Leo B Waterston

List of Publications by Year in Descending Order

106 papers	3,002 citations	27 h-index	52 g-index
109 ext. papers	4,235 ext. citations	7.7 avg, IF	5.6 L-index

#	Paper	IF	Citations
106	PALLISTER□HALL SYNDROME AND GREIG CEPHALOPOLYSYNDACTYLY SYNDROME **2021**, 707-716		
105	Variant curation expert panel recommendations for RYR1 pathogenicity classifications in malignant hyperthermia susceptibility. *Genetics in Medicine*, **2021**, 23, 1288-1295	8.1	12
104	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	8.1	4
103	SomatoSim: precision simulation of somatic single nucleotide variants. *BMC Bioinformatics*, **2021**, 22, 109	3.6	2
102	Clinical diagnosis of presumed SOX2 gonadosomatic mosaicism. *Ophthalmic Genetics*, **2021**, 42, 320-325	1.2	0
101	Communicating Scientific Uncertainty About the COVID-19 Pandemic: Online Experimental Study of an Uncertainty-Normalizing Strategy. *Journal of Medical Internet Research*, **2021**, 23, e27832	7.6	14
100	Preferences for and acceptability of receiving pharmacogenomic results by mail: A focus group study with a primarily African-American cohort. *Journal of Genetic Counseling*, **2021**, 30, 1582-1590	2.5	0
99	Engagement and return of results preferences among a primarily African American genomic sequencing research cohort. *American Journal of Human Genetics*, **2021**, 108, 894-902	11	0
98	Correspondence on: "Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features". *Atherosclerosis*, **2021**, 326, 63-64	3.1	2
97	Correspondence on "The role of clinical response to treatment in determining pathogenicity of genomic variants" by Shen et al. *Genetics in Medicine*, **2021**, 23, 586	8.1	1
96	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	5.6	8
95	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
94	Adaptation of the working alliance inventory for the assessment of the therapeutic alliance in genetic counseling. *Journal of Genetic Counseling*, **2021**, 30, 11-21	2.5	0
93	The role of future-oriented affect in engagement with genomic testing results. *Journal of Behavioral Medicine*, **2021**, 1	3.6	0
92	Response to Hamosh et□al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	
91	Prophylactic anticoagulation of individuals with Proteus syndrome and COVID-19. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2829-2831	2.5	1
90	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	11	17

89	Ubiquitous expression of Akt1 p.(E17K) results in vascular defects and embryonic lethality in mice. *Human Molecular Genetics*, **2020**, 29, 3350-3360	5.6	
88	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. *Human Mutation*, **2020**, 41, 1577-1587	4.7	4
87	Management of Secondary Genomic Findings. *American Journal of Human Genetics*, **2020**, 107, 3-14	11	9
86	Clinical report: one year of treatment of Proteus syndrome with miransertib (ARQ 092). *Journal of Physical Education and Sports Management*, **2020**, 6,	2.8	17
85	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	8.4	10
84	Acute Statin Administration Reduces Levels of Steroid Hormone Precursors. *Hormone and Metabolic Research*, **2020**, 52, 742-746	3.1	
83	Allelic heterogeneity of Proteus syndrome. *Journal of Physical Education and Sports Management*, **2020**, 6,	2.8	7
82	Advancing RAS/RASopathy therapies: An NCI-sponsored intramural and extramural collaboration for the study of RASopathies. *American Journal of Medical Genetics, Part A*, **2020**, 182, 866-876	2.5	24
81	Predictive and Precision Medicine with Genomic Data. *Clinical Chemistry*, **2020**, 66, 33-41	5.5	5
80	Orofacial overgrowth with peripheral nerve enlargement and perineuriomatous pseudo-onion bulb proliferations is part of the -related overgrowth spectrum.. *Human Genetics and Genomics Advances*, **2020**, 1, 100009	0.8	3
79	A six-attribute classification of genetic mosaicism. *Genetics in Medicine*, **2020**, 22, 1743-1757	8.1	14
78	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines. *Human Mutation*, **2020**, 41, 1734-1737	4.7	12
77	Area Deprivation Index and Rurality in Relation to Lung Cancer Prevalence and Mortality in a Rural State. *JNCI Cancer Spectrum*, **2020**, 4, pkaa011	4.6	12
76	Perceptions of uncertainties about carrier results identified by exome sequencing in a randomized controlled trial. *Translational Behavioral Medicine*, **2020**, 10, 441-450	3.2	1
75	Redefining the Etiologic Landscape of Cerebellar Malformations. *American Journal of Human Genetics*, **2019**, 105, 606-615	11	40
74	A mouse model of Proteus syndrome. *Human Molecular Genetics*, **2019**, 28, 2920-2936	5.6	5
73	Generation of human induced pluripotent stem cells from individuals with a homozygous CCR5Δ32 mutation. *Stem Cell Research*, **2019**, 38, 101481	1.6	3
72	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	8.1	32

