce congenital cervical vertebral malformations in humans and mice. Journal of Medical Genetics, 2020, 57, 371-379. | 3.2 | 23 |

| # | Article | IF | Citations |
|---|---|---|---|
| 307 | Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. Human Genetics, 2021, 140, 1011-1029. | 3.8 | 23 |
| 308 | Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25. | 8.2 | 22 |
| 309 | Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725. | 2.4 | 22 |
| 310 | An evolution revolution provides further revelation. BioEssays, 2007, 29, 1182-1184. | 2.5 | 21 |
| 311 | Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. PLoS Genetics, 2015, 11, e1005686. | 3.5 | 21 |
| 312 | Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. Journal of Genetics and Genomics, 2021, 48, 396-402. | 3.9 | 21 |
| 313 | Prenatal diagnosis of Charcot-Marie-Tooth disease type 1A by interphase fluorescencein situ hybridization. , 1999, 19, 446-449. | | 20 |
| 314 | Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. Genome Medicine, 2016, 8, 105. | 8.2 | 20 |
| 315 | Efficient CNV breakpoint analysis reveals unexpected structural complexity and correlation of dosage-sensitive genes with clinical severity in genomic disorders. Human Molecular Genetics, 2017, 26, 1927-1941. | 2.9 | 20 |
| 316 | Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406. | 3.7 | 20 |
| 317 | Missense mutations in the 3' end of the Escherichia coli dnaG gene do not abolish primase activity but do confer the chromosome-segregation-defective (par) phenotype. Microbiology (United Kingdom), 1997, 143, 585-594. | 1.8 | 19 |
| 318 | Prenatal ultrasonographic description and postnatal pathological findings in atelosteogenesis type 1. , 1998, 79, 392-395. | | 19 |
| 319 | Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia. European Journal of Human Genetics, 2014, 22, 1145-1148. | 2.8 | 19 |
| 320 | Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1â€“q35.3 susceptibility locus identified by whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 73-78. | 2.8 | 19 |
| 321 | A large CRISPR-induced bystander mutation causes immune dysregulation. Communications Biology, 2019, 2, 70. | 4.4 | 19 |
| 322 | Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. Molecular Genetics & Genomic Medicine, 2020, 8, e1023. | 1.2 | 19 |
| 323 | Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873. | 2.4 | 19 |
| 324 | Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. Human Mutation, 2016, 37, 231-234. | 2.5 | 18 |

| # | Article | IF | Citations |
|---|---|---|---|
| 325 | The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. Genetics in Medicine, 2016, 18, 443-451. | 2.4 | 18 |
| 326 | An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984. | 1.2 | 18 |
| 327 | Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807. | 6.2 | 18 |
| 328 | Novel Heterozygous Mutation in NFKB2 Is Associated With Early Onset CVID and a Functional Defect in NK Cells Complicated by Disseminated CMV Infection and Severe Nephrotic Syndrome. Frontiers in Pediatrics, 2019, 7, 303. | 1.9 | 18 |
| 329 | Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60. | 4.1 | 18 |
| 330 | Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776. | 2.5 | 18 |
| 331 | Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464. | 2.5 | 17 |
| 332 | Wholeâ€genome sequencing reveals complex chromosome rearrangement disrupting <scp>NIPBL</scp> in infant with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1143-1151. | 1.2 | 17 |
| 333 | Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924. | 7.6 | 17 |
| 334 | IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312. | 3.8 | 17 |
| 335 | Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. Kidney International, 2020, 98, 1020-1030. | 5.2 | 17 |
| 336 | Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. Human Mutation, 2016, 37, 160-164. | 2.5 | 16 |
| 337 | The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075. | 2.9 | 16 |
| 338 | Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600. | 1.2 | 16 |
