

# Bruce R Korf

## List of Publications by Year in descending order

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Version: 2024-02-01

113  
papers

13,595  
citations

38742

50  
h-index

23533

111  
g-index

123  
all docs

123  
docs citations

123  
times ranked

16343  
citing authors

#	ARTICLE	IF	CITATIONS
1	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. <i>Genetics in Medicine</i> , 2013, 15, 565-574.	2.4	2,186
2	Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. <i>Genetics in Medicine</i> , 2017, 19, 249-255.	2.4	1,398
3	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. <i>Pediatric Neurology</i> , 2013, 49, 243-254.	2.1	1,185
4	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. <i>Pediatric Neurology</i> , 2013, 49, 255-265.	2.1	693
5	Neurofibromatosis Type 1 Revisited. <i>Pediatrics</i> , 2009, 123, 124-133.	2.1	562
6	Neurofibromatosis type 1. <i>Nature Reviews Disease Primers</i> , 2017, 3, 17004.	30.5	498
7	Implementing genomic medicine in the clinic: the future is here. <i>Genetics in Medicine</i> , 2013, 15, 258-267.	2.4	472
8	Neurofibromatosis type 1. <i>Journal of the American Academy of Dermatology</i> , 2009, 61, 1-14.	1.2	443
9	Cardiovascular disease in neurofibromatosis 1: Report of the NF1 Cardiovascular Task Force. <i>Genetics in Medicine</i> , 2002, 4, 105-111.	2.4	330
10	Malignancy in Neurofibromatosis Type 1. <i>Oncologist</i> , 2000, 5, 477-485.	3.7	292
11	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. <i>Genetics in Medicine</i> , 2021, 23, 1506-1513.	2.4	290
12	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. <i>Nature Genetics</i> , 2014, 46, 182-187.	21.4	242
13	Identification of a Novel Genetic Locus for Familial Cardiac Myxomas and Carney Complex. <i>Circulation</i> , 1998, 98, 2560-2566.	1.6	209
14	Genetic Heterogeneity of Saethre-Chotzen Syndrome, Due to TWIST and FGFR Mutations. <i>American Journal of Human Genetics</i> , 1998, 62, 1370-1380.	6.2	202
15	American College of Medical Genetics Consensus Statement on Factor V Leiden Mutation Testing. <i>Genetics in Medicine</i> , 2001, 3, 139-148.	2.4	166
16	Clinical and Mutational Spectrum of Neurofibromatosis Type 1-like Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2009, 302, 2111.	7.4	160
17	Clinically relevant single gene or intragenic deletions encompassing critical neurodevelopmental genes in patients with developmental delay, mental retardation, and/or autism spectrum disorders. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2386-2396.	1.2	159
18	Exploring concordance and discordance for return of incidental findings from clinical sequencing. <i>Genetics in Medicine</i> , 2012, 14, 405-410.	2.4	149

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19	Global implementation of genomic medicine: We are not alone. <i>Science Translational Medicine</i> , 2015, 7, 290ps13.	12.4	146
20	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. <i>American Journal of Human Genetics</i> , 2018, 102, 69-87.	6.2	144
21	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. <i>Human Mutation</i> , 2015, 36, 1052-1063.	2.5	143
22	The case for strategic international alliances to harness nutritional genomics for public and personal health. <i>British Journal of Nutrition</i> , 2005, 94, 623-632.	2.3	137
23	Future Health Applications of Genomics. <i>American Journal of Preventive Medicine</i> , 2010, 38, 556-565.	3.0	136
24	Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2018, 20, 671-682.	2.4	128
25	Recommendations for the integration of genomics into clinical practice. <i>Genetics in Medicine</i> , 2016, 18, 1075-1084.	2.4	125
26	Framework for development of physician competencies in genomic medicine: report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics. <i>Genetics in Medicine</i> , 2014, 16, 804-809.	2.4	123
27	Pathophysiology of Neurofibromatosis Type 1. <i>Annals of Internal Medicine</i> , 2006, 144, 842.	3.9	121
28	Sirolimus for progressive neurofibromatosis type 1-associated plexiform neurofibromas: a Neurofibromatosis Clinical Trials Consortium phase II study. <i>Neuro-Oncology</i> , 2015, 17, 596-603.	1.2	118
29	New Approaches to Molecular Diagnosis. <i>JAMA - Journal of the American Medical Association</i> , 2013, 309, 1511.	7.4	116
30	NF1 Microdeletion Syndrome: Refined FISH Characterization of Sporadic and Familial Deletions with Locus-Specific Probes. <i>American Journal of Human Genetics</i> , 2000, 66, 100-109.	6.2	105
31	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. <i>American Journal of Human Genetics</i> , 2008, 82, 712-722.	6.2	95
32	Phase 2 randomized, flexible crossover, double-blinded, placebo-controlled trial of the farnesyltransferase inhibitor tipifarnib in children and young adults with neurofibromatosis type 1 and progressive plexiform neurofibromas. <i>Neuro-Oncology</i> , 2014, 16, 707-718.	1.2	93
33	Clinical spectrum of individuals with pathogenic <i>NF1</i> missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. <i>Human Mutation</i> , 2020, 41, 299-315.	2.5	80
34	The effect of everolimus on renal angiomyolipoma in patients with tuberous sclerosis complex being treated for subependymal giant cell astrocytoma: subgroup results from the randomized, placebo-controlled, Phase 3 trial EXIST-1. <i>Nephrology Dialysis Transplantation</i> , 2014, 29, 1203-1210.	0.7	79
35	Cutaneous neurofibromas. <i>Neurology</i> , 2018, 91, S5-S13.	1.1	79
36	Neurofibromatosis. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 111, 333-340.	1.8	77

