

# Patricia H Birch

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9015189/publications.pdf>

Version: 2024-02-01

49  
papers

2,418  
citations

279778

23  
h-index

214788

47  
g-index

51  
all docs

51  
docs citations

51  
times ranked

2840  
citing authors

#	ARTICLE	IF	CITATIONS
1	Type 1 neurofibromatosis: A descriptive analysis of the disorder in 1,728 patients. American Journal of Medical Genetics Part A, 1997, 70, 138-143.	2.4	381
2	Oligonucleotide Microarray Analysis of Genomic Imbalance in Children with Mental Retardation. American Journal of Human Genetics, 2006, 79, 500-513.	6.2	261
3	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. American Journal of Medical Genetics Part A, 2000, 95, 108-117.	2.4	214
4	â€œI want to know what's in Pandora's boxâ€ Comparing stakeholder perspectives on incidental findings in clinical whole genomic sequencing. American Journal of Medical Genetics, Part A, 2012, 158A, 2519-2525.	1.2	135
5	Skeletal abnormalities in neurofibromatosis type 1: Approaches to therapeutic options. American Journal of Medical Genetics, Part A, 2009, 149A, 2327-2338.	1.2	128
6	Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1. , 1999, 84, 413-419.		114
7	Growth in North American white children with neurofibromatosis 1 (NF1). Journal of Medical Genetics, 2000, 37, 933-938.	3.2	85
8	Subcutaneous neurofibromas are associated with mortality in neurofibromatosis 1: A cohort study of 703 patients. American Journal of Medical Genetics, Part A, 2005, 132A, 49-53.	1.2	73
9	Parental perceived value of a diagnosis for intellectual disability (ID): A qualitative comparison of families with and without a diagnosis for their child's ID. American Journal of Medical Genetics, Part A, 2009, 149A, 2393-2402.	1.2	69
10	The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review. Genetics in Medicine, 2020, 22, 1437-1449.	2.4	64
11	Genetics professionals' perspectives on reporting incidental findings from clinical genome-wide sequencing. American Journal of Medical Genetics, Part A, 2013, 161, 542-549.	1.2	55
12	Paternalism and the ACMG recommendations on genomic incidental findings: patients seen but not heard. Genetics in Medicine, 2013, 15, 751-752.	2.4	50
13	Assessment of algorithms for high throughput detection of genomic copy number variation in oligonucleotide microarray data. BMC Bioinformatics, 2007, 8, 368.	2.6	49
14	An Association Between Optic Glioma and Other Tumours of the Central Nervous System in Neurofibromatosis Type 1. Neuropediatrics, 1997, 28, 131-132.	0.6	42
15	Incidental Findings from Clinical Genome-wide Sequencing: A Review. Journal of Genetic Counseling, 2014, 23, 463-473.	1.6	41
16	Interactive e-counselling for genetics pre-test decisions: where are we now?. Clinical Genetics, 2015, 87, 209-217.	2.0	40
17	National Neurofibromatosis Foundation International Database. American Journal of Medical Genetics Part A, 1993, 45, 88-91.	2.4	39
18	Costs of caring for children with an intellectual developmental disorder. Disability and Health Journal, 2015, 8, 646-651.	2.8	39