| 339 | NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. Human Molecular Genetics, 2021, 29, 3516-3531. | 2.9 | 16 |
| 340 | Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55. | 8.2 | 16 |
| 341 | A novel homozygous <scp>SLC13A5</scp> wholeâ€gene deletion generated by <scp>Alu/Alu</scp>â€mediated rearrangement in an Iraqi family with epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 1972-1980. | 1.2 | 16 |
| 342 | Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137. | 2.4 | 16 |

| # | Article | IF | Citations |
|---|---|---|---|
| 343 | Dna ? DNA, and DNA ? RNA ? protein: Orchestration by a single complex operon. BioEssays, 1989, 10, 152-157. | 2.5 | 15 |
| 344 | Cerebral visual impairment and intellectual disability caused by PGAP1 variants. European Journal of Human Genetics, 2015, 23, 1689-1693. | 2.8 | 15 |
| 345 | CNV instability associated with DNA replication dynamics: evidence for replicative mechanisms in CNV mutagenesis. Human Molecular Genetics, 2015, 24, 1574-1583. | 2.9 | 15 |
| 346 | Clinical genomics: from a truly personal genome viewpoint. Human Genetics, 2016, 135, 591-601. | 3.8 | 15 |
| 347 | Prevalence of spinocerebellar ataxia 36 in a US population. Neurology: Genetics, 2017, 3, e174. | 1.9 | 15 |
| 348 | Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173. | 0.7 | 15 |
| 349 | Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066. | 1.2 | 15 |
| 350 | Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. Human Mutation, 2020, 41, 150-168. | 2.5 | 15 |
| 351 | Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627. | 3.7 | 15 |
| 352 | Biology in balance: human diploid genome integrity, gene dosage, and genomic medicine. Trends in Genetics, 2022, 38, 554-571. | 6.7 | 15 |
| 353 | Severe clinical phenotype due to an interstitial deletion of the short arm of chromosome 1: A brief review. , 1997, 71, 189-193. | | 14 |
| 354 | Heterozygous CTNNB1 and TBX4 variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. Clinical Genetics, 2019, 96, 366-370. | 2.0 | 14 |
| 355 | Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002. | 3.1 | 14 |
| 356 | Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941. | 2.9 | 14 |
| 357 | <scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399. | 1.2 | 14 |
| 358 | <scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833. | 5.3 | 14 |
| 359 | Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic proteinâ€truncating alleles in Xiaâ€“Gibbs syndrome. Human Mutation, 2021, 42, 577-591. | 2.5 | 14 |
| 360 | Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. American Journal of Medical Genetics, Part A, 2022, 188, 735-750. | 1.2 | 14 |

| # | Article | IF | Citations |
|---|---|---|---|
| 361 | Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074. | 1.7 | 14 |
| 362 | Di George anomaly associated with ade novo Y;22 translocation resulting in monosomy del(22)(q11.2). American Journal of Medical Genetics Part A, 1991, 40, 196-198. | 2.4 | 13 |
| 363 | Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 2014, 2, 144-146. | 1.3 | 13 |
| 364 | From genes to genomes in the clinic. Genome Medicine, 2015, 7, 78. | 8.2 | 13 |
| 365 | First Case of CD40LG Deficiency in Ecuador, Diagnosed after Whole Exome Sequencing in a Patient with Severe Cutaneous Histoplasmosis. Frontiers in Pediatrics, 2017, 5, 17. | 1.9 | 13 |
| 366 | Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. Molecular Genetics and Metabolism, 2018, 125, 302-304. | 1.1 | 13 |
| 367 | Neurodevelopmental disorder in an Egyptian family with a biallelic <scp>ALKBH8</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 1288-1293. | 1.2 | 13 |
| 368 | Isolation and Characterization of Suppressors of Two Escherichia coli dnaG Mutations, dnaG2903 and parB. Genetics, 1997, 145, 867-875. | 2.9 | 13 |
| 369 | Opposing phenotypes in mice with Smithâ€“Magenis deletion and Potockâ€“Lupski duplication syndromes suggest gene dosage effects on fluid consumption behavior. American Journal of Medical Genetics, Part A, 2012, 158A, 2807-2814. | 1.2 | 12 |