71	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. *Cell*, **2019**, 177, 32-37	56.2	53
70	DENND5B Regulates Intestinal Triglyceride Absorption and Body Mass. *Scientific Reports*, **2019**, 9, 3597	4.9	6
69	Pharmacodynamic Study of Miransertib in Individuals with Proteus Syndrome. *American Journal of Human Genetics*, **2019**, 104, 484-491	11	39
68	Response to Mendelsohn and Sabbadini. *Genetics in Medicine*, **2019**, 21, 763	8.1	1
67	Health behaviors among unaffected participants following receipt of variants of uncertain significance in cardiomyopathy-associated genes. *Genetics in Medicine*, **2019**, 21, 748-752	8.1	3
66	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. *Current Protocols in Human Genetics*, **2019**, 103, e93	3.2	14
65	Thrombosis risk factors in PIK3CA-related overgrowth spectrum and Proteus syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 571-581	3.1	17
64	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. *Genome Medicine*, **2019**, 12, 3	14.4	106
63	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	11	17
62	Ethnic identity and engagement with genome sequencing research. *Genetics in Medicine*, **2019**, 21, 1735-1743	8.1	6
61	Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. *Clinical Pharmacology and Therapeutics*, **2019**, 105, 1338-1344	6.1	35
60	Response to Esplin et al. *Genetics in Medicine*, **2019**, 21, 1252-1253	8.1	1
59	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. *Genetics in Medicine*, **2019**, 21, 1189-1198	8.1	67
58	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	8.1	61
57	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
56	Intentions to share exome sequencing results with family members: exploring spousal beliefs and attitudes. *European Journal of Human Genetics*, **2018**, 26, 735-739	5.3	7
55	Web Platform vs In-Person Genetic Counselor for Return of Carrier Results From Exome Sequencing: A Randomized Clinical Trial. *JAMA Internal Medicine*, **2018**, 178, 338-346	11.5	53
54	Pathogenetic insights from quantification of the cerebriform connective tissue nevus in Proteus syndrome. *Journal of the American Academy of Dermatology*, **2018**, 78, 725-732	4.5	12

53	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). *Genetics in Medicine*, **2018**, 20, 169-171	8.1	6
52	Mosaic disorders and the Taxonomy of Human Disease. *Genetics in Medicine*, **2018**, 20, 800-801	8.1	5
51	Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework. *Genetics in Medicine*, **2018**, 20, 1054-1060	8.1	159
50	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	8.1	17
49	Outcomes of Counseling after Education about Carrier Results: A Randomized Controlled Trial. *American Journal of Human Genetics*, **2018**, 102, 540-546	11	14
48	The ACMG/AMP reputable source criteria for the interpretation of sequence variants. *Genetics in Medicine*, **2018**, 20, 1687-1688	8.1	74
47	A common haplotype containing functional CACNA1H variants is frequently coinherited with increased TPSAB1 copy number. *Genetics in Medicine*, **2018**, 20, 503-512	8.1	16
46	Web-Based Platform vs Genetic Counselors in Educating Patients About Carrier Results From Exome Sequencing-Reply. *JAMA Internal Medicine*, **2018**, 178, 999	11.5	7
45	Genomic screening for monogenic forms of diabetes. *BMC Medicine*, **2018**, 16, 25	11.4	2
44	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	11	24
43	Ability of Patients to Distinguish Among Cardiac Genomic Variant Subclassifications. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001975	5.2	5
42	Associations of perceived norms with intentions to learn genomic sequencing results: Roles for attitudes and ambivalence. *Health Psychology*, **2018**, 37, 553-561	5	6
41	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. *Genetics in Medicine*, **2018**, 20, 855-866	8.1	16
40	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	14.4	9
39	Characterization of the hepatosplenic and portal venous findings in patients with Proteus syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2677-2684	2.5	4
38	Evidence-based assessments of clinical actionability in the context of secondary findings: Updates from ClinGen's Actionability Working Group. *Human Mutation*, **2018**, 39, 1677-1685	4.7	21
37	Updated recommendation for the benign stand-alone ACMG/AMP criterion. *Human Mutation*, **2018**, 39, 1525-1530	4.7	48
36	Distinguishing Variant Pathogenicity From Genetic Diagnosis: How to Know Whether a Variant Causes a Condition. *JAMA - Journal of the American Medical Association*, **2018**, 320, 1929-1930	27.4	23

