Shannon Rego

List of Publications by Year
in descending order

24 papers	1,203 citations	840776 11 h-index	610901 24 g-index
24 all docs	24 docs citations	24 times ranked	2536 citing authors

#	Article	IF	Citations
1	Preference for secondary findings in prenatal and pediatric exome sequencing. Prenatal Diagnosis, 2022, 42, 753-761.	2.3	11
2	Integration of stakeholder engagement from development to dissemination in genomic medicine research: Approaches and outcomes from the CSER Consortium. Genetics in Medicine, 2022, 24, 1108-1119.	2.4	5
3	Predicting genes from phenotypes using human phenotype ontology (HPO) terms. Human Genetics, 2022, 141, 1749-1760.	3.8	1
4	Perspectives and preferences regarding genomic secondary findings in underrepresented prenatal and pediatric populations: A mixed-methods approach. Genetics in Medicine, 2022, 24, 1206-1216.	2.4	8
5	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
6	A review and definition of â€˜usual careâ€™ in genetic counseling trials to standardize use in research. Journal of Genetic Counseling, 2021, 30, 42-50.	1.6	12
7	Genomic Sequencing Results Disclosure in Diverse and Medically Underserved Populations: Themes, Challenges, and Strategies from the CSER Consortium. Journal of Personalized Medicine, 2021, 11, 202.	2.5	6
8	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	2.5	16
9	A novel truncating variant in ring finger protein 113A (RNF113A) confirms the association of this gene with Xâ€linked trichothiodystrophy. American Journal of Medical Genetics, Part A, 2020, 182, 513-520.	1.2	12
10	The expanding spectrum of NFIB â€associated phenotypes in a diverse patient populationâ€”A report of two new patients. American Journal of Medical Genetics, Part A, 2020, 182, 2959-2963.	1.2	3
11	iPSC Modeling of RBM20-Deficient DCM Identifies Upregulation of RBM20 as a Therapeutic Strategy. Cell Reports, 2020, 32, 108117.	6.4	40
12	Candidate variants in TUB are associated with familial tremor. PLoS Genetics, 2020, 16, e1009010.	3.5	3
13	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
14	Informed Consent in the Genomics Era. Cold Spring Harbor Perspectives in Medicine, 2020, 10, a036582.	6.2	16
15	Developmental and epileptic encephalopathy in two siblings with a novel, homozygous missense variant in SCN1B. American Journal of Medical Genetics, Part A, 2019, 179, 2190-2195.	1.2	10
16	Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students. Journal of Genetic Counseling, 2019, 28, 466-476.	1.6	10
17	Longitudinal multi-omics of hostâ€“microbe dynamics in prediabetes. Nature, 2019, 569, 663-671.	27.8	391
18	A longitudinal big data approach for precision health. Nature Medicine, 2019, 25, 792-804.	30.7	329

#	Article	IF	Citations
19	Much ado about nothing: A qualitative study of the experiences of an averageâ€risk population receiving results of exome sequencing. Journal of Genetic Counseling, 2019, 28, 428-437.	1.6	15
20	Integrative Personal Omics Profiles during Periods of Weight Gain and Loss. Cell Systems, 2018, 6, 157-170.e8.	6.2	183
21	High-frequency actionable pathogenic exome variants in an average-risk cohort. Journal of Physical Education and Sports Management, 2018, 4, a003178.	1.2	23
22	Evidenceâ€based assessments of clinical actionability in the context of secondary findings: Updates from ClinGen's Actionability Working Group. Human Mutation, 2018, 39, 1677-1685.	2.5	34
23	Association of AHSG with alopecia and mental retardation (APMR) syndrome. Human Genetics, 2017, 136, 287-296.	3.8	14
24	A Case Report of Hypoglycemia and Hypogammaglobulinemia: DAVID Syndrome in a Patient With a Novel NFKB2 Mutation. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2127-2130.	3.6	26