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37	Optimizing biologically targeted clinical trials for neurofibromatosis. Expert Opinion on Investigational Drugs, 2013, 22, 443-462.	4.1	77
38	Randomized placebo-controlled study of lovastatin in children with neurofibromatosis type 1. Neurology, 2016, 87, 2575-2584.	1.1	76
39	Superficial Neurofibroma: A Lesion with Unique MRI Characteristics in Patients with Neurofibromatosis Type 1. American Journal of Roentgenology, 2005, 184, 962-968.	2.2	75
40	Consensus Recommendations to Accelerate Clinical Trials for Neurofibromatosis Type 2. Clinical Cancer Research, 2009, 15, 5032-5039.	7.0	74
41	Sirolimus for nonâ€progressive NF1â€associated plexiform neurofibromas: An NF clinical trials consortium phase II study. Pediatric Blood and Cancer, 2014, 61, 982-986.	1.5	73
42	Phase I trial and pharmacokinetic study of sorafenib in children with neurofibromatosis type I and plexiform neurofibromas. Pediatric Blood and Cancer, 2013, 60, 396-401.	1.5	67
43	Psychiatric Genetics: A Survey of Psychiatrists' Knowledge, Opinions, and Practice Patterns. Journal of Clinical Psychiatry, 2005, 66, 821-830.	2.2	67
44	Consistent cytogenetic aberrations in hepatoblastoma: A common pathway of genetic alterations in embryonal liver and skeletal muscle malignancies?. Genes Chromosomes and Cancer, 1991, 3, 37-43.	2.8	62
45	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
46	Genetic Testing in Cardiovascular Disease. Journal of the American College of Cardiology, 2007, 50, 727-737.	2.8	59
47	Review Article : Clinical Features and Pathobiology of Neurofibromatosis 1. Journal of Child Neurology, 2002, 17, 573-577.	1.4	57
48	Effectiveness of sequencing connexin 26 (GJB2) in cases of familial or sporadic childhood deafness referred for molecular diagnostic testing. Genetics in Medicine, 2002, 4, 279-288.	2.4	56
49	How to know when physicians are ready for genomic medicine. Science Translational Medicine, 2015, 7, 287fs19.	12.4	54
50	NF106: A Neurofibromatosis Clinical Trials Consortium Phase II Trial of the MEK Inhibitor Mirdametinib (PD-0325901) in Adolescents and Adults With NF1-Related Plexiform Neurofibromas. Journal of Clinical Oncology, 2021, 39, 797-806.	1.6	54
51	Stage IV neuroblastoma in infants. Long-term survival. Cancer, 1991, 67, 1493-1497.	4.1	50
52	An Update on Neurofibromatosis Type 1-Associated Gliomas. Cancers, 2020, 12, 114.	3.7	50
53	Patterns of Seizures Observed in Association with Neurofibromatosis 1. Epilepsia, 1993, 34, 616-620.	5.1	48
54	Genetic Heterogeneity of Familial Atrial Myxoma Syndromes (Carney Complex). American Journal of Cardiology, 1997, 79, 994-995.	1.6	48