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19	DECIDE: a Decision Support Tool to Facilitate Parentsâ€™ Choices Regarding Genomeâ€Wide Sequencing. <i>Journal of Genetic Counseling</i> , 2016, 25, 1298-1308.	1.6	36
20	The cost trajectory of the diagnostic care pathway for children with suspected genetic disorders. <i>Genetics in Medicine</i> , 2020, 22, 292-300.	2.4	35
21	Non-optic glioma in adults and children with neurofibromatosis 1. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 34.	2.7	31
22	Genital tract and plasma human immunodeficiency virus viral load throughout the menstrual cycle in women who are infected with ovulatory human immunodeficiency virus. <i>American Journal of Obstetrics and Gynecology</i> , 2003, 188, 122-128.	1.3	30
23	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. <i>BMC Genomics</i> , 2009, 10, 526.	2.8	30
24	Genome-wide sequencing as a first-tier screening test for short tandem repeat expansions. <i>Genome Medicine</i> , 2021, 13, 126.	8.2	27
25	Assessing an Interactive Online Tool to Support Parents' Genomic Testing Decisions. <i>Journal of Genetic Counseling</i> , 2019, 28, 10-17.	1.6	26
26	Growth charts for young children with neurofibromatosis 1 (NF1)., 2000, 92, 224-227.		25
27	The generalized bone phenotype in children with neurofibromatosis 1: A sibling matched caseâ€control study. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1654-1661.	1.2	25
28	Serial MRIs provide novel insight into natural history of optic pathway gliomas in patients with neurofibromatosis 1. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 62.	2.7	25
29	Linkage analysis of X-linked cleft palate and ankyloglossia in Manitoba Mennonite and British Columbia Native kindreds. <i>Human Genetics</i> , 1994, 94, 141-8.	3.8	21
30	Comparing the ability of OPTION12 and OPTION5 to assess shared decision-making in genetic counselling.. <i>Patient Education and Counseling</i> , 2016, 99, 1717-1723.	2.2	21
31	Outâ€ofâ€pocket and private pay in clinical genetic testing: A scoping review. <i>Clinical Genetics</i> , 2021, 100, 504-521.	2.0	21
32	Increased dental caries in people with neurofibromatosis 1. <i>Clinical Genetics</i> , 2007, 72, 524-527.	2.0	19
33	Carbamazepine poisoning in children. <i>Pediatric Emergency Care</i> , 1993, 9, 195-198.	0.9	17
34	Parentsâ€™ Perspectives on Supporting Their Decision Making in Genomeâ€Wide Sequencing. <i>Journal of Nursing Scholarship</i> , 2016, 48, 265-275.	2.4	17
35	MIA is a potential biomarker for tumour load in neurofibromatosis type 1. <i>BMC Medicine</i> , 2011, 9, 82.	5.5	15
36	Indigenous Peoples and genomics: Starting a conversation. <i>Journal of Genetic Counseling</i> , 2019, 28, 407-418.	1.6	15

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37	Assessing Shared Decision-Making Clinical Behaviors Among Genetic Counsellors. <i>Journal of Genetic Counseling</i> , 2019, 28, 40-49.	1.6	11
38	Development and pilot testing of a tool to calculate parental and societal costs of raising a child with intellectual disability. <i>Journal of Intellectual and Developmental Disability</i> , 2016, 41, 11-20.	1.6	10
39	After genomic testing results: <i>Parents' long-term views</i> . <i>Journal of Genetic Counseling</i> , 2022, 31, 82-95.	1.6	10
40	Autonomy and the patient's right "not to know" in clinical whole-genomic sequencing. <i>European Journal of Human Genetics</i> , 2014, 22, 6-6.	2.8	9
41	Quantitative associations of scalp and body subcutaneous neurofibromas with internal plexiform tumors in neurofibromatosis 1. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1518-1524.	1.2	9
42	Utilization and uptake of clinical genetics services in high-income countries: A scoping review. <i>Health Policy</i> , 2021, 125, 877-887.	3.0	9
43	Vertebral scalloping in neurofibromatosis type 1: a quantitative approach. <i>Canadian Journal of Surgery</i> , 2002, 45, 181-4.	1.2	8
44	Prevalence of dental caries in children with neurofibromatosis 1. <i>Clinical Oral Investigations</i> , 2010, 14, 479-480.	3.0	7
45	Utility and limitations of genetic disease databases in clinical genetics research: A neurofibromatosis 1 database example. <i>American Journal of Medical Genetics Part A</i> , 2004, 125C, 42-49.	2.4	6
46	A personalized genomic results e-booklet, co-designed and pilot-tested by families. <i>PEC Innovation</i> , 2022, 1, 100039.	0.8	6
47	Valuing gene testing in children with possible neurofibromatosis 1. <i>Clinical Genetics</i> , 2012, 82, 591-593.	2.0	5
48	The long-term impact of receiving incidental findings on parents undergoing genome-wide sequencing. <i>Journal of Genetic Counseling</i> , 2022, 31, 887-900.	1.6	5
49	Quality of Life in NF1. , 2012, , 93-103.		3