# Wendy Chung

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8826911/publications.pdf

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|---|---|---|---|
| 586 papers | 46,279 citations | 3264 94 h-index | 3782 185 g-index |
| 621 all docs | 621 docs citations | 621 times ranked | 58787 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Questioning the validity of clinically available breast cancer polygenic risk scores: comparison of two labs reveals discrepancies. Familial Cancer, 2022, 21, 125-127. | 0.9 | 0 |
| 2 | Neurodevelopmental phenotypes associated with pathogenic variants in SLC6A1. Journal of Medical Genetics, 2022, 59, 536-543. | 1.5 | 18 |
| 3 | Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677. | 1.5 | 5 |
| 4 | Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. Amino Acids, 2022, 54, 485-499. | 1.2 | 19 |
| 5 | Biallelic variants of ATP13A3 cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. Journal of Medical Genetics, 2022, 59, 906-911. | 1.5 | 22 |
| 6 | Neurodevelopmental profile of HIVEP2â€related disorder. Developmental Medicine and Child Neurology, 2022, 64, 654-661. | 1.1 | 3 |
| 7 | Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074. | 1.0 | 14 |
| 8 | Genetics Dictating Therapeutic Decisions in Pediatric Pulmonary Hypertension? A Case Report Suggesting We Are Getting Closer.. Pulmonary Circulation, 2022, 12, e12033. | 0.8 | 5 |
| 9 | Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541. | 0.8 | 90 |
| 10 | The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298. | 2.6 | 21 |
| 11 | Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362. | 1.4 | 23 |
| 12 | Extracorporeal membrane oxygenation (ECMO) and its complications in newborns with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2022, , . | 0.8 | 4 |
| 13 | Genotype-Phenotype Comparison in POGZ-Related Neurodevelopmental Disorders by Using Clinical Scoring. Genes, 2022, 13, 154. | 1.0 | 6 |
| 14 | Neurogenetic disorders across the lifespan: from aberrant development to degeneration. Nature Reviews Neurology, 2022, 18, 117-124. | 4.9 | 19 |
| 15 | Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003500. | 1.6 | 8 |
| 16 | Channelopathy Genes in Pulmonary Arterial Hypertension. Biomolecules, 2022, 12, 265. | 1.8 | 6 |
| 17 | The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network. Genetics in Medicine, 2022, 24, 1130-1138. | 1.1 | 12 |
| 18 | Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. Circulation, 2022, 145, 877-891. | 1.6 | 18 |

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|---|---|---|---|
| 19 | OP035: Rapid Whole Genome Sequencing (rWGS) in the cardiac NICU. Genetics in Medicine, 2022, 24, S362-S363. | 1.1 | 1 |
| 20 | eP123: Design of a phase 2, double-blind, placebo-controlled trial of setmelanotide in patients with genetic variants in the melanocortin-4 receptor pathway. Genetics in Medicine, 2022, 24, S77. | 1.1 | 0 |
| 21 | Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617. | 2.6 | 16 |
| 22 | Neither cardiac mitochondrial DNA variation nor copy number contribute to congenital heart disease risk. American Journal of Human Genetics, 2022, 109, 961-966. | 2.6 | 5 |
| 23 | Generation of three induced pluripotent stem cells lines from patients with esophageal atresia/tracheoesophageal fistula type C. Stem Cell Research, 2022, 60, 102711. | 0.3 | 1 |
| 24 | Loss-of-function variants in TIAM1 are associated with developmental delay, intellectual disability, and seizures. American Journal of Human Genetics, 2022, 109, 571-586. | 2.6 | 19 |
| 25 | Identification and validation of candidate risk genes in endocytic vesicular trafficking associated with esophageal atresia and tracheoesophageal fistulas. Human Genetics and Genomics Advances, 2022, 3, 100107. | 1.0 | 2 |
| 26 | Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52. | 1.8 | 10 |
| 27 | Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835. | 3.4 | 25 |
| 28 | Discovering the Developmental Basis of Tracheaâ€Esophageal Birth Defects: Evidence for Endosomeâ€opathies. FASEB Journal, 2022, 36, . | 0.2 | 0 |
| 29 | Information is power: The experiences, attitudes and needs of individuals who chose to have prenatal genomic sequencing for fetal anomalies. Prenatal Diagnosis, 2022, 42, 947-954. | 1.1 | 3 |
| 30 | Is there a way to reduce the inequity in variant interpretation on the basis of ancestry?. American Journal of Human Genetics, 2022, 109, 981-988. | 2.6 | 13 |
| 31 | ACMG SF v3.1 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1407-1414. | 1.1 | 119 |
| 32 | Newborn screening for neurodevelopmental diseases: Are we there yet?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 222-230. | 0.7 | 10 |
| 33 | Response to Faulkner et al.. Genetics in Medicine, 2021, 23, 243. | 1.1 | 0 |
| 34 | A novel homozygous variant in TRAPPC2L results in a neurodevelopmental disorder and disrupts TRAPP complex function. Journal of Medical Genetics, 2021, 58, 592-601. | 1.5 | 10 |
| 35 | Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, . | 1.6 | 29 |
| 36 | Common Childhood Viruses and Pubertal Timing: The LEGACY Girls Study. American Journal of Epidemiology, 2021, 190, 766-778. | 1.6 | 3 |

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|---|---|---|---|
| 37 | Role of Aberrant Spontaneous Neurotransmission in SNAP25-Associated Encephalopathies. Neuron, 2021, 109, 59-72.e5. | 3.8 | 31 |
| 38 | PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317. | 1.0 | 7 |
| 39 | Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130. | 1.4 | 17 |
| 40 | Prepubertal Internalizing Symptoms and Timing of Puberty Onset in Girls. American Journal of Epidemiology, 2021, 190, 431-438. | 1.6 | 14 |
| 41 | Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457. | 3.0 | 34 |
| 42 | Cases in Precision Medicine: The Role of Polygenic Risk Scores in Breast Cancer Risk Assessment. Annals of Internal Medicine, 2021, 174, 408-412. | 2.0 | 13 |
| 43 | Weightâ€loss response to naltrexone/bupropion is modulated by the <scp>Taq1A</scp> genetic variant near <scp>DRD2</scp> (<scp>rs1800497</scp>): A pilot study. Diabetes, Obesity and Metabolism, 2021, 23, 850-853. | 2.2 | 10 |
| 44 | De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660. | 1.1 | 20 |
| 45 | Comparing 5-Year and Lifetime Risks of Breast CancerÂusing the Prospective Family Study Cohort. Journal of the National Cancer Institute, 2021, 113, 785-791. | 3.0 | 13 |
| 46 | The Steroid Metabolome and Breast Cancer Risk in Women with a Family History of Breast Cancer: The Novel Role of Adrenal Androgens and Glucocorticoids. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 89-96. | 1.1 | 8 |
| 47 | Returning negative results from <scp>largeâ€scale</scp> genomic screening: Experiences from the <scp>eMERGE III</scp> network. American Journal of Medical Genetics, Part A, 2021, 185, 508-516. | 0.7 | 5 |
| 48 | Reply to â€œ<scp>PPP2R5D</scp> Genetic Mutations and Early Onset Parkinsonismâ€• Annals of Neurology, 2021, 89, 195-196. | 2.8 | 1 |
| 49 | The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362. | 1.1 | 23 |
| 50 | Heterozygous variants that disturb the transcriptional repressor activity of FOXP4 cause a developmental disorder with speech/language delays and multiple congenital abnormalities. Genetics in Medicine, 2021, 23, 534-542. | 1.1 | 17 |
| 51 | Genetic Variant Reinterpretation: Economic and Population Health Management Challenges. Population Health Management, 2021, 24, 310-313. | 0.8 | 5 |
| 52 | United States Pulmonary Hypertension Scientific Registry. Chest, 2021, 159, 311-327. | 0.4 | 25 |
| 53 | Genes that drive the pathobiology of pediatric pulmonary arterial hypertension. Pediatric Pulmonology, 2021, 56, 614-620. | 1.0 | 16 |
| 54 | Detailed Clinical and Psychological Phenotype of the X-linked HNRNPH2-Related Neurodevelopmental Disorder. Neurology: Genetics, 2021, 7, e551. | 0.9 | 16 |

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|---|---|---|---|
| 55 | A Genetics-First Approach to Dissecting the Heterogeneity of Autism: Phenotypic Comparison of Autism Risk Copy Number Variants. American Journal of Psychiatry, 2021, 178, 77-86. | 4.0 | 62 |
| 56 | Brief Report: Impact of COVID-19 on Individuals with ASD and Their Caregivers: A Perspective from the SPARK Cohort. Journal of Autism and Developmental Disorders, 2021, 51, 3766-3773. | 1.7 | 97 |
| 57 | MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510. | 5.8 | 85 |
| 58 | An electronic health record (EHR) log analysis shows limited clinician engagement with unsolicited genetic test results. JAMIA Open, 2021, 4, ooab014. | 1.0 | 5 |
| 59 | Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636. | 1.4 | 17 |
| 60 | Does the law require reinterpretation and return of revised genomic results?. Genetics in Medicine, 2021, 23, 833-836. | 1.1 | 14 |
| 61 | A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078. | 5.8 | 19 |
| 62 | The psychiatric phenotypes of 1q21 distal deletion and duplication. Translational Psychiatry, 2021, 11, 105. | 2.4 | 6 |
| 63 | Early Pandemic Experiences of Autistic Adults: Predictors of Psychological Distress. Autism Research, 2021, 14, 1209-1219. | 2.1 | 48 |
| 64 | Response to Li and Hopper. American Journal of Human Genetics, 2021, 108, 527-529. | 2.6 | 5 |
| 65 | Availability of Services and Caregiver Burden: Supporting Individuals With Neurogenetic Conditions During the COVID-19 Pandemic. Journal of Child Neurology, 2021, 36, 760-767. | 0.7 | 16 |
| 66 | Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1399-1415. | 1.1 | 64 |
| 67 | Frequency and characterization of mutations in genes in a large cohort of patients referred to MODY registry. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 633-638. | 0.4 | 9 |
| 68 | Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. Journal of Crohn's and Colitis, 2021, 15, 1908-1919. | 0.6 | 7 |
| 69 | Newborn screening pilot study in time of pandemic: Duchenne Muscular Dystrophy pilot project experience. Molecular Genetics and Metabolism, 2021, 132, S359. | 0.5 | 0 |
| 70 | Reproductive decisionâ€making in families containing multiple individuals with epilepsy. Epilepsia, 2021, 62, 1220-1230. | 2.6 | 5 |
| 71 | Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169. | 2.0 | 27 |
| 72 | Genotype and defects in microtubule-based motility correlate with clinical severity in KIF1A-associated neurological disorder. Human Genetics and Genomics Advances, 2021, 2, 100026. | 1.0 | 34 |

