Leo B Waterston

List of Publications by Year
in descending order

105 papers	5,274 citations	109321 35 h-index	98798 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 standâ€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 â€œuncertaintyâ€•in genome sequencing?. Genetics in Medicine, 2014, 16, 977-980.	2.4	71
20	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
21	A Point Mutation in PDGFRB Causes Autosomal-Dominant Penttinen Syndrome. American Journal of Human Genetics, 2015, 97, 465-474.	6.2	64
22	Web Platform vs In-Person Genetic Counselor for Return of Carrier Results From Exome Sequencing. JAMA Internal Medicine, 2018, 178, 338.	5.1	64
23	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
24	Participant use and communication of findings from exome sequencing: a mixed-methods study. Genetics in Medicine, 2016, 18, 577-583.	2.4	56
25	Pharmacodynamic Study of Miransertib in Individuals with Proteus Syndrome. American Journal of Human Genetics, 2019, 104, 484-491.	6.2	56
26	Clinical Pharmacogenetics Implementation Consortium (<scp>CPIC</scp>) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of <scp>RYR</scp>1 or <scp>CACNA</scp>1S Genotypes. Clinical Pharmacology and Therapeutics, 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. Genetics in Medicine, 2019, 21, 2781-2790.	2.4	55
28	Variant Classification Concordance using the ACMG-AMP Variant Interpretation Guidelines across Nine Genomic Implementation Research Studies. American Journal of Human Genetics, 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. Cognition and Emotion, 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. JNCI Cancer Spectrum, 2020, 4, pkaa011.	2.9	48
31	Variant curation expert panel recommendations for RYR1 pathogenicity classifications in malignant hyperthermia susceptibility. Genetics in Medicine, 2021, 23, 1288-1295.	2.4	46
32	Assessing the reproducibility of exome copy number variations predictions. Genome Medicine, 2016, 8, 82.	8.2	44
33	Communicating Scientific Uncertainty About the COVID-19 Pandemic: Online Experimental Study of an Uncertainty-Normalizing Strategy. Journal of Medical Internet Research, 2021, 23, e27832.	4.3	44
34	Characterizing Participants in the ClinSeq Genome Sequencing Cohort as Early Adopters of a New Health Technology. PLoS ONE, 2015, 10, e0132690.	2.5	42
35	Advancing <scp>RAS/RASopathy</scp> therapies: An NCIâ€sponsored intramural and extramural collaboration for the study of <scp>RASopathies</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 866-876.	1.2	40
36	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36

