

# Joann N Bodurtha

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3193721/publications.pdf>

Version: 2024-02-01

42  
papers

645  
citations

686830

13  
h-index

610482

24  
g-index

43  
all docs

43  
docs citations

43  
times ranked

1292  
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. <i>Genetics in Medicine</i> , 2017, 19, 412-420.	1.1	73
2	The 2019 US medical genetics workforce: a focus on clinical genetics. <i>Genetics in Medicine</i> , 2021, 23, 1458-1464.	1.1	70
3	Hereditary Renal Hypouricemia: A New Role for Allopurinol?. <i>American Journal of Medicine</i> , 2014, 127, e3-e4.	0.6	47
4	Genotype-phenotype correlation of congenital anomalies in multiple congenital anomalies hypotonia seizures syndrome (MCAHS1)/PIGN-related epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 77-86.	0.7	41
5	Genomics and Perinatal Care. <i>New England Journal of Medicine</i> , 2012, 366, 64-73.	13.9	40
6	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. <i>Human Genetics</i> , 2016, 135, 1399-1409.	1.8	40
7	Factors affecting quality of life in children and adolescents with hypermobile Ehlers-Danlos syndrome/hypermobility spectrum disorders. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 561-569.	0.7	39
8	“It’s a Little Different for Men” Sponsorship and Gender in Academic Medicine: a Qualitative Study. <i>Journal of General Internal Medicine</i> , 2021, 36, 1-8.	1.3	35
9	The KinFact Intervention – A Randomized Controlled Trial to Increase Family Communication About Cancer History. <i>Journal of Women's Health</i> , 2014, 23, 806-816.	1.5	28
10	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. <i>Science Advances</i> , 2021, 7, .	4.7	25
11	Physicians' Current Practices and Opportunities for DNA Banking of Dying Patients With Cancer. <i>Journal of Oncology Practice</i> , 2011, 7, 183-187.	2.5	20
12	Family Ties: The Role of Family Context in Family Health History Communication About Cancer. <i>Journal of Health Communication</i> , 2016, 21, 346-355.	1.2	19
13	Exploring Hereditary Cancer Among Dying Cancer Patients – A Cross-Sectional Study of Hereditary Risk and Perceived Awareness of DNA Testing and Banking. <i>Journal of Genetic Counseling</i> , 2010, 19, 497-525.	0.9	18
14	Identifying Gender Disparities and Barriers to Measuring the Status of Female Faculty: The Experience of a Large School of Medicine. <i>Journal of Women's Health</i> , 2019, 28, 1569-1575.	1.5	17
15	Uncertainty, hope, and coping efficacy among mothers of children with Duchenne/Becker muscular dystrophy. <i>Clinical Genetics</i> , 2019, 95, 677-683.	1.0	13
16	Pain and sleep quality in children with non-vascular Ehlers-Danlos syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1858-1864.	0.7	12
17	Patient-reported hereditary breast and ovarian cancer in a primary care practice. <i>Journal of Community Genetics</i> , 2014, 5, 179-183.	0.5	10
18	Regional models of genetic services in the United States. <i>Genetics in Medicine</i> , 2020, 22, 381-388.	1.1	10

#	ARTICLE	IF	CITATIONS
19	A Cost Analysis of Universal versus Targeted Cholesterol Screening in Pediatrics. <i>Journal of Pediatrics</i> , 2018, 196, 201-207.e2.	0.9	9
20	A structured genetics rotation for pediatric residents: an important educational opportunity. <i>Genetics in Medicine</i> , 2020, 22, 793-796.	1.1	9
21	Family health history and genetic services—the East Baltimore community stakeholder interview project. <i>Journal of Community Genetics</i> , 2019, 10, 219-227.	0.5	8
22	Trends in Unmet Need for Genetic Counseling Among Children With Special Health Care Needs, 2001–2010. <i>Academic Pediatrics</i> , 2015, 15, 544-550.	1.0	7
23	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2022, 141, 853-863.	1.8	7
24	Horizontal integration of OMIM across the medical school preclinical curriculum for early reinforcement of clinical genetics principles. <i>Genetics in Medicine</i> , 2015, 17, 158-163.	1.1	6
25	High-Risk Palliative Care Patients' Knowledge and Attitudes about Hereditary Cancer Testing and DNA Banking. <i>Journal of Genetic Counseling</i> , 2018, 27, 834-843.	0.9	6
26	Milestones for medical students completing a clinical genetics elective. <i>Genetics in Medicine</i> , 2017, 19, 236-239.	1.1	5
27	The Role of Palliative Medicine in Assessing Hereditary Cancer Risk. <i>American Journal of Hospice and Palliative Medicine</i> , 2018, 35, 1490-1497.	0.8	5
28	Lung cancer and family-centered patient concerns. <i>Supportive Care in Cancer</i> , 2018, 26, 3047-3053.	1.0	4
29	Incontinentia Pigmenti with Persistent Hypercalcemia: Case Report. <i>Pediatric Dermatology</i> , 2016, 33, e315-7.	0.5	3
30	Juvenile Idiopathic Arthritis Associated with Combined JPA–HHT Syndrome: A Novel Phenotype Associated with a Novel Variant in SMAD4. <i>Journal of Pediatric Genetics</i> , 2018, 07, 078-082.	0.3	3
31	Response to Biesecker et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1807-1808.	2.6	3
32	Five Pediatric Cancers – Update on Genetic Implications. <i>Current Pediatric Reviews</i> , 2017, 13, 42-48.	0.4	3
33	46 Chromosomes and Me. <i>Journal of Genetic Counseling</i> , 2012, 21, 173-174.	0.9	2
34	PhenX measures for phenotyping rare genetic conditions. <i>Genetics in Medicine</i> , 2017, 19, 834-837.	1.1	2
35	Lung cancer and family-centered concerns. <i>Supportive Care in Cancer</i> , 2020, 28, 497-505.	1.0	2
36	The Wills Eye Handbook of Ocular Genetics, 1st Edition. <i>Journal of Pediatric Genetics</i> , 2018, 07, 191-192.	0.3	1

#	ARTICLE	IF	CITATIONS
37	Post Graduate Education: Is Genomics Included on Board Certification Exams?. Medical Science Educator, 2020, 30, 1379-1382.	0.7	1
38	Gene Therapy. Pediatrics in Review, 2020, 41, 606-608.	0.2	1
39	Demographic and socioeconomic trends in DNA banking utilization in the USA. Journal of Community Genetics, 2021, 12, 593-602.	0.5	1
40	Response to Laissue et al.. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	0
41	Essential Mednotesâ€™ Comprehensive Medical Reference & Review for USMLE II and MCCQE I. Journal of Pediatric Genetics, 2017, 06, 128-128.	0.3	0
42	Successful Treatment of Refractory Orthostatic Intolerance (OI) With Droxidopa. Clinical Pediatrics, 0, , 000992282210926.	0.4	0