

# Monica A Giovanni

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/11920535/publications.pdf>

Version: 2024-02-01

14  
papers

1,920  
citations

840776

11  
h-index

1058476

14  
g-index

15  
all docs

15  
docs citations

15  
times ranked

4970  
citing authors

#	ARTICLE	IF	CITATIONS
1	Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. <i>Science</i> , 2016, 354, .	12.6	464
2	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. <i>American Journal of Human Genetics</i> , 2017, 100, 895-906.	6.2	403
3	Genetic identification of familial hypercholesterolemia within a single U.S. health care system. <i>Science</i> , 2016, 354, .	12.6	349
4	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 4667-4672.	7.1	193
5	Exome Sequencing-Based Screening for <i>BRCA1/2</i> Expected Pathogenic Variants Among Adult Biobank Participants. <i>JAMA Network Open</i> , 2018, 1, e182140.	5.9	163
6	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort. <i>American Journal of Human Genetics</i> , 2018, 103, 328-337.	6.2	130
7	Health-Care Referrals from Direct-to-Consumer Genetic Testing. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 817-819.	0.7	53
8	COVID-19 outcomes and the human genome. <i>Genetics in Medicine</i> , 2020, 22, 1175-1177.	2.4	49
9	DNA-based screening and population health: a points to consider statement for programs and sponsoring organizations from the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2021, 23, 989-995.	2.4	43
10	Comparing Electronic Health Record Portals to Obtain Patient-Entered Family Health History in Primary Care. <i>Journal of General Internal Medicine</i> , 2013, 28, 1558-1564.	2.6	35
11	The Application of Computer-Based Tools in Obtaining the Genetic Family History. <i>Current Protocols in Human Genetics</i> , 2010, 66, Unit 9.21.	3.5	20
12	Obtaining a Genetic Family History Using Computer-Based Tools. <i>Current Protocols in Human Genetics</i> , 2019, 100, e72.	3.5	7
13	Bringing monogenic disease screening to the clinic. <i>Nature Medicine</i> , 2020, 26, 1172-1174.	30.7	6
14	Genome-first findings require precision phenotyping. <i>Genetics in Medicine</i> , 2018, 20, 1510-1511.	2.4	5