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55	Cabozantinib for neurofibromatosis type 1-related plexiform neurofibromas: a phase 2 trial. <i>Nature Medicine</i> , 2021, 27, 165-173.	30.7	46
56	Diagnosis and management of neurofibromatosis type 1. <i>Current Neurology and Neuroscience Reports</i> , 2001, 1, 162-167.	4.2	45
57	A phase II study of continuous oral mTOR inhibitor everolimus for recurrent, radiographic-progressive neurofibromatosis type 1-associated pediatric low-grade glioma: a Neurofibromatosis Clinical Trials Consortium study. <i>Neuro-Oncology</i> , 2020, 22, 1527-1535.	1.2	45
58	The Genomic Medicine Integrative Research Framework: A Conceptual Framework for Conducting Genomic Medicine Research. <i>American Journal of Human Genetics</i> , 2019, 104, 1088-1096.	6.2	35
59	Clinical response to bevacizumab in schwannomatosis. <i>Neurology</i> , 2014, 83, 1986-1987.	1.1	33
60	A Proposed Approach for Implementing Genomics-Based Screening Programs for Healthy Adults. <i>NAM Perspectives</i> , 0, , .	2.9	30
61	The phakomatoses. <i>Clinics in Dermatology</i> , 2005, 23, 78-84.	1.6	25
62	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. <i>Genetics in Medicine</i> , 2017, 19, 377-385.	2.4	24
63	Reproducibility of cognitive endpoints in clinical trials: lessons from neurofibromatosis type 1. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 2555-2565.	3.7	24
64	Characterization and utilization of an international neurofibromatosis web-based, patient-entered registry: An observational study. <i>PLoS ONE</i> , 2017, 12, e0178639.	2.5	24
65	Mice with missense and nonsense <i>NF1</i> mutations display divergent phenotypes compared to <i>NF1</i> patients. <i>DMM Disease Models and Mechanisms</i> , 2016, 9, 759-67.	2.4	23
66	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. <i>Frontiers in Physiology</i> , 2019, 10, 388.	2.8	23
67	Breast cancer risk and germline genomic profiling of women with neurofibromatosis type 1 who developed breast cancer. <i>Genes Chromosomes and Cancer</i> , 2018, 57, 19-27.	2.8	22
68	Genome sequencing as a first-line diagnostic test for hospitalized infants. <i>Genetics in Medicine</i> , 2022, 24, 851-861.	2.4	22
69	Report of the Banbury Summit Meeting on the evolving role of the medical geneticist, February 12-14, 2006. <i>Genetics in Medicine</i> , 2008, 10, 502-507.	2.4	19
70	Genetics and genomics education: The next generation. <i>Genetics in Medicine</i> , 2011, 13, 201-202.	2.4	19
71	A state-based approach to genomics for rare disease and population screening. <i>Genetics in Medicine</i> , 2021, 23, 777-781.	2.4	19
72	Case 13-2005. <i>New England Journal of Medicine</i> , 2005, 352, 1800-1808.	27.0	17

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73	Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics). <i>Genetics in Medicine</i> , 2021, 23, 1356-1365.	2.4	17
74	Mutation-Directed Therapeutics for Neurofibromatosis Type I. <i>Molecular Therapy - Nucleic Acids</i> , 2020, 20, 739-753.	5.1	16
75	Neurofibromin (<i>NF1</i> ) genetic variant structure-function analyses using a full-length mouse cDNA. <i>Human Mutation</i> , 2018, 39, 816-821.	2.5	15
76	Pitfalls and challenges in genetic test interpretation: An exploration of genetic professionals experience with interpretation of results. <i>Clinical Genetics</i> , 2021, 99, 638-649.	2.0	15
77	Genomic medicine: educational challenges. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2013, 1, 119-122.	1.2	14
78	Education and Training of Non-Genetics Providers on the Return of Genome Sequencing Results in a NICU Setting. <i>Journal of Personalized Medicine</i> , 2022, 12, 405.	2.5	13
79	Clinical trial design for cutaneous neurofibromas. <i>Neurology</i> , 2018, 91, S31-S37.	1.1	11
80	Affinity Purification of NF1 Proteinâ€“Protein Interactors Identifies Keratins and Neurofibromin Itself as Binding Partners. <i>Genes</i> , 2019, 10, 650.	2.4	11
81	Two novel cases further expand the phenotype of TOR1AIP1-associated nuclear envelopathies. <i>Human Genetics</i> , 2020, 139, 483-498.	3.8	11
82	Recruiting diversity where it exists: The Alabama Genomic Health Initiative. <i>Journal of Genetic Counseling</i> , 2020, 29, 471-478.	1.6	11
83	Comparison of family health history in surveys vs electronic health record data mapped to the observational medical outcomes partnership data model in the <i>All of Us</i> Research Program. <i>Journal of the American Medical Informatics Association: JAMIA</i> , 2021, 28, 695-703.	4.4	11
84	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. <i>Human Mutation</i> , 2021, 42, 3-7.	2.5	10
85	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€”expanding knowledge, enhancing research, and therapeutic discovery. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1915-1927.	1.2	10
86	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. <i>Genetics in Medicine</i> , 2021, 23, 280-288.	2.4	9
87	Visual outcomes following everolimus targeted therapy for neurofibromatosis type Iâ€“associated optic pathway gliomas in children. <i>Pediatric Blood and Cancer</i> , 2021, 68, e28833.	1.5	9
88	Integration of genomics into medical practice. <i>Discovery Medicine</i> , 2013, 16, 241-8.	0.5	9
89	The phakomatoses. <i>Neuroimaging Clinics of North America</i> , 2004, 14, 139-148.	1.0	7
90	Overview of Clinical Cytogenetics. <i>Current Protocols in Human Genetics</i> , 2016, 89, 8.1.1-8.1.13.	3.5	7