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|---|---|---|---|
| 73 | Parental perceptions of genomic sequencing for expanded newborn screening. Molecular Genetics and Metabolism, 2021, 132, S339-S340. | 0.5 | 0 |
| 74 | Genetic Causes of Cardiomyopathy in Children: First Results From the Pediatric Cardiomyopathy Genes Study. Journal of the American Heart Association, 2021, 10, e017731. | 1.6 | 29 |
| 75 | Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044. | 1.4 | 14 |
| 76 | Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2021 update: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1391-1398. | 1.1 | 145 |
| 77 | Practice Patterns After Return of Rare Variants Associated With Cardiomyopathy in the Electronic Medical Records and Genomics Network. Circulation: Heart Failure, 2021, 14, e008155. | 1.6 | 1 |
| 78 | Genomic medicine implementation protocols in the PhenX Toolkit: tools for standardized data collection. Genetics in Medicine, 2021, 23, 1783-1788. | 1.1 | 2 |
| 79 | Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. Genome Medicine, 2021, 13, 80. | 3.6 | 43 |
| 80 | 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. | 1.1 | 22 |
| 81 | ACMG SF v3.0 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1381-1390. | 1.1 | 356 |
| 82 | UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635. | 1.1 | 7 |
| 83 | Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873. | 2.6 | 19 |
| 84 | Neurodevelopmental phenotypes in individuals with pathogenic variants in CHAMP1. Journal of Physical Education and Sports Management, 2021, 7, a006092. | 0.5 | 9 |
| 85 | 16p11.2 deletion syndrome. Current Opinion in Genetics and Development, 2021, 68, 49-56. | 1.5 | 39 |
| 86 | The predictive ability of the 313 variantâ€“based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. Genetics in Medicine, 2021, 23, 1726-1737. | 1.1 | 16 |
| 87 | Recommendation of premarital genetic screening in the Syrian Jewish community based on mutation carrier frequencies within Syrian Jewish cohorts. Molecular Genetics & Genomic Medicine, 2021, 9, e1756. | 0.6 | 5 |
| 88 | De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341. | 2.6 | 18 |
| 89 | Clinical and genomic characterization of 8p cytogenomic disorders. Genetics in Medicine, 2021, 23, 2342-2351. | 1.1 | 3 |
| 90 | Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846. | 1.1 | 3 |

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|---|---|---|---|
| 91 | Case Report: Esophageal Bronchus in a Neonate, With Image, Histological, and Molecular Analysis. Frontiers in Pediatrics, 2021, 9, 707822. | 0.9 | 3 |
| 92 | Impact of Genetic Testing for Cardiomyopathy on Emotional Well-Being and Family Dynamics: A Study of Parents and Adolescents. Circulation Genomic and Precision Medicine, 2021, 14, e003189. | 1.6 | 2 |
| 93 | Generalizability of Polygenic Risk Scores for Breast Cancer Among Women With European, African, and Latinx Ancestry. JAMA Network Open, 2021, 4, e2119084. | 2.8 | 31 |
| 94 | Harnessing rare variants in neuropsychiatric and neurodevelopment disordersâ€"a Keystone Symposia report. Annals of the New York Academy of Sciences, 2021, , . | 1.8 | 2 |
| 95 | PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425. | 1.1 | 8 |
| 96 | Neuropathological Findings in a Case of Parkinsonism and Developmental Delay Associated with a Monoallelic Variant in <scp>PLXNA1</scp>. Movement Disorders, 2021, 36, 2681-2687. | 2.2 | 5 |
| 97 | Gene expression atlas of energy balance brain regions. JCI Insight, 2021, 6, . | 2.3 | 6 |
| 98 | Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. Genome Medicine, 2021, 13, 147. | 3.6 | 4 |
| 99 | A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. New England Journal of Medicine, 2021, 385, 1292-1301. | 13.9 | 23 |
| 100 | Biâ€allelic PAGR1 variants are associated with microcephaly and a severe neurodevelopmental disorder: Genetic evidence from two families. American Journal of Medical Genetics, Part A, 2021, , . | 0.7 | 3 |
| 101 | Developmental basis of trachea-esophageal birth defects. Developmental Biology, 2021, 477, 85-97. | 0.9 | 21 |
| 102 | Rare and de novo variants in 827 congenital diaphragmatic hernia probands implicate LONP1 as candidate risk gene. American Journal of Human Genetics, 2021, 108, 1964-1980. | 2.6 | 22 |
| 103 | Implementation of Population-Based Newborn Screening Reveals Low Incidence of Spinal Muscular Atrophy. Obstetrical and Gynecological Survey, 2021, 76, 17-19. | 0.2 | 1 |
| 104 | GeneLiFT: A novel test to facilitate rapid screening of genetic literacy in a diverse population undergoing genetic testing. Journal of Genetic Counseling, 2021, 30, 742-754. | 0.9 | 16 |
| 105 | Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016. | 2.6 | 11 |
| 106 | Biodistribution of onasemnogene abeparvovec DNA, mRNA and SMN protein in human tissue. Nature Medicine, 2021, 27, 1701-1711. | 15.2 | 49 |
| 107 | Reimbursement for genetic variant reinterpretation: five questions payers should ask. American Journal of Managed Care, 2021, 27, e336-e338. | 0.8 | 3 |
| 108 | Association Between Genetic Testing for Hereditary Breast Cancer and Contralateral Prophylactic Mastectomy Among Multiethnic Women Diagnosed With Early-Stage Breast Cancer. JCO Oncology Practice, 2021, , OP2100322. | 1.4 | 2 |

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|---|---|---|---|
| 109 | Powerful gene-based testing by integrating long-range chromatin interactions and knockoff genotypes. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, . | 3.3 | 5 |
| 110 | Cross-sectional, quantitative analysis of motor function in females with HNRNPH2-related disorder. Research in Developmental Disabilities, 2021, 119, 104110. | 1.2 | 6 |
| 111 | Recreational Physical Activity and Outcomes After Breast Cancer in Women at High Familial Risk. JNCI Cancer Spectrum, 2021, 5, pkab090. | 1.4 | 1 |
| 112 | PH Roundtable: Genetics and Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 168-175. | 0.1 | 0 |
| 113 | Genomics of Pulmonary Hypertension. Advances in Pulmonary Hypertension, 2021, 20, 142-149. | 0.1 | 0 |
| 114 | Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638. | 0.4 | 39 |
| 115 | Recreational Physical Activity Is Associated with Reduced Breast Cancer Risk in Adult Women at High Risk for Breast Cancer: A Cohort Study of Women Selected for Familial and Genetic Risk. Cancer Research, 2020, 80, 116-125. | 0.4 | 37 |
| 116 | Is there a duty to reinterpret genetic data? The ethical dimensions. Genetics in Medicine, 2020, 22, 633-639. | 1.1 | 51 |
| 117 | The influence of genetics in congenital diaphragmatic hernia. Seminars in Perinatology, 2020, 44, 151169. | 1.1 | 39 |
| 118 | Comparative outcomes of right versus left congenital diaphragmatic hernia: A multicenter analysis. Journal of Pediatric Surgery, 2020, 55, 33-38. | 0.8 | 22 |
| 119 | Psychotic symptoms in 16p11.2 copyâ€number variant carriers. Autism Research, 2020, 13, 187-198. | 2.1 | 11 |
| 120 | Impact of patient education videos on genetic counseling outcomes after exome sequencing. Patient Education and Counseling, 2020, 103, 127-135. | 1.0 | 18 |
| 121 | Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73. | 9.4 | 120 |
| 122 | Genetic Basis of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Biology, 2020, 12, a036749. | 2.3 | 34 |
| 123 | Abnormal Auditory Mismatch Fields in Children and Adolescents With 16p11.2 Deletion and 16p11.2 Duplication. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2020, 5, 942-950. | 1.1 | 1 |
| 124 | Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756. | 13.7 | 369 |
| 125 | Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918. | 4.9 | 139 |
| 126 | Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666. | 1.1 | 82 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348. | 1.7 | 24 |
| 128 | Language characterization in 16p11.2 deletion and duplication syndromes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 380-391. | 1.1 | 16 |
| 129 | Common germline-somatic variant interactions in advanced urothelial cancer. Nature Communications, 2020, 11, 6195. | 5.8 | 21 |
| 130 | Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, . | 4.7 | 43 |
| 131 | Human iPSC-Derived Neuronal Cells From CTBP1-Mutated Patients Reveal Altered Expression of Neurodevelopmental Gene Networks. Frontiers in Neuroscience, 2020, 14, 562292. | 1.4 | 6 |
| 132 | Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444. | 2.6 | 124 |
| 133 | Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829. | 1.1 | 25 |
| 134 | Likely damaging de novo variants in congenital diaphragmatic hernia patients are associated with worse clinical outcomes. Genetics in Medicine, 2020, 22, 2020-2028. | 1.1 | 21 |
| 135 | Influence of pubertal development on urinary oxidative stress biomarkers in adolescent girls in the New York LEGACY cohort. Free Radical Research, 2020, 54, 431-441. | 1.5 | 5 |
| 136 | Earlyâ€Onset Parkinsonism Is a Manifestation of the <scp>PPP2R5D</scp> p.<scp>E200K</scp> Mutation. Annals of Neurology, 2020, 88, 1028-1033. | 2.8 | 34 |
| 137 | Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513. | 2.6 | 48 |
| 138 | Deep whole-genome sequencing of multiple proband tissues and parental blood reveals the complex genetic etiology of congenital diaphragmatic hernias. Human Genetics and Genomics Advances, 2020, 1, 100008. | 1.0 | 5 |
| 139 | Genetics and Genomics of Pediatric Pulmonary Arterial Hypertension. Genes, 2020, 11, 1213. | 1.0 | 24 |
| 140 | Efficacy and safety of setmelanotide, an MC4R agonist, in individuals with severe obesity due to LEPR or POMC deficiency: single-arm, open-label, multicentre, phase 3 trials. Lancet Diabetes and Endocrinology,the, 2020, 8, 960-970. | 5.5 | 235 |
| 141 | Impact of Coronavirus Disease 2019 (COVIDâ€19) on Patients With Congenital Heart Disease Across the Lifespan: The Experience of an Academic Congenital Heart Disease Center in New York City. Journal of the American Heart Association, 2020, 9, e017580. | 1.6 | 46 |
| 142 | NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976. | 2.6 | 18 |
| 143 | Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595. | 2.6 | 110 |
| 144 | Implementation of population-based newborn screening reveals low incidence of spinal muscular atrophy. Genetics in Medicine, 2020, 22, 1296-1302. | 1.1 | 57 |

