

Leo B Waterston

List of Publications by Year in descending order

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Version: 2024-02-01

105
papers

5,274
citations

109311

35
h-index

98792

67
g-index

109
all docs

109
docs citations

109
times ranked

8648
citing authors

#	ARTICLE	IF	CITATIONS
1	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. Human Mutation, 2018, 39, 1517-1524.	2.5	511
2	Diagnostic Clinical Genome and Exome Sequencing. New England Journal of Medicine, 2014, 370, 2418-2425.	27.0	488
3	Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework. Genetics in Medicine, 2018, 20, 1054-1060.	2.4	366
4	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	8.2	312
5	Elevated basal serum tryptase identifies a multisystem disorder associated with increased TPSAB1 copy number. Nature Genetics, 2016, 48, 1564-1569.	21.4	279
6	The ACMG/AMP reputable source criteria for the interpretation of sequence variants. Genetics in Medicine, 2018, 20, 1687-1688.	2.4	152
7	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
8	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
9	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. American Journal of Human Genetics, 2018, 103, 319-327.	6.2	122
10	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. Genetics in Medicine, 2019, 21, 1189-1198.	2.4	115
11	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	28.9	113
12	Mosaic Disorders of the PI3K/PTEN/AKT/TSC/mTORC1 Signaling Pathway. Dermatologic Clinics, 2017, 35, 51-60.	1.7	111
13	Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study. Genetics in Medicine, 2019, 21, 1100-1110.	2.4	111
14	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines. Human Mutation, 2020, 41, 1734-1737.	2.5	105
15	Updated recommendation for the benign stand-alone ACMG/AMP criterion. Human Mutation, 2018, 39, 1525-1530.	2.5	102
16	A taxonomy of medical uncertainties in clinical genome sequencing. Genetics in Medicine, 2017, 19, 918-925.	2.4	91
17	A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation. Genetics in Medicine, 2016, 18, 1258-1268.	2.4	89
18	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. Current Protocols in Human Genetics, 2019, 103, e93.	3.5	88

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19	How do research participants perceive "uncertainty" in genome sequencing?. <i>Genetics in Medicine</i> , 2014, 16, 977-980.	2.4	71
20	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021, 108, 8-15.	6.2	71
21	A Point Mutation in PDGFRB Causes Autosomal-Dominant Penttinen Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 465-474.	6.2	64
22	Web Platform vs In-Person Genetic Counselor for Return of Carrier Results From Exome Sequencing. <i>JAMA Internal Medicine</i> , 2018, 178, 338.	5.1	64
23	Redefining the Etiologic Landscape of Cerebellar Malformations. <i>American Journal of Human Genetics</i> , 2019, 105, 606-615.	6.2	61
24	Participant use and communication of findings from exome sequencing: a mixed-methods study. <i>Genetics in Medicine</i> , 2016, 18, 577-583.	2.4	56
25	Pharmacodynamic Study of Miransertib in Individuals with Proteus Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 484-491.	6.2	56
26	Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. <i>Clinical Pharmacology and Therapeutics</i> , 2019, 105, 1338-1344.	4.7	56
27	Psychological outcomes related to exome and genome sequencing result disclosure: a meta-analysis of seven Clinical Sequencing Exploratory Research (CSER) Consortium studies. <i>Genetics in Medicine</i> , 2019, 21, 2781-2790.	2.4	55
28	Variant Classification Concordance using the ACMG-AMP Variant Interpretation Guidelines across Nine Genomic Implementation Research Studies. <i>American Journal of Human Genetics</i> , 2020, 107, 932-941.	6.2	51
29	The role of current affect, anticipated affect and spontaneous self-affirmation in decisions to receive self-threatening genetic risk information. <i>Cognition and Emotion</i> , 2015, 29, 1456-1465.	2.0	48
30	Area Deprivation Index and Rurality in Relation to Lung Cancer Prevalence and Mortality in a Rural State. <i>JNCI Cancer Spectrum</i> , 2020, 4, pkaa011.	2.9	48
31	Variant curation expert panel recommendations for RYR1 pathogenicity classifications in malignant hyperthermia susceptibility. <i>Genetics in Medicine</i> , 2021, 23, 1288-1295.	2.4	46
32	Assessing the reproducibility of exome copy number variations predictions. <i>Genome Medicine</i> , 2016, 8, 82.	8.2	44
33	Communicating Scientific Uncertainty About the COVID-19 Pandemic: Online Experimental Study of an Uncertainty-Normalizing Strategy. <i>Journal of Medical Internet Research</i> , 2021, 23, e27832.	4.3	44
34	Characterizing Participants in the ClinSeq Genome Sequencing Cohort as Early Adopters of a New Health Technology. <i>PLoS ONE</i> , 2015, 10, e0132690.	2.5	42
35	Advancing RAS/RASopathy therapies: An NCI-sponsored intramural and extramural collaboration for the study of RASopathies. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 866-876.	1.2	40
36	Nomenclature and definition in asymmetric regional body overgrowth. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1735-1738.	1.2	36

