Jonathan S Berg

List of Publications by Year
in descending order

		87888	37204
104	10,158	38	96
papers	citations	h-index	g-index

118	118	118	13478
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Testing and extending strategies for identifying genetic diseaseâ€“related encounters in pediatric patients. Genetics in Medicine, 2022, 24, 831-838.	2.4	0
2	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.	8.2	34
3	A systematic review of the methodological quality of economic evaluations in genetic screening and testing for monogenic disorders. Genetics in Medicine, 2022, 24, 262-288.	2.4	4
4	The Rise of Population Genomic Screening: Characteristics of Current Programs and the Need for Evidence Regarding Optimal Implementation. Journal of Personalized Medicine, 2022, 12, 692.	2.5	11
5	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. Cell Genomics, 2022, 2, 100131.	6.5	11
6	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
7	Newborn screening for neurodevelopmental diseases: Are we there yet?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 222-230.	1.6	10
8	A behavior-theoretic evaluation of values clarification on parental beliefs and intentions toward genomic sequencing for newborns. Social Science and Medicine, 2021, 271, 112037.	3.8	13
9	Actionability of commercial laboratory sequencing panels for newborn screening and the importance of transparency for parental decision-making. Genome Medicine, 2021, 13, 50.	8.2	20
10	Burden or benefit? Effects of providing education about and the option to request additional genomic findings from diagnostic exome sequencing: A randomized controlled trial. Patient Education and Counseling, 2021, 104, 2989-2998.	2.2	0
11	Evaluating the clinical utility of early exome sequencing in diverse pediatric outpatient populations in the North Carolina Clinical Genomic Evaluation of Next-generation Exome Sequencing (NCGENES) 2 study: a randomized controlled trial. Trials, 2021, 22, 395.	1.6	5
12	Pre-capture multiplexing provides additional power to detect copy number variation in exome sequencing. BMC Bioinformatics, 2021, 22, 374.	2.6	3
13	Noninvasive prenatal exome sequencing diagnostic utility limited by sequencing depth and fetal fraction. Prenatal Diagnosis, 2021, , .	2.3	2
14	A Validated Functional Analysis of Partner and Localizer of BRCA2 Missense Variants for Use in Clinical Variant Interpretation. Journal of Molecular Diagnostics, 2021, 23, 847-864.	2.8	1
15	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	8.2	312
16	Parentsâ€™ perceptions of personal utility of exome sequencing results. Genetics in Medicine, 2020, 22, 752-757.	2.4	37
17	Treatment-resistant psychotic symptoms and early-onset dementia: A case report of the 3q29 deletion syndrome. Schizophrenia Research, 2020, 224, 195-197.	2.0	8
18	Genomic Sequencing for Newborn Screening: Results of the NC NEXUS Project. American Journal of Human Genetics, 2020, 107, 596-611.	6.2	63

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19	Parental Views on Newborn Next Generation Sequencing: Implications for Decision Support. Maternal and Child Health Journal, 2020, 24, 856-864.	1.5	15
20	Lessons learned about harmonizing survey measures for the CSER consortium. Journal of Clinical and Translational Science, 2020, 4, 537-546.	0.6	16
21	Assessing the implications of positive genomic screening results. Personalized Medicine, 2020, 17, 101-109.	1.5	6
22	An approach to integrating exome sequencing for fetal structural anomalies into clinical practice. Genetics in Medicine, 2020, 22, 954-961.	2.4	49
23	Referencing BRCA in hereditary cancer risk discussions: In search of an anchor in a sea of uncertainty. Journal of Genetic Counseling, 2020, 29, 949-959.	1.6	4
24	Selecting secondary findings to report: Creating a list that suits your study. , 2020, , 43-58.		0
25	Values clarification and parental decision making about newborn genomic sequencing.. Health Psychology, 2020, 39, 335-344.	1.6	9
26	Treatment-resistant psychotic symptoms and the 15q11.2 BP1â€“BP2 (Burnside-Butler) deletion syndrome: case report and review of the literature. Translational Psychiatry, 2020, 10, 42.	4.8	11
27	Engaging community stakeholders in research on best practices for clinical genomic sequencing. Personalized Medicine, 2020, 17, 435-444.	1.5	9
28	Expert and lay perspectives on burden, risk, tolerability, and acceptability of clinical interventions for genetic disorders. Genetics in Medicine, 2019, 21, 2561-2568.	2.4	4
29	An Age-Based Framework for Evaluating Genome-Scale Sequencing Results in Newborn Screening. Journal of Pediatrics, 2019, 209, 68-76.	1.8	50
30	Selective serotonin reuptake inhibitors ameliorate MEGF10 myopathy. Human Molecular Genetics, 2019, 28, 2365-2377.	2.9	7
31	FDA oversight of NSIGHT genomic research: the need for an integrated systems approach to regulation. Npj Genomic Medicine, 2019, 4, 32.	3.8	6
32	Comparative analysis of functional assay evidence use by ClinGen Variant Curation Expert Panels. Genome Medicine, 2019, 11, 77.	8.2	34
33	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
34	Factors influencing NCGENES research participants- requests for non-medically actionable secondary findings. Genetics in Medicine, 2019, 21, 1092-1099.	2.4	11
35	Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study. Genetics in Medicine, 2019, 21, 1100-1110.	2.4	111
36	Anticipated responses of early adopter genetic specialists and nongenetic specialists to unsolicited genomic secondary findings. Genetics in Medicine, 2018, 20, 1186-1195.	2.4	11