| 370 | Duplications, deletions, and single-nucleotide variations: the complexity of genetic arithmetic. Genetics in Medicine, 2013, 15, 172-173. | 2.4 | 12 |
| 371 | Exome sequencing identifies a homozygous C5orf42 variant in a Turkish kindred with oralâ€facialâ€digital syndrome type VI. American Journal of Medical Genetics, Part A, 2015, 167, 2132-2137. | 1.2 | 12 |
| 372 | Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998. | 2.5 | 12 |
| 373 | Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the Literature. Frontiers in Pediatrics, 2021, 9, 673957. | 1.9 | 12 |
| 374 | Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373. | 2.4 | 12 |
| 375 | Cognitive Phenotypes and Genomic Copy Number Variations. JAMA - Journal of the American Medical Association, 2015, 313, 2029. | 7.4 | 11 |
| 376 | Phenotypic Expansion of Congenital Disorder of Glycosylation Due to SRD5A3 Null Mutation. JIMD Reports, 2015, 26, 7-12. | 1.5 | 11 |
| 377 | Two male sibs with severe micrognathia and a missense variant in MED12. European Journal of Medical Genetics, 2016, 59, 367-372. | 1.3 | 11 |
| 378 | Divergent Levels of Marker Chromosomes in an hiPSC-Based Model ofÂPsychosis. Stem Cell Reports, 2017, 8, 519-528. | 4.8 | 11 |

| # | Article | IF | Citations |
|---|---|---|---|
| 379 | A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1015-1022. | 1.2 | 11 |
| 380 | 2018 Victor A. McKusick Leadership Award: Molecular Mechanisms for Genomic and Chromosomal Rearrangements. American Journal of Human Genetics, 2019, 104, 391-406. | 6.2 | 11 |
| 381 | Ten years of Genome Medicine. Genome Medicine, 2019, 11, 7. | 8.2 | 11 |
| 382 | Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498. | 1.9 | 11 |
| 383 | Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317. | 6.2 | 11 |
| 384 | Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , . | 2.5 | 11 |
| 385 | Whole-exome sequencing reveals an inherited R566X mutation of the epithelial sodium channel Î²-subunit in a case of early-onset phenotype of Liddle syndrome. Journal of Physical Education and Sports Management, 2016, 2, a001255. | 1.2 | 10 |
| 386 | Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. Parkinsonism and Related Disorders, 2021, 82, 84-86. | 2.2 | 10 |
| 387 | Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014. | 1.7 | 10 |
| 388 | Prenatal Diagnosis of Charcot-Marie-Tooth Disease Type 1A. Annals of the New York Academy of Sciences, 1999, 883, 457-459. | 3.8 | 9 |
| 389 | CHRNA7 Deletions are Enriched in Risperidone-Treated Children and Adolescents. Journal of Child and Adolescent Psychopharmacology, 2017, 27, 908-915. | 1.3 | 9 |
| 390 | Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911. | 2.4 | 9 |
| 391 | Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460. | 2.4 | 9 |
| 392 | Balancing between Adaptive and Maladaptive Cellular Stress Responses in Peripheral Neuropathy. Neuron, 2008, 57, 329-330. | 8.1 | 8 |
| 393 | Brain Copy Number Variants and Neuropsychiatric Traits. Biological Psychiatry, 2012, 72, 617-619. | 1.3 | 8 |
| 394 | CAV3 mutation in a patient with transient hyperCKemia and myalgia. Neurologia I Neurochirurgia Polska, 2016, 50, 468-473. | 1.2 | 8 |
| 395 | Chimeric transcripts resulting from complex duplications in chromosome Xq28. Human Genetics, 2016, 135, 253-256. | 3.8 | 8 |
| 396 | Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753. | 0.9 | 8 |

| # | Article | IF | Citations |
|---|---|---|---|
| 397 | Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83. | 7.6 | 8 |
| 398 | Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925. | 1.2 | 8 |
| 399 | Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104. | 2.5 | 8 |
| 400 | Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082. | 6.2 | 8 |
| 401 | Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney International, 2022, 101, 1039-1053. | 5.2 | 8 |