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37	Evidenceâ€based assessments of clinical actionability in the context of secondary findings: Updates from ClinGen's Actionability Working Group. Human Mutation, 2018, 39, 1677-1685.	2.5	34
38	A six-attribute classification of geneticmosaicism. Genetics in Medicine, 2020, 22, 1743-1757.	2.4	34
39	Characterization of Courtesy Stigma Perceived by Parents of Overweight Children with Bardet-Biedl Syndrome. PLoS ONE, 2015, 10, e0140705.	2.5	33
40	Distinguishing Variant Pathogenicity From Genetic Diagnosis. JAMA - Journal of the American Medical Association, 2018, 320, 1929.	7.4	32
41	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
42	A Clinical Service to Support the Return of Secondary Genomic Findings in Human Research. American Journal of Human Genetics, 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. American Journal of Human Genetics, 2018, 103, 358-366.	6.2	29
44	Management of Secondary Genomic Findings. American Journal of Human Genetics, 2020, 107, 3-14.	6.2	29
45	Thrombosis risk factors in PIK3CAâ€related overgrowth spectrum and Proteus syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 571-581.	1.6	28
46	Clinical report: one year of treatment of Proteus syndrome with miransertib (ARQ 092). Journal of Physical Education and Sports Management, 2020, 6, a004549.	1.2	27
47	Characterization of thrombosis in patients with Proteus syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2359-2365.	1.2	26
48	A common haplotype containing functional CACNA1H variants is frequently coinherited with increased TPSAB1 copy number. Genetics in Medicine, 2018, 20, 503-512.	2.4	25
49	Somatic AKT1 mutations cause meningiomas colocalizing with a characteristic pattern of cranial hyperostosis. American Journal of Medical Genetics, Part A, 2016, 170, 2605-2610.	1.2	24
50	An approach to pediatric exome and genome sequencing. Current Opinion in Pediatrics, 2014, 26, 639-645.	2.0	23
51	Compound heterozygous alterations in intraflagellar transport protein CLUAP1 in a child with a novel Joubert and oralâ€“facialâ€“digital overlap syndrome. Journal of Physical Education and Sports Management, 2017, 3, a001321.	1.2	23
52	Lack of mutationâ€“histopathology correlation in a patient with Proteus syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1422-1432.	1.2	22
53	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in Medicine, 2018, 20, 855-866.	2.4	22
54	Family functioning mediates adaptation in caregivers of individuals with Rett syndrome. Patient Education and Counseling, 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). Genetics in Medicine, 2018, 20, 1077-1081.	2.4	21
56	Exome sequencing identifies novel mutations in C5orf42 in patients with Joubert syndrome with oralâ€“facialâ€“digital anomalies. Human Genome Variation, 2015, 2, 15045.	0.7	20
57	Outcomes of Counseling after Education about Carrier Results: A Randomized Controlled Trial. American Journal of Human Genetics, 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. Frontiers in Immunology, 2020, 11, 397.	4.8	16
59	Exome sequencing identifies a mutation in OFD1 in a male with Joubert syndrome, orofaciodigital spectrum anomalies and complex polydactyly. Human Genome Variation, 2016, 3, 15069.	0.7	15
60	Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. Genetics in Medicine, 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. Genome Medicine, 2018, 10, 99.	8.2	15
62	Pathogenetic insights from quantification of the cerebriform connective tissue nevus in Proteus syndrome. Journal of the American Academy of Dermatology, 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). Genetics in Medicine, 2021, 23, 979-988.	2.4	14
64	Preliminary validation of a consumer-oriented colorectal cancer risk assessment tool compatible with the US Surgeon Generalâ€™s My Family Health Portrait. Genetics in Medicine, 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 considerâ€”a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 169-171.	2.4	13
66	Secondary findings in exome slices, virtual panels, and anticipatory sequencing. Genetics in Medicine, 2019, 21, 41-43.	2.4	13
67	Increased Burden of Rare Sequence Variants in GnRH-Associated Genes in Women With Hypothalamic Amenorrhea. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e1441-e1452.	3.6	13
68	Web-Based Platform vs Genetic Counselors in Educating Patients About Carrier Results From Exome Sequencingâ€”Reply. JAMA Internal Medicine, 2018, 178, 999.	5.1	11
69	A mouse model of Proteus syndrome. Human Molecular Genetics, 2019, 28, 2920-2936.	2.9	11
70	Engagement and return of results preferences among a primarily African American genomic sequencing research cohort. American Journal of Human Genetics, 2021, 108, 894-902.	6.2	11
71	A genotypic ascertainment approach to refute the association of MYO1A variants with non-syndromic deafness. European Journal of Human Genetics, 2017, 25, 147-149.	2.8	10
72	Intentions to share exome sequencing results with family members: exploring spousal beliefs and attitudes. European Journal of Human Genetics, 2018, 26, 735-739.	2.8	10

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73	DENND5B Regulates Intestinal Triglyceride Absorption and Body Mass. Scientific Reports, 2019, 9, 3597.	3.3	10
74	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
75	Ethnic identity and engagement with genome sequencing research. Genetics in Medicine, 2019, 21, 1735-1743.	2.4	9
76	Associations of perceived norms with intentions to learn genomic sequencing results: Roles for attitudes and ambivalence.. Health Psychology, 2018, 37, 553-561.	1.6	9
77	Allelic heterogeneity of Proteus syndrome. Journal of Physical Education and Sports Management, 2020, 6, a005181.	1.2	8
78	ACMG secondary findings 2.0. Genetics in Medicine, 2017, 19, 604-604.	2.4	7
79	Predictive and Precision Medicine with Genomic Data. Clinical Chemistry, 2020, 66, 33-41.	3.2	7
80	SomatoSim: precision simulation of somatic single nucleotide variants. BMC Bioinformatics, 2021, 22, 109.	2.6	7
81	Social Media Use in Research: Engaging Communities in Cohort Studies to Support Recruitment and Retention. JMIR Research Protocols, 2015, 4, e90.	1.0	7
82	Mosaic disorders and the Taxonomy of Human Disease. Genetics in Medicine, 2018, 20, 800-801.	2.4	6
83	Ability of Patients to Distinguish Among Cardiac Genomic Variant Subclassifications. Circulation Genomic and Precision Medicine, 2018, 11, e001975.	3.6	6
84	Generation of human induced pluripotent stem cells from individuals with a homozygous CCR5Î”32 mutation. Stem Cell Research, 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. Human Genetics and Genomics Advances, 2020, 1, 100009.	1.7	6
86	Overcalling secondary findings. Genetics in Medicine, 2016, 18, 416.	2.4	5
87	Genomic screening for monogenic forms of diabetes. BMC Medicine, 2018, 16, 25.	5.5	5
88	Adaptation of the working alliance inventory for the assessment of the therapeutic alliance in genetic counseling. Journal of Genetic Counseling, 2021, 30, 11-21.	1.6	5
89	Characterization of the hepatosplenic and portal venous findings in patients with Proteus syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2677-2684.	1.2	4
90	The role of future-oriented affect in engagement with genomic testing results. Journal of Behavioral Medicine, 2022, 45, 103-114.	2.1	4

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