

Christine Patch

List of Publications by Year in descending order

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Version: 2024-02-01

84
papers

3,129
citations

293460

24
h-index

214428

50
g-index

90
all docs

90
docs citations

90
times ranked

5582
citing authors

#	ARTICLE	IF	CITATIONS
1	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care – Preliminary Report. <i>New England Journal of Medicine</i> , 2021, 385, 1868-1880.	13.9	352
2	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	2.6	343
3	The 100,000 Genomes Project: bringing whole genome sequencing to the NHS. <i>BMJ: British Medical Journal</i> , 2018, 361, k1687.	2.4	312
4	Whole-genome sequencing reveals host factors underlying critical COVID-19. <i>Nature</i> , 2022, 607, 97-103.	13.7	174
5	Penetrance for copy number variants associated with schizophrenia. <i>Human Molecular Genetics</i> , 2010, 19, 3477-3481.	1.4	132
6	How can the evaluation of genetic tests be enhanced? Lessons learned from the ACCE framework and evaluating genetic tests in the United Kingdom. <i>Genetics in Medicine</i> , 2005, 7, 495-500.	1.1	115
7	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
8	Genetic counselling in the era of genomic medicine. <i>British Medical Bulletin</i> , 2018, 126, 27-36.	2.7	85
9	Use of Antihypertensive Medications and Mortality of Patients With Autosomal Dominant Polycystic Kidney Disease: A Population-Based Study. <i>American Journal of Kidney Diseases</i> , 2011, 57, 856-862.	2.1	81
10	European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. <i>European Journal of Human Genetics</i> , 2019, 27, 1763-1773.	1.4	78
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	Opportunistic genomic screening. Recommendations of the European Society of Human Genetics. <i>European Journal of Human Genetics</i> , 2021, 29, 365-377.	1.4	76
13	Genetic testing and common disorders in a public health framework: how to assess relevance and possibilities. <i>European Journal of Human Genetics</i> , 2011, 19, S6-S44.	1.4	75
14	Developing a policy for paediatric biobanks: principles for good practice. <i>European Journal of Human Genetics</i> , 2013, 21, 2-7.	1.4	63
15	Living a normal life in an extraordinary way: A systematic review investigating experiences of families of young people's transition into adulthood when affected by a genetic and chronic childhood condition. <i>International Journal of Nursing Studies</i> , 2016, 62, 44-59.	2.5	53
16	Genetic horoscopes: is it all in the genes? Points for regulatory control of direct-to-consumer genetic testing. <i>European Journal of Human Genetics</i> , 2009, 17, 857-859.	1.4	50
17	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 3-18.	2.6	46
18	Communication about genetic testing with breast and ovarian cancer patients: a scoping review. <i>European Journal of Human Genetics</i> , 2019, 27, 511-524.	1.4	39

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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	Factors affecting the clinical use of noninvasive prenatal testing: a mixed methods systematic review. <i>Prenatal Diagnosis</i> , 2013, 33, 532-541.	1.1	37
21	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	2.6	36
22	Developing an intervention to facilitate family communication about inherited genetic conditions, and training genetic counsellors in its delivery. <i>European Journal of Human Genetics</i> , 2016, 24, 794-802.	1.4	35
23	Towards equitable and trustworthy genomics research. <i>EBioMedicine</i> , 2022, 76, 103879.	2.7	34
24	The role of genetic counsellors in genomic healthcare in the United Kingdom: a statement by the Association of Genetic Nurses and Counsellors. <i>European Journal of Human Genetics</i> , 2017, 25, 659-661.	1.4	32
25	Return of individual research results from genomic research: A systematic review of stakeholder perspectives. <i>PLoS ONE</i> , 2021, 16, e0258646.	1.1	32
26	Building the Genetic Counsellor Profession in the United Kingdom: Two Decades of Growth and Development. <i>Journal of Genetic Counseling</i> , 2013, 22, 902-906.	0.9	31
27	Delivering genome sequencing in clinical practice: an interview study with healthcare professionals involved in the 100 000 Genomes Project. <i>BMJ Open</i> , 2019, 9, e029699.	0.8	30
28	Parents' motivations, concerns and understanding of genome sequencing: a qualitative interview study. <i>European Journal of Human Genetics</i> , 2020, 28, 874-884.	1.4	30
29	Genetic counselors and Genomic Counseling in the United Kingdom. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 79-83.	0.6	28
30	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017, 100, 334-342.	2.6	26
31	Using a community of practice to develop standards of practice and education for genetic counsellors in Europe. <i>Journal of Community Genetics</i> , 2010, 1, 169-173.	0.5	25
32	Position statement on opportunistic genomic screening from the Association of Genetic Nurses and Counsellors (UK and Ireland). <i>European Journal of Human Genetics</i> , 2014, 22, 955-956.	1.4	25
33	A Roadmap for Global Acceleration of Genomics Integration Across Nursing. <i>Journal of Nursing Scholarship</i> , 2020, 52, 329-338.	1.1	24
34	Protelomeric sequences are deleted in cases of short arm inverted duplication of chromosome 8. <i>American Journal of Medical Genetics Part A</i> , 1994, 50, 296-299.	2.4	21
35	Comparison of genotypic and phenotypic strategies for population screening in hemochromatosis: Assessment of anxiety, depression, and perception of health. <i>Genetics in Medicine</i> , 2005, 7, 550-556.	1.1	21
36	An ancestral 10-bp repeat expansion in <i>VWA1</i> causes recessive hereditary motor neuropathy. <i>Brain</i> , 2021, 144, 584-600.	3.7	20

