

# Erin Turbitt

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5578614/publications.pdf>

Version: 2024-02-01

61  
papers

1,116  
citations

687363

13  
h-index

434195

31  
g-index

61  
all docs

61  
docs citations

61  
times ranked

1819  
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of SOX3 as an XX male sex reversal gene in mice and humans. <i>Journal of Clinical Investigation</i> , 2011, 121, 328-341.	8.2	234
2	Personal utility in genomic testing: a systematic literature review. <i>European Journal of Human Genetics</i> , 2017, 25, 662-668.	2.8	122
3	Copy Number Variation in Patients with Disorders of Sex Development Due to 46,XY Gonadal Dysgenesis. <i>PLoS ONE</i> , 2011, 6, e17793.	2.5	116
4	Defining personal utility in genomics: A Delphi study. <i>Clinical Genetics</i> , 2017, 92, 290-297.	2.0	75
5	Web Platform vs In-Person Genetic Counselor for Return of Carrier Results From Exome Sequencing. <i>JAMA Internal Medicine</i> , 2018, 178, 338.	5.1	64
6	A multi-exon deletion within WWOX is associated with a 46,XY disorder of sex development. <i>European Journal of Human Genetics</i> , 2012, 20, 348-351.	2.8	48
7	Novel methylation markers of the dysexecutive-psychiatric phenotype in <i>FMR1</i> premutation women. <i>Neurology</i> , 2015, 84, 1631-1638.	1.1	32
8	Evaluation of Recipients of Positive and Negative Secondary Findings Evaluations in a Hybrid CLIA-Research Sequencing Pilot. <i>American Journal of Human Genetics</i> , 2018, 103, 358-366.	6.2	29
9	Advancing precision public health using human genomics: examples from the field and future research opportunities. <i>Genome Medicine</i> , 2021, 13, 97.	8.2	26
10	Preferences for results from genomic microarrays: comparing parents and health care providers. <i>Clinical Genetics</i> , 2015, 87, 21-29.	2.0	18
11	Australians'™ perspectives on support around use of personal genomic testing: Findings from the Genioz study. <i>European Journal of Medical Genetics</i> , 2019, 62, 290-299.	1.3	17
12	CITED2 mutations potentially cause idiopathic premature ovarian failure. <i>Translational Research</i> , 2012, 160, 384-388.	5.0	15
13	SRY mutation analysis by next generation (deep) sequencing in a cohort of chromosomal Disorders of Sex Development (DSD) patients with a mosaic karyotype. <i>BMC Medical Genetics</i> , 2012, 13, 108.	2.1	15
14	Australians'™ views on personal genomic testing: focus group findings from the Genioz study. <i>European Journal of Human Genetics</i> , 2018, 26, 1101-1112.	2.8	14
15	Australians'™ views and experience of personal genomic testing: survey findings from the Genioz study. <i>European Journal of Human Genetics</i> , 2019, 27, 711-720.	2.8	14
16	Availability of treatment drives decisions of genetic health professionals about disclosure of incidental findings. <i>European Journal of Human Genetics</i> , 2014, 22, 1225-1228.	2.8	13
17	Genetic counseling, genetic testing, and risk perceptions for breast and colorectal cancer: Results from the 2015 National Health Interview Survey. <i>Preventive Medicine</i> , 2019, 123, 12-19.	3.4	13
18	A randomized controlled study of a consent intervention for participating in an NIH genome sequencing study. <i>European Journal of Human Genetics</i> , 2018, 26, 622-630.	2.8	12

#	ARTICLE	IF	CITATIONS
19	Engagement and return of results preferences among a primarily African American genomic sequencing research cohort. <i>American Journal of Human Genetics</i> , 2021, 108, 894-902.	6.2	11
20	Key Informantsâ€™ Perspectives of Implementing Chromosomal Microarrays Into Clinical Practice in Australia. <i>Twin Research and Human Genetics</i> , 2013, 16, 833-839.	0.6	10
21	Evidence linking FMR1 mRNA and attentional demands of stepping and postural control in women with the premutation. <i>Neurobiology of Aging</i> , 2015, 36, 1400-1408.	3.1	10
22	Public or private care: where do specialists spend their time?. <i>Australian Health Review</i> , 2017, 41, 541.	1.1	10
23	Intentions to share exome sequencing results with family members: exploring spousal beliefs and attitudes. <i>European Journal of Human Genetics</i> , 2018, 26, 735-739.	2.8	10
24	Fragile X syndrome clinical trials: exploring parental decisionâ€™making. <i>Journal of Intellectual Disability Research</i> , 2019, 63, 926-935.	2.0	10
25	Multiple-locus variable-number tandem-repeat analysis for discriminating within<i>Salmonella enterica</i> serovar Typhimurium definitive types and investigation of outbreaks. <i>Epidemiology and Infection</i> , 2011, 139, 1050-1059.	2.1	9
26	Lower urgency paediatric injuries: Parent preferences for emergency department or general practitioner care. <i>EMA - Emergency Medicine Australasia</i> , 2016, 28, 564-568.	1.1	9
27	Children referred for specialty care: Parental perspectives and preferences on referral, followâ€™up and primary care. <i>Journal of Paediatrics and Child Health</i> , 2017, 53, 18-25.	0.8	9
28	Ethnic identity and engagement with genome sequencing research. <i>Genetics in Medicine</i> , 2019, 21, 1735-1743.	2.4	9
29	The Many Faces of MLPA. <i>Methods in Molecular Biology</i> , 2011, 687, 193-205.	0.9	8
30	Characterization of <i>Escherichia coli</i> O157:H7 in New Zealand using multiple-locus variable-number tandem-repeat analysis. <i>Epidemiology and Infection</i> , 2011, 139, 464-471.	2.1	8
31	Parent perspectives and reasons for lower urgency paediatric presentations to emergency departments. <i>EMA - Emergency Medicine Australasia</i> , 2016, 28, 211-215.	1.1	8
32	Psychosocial, attitudinal, and demographic correlates of cancer-related germline genetic testing in the 2017 Health Information National Trends Survey. <i>Journal of Community Genetics</i> , 2019, 10, 453-459.	1.2	8
33	Decision-making about genetic health information among family dyads: a systematic literature review. <i>Health Psychology Review</i> , 2022, 16, 412-429.	8.6	8
34	Use of a telenursing triage service by <sc>V</sc>ictorian parents attending the emergency department for their child's lower urgency condition. <i>EMA - Emergency Medicine Australasia</i> , 2015, 27, 558-562.	1.1	7
35	Regular source of primary care and emergency department use of children in <sc>V</sc>ictoria. <i>Journal of Paediatrics and Child Health</i> , 2016, 52, 303-307.	0.8	7
36	Feasibility of Coping Effectiveness Training for Caregivers of Children with Autism Spectrum Disorder: a Genetic Counseling Intervention. <i>Journal of Genetic Counseling</i> , 2018, 27, 252-262.	1.6	7

