Birgit H Funke

List of Publications by Year
in descending order

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	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
2	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
3	The genetic architecture of Plakophilin 2 cardiomyopathy. Genetics in Medicine, 2021, 23, 1961-1968.	2.4	13
4	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	2.4	3
5	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
6	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
7	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
8	Use of â€œColdspotâ€•Regions in Variant Classification. Clinical Chemistry, 2020, 66, 1263-1265.	3.2	0
9	Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477.	2.4	61
10	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
11	Design and Reporting Considerations for Genetic Screening Tests. Journal of Molecular Diagnostics, 2020, 22, 599-609.	2.8	15
12	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
13	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	6.2	99
14	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460.	3.6	267
15	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
16	Best practices for benchmarking germline small-variant calls in human genomes. Nature Biotechnology, 2019, 37, 555-560.	17.5	273
17	Regional Variation in RBM20 Causes a Highly Penetrant Arrhythmogenic Cardiomyopathy. Circulation: Heart Failure, 2019, 12, e005371.	3.9	96
18	121â€...Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1

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19	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
20	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
21	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
22	Designing and Implementing NGS Tests for Inherited Disorders. Journal of Molecular Diagnostics, 2019, 21, 369-374.	2.8	23
23	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
24	NGS testing for cardiomyopathy: Utility of adding RASopathy-associated genes. Human Mutation, 2018, 39, 954-958.	2.5	11
25	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
26	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603.	2.2	142
27	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	2.4	355
28	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
29	Pathogenicity of Hypertrophic Cardiomyopathy Variants. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	10
30	The Case for Laboratory Developed Procedures. Academic Pathology, 2017, 4, 2374289517708309.	1.1	24
31	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
32	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
33	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126.	2.5	37
34	Genetic Misdiagnoses and the Potential for Health Disparities. New England Journal of Medicine, 2016, 375, 655-665.	27.0	602
35	Multiplexed Reference Materials as Controls for Diagnostic Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 882-889.	2.8	13
36	Classifying Germline Sequence Variants in the Era of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 799-806.	3.2	0

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37	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
38	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
39	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 1-2.	2.4	3
40	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
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	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
43	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
44	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
45	Comprehensive Diagnostic Testing for Stereocilin. Journal of Molecular Diagnostics, 2014, 16, 639-647.	2.8	53
46	The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. Genetics in Medicine, 2014, 16, 601-608.	2.4	284
47	ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747.	2.4	794
48	Inherited Cardiomyopathies. Journal of Molecular Diagnostics, 2013, 15, 158-170.	2.8	172
49	Dilated Cardiomyopathy. Circulation: Arrhythmia and Electrophysiology, 2013, 6, 228-237.	4.8	93
50	The CHC22 Clathrin-GLUT4 Transport Pathway Contributes to Skeletal Muscle Regeneration. PLoS ONE, 2013, 8, e77787.	2.5	19
51	Genetic Testing for Dilated Cardiomyopathy in Clinical Practice. Journal of Cardiac Failure, 2012, 18, 296-303.	1.7	145
52	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
53	Assuring the quality of next-generation sequencing in clinical laboratory practice. Nature Biotechnology, 2012, 30, 1033-1036.	17.5	437
54	Alcama mediates Edn1 signaling during zebrafish cartilage morphogenesis. Developmental Biology, 2011, 349, 483-493.	2.0	11

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55	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
56	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
57	LAMP2 Microdeletions in Patients With Danon Disease. Circulation: Cardiovascular Genetics, 2010, 3, 129-137.	5.1	31
58	A novel custom resequencing array for dilated cardiomyopathy. Genetics in Medicine, 2010, 12, 268-278.	2.4	71
59	Familial Dilated Cardiomyopathy Caused by an Alpha-Tropomyosin Mutation. Journal of the American College of Cardiology, 2010, 55, 320-329.	2.8	104
60	Evaluation of Second-Generation Sequencing of 19 Dilated Cardiomyopathy Genes for Clinical Applications. Journal of Molecular Diagnostics, 2010, 12, 818-827.	2.8	43
61	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
62	A Role for the CHC22 Clathrin Heavy-Chain Isoform in Human Glucose Metabolism. Science, 2009, 324, 1192-1196.	12.6	98
63	Platform evaluation for rapid genotyping ofCYP2C9andVKORC1alleles. Personalized Medicine, 2009, 6, 449-457.	1.5	1
64	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
65	Sarcomeric Dilated Cardiomyopathy: Onset from Infancy to Late Adulthood. Journal of Cardiac Failure, 2009, 15, S41-S42.	1.7	0
66	A Novel, Single Nucleotide Polymorphism-Based Assay to Detect 22q11 Deletions. Genetic Testing and Molecular Biomarkers, 2007, 11, 91-100.	1.7	5
67	DTNBP1 genotype influences cognitive decline in schizophrenia. Schizophrenia Research, 2007, 89, 169-172.	2.0	102
68	Analysis of TBX1 Variation in Patients with Psychotic and Affective Disorders. Molecular Medicine, 2007, 13, 407-414.	4.4	16
69	COMT genotype increases risk for bipolar I disorder and influences neurocognitive performance. Bipolar Disorders, 2007, 9, 370-376.	1.9	80
70	COMT genotype and manic symptoms in schizophrenia. Schizophrenia Research, 2006, 87, 28-31.	2.0	22
71	Behavior of mice with mutations in the conserved region deleted in velocardiofacial/DiGeorge syndrome. Neurogenetics, 2006, 7, 247-257.	1.4	70
72	Dysbindin Genotype and Negative Symptoms in Schizophrenia. American Journal of Psychiatry, 2006, 163, 532-534.	7.2	101

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73	Genetic variation in DTNBP1 influences general cognitive ability. Human Molecular Genetics, 2006, 15, 1563-1568.	2.9	160
74	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
75	Association of the DTNBP1 Locus with Schizophrenia in a U.S. Population. American Journal of Human Genetics, 2004, 75, 891-898.	6.2	155
76	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
77	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
78	TBX1 Is Responsible for Cardiovascular Defects in Velo-Cardio-Facial/DiGeorge Syndrome. Cell, 2001, 104, 619-629.	28.9	884
79	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
80	A common molecular basis for rearrangement disorders on chromosome 22q11. Human Molecular Genetics, 1999, 8, 1157-1167.	2.9	385
81	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
82	Murine protein which binds preferentially to oligo-C-rich single-stranded nucleic acids. Nucleic Acids Research, 1994, 22, 1885-1889.	14.5	35