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91	Patterns of Disease Monitoring and Treatment Among Patients With Tuberous Sclerosis Complex-related Angiomyolipomas. <i>Urology</i> , 2017, 104, 110-114.	1.0	7
92	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. <i>Neurology</i> , 2019, 93, 510-514.	1.1	7
93	Targeted exon skipping of NF1 exon 17 as a therapeutic for neurofibromatosis type I. <i>Molecular Therapy - Nucleic Acids</i> , 2022, 28, 261-278.	5.1	7
94	Statins, bone, and neurofibromatosis type 1. <i>BMC Medicine</i> , 2008, 6, 22.	5.5	6
95	Spinal neurofibromatosis and phenotypic heterogeneity in NF1. <i>Clinical Genetics</i> , 2015, 87, 399-400.	2.0	6
96	Analysis of patient-specific <i>NF1</i> variants leads to functional insights for Ras signaling that can impact personalized medicine. <i>Human Mutation</i> , 2022, 43, 30-41.	2.5	6
97	Genetic and Genomic Competency in Medical Practice. <i>AMA Journal of Ethics</i> , 2012, 14, 622-626.	0.7	5
98	Multi-Omics Profiling for NF1 Target Discovery in Neurofibromin (NF1) Deficient Cells. <i>Proteomics</i> , 2019, 19, e1800334.	2.2	5
99	Genomic Privacy in the Information Age. <i>Clinical Chemistry</i> , 2013, 59, 1148-1150.	3.2	4
100	Pushing the envelope in genomics education. <i>Genetics in Medicine</i> , 2015, 17, 857-858.	2.4	4
101	Germline and Somatic <i>NF1</i> Alterations Are Linked to Increased HER2 Expression in Breast Cancer. <i>Cancer Prevention Research</i> , 2018, 11, 655-664.	1.5	4
102	Genetics training in the genomic era. <i>Current Opinion in Pediatrics</i> , 2005, 17, 747-750.	2.0	3
103	Restoration of Normal NF1 Function with Antisense Morpholino Treatment of Recurrent Pathogenic Patient-Specific Variant c.1466A>G; p.Y489C. <i>Journal of Personalized Medicine</i> , 2021, 11, 1320.	2.5	3
104	Integration of genetics into medical practice. <i>Growth Hormone and IGF Research</i> , 2004, 14, 146-149.	1.1	2
105	Overview of Genetic Diagnosis in Cancer. <i>Current Protocols in Human Genetics</i> , 2017, 93, 10.1.1-10.1.9.	3.5	2
106	Return of raw data in genomic testing and research: ownership, partnership, and risk-benefit. <i>Genetics in Medicine</i> , 2020, 22, 12-14.	2.4	2
107	Status and Recommendations for Incorporating Biomarkers for Cutaneous Neurofibromas Into Clinical Research. <i>Neurology</i> , 2021, 97, S42-S49.	1.1	2
108	What's new in Neurogenetics? Amish microcephaly. <i>European Journal of Paediatric Neurology</i> , 2003, 7, 393-394.	1.6	1

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109	An evaluation of selumetinib for the treatment of neurofibromatosis type 1-associated symptomatic, inoperable plexiform neurofibromas. Expert Review of Precision Medicine and Drug Development, 2021, 6, 239-246.	0.7	1
110	Basic genetics. Primary Care - Clinics in Office Practice, 2004, 31, 461-478.	1.6	0
111	Unusual presentation of hereditary leiomyomatosis mimicking neurofibromatosis. JAAD Case Reports, 2018, 4, 440-441.	0.8	0
112	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. Neurology: Genetics, 2020, 6, e476.	1.9	0
113	An interview on rare and genetic diseases with Dr Bruce Korf, Associate Dean for Genomic Medicine at the University of Alabama at Birmingham. Current Medical Research and Opinion, 2022, 38, 161-163.	1.9	0