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|---|---|---|---|
| 145 | Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581. | 9.4 | 265 |
| 146 | Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226. | 1.1 | 22 |
| 147 | A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586. | 0.9 | 16 |
| 148 | Choices, attitudes, and experiences of genetic screening in Latino/a and Ashkenazi Jewish individuals. Journal of Community Genetics, 2020, 11, 391-403. | 0.5 | 4 |
| 149 | Returning Results in the Genomic Era: Initial Experiences of the eMERGE Network. Journal of Personalized Medicine, 2020, 10, 30. | 1.1 | 39 |
| 150 | Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477. | 1.1 | 61 |
| 151 | COVID-19â€™s Impact on Genetics at One Medical Center in New York. Genetics in Medicine, 2020, 22, 1467-1469. | 1.1 | 17 |
| 152 | Precision Medicine in Diabetes: A Consensus Report From the American Diabetes Association (ADA) and the European Association for the Study of Diabetes (EASD). Diabetes Care, 2020, 43, 1617-1635. | 4.3 | 204 |
| 153 | An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587. | 1.1 | 10 |
| 154 | Novel Mutations and Decreased Expression of the Epigenetic Regulator TET2 in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000. | 1.6 | 75 |
| 155 | Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777. | 9.4 | 97 |
| 156 | Evaluating heterogeneity in <scp>ASD</scp> symptomatology, cognitive ability, and adaptive functioning among 16p11.2 <scp>CNV</scp> carriers. Autism Research, 2020, 13, 1300-1310. | 2.1 | 23 |
| 157 | SARS-CoV-2 Infection in Patients with Down Syndrome, Congenital Heart Disease, and Pulmonary Hypertension: Is Down Syndrome a Risk Factor?. Journal of Pediatrics, 2020, 225, 246-248. | 0.9 | 27 |
| 158 | Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218. | 3.4 | 48 |
| 159 | Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1 0.784314 rgBT /Overlo | 1.1 | 16 |
| 160 | De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. Circulation Genomic and Precision Medicine, 2020, 13, e002836. | 1.6 | 30 |
| 161 | De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973. | 0.7 | 8 |
| 162 | Psychiatric and Medical Profiles of Autistic Adults in the SPARK Cohort. Journal of Autism and Developmental Disorders, 2020, 50, 3679-3698. | 1.7 | 33 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468. | 0.6 | 32 |
| 164 | Genetics and Other Omics in Pediatric Pulmonary Arterial Hypertension. Chest, 2020, 157, 1287-1295. | 0.4 | 20 |
| 165 | Phenotypic expansion of <scp>Boschâ€“Boonstraâ€“Schaaf</scp> optic atrophy syndrome and further evidence for genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2020, 182, 1426-1437. | 0.7 | 27 |
| 166 | Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483. | 2.6 | 31 |
| 167 | Rapid exome sequencing in PICU patients with new-onset metabolic or neurological disorders. Pediatric Research, 2020, 88, 761-768. | 1.1 | 19 |
| 168 | Differences in brain structure and function in children with the FTO obesityâ€risk allele. Obesity Science and Practice, 2020, 6, 409-424. | 1.0 | 11 |
| 169 | A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376. | 7.7 | 83 |
| 170 | Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932. | 5.8 | 105 |
| 171 | Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. PLoS Genetics, 2020, 16, e1009189. | 1.5 | 27 |
| 172 | GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. ELife, 2020, 9, . | 2.8 | 31 |
| 173 | A Novel On-Site Volunteer Community Infection Prevention Team Prevented Outbreaks at a Hurricane Harvey Mega-Shelter. Infection Control and Hospital Epidemiology, 2020, 41, s100-s100. | 1.0 | 0 |
| 174 | Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147. | 1.7 | 1 |
| 175 | Increased yield of full GBA sequencing in Ashkenazi Jews with Parkinson's disease. European Journal of Medical Genetics, 2019, 62, 65-69. | 0.7 | 49 |
| 176 | Assessing patient readiness for personalized genomic medicine. Journal of Community Genetics, 2019, 10, 109-120. | 0.5 | 10 |
| 177 | Response to ten Broeke et al.. Genetics in Medicine, 2019, 21, 258-259. | 1.1 | 2 |
| 178 | Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19. | 1.7 | 163 |
| 179 | Applying Deep Neural Network Analysis to High-Content Image-Based Assays. SLAS Discovery, 2019, 24, 829-841. | 1.4 | 22 |
| 180 | De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424. | 2.6 | 43 |

| # | Article | IF | Citations |
|---|---|---|---|
| 181 | Clinical and genetic characterization of individuals with predicted deleterious PHIP variants. Journal of Physical Education and Sports Management, 2019, 5, a004200. | 0.5 | 14 |
| 182 | Genomic Sequencing for Infants and Children in Intensive Care Units. Current Pediatrics Reports, 2019, 7, 78-82. | 1.7 | 1 |
| 183 | De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. Nature Communications, 2019, 10, 4722. | 5.8 | 58 |
| 184 | Biallelic variants in AGMO with diminished enzyme activity are associated with a neurodevelopmental disorder. Human Genetics, 2019, 138, 1259-1266. | 1.8 | 10 |
| 185 | Functional Consequences of the SCN5A-p.Y1977N Mutation within the PY Ubiquitylation Motif: Discrepancy between HEK293 Cells and Transgenic Mice. International Journal of Molecular Sciences, 2019, 20, 5033. | 1.8 | 11 |
| 186 | The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38. | 2.3 | 28 |
| 187 | Genomic Information for Clinicians in the Electronic Health Record: Lessons Learned From the Clinical Genome Resource Project and the Electronic Medical Records and Genomics Network. Frontiers in Genetics, 2019, 10, 1059. | 1.1 | 40 |
| 188 | Accuracy of Risk Estimates from the iPrevent Breast Cancer Risk Assessment and Management Tool. JNCI Cancer Spectrum, 2019, 3, pkz066. | 1.4 | 8 |
| 189 | Genetic attribution and perceived impact of epilepsy in multiplex epilepsy families. Epilepsia, 2019, 60, 2286-2293. | 2.6 | 4 |
| 190 | Familial X-Linked Acrogigantism: Postnatal Outcomes and Tumor Pathology in a Prenatally Diagnosed Infant and His Mother. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 4667-4675. | 1.8 | 20 |
| 191 | Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605. | 2.6 | 99 |
| 192 | Psychiatric disorders in children with 16p11.2 deletion and duplication. Translational Psychiatry, 2019, 9, 8. | 2.4 | 93 |
| 193 | Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2019, 121, 180-192. | 2.9 | 19 |
| 194 | Identification of a secondary RET mutation in a pediatric patient with relapsed acute myeloid leukemia leads to the diagnosis and treatment of asymptomatic metastatic medullary thyroid cancer in a parent: a case for sequencing the germline. Journal of Physical Education and Sports Management, 2019, 5, a003889. | 0.5 | 2 |
| 195 | Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 14049-14054. | 3.3 | 30 |
| 196 | Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 2413-2421. | 1.1 | 378 |
| 197 | Psychological outcomes related to exome and genome sequencing result disclosure: a meta-analysis of seven Clinical Sequencing Exploratory Research (CSER) Consortium studies. Genetics in Medicine, 2019, 21, 2781-2790. | 1.1 | 55 |
| 198 | United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. Pulmonary Circulation, 2019, 9, 204589401985169. | 0.8 | 7 |

| # | Article | IF | Citations |
|---|---|---|---|
| 199 | A GENETIC FIRST APPROACH TO DISSECTING THE HETEROGENEITY OF AUTISM: PHENOTYPIC COMPARISON OF AUTISM RISK COPY NUMBER VARIANTS. European Neuropsychopharmacology, 2019, 29, S783-S784. | 0.3 | 6 |
| 200 | Points to consider in the reevaluation and reanalysis of genomic test results: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 1267-1270. | 1.1 | 147 |
| 201 | Homozygous noncanonical splice variant in LSM1 in two siblings with multiple congenital anomalies and global developmental delay. Journal of Physical Education and Sports Management, 2019, 5, a004101. | 0.5 | 4 |
| 202 | Regular use of aspirin and other non-steroidal anti-inflammatory drugs and breast cancer risk for women at familial or genetic risk: a cohort study. Breast Cancer Research, 2019, 21, 52. | 2.2 | 44 |
| 203 | A pathogenic CtBP1 missense mutation causes altered cofactor binding and transcriptional activity. Neurogenetics, 2019, 20, 129-143. | 0.7 | 16 |
| 204 | Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741. | 5.8 | 90 |
| 205 | Rethinking the â€œopen futureâ€•argument against predictive genetic testing of children. Genetics in Medicine, 2019, 21, 2190-2198. | 1.1 | 43 |
| 206 | Evaluation of the cost and effectiveness of diverse recruitment methods for a genetic screening study. Genetics in Medicine, 2019, 21, 2371-2380. | 1.1 | 10 |
| 207 | SMPD1 mutations, activity, and Î±â€synuclein accumulation in Parkinson's disease. Movement Disorders, 2019, 34, 526-535. | 2.2 | 81 |
| 208 | Association of Prepubertal and Adolescent Androgen Concentrations With Timing of Breast Development and Family History of Breast Cancer. JAMA Network Open, 2019, 2, e190083. | 2.8 | 7 |
| 209 | Benign breast disease increases breast cancer risk independent of underlying familial risk profile: Findings from a Prospective Family Study Cohort. International Journal of Cancer, 2019, 145, 370-379. | 2.3 | 9 |
| 210 | Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: Update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147-158. | 1.7 | 26 |
| 211 | Response to Wang et al.. Genetics in Medicine, 2019, 21, 2158. | 1.1 | 0 |
| 212 | A phenome-wide association study to discover pleiotropic effects of PCSK9, APOB, and LDLR. Npj Genomic Medicine, 2019, 4, 3. | 1.7 | 26 |
| 213 | De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089. | 1.4 | 19 |
| 214 | 10-year performance of four models of breast cancer risk: a validation study. Lancet Oncology, The, 2019, 20, 504-517. | 5.1 | 116 |
| 215 | Precision Medicine in Internal Medicine. Annals of Internal Medicine, 2019, 170, 635. | 2.0 | 12 |
| 216 | Cases in Precision Medicine: When Patients Present With Direct-to-Consumer Genetic Test Results. Annals of Internal Medicine, 2019, 170, 643. | 2.0 | 7 |

| # | Article | IF | Citations |
|---|---|---|---|
| 217 | Cases in Precision Medicine: Should You Participate in a Study Involving Genomic Sequencing of Your Patients?. Annals of Internal Medicine, 2019, 171, 568. | 2.0 | 0 |
| 218 | Cases in Precision Medicine: The Role of Pharmacogenetics in Precision Prescribing. Annals of Internal Medicine, 2019, 170, 796. | 2.0 | 8 |
| 219 | Cases in Precision Medicine: Genetic Assessment After a Sudden Cardiac Death in the Family. Annals of Internal Medicine, 2019, 170, 710. | 2.0 | 2 |
| 220 | Cases in Precision Medicine: The Role of Tumor and Germline Genetic Testing in Breast Cancer Management. Annals of Internal Medicine, 2019, 171, 925. | 2.0 | 10 |
| 221 | Sensorimotor Cortical Oscillations during Movement Preparation in 16p11.2 Deletion Carriers. Journal of Neuroscience, 2019, 39, 7321-7331. | 1.7 | 11 |
| 222 | T148. Structural and Functional Brain Differences in Children With the FTO Obesity Risk Allele. Biological Psychiatry, 2019, 85, S186. | 0.7 | 0 |
| 223 | Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69. | 3.6 | 86 |
| 224 | Increasing genomic literacy among adolescents. Genetics in Medicine, 2019, 21, 994-1000. | 1.1 | 14 |
| 225 | Response to Evans et al.. Genetics in Medicine, 2019, 21, 1880-1881. | 1.1 | 1 |
| 226 | The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11. | 2.0 | 60 |
| 227 | Recessive Rare Variants in Deoxyhypusine Synthase, an Enzyme Involved in the Synthesis of Hypusine, Are Associated with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 287-298. | 2.6 | 38 |
| 228 | Diagnosis and management of glycogen storage diseases type VI and IX: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 772-789. | 1.1 | 81 |
| 229 | 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. | 2.6 | 90 |
| 230 | Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. Journal of the National Cancer Institute, 2019, 111, 350-364. | 3.0 | 30 |
| 231 | Genetics and genomics of pulmonary arterial hypertension. European Respiratory Journal, 2019, 53, 1801899. | 3.1 | 306 |
| 232 | Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297. | 0.7 | 108 |
| 233 | Dallas MegaShelter Medical Operations Response to Hurricane Harvey. Disaster Medicine and Public Health Preparedness, 2019, 13, 90-93. | 0.7 | 6 |
| 234 | User engagement with web-based genomics education videos and implications for designing scalable patient education materials. AMIA ... Annual Symposium proceedings, 2019, 2019, 923-932. | 0.2 | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 235 | Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115. | 0.3 | 584 |
| 236 | FTO genotype impacts food intake and corticolimbic activation. American Journal of Clinical Nutrition, 2018, 107, 145-154. | 2.2 | 60 |
| 237 | Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456. | 4.2 | 210 |
| 238 | Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264. | 0.7 | 56 |
| 239 | Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887. | 1.6 | 104 |
| 240 | Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620. | 1.1 | 224 |
| 241 | SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493. | 3.8 | 265 |
| 242 | Breast cancer family history and allele-specific DNA methylation in the legacy girls study. Epigenetics, 2018, 13, 240-250. | 1.3 | 10 |
| 243 | MSH6 and PMS2 germ-line pathogenic variants implicated in Lynch syndrome are associated with breast cancer. Genetics in Medicine, 2018, 20, 1167-1174. | 1.1 | 116 |
| 244 | Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193. | 1.8 | 43 |
| 245 | Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935. | 0.7 | 57 |
| 246 | Examining the Psychosocial Impact of Genetic Testing for Cardiomyopathies. Journal of Genetic Counseling, 2018, 27, 927-934. | 0.9 | 16 |
| 247 | A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67. | 5.8 | 64 |
| 248 | Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, . | 0.7 | 0 |
| 249 | 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. | 2.6 | 59 |
| 250 | Advances in the Understanding of the Genetic Determinants of Congenital Heart Disease and Their Impact on Clinical Outcomes. Journal of the American Heart Association, 2018, 7, . | 1.6 | 82 |
| 251 | Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. Human Mutation, 2018, 39, 870-881. | 1.1 | 3 |
| 252 | Tandem mass spectrometry assay of Î²-glucocerebrosidase activity in dried blood spots eliminates false positives detected in fluorescence assay. Molecular Genetics and Metabolism, 2018, 123, 135-139. | 0.5 | 12 |

