

Jonathan S Berg

List of Publications by Year in descending order

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Version: 2024-02-01

104
papers

10,158
citations

87723

38
h-index

37111

96
g-index

118
all docs

118
docs citations

118
times ranked

13478
citing authors

#	ARTICLE	IF	CITATIONS
1	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. <i>Genetics in Medicine</i> , 2013, 15, 565-574.	1.1	2,186
2	ClinGen – The Clinical Genome Resource. <i>New England Journal of Medicine</i> , 2015, 372, 2235-2242.	13.9	1,016
3	ACMG clinical laboratory standards for next-generation sequencing. <i>Genetics in Medicine</i> , 2013, 15, 733-747.	1.1	794
4	Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents. <i>American Journal of Human Genetics</i> , 2015, 97, 6-21.	2.6	453
5	Deploying whole genome sequencing in clinical practice and public health: Meeting the challenge one bin at a time. <i>Genetics in Medicine</i> , 2011, 13, 499-504.	1.1	451
6	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. <i>American Journal of Human Genetics</i> , 2016, 98, 1067-1076.	2.6	432
7	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. <i>American Journal of Human Genetics</i> , 2017, 100, 895-906.	2.6	403
8	Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices In Between. <i>American Journal of Human Genetics</i> , 2014, 94, 818-826.	2.6	342
9	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. <i>Genome Research</i> , 2015, 25, 305-315.	2.4	313
10	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. <i>Genome Medicine</i> , 2020, 12, 3.	3.6	312
11	Newborn Sequencing in Genomic Medicine and Public Health. <i>Pediatrics</i> , 2017, 139, .	1.0	174
12	Exploring concordance and discordance for return of incidental findings from clinical sequencing. <i>Genetics in Medicine</i> , 2012, 14, 405-410.	1.1	149
13	An informatics approach to analyzing the incidentalome. <i>Genetics in Medicine</i> , 2013, 15, 36-44.	1.1	148
14	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. <i>American Journal of Human Genetics</i> , 2016, 98, 1051-1066.	2.6	137
15	Prenatal exome sequencing in anomalous fetuses: new opportunities and challenges. <i>Genetics in Medicine</i> , 2017, 19, 1207-1216.	1.1	135
16	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and gene-level specification of the ACMG/AMP guidelines for sequence variant interpretation. <i>Human Mutation</i> , 2018, 39, 1614-1622.	1.1	132
17	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. <i>American Journal of Human Genetics</i> , 2018, 103, 319-327.	2.6	122
18	Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study. <i>Genetics in Medicine</i> , 2019, 21, 1100-1110.	1.1	111

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19	Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. <i>Genetics in Medicine</i> , 2013, 15, 860-867.	1.1	99
20	A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation. <i>Genetics in Medicine</i> , 2016, 18, 1258-1268.	1.1	89
21	Identification of Clonal Hematopoiesis Mutations in Solid Tumor Patients Undergoing Unpaired Next-Generation Sequencing Assays. <i>Clinical Cancer Research</i> , 2018, 24, 5918-5924.	3.2	84
22	Germline Analysis from Tumorâ€™Germline Sequencing Dyads to Identify Clinically Actionable Secondary Findings. <i>Clinical Cancer Research</i> , 2016, 22, 4087-4094.	3.2	75
23	A semiquantitative metric for evaluating clinical actionability of incidental or secondary findings from genome-scale sequencing. <i>Genetics in Medicine</i> , 2016, 18, 467-475.	1.1	74
24	Incidental Findings with Genomic Testing: Implications for Genetic Counseling Practice. <i>Current Genetic Medicine Reports</i> , 2015, 3, 166-176.	1.9	68
25	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. <i>Genetics in Medicine</i> , 2017, 19, 575-582.	1.1	68
26	We screen newborns, donâ€™t we?: realizing the promise of public health genomics. <i>Genetics in Medicine</i> , 2013, 15, 332-334.	1.1	64
27	Genomic Sequencing for Newborn Screening: Results of the NC NEXUS Project. <i>American Journal of Human Genetics</i> , 2020, 107, 596-611.	2.6	63
28	Next generation massively parallel sequencing of targeted exomes to identify genetic mutations in primary ciliary dyskinesia: Implications for application to clinical testing. <i>Genetics in Medicine</i> , 2011, 13, 218-229.	1.1	59
29	Increasing the diagnostic yield of exome sequencing by copy number variant analysis. <i>PLoS ONE</i> , 2018, 13, e0209185.	1.1	58
30	The Gene Curation Coalition: A global effort to harmonize geneâ€™disease evidence resources. <i>Genetics in Medicine</i> , 2022, 24, 1732-1742.	1.1	56
31	The promise and peril of genomic screening in the general population. <i>Genetics in Medicine</i> , 2016, 18, 593-599.	1.1	53
32	Quantifying the potential of functional evidence to reclassify variants of uncertain significance in the categorical and Bayesian interpretation frameworks. <i>Human Mutation</i> , 2018, 39, 1531-1541.	1.1	52
33	An Age-Based Framework for Evaluating Genome-Scale Sequencing Results in Newborn Screening. <i>Journal of Pediatrics</i> , 2019, 209, 68-76.	0.9	50
34	An approach to integrating exome sequencing for fetal structural anomalies into clinical practice. <i>Genetics in Medicine</i> , 2020, 22, 954-961.	1.1	49
35	Supporting Parental Decisions About Genomic Sequencing for Newborn Screening: The NC NEXUS Decision Aid. <i>Pediatrics</i> , 2016, 137, S16-S23.	1.0	45
36	Defining the Clinical Value of a Genomic Diagnosis in the Era of Next-Generation Sequencing. <i>Annual Review of Genomics and Human Genetics</i> , 2016, 17, 303-332.	2.5	43