#	ARTICLE	IF	CITATIONS
37	Managing the need to tell: Triggers and strategic disclosure of thalassemia major in Singapore. American Journal of Medical Genetics, Part A, 2019, 179, 762-769.	1.2	7
38	General practitioner referrals to paediatric specialist outpatient clinics: referral goals and parental influence. Journal of Primary Health Care, 2018, 10, 76.	0.6	6
39	Early career investigators and precision public health. Lancet, The, 2019, 394, 382-383.	13.7	6
40	Uptake of Genetic Testing Among Patients with Cancer At Risk for Lynch Syndrome in the National Health Interview Survey. Cancer Prevention Research, 2021, 14, 927-932.	1.5	6
41	Adaptation of couples living with a high risk of breast/ovarian cancer and the association with risk-reducing surgery. Familial Cancer, 2018, 17, 485-493.	1.9	5
42	Perspectives From Early Career Investigators Who Are "Staying in the Game" of Precision Public Health Research. American Journal of Public Health, 2019, 109, 1186-1187.	2.7	5
43	General practitioner perspectives on referrals to paediatric public specialty clinics. Australian Family Physician, 2016, 45, 747-753.	0.5	5
44	Perspectives of Australian general practitioners on shared care for paediatric patients. Australian Journal of Primary Health, 2017, 23, 147.	0.9	4
45	A primer in genomics for social and behavioral investigators. Translational Behavioral Medicine, 2020, 10, 451-456.	2.4	4
46	Molecular phylogenetic analysis of New Zealand mosquito species. New Zealand Journal of Zoology, 2020, 47, 324-349.	1.1	4
47	Parent clinical trial priorities for fragile X syndrome: a best-worst scaling. European Journal of Human Genetics, 2021, 29, 1245-1251.	2.8	4
48	Motivators and barriers for paediatricians discharging patients. Australian Journal of Primary Health, 2017, 23, 284.	0.9	4
49	Paediatric emergency department referrals from primary care. Australian Health Review, 2016, 40, 691.	1.1	2
50	Paediatrician perceptions of patient referral and discharge. Australian Health Review, 2017, 41, 561.	1.1	2
51	Parental preferences for paediatric specialty follow-up care. Australian Health Review, 2017, 41, 401.	1.1	2
52	What proportion of paediatric specialist referrals originates from general practitioners?. Journal of Paediatrics and Child Health, 2018, 54, 183-187.	0.8	2
53	Perceptions of uncertainties about carrier results identified by exome sequencing in a randomized controlled trial. Translational Behavioral Medicine, 2020, 10, 441-450.	2.4	2
54	Preferences for and acceptability of receiving pharmacogenomic results by mail: A focus group study with a primarily African-American cohort. Journal of Genetic Counseling, 2021, 30, 1582-1590.	1.6	2

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55	Dyadic concordance and associations of beliefs with intentions to learn carrier results from genomic sequencing. <i>Journal of Behavioral Medicine</i> , 2021, 44, 860-866.	2.1	2
56	Parental decision making about clinical trial enrollment: A survey of parents of children with Fragile X syndrome.. <i>Health Psychology</i> , 2020, 39, 1070-1077.	1.6	2
57	Genetic testing decisions in non-western cultures: an opportunity for intergenerational decision making. <i>European Journal of Human Genetics</i> , 2022, 30, 391-391.	2.8	2
58	Using a Participatory Approach to Develop Research Priorities for Future Leaders in Cancer-Related Precision Public Health. <i>Frontiers in Genetics</i> , 0, 13, .	2.3	2
59	Judgment and Decision Making in Genome Sequencing. , 2019, , 57-73.		1
60	Considerations for Reporting Genome Results to Patients. <i>Journal of Paediatrics and Child Health</i> , 2013, 49, 82-82.	0.8	0
61	Enrolling Children in Clinical Trials for Genetic Neurodevelopmental Conditions: Ethics, Parental Decisions, and Children's Identities. <i>Ethics &amp; Human Research</i> , 2021, 43, 27-36.	0.9	0