| # | Article | IF | Citations |
|---|---|---|---|
| 253 | Pilot study of population-based newborn screening for spinal muscular atrophy in New York state. Genetics in Medicine, 2018, 20, 608-613. | 1.1 | 98 |
| 254 | Genetics of pediatric cardiomyopathies. Progress in Pediatric Cardiology, 2018, 49, 18-19. | 0.2 | 3 |
| 255 | Missense variants in the chromatin remodeler CHD1 are associated with neurodevelopmental disability. Journal of Medical Genetics, 2018, 55, 561-566. | 1.5 | 49 |
| 256 | Navigating the researchâ€“clinical interface in genomic medicine: analysis from the CSER Consortium. Genetics in Medicine, 2018, 20, 545-553. | 1.1 | 34 |
| 257 | Impact of Receiving Secondary Results from Genomic Research: A 12â€Month Longitudinal Study. Journal of Genetic Counseling, 2018, 27, 709-722. | 0.9 | 26 |
| 258 | Whole-Genome and Whole-Exome Sequencing in Pediatric Oncology: An Assessment of Parent and Young Adult Patient Knowledge, Attitudes, and Expectations. JCO Precision Oncology, 2018, 2, 1-11. | 1.5 | 5 |
| 259 | Understanding Factors Associated with Uptake of<b><i> BRCA1/2</i></b> Genetic Testing<b><i></i></b> among Orthodox Jewish Women in the USA Using a Mixed-Methods Approach. Public Health Genomics, 2018, 21, 186-196. | 0.6 | 4 |
| 260 | De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822. | 1.5 | 79 |
| 261 | Age-specific breast cancer risk by body mass index and familial risk: prospective family study cohort (ProF-SC). Breast Cancer Research, 2018, 20, 132. | 2.2 | 51 |
| 262 | Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. Circulation, 2018, 138, e653-e711. | 1.6 | 387 |
| 263 | Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478. | 0.7 | 19 |
| 264 | Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. JNCI Cancer Spectrum, 2018, 2, pky023. | 1.4 | 33 |
| 265 | Biallelic variants in VARS in a family with two siblings with intellectual disability and microcephaly: case report and review of the literature. Journal of Physical Education and Sports Management, 2018, 4, a003301. | 0.5 | 7 |
| 266 | Loss-of-Function ABCC8 Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087. | 1.6 | 62 |
| 267 | Ethical Considerations Related to Return of Results from Genomic Medicine Projects: The eMERGE Network (Phase III) Experience. Journal of Personalized Medicine, 2018, 8, 2. | 1.1 | 44 |
| 268 | Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793. | 2.6 | 17 |
| 269 | KCNJ11 Mutation in One Family is Associated with Adult-Onset Rather than Neonatal-Onset Diabetes Mellitus. AACE Clinical Case Reports, 2018, 4, e411-e414. | 0.4 | 2 |
| 270 | Harmonizing Outcomes for Genomic Medicine: Comparison of eMERGE Outcomes to ClinGen Outcome/Intervention Pairs. Healthcare (Switzerland), 2018, 6, 83. | 1.0 | 18 |

| # | Article | IF | Citations |
|---|---|---|---|
| 271 | Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. Molecular Genetics & Genomic Medicine, 2018, 6, 898-909. | 0.6 | 15 |
| 272 | Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884. | 1.1 | 23 |
| 273 | Deep Phenotyping on Electronic Health Records Facilitates Genetic Diagnosis by Clinical Exomes. American Journal of Human Genetics, 2018, 103, 58-73. | 2.6 | 99 |
| 274 | Impacts of variants of uncertain significance on parental perceptions of children after prenatal chromosome microarray testing. Prenatal Diagnosis, 2018, 38, 740-747. | 1.1 | 25 |
| 275 | Laboratory considerations for prenatal genetic testing. Seminars in Perinatology, 2018, 42, 307-313. | 1.1 | 7 |
| 276 | Pulmonary hypertension in patients with 9q34.3 microdeletionâ€associated Kleefstra syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1773-1777. | 0.7 | 13 |
| 277 | Parental perceptions of prenatal whole exome sequencing (PPPWES) study. Prenatal Diagnosis, 2018, 38, 801-811. | 1.1 | 31 |
| 278 | Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114. | 2.6 | 34 |
| 279 | Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56. | 3.6 | 112 |
| 280 | Recurrent diffuse lung disease due to surfactant protein C deficiency. Respiratory Medicine Case Reports, 2018, 25, 91-95. | 0.2 | 11 |
| 281 | Features of Feingold syndrome 1 dominate in subjects with 2p deletions including MYCN. American Journal of Medical Genetics, Part A, 2018, 176, 1956-1963. | 0.7 | 6 |
| 282 | Clinical presentation and natural history of infantile-onset ascending spastic paralysis from three families with an ALS2 founder variant. Neurological Sciences, 2018, 39, 1917-1925. | 0.9 | 18 |
| 283 | The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319. | 1.1 | 82 |
| 284 | USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312. | 1.4 | 45 |
| 285 | Detecting potential pleiotropy across cardiovascular and neurological diseases using univariate, bivariate, and multivariate methods on 43,870 individuals from the eMERGE network. , 2018, , . | | 6 |
| 286 | Abnormal Vertical Eye Movements as a Clue for Diagnosis of Niemann-Pick Type C. Tremor and Other Hyperkinetic Movements, 2018, 8, 560. | 1.1 | 3 |
| 287 | A definition of gentle ventilation in congenital diaphragmatic hernia: a survey of neonatologists and pediatric surgeons. Journal of Perinatal Medicine, 2017, 45, 1031-1038. | 0.6 | 7 |
| 288 | Association of the missense variant p.<scp>Arg203Trp</scp> in <scp>PACS1</scp> as a cause of intellectual disability and seizures. Clinical Genetics, 2017, 92, 221-223. | 1.0 | 26 |

| # | Article | IF | Citations |
|---|---|---|---|
| 289 | MC4R-dependent suppression of appetite by bone-derived lipocalin 2. Nature, 2017, 543, 385-390. | 13.7 | 299 |
| 290 | De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86. | 1.5 | 46 |
| 291 | Single-Cell Analysis of SMN Reveals Its Broader Role in Neuromuscular Disease. Cell Reports, 2017, 18, 1484-1498. | 2.9 | 38 |
| 292 | Mutations in BMPR2 are not present in patients with pulmonary hypertension associated with congenital diaphragmatic hernia. Journal of Pediatric Surgery, 2017, 52, 1747-1750. | 0.8 | 3 |
| 293 | Selfâ€Reported Questionnaire Detects Family History of Cancer in a Pancreatic Cancer Screening Program. Journal of Genetic Counseling, 2017, 26, 806-813. | 0.9 | 3 |
| 294 | Differences in Presentation and Outcomes Between Children With Familial Dilated Cardiomyopathy and Children With Idiopathic Dilated Cardiomyopathy. Circulation: Heart Failure, 2017, 10, . | 1.6 | 30 |
| 295 | Research Participantsâ€™ Preferences for Hypothetical Secondary Results from Genomic Research. Journal of Genetic Counseling, 2017, 26, 841-851. | 0.9 | 39 |
| 296 | Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924. | 1.5 | 49 |
| 297 | Rapidly progressive mitral valve stenosis in patients with acromelic dysplasia. Cardiology in the Young, 2017, 27, 797-800. | 0.4 | 5 |
| 298 | Population-Based Study of Attitudes towardBRCAGenetic Testing among Orthodox Jewish Women. Breast Journal, 2017, 23, 333-337. | 0.4 | 13 |
| 299 | Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. JAMA - Journal of the American Medical Association, 2017, 317, 2402. | 3.8 | 1,898 |
| 300 | Pulmonary arterial hypertension: Specialistsâ€™ knowledge, practices, and attitudes of genetic counseling and genetic testing in the USA. Pulmonary Circulation, 2017, 7, 372-383. | 0.8 | 12 |
| 301 | Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829. | 1.5 | 27 |
| 302 | ACSS2 gene variant associated with cleft lip and palate in two independent Hispanic populations. Laryngoscope, 2017, 127, E336-E339. | 1.1 | 2 |
| 303 | 23andMe Paves the Way for Direct-to-Consumer Genetic Health Risk Tests of Limited Clinical Utility. Annals of Internal Medicine, 2017, 167, 125. | 2.0 | 13 |
| 304 | Developmental trajectories for young children with 16p11.2 copy number variation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 367-380. | 1.1 | 42 |
| 305 | Identification and characterization of a novel DGAT1 missense mutation associated with congenital diarrhea. Journal of Lipid Research, 2017, 58, 1230-1237. | 2.0 | 44 |
| 306 | Frequency of GBA Variants in Autopsyâ€proven Multiple System Atrophy. Movement Disorders Clinical Practice, 2017, 4, 574-581. | 0.8 | 47 |

| # | Article | IF | Citations |
|---|---|---|---|
| 307 | IMPACT OF DAMAGING DE NOVO VARIANTS ON CLINICAL OUTCOMES IN CONGENITAL HEART DISEASE. Journal of the American College of Cardiology, 2017, 69, 2553. | 1.2 | 0 |
| 308 | Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691. | 9.4 | 356 |
| 309 | Impact of Panel Gene Testing for Hereditary Breast and Ovarian Cancer on Patients. Journal of Genetic Counseling, 2017, 26, 1116-1129. | 0.9 | 90 |
| 310 | Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. Human Genetics, 2017, 136, 679-691. | 1.8 | 53 |
| 311 | Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127. | 2.6 | 62 |
| 312 | High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685. | 2.6 | 337 |
| 313 | Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601. | 9.4 | 624 |
| 314 | Pediatric Cardiomyopathies. Circulation Research, 2017, 121, 855-873. | 2.0 | 207 |
| 315 | The Impact of Heterozygous KCNK3 Mutations Associated With Pulmonary Arterial Hypertension on Channel Function and Pharmacological Recovery. Journal of the American Heart Association, 2017, 6, . | 1.6 | 34 |
| 316 | Analysis of myocardial fibrosis in children with hypertrophic cardiomyopathy: A report from the pediatric cardiomyopathy registry. Progress in Pediatric Cardiology, 2017, 46, 33. | 0.2 | 0 |
| 317 | Congenital diaphragmatic hernias: from genes to mechanisms to therapies. DMM Disease Models and Mechanisms, 2017, 10, 955-970. | 1.2 | 143 |
| 318 | Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015. | 2.8 | 63 |
| 319 | The impact of hereditary cancer gene panels on clinical care and lessons learned. Journal of Physical Education and Sports Management, 2017, 3, a002154. | 0.5 | 33 |
| 320 | Response to Dr. Sorscher. Journal of Genetic Counseling, 2017, 26, 1164-1164. | 0.9 | 0 |
| 321 | Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. Circulation, 2017, 136, 1037-1048. | 1.6 | 204 |
| 322 | Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134. | 1.1 | 18 |
| 323 | A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582. | 1.1 | 68 |
| 324 | Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2017, 19, 249-255. | 1.1 | 1,398 |