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37	Evidence-based assessments of clinical actionability in the context of secondary findings: Updates from ClinGen's Actionability Working Group. <i>Human Mutation</i> , 2018, 39, 1677-1685.	2.5	34
38	A six-attribute classification of genetic mosaicism. <i>Genetics in Medicine</i> , 2020, 22, 1743-1757.	2.4	34
39	Characterization of Courtesy Stigma Perceived by Parents of Overweight Children with Bardet-Biedl Syndrome. <i>PLoS ONE</i> , 2015, 10, e0140705.	2.5	33
40	Distinguishing Variant Pathogenicity From Genetic Diagnosis. <i>JAMA - Journal of the American Medical Association</i> , 2018, 320, 1929.	7.4	32
41	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	6.2	30
42	A Clinical Service to Support the Return of Secondary Genomic Findings in Human Research. <i>American Journal of Human Genetics</i> , 2016, 98, 435-441.	6.2	29
43	Evaluation of Recipients of Positive and Negative Secondary Findings Evaluations in a Hybrid CLIA-Research Sequencing Pilot. <i>American Journal of Human Genetics</i> , 2018, 103, 358-366.	6.2	29
44	Management of Secondary Genomic Findings. <i>American Journal of Human Genetics</i> , 2020, 107, 3-14.	6.2	29
45	Thrombosis risk factors in PIK3CA-related overgrowth spectrum and Proteus syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 571-581.	1.6	28
46	Clinical report: one year of treatment of Proteus syndrome with miransertib (ARQ 092). <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a004549.	1.2	27
47	Characterization of thrombosis in patients with Proteus syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2359-2365.	1.2	26
48	A common haplotype containing functional CACNA1H variants is frequently coinherited with increased TPSAB1 copy number. <i>Genetics in Medicine</i> , 2018, 20, 503-512.	2.4	25
49	Somatic <i>AKT1</i> mutations cause meningiomas colocalizing with a characteristic pattern of cranial hyperostosis. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2605-2610.	1.2	24
50	An approach to pediatric exome and genome sequencing. <i>Current Opinion in Pediatrics</i> , 2014, 26, 639-645.	2.0	23
51	Compound heterozygous alterations in intraflagellar transport protein <i>CLUAP1</i> in a child with a novel Joubert and oral-facial-digital overlap syndrome. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001321.	1.2	23
52	Lack of mutation-histopathology correlation in a patient with Proteus syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1422-1432.	1.2	22
53	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. <i>Genetics in Medicine</i> , 2018, 20, 855-866.	2.4	22
54	Family functioning mediates adaptation in caregivers of individuals with Rett syndrome. <i>Patient Education and Counseling</i> , 2016, 99, 1873-1879.	2.2	21