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37	Diagnostic utility of exome sequencing in the evaluation of neuromuscular disorders. <i>Neurology: Genetics</i> , 2018, 4, e212.	0.9	42
38	Description and pilot results from a novel method for evaluating return of incidental findings from next-generation sequencing technologies. <i>Genetics in Medicine</i> , 2013, 15, 721-728.	1.1	40
39	Navigating the nuances of clinical sequence variant interpretation in Mendelian disease. <i>Genetics in Medicine</i> , 2018, 20, 918-926.	1.1	40
40	“Possibly positive or certainly uncertain?” participants’ responses to uncertain diagnostic results from exome sequencing. <i>Genetics in Medicine</i> , 2018, 20, 313-319.	1.1	39
41	Finding the Rare Pathogenic Variants in a Human Genome. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 1904.	3.8	38
42	Parents’ perceptions of personal utility of exome sequencing results. <i>Genetics in Medicine</i> , 2020, 22, 752-757.	1.1	37
43	Whole Exome Sequencing Identifies Truncating Variants in Nuclear Envelope Genes in Patients With Cardiovascular Disease. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	34
44	Navigating the research-clinical interface in genomic medicine: analysis from the CSER Consortium. <i>Genetics in Medicine</i> , 2018, 20, 545-553.	1.1	34
45	Comparative analysis of functional assay evidence use by ClinGen Variant Curation Expert Panels. <i>Genome Medicine</i> , 2019, 11, 77.	3.6	34
46	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. <i>Genome Medicine</i> , 2022, 14, 6.	3.6	34
47	Common recurrent microduplication syndromes: Diagnosis and management in clinical practice. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1066-1078.	0.7	32
48	Development and Validation of a Genomic Knowledge Scale to Advance Informed Decision-Making Research in Genomic Sequencing. <i>MDM Policy and Practice</i> , 2017, 2, 238146831769258.	0.5	32
49	Potential Uses and Inherent Challenges of Using Genome-Scale Sequencing to Augment Current Newborn Screening. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2015, 5, a023150.	2.9	28
50	Evaluating parents’ 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. <i>Trials</i> , 2018, 19, 344.	0.7	28
51	High Diagnostic Yield of Whole Exome Sequencing in Participants With Retinal Dystrophies in a Clinical Ophthalmology Setting. <i>American Journal of Ophthalmology</i> , 2015, 160, 354-363.e9.	1.7	27
52	Generating a taxonomy for genetic conditions relevant to reproductive planning. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 565-573.	0.7	25
53	<i>CALCRL</i> mutation causes autosomal recessive nonimmune hydrops fetalis with lymphatic dysplasia. <i>Journal of Experimental Medicine</i> , 2018, 215, 2339-2353.	4.2	25
54	The phenotype of multiple congenital anomalies-hypotonia-seizures syndrome 1: Report and review. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2176-2181.	0.7	24