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37	Genetic Counsellors: A Registration System to Assure Competence in Practice in the United Kingdom. <i>Public Health Genomics</i> , 2003, 6, 182-183.	0.6	16
38	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 397-405.	1.1	16
39	Factors affecting the uptake of screening: A randomised controlled non-inferiority trial comparing a genotypic and a phenotypic strategy for screening for haemochromatosis. <i>Journal of Hepatology</i> , 2005, 43, 149-155.	1.8	15
40	Opening the 'black box' of informed consent appointments for genome sequencing: a multisite observational study. <i>Genetics in Medicine</i> , 2019, 21, 1083-1091.	1.1	15
41	A Maturity Matrix for Nurse Leaders to Facilitate and Benchmark Progress in Genomic Healthcare Policy, Infrastructure, Education, and Delivery. <i>Journal of Nursing Scholarship</i> , 2020, 52, 583-592.	1.1	14
42	Genetics for Healthcare Professionals. , 0, , .		14
43	Psychosocial Aspects of DNA Testing for Hereditary Hemochromatosis in At-Risk Individuals: A Systematic Review. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 7-14.	0.3	13
44	Duplication of 10q24 locus: broadening the clinical and radiological spectrum. <i>European Journal of Human Genetics</i> , 2019, 27, 525-534.	1.4	13
45	Young people's understanding, attitudes and involvement in decision-making about genome sequencing for rare diseases: A qualitative study with participants in the UK 100,000 Genomes Project. <i>European Journal of Medical Genetics</i> , 2020, 63, 104043.	0.7	13
46	Point of View: An evolution from genetic counselling to genomic counselling. <i>European Journal of Medical Genetics</i> , 2019, 62, 288-289.	0.7	12
47	Web-based return of BRCA2 research results: one-year genetic counselling experience in Iceland. <i>European Journal of Human Genetics</i> , 2020, 28, 1656-1661.	1.4	12
48	A Delphi study to determine the European core curriculum for Master programmes in genetic counselling. <i>European Journal of Human Genetics</i> , 2013, 21, 1060-1066.	1.4	11
49	Training Genetic Counsellors to Deliver an Innovative Therapeutic Intervention: their Views and Experience of Facilitating Multi-€Family Discussion Groups. <i>Journal of Genetic Counseling</i> , 2017, 26, 199-214.	0.9	11
50	Study of the relationship between Black men, culture and prostate cancer beliefs. <i>Cogent Medicine</i> , 2018, 5, 1442636.	0.7	10
51	Development and mixed-methods evaluation of an online animation for young people about genome sequencing. <i>European Journal of Human Genetics</i> , 2020, 28, 896-906.	1.4	10
52	Participant experiences of genome sequencing for rare diseases in the 100,000 Genomes Project: a mixed methods study. <i>European Journal of Human Genetics</i> , 2022, 30, 604-610.	1.4	10
53	A systematic review of the clinical validity and clinical utility of DNA testing for hereditary haemochromatosis type 1 in at-risk populations. <i>Journal of Medical Genetics</i> , 2008, 45, 513-518.	1.5	9
54	Black and Minority Ethnic women's decision-making for risk reduction strategies after BRCA testing: Use of context and knowledge. <i>European Journal of Medical Genetics</i> , 2019, 62, 376-384.	0.7	9