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55	Urine cell-free DNA is a biomarker for nephroblastomatosis or Wilms tumor in PIK3CA-related overgrowth spectrum (PROS). <i>Genetics in Medicine</i> , 2018, 20, 1077-1081.	2.4	21
56	Exome sequencing identifies novel mutations in C5orf42 in patients with Joubert syndrome with oral-facial-digital anomalies. <i>Human Genome Variation</i> , 2015, 2, 15045.	0.7	20
57	Outcomes of Counseling after Education about Carrier Results: A Randomized Controlled Trial. <i>American Journal of Human Genetics</i> , 2018, 102, 540-546.	6.2	18
58	TREML4 Promotes Inflammatory Programs in Human and Murine Macrophages and Alters Atherosclerosis Lesion Composition in the Apolipoprotein E Deficient Mouse. <i>Frontiers in Immunology</i> , 2020, 11, 397.	4.8	16
59	Exome sequencing identifies a mutation in OFD1 in a male with Joubert syndrome, orofacioidigital spectrum anomalies and complex polydactyly. <i>Human Genome Variation</i> , 2016, 3, 15069.	0.7	15
60	Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. <i>Genetics in Medicine</i> , 2017, 19, 357-361.	2.4	15
61	Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. <i>Genome Medicine</i> , 2018, 10, 99.	8.2	15
62	Pathogenetic insights from quantification of the cerebriform connective tissue nevus in Proteus syndrome. <i>Journal of the American Academy of Dermatology</i> , 2018, 78, 725-732.	1.2	14
63	DNA-based screening and personal health: a points to consider statement for individuals and health-care providers from the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2021, 23, 979-988.	2.4	14
64	Preliminary validation of a consumer-oriented colorectal cancer risk assessment tool compatible with the US Surgeon General's My Family Health Portrait. <i>Genetics in Medicine</i> , 2015, 17, 753-756.	2.4	13
65	Professional responsibilities regarding the provision, publication, and dissemination of patient phenotypes in the context of clinical genetic and genomic testing: points to consider—a statement of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2018, 20, 169-171.	2.4	13
66	Secondary findings in exome slices, virtual panels, and anticipatory sequencing. <i>Genetics in Medicine</i> , 2019, 21, 41-43.	2.4	13
67	Increased Burden of Rare Sequence Variants in GnRH-Associated Genes in Women With Hypothalamic Amenorrhea. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e1441-e1452.	3.6	13
68	Web-Based Platform vs Genetic Counselors in Educating Patients About Carrier Results From Exome Sequencing—Reply. <i>JAMA Internal Medicine</i> , 2018, 178, 999.	5.1	11
69	A mouse model of Proteus syndrome. <i>Human Molecular Genetics</i> , 2019, 28, 2920-2936.	2.9	11
70	Engagement and return of results preferences among a primarily African American genomic sequencing research cohort. <i>American Journal of Human Genetics</i> , 2021, 108, 894-902.	6.2	11
71	A genotypic ascertainment approach to refute the association of MYO1A variants with non-syndromic deafness. <i>European Journal of Human Genetics</i> , 2017, 25, 147-149.	2.8	10
72	Intentions to share exome sequencing results with family members: exploring spousal beliefs and attitudes. <i>European Journal of Human Genetics</i> , 2018, 26, 735-739.	2.8	10

