

# Shannon Rego

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8095999/publications.pdf>

Version: 2024-02-01

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	Longitudinal multi-omics of host-microbe dynamics in prediabetes. <i>Nature</i> , 2019, 569, 663-671.	27.8	391
2	A longitudinal big data approach for precision health. <i>Nature Medicine</i> , 2019, 25, 792-804.	30.7	329
3	Integrative Personal Omics Profiles during Periods of Weight Gain and Loss. <i>Cell Systems</i> , 2018, 6, 157-170.e8.	6.2	183
4	iPSC Modeling of RBM20-Deficient DCM Identifies Upregulation of RBM20 as a Therapeutic Strategy. <i>Cell Reports</i> , 2020, 32, 108117.	6.4	40
5	Evidence-based assessments of clinical actionability in the context of secondary findings: Updates from ClinGen's Actionability Working Group. <i>Human Mutation</i> , 2018, 39, 1677-1685.	2.5	34
6	A Case Report of Hypoglycemia and Hypogammaglobulinemia: DAVID Syndrome in a Patient With a Novel <i>NFKB2</i> Mutation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 2127-2130.	3.6	26
7	High-frequency actionable pathogenic exome variants in an average-risk cohort. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003178.	1.2	23
8	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020, 22, 1215-1226.	2.4	22
9	Informed Consent in the Genomics Era. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2020, 10, a036582.	6.2	16
10	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. <i>Human Mutation</i> , 2021, 42, 862-876.	2.5	16
11	Much ado about nothing: A qualitative study of the experiences of an average-risk population receiving results of exome sequencing. <i>Journal of Genetic Counseling</i> , 2019, 28, 428-437.	1.6	15
12	Association of AHSG with alopecia and mental retardation (APMR) syndrome. <i>Human Genetics</i> , 2017, 136, 287-296.	3.8	14
13	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 750-758.	6.2	13
14	A novel truncating variant in ring finger protein 113A ( <i>RNF113A</i> ) confirms the association of this gene with X-linked trichothiodystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 513-520.	1.2	12
15	A review and definition of "usual care" in genetic counseling trials to standardize use in research. <i>Journal of Genetic Counseling</i> , 2021, 30, 42-50.	1.6	12
16	Preference for secondary findings in prenatal and pediatric exome sequencing. <i>Prenatal Diagnosis</i> , 2022, 42, 753-761.	2.3	11
17	Developmental and epileptic encephalopathy in two siblings with a novel, homozygous missense variant in <i>SCN1B</i> . <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2190-2195.	1.2	10
18	Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students. <i>Journal of Genetic Counseling</i> , 2019, 28, 466-476.	1.6	10

#	ARTICLE	IF	CITATIONS
19	Perspectives and preferences regarding genomic secondary findings in underrepresented prenatal and pediatric populations: A mixed-methods approach. <i>Genetics in Medicine</i> , 2022, 24, 1206-1216.	2.4	8
20	Genomic Sequencing Results Disclosure in Diverse and Medically Underserved Populations: Themes, Challenges, and Strategies from the CSER Consortium. <i>Journal of Personalized Medicine</i> , 2021, 11, 202.	2.5	6
21	Integration of stakeholder engagement from development to dissemination in genomic medicine research: Approaches and outcomes from the CSER Consortium. <i>Genetics in Medicine</i> , 2022, 24, 1108-1119.	2.4	5
22	The expanding spectrum of NFIB-associated phenotypes in a diverse patient population—A report of two new patients. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2959-2963.	1.2	3
23	Candidate variants in TUB are associated with familial tremor. <i>PLoS Genetics</i> , 2020, 16, e1009010.	3.5	3
24	Predicting genes from phenotypes using human phenotype ontology (HPO) terms. <i>Human Genetics</i> , 2022, 141, 1749-1760.	3.8	1