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55	Genome sequencing and carrier testing: decisions on categorization and whether to disclose results of carrier testing. <i>Genetics in Medicine</i> , 2017, 19, 803-808.	1.1	24
56	Parental preferences toward genomic sequencing for non-medically actionable conditions in children: a discrete-choice experiment. <i>Genetics in Medicine</i> , 2018, 20, 181-189.	1.1	24
57	The who, what, and why of research participants'™ intentions to request a broad range of secondary findings in a diagnostic genomic sequencing study. <i>Genetics in Medicine</i> , 2018, 20, 760-769.	1.1	22
58	Genome-Scale Sequencing in Clinical Care. <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 1865.	3.8	20
59	Patients' ratings of genetic conditions validate a taxonomy to simplify decisions about preconception carrier screening via genome sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 574-582.	0.7	20
60	Actionability of commercial laboratory sequencing panels for newborn screening and the importance of transparency for parental decision-making. <i>Genome Medicine</i> , 2021, 13, 50.	3.6	20
61	Genomic screening of the general adult population: key concepts for assessing net benefit with systematic evidence reviews. <i>Genetics in Medicine</i> , 2015, 17, 441-443.	1.1	19
62	Development of Clinical Domain Working Groups for the Clinical Genome Resource (ClinGen): lessons learned and plans for the future. <i>Genetics in Medicine</i> , 2019, 21, 987-993.	1.1	17
63	The progression of the ClinGen gene clinical validity classification over time. <i>Human Mutation</i> , 2018, 39, 1494-1504.	1.1	16
64	Lessons learned about harmonizing survey measures for the CSER consortium. <i>Journal of Clinical and Translational Science</i> , 2020, 4, 537-546.	0.3	16
65	Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 898-909.	0.6	15
66	Parental Views on Newborn Next Generation Sequencing: Implications for Decision Support. <i>Maternal and Child Health Journal</i> , 2020, 24, 856-864.	0.7	15
67	ClinGen and ClinVar â€œ Enabling Genomics in Precision Medicine. <i>Human Mutation</i> , 2018, 39, 1473-1475.	1.1	14
68	A behavior-theoretic evaluation of values clarification on parental beliefs and intentions toward genomic sequencing for newborns. <i>Social Science and Medicine</i> , 2021, 271, 112037.	1.8	13
69	Exploring the importance of case-level clinical information for variant interpretation. <i>Genetics in Medicine</i> , 2017, 19, 3-5.	1.1	12
70	Anticipated responses of early adopter genetic specialists and nongenetic specialists to unsolicited genomic secondary findings. <i>Genetics in Medicine</i> , 2018, 20, 1186-1195.	1.1	11
71	Too much of a good thing? Overdiagnosis, or overestimating risk in preventive genomic screening. <i>Personalized Medicine</i> , 2018, 15, 343-346.	0.8	11
72	Factors influencing NCGENES research participants- requests for non-medically actionable secondary findings. <i>Genetics in Medicine</i> , 2019, 21, 1092-1099.	1.1	11

