# Birgit H Funke

List of Publications by Year
in descending order



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|---|---|---|---|
| 82 papers | 9,779 citations | 61984 43 h-index | 64796 79 g-index |
| 92 all docs | 92 docs citations | 92 times ranked | 14125 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | TBX1 Is Responsible for Cardiovascular Defects in Velo-Cardio-Facial/DiGeorge Syndrome. Cell, 2001, 104, 619-629. | 28.9 | 884 |
| 2 | ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747. | 2.4 | 794 |
| 3 | Genetic Misdiagnoses and the Potential for Health Disparities. New England Journal of Medicine, 2016, 375, 655-665. | 27.0 | 602 |
| 4 | Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. Genetics in Medicine, 2017, 19, 192-203. | 2.4 | 585 |
| 5 | Assuring the quality of next-generation sequencing in clinical laboratory practice. Nature Biotechnology, 2012, 30, 1033-1036. | 17.5 | 437 |
| 6 | A common molecular basis for rearrangement disorders on chromosome 22q11. Human Molecular Genetics, 1999, 8, 1157-1167. | 2.9 | 385 |
| 7 | Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158. | 2.4 | 355 |
| 8 | Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. Genetics in Medicine, 2015, 17, 880-888. | 2.4 | 344 |
| 9 | Cardiovascular Pathology in Hutchinson-Gilford Progeria: Correlation With the Vascular Pathology of Aging. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 2301-2309. | 2.4 | 332 |
| 10 | College of American Pathologists' Laboratory Standards for Next-Generation Sequencing Clinical Tests. Archives of Pathology and Laboratory Medicine, 2015, 139, 481-493. | 2.5 | 315 |
| 11 | The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. Genetics in Medicine, 2014, 16, 601-608. | 2.4 | 284 |
| 12 | Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGenâ€™s Inherited Cardiomyopathy Expert Panel. Genetics in Medicine, 2018, 20, 351-359. | 2.4 | 283 |
| 13 | Best practices for benchmarking germline small-variant calls in human genomes. Nature Biotechnology, 2019, 37, 555-560. | 17.5 | 273 |
| 14 | Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460. | 3.6 | 267 |
| 15 | Inherited Cardiomyopathies. Journal of Molecular Diagnostics, 2013, 15, 158-170. | 2.8 | 172 |
| 16 | Navigating highly homologous genes in a molecular diagnostic setting: a resource for clinical next-generation sequencing. Genetics in Medicine, 2016, 18, 1282-1289. | 2.4 | 170 |
| 17 | Genetic variation in DTNBP1 influences general cognitive ability. Human Molecular Genetics, 2006, 15, 1563-1568. | 2.9 | 160 |
| 18 | Association of the DTNBP1 Locus with Schizophrenia in a U.S. Population. American Journal of Human Genetics, 2004, 75, 891-898. | 6.2 | 155 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398. | 1.6 | 148 |
| 20 | Genetic Testing for Dilated Cardiomyopathy in Clinical Practice. Journal of Cardiac Failure, 2012, 18, 296-303. | 1.7 | 145 |
| 21 | Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603. | 2.2 | 142 |
| 22 | Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693. | 17.5 | 134 |
| 23 | ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622. | 2.5 | 132 |
| 24 | Burden of Rare Sarcomere Gene Variants in the Framingham and Jackson Heart Study Cohorts. American Journal of Human Genetics, 2012, 91, 513-519. | 6.2 | 116 |
| 25 | Familial Dilated Cardiomyopathy Caused by an Alpha-Tropomyosin Mutation. Journal of the American College of Cardiology, 2010, 55, 320-329. | 2.8 | 104 |
| 26 | DTNBP1 genotype influences cognitive decline in schizophrenia. Schizophrenia Research, 2007, 89, 169-172. | 2.0 | 102 |
| 27 | Dysbindin Genotype and Negative Symptoms in Schizophrenia. American Journal of Psychiatry, 2006, 163, 532-534. | 7.2 | 101 |
| 28 | Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605. | 6.2 | 99 |
| 29 | A Role for the CHC22 Clathrin Heavy-Chain Isoform in Human Glucose Metabolism. Science, 2009, 324, 1192-1196. | 12.6 | 98 |
| 30 | Regional Variation in RBM20 Causes a Highly Penetrant Arrhythmogenic Cardiomyopathy. Circulation: Heart Failure, 2019, 12, e005371. | 3.9 | 96 |
| 31 | Dilated Cardiomyopathy. Circulation: Arrhythmia and Electrophysiology, 2013, 6, 228-237. | 4.8 | 93 |
| 32 | COMT genotype increases risk for bipolar I disorder and influences neurocognitive performance. Bipolar Disorders, 2007, 9, 370-376. | 1.9 | 80 |
| 33 | Diagnostic gene sequencing panels: from design to reportâ€”a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 453-461. | 2.4 | 77 |
| 34 | The GeneInsight suite: a platform to support laboratory and provider use of DNA-based genetic testing. Human Mutation, 2011, 32, 532-536. | 2.5 | 75 |
| 35 | Development and Validation of a Computational Method for Assessment of Missense Variants in Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 183-192. | 6.2 | 73 |
| 36 | A novel custom resequencing array for dilated cardiomyopathy. Genetics in Medicine, 2010, 12, 268-278. | 2.4 | 71 |

