Erin R Riggs

List of Publications by Year
in descending order

25 papers	2,722 citations	471509 17 h-index	552781 26 g-index
32 all docs	32 docs citations	32 times ranked	5739 citing authors

#	Article	IF	Citations
1	Utilizing ClinGen geneâ€disease validity and dosage sensitivity curations to inform variant classification. Human Mutation, 2022, 43, 1031-1040.	2.5	20
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	OP029: Genetic updates returned by GenomeConnect, the ClinGen patient registry: A pilot study of participant experience. Genetics in Medicine, 2022, 24, S357-S358.	2.4	0
4	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
5	Clinical validity assessment of genes frequently tested on intellectual disability/autism sequencing panels. Genetics in Medicine, 2022, 24, 1899-1908.	2.4	9
6	Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics). Genetics in Medicine, 2021, 23, 1356-1365.	2.4	17
7	Utilizing population-based genomic data to expedite the curation of genes and genomic regions for the ClinGen â€œdosage sensitivity unlikelyâ€•classification. Molecular Genetics and Metabolism, 2021, 132, S228-S229.	1.1	0
8	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. Genetics in Medicine, 2021, 23, 1738-1745.	2.4	7
9	The Monarch Initiative in 2019: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2020, 48, D704-D715.	14.5	178
10	Variant interpretation is a component of clinical practice among genetic counselors in multiple specialties. Genetics in Medicine, 2020, 22, 785-792.	2.4	14
11	Response to Maya et al.. Genetics in Medicine, 2020, 22, 1278-1279.	2.4	4
12	Development of a consent resource for genomic data sharing in the clinical setting. Genetics in Medicine, 2019, 21, 81-88.	2.4	20
13	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
14	Incorporating Social Media into your Support Tool Box: Points to Consider from Geneticsâ€Based Communities. Journal of Genetic Counseling, 2018, 27, 470-480.	1.6	33
15	Points to consider for sharing variant-level information from clinical genetic testing with ClinVar. Journal of Physical Education and Sports Management, 2018, 4, a002345.	1.2	23
16	The ClinGen Epilepsy Gene Curation Expert Panelâ€”Bridging the divide between clinical domain knowledge and formal gene curation criteria. Human Mutation, 2018, 39, 1476-1484.	2.5	33
17	ClinGen's GenomeConnect registry enables patientâ€centered data sharing. Human Mutation, 2018, 39, 1668-1676.	2.5	25
18	Copy number variant discrepancy resolution using the ClinGen dosage sensitivity map results in updated clinical interpretations in ClinVar. Human Mutation, 2018, 39, 1650-1659.	2.5	23

#	Article	IF	Citations
19	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
20	GenomeConnect: Matchmaking Between Patients, Clinical Laboratories, and Researchers to Improve Genomic Knowledge. Human Mutation, 2015, 36, 974-978.	2.5	56
21	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
22	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
23	Genomic Variation: Lessons Learned from Whole-Genome CNV Analysis. Current Genetic Medicine Reports, 2014, 2, 146-150.	1.9	16
24	The Laboratoryâ€Clinician Team: A Professional Call to Action to Improve Communication and Collaboration for Optimal Patient Care in Chromosomal Microarray Testing. Journal of Genetic Counseling, 2012, 21, 631-637.	1.6	12
25	Phenotypic information in genomic variant databases enhances clinical care and research: The international standards for cytogenomic arrays consortium experience. Human Mutation, 2012, 33, 787-796.	2.5	49