| 402 | Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918. | 2.5 | 8 |
| 403 | 2012 highlights in translational 'omics. Genome Medicine, 2013, 5, 10. | 8.2 | 7 |
| 404 | SVachra: a tool to identify genomic structural variation in mate pair sequencing data containing inward and outward facing reads. BMC Genomics, 2017, 18, 691. | 2.8 | 7 |
| 405 | Xq26.3 Duplication in a Boy With Motor Delay and Low Muscle Tone Refines the X-Linked Acrogigantism Genetic Locus. Journal of the Endocrine Society, 2018, 2, 1100-1108. | 0.2 | 7 |
| 406 | A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. American Journal of Medical Genetics, Part A, 2018, 176, 1897-1909. | 1.2 | 7 |
| 407 | Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. Journal of NeuroInterventional Surgery, 2020, 12, 221-226. | 3.3 | 7 |
| 408 | Quantitative Assessment of Parental Somatic Mosaicism for Copyâ€Number Variant (CNV) Deletions. Current Protocols in Human Genetics, 2020, 106, e99. | 3.5 | 7 |
| 409 | Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249. | 1.2 | 7 |
| 410 | Clinical presentation and evolution of Xiaâ€Gibbs syndrome due to p.Gly375ArgfsTer3 variant in a patient from DR Congo (Central Africa). American Journal of Medical Genetics, Part A, 2021, 185, 990-994. | 1.2 | 7 |
| 411 | Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11. | 3.8 | 7 |
| 412 | <scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540. | 2.0 | 7 |
| 413 | De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. Npj Genomic Medicine, 2021, 6, 104. | 3.8 | 7 |
| 414 | Dominant Transmission Observed in Adolescents and Families With Orthostatic Intolerance. Pediatric Neurology, 2017, 66, 53-58.e5. | 2.1 | 6 |

| # | Article | IF | Citations |
|---|---|---|---|
| 415 | Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. Neurology: Genetics, 2021, 7, e589. | 1.9 | 6 |
| 416 | Risk of sudden cardiac death in <scp>EXOSC5</scp>â€related disease. American Journal of Medical Genetics, Part A, 2021, 185, 2532-2540. | 1.2 | 6 |
| 417 | Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. Journal of Allergy and Clinical Immunology, 2022, 149, 758-766. | 2.9 | 6 |
| 418 | A Patient with Berardinelli-Seip Syndrome, Novel <i>AGPAT2</i> Splicesite Mutation and Concomitant Development of Non-diabetic Polyneuropathy. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 319-326. | 0.9 | 6 |
| 419 | Niacin therapy improves outcome and normalizes metabolic abnormalities in an NAXD-deficient patient. Brain, 2022, 145, e36-e40. | 7.6 | 6 |
| 420 | Settling the myelin protein zero question in CMT1B. Nature Genetics, 1995, 11, 119-120. | 21.4 | 5 |
| 421 | Animal models for human contiguous gene syndromes and other genomic disorders. Genetics and Molecular Biology, 2004, 27, 305-320. | 1.3 | 5 |
| 422 | Short stature and growth hormone deficiency in a subset of patients with <scp>Potockiâ€“Lupski</scp> syndrome: Expanding the phenotype of <scp>PTLS</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2077-2084. | 1.2 | 5 |
| 423 | Variants in FLRT3 and SLC35E2B identified using exome sequencing in seven high myopia families from Central Europe. Advances in Medical Sciences, 2021, 66, 192-198. | 2.1 | 5 |
| 424 | Chromoanagenesis Event Underlies a de novo Pericentric and Multiple Paracentric Inversions in a Single Chromosome Causing Coffinâ€“Siris Syndrome. Frontiers in Genetics, 2021, 12, 708348. | 2.3 | 5 |
| 425 | AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049. | 1.7 | 5 |
| 426 | Functional analysis of mutations in the transcription terminator T1 that suppress two dnaG alleles in Escherichia coli. Molecular Genetics and Genomics, 1995, 246, 729-733. | 2.4 | 4 |
| 427 | Duplication of thePMP22 gene in 17p partial trisomy patients with Charcot-Marie-Tooth type-1A neuropathy. Human Genetics, 1996, 97, 642-649. | 3.8 | 4 |
| 428 | Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smithâ€“Magenis syndrome with evident peripheral neuropathy. Human Genetics, 2016, 135, 1161-1174. | 3.8 | 4 |
