Erin R Riggs

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5576983/publications.pdf

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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	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
2	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
3	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
4	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
5	GenomeConnect: Matchmaking Between Patients, Clinical Laboratories, and Researchers to Improve Genomic Knowledge. Human Mutation, 2015, 36, 974-978.	2.5	56
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	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
8	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
9	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
10	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
11	ClinGen's GenomeConnect registry enables patientâ€centered data sharing. Human Mutation, 2018, 39, 1668-1676.	2.5	25
12	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
13	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
14	Development of a consent resource for genomic data sharing in the clinical setting. Genetics in Medicine, 2019, 21, 81-88.	2.4	20
15	Utilizing ClinGen geneâ€disease validity and dosage sensitivity curations to inform variant classification. Human Mutation, 2022, 43, 1031-1040.	2.5	20
16	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
17	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
18	Genomic Variation: Lessons Learned from Whole-Genome CNV Analysis. Current Genetic Medicine Reports, 2014, 2, 146-150.	1.9	16

#	Article	IF	Citations
19	Variant interpretation is a component of clinical practice among genetic counselors in multiple specialties. Genetics in Medicine, 2020, 22, 785-792.	2.4	14
20	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
21	Clinical validity assessment of genes frequently tested on intellectual disability/autism sequencing panels. Genetics in Medicine, 2022, 24, 1899-1908.	2.4	9
22	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. Genetics in Medicine, 2021, 23, 1738-1745.	2.4	7
23	Response to Maya et al.. Genetics in Medicine, 2020, 22, 1278-1279.	2.4	4
24	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
25	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