Bruce R Korf

List of Publications by Citations

113 papers	10,266 citations	47 h-index	101 g-index
123 ext. papers	12,146 ext. citations	7.4 avg, IF	5.81 L-index

#	Paper	IF	Citations
113	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genetics in Medicine*, **2013**, 15, 565-74	8.1	1787
112	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. *Genetics in Medicine*, **2017**, 19, 249-255	8.1	1017
111	Tuberous sclerosis complex diagnostic criteria update: recommendations of the 2012 Iinternational Tuberous Sclerosis Complex Consensus Conference. *Pediatric Neurology*, **2013**, 49, 243-54	2.9	916
110	Tuberous sclerosis complex surveillance and management: recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. *Pediatric Neurology*, **2013**, 49, 255-65	2.9	553
109	Neurofibromatosis type 1 revisited. *Pediatrics*, **2009**, 123, 124-33	7.4	439
108	Implementing genomic medicine in the clinic: the future is here. *Genetics in Medicine*, **2013**, 15, 258-67	8.1	385
107	Neurofibromatosis type 1. *Journal of the American Academy of Dermatology*, **2009**, 61, 1-14; quiz 15-6	4.5	323
106	Neurofibromatosis type 1. *Nature Reviews Disease Primers*, **2017**, 3, 17004	51.1	299
105	Cardiovascular disease in neurofibromatosis 1: report of the NF1 Cardiovascular Task Force. *Genetics in Medicine*, **2002**, 4, 105-11	8.1	276
104	Malignancy in neurofibromatosis type 1. *Oncologist*, **2000**, 5, 477-85	5.7	259
103	Identification of a novel genetic locus for familial cardiac myxomas and Carney complex. *Circulation*, **1998**, 98, 2560-6	16.7	183
102	Genetic heterogeneity of Saethre-Chotzen syndrome, due to TWIST and FGFR mutations. *American Journal of Human Genetics*, **1998**, 62, 1370-80	11	182
101	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. *Nature Genetics*, **2014**, 46, 182-7	36.3	177
100	American College of Medical Genetics consensus statement on factor V Leiden mutation testing. *Genetics in Medicine*, **2001**, 3, 139-48	8.1	141
99	Exploring concordance and discordance for return of incidental findings from clinical sequencing. *Genetics in Medicine*, **2012**, 14, 405-10	8.1	136
98	Clinical and mutational spectrum of neurofibromatosis type 1-like syndrome. *JAMA - Journal of the American Medical Association*, **2009**, 302, 2111-8	27.4	126
97	Clinically relevant single gene or intragenic deletions encompassing critical neurodevelopmental genes in patients with developmental delay, mental retardation, and/or autism spectrum disorders. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2386-96	2.5	125

