Christine Patch

List of Publications by Year
in descending order

84 papers	3,129 citations	257450 24 h-index	189892 50 g-index
90 all docs	90 docs citations	90 times ranked	5205 citing authors

#	Article	IF	Citations
1	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
2	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
3	The 100â€‰000 Genomes Project: bringing whole genome sequencing to the NHS. BMJ: British Medical Journal, 2018, 361, k1687.	2.3	312
4	Whole-genome sequencing reveals host factors underlying critical COVID-19. Nature, 2022, 607, 97-103.	27.8	174
5	Penetrance for copy number variants associated with schizophrenia. Human Molecular Genetics, 2010, 19, 3477-3481.	2.9	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. Genetics in Medicine, 2005, 7, 495-500.	2.4	115
7	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
8	Genetic counselling in the era of genomic medicine. British Medical Bulletin, 2018, 126, 27-36.	6.9	85
9	Use of Antihypertensive Medications and Mortality of Patients With Autosomal Dominant Polycystic Kidney Disease: A Population-Based Study. American Journal of Kidney Diseases, 2011, 57, 856-862.	1.9	81
10	European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. European Journal of Human Genetics, 2019, 27, 1763-1773.	2.8	78
11	Global Public Perceptions of Genomic Data Sharing: What Shapes the Willingness to Donate DNA and Health Data?. American Journal of Human Genetics, 2020, 107, 743-752.	6.2	76
12	Opportunistic genomic screening. Recommendations of the European Society of Human Genetics. European Journal of Human Genetics, 2021, 29, 365-377.	2.8	76
13	Genetic testing and common disorders in a public health framework: how to assess relevance and possibilities. European Journal of Human Genetics, 2011, 19, S6-S44.	2.8	75
14	Developing a policy for paediatric biobanks: principles for good practice. European Journal of Human Genetics, 2013, 21, 2-7.	2.8	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. International Journal of Nursing Studies, 2016, 62, 44-59.	5.6	53
16	Genetic horoscopes: is it all in the genes? Points for regulatory control of direct-to-consumer genetic testing. European Journal of Human Genetics, 2009, 17, 857-859.	2.8	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. American Journal of Human Genetics, 2018, 103, 3-18.	6.2	46
18	Communication about genetic testing with breast and ovarian cancer patients: a scoping review. European Journal of Human Genetics, 2019, 27, 511-524.	2.8	39

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19	Demonstrating trustworthiness when collecting and sharing genomic data: public views across 22 countries. Genome Medicine, 2021, 13, 92.	8.2	39
20	Factors affecting the clinical use of nonâ€invasive prenatal testing: a mixed methods systematic review. Prenatal Diagnosis, 2013, 33, 532-541.	2.3	37
21	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
22	Developing an intervention to facilitate family communication about inherited genetic conditions, and training genetic counsellors in its delivery. European Journal of Human Genetics, 2016, 24, 794-802.	2.8	35
23	Towards equitable and trustworthy genomics research. EBioMedicine, 2022, 76, 103879.	6.1	34
24	The role of genetic counsellors in genomic healthcare in the United Kingdom: a statement by the Association of Genetic Nurses and Counsellors. European Journal of Human Genetics, 2017, 25, 659-661.	2.8	32
25	Return of individual research results from genomic research: A systematic review of stakeholder perspectives. PLoS ONE, 2021, 16, e0258646.	2.5	32
26	Building the Genetic Counsellor Profession in the United Kingdom: Two Decades of Growth and Development. Journal of Genetic Counseling, 2013, 22, 902-906.	1.6	31
27	Delivering genome sequencing in clinical practice: an interview study with healthcare professionals involved in the 100 000 Genomes Project. BMJ Open, 2019, 9, e029699.	1.9	30
28	Parentsâ€™ motivations, concerns and understanding of genome sequencing: a qualitative interview study. European Journal of Human Genetics, 2020, 28, 874-884.	2.8	30
29	Genetic counselors and Genomic Counseling in the United Kingdom. Molecular Genetics & Genomic Medicine, 2015, 3, 79-83.	1.2	28
30	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
31	Using a community of practice to develop standards of practice and education for genetic counsellors in Europe. Journal of Community Genetics, 2010, 1, 169-173.	1.2	25
32	Position statement on opportunistic genomic screening from the Association of Genetic Nurses and Counsellors (UK and Ireland). European Journal of Human Genetics, 2014, 22, 955-956.	2.8	25
33	A Roadmap for Global Acceleration of Genomics Integration Across Nursing. Journal of Nursing Scholarship, 2020, 52, 329-338.	2.4	24
34	Protelomeric sequences are deleted in cases of short arm inverted duplication of chromosome 8. American Journal of Medical Genetics Part A, 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. Genetics in Medicine, 2005, 7, 550-556.	2.4	21
36	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20