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73	Treatment-resistant psychotic symptoms and the 15q11.2 BP1-BP2 (Burnside-Butler) deletion syndrome: case report and review of the literature. <i>Translational Psychiatry</i> , 2020, 10, 42.	2.4	11
74	The Rise of Population Genomic Screening: Characteristics of Current Programs and the Need for Evidence Regarding Optimal Implementation. <i>Journal of Personalized Medicine</i> , 2022, 12, 692.	1.1	11
75	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. <i>Cell Genomics</i> , 2022, 2, 100131.	3.0	11
76	Newborn screening for neurodevelopmental diseases: Are we there yet?. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, 190, 222-230.	0.7	10
77	Developmental Delay, Treatment-Resistant Psychosis, and Early-Onset Dementia in a Man With 22q11 Deletion Syndrome and Huntington's Disease. <i>American Journal of Psychiatry</i> , 2018, 175, 400-407.	4.0	9
78	Values clarification and parental decision making about newborn genomic sequencing.. <i>Health Psychology</i> , 2020, 39, 335-344.	1.3	9
79	Engaging community stakeholders in research on best practices for clinical genomic sequencing. <i>Personalized Medicine</i> , 2020, 17, 435-444.	0.8	9
80	Examining the Cascade of Participant Attrition in a Genomic Medicine Research Study: Barriers and Facilitators to Achieving Diversity. <i>Public Health Genomics</i> , 2017, 20, 332-342.	0.6	8
81	Treatment-resistant psychotic symptoms and early-onset dementia: A case report of the 3q29 deletion syndrome. <i>Schizophrenia Research</i> , 2020, 224, 195-197.	1.1	8
82	Identifying gene disruptions in novel balanced de novo constitutional translocations in childhood cancer patients by whole-genome sequencing. <i>Genetics in Medicine</i> , 2015, 17, 831-835.	1.1	7
83	Genetic screening: birthright or earned with age?. <i>Expert Review of Molecular Diagnostics</i> , 2017, 17, 735-738.	1.5	7
84	Selective serotonin reuptake inhibitors ameliorate MEGF10 myopathy. <i>Human Molecular Genetics</i> , 2019, 28, 2365-2377.	1.4	7
85	Look Before You Leap. <i>Obstetrics and Gynecology</i> , 2015, 125, 1299-1305.	1.2	6
86	A missing link in the bench-to-bedside paradigm: engaging regulatory stakeholders in clinical genomics research. <i>Genome Medicine</i> , 2016, 8, 95.	3.6	6
87	Combination of exome sequencing and immune testing confirms Aicardi-Goutières syndrome type 5 in a challenging pediatric neurology case. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002758.	0.5	6
88	FDA oversight of NSIGHT genomic research: the need for an integrated systems approach to regulation. <i>Npj Genomic Medicine</i> , 2019, 4, 32.	1.7	6
89	Assessing the implications of positive genomic screening results. <i>Personalized Medicine</i> , 2020, 17, 101-109.	0.8	6
90	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. <i>Trials</i> , 2021, 22, 395.	0.7	5

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91	Expert and lay perspectives on burden, risk, tolerability, and acceptability of clinical interventions for genetic disorders. <i>Genetics in Medicine</i> , 2019, 21, 2561-2568.	1.1	4
92	Referencing <i>BRCA</i> in hereditary cancer risk discussions: In search of an anchor in a sea of uncertainty. <i>Journal of Genetic Counseling</i> , 2020, 29, 949-959.	0.9	4
93	A systematic review of the methodological quality of economic evaluations in genetic screening and testing for monogenic disorders. <i>Genetics in Medicine</i> , 2022, 24, 262-288.	1.1	4
94	Pre-capture multiplexing provides additional power to detect copy number variation in exome sequencing. <i>BMC Bioinformatics</i> , 2021, 22, 374.	1.2	3
95	Loss of De Novo DNA Methylation Causes Expansion of the Mouse Hematopoietic Stem Cell Pool. <i>Blood</i> , 2010, 116, 835-835.	0.6	3
96	Noninvasive prenatal exome sequencing diagnostic utility limited by sequencing depth and fetal fraction. <i>Prenatal Diagnosis</i> , 2021, , .	1.1	2
97	Response to Lindor et al.. <i>Genetics in Medicine</i> , 2013, 15, 409-410.	1.1	1
98	A Validated Functional Analysis of Partner and Localizer of BRCA2 Missense Variants for Use in Clinical Variant Interpretation. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 847-864.	1.2	1
99	Identification of clonal hematopoiesis mutations in solid tumor patients undergoing unpaired commercial next-generation sequencing assays.. <i>Journal of Clinical Oncology</i> , 2018, 36, 12068-12068.	0.8	1
100	Genetic Complexity of Mitral Valve Prolapse Revealed by Clinical and Genetic Evaluation of a Large Family. <i>Journal of Heart Valve Disease</i> , 2017, 26, 569-580.	0.5	1
101	Crowdsourcing to define the clinical actionability of incidental findings of genetic testing. <i>North Carolina Medical Journal</i> , 2013, 74, 501-2.	0.1	1
102	Selecting secondary findings to report: Creating a list that suits your study. , 2020, , 43-58.		0
103	Burden or benefit? Effects of providing education about and the option to request additional genomic findings from diagnostic exome sequencing: A randomized controlled trial. <i>Patient Education and Counseling</i> , 2021, 104, 2989-2998.	1.0	0
104	Testing and extending strategies for identifying genetic disease-related encounters in pediatric patients. <i>Genetics in Medicine</i> , 2022, 24, 831-838.	1.1	0