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37	Diagnostic utility of exome sequencing in the evaluation of neuromuscular disorders. Neurology: Genetics, 2018, 4, e212.	1.9	42
38	Developmental Delay, Treatment-Resistant Psychosis, and Early-Onset Dementia in a Man With 22q11 Deletion Syndrome and Huntingtonâ€™s Disease. American Journal of Psychiatry, 2018, 175, 400-407.	7.2	9
39	â€œPossibly positive or certainly uncertain?â€ participantsâ€™ responses to uncertain diagnostic results from exome sequencing. Genetics in Medicine, 2018, 20, 313-319.	2.4	39
40	Navigating the researchâ€“clinical interface in genomic medicine: analysis from the CSER Consortium. Genetics in Medicine, 2018, 20, 545-553.	2.4	34
41	The who, what, and why of research participantsâ€™ intentions to request a broad range of secondary findings in a diagnostic genomic sequencing study. Genetics in Medicine, 2018, 20, 760-769.	2.4	22
42	Parental preferences toward genomic sequencing for non-medically actionable conditions in children: a discrete-choice experiment. Genetics in Medicine, 2018, 20, 181-189.	2.4	24
43	Increasing the diagnostic yield of exome sequencing by copy number variant analysis. PLoS ONE, 2018, 13, e0209185.	2.5	58
44	Combination of exome sequencing and immune testing confirms Aicardiâ€“GoutiÃ¨res syndrome type 5 in a challenging pediatric neurology case. Journal of Physical Education and Sports Management, 2018, 4, a002758.	1.2	6
45	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
46	Too much of a good thing? Overdiagnosis, or overestimating risk in preventive genomic screening. Personalized Medicine, 2018, 15, 343-346.	1.5	11
47	ClinGen and ClinVar â€“ Enabling Genomics in Precision Medicine. Human Mutation, 2018, 39, 1473-1475.	2.5	14
48	The progression of the ClinGen gene clinical validity classification over time. Human Mutation, 2018, 39, 1494-1504.	2.5	16
49	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. American Journal of Human Genetics, 2018, 103, 319-327.	6.2	122
50	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.	1.2	15
51	Evaluating parentsâ€™ decisions about next-generation sequencing for their child in the NC NEXUS (North Carolina Newborn Exome Sequencing for Universal Screening) study: a randomized controlled trial protocol. Trials, 2018, 19, 344.	1.6	28
52	Navigating the nuances of clinical sequence variant interpretation in Mendelian disease. Genetics in Medicine, 2018, 20, 918-926.	2.4	40
53	Quantifying the potential of functional evidence to reclassify variants of uncertain significance in the categorical and Bayesian interpretation frameworks. Human Mutation, 2018, 39, 1531-1541.	2.5	52
54	hCALCRL mutation causes autosomal recessive nonimmune hydrops fetalis with lymphatic dysplasia. Journal of Experimental Medicine, 2018, 215, 2339-2353.	8.5	25

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55	Identification of Clonal Hematopoiesis Mutations in Solid Tumor Patients Undergoing Unpaired Next-Generation Sequencing Assays. Clinical Cancer Research, 2018, 24, 5918-5924.	7.0	84
56	Identification of clonal hematopoiesis mutations in solid tumor patients undergoing unpaired commercial next-generation sequencing assays.. Journal of Clinical Oncology, 2018, 36, 12068-12068.	1.6	1
57	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
58	Genome sequencing and carrier testing: decisions on categorization and whether to disclose results of carrier testing. Genetics in Medicine, 2017, 19, 803-808.	2.4	24
59	Development and Validation of a Genomic Knowledge Scale to Advance Informed Decision-Making Research in Genomic Sequencing. MDM Policy and Practice, 2017, 2, 238146831769258.	0.9	32
60	Finding the Rare Pathogenic Variants in a Human Genome. JAMA - Journal of the American Medical Association, 2017, 317, 1904.	7.4	38
61	Prenatal exome sequencing in anomalous fetuses: new opportunities and challenges. Genetics in Medicine, 2017, 19, 1207-1216.	2.4	135
62	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. American Journal of Human Genetics, 2017, 100, 895-906.	6.2	403
63	Whole Exome Sequencing Identifies Truncating Variants in Nuclear Envelope Genes in Patients With Cardiovascular Disease. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	34
64	Genetic screening: birthright or earned with age?. Expert Review of Molecular Diagnostics, 2017, 17, 735-738.	3.1	7
65	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68
66	Exploring the importance of case-level clinical information for variant interpretation. Genetics in Medicine, 2017, 19, 3-5.	2.4	12
67	Examining the Cascade of Participant Attrition in a Genomic Medicine Research Study: Barriers and Facilitators to Achieving Diversity. Public Health Genomics, 2017, 20, 332-342.	1.0	8
68	Genetic Complexity of Mitral Valve Prolapse Revealed by Clinical and Genetic Evaluation of a Large Family. Journal of Heart Valve Disease, 2017, 26, 569-580.	0.5	1
69	A missing link in the bench-to-bedside paradigm: engaging regulatory stakeholders in clinical genomics research. Genome Medicine, 2016, 8, 95.	8.2	6
70	Generating a taxonomy for genetic conditions relevant to reproductive planning. American Journal of Medical Genetics, Part A, 2016, 170, 565-573.	1.2	25
71	Germline Analysis from Tumorâ€“Germline Sequencing Dyads to Identify Clinically Actionable Secondary Findings. Clinical Cancer Research, 2016, 22, 4087-4094.	7.0	75
72	A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation. Genetics in Medicine, 2016, 18, 1258-1268.	2.4	89