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|---|---|---|---|
| 37 | Behavior of mice with mutations in the conserved region deleted in velocardiofacial/DiGeorge syndrome. Neurogenetics, 2006, 7, 247-257. | 1.4 | 70 |
| 38 | Improving hearing loss gene testing: a systematic review of gene evidence toward more efficient next-generation sequencingâ€“based diagnostic testing and interpretation. Genetics in Medicine, 2016, 18, 545-553. | 2.4 | 63 |
| 39 | VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. Genetics in Medicine, 2016, 18, 712-719. | 2.4 | 61 |
| 40 | Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477. | 2.4 | 61 |
| 41 | Mouse and Human CRKL Is Dosage Sensitive for Cardiac Outflow Tract Formation. American Journal of Human Genetics, 2015, 96, 235-244. | 6.2 | 58 |
| 42 | Over-expression of a human chromosome 22q11.2 segment including TXNRD2, COMT and ARVCF developmentally affects incentive learning and working memory in mice. Human Molecular Genetics, 2009, 18, 3914-3925. | 2.9 | 53 |
| 43 | Comprehensive Diagnostic Testing for Stereocilin. Journal of Molecular Diagnostics, 2014, 16, 639-647. | 2.8 | 53 |
| 44 | Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. Genetics in Medicine, 2021, 23, 856-864. | 2.4 | 45 |
| 45 | A 200-kb region of human chromosome 22q11.2 confers antipsychotic-responsive behavioral abnormalities in mice. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19132-19137. | 7.1 | 44 |
| 46 | Evaluation of Second-Generation Sequencing of 19 Dilated Cardiomyopathy Genes for Clinical Applications. Journal of Molecular Diagnostics, 2010, 12, 818-827. | 2.8 | 43 |
| 47 | Gene expression profile of trisomy 21 placentas: A potential approach for designing noninvasive techniques of prenatal diagnosis. American Journal of Obstetrics and Gynecology, 2002, 187, 457-462. | 1.3 | 42 |
| 48 | Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126. | 2.5 | 37 |
| 49 | Murine protein which binds preferentially to oligo-C-rich single-stranded nucleic acids. Nucleic Acids Research, 1994, 22, 1885-1889. | 14.5 | 35 |
| 50 | Development and Validation of Targeted Next-Generation Sequencing Panels for Detection of Germline Variants in Inherited Diseases. Archives of Pathology and Laboratory Medicine, 2017, 141, 787-797. | 2.5 | 35 |
| 51 | LAMP2 Microdeletions in Patients With Danon Disease. Circulation: Cardiovascular Genetics, 2010, 3, 129-137. | 5.1 | 31 |
| 52 | Next generation sequencingâ€based copy number analysis reveals low prevalence of deletions and duplications in 46 genes associated with genetic cardiomyopathies. Molecular Genetics & Genomic Medicine, 2016, 4, 143-151. | 1.2 | 29 |
| 53 | The Case for Laboratory Developed Procedures. Academic Pathology, 2017, 4, 2374289517708309. | 1.1 | 24 |
| 54 | Designing and Implementing NGS Tests for Inherited Disorders. Journal of Molecular Diagnostics, 2019, 21, 369-374. | 2.8 | 23 |