| # | Article | IF | Citations |
|---|---|---|---|
| 325 | The role of genetics in pulmonary arterial hypertension. Journal of Pathology, 2017, 241, 273-280. | 2.1 | 52 |
| 326 | De novo loss of function mutations in <scp>KIAA2022</scp> are associated with epilepsy and neurodevelopmental delay in females. Clinical Genetics, 2017, 91, 756-763. | 1.0 | 24 |
| 327 | Precision Medicine in Children and Young Adults with Hematologic Malignancies and Blood Disorders: The Columbia University Experience. Frontiers in Pediatrics, 2017, 5, 265. | 0.9 | 29 |
| 328 | PVDOMICS. Circulation Research, 2017, 121, 1136-1139. | 2.0 | 113 |
| 329 | iPSC-derived Î² cells model diabetes due to glucokinase deficiency. Journal of Clinical Investigation, 2017, 127, 1115-1115. | 3.9 | 2 |
| 330 | Hypertrophic cardiomyopathy: New approaches and a time to reappraise older approaches. Journal of Thoracic and Cardiovascular Surgery, 2016, 152, 983-988. | 0.4 | 7 |
| 331 | De novo pathogenic variants in CHAMP1 are associated with global developmental delay, intellectual disability, and dysmorphic facial features. Journal of Physical Education and Sports Management, 2016, 2, a000661. | 0.5 | 31 |
| 332 | Relationship between M100 Auditory Evoked Response and Auditory Radiation Microstructure in 16p11.2 Deletion and Duplication Carriers. American Journal of Neuroradiology, 2016, 37, 1178-1184. | 1.2 | 19 |
| 333 | Parentsâ€™ interest in genetic testing of their offspring in multiplex epilepsy families. Epilepsia, 2016, 57, 279-287. | 2.6 | 8 |
| 334 | Implementation of next generation sequencing into pediatric hematology-oncology practice: moving beyond actionable alterations. Genome Medicine, 2016, 8, 133. | 3.6 | 147 |
| 335 | Mutations in HIVEP2 are associated with developmental delay, intellectual disability, and dysmorphic features. Neurogenetics, 2016, 17, 159-164. | 0.7 | 31 |
| 336 | A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178. | 0.7 | 32 |
| 337 | Autism Spectrum Disorder, Developmental and Psychiatric Features in 16p11.2 Duplication. Journal of Autism and Developmental Disorders, 2016, 46, 2734-2748. | 1.7 | 47 |
| 338 | De novo mutations in CSNK2A1 are associated with neurodevelopmental abnormalities and dysmorphic features. Human Genetics, 2016, 135, 699-705. | 1.8 | 47 |
| 339 | Report of the National Heart, Lung, and Blood Institute Working Group. Circulation, 2016, 133, 1410-1418. | 1.6 | 33 |
| 340 | BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. Lancet Respiratory Medicine,the, 2016, 4, 129-137. | 5.2 | 307 |
| 341 | Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. Journal of Clinical Oncology, 2016, 34, 2172-2181. | 0.8 | 132 |
| 342 | Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066. | 2.6 | 137 |

| # | Article | IF | Citations |
|---|---|---|---|
| 343 | De novo PHIP-predicted deleterious variants are associated with developmental delay, intellectual disability, obesity, and dysmorphic features. Journal of Physical Education and Sports Management, 2016, 2, a001172. | 0.5 | 42 |
| 344 | De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409. | 1.8 | 40 |
| 345 | Characterization of a caveolinâ€1 mutation associated with both pulmonary arterial hypertension and congenital generalized lipodystrophy. Traffic, 2016, 17, 1297-1312. | 1.3 | 48 |
| 346 | Depression and genetic causal attribution of epilepsy in multiplex epilepsy families. Epilepsia, 2016, 57, 1643-1650. | 2.6 | 7 |
| 347 | Deep Genetic Connection Between Cancer and Developmental Disorders. Human Mutation, 2016, 37, 1042-1050. | 1.1 | 24 |
| 348 | Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734. | 2.6 | 75 |
| 349 | 16p11.2 deletion and duplication: Characterizing neurologic phenotypes in a large clinically ascertained cohort. American Journal of Medical Genetics, Part A, 2016, 170, 2943-2955. | 0.7 | 131 |
| 350 | Childhood acromegaly due to X-linked acrogigantism: long term follow-up. Pituitary, 2016, 19, 560-564. | 1.6 | 25 |
| 351 | Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375. | 5.8 | 93 |
| 352 | Rare variant phasing and haplotypic expression from RNA sequencing with phASER. Nature Communications, 2016, 7, 12817. | 5.8 | 105 |
| 353 | Mutations in TKT Are the Cause of a Syndrome Including Short Stature, Developmental Delay, and Congenital Heart Defects. American Journal of Human Genetics, 2016, 98, 1235-1242. | 2.6 | 31 |
| 354 | Cohort Profile: The Breast Cancer Prospective Family Study Cohort (ProF-SC). International Journal of Epidemiology, 2016, 45, 683-692. | 0.9 | 48 |
| 355 | Genetics and Hypertrophic Cardiomyopathy. Current Pediatrics Reports, 2016, 4, 35-44. | 1.7 | 0 |
| 356 | Reciprocal white matter alterations due to 16p11.2 chromosomal deletions versus duplications. Human Brain Mapping, 2016, 37, 2833-2848. | 1.9 | 37 |
| 357 | Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. Progress in Pediatric Cardiology, 2016, 40, 41-45. | 0.2 | 23 |
| 358 | Future research directions in pediatric cardiomyopathy. Progress in Pediatric Cardiology, 2016, 40, 35-39. | 0.2 | 1 |
| 359 | Auditory Evoked M100 Response Latency is Delayed in Children with 16p11.2 Deletion but not 16p11.2 Duplication. Cerebral Cortex, 2016, 26, 1957-1964. | 1.6 | 29 |
| 360 | Clinical application of whole-exome sequencing across clinical indications. Genetics in Medicine, 2016, 18, 696-704. | 1.1 | 780 |

| # | Article | IF | Citations |
|---|---|---|---|
| 361 | The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150. | 1.1 | 64 |
| 362 | A Quality Improvement Collaborative to Improve Pediatric Primary Care Genetic Services. Pediatrics, 2016, 137, e20143874. | 1.0 | 9 |
| 363 | 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. | 2.6 | 66 |
| 364 | Pathogenic and likely pathogenic variant prevalence among the first 10,000 patients referred for next-generation cancer panel testing. Genetics in Medicine, 2016, 18, 823-832. | 1.1 | 227 |
| 365 | Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20. | 6.0 | 195 |
| 366 | De novo missense variants in PPP2R5D are associated with intellectual disability, macrocephaly, hypotonia, and autism. Neurogenetics, 2016, 17, 43-49. | 0.7 | 61 |
| 367 | The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659. | 1.4 | 108 |
| 368 | Clinical phenotype of the recurrent 1q21.1 copy-number variant. Genetics in Medicine, 2016, 18, 341-349. | 1.1 | 134 |
| 369 | Genetics of Pulmonary Vascular Disease. , 2016, , 105-121. | | 0 |
| 370 | Evaluation of the <scp>CAV</scp>1 gene in clinically, sonographically and histologically proven morphea patients. Experimental Dermatology, 2015, 24, 718-720. | 1.4 | 4 |
| 371 | Abnormal auditory and language pathways in children with 16p11.2 deletion. NeuroImage: Clinical, 2015, 9, 50-57. | 1.4 | 19 |
| 372 | Delineation of New Disorders and Phenotypic Expansion of Known Disorders Through Whole Exome Sequencing. Current Genetic Medicine Reports, 2015, 3, 209-218. | 1.9 | 2 |
| 373 | An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61. | 2.2 | 26 |
| 374 | KCNK3 Mutations Are Found in Patients With Pulmonary Arterial Hypertension Associated With Fenfluramine or Dexfenfluramine Ingestion. Chest, 2015, 148, 944A. | 0.4 | 0 |
| 375 | Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127. | 1.1 | 185 |
| 376 | Genetic causal attribution of epilepsy and its implications for felt stigma. Epilepsia, 2015, 56, 1542-1550. | 2.6 | 18 |
| 377 | Overcoming challenges to meaningful informed consent for whole genome sequencing in pediatric cancer research. Pediatric Blood and Cancer, 2015, 62, 1374-1380. | 0.8 | 27 |
| 378 | Association of a Best-Practice Alert and Prenatal Administration With Tetanus Toxoid, Reduced Diphtheria Toxoid, and Acellular Pertussis Vaccination Rates. Obstetrics and Gynecology, 2015, 126, 333-337. | 1.2 | 23 |

| # | Article | IF | Citations |
|---|---|---|---|
| 379 | Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020. | 1.1 | 34 |
| 380 | Spectrum of Neuropathophysiology in Spinal Muscular Atrophy Type I. Journal of Neuropathology and Experimental Neurology, 2015, 74, 15-24. | 0.9 | 80 |
| 381 | Second primary breast cancer in BRCA1 and BRCA2 mutation carriers: 10-year cumulative incidence in the Breast Cancer Family Registry. Breast Cancer Research and Treatment, 2015, 151, 653-660. | 1.1 | 25 |
| 382 | New Insights into the Genetics of Fetal Megacystis: ACTG2 Mutations, Encoding γ-2 Smooth Muscle Actin in Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (Berdon Syndrome). Fetal Diagnosis and Therapy, 2015, 38, 296-306. | 0.6 | 48 |
| 383 | Psychosocial Adjustment in School-age Girls With a Family History of Breast Cancer. Pediatrics, 2015, 136, 927-937. | 1.0 | 13 |
| 384 | Pediatric Pulmonary Hypertension. Circulation, 2015, 132, 2037-2099. | 1.6 | 879 |
| 385 | Increased burden ofde novopredicted deleterious variants in complex congenital diaphragmatic hernia. Human Molecular Genetics, 2015, 24, 4764-4773. | 1.4 | 65 |
| 386 | De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266. | 6.0 | 646 |
| 387 | De novo POGZ mutations are associated with neurodevelopmental disorders and microcephaly. Journal of Physical Education and Sports Management, 2015, 1, a000455. | 0.5 | 51 |
| 388 | How to effectively utilize genetic testing in the care of children with cardiomyopathies. Progress in Pediatric Cardiology, 2015, 39, 3-11. | 0.2 | 4 |
| 389 | Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441. | 6.0 | 823 |
| 390 | Serum Endostatin Is a Genetically Determined Predictor of Survival in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2015, 191, 208-218. | 2.5 | 92 |
| 391 | Dual Optical Recordings for Action Potentials and Calcium Handling in Induced Pluripotent Stem Cell Models of Cardiac Arrhythmias Using Genetically Encoded Fluorescent Indicators. Stem Cells Translational Medicine, 2015, 4, 468-475. | 1.6 | 36 |
| 392 | Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352. | 2.6 | 230 |
| 393 | The association between congenital diaphragmatic hernia and undescended testes. Journal of Pediatric Surgery, 2015, 50, 744-745. | 0.8 | 8 |
| 394 | Brief Report: SETD2 Mutation in a Child with Autism, Intellectual Disabilities and Epilepsy. Journal of Autism and Developmental Disorders, 2015, 45, 3764-3770. | 1.7 | 64 |
| 395 | Glucocerebrosidase activity in Parkinsonâ€™s disease with and withoutGBAmutations. Brain, 2015, 138, 2648-2658. | 3.7 | 326 |
| 396 | When to Offer Genetic Testing for Pulmonary Arterial Hypertension. Canadian Journal of Cardiology, 2015, 31, 544-547. | 0.8 | 9 |