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37	Genetic Counsellors: A Registration System to Assure Competence in Practice in the United Kingdom. Public Health Genomics, 2003, 6, 182-183.	1.0	16
38	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 397-405.	1.7	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. Journal of Hepatology, 2005, 43, 149-155.	3.7	15
40	Opening the ìblack boxì of informed consent appointments for genome sequencing: a multisite observational study. Genetics in Medicine, 2019, 21, 1083-1091.	2.4	15
41	A Maturity Matrix for Nurse Leaders to Facilitate and Benchmark Progress in Genomic Healthcare Policy, Infrastructure, Education, and Delivery. Journal of Nursing Scholarship, 2020, 52, 583-592.	2.4	14
42	Genetics for Healthcare Professionals. , 0, , .		14
43	Psychosocial Aspects of DNA Testing for Hereditary Hemochromatosis in At-Risk Individuals: A Systematic Review. Genetic Testing and Molecular Biomarkers, 2009, 13, 7-14.	0.7	13
44	Duplication of 10q24 locus: broadening the clinical and radiological spectrum. European Journal of Human Genetics, 2019, 27, 525-534.	2.8	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. European Journal of Medical Genetics, 2020, 63, 104043.	1.3	13
46	Point of View: An evolution from genetic counselling to genomic counselling. European Journal of Medical Genetics, 2019, 62, 288-289.	1.3	12
47	Web-based return of BRCA2 research results: one-year genetic counselling experience in Iceland. European Journal of Human Genetics, 2020, 28, 1656-1661.	2.8	12
48	A Delphi study to determine the European core curriculum for Master programmes in genetic counselling. European Journal of Human Genetics, 2013, 21, 1060-1066.	2.8	11
49	Training Genetic Counsellors to Deliver an Innovative Therapeutic Intervention: their Views and Experience of Facilitating Multiâ€Family Discussion Groups. Journal of Genetic Counseling, 2017, 26, 199-214.	1.6	11
50	Study of the relationship between Black men, culture and prostate cancer beliefs. Cogent Medicine, 2018, 5, 1442636.	0.7	10
51	Development and mixed-methods evaluation of an online animation for young people about genome sequencing. European Journal of Human Genetics, 2020, 28, 896-906.	2.8	10
52	Participant experiences of genome sequencing for rare diseases in the 100,000 Genomes Project: a mixed methods study. European Journal of Human Genetics, 2022, 30, 604-610.	2.8	10
53	A systematic review of the clinical validity and clinical utility of DNA testing for hereditary haemochromatosis type 1 in at-risk populations. Journal of Medical Genetics, 2008, 45, 513-518.	3.2	9
54	Black and Minority Ethnic women's decision-making for risk reduction strategies after BRCA testing: Use of context and knowledge. European Journal of Medical Genetics, 2019, 62, 376-384.	1.3	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.	13.7	9
56	The family transition experience when living with childhood neuromuscular disease: A grounded theory study. Journal of Advanced Nursing, 2021, 77, 1921-1933.	3.3	8
57	Return of genomic results does not motivate intentÂtoÂparticipate in research for all: Perspectives across 22 countries. Genetics in Medicine, 2022, 24, 1120-1129.	2.4	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.	0.5	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.	2.4	7
60	Haemochromatosis: the need for an agreed case definition. Journal of Hepatology, 2005, 43, 911.	3.7	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.	0.7	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.	1.8	6
63	Direct to consumer genetic tests. European Journal of Human Genetics, 2009, 17, 1111-1111.	2.8	5
64	The â€˜new geneticsâ€™ and nursing: what does it have to do with me?. Nursing Standard (Royal College of) Tj ETQq0 0 0 rgBT /Overlo	0.1	4
65	Development of a measure of genome sequencing knowledge for young people: The kidsâ€KOGS. Clinical Genetics, 2019, 96, 411-417.	2.0	4
66	ESHG PPPC Comments on postmortem use of genetic data for research purposes. European Journal of Human Genetics, 2020, 28, 144-146.	2.8	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.	1.2	3
69	Interventions to improve patient access to and utilisation of genetic and genomic counselling services.. The Cochrane Library, 2015, 2015, .	2.8	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.	2.2	2
71	Correspondence on â€œEnsuring best practice in genomics education and evaluation: Reporting item standards for education and its evaluation in genomics (RISE2 Genomics)â€•by Nisselle etÂal. Genetics in Medicine, 2022, 24, 962-963.	2.4	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.	1.6	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.	3.0	1
75	World congress on genetic counselling. European Journal of Medical Genetics, 2019, 62, 287.	1.3	1
76	Basic cancer genetics. , 2014, , 26-35.		1
77	â€œIt didnâ€™t mean anythingâ€•â€“ 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.	1.2	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.	1.6	0
80	Reply to Blendis. Gastroenterology, 2005, 129, 1800.	1.3	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.	1.6	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