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55	Should doctors have a legal duty to warn relatives of their genetic risks?. Lancet, The, 2019, 394, 2133-2135.	6.3	9
56	The family transition experience when living with childhood neuromuscular disease: A grounded theory study. Journal of Advanced Nursing, 2021, 77, 1921-1933.	1.5	8
57	Return of genomic results does not motivate intent to participate in research for all: Perspectives across 22 countries. Genetics in Medicine, 2022, 24, 1120-1129.	1.1	8
58	A decision analysis model for diagnostic strategies using DNA testing for hereditary haemochromatosis in at risk populations. QJM - Monthly Journal of the Association of Physicians, 2008, 101, 631-641.	0.2	7
59	Decision-making, attitudes, and understanding among patients and relatives invited to undergo genome sequencing in the 100,000 Genomes Project: A multisite survey study. Genetics in Medicine, 2022, 24, 61-74.	1.1	7
60	Haemochromatosis: the need for an agreed case definition. Journal of Hepatology, 2005, 43, 911.	1.8	6
61	Newborn Screening Policy in the United Kingdom & the United States: Two Different Communities of Practice. MCN the American Journal of Maternal Child Nursing, 2006, 31, 164-168.	0.3	6
62	Engaged genomic science produces better and fairer outcomes: an engagement framework for engaging and involving participants, patients and publics in genomics research and healthcare implementation. Wellcome Open Research, 2021, 6, 311.	0.9	6
63	Direct to consumer genetic tests. European Journal of Human Genetics, 2009, 17, 1111-1111.	1.4	5
64	The "new genetics" and nursing: what does it have to do with me?. Nursing Standard (Royal College of Nursing), 2011, 5, 40-44.	0.1	4
65	Development of a measure of genome sequencing knowledge for young people: The kids-KOGS. Clinical Genetics, 2019, 96, 411-417.	1.0	4
66	ESHG PPPC Comments on postmortem use of genetic data for research purposes. European Journal of Human Genetics, 2020, 28, 144-146.	1.4	3
67	Advances in the genetics of schizophrenia: will high-risk copy number variants be useful in clinical genetics or diagnostics?. F1000 Medicine Reports, 2009, 1, .	2.9	3
68	Co-designing models for the communication of genomic results for rare diseases: a comparative study in the Czech Republic and the United Kingdom. Journal of Community Genetics, 2022, 13, 313-327.	0.5	3
69	Interventions to improve patient access to and utilisation of genetic and genomic counselling services.. The Cochrane Library, 2015, 2015, .	1.5	2
70	Animation or leaflet: Does it make a difference when educating young people about genome sequencing?. Patient Education and Counseling, 2021, 104, 2522-2530.	1.0	2
71	Correspondence on "Ensuring best practice in genomics education and evaluation: Reporting item standards for education and its evaluation in genomics (RISE2 Genomics)" by Nisselle et al. Genetics in Medicine, 2022, 24, 962-963.	1.1	2
72	The new genetics: A research agenda for nurses?. Journal of Research in Nursing, 2002, 7, 161-163.	0.4	1

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73	Prevalence and Burden of Disease in Hemochromatosis: Estimates Derived from Routine Data. Australian Journal of Cancer Nursing, 2006, 8, 128-129.	0.8	1
74	Predictive or not predictive: understanding the mixed messages from the patient's <scp>DNA</scp> sequence. Journal of Clinical Nursing, 2015, 24, 3730-3735.	1.4	1
75	World congress on genetic counselling. European Journal of Medical Genetics, 2019, 62, 287.	0.7	1
76	Basic cancer genetics. , 2014, , 26-35.		1
77	“It didn’t mean anything” moving within a landscape of knowledge to interpret genetics and genetic test results within familial cancer concerns. New Genetics and Society, 2021, 40, 570-598.	0.7	1
78	Mixed-methods evaluation of the NHS Genomic Medicine Service for paediatric rare diseases: study protocol. NIHR Open Research, 0, 1, 23.	0.0	1
79	A comparison of a genetic screening strategy and a biochemical strategy for population screening for hemochromatosis. Australian Journal of Cancer Nursing, 2005, 7, 145-145.	0.8	0
80	Reply to Blendis. Gastroenterology, 2005, 129, 1800.	0.6	0
81	Identifying individuals who might benefit from genetic services and information. Nursing Standard (Royal College of Nursing (Great Britain): 1987), 2013, 28, 37-42.	0.1	0
82	Second World Congress on Genetic Counseling: An introduction to the special issue. Journal of Genetic Counseling, 2021, 30, 5-6.	0.9	0
83	Psychosocial Aspects of DNA Testing for Hereditary Hemochromatosis in At-Risk Individuals: A Systematic Review. Genetic Testing and Molecular Biomarkers, 2009, .	1.7	0
84	Genetic Counselling in Disorders of Low Penetrance. , 2010, , 371-379.		0