

# Deborah Mascalzoni

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9171197/publications.pdf>

Version: 2024-02-01

50  
papers

2,920  
citations

279701  
23  
h-index

197736  
49  
g-index

55  
all docs

55  
docs citations

55  
times ranked

5471  
citing authors

#	ARTICLE	IF	CITATIONS
1	A catalog of genetic loci associated with kidney function from analyses of a million individuals. <i>Nature Genetics</i> , 2019, 51, 957-972.	9.4	549
2	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. <i>Nature Genetics</i> , 2019, 51, 1459-1474.	9.4	251
3	From patients to partners: participant-centric initiatives in biomedical research. <i>Nature Reviews Genetics</i> , 2012, 13, 371-376.	7.7	250
4	Dynamic Consent: a potential solution to some of the challenges of modern biomedical research. <i>BMC Medical Ethics</i> , 2017, 18, 4.	1.0	223
5	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. <i>Nature Communications</i> , 2018, 9, 4455.	5.8	181
6	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. <i>Nature Communications</i> , 2019, 10, 4130.	5.8	133
7	Recommendations for Improving the Quality of Rare Disease Registries. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 1644.	1.2	116
8	International Charter of principles for sharing bio-specimens and data. <i>European Journal of Human Genetics</i> , 2015, 23, 721-728.	1.4	112
9	Large-scale genome-wide analysis identifies genetic variants associated with cardiac structure and function. <i>Journal of Clinical Investigation</i> , 2017, 127, 1798-1812.	3.9	106
10	Informed Consent in the Genomics Era. <i>PLoS Medicine</i> , 2008, 5, e192.	3.9	81
11	Global Public Perceptions of Genomic Data Sharing: What Shapes the Willingness to Donate DNA and Health Data?. <i>American Journal of Human Genetics</i> , 2020, 107, 743-752.	2.6	76
12	“You should at least ask”. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. <i>European Journal of Human Genetics</i> , 2016, 24, 1403-1408.	1.4	70
13	The GDPR and the research exemption: considerations on the necessary safeguards for research biobanks. <i>European Journal of Human Genetics</i> , 2019, 27, 1159-1167.	1.4	70
14	The Cooperative Health Research in South Tyrol (CHRIS) study: rationale, objectives, and preliminary results. <i>Journal of Translational Medicine</i> , 2015, 13, 348.	1.8	63
15	The genetic study of three population microisolates in South Tyrol (MICROS): study design and epidemiological perspectives. <i>BMC Medical Genetics</i> , 2007, 8, 29.	2.1	56
16	Improving the informed consent process in international collaborative rare disease research: effective consent for effective research. <i>European Journal of Human Genetics</i> , 2016, 24, 1248-1254.	1.4	47
17	The challenges of the expanded availability of genomic information: an agenda-setting paper. <i>Journal of Community Genetics</i> , 2018, 9, 103-116.	0.5	45
18	Meeting Patients’ Right to the Correct Diagnosis: Ongoing International Initiatives on Undiagnosed Rare Diseases and Ethical and Social Issues. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 2072.	1.2	40

