Christine Patch

List of Publications by Citations

75 papers	1,666 citations	21 h-index	39 g-index
90 ext. papers	2,451 ext. citations	6.6 avg, IF	4.43 L-index

#	Paper	IF	Citations
75	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	11	235
74	The 100 000 Genomes Project: bringing whole genome sequencing to the NHS. *BMJ, The*, **2018**, 361, k1687	5.9	184
73	Penetrance for copy number variants associated with schizophrenia. *Human Molecular Genetics*, **2010**, 19, 3477-81	5.6	117
72	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	8.1	102
71	Phenotypic Characterization of Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. *Circulation*, **2017**, 136, 2022-2033	16.7	75
70	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, 856-62	7.4	67
69	Genetic testing and common disorders in a public health framework: how to assess relevance and possibilities. Background Document to the ESHG recommendations on genetic testing and common disorders. *European Journal of Human Genetics*, **2011**, 19 Suppl 1, S6-44	5.3	60
68	Developing a policy for paediatric biobanks: principles for good practice. *European Journal of Human Genetics*, **2013**, 21, 2-7	5.3	52
67	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, 857-9	5.3	42
66	Genetic counselling in the era of genomic medicine. *British Medical Bulletin*, **2018**, 126, 27-36	5.4	40
65	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
64	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	5.8	33
63	European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. *European Journal of Human Genetics*, **2019**, 27, 1763-1773	5.3	31
62	Factors affecting the clinical use of non-invasive prenatal testing: a mixed methods systematic review. *Prenatal Diagnosis*, **2013**, 33, 532-41	3.2	31
61	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	5.3	27
60	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	11	27
59	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	11	26

58	Genetic counselors and Genomic Counseling in the United Kingdom. *Molecular Genetics & Genomic Medicine*, **2015**, 3, 79-83	2.3	24
57	Using a community of practice to develop standards of practice and education for genetic counsellors in Europe. *Journal of Community Genetics*, **2010**, 1, 169-73	2.5	23
56	Building the genetic counsellor profession in the United Kingdom: two decades of growth and development. *Journal of Genetic Counseling*, **2013**, 22, 902-6	2.5	22
55	Communication about genetic testing with breast and ovarian cancer patients: a scoping review. *European Journal of Human Genetics*, **2019**, 27, 511-524	5.3	22
54	Position statement on opportunistic genomic screening from the Association of Genetic Nurses and Counsellors (UK and Ireland). *European Journal of Human Genetics*, **2014**, 22, 955-6	5.3	21
53	Opportunistic genomic screening. Recommendations of the European Society of Human Genetics. *European Journal of Human Genetics*, **2021**, 29, 365-377	5.3	21
52	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	5.3	20
51	Protelomeric sequences are deleted in cases of short arm inverted duplication of chromosome 8. *American Journal of Medical Genetics Part A*, **1994**, 50, 296-9		20
50	Comparison of genotypic and phenotypic strategies for population screening in hemochromatosis: assessment of anxiety, depression, and perception of health. *Genetics in Medicine*, **2005**, 7, 550-6	8.1	19
49	Applied Genetics in Healthcare		18
48	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
47	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	11	14
46	Parents' motivations, concerns and understanding of genome sequencing: a qualitative interview study. *European Journal of Human Genetics*, **2020**, 28, 874-884	5.3	13
45	A Roadmap for Global Acceleration of Genomics Integration Across Nursing. *Journal of Nursing Scholarship*, **2020**, 52, 329-338	3.6	12
44	Psychosocial aspects of DNA testing for hereditary hemochromatosis in at-risk individuals: a systematic review. *Genetic Testing and Molecular Biomarkers*, **2009**, 13, 7-14	1.6	12
43	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, 149-55	13.4	12
42	Delivering genome sequencing in clinical practice: an interview study with healthcare professionals involved in the 100 000 Genomes Project. *BMJ Open*, **2019**, 9, e029699	3	12
41	Genetic counsellors: a registration system to assure competence in practice in the United kingdom. *Public Health Genomics*, **2003**, 6, 182-3	1.9	11