96	Future health applications of genomics: priorities for communication, behavioral, and social sciences research. *American Journal of Preventive Medicine*, **2010**, 38, 556-65	6.1	122
95	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotype-Phenotype Correlation. *Human Mutation*, **2015**, 36, 1052-63	4.7	112
94	Global implementation of genomic medicine: We are not alone. *Science Translational Medicine*, **2015** , 7, 290ps13	17.5	112
93	The case for strategic international alliances to harness nutritional genomics for public and personal health. *British Journal of Nutrition*, **2005**, 94, 623-32	3.6	112
92	Pathophysiology of neurofibromatosis type 1. *Annals of Internal Medicine*, **2006**, 144, 842-9	8	96
91	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. *Genetics in Medicine*, **2014**, 16, 804-9	8.1	95
90	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848. *American Journal of Human Genetics*, **2018**, 102, 69-87	11	93
89	NF1 microdeletion syndrome: refined FISH characterization of sporadic and familial deletions with locus-specific probes. *American Journal of Human Genetics*, **2000**, 66, 100-9	11	93
88	Recommendations for the integration of genomics into clinical practice. *Genetics in Medicine*, **2016**, 18, 1075-1084	8.1	92
87	Sirolimus for progressive neurofibromatosis type 1-associated plexiform neurofibromas: a neurofibromatosis Clinical Trials Consortium phase II study. *Neuro-Oncology*, **2015**, 17, 596-603	1	91
86	New approaches to molecular diagnosis. *JAMA - Journal of the American Medical Association*, **2013**, 309, 1511-21	27.4	91
85	Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. *American Journal of Human Genetics*, **2008**, 82, 712-22	11	84
84	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. *Neuro-Oncology*, **2014**, 16, 707-18	1	77
83	Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*, **2018**, 20, 671-682	8.1	74
82	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. *Nephrology Dialysis Transplantation*, **2014**, 29, 1203-10	4.3	67
81	Sirolimus for non-progressive NF1-associated plexiform neurofibromas: an NF clinical trials consortium phase II study. *Pediatric Blood and Cancer*, **2014**, 61, 982-6	3	65
80	Superficial neurofibroma: a lesion with unique MRI characteristics in patients with neurofibromatosis type 1. *American Journal of Roentgenology*, **2005**, 184, 962-8	5.4	62
79	Optimizing biologically targeted clinical trials for neurofibromatosis. *Expert Opinion on Investigational Drugs*, **2013**, 22, 443-62	5.9	61

78	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	3	61
77	Consensus recommendations to accelerate clinical trials for neurofibromatosis type 2. *Clinical Cancer Research*, **2009**, 15, 5032-5039	12.9	61
76	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	5	60
75	Psychiatric genetics: a survey of psychiatristsSknowledge, opinions, and practice patterns. *Journal of Clinical Psychiatry*, **2005**, 66, 821-30	4.6	56
74	Effectiveness of sequencing connexin 26 (GJB2) in cases of familial or sporadic childhood deafness referred for molecular diagnostic testing. *Genetics in Medicine*, **2002**, 4, 279-88	8.1	54
73	Randomized placebo-controlled study of lovastatin in children with neurofibromatosis type 1. *Neurology*, **2016**, 87, 2575-2584	6.5	53
72	Neurofibromatosis. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 111, 333-40	3	53
71	Genetic testing in cardiovascular disease. *Journal of the American College of Cardiology*, **2007**, 50, 727-37	15.1	51
70	Clinical features and pathobiology of neurofibromatosis 1. *Journal of Child Neurology*, **2002**, 17, 573-7; discussion 602-4, 646-51	2.5	50
69	How to know when physicians are ready for genomic medicine. *Science Translational Medicine*, **2015**, 7, 287fs19	17.5	48
68	Cutaneous neurofibromas: Current clinical and pathologic issues. *Neurology*, **2018**, 91, S5-S13	6.5	47
67	Clinical spectrum of individuals with pathogenic NF1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. *Human Mutation*, **2020**, 41, 299-315	4.7	47
66	Genetic heterogeneity of familial atrial myxoma syndromes (Carney complex). *American Journal of Cardiology*, **1997**, 79, 994-5	3	44
65	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. *Genetics in Medicine*, **2021**, 23, 1506-1513	8.1	43
64	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotype-phenotype correlation. *Genetics in Medicine*, **2019**, 21, 867-876	8.1	43
63	Patterns of seizures observed in association with neurofibromatosis 1. *Epilepsia*, **1993**, 34, 616-20	6.4	40
62	Stage IV neuroblastoma in infants. Long-term survival. *Cancer*, **1991**, 67, 1493-7	6.4	40
61	Diagnosis and management of neurofibromatosis type 1. *Current Neurology and Neuroscience Reports*, **2001**, 1, 162-7	6.6	38