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73	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. American Journal of Human Genetics, 2016, 98, 1067-1076.	6.2	432
74	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
75	Patients' ratings of genetic conditions validate a taxonomy to simplify decisions about preconception carrier screening via genome sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 574-582.	1.2	20
76	Defining the Clinical Value of a Genomic Diagnosis in the Era of Next-Generation Sequencing. Annual Review of Genomics and Human Genetics, 2016, 17, 303-332.	6.2	43
77	Supporting Parental Decisions About Genomic Sequencing for Newborn Screening: The NC NEXUS Decision Aid. Pediatrics, 2016, 137, S16-S23.	2.1	45
78	The promise and peril of genomic screening in the general population. Genetics in Medicine, 2016, 18, 593-599.	2.4	53
79	A semiquantitative metric for evaluating clinical actionability of incidental or secondary findings from genome-scale sequencing. Genetics in Medicine, 2016, 18, 467-475.	2.4	74
80	Look Before You Leap. Obstetrics and Gynecology, 2015, 125, 1299-1305.	2.4	6
81	Genomic screening of the general adult population: key concepts for assessing net benefit with systematic evidence reviews. Genetics in Medicine, 2015, 17, 441-443.	2.4	19
82	ClinGen â€" The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	27.0	1,016
83	High Diagnostic Yield of Whole Exome Sequencing in Participants With Retinal Dystrophies in a Clinical Ophthalmology Setting. American Journal of Ophthalmology, 2015, 160, 354-363.e9.	3.3	27
84	Incidental Findings with Genomic Testing: Implications for Genetic Counseling Practice. Current Genetic Medicine Reports, 2015, 3, 166-176.	1.9	68
85	Identifying gene disruptions in novel balanced de novo constitutional translocations in childhood cancer patients by whole-genome sequencing. Genetics in Medicine, 2015, 17, 831-835.	2.4	7
86	Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents. American Journal of Human Genetics, 2015, 97, 6-21.	6.2	453
87	The phenotype of multiple congenital anomaliesâ€hypotoniaâ€seizures syndrome 1: Report and review. American Journal of Medical Genetics, Part A, 2015, 167, 2176-2181.	1.2	24
88	Potential Uses and Inherent Challenges of Using Genome-Scale Sequencing to Augment Current Newborn Screening. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a023150.	6.2	28
89	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
90	Genome-Scale Sequencing in Clinical Care. JAMA - Journal of the American Medical Association, 2014, 312, 1865.	7.4	20

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91	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.	6.2	342
92	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in Medicine, 2013, 15, 565-574.	2.4	2,186
93	ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747.	2.4	794
94	Description and pilot results from a novel method for evaluating return of incidental findings from next-generation sequencing technologies. Genetics in Medicine, 2013, 15, 721-728.	2.4	40
95	Response to Lindor et al.. Genetics in Medicine, 2013, 15, 409-410.	2.4	1
96	We screen newborns, donâ€™t we?: realizing the promise of public health genomics. Genetics in Medicine, 2013, 15, 332-334.	2.4	64
97	Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. Genetics in Medicine, 2013, 15, 860-867.	2.4	99
98	An informatics approach to analyzing the incidentalome. Genetics in Medicine, 2013, 15, 36-44.	2.4	148
99	Crowdsourcing to define the clinical actionability of incidental findings of genetic testing. North Carolina Medical Journal, 2013, 74, 501-2.	0.2	1
100	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
101	Next generation massively parallel sequencing of targeted exomes to identify genetic mutations in primary ciliary dyskinesia: Implications for application to clinical testing. Genetics in Medicine, 2011, 13, 218-229.	2.4	59
102	Deploying whole genome sequencing in clinical practice and public health: Meeting the challenge one bin at a time. Genetics in Medicine, 2011, 13, 499-504.	2.4	451
103	Common recurrent microduplication syndromes: Diagnosis and management in clinical practice. American Journal of Medical Genetics, Part A, 2010, 152A, 1066-1078.	1.2	32
104	Loss of De Novo DNA Methylation Causes Expansion of the Mouse Hematopoietic Stem Cell Pool. Blood, 2010, 116, 835-835.	1.4	3