#	ARTICLE	IF	CITATIONS
19	Demonstrating trustworthiness when collecting and sharing genomic data: public views across 22 countries. <i>Genome Medicine</i> , 2021, 13, 92.	3.6	39
20	Collaboration to Understand Complex Diseases. <i>Hypertension</i> , 2016, 67, 681-687.	1.3	36
21	The case for open science: rare diseases. <i>JAMIA Open</i> , 2020, 3, 472-486.	1.0	33
22	Comparison of participant information and informed consent forms of five European studies in genetic isolated populations. <i>European Journal of Human Genetics</i> , 2010, 18, 296-302.	1.4	31
23	Are Requirements to Deposit Data in Research Repositories Compatible With the European Union's General Data Protection Regulation?. <i>Annals of Internal Medicine</i> , 2019, 170, 332.	2.0	27
24	Rare disease research: Breaking the privacy barrier. <i>Applied &amp; Translational Genomics</i> , 2014, 3, 23-29.	2.1	26
25	Feedback of Individual Genetic Results to Research Participants: Is It Feasible in Europe?. <i>Biopreservation and Biobanking</i> , 2016, 14, 241-248.	0.5	24
26	Practical Barriers and Ethical Challenges in Genetic Data Sharing. <i>International Journal of Environmental Research and Public Health</i> , 2014, 11, 8383-8398.	1.2	20
27	Rare diseases and now rare data?. <i>Nature Reviews Genetics</i> , 2013, 14, 372-372.	7.7	19
28	Data in question: A survey of European biobank professionals on ethical, legal and societal challenges of biobank research. <i>PLoS ONE</i> , 2019, 14, e0221496.	1.1	18
29	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. <i>Human Mutation</i> , 2022, , .	1.1	18
30	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. <i>Communications Biology</i> , 2022, 5, .	2.0	17
31	The commercial genetic testing landscape for Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2021, 92, 107-111.	1.1	16
32	How to responsibly acknowledge research work in the era of big data and biobanks: ethical aspects of the Bioresource Research Impact Factor (BRIF). <i>Journal of Community Genetics</i> , 2018, 9, 169-176.	0.5	15
33	Compound heterozygous SZT2 mutations in two siblings with early-onset epilepsy, intellectual disability and macrocephaly. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 66, 81-85.	0.9	14
34	Preferences of the Public for Sharing Health Data: Discrete Choice Experiment. <i>JMIR Medical Informatics</i> , 2021, 9, e29614.	1.3	11
35	Consenting in Population Genomics as an Open Communication Process. <i>Studies in Ethics, Law, and Technology</i> , 2009, 3, .	0.3	10
36	The Role of Solidarity(-ies) in Rare Diseases Research. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 589-604.	0.8	9

#	ARTICLE	IF	CITATIONS
37	Return of genomic results does not motivate intent to participate in research for all: Perspectives across 22 countries. <i>Genetics in Medicine</i> , 2022, 24, 1120-1129.	1.1	8
38	Appropriate Safeguards and Article 89 of the GDPR: Considerations for Biobank, Databank and Genetic Research. <i>Frontiers in Genetics</i> , 2022, 13, 719317.	1.1	8
39	What ethical approaches are used by scientists when sharing health data? An interview study. <i>BMC Medical Ethics</i> , 2022, 23, 41.	1.0	7
40	Task matters - challenging the motor system allows distinguishing unaffected Parkin mutation carriers from mutation-free controls. <i>Parkinsonism and Related Disorders</i> , 2021, 86, 101-104.	1.1	6
41	Balancing scientific interests and the rights of participants in designing a recall by genotype study. <i>European Journal of Human Genetics</i> , 2021, 29, 1146-1157.	1.4	6
42	Return of research results (RoRR) to the healthy CHRIS cohort: designing a policy with the participants. <i>Journal of Community Genetics</i> , 2021, 12, 577-592.	0.5	6
43	Governance mechanisms for sharing of health data: An approach towards selecting attributes for complex discrete choice experiment studies. <i>Technology in Society</i> , 2021, 66, 101625.	4.8	6
44	Italian public's views on sharing genetic information and medical information: findings from the "Your DNA, Your Say" study. <i>Wellcome Open Research</i> , 2021, 6, 180.	0.9	4
45	Prospective epidemiological, molecular, and genetic characterization of a novel coronavirus disease in the Val Venosta/Vinschgau: the CHRIS COVID-19 study protocol. <i>Pathogens and Global Health</i> , 2022, 116, 128-136.	1.0	4
46	Legal issues in governing genetic biobanks: the Italian framework as a case study for the implications for citizen's health through public-private initiatives. <i>Journal of Community Genetics</i> , 2018, 9, 177-190.	0.5	3
47	Ethics Law and Governance of Biobanking: A Very Complex Normative Puzzle. <i>The International Library of Ethics, Law and Technology</i> , 2015, , 1-14.	0.2	3
48	Digital Biomarkers in Psychiatric Research: Data Protection Qualifications in a Complex Ecosystem. <i>Frontiers in Psychiatry</i> , 0, 13, .	1.3	3
49	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. <i>Biopreservation and Biobanking</i> , 2021, 19, 483-492.	0.5	1
50	Ethical, legal and social/societal implications (ELSI) of recall-by-genotype (RbG) and genotype-driven-research (GDR) approaches: a scoping review. <i>European Journal of Human Genetics</i> , 0, , .	1.4	1