60	An Update on Neurofibromatosis Type 1-Associated Gliomas. *Cancers*, **2020**, 12,	6.6	28
59	Clinical response to bevacizumab in schwannomatosis. *Neurology*, **2014**, 83, 1986-7	6.5	26
58	The Genomic Medicine Integrative Research Framework: A Conceptual Framework for Conducting Genomic Medicine Research. *American Journal of Human Genetics*, **2019**, 104, 1088-1096	11	24
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. *Neuro-Oncology*, **2020**, 22, 1527-1535	1	20
56	A Proposed Approach for Implementing Genomics-Based Screening Programs for Healthy Adults. *NAM Perspectives*,	2.8	19
55	Cabozantinib for neurofibromatosis type 1-related plexiform neurofibromas: a phase 2 trial. *Nature Medicine*, **2021**, 27, 165-173	50.5	19
54	The phakomatoses. *Clinics in Dermatology*, **2005**, 23, 78-84	3	18
53	Breast cancer risk and germline genomic profiling of women with neurofibromatosis type 1 who developed breast cancer. *Genes Chromosomes and Cancer*, **2018**, 57, 19-27	5	16
52	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	2.2	16
51	Mice with missense and nonsense NF1 mutations display divergent phenotypes compared with human neurofibromatosis type I. *DMM Disease Models and Mechanisms*, **2016**, 9, 759-67	4.1	16
50	Case records of the Massachusetts General Hospital. Case 13-2005. A 48-year-old man with weakness of the limbs and multiple tumors of spinal nerves. *New England Journal of Medicine*, **2005**, 352, 1800-8	59.2	15
49	Characterization and utilization of an international neurofibromatosis web-based, patient-entered registry: An observational study. *PLoS ONE*, **2017**, 12, e0178639	3.7	15
48	Genetics and genomics education: the next generation. *Genetics in Medicine*, **2011**, 13, 201-2	8.1	14
47	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. *Genetics in Medicine*, **2017**, 19, 377-385	8.1	13
46	Report of the Banbury Summit Meeting on the evolving role of the medical geneticist, February 12-14, 2006. *Genetics in Medicine*, **2008**, 10, 502-7	8.1	13
45	Neurofibromin (NF1) genetic variant structure-function analyses using a full-length mouse cDNA. *Human Mutation*, **2018**, 39, 816-821	4.7	12
44	Genomic medicine: educational challenges. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 119-22	2.3	12
43	Reproducibility of cognitive endpoints in clinical trials: lessons from neurofibromatosis type 1. *Annals of Clinical and Translational Neurology*, **2019**, 6, 2555-2565	5.3	11

42	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. *Frontiers in Physiology*, **2019**, 10, 388	4.6	9
41	Two novel cases further expand the phenotype of TOR1AIP1-associated nuclear envelopathies. *Human Genetics*, **2020**, 139, 483-498	6.3	9
40	Mutation-Directed Therapeutics for Neurofibromatosis Type I. *Molecular Therapy - Nucleic Acids*, **2020**, 20, 739-753	10.7	8
39	Clinical trial design for cutaneous neurofibromas. *Neurology*, **2018**, 91, S31-S37	6.5	7
38	Integration of genomics into medical practice. *Discovery Medicine*, **2013**, 16, 241-8	2.5	7
37	Affinity Purification of NF1 Protein-Protein Interactors Identifies Keratins and Neurofibromin Itself as Binding Partners. *Genes*, **2019**, 10,	4.2	6
36	The phakomatoses. *Neuroimaging Clinics of North America*, **2004**, 14, 139-48, vii	3	6
35	A state-based approach to genomics for rare disease and population screening. *Genetics in Medicine* , **2021**, 23, 777-781	8.1	6
34	Patterns of Disease Monitoring and Treatment Among Patients With Tuberous Sclerosis Complex-related Angiomyolipomas. *Urology*, **2017**, 104, 110-114	1.6	5
33	Spinal neurofibromatosis and phenotypic heterogeneity in NF1. *Clinical Genetics*, **2015**, 87, 399-400	4	5
32	Overview of Clinical Cytogenetics. *Current Protocols in Human Genetics*, **2016**, 89, 8.1.1-8.1.13	3.2	5
31	The All of Us Research Program: data quality, utility, and diversity		5
30	Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics). *Genetics in Medicine*, **2021**, 23, 1356-1365	8.1	5
29	Multi-Omics Profiling for NF1 Target Discovery in Neurofibromin (NF1) Deficient Cells. *Proteomics*, **2019**, 19, e1800334	4.8	4
28	Recruiting diversity where it exists: The Alabama Genomic Health Initiative. *Journal of Genetic Counseling*, **2020**, 29, 471-478	2.5	4
27	Germline and Somatic Alterations Are Linked to Increased HER2 Expression in Breast Cancer. *Cancer Prevention Research*, **2018**, 11, 655-664	3.2	4
26	Genetic and genomic competency in medical practice. *AMA Journal of Ethics*, **2012**, 14, 622-6	1.4	4
25	Genomic privacy in the information age. *Clinical Chemistry*, **2013**, 59, 1148-50	5.5	4