| # | Article | IF | Citations |
|---|---|---|---|
| 397 | Loss of function mutation in glutamic pyruvate transaminase 2 (GPT2) causes developmental encephalopathy. Journal of Inherited Metabolic Disease, 2015, 38, 941-948. | 1.7 | 28 |
| 398 | Association of Researcher Characteristics with Views on Return of Incidental Findings from Genomic Research. Journal of Genetic Counseling, 2015, 24, 833-841. | 0.9 | 17 |
| 399 | Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347. | 3.8 | 390 |
| 400 | The Role of Parental Cognitive, Behavioral, and Motor Profiles in Clinical Variability in Individuals With Chromosome 16p11.2 Deletions. JAMA Psychiatry, 2015, 72, 119. | 6.0 | 112 |
| 401 | Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513. | 3.8 | 258 |
| 402 | Whole-Exome Sequencing Reveals CLCNKB Mutations in a Case of Sudden Unexpected Infant Death. Pediatric and Developmental Pathology, 2015, 18, 324-326. | 0.5 | 8 |
| 403 | Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547. | 1.5 | 68 |
| 404 | Nonspecific phenotype of Noonan syndrome diagnosed by whole exome sequencing. Clinical Case Reports (discontinued), 2015, 3, 237-239. | 0.2 | 4 |
| 405 | Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314. | 0.7 | 54 |
| 406 | CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record. Journal of the American Medical Informatics Association: JAMIA, 2015, 22, 1231-1242. | 2.2 | 73 |
| 407 | Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635. | 1.5 | 48 |
| 408 | Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464. | 2.6 | 134 |
| 409 | Researchersâ€™ views on informed consent for return of secondary results in genomic research. Genetics in Medicine, 2015, 17, 644-650. | 1.1 | 16 |
| 410 | Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice. Pediatrics, 2015, 136, e974-e982. | 1.0 | 28 |
| 411 | Adrenergic receptor genotype influences heart failure severity and Î²-blocker response in children with dilated cardiomyopathy. Pediatric Research, 2015, 77, 363-369. | 1.1 | 8 |
| 412 | Development of a tiered and binned genetic counseling model for informed consent in the era of multiplex testing for cancer susceptibility. Genetics in Medicine, 2015, 17, 485-492. | 1.1 | 79 |
| 413 | Intermediate filament protein accumulation in motor neurons derived from giant axonal neuropathy iPSCs rescued by restoration of gigaxonin. Human Molecular Genetics, 2015, 24, 1420-1431. | 1.4 | 31 |
| 414 | Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316. | 1.1 | 22 |

| # | Article | IF | Citations |
|---|---|---|---|
| 415 | The Cognitive and Behavioral Phenotype of the 16p11.2 Deletion in a Clinically Ascertained Population. Biological Psychiatry, 2015, 77, 785-793. | 0.7 | 198 |
| 416 | Identifying monogenic diabetes in a pediatric cohort with presumed type 1 diabetes. Pediatric Diabetes, 2015, 16, 227-233. | 1.2 | 24 |
| 417 | Abstract 17301: Functional Characterization of KCNK3 Mutants Associated With Pulmonary Arterial Hypertension Under Physiologically Relevant Heterozygous Conditions. Circulation, 2015, 132, . | 1.6 | 0 |
| 418 | Eating in the absence of hunger but not loss of control behaviors are associated with 16p11.2 deletions. Obesity, 2014, 22, 2625-2631. | 1.5 | 12 |
| 419 | Dilated cardiomyopathy due to a phospholamban duplication. Cardiology in the Young, 2014, 24, 953-954. | 0.4 | 6 |
| 420 | Informed consent for return of incidental findings in genomic research. Genetics in Medicine, 2014, 16, 367-373. | 1.1 | 58 |
| 421 | The Expanding MEGDEL Phenotype: Optic Nerve Atrophy, Microcephaly, and Myoclonic Epilepsy in a Child with SERAC1 Mutations. JIMD Reports, 2014, 16, 75-79. | 0.7 | 17 |
| 422 | CANOES: detecting rare copy number variants from whole exome sequencing data. Nucleic Acids Research, 2014, 42, e97-e97. | 6.5 | 123 |
| 423 | Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2014, 16, e1-e29. | 1.1 | 318 |
| 424 | Weight loss after bariatric surgery in morbidly obese adolescents with MC4R mutations. Obesity, 2014, 22, 225-231. | 1.5 | 45 |
| 425 | Correlation of DNA methylation levels in blood and saliva DNA in young girls of the LEGACY Girls study. Epigenetics, 2014, 9, 929-933. | 1.3 | 32 |
| 426 | Whole-Exome sequencing identifies novel LEPR mutations in individuals with severe early onset obesity. Obesity, 2014, 22, 576-584. | 1.5 | 45 |
| 427 | Genetic causes of congenital diaphragmatic hernia. Seminars in Fetal and Neonatal Medicine, 2014, 19, 324-330. | 1.1 | 77 |
| 428 | Psychiatristsâ€™ Views of the Genetic Bases of Mental Disorders and Behavioral Traits and Their Use of Genetic Tests. Journal of Nervous and Mental Disease, 2014, 202, 530-538. | 0.5 | 18 |
| 429 | Genetics of pulmonary hypertension. Current Opinion in Cardiology, 2014, 29, 520-527. | 0.8 | 38 |
| 430 | Genetic Testing of Children for Diseases That Have Onset in Adulthood: The Limits of Family Interests. Pediatrics, 2014, 134, S104-S110. | 1.0 | 17 |
| 431 | Should Life Insurers Have Access to Genetic Test Results?. JAMA - Journal of the American Medical Association, 2014, 312, 1855. | 3.8 | 11 |
| 432 | Comparison of Parkinson Risk in Ashkenazi Jewish Patients With Gaucher Disease andGBAHeterozygotes. JAMA Neurology, 2014, 71, 752. | 4.5 | 172 |

| # | Article | IF | Citations |
|---|---|---|---|
| 433 | Processes and factors involved in decisions regarding return of incidental genomic findings in research. Genetics in Medicine, 2014, 16, 311-317. | 1.1 | 28 |
| 434 | Clinical features of West Nile virus epidemic in Dallas, Texas, 2012. Diagnostic Microbiology and Infectious Disease, 2014, 78, 132-136. | 0.8 | 23 |
| 435 | The effect of cardiac genetic testing on psychological well-being and illness perceptions. Heart and Lung: Journal of Acute and Critical Care, 2014, 43, 127-132. | 0.8 | 14 |
| 436 | Knowledge of and Interest in Genetic Results Among Parkinson Disease Patients and Caregivers. Journal of Genetic Counseling, 2014, 23, 114-120. | 0.9 | 29 |
| 437 | The genetic basis of pulmonary arterial hypertension. Human Genetics, 2014, 133, 471-479. | 1.8 | 75 |
| 438 | The contribution of de novo and rare inherited copy number changes to congenital heart disease in an unselected sample of children with conotruncal defects or hypoplastic left heart disease. Human Genetics, 2014, 133, 11-27. | 1.8 | 112 |
| 439 | Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices In Between. American Journal of Human Genetics, 2014, 94, 818-826. | 2.6 | 342 |
| 440 | Whole exome sequencing identifies de novo mutations inGATA6associated with congenital diaphragmatic hernia. Journal of Medical Genetics, 2014, 51, 197-202. | 1.5 | 55 |
| 441 | Î²-Cell Dysfunction Due to Increased ER Stress in a Stem Cell Model of Wolfram Syndrome. Diabetes, 2014, 63, 923-933. | 0.3 | 144 |
| 442 | Complex Genetics and the Etiology of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a013953-a013953. | 2.9 | 118 |
| 443 | Aberrant White Matter Microstructure in Children with 16p11.2 Deletions. Journal of Neuroscience, 2014, 34, 6214-6223. | 1.7 | 70 |
| 444 | The motor neuron response to SMN1 deficiency in spinal muscular atrophy. Muscle and Nerve, 2014, 49, 636-644. | 1.0 | 34 |
| 445 | Catecholaminergic polymorphic ventricular tachycardia in a child with Brugada pattern on ECG: One patient with two diseases?. Heart Rhythm, 2014, 11, 2101-2104. | 0.3 | 1 |
| 446 | Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. Circulation Research, 2014, 115, 884-896. | 2.0 | 229 |
| 447 | Reply. Muscle and Nerve, 2014, 50, 458-459. | 1.0 | 0 |
| 448 | The usefulness of whole-exome sequencing in routine clinical practice. Genetics in Medicine, 2014, 16, 922-931. | 1.1 | 196 |
| 449 | Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology, 2014, 83, 810-817. | 1.5 | 367 |
| 450 | Opposing Brain Differences in 16p11.2 Deletion and Duplication Carriers. Journal of Neuroscience, 2014, 34, 11199-11211. | 1.7 | 149 |

| # | Article | IF | Citations |
|---|---|---|---|
| 451 | Genetic testing preferences in families containing multiple individuals with epilepsy. Epilepsia, 2014, 55, 1705-1713. | 2.6 | 16 |
| 452 | BRCA1 and BRCA2 germline mutations are frequently demonstrated in both highâ€risk pancreatic cancer screening and pancreatic cancer cohorts. Cancer, 2014, 120, 1960-1967. | 2.0 | 59 |
| 453 | Hypomorphism for RPGRIP1L, a Ciliary Gene Vicinal to the FTO Locus, Causes Increased Adiposity in Mice. Cell Metabolism, 2014, 19, 767-779. | 7.2 | 145 |
| 454 | Human Subjects Protection: An Event Monitoring Committee for Research Studies of Girls From Breast Cancer Families. Journal of Adolescent Health, 2014, 55, 352-357. | 1.2 | 5 |
| 455 | Use of Genetic Tests among Neurologists and Psychiatrists: Knowledge, Attitudes, Behaviors, and Needs for Training. Journal of Genetic Counseling, 2014, 23, 156-163. | 0.9 | 87 |
| 456 | EIF2AK4 Mutations in Pulmonary Capillary Hemangiomatosis. Chest, 2014, 145, 231-236. | 0.4 | 176 |
| 457 | Views of preimplantation genetic diagnosis among psychiatrists and neurologists. Journal of reproductive medicine, The, 2014, 59, 385-92. | 0.2 | 10 |
| 458 | A Novel Channelopathy in Pulmonary Arterial Hypertension. New England Journal of Medicine, 2013, 369, 351-361. | 13.9 | 412 |
| 459 | Genetic variants associated with breast cancer risk for Ashkenazi Jewish women with strong family histories but no identifiable BRCA1/2 mutation. Human Genetics, 2013, 132, 523-536. | 1.8 | 26 |
| 460 | Variants in GATA4 are a rare cause of familial and sporadic congenital diaphragmatic hernia. Human Genetics, 2013, 132, 285-292. | 1.8 | 81 |
| 461 | CDKL5 and ARX Mutations in Males With Early-Onset Epilepsy. Pediatric Neurology, 2013, 48, 367-377. | 1.0 | 53 |
| 462 | Return of Secondary Genomic Findings vs Patient Autonomy. JAMA - Journal of the American Medical Association, 2013, 310, 369. | 3.8 | 68 |
| 463 | Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2013, 62, D13-D21. | 1.2 | 367 |
| 464 | Developmental outcomes of children with congenital diaphragmatic hernia: A multicenter prospective study. Journal of Pediatric Surgery, 2013, 48, 1995-2004. | 0.8 | 68 |
| 465 | Attitudes and Practices Among Internists Concerning Genetic Testing. Journal of Genetic Counseling, 2013, 22, 90-100. | 0.9 | 147 |
| 466 | A Recurrent PDGFRB Mutation Causes Familial Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 996-1000. | 2.6 | 135 |
| 467 | Views of internists towards uses of PGD. Reproductive BioMedicine Online, 2013, 26, 142-147. | 1.1 | 21 |
| 468 | Outcomes of Congenital Diaphragmatic Hernia in the Modern Era ofÂManagement. Journal of Pediatrics, 2013, 163, 114-119.e1. | 0.9 | 185 |

