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List of Publications by Year
in descending order

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		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	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in Medicine, 2013, 15, 565-574.	2.4	2,186
2	ClinGen â€" The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	27.0	1,016
3	ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747.	2.4	794
4	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
5	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
6	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
7	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
8	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
9	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
10	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
11	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
12	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
13	An informatics approach to analyzing the incidentalome. Genetics in Medicine, 2013, 15, 36-44.	2.4	148
14	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
15	Prenatal exome sequencing in anomalous fetuses: new opportunities and challenges. Genetics in Medicine, 2017, 19, 1207-1216.	2.4	135
16	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
17	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
18	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

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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. Genetics in Medicine, 2013, 15, 860-867.	2.4	99
20	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
21	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
22	Germline Analysis from Tumorâ€“Germline Sequencing Dyads to Identify Clinically Actionable Secondary Findings. Clinical Cancer Research, 2016, 22, 4087-4094.	7.0	75
23	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
24	Incidental Findings with Genomic Testing: Implications for Genetic Counseling Practice. Current Genetic Medicine Reports, 2015, 3, 166-176.	1.9	68
25	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
26	We screen newborns, donâ€™t we?: realizing the promise of public health genomics. Genetics in Medicine, 2013, 15, 332-334.	2.4	64
27	Genomic Sequencing for Newborn Screening: Results of the NC NEXUS Project. American Journal of Human Genetics, 2020, 107, 596-611.	6.2	63
28	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
29	Increasing the diagnostic yield of exome sequencing by copy number variant analysis. PLoS ONE, 2018, 13, e0209185.	2.5	58
30	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
31	The promise and peril of genomic screening in the general population. Genetics in Medicine, 2016, 18, 593-599.	2.4	53
32	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
33	An Age-Based Framework for Evaluating Genome-Scale Sequencing Results in Newborn Screening. Journal of Pediatrics, 2019, 209, 68-76.	1.8	50
34	An approach to integrating exome sequencing for fetal structural anomalies into clinical practice. Genetics in Medicine, 2020, 22, 954-961.	2.4	49
35	Supporting Parental Decisions About Genomic Sequencing for Newborn Screening: The NC NEXUS Decision Aid. Pediatrics, 2016, 137, S16-S23.	2.1	45
36	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

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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	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
39	Navigating the nuances of clinical sequence variant interpretation in Mendelian disease. Genetics in Medicine, 2018, 20, 918-926.	2.4	40
40	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
41	Finding the Rare Pathogenic Variants in a Human Genome. JAMA - Journal of the American Medical Association, 2017, 317, 1904.	7.4	38
42	Parentsâ€™ perceptions of personal utility of exome sequencing results. Genetics in Medicine, 2020, 22, 752-757.	2.4	37
43	Whole Exome Sequencing Identifies Truncating Variants in Nuclear Envelope Genes in Patients With Cardiovascular Disease. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	34
44	Navigating the researchâ€“clinical interface in genomic medicine: analysis from the CSER Consortium. Genetics in Medicine, 2018, 20, 545-553.	2.4	34
45	Comparative analysis of functional assay evidence use by ClinGen Variant Curation Expert Panels. Genome Medicine, 2019, 11, 77.	8.2	34
46	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
47	Common recurrent microduplication syndromes: Diagnosis and management in clinical practice. American Journal of Medical Genetics, Part A, 2010, 152A, 1066-1078.	1.2	32
48	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
49	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
50	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
51	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
52	Generating a taxonomy for genetic conditions relevant to reproductive planning. American Journal of Medical Genetics, Part A, 2016, 170, 565-573.	1.2	25
53	hCALCRL mutation causes autosomal recessive nonimmune hydrops fetalis with lymphatic dysplasia. Journal of Experimental Medicine, 2018, 215, 2339-2353.	8.5	25
54	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

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55	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
56	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
57	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
58	Genome-Scale Sequencing in Clinical Care. JAMA - Journal of the American Medical Association, 2014, 312, 1865.	7.4	20
59	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
60	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
61	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
62	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
63	The progression of the ClinGen gene clinical validity classification over time. Human Mutation, 2018, 39, 1494-1504.	2.5	16
64	Lessons learned about harmonizing survey measures for the CSER consortium. Journal of Clinical and Translational Science, 2020, 4, 537-546.	0.6	16
65	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
66	Parental Views on Newborn Next Generation Sequencing: Implications for Decision Support. Maternal and Child Health Journal, 2020, 24, 856-864.	1.5	15
67	ClinGen and ClinVar â€“ Enabling Genomics in Precision Medicine. Human Mutation, 2018, 39, 1473-1475.	2.5	14
68	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
69	Exploring the importance of case-level clinical information for variant interpretation. Genetics in Medicine, 2017, 19, 3-5.	2.4	12
70	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
71	Too much of a good thing? Overdiagnosis, or overestimating risk in preventive genomic screening. Personalized Medicine, 2018, 15, 343-346.	1.5	11
72	Factors influencing NCGENES research participants- requests for non-medically actionable secondary findings. Genetics in Medicine, 2019, 21, 1092-1099.	2.4	11

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73	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
74	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
75	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. Cell Genomics, 2022, 2, 100131.	6.5	11
76	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
77	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
78	Values clarification and parental decision making about newborn genomic sequencing.. Health Psychology, 2020, 39, 335-344.	1.6	9
79	Engaging community stakeholders in research on best practices for clinical genomic sequencing. Personalized Medicine, 2020, 17, 435-444.	1.5	9
80	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
81	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
82	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
83	Genetic screening: birthright or earned with age?. Expert Review of Molecular Diagnostics, 2017, 17, 735-738.	3.1	7
84	Selective serotonin reuptake inhibitors ameliorate MEGF10 myopathy. Human Molecular Genetics, 2019, 28, 2365-2377.	2.9	7
85	Look Before You Leap. Obstetrics and Gynecology, 2015, 125, 1299-1305.	2.4	6
86	A missing link in the bench-to-bedside paradigm: engaging regulatory stakeholders in clinical genomics research. Genome Medicine, 2016, 8, 95.	8.2	6
87	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
88	FDA oversight of NSIGHT genomic research: the need for an integrated systems approach to regulation. Npj Genomic Medicine, 2019, 4, 32.	3.8	6
89	Assessing the implications of positive genomic screening results. Personalized Medicine, 2020, 17, 101-109.	1.5	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. Trials, 2021, 22, 395.	1.6	5

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91	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
92	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
93	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
94	Pre-capture multiplexing provides additional power to detect copy number variation in exome sequencing. BMC Bioinformatics, 2021, 22, 374.	2.6	3
95	Loss of De Novo DNA Methylation Causes Expansion of the Mouse Hematopoietic Stem Cell Pool. Blood, 2010, 116, 835-835.	1.4	3
96	Noninvasive prenatal exome sequencing diagnostic utility limited by sequencing depth and fetal fraction. Prenatal Diagnosis, 2021, , .	2.3	2
97	Response to Lindor et al.. Genetics in Medicine, 2013, 15, 409-410.	2.4	1
98	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
99	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
100	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
101	Crowdsourcing to define the clinical actionability of incidental findings of genetic testing. North Carolina Medical Journal, 2013, 74, 501-2.	0.2	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. Patient Education and Counseling, 2021, 104, 2989-2998.	2.2	0
104	Testing and extending strategies for identifying genetic diseaseâ€“related encounters in pediatric patients. Genetics in Medicine, 2022, 24, 831-838.	2.4	0