24	Child Neurology: Spastic paraparesis and dystonia with a novel mutation. *Neurology*, **2019**, 93, 510-514	6.5	4
23	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. *Human Mutation*, **2021**, 42, 3-7	4.7	4
22	Genetics training in the genomic era. *Current Opinion in Pediatrics*, **2005**, 17, 747-50	3.2	3
21	Visual outcomes following everolimus targeted therapy for neurofibromatosis type 1-associated optic pathway gliomas in children. *Pediatric Blood and Cancer*, **2021**, 68, e28833	3	3
20	Comparison of family health history in surveys vs electronic health record data mapped to the observational medical outcomes partnership data model in the All of Us Research Program. *Journal of the American Medical Informatics Association: JAMIA*, **2021**, 28, 695-703	8.6	3
19	Overview of Genetic Diagnosis in Cancer. *Current Protocols in Human Genetics*, **2017**, 93, 10.1.1-10.1.9	3.2	2
18	Pushing the envelope in genomics education. *Genetics in Medicine*, **2015**, 17, 857-8	8.1	2
17	Analysis of patient-specific NF1 variants leads to functional insights for Ras signaling that can impact personalized medicine. *Human Mutation*, **2021**,	4.7	2
16	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. *Genetics in Medicine*, **2021**, 23, 280-288	8.1	2
15	The seventh international RASopathies symposium: Pathways to a cure-expanding knowledge, enhancing research, and therapeutic discovery.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	2
14	The Neurofibromatoses **2011**, 128.1-128.14		1
13	What's new in neurogenetics? Amish microcephaly. *European Journal of Paediatric Neurology*, **2003**, 7, 393-4	3.8	1
12	Integration of genetics into medical practice. *Growth Hormone and IGF Research*, **2004**, 14 Suppl A, S146-9	2	1
11	Pitfalls and challenges in genetic test interpretation: An exploration of genetic professionals experience with interpretation of results. *Clinical Genetics*, **2021**, 99, 638-649	4	1
10	Return of raw data in genomic testing and research: ownership, partnership, and risk-benefit. *Genetics in Medicine*, **2020**, 22, 12-14	8.1	1
9	Targeted exon skipping of exon 17 as a therapeutic for neurofibromatosis type I.. *Molecular Therapy - Nucleic Acids*, **2022**, 28, 261-278	10.7	1
8	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	1.6	0
7	Status and Recommendations for Incorporating Biomarkers for Cutaneous Neurofibromas Into Clinical Research. *Neurology*, **2021**, 97, S42-S49	6.5	0

6	Unusual presentation of hereditary leiomyomatosis mimicking neurofibromatosis. *JAAD Case Reports*, **2018**, 4, 440-441	1.4
5	Carrier Screening **2006**, 238-267	
4	Basic genetics. *Primary Care - Clinics in Office Practice*, **2004**, 31, 461-78, vii	2.2
3	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**, 1-3	2.5
2	mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. *Neurology: Genetics*, **2020**, 6, e476	3.8
1	The Neurofibromatoses **2019**, 1823-1836	