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73	DENND5B Regulates Intestinal Triglyceride Absorption and Body Mass. <i>Scientific Reports</i> , 2019, 9, 3597.	3.3	10
74	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. <i>Human Mutation</i> , 2020, 41, 1577-1587.	2.5	10
75	Ethnic identity and engagement with genome sequencing research. <i>Genetics in Medicine</i> , 2019, 21, 1735-1743.	2.4	9
76	Associations of perceived norms with intentions to learn genomic sequencing results: Roles for attitudes and ambivalence.. <i>Health Psychology</i> , 2018, 37, 553-561.	1.6	9
77	Allelic heterogeneity of Proteus syndrome. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005181.	1.2	8
78	ACMG secondary findings 2.0. <i>Genetics in Medicine</i> , 2017, 19, 604-604.	2.4	7
79	Predictive and Precision Medicine with Genomic Data. <i>Clinical Chemistry</i> , 2020, 66, 33-41.	3.2	7
80	SomatoSim: precision simulation of somatic single nucleotide variants. <i>BMC Bioinformatics</i> , 2021, 22, 109.	2.6	7
81	Social Media Use in Research: Engaging Communities in Cohort Studies to Support Recruitment and Retention. <i>JMIR Research Protocols</i> , 2015, 4, e90.	1.0	7
82	Mosaic disorders and the Taxonomy of Human Disease. <i>Genetics in Medicine</i> , 2018, 20, 800-801.	2.4	6
83	Ability of Patients to Distinguish Among Cardiac Genomic Variant Subclassifications. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e001975.	3.6	6
84	Generation of human induced pluripotent stem cells from individuals with a homozygous CCR5 Δ 32 mutation. <i>Stem Cell Research</i> , 2019, 38, 101481.	0.7	6
85	Orofacial overgrowth with peripheral nerve enlargement and perineuriomatous pseudo-onion bulb proliferations is part of the PIK3CA-related overgrowth spectrum. <i>Human Genetics and Genomics Advances</i> , 2020, 1, 100009.	1.7	6
86	Overcalling secondary findings. <i>Genetics in Medicine</i> , 2016, 18, 416.	2.4	5
87	Genomic screening for monogenic forms of diabetes. <i>BMC Medicine</i> , 2018, 16, 25.	5.5	5
88	Adaptation of the working alliance inventory for the assessment of the therapeutic alliance in genetic counseling. <i>Journal of Genetic Counseling</i> , 2021, 30, 11-21.	1.6	5
89	Characterization of the hepatosplenic and portal venous findings in patients with Proteus syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2677-2684.	1.2	4
90	The role of future-oriented affect in engagement with genomic testing results. <i>Journal of Behavioral Medicine</i> , 2022, 45, 103-114.	2.1	4

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91	Engagement and communication among participants in the ClinSeq Genomic Sequencing Study. <i>Genetics in Medicine</i> , 2017, 19, 98-103.	2.4	3
92	Health behaviors among unaffected participants following receipt of variants of uncertain significance in cardiomyopathy-associated genes. <i>Genetics in Medicine</i> , 2019, 21, 748-752.	2.4	3
93	Correspondence on: "Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features". <i>Atherosclerosis</i> , 2021, 326, 63-64.	0.8	3
94	Acute Statin Administration Reduces Levels of Steroid Hormone Precursors. <i>Hormone and Metabolic Research</i> , 2020, 52, 742-746.	1.5	3
95	Perceptions of uncertainties about carrier results identified by exome sequencing in a randomized controlled trial. <i>Translational Behavioral Medicine</i> , 2020, 10, 441-450.	2.4	2
96	Clinical diagnosis of presumed SOX2 gonadosomatic mosaicism. <i>Ophthalmic Genetics</i> , 2021, 42, 320-325.	1.2	2
97	Preferences for and acceptability of receiving pharmacogenomic results by mail: A focus group study with a primarily African-American cohort. <i>Journal of Genetic Counseling</i> , 2021, 30, 1582-1590.	1.6	2
98	Response to Mendelsohn and Sabbadini. <i>Genetics in Medicine</i> , 2019, 21, 763-763.	2.4	1
99	Response to Esplin et al.. <i>Genetics in Medicine</i> , 2019, 21, 1252-1253.	2.4	1
100	Prophylactic anticoagulation of individuals with Proteus syndrome and COVID-19. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2829-2831.	1.2	1
101	Ubiquitous expression of Akt1 p.(E17K) results in vascular defects and embryonic lethality in mice. <i>Human Molecular Genetics</i> , 2020, 29, 3350-3360.	2.9	1
102	Correspondence on "The role of clinical response to treatment in determining pathogenicity of genomic variants" by Shen et al.. <i>Genetics in Medicine</i> , 2021, 23, 586.	2.4	1
103	Response to Nogales-Gadea et al.. <i>Genetics in Medicine</i> , 2015, 17, 680-681.	2.4	0
104	Cover Image, Volume 173A, Number 7, July 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.	1.2	0
105	Response to Hamosh et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1809-1810.	6.2	0