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|---|---|---|---|
| 55 | Isolation and Characterization of a Human Gene Containing a Nuclear Localization Signal from the Critical Region for Veloâ€“Cardioâ€“Facial Syndrome on 22q11. Genomics, 1998, 53, 146-154. | 2.9 | 22 |
| 56 | COMT genotype and manic symptoms in schizophrenia. Schizophrenia Research, 2006, 87, 28-31. | 2.0 | 22 |
| 57 | Considerations for clinical curation, classification, and reporting of low-penetrance and low effect size variants associated with disease risk. Genetics in Medicine, 2019, 21, 2765-2773. | 2.4 | 20 |
| 58 | The CHC22 Clathrin-GLUT4 Transport Pathway Contributes to Skeletal Muscle Regeneration. PLoS ONE, 2013, 8, e77787. | 2.5 | 19 |
| 59 | Development of Clinical Domain Working Groups for the Clinical Genome Resource (ClinGen): lessons learned and plans for the future. Genetics in Medicine, 2019, 21, 987-993. | 2.4 | 17 |
| 60 | Analysis of TBX1 Variation in Patients with Psychotic and Affective Disorders. Molecular Medicine, 2007, 13, 407-414. | 4.4 | 16 |
| 61 | Isolation and Characterization of a Novel Gene Containing WD40 Repeats from the Region Deleted in Veloâ€“cardioâ€“facial/ DiGeorge Syndrome on Chromosome 22q11. Genomics, 2001, 73, 264-271. | 2.9 | 15 |
| 62 | Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190. | 6.2 | 15 |
| 63 | Design and Reporting Considerations for Genetic Screening Tests. Journal of Molecular Diagnostics, 2020, 22, 599-609. | 2.8 | 15 |
| 64 | Multiplexed Reference Materials as Controls for Diagnostic Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 882-889. | 2.8 | 13 |
| 65 | The genetic architecture of Plakophilin 2 cardiomyopathy. Genetics in Medicine, 2021, 23, 1961-1968. | 2.4 | 13 |
| 66 | Expression of Cdcrel-1 (Pnutl1), a gene frequently deleted in velo-cardio-facial syndrome/DiGeorge syndrome. Mechanisms of Development, 2000, 96, 121-124. | 1.7 | 12 |
| 67 | Alcama mediates Edn1 signaling during zebrafish cartilage morphogenesis. Developmental Biology, 2011, 349, 483-493. | 2.0 | 11 |
| 68 | NGS testing for cardiomyopathy: Utility of adding RASopathy-associated genes. Human Mutation, 2018, 39, 954-958. | 2.5 | 11 |
| 69 | Pathogenicity of Hypertrophic Cardiomyopathy Variants. Circulation: Cardiovascular Genetics, 2017, 10, . | 5.1 | 10 |
| 70 | Clinical diversity of MYH7â€related cardiomyopathies: Insights into genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2019, 179, 365-372. | 1.2 | 10 |
| 71 | An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587. | 2.5 | 10 |
| 72 | A Novel, Single Nucleotide Polymorphism-Based Assay to Detect 22q11 Deletions. Genetic Testing and Molecular Biomarkers, 2007, 11, 91-100. | 1.7 | 5 |

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|---|---|---|---|
| 73 | Harmonizing the Collection of Clinical Data on Genetic Testing Requisition Forms to Enhance Variant Interpretation in Hypertrophic Cardiomyopathy (HCM). Journal of Molecular Diagnostics, 2021, 23, 589-598. | 2.8 | 5 |
| 74 | A Framework of Critical Considerations in Clinical Exome Reanalyses by Clinical and Laboratory Standards Institute. Journal of Molecular Diagnostics, 2022, 24, 177-188. | 2.8 | 4 |
| 75 | Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 1-2. | 2.4 | 3 |
| 76 | Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846. | 2.4 | 3 |
| 77 | Creation of an Expert Curated Variant List for Clinical Genomic Test Development and Validation. Journal of Molecular Diagnostics, 2021, 23, 1500-1505. | 2.8 | 2 |
| 78 | Platform evaluation for rapid genotyping ofCYP2C9andVKORC1alleles. Personalized Medicine, 2009, 6, 449-457. | 1.5 | 1 |
| 79 | 121â€...Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , . | | 1 |
| 80 | Sarcomeric Dilated Cardiomyopathy: Onset from Infancy to Late Adulthood. Journal of Cardiac Failure, 2009, 15, S41-S42. | 1.7 | 0 |
| 81 | Classifying Germline Sequence Variants in the Era of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 799-806. | 3.2 | 0 |
| 82 | Use of â€œColdspotâ€•Regions in Variant Classification. Clinical Chemistry, 2020, 66, 1263-1265. | 3.2 | 0 |