| # | Article | IF | Citations |
|---|---|---|---|
| 469 | Genome-wide association analysis identifies a susceptibility locus for pulmonary arterial hypertension. Nature Genetics, 2013, 45, 518-521. | 9.4 | 93 |
| 470 | De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223. | 13.7 | 798 |
| 471 | Defining a comprehensive verotype using electronic health records for personalized medicine. Journal of the American Medical Informatics Association: JAMIA, 2013, 20, e232-e238. | 2.2 | 59 |
| 472 | Researchersâ€™ views on return of incidental genomic research results: qualitative and quantitative findings. Genetics in Medicine, 2013, 15, 888-895. | 1.1 | 103 |
| 473 | Novel Association of Early Onset Hepatocellular Carcinoma with Transaldolase Deficiency. JIMD Reports, 2013, 12, 121-127. | 0.7 | 33 |
| 474 | Clinical Application of Whole-Exome Sequencing. JAMA Neurology, 2013, 70, 788. | 4.5 | 15 |
| 475 | Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212. | 1.5 | 244 |
| 476 | Risk of Pancreatic Cancer in Breast Cancer Families from the Breast Cancer Family Registry. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 803-811. | 1.1 | 83 |
| 477 | A human de novo mutation inMYH10phenocopies the loss of function mutation in mice. Rare Diseases (Austin, Tex ), 2013, 1, e26144. | 1.8 | 34 |
| 478 | Induced pluripotent stem cells used to reveal drug actions in a long QT syndrome family with complex genetics. Journal of General Physiology, 2013, 141, 61-72. | 0.9 | 189 |
| 479 | Effectiveness of a School District Closure for Pandemic Influenza A (H1N1) on Acute Respiratory Illnesses in the Community: A Natural Experiment. Clinical Infectious Diseases, 2013, 56, 509-516. | 2.9 | 41 |
| 480 | High Prevalence of BRCA1 and BRCA2 Germline Mutations with Loss of Heterozygosity in a Series of Resected Pancreatic Adenocarcinoma and Other Neoplastic Lesions. Clinical Cancer Research, 2013, 19, 3396-3403. | 3.2 | 65 |
| 481 | RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641. | 1.1 | 221 |
| 482 | Incidental Findings in the Era of Whole Genome Sequencing?. Hastings Center Report, 2013, 43, 16-19. | 0.7 | 45 |
| 483 | Mutation inSNAP25as a novel genetic cause of epilepsy and intellectual disability. Rare Diseases (Austin, Tex ), 2013, 1, e26314. | 1.8 | 55 |
| 484 | The Congenital Heart Disease Genetic Network Study. Circulation Research, 2013, 112, 698-706. | 2.0 | 142 |
| 485 | Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. Circulation: Cardiovascular Genetics, 2013, 6, 444-451. | 5.1 | 89 |
| 486 | Genetic loss of SH2B3 in acute lymphoblastic leukemia. Blood, 2013, 122, 2425-2432. | 0.6 | 101 |

| # | Article | IF | Citations |
|---|---|---|---|
| 487 | Are KCNK3 Mutations Found in Vasoreactive Patients With Idiopathic Pulmonary Arterial Hypertension?. Chest, 2013, 144, 878A. | 0.4 | 0 |
| 488 | iPSC-derived Î² cells model diabetes due to glucokinase deficiency. Journal of Clinical Investigation, 2013, 123, 3146-3153. | 3.9 | 84 |
| 489 | SMA-MAP: A Plasma Protein Panel for Spinal Muscular Atrophy. PLoS ONE, 2013, 8, e60113. | 1.1 | 40 |
| 490 | A Nonsynonymous Polymorphism in IRS1 Modifies Risk of Developing Breast and Ovarian Cancers in BRCA1 and Ovarian Cancer in BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1362-1370. | 1.1 | 23 |
| 491 | A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668. | 1.5 | 251 |
| 492 | Whole Exome Sequencing to Identify a Novel Gene (Caveolin-1) Associated With Human Pulmonary Arterial Hypertension. Circulation: Cardiovascular Genetics, 2012, 5, 336-343. | 5.1 | 333 |
| 493 | A public resource facilitating clinical use of genomes. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 11920-11927. | 3.3 | 194 |
| 494 | Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657. | 1.1 | 47 |
| 495 | Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997. | 2.6 | 201 |
| 496 | De novo copy number variants are associated with congenital diaphragmatic hernia. Journal of Medical Genetics, 2012, 49, 650-659. | 1.5 | 68 |
| 497 | Genetic Testing for Dilated Cardiomyopathy in Clinical Practice. Journal of Cardiac Failure, 2012, 18, 296-303. | 0.7 | 145 |
| 498 | Association of candidate genes with nonsyndromic clefts in Honduran and Colombian populations. Laryngoscope, 2012, 122, 2082-2087. | 1.1 | 35 |
| 499 | Mutations in ZIC3 and ACVR2B are a common cause of heterotaxy and associated cardiovascular anomalies. Cardiology in the Young, 2012, 22, 194-201. | 0.4 | 37 |
| 500 | An overview of mongenic and syndromic obesities in humans. Pediatric Blood and Cancer, 2012, 58, 122-128. | 0.8 | 49 |
| 501 | Fat discrimination: A phenotype with potential implications for studying fat intake behaviors and obesity. Physiology and Behavior, 2012, 105, 470-475. | 1.0 | 30 |
| 502 | Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702. | 1.1 | 34 |
| 503 | Combined OXPHOS complex I and IV defect, due to mutated complex I assembly factor C20ORF7. Journal of Inherited Metabolic Disease, 2012, 35, 125-131. | 1.7 | 52 |
| 504 | Familial and Acquired SH2B3 mutations in ALL. Blood, 2012, 120, 1326-1326. | 0.6 | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 505 | Novel frameshift mutation in Troponin C (TNNC1) associated with hypertrophic cardiomyopathy and sudden death. Cardiology in the Young, 2011, 21, 345-348. | 0.4 | 27 |
| 506 | A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846. | 9.4 | 1,141 |
| 507 | Clinical and Molecular Genetic Features of Hereditary Pulmonary Arterial Hypertension. , 2011, 1, 1721-1728. | | 5 |
| 508 | Intragenic deletion as a novel type of mutation in Wolman disease. Molecular Genetics and Metabolism, 2011, 104, 703-705. | 0.5 | 5 |
| 509 | Developments in Molecular Genetic Diagnostics: An Update for the Pediatric Epilepsy Specialist. Pediatric Neurology, 2011, 44, 317-327. | 1.0 | 22 |
| 510 | Association of Allelic Variation in Genes Mediating Aspects of Energy Homeostasis with Weight Gain during Administration of Antipsychotic Drugs (CATIE Study). Frontiers in Genetics, 2011, 2, 56. | 1.1 | 19 |
| 511 | Novel SLC39A4 Mutation in Acrodermatitis Enteropathica. Pediatric Dermatology, 2011, 28, 697-700. | 0.5 | 10 |
| 512 | Novel gene discovery in pediatric cardiomyopathy. Progress in Pediatric Cardiology, 2011, 31, 89-91. | 0.2 | 4 |
| 513 | Genetic issues in pediatric cardiomyopathy: Future research directions. Progress in Pediatric Cardiology, 2011, 32, 3-4. | 0.2 | 3 |
| 514 | Comparison of Endoscopic and Clinical Characteristics of Patients with Familial and Sporadic Barrettâ€™s Esophagus. Digestive Diseases and Sciences, 2011, 56, 1702-1706. | 1.1 | 13 |
| 515 | A complete deficiency of Hyaluronoglucosaminidase 1 (HYAL1) presenting as familial juvenile idiopathic arthritis. Journal of Inherited Metabolic Disease, 2011, 34, 1013-1022. | 1.7 | 68 |
| 516 | Glut1 deficiency syndrome and erythrocyte glucose uptake assay. Annals of Neurology, 2011, 70, 996-1005. | 2.8 | 83 |
| 517 | Global DNA methylation levels in girls with and without a family history of breast cancer. Epigenetics, 2011, 6, 29-33. | 1.3 | 31 |
| 518 | Two Cases of Pulmonary Hypertension Associated with Type III Glycogen Storage Disease. JIMD Reports, 2011, 1, 79-82. | 0.7 | 11 |
| 519 | Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function. European Journal of Human Genetics, 2011, 19, 1238-1245. | 1.4 | 74 |
| 520 | Determinants of extracellular matrix remodelling are differentially expressed in paediatric and adult dilated cardiomyopathy. European Journal of Heart Failure, 2011, 13, 271-277. | 2.9 | 10 |
| 521 | Validation of the Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP) Tj ETQq1 1 0.784314 rgBT /Overl | 0.3 | 106 |
| 522 | Renin-Angiotensin-Aldosterone Genotype Influences Ventricular Remodeling in Infants With Single Ventricle. Circulation, 2011, 123, 2353-2362. | 1.6 | 63 |