| 429 | Linking newborn severe combined immunodeficiency screening with targeted exome sequencing: A case report. Journal of Allergy and Clinical Immunology: in Practice, 2017, 5, 1442-1444. | 3.8 | 4 |
| 430 | Diagnosis of CMT1A duplications and HNPP deletions by interphase FISH: Implications for testing in the cytogenetics laboratory. American Journal of Medical Genetics Part A, 1997, 69, 325-331. | 2.4 | 4 |
| 431 | Novel RETREG1 (<scp>FAM134B</scp> founder allele is linked to <scp>HSAN2B</scp> and renal disease in a Turkish family. American Journal of Medical Genetics, Part A, 2022, 188, 2153-2161. | 1.2 | 4 |
| 432 | DNA rearrangements affecting dosage sensitive genes. Mental Retardation and Developmental Disabilities Research Reviews, 1996, 2, 139-146. | 3.6 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 433 | Regional localization of 10 mariner transposon-like ESTs by means of FISHâ€"evidence for a correlation with fragile sites. Mammalian Genome, 2001, 12, 326-328. | 2.2 | 3 |
| 434 | Objective measures of sleep disturbances in children with Potockiâ€"Lupski syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1982-1986. | 1.2 | 3 |
| 435 | Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808. | 6.2 | 3 |
| 436 | Longitudinal studies of the duplication form of Charcotâ€Marieâ€Tooth polyneuropathy. Muscle and Nerve, 1996, 19, 74-78. | 2.2 | 3 |
| 437 | Expanding the phenotypic and allelic spectrum of <scp>SMG8</scp>: Clinical observations reveal overlap with <scp>SMG9</scp>â€associated disease trait. American Journal of Medical Genetics, Part A, 2022, 188, 648-657. | 1.2 | 3 |
| 438 | Novel dominant and recessive variants in human ROBO1 cause distinct neurodevelopmental defects through different mechanisms. Human Molecular Genetics, 2022, 31, 2751-2765. | 2.9 | 3 |
| 439 | De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366. | 1.2 | 3 |
| 440 | Short stature and combined immunodeficiency associated with mutations in RGS10. Science Signaling, 2021, 14, . | 3.6 | 2 |
| 441 | Proteolipid protein gene duplications causing Pelizaeusâ€Merzbacher disease: Molecular mechanism and phenotypic manifestations. Annals of Neurology, 1999, 45, 624-632. | 5.3 | 2 |
| 442 | Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321. | 5.3 | 2 |
| 443 | Copy number analysis of the low-copy repeats at the primate NPHP1 locus by array comparative genomic hybridization. Genomics Data, 2016, 8, 106-109. | 1.3 | 1 |
| 444 | Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. Annals of Clinical and Translational Neurology, 2021, 8, 2052-2058. | 3.7 | 1 |
| 445 | The CMT1A Duplication. , 2006, , 3-17. | | 1 |
| 446 | CRISPR/Cas9-induced gene conversion between ATAD3 paralogs. Human Genetics and Genomics Advances, 2022, 3, 100092. | 1.7 | 1 |
| 447 | Introduction of Arthur L. Beaudet, Harland Sanders Award Recipient. Genetics in Medicine, 2002, 4, 396-398. | 2.4 | 0 |
| 448 | Reply to InÃ¡cio et al. European Journal of Human Genetics, 2007, 15, 534-534. | 2.8 | 0 |
| 449 | Allan Award Introduction: Arthur L. Beaudet. American Journal of Human Genetics, 2008, 82, 1032-1033. | 6.2 | 0 |
| 450 | A Rare Novel Tyrosine Hydroxylase Gene Deletion in Parkinson Disease. Human Mutation, 2010, 31, v-v. | 2.5 | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 451 | Front Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, i. | 2.5 | 0 |
| 452 | Inside Back Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, ii. | 2.5 | 0 |
| 453 | Abstract 11800: Whole Exome Sequencing in a Large Pedigree With DCM Identifies a Novel Mutation in RBM20. Circulation, 2015, 132, . | 1.6 | 0 |
| 454 | Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, . | | 0 |
| 455 | Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644. | 2.4 | 0 |
| 456 | MO047: Biallelic pathogenic variants in ROBO1 associate with syndromic CAKUT. Nephrology Dialysis Transplantation, 2022, 37, . | 0.7 | 0 |
| 457 | Back Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, . | 2.5 | 0 |