40	Genetics for Healthcare Professionals		11
39	Demonstrating trustworthiness when collecting and sharing genomic data: public views across 22 countries. *Genome Medicine*, **2021**, 13, 92	14.4	11
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	3.5	10
37	A Delphi study to determine the European core curriculum for Master programmes in genetic counselling. *European Journal of Human Genetics*, **2013**, 21, 1060-6	5.3	10
36	Training Genetic Counsellors to Deliver an Innovative Therapeutic Intervention: their Views and Experience of Facilitating Multi-Family Discussion Groups. *Journal of Genetic Counseling*, **2017**, 26, 199-214	2.5	10
35	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. *Brain*, **2021**, 144, 584-600	11.2	10
34	Opening the "black box" of informed consent appointments for genome sequencing: a multisite observational study. *Genetics in Medicine*, **2019**, 21, 1083-1091	8.1	9
33	Whole genome sequencing reveals host factors underlying critical Covid-19.. *Nature*, **2022**,	50.4	8
32	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, 513-8	5.8	6
31	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	3.6	5
30	Direct to consumer genetic tests. *European Journal of Human Genetics*, **2009**, 17, 1111	5.3	5
29	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, 631-41	2.7	5
28	Haemochromatosis: the need for an agreed case definition. *Journal of Hepatology*, **2005**, 43, 911	13.4	5
27	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, 164-8	1	5
26	Should doctors have a legal duty to warn relatives of their genetic risks?. *Lancet, The*, **2019**, 394, 2133-2135	45	5
25	Study of the relationship between Black men, culture and prostate cancer beliefs. *Cogent Medicine*, **2018**, 5, 1442636	1.4	5
24	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	2.6	4
23	Development of a measure of genome sequencing knowledge for young people: The kids-KOGS. *Clinical Genetics*, **2019**, 96, 411-417	4	4

22	Duplication of 10q24 locus: broadening the clinical and radiological spectrum. *European Journal of Human Genetics*, **2019**, 27, 525-534	5.3	4
21	Web-based return of BRCA2 research results: one-year genetic counselling experience in Iceland. *European Journal of Human Genetics*, **2020**, 28, 1656-1661	5.3	3
20	Development and mixed-methods evaluation of an online animation for young people about genome sequencing. *European Journal of Human Genetics*, **2020**, 28, 896-906	5.3	3
19	Return of individual research results from genomic research: A systematic review of stakeholder perspectives. *PLoS ONE*, **2021**, 16, e0258646	3.7	3
18	Advances in the genetics of schizophrenia: will high-risk copy number variants be useful in clinical genetics or diagnostics?. *F1000 Medicine Reports*, **2009**, 1,		3
17	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	2.6	3
16	The 'new genetics' and nursing: what does it have to do with me?. *Nursing Standard (Royal College of Nursing (Great Britain): 1987)*, **2000**, 14, 42-6	1.1	2
15	Interventions to improve patient access to and utilisation of genetic and genomic counselling services. *The Cochrane Library*, **2015**, 2015,	5.2	1
14	Towards equitable and trustworthy genomics research.. *EBioMedicine*, **2022**, 76, 103879	8.8	1
13	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	4.8	1
12	ESHG PPPC Comments on postmortem use of genetic data for research purposes. *European Journal of Human Genetics*, **2020**, 28, 144-146	5.3	1
11	Predictive or not predictive: understanding the mixed messages from the patient's DNA sequence. *Journal of Clinical Nursing*, **2015**, 24, 3730-5	3.2	0
10	The family transition experience when living with childhood neuromuscular disease: A grounded theory study. *Journal of Advanced Nursing*, **2021**, 77, 1921-1933	3.1	0
9	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**, 1	2.5	0
8	Identifying individuals who might benefit from genetic services and information. *Nursing Standard (Royal College of Nursing (Great Britain): 1987)*, **2013**, 28, 37-42	1.1	
7	Prevalence and Burden of Disease in Hemochromatosis: Estimates Derived from Routine Data. *Australian Journal of Cancer Nursing*, **2006**, 8, 128-129	1.9	
6	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	1.9	
5	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*,1-29	1.9	

4	Psychosocial Aspects of DNA Testing for Hereditary Hemochromatosis in At-Risk Individuals: A Systematic Review. *Genetic Testing and Molecular Biomarkers*, **2009**, 090108090224061	
3	Genetic Counselling in Disorders of Low Penetrance **2010**, 371-379	
2	Animation or leaflet: Does it make a difference when educating young people about genome sequencing?. *Patient Education and Counseling*, **2021**, 104, 2522-2530	3.1
1	Second World Congress on Genetic Counseling: An introduction to the special issue. *Journal of Genetic Counseling*, **2021**, 30, 5-6	2.5