| # | Article | IF | Citations |
|---|---|---|---|
| 523 | Validation of the Expanded Hammersmith Functional Motor Scale in Spinal Muscular Atrophy Type II and III. Journal of Child Neurology, 2011, 26, 1499-1507. | 0.7 | 143 |
| 524 | Cut-like Homeobox 1 (CUX1) Regulates Expression of the Fat Mass and Obesity-associated and Retinitis Pigmentosa GTPase Regulator-interacting Protein-1-like (RPGRIP1L) Genes and Coordinates Leptin Receptor Signaling. Journal of Biological Chemistry, 2011, 286, 2155-2170. | 1.6 | 129 |
| 525 | Observational Study of Spinal Muscular Atrophy Type 2 and 3. Archives of Neurology, 2011, 68, 779. | 4.9 | 142 |
| 526 | Glut1 deficiency: Inheritance pattern determined by haploinsufficiency. Annals of Neurology, 2010, 68, 955-958. | 2.8 | 78 |
| 527 | Association of Plastin 3 Expression With Disease Severity in Spinal Muscular Atrophy Only in Postpubertal Females. Archives of Neurology, 2010, 67, 1252-6. | 4.9 | 53 |
| 528 | Enalapril in Infants With Single Ventricle. Circulation, 2010, 122, 333-340. | 1.6 | 267 |
| 529 | Pancreatic Cancer Screening in a Prospective Cohort of High-Risk Patients: A Comprehensive Strategy of Imaging and Genetics. Clinical Cancer Research, 2010, 16, 5028-5037. | 3.2 | 189 |
| 530 | Genetic evaluation and counseling for epilepsy. Nature Reviews Neurology, 2010, 6, 445-453. | 4.9 | 75 |
| 531 | Short Communication: The Cardiac Myosin Binding Protein C Arg502Trp Mutation. Circulation Research, 2010, 106, 1549-1552. | 2.0 | 67 |
| 532 | Novel loci interacting epistatically with bone morphogenetic protein receptor 2 cause familial pulmonary arterial hypertension. Journal of Heart and Lung Transplantation, 2010, 29, 174-180. | 0.3 | 17 |
| 533 | Uncovering microdeletions in patients with severe Glut-1 deficiency syndrome using SNP oligonucleotide microarray analysis. Molecular Genetics and Metabolism, 2010, 100, 129-135. | 0.5 | 24 |
| 534 | Glycogen Storage Disease Type III diagnosis and management guidelines. Genetics in Medicine, 2010, 12, 446-463. | 1.1 | 236 |
| 535 | Blood leukocyte microarrays to diagnose systemic onset juvenile idiopathic arthritis and follow the response to IL-1 blockade. Journal of Experimental Medicine, 2009, 206, 2299-2299. | 4.2 | 0 |
| 536 | Analysis of 30 Genes (355 SNPS) Related to Energy Homeostasis for Association with Adiposity in European-American and Yupâ€™ik Eskimo Populations. Human Heredity, 2009, 67, 193-205. | 0.4 | 16 |
| 537 | Absence epilepsy in apathetic, a spontaneous mutant mouse lacking the h channel subunit, HCN2. Neurobiology of Disease, 2009, 33, 499-508. | 2.1 | 67 |
| 538 | BRCA1 and BRCA2 mutation carriers in the Breast Cancer Family Registry: an open resource for collaborative research. Breast Cancer Research and Treatment, 2009, 116, 379-386. | 1.1 | 52 |
| 539 | Founder Fukutin mutation causes Walkerâ€“Warburg syndrome in four Ashkenazi Jewish families. Prenatal Diagnosis, 2009, 29, 560-569. | 1.1 | 24 |
| 540 | Mild fasting hyperglycemia in children: high rate of glucokinase mutations and some risk of developing type 1 diabetes mellitus. Pediatric Diabetes, 2009, 10, 382-388. | 1.2 | 28 |

| # | Article | IF | Citations |
|---|---|---|---|
| 541 | Oligonucleotide array CGH studies in myeloproliferative neoplasms: Comparison with JAK2V617F mutational status and conventional chromosome analysis. Leukemia Research, 2009, 33, 662-664. | 0.4 | 18 |
| 542 | Preimplantation genetic diagnosis on in vitro fertilization clinic websites: presentations of risks, benefits and other information. Fertility and Sterility, 2009, 92, 1276-1283. | 0.5 | 17 |
| 543 | Polymorphism in the Angiotensin II Type 1 Receptor (AGTR1) is Associated With Age at Diagnosis in Pulmonary Arterial Hypertension. Journal of Heart and Lung Transplantation, 2009, 28, 373-379. | 0.3 | 34 |
| 544 | Recipient Genotype Is a Predictor of Allograft Cytokine Expression and Outcomes After Pediatric Cardiac Transplantation. Journal of the American College of Cardiology, 2009, 53, 1909-1917. | 1.2 | 16 |
| 545 | Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2009, 54, S32-S42. | 1.2 | 342 |
| 546 | Prenatal diagnosis of congenital lipoid adrenal hyperplasia (CLAH) by estriol amniotic fluid analysis and molecular genetic testing. Prenatal Diagnosis, 2008, 28, 11-14. | 1.1 | 13 |
| 547 | Induced Pluripotent Stem Cells Generated from Patients with ALS Can Be Differentiated into Motor Neurons. Science, 2008, 321, 1218-1221. | 6.0 | 1,826 |
| 548 | Considerations Regarding the Genetics of Obesity. Obesity, 2008, 16, S33-9. | 1.5 | 47 |
| 549 | Positional Cloning of â€œLisch-likeâ€, a Candidate Modifier of Susceptibility to Type 2 Diabetes in Mice. PLoS Genetics, 2008, 4, e1000137. | 1.5 | 58 |
| 550 | A KCNE2 mutation in a patient with cardiac arrhythmia induced by auditory stimuli and serum electrolyte imbalance. Cardiovascular Research, 2008, 77, 98-106. | 1.8 | 35 |
| 551 | Utility of Oligonucleotide Array Comparative Genomic Hybridization to Identify Cryptic Copy Number Alterations in Myelodysplastic Syndromes. Blood, 2008, 112, 5076-5076. | 0.6 | 1 |
| 552 | A Novel LQT-3 Mutation Disrupts an Inactivation Gate Complex with Distinct Rate-Dependent Phenotypic Consequences. Channels, 2007, 1, 273-280. | 1.5 | 34 |
| 553 | Variants of the CFC1 gene in patients with laterality defects associated with congenital cardiac disease. Cardiology in the Young, 2007, 17, 268-274. | 0.4 | 21 |
| 554 | Implementation of Genetics to Personalize Medicine. Gender Medicine, 2007, 4, 248-265. | 1.4 | 21 |
| 555 | Strong Association of De Novo Copy Number Mutations with Autism. Science, 2007, 316, 445-449. | 6.0 | 2,497 |
| 556 | Role of a Founder c.201_202delCT Mutation and New Phenotypic Features of Congenital Lipoid Adrenal Hyperplasia in Palestinians. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 4000-4008. | 1.8 | 30 |
| 557 | A Novel and Lethal De Novo LQT-3 Mutation in a Newborn with Distinct Molecular Pharmacology and Therapeutic Response. PLoS ONE, 2007, 2, e1258. | 1.1 | 50 |
| 558 | Predictive genetic testing for cardiomyopathies. Progress in Pediatric Cardiology, 2007, 23, 33-38. | 0.2 | 7 |

| # | Article | IF | Citations |
|---|---|---|---|
| 559 | Analysis of GNAS mutations in 60 growth hormone secreting pituitary tumors: correlation with clinical and pathological characteristics and surgical outcome based on highly sensitive GH and IGF-I criteria for remission. Pituitary, 2007, 10, 275-282. | 1.6 | 316 |
| 560 | Decision-Making About Reproductive Choices Among Individuals At-Risk for Huntington's Disease. Journal of Genetic Counseling, 2007, 16, 347-362. | 0.9 | 63 |
| 561 | RAAS gene polymorphisms influence progression of pediatric hypertrophic cardiomyopathy. Human Genetics, 2007, 122, 515-523. | 1.8 | 36 |
| 562 | Oligonucleotide Array CGH Studies in Myeloproliferative Neoplasms and Comparison with Conventional Cytogenetic Analysis. Blood, 2007, 110, 1550-1550. | 0.6 | 16 |
| 563 | The Links Between Obesity, Leptin, and Prostate Cancer. Cancer Journal (Sudbury, Mass ), 2006, 12, 178-181. | 1.0 | 11 |
| 564 | Glucokinase mutations in young children with hyperglycemia. Diabetes/Metabolism Research and Reviews, 2006, 22, 348-355. | 1.7 | 14 |
| 565 | Application of ROMA (representational oligonucleotide microarray analysis) to patients with cytogenetic rearrangements. Genetics in Medicine, 2005, 7, 111-118. | 1.1 | 32 |
| 566 | Analysis of Significance Patterns Identifies Ubiquitous and Disease-Specific Gene-Expression Signatures in Patient Peripheral Blood Leukocytes. Annals of the New York Academy of Sciences, 2005, 1062, 146-154. | 1.8 | 43 |
| 567 | Alpha-Thalassemia Major Presenting in a Term Neonate without Hydrops. Pediatric and Developmental Pathology, 2005, 8, 706-709. | 0.5 | 9 |
| 568 | A unique case of der(11)t(11;22),-22 arising from 3:1 segregation of a maternal t(11;22) in a family with co-segregation of the translocation and breast cancer. Prenatal Diagnosis, 2005, 25, 683-686. | 1.1 | 15 |
| 569 | Congenital Disorder of Glycosylation Id Presenting with Hyperinsulinemic Hypoglycemia and Islet Cell Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 4371-4375. | 1.8 | 88 |
| 570 | Molecular physiology of syndromic obesities in humans. Trends in Endocrinology and Metabolism, 2005, 16, 267-272. | 3.1 | 39 |
| 571 | Molecular and functional analysis ofSLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. Human Mutation, 2004, 24, 312-320. | 1.1 | 63 |
| 572 | Fatal infantile neuromuscular presentation of glycogen storage disease type IV. Neuromuscular Disorders, 2004, 14, 253-260. | 0.3 | 185 |
| 573 | Foamy podocytes. American Journal of Kidney Diseases, 2003, 41, 891-896. | 2.1 | 4 |
| 574 | Solid phase capturable dideoxynucleotides for multiplex genotyping using mass spectrometry. Nucleic Acids Research, 2002, 30, 85e-85. | 6.5 | 41 |
| 575 | The mouse mahoganoid coat color mutation disrupts a novel C3HC4 RING domain protein. Journal of Clinical Investigation, 2002, 110, 1449-1459. | 3.9 | 34 |
| 576 | Comparative Maps of Human 19p13.3 and Mouse Chromosome 10 Allow Identification of Sequences at Evolutionary Breakpoints. Genome Research, 2000, 10, 1369-1380. | 2.4 | 36 |

| # | Article | IF | Citations |
|---|---|---|---|
| 577 | The Molecular Genetics of Rodent Single Gene Obesities. Journal of Biological Chemistry, 1997, 272, 31937-31940. | 1.6 | 245 |
| 578 | Molecular Genetic Analysis of a Human Neuropeptide Y Receptor. Journal of Biological Chemistry, 1997, 272, 3622-3627. | 1.6 | 46 |
| 579 | Polymerase Chain Reactionâ€Restriction Fragment Length Polymorphisms (PCRâ€RFLP) and Electrophoretic Assays for the Mouse Obese (Lep<sup>ob</sup>) Mutation. Obesity, 1997, 5, 183-185. | 4.0 | 16 |
| 580 | Suggestive Linkages Between Markers on Human 1p32â€p22 and Body Fat and Insulin Levels in the QuÃ©bec Family Study. Obesity, 1997, 5, 115-121. | 4.0 | 56 |
| 581 | Amino Acid Variants in the Human Leptin Receptor: Lack of Association to Juvenile Onset Obesity. Biochemical and Biophysical Research Communications, 1997, 233, 248-252. | 1.0 | 74 |
| 582 | Genetic Modifiers ofLeprfaAssociated with Variability in Insulin Production and Susceptibility to NIDDM. Genomics, 1997, 41, 332-344. | 1.3 | 57 |
| 583 | Fine Structure of the Murine Leptin Receptor Gene: Splice Site Suppression Is Required to Form Two Alternatively Spliced Transcripts. Genomics, 1997, 45, 264-270. | 1.3 | 130 |
| 584 | Molecular Mapping of the Tubby (tub) Mutation on Mouse Chromosome 7. Genomics, 1996, 32, 210-217. | 1.3 | 15 |
| 585 | A homozygous splice variant in ATP5PO , disrupts mitochondrial complex V function and causes Leigh syndrome in two unrelated families. Journal of Inherited Metabolic Disease, 0, , . | 1.7 | 1 |
| 586 | Women's thoughts on receiving and sharing genetic information: Considerations for genetic counseling. Journal of Genetic Counseling, 0, , . | 0.9 | 1 |