35	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	11	64
34	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. *Human Mutation*, **2018**, 39, 1517-1524	4.7	215
33	A taxonomy of medical uncertainties in clinical genome sequencing. *Genetics in Medicine*, **2017**, 19, 918-925	8.1	62
32	ACMG secondary findings 2.0. *Genetics in Medicine*, **2017**, 19, 604	8.1	7
31	Nomenclature and definition in asymmetric regional body overgrowth. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1735-1738	2.5	24
30	Characterization of thrombosis in patients with Proteus syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2359-2365	2.5	19
29	Compound heterozygous alterations in intraflagellar transport protein in a child with a novel Joubert and oral-facial-digital overlap syndrome. *Journal of Physical Education and Sports Management*, **2017**, 3,	2.8	12
28	Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. *Genetics in Medicine*, **2017**, 19, 357-361	8.1	10
27	Engagement and communication among participants in the ClinSeq Genomic Sequencing Study. *Genetics in Medicine*, **2017**, 19, 98-103	8.1	2
26	Mosaic Disorders of the PI3K/PTEN/AKT/TSC/mTORC1 Signaling Pathway. *Dermatologic Clinics*, **2017**, 35, 51-60	4.2	76
25	Assessing the reproducibility of exome copy number variations predictions. *Genome Medicine*, **2016**, 8, 82	14.4	28
24	Somatic AKT1 mutations cause meningiomas colocalizing with a characteristic pattern of cranial hyperostosis. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2605-10	2.5	19
23	Elevated basal serum tryptase identifies a multisystem disorder associated with increased TPSAB1 copy number. *Nature Genetics*, **2016**, 48, 1564-1569	36.3	185
22	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	1.8	11
21	Participant use and communication of findings from exome sequencing: a mixed-methods study. *Genetics in Medicine*, **2016**, 18, 577-83	8.1	49
20	A Clinical Service to Support the Return of Secondary Genomic Findings in Human Research. *American Journal of Human Genetics*, **2016**, 98, 435-441	11	20
19	A genotypic ascertainment approach to refute the association of MYO1A variants with non-syndromic deafness. *European Journal of Human Genetics*, **2016**, 25, 147-149	5.3	7
18	Lack of mutation-histopathology correlation in a patient with Proteus syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1422-1432	2.5	16

17	Overcalling secondary findings. *Genetics in Medicine*, **2016**, 18, 416	8.1	4
16	A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation. *Genetics in Medicine*, **2016**, 18, 1258-1268	8.1	64
15	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. *American Journal of Human Genetics*, **2016**, 98, 1051-1066	11	107
14	Family functioning mediates adaptation in caregivers of individuals with Rett syndrome. *Patient Education and Counseling*, **2016**, 99, 1873-1879	3.1	12
13	Preliminary validation of a consumer-oriented colorectal cancer risk assessment tool compatible with the US Surgeon General's My Family Health Portrait. *Genetics in Medicine*, **2015**, 17, 753-6	8.1	11
12	Response to Nogales-Gadea et al. *Genetics in Medicine*, **2015**, 17, 680-1	8.1	
11	A Point Mutation in PDGFRB Causes Autosomal-Dominant Penttinen Syndrome. *American Journal of Human Genetics*, **2015**, 97, 465-74	11	52
10	Characterizing Participants in the ClinSeq Genome Sequencing Cohort as Early Adopters of a New Health Technology. *PLoS ONE*, **2015**, 10, e0132690	3.7	38
9	Exome sequencing identifies novel mutations in C5orf42 in patients with Joubert syndrome with oral-facial-digital anomalies. *Human Genome Variation*, **2015**, 2, 15045	1.8	16
8	Characterization of Courtesy Stigma Perceived by Parents of Overweight Children with Bardet-Biedl Syndrome. *PLoS ONE*, **2015**, 10, e0140705	3.7	20
7	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, 1456-65	2.3	37
6	Social Media Use in Research: Engaging Communities in Cohort Studies to Support Recruitment and Retention. *JMIR Research Protocols*, **2015**, 4, e90	2	6
5	How do research participants perceive "uncertainty" in genome sequencing?. *Genetics in Medicine*, **2014**, 16, 977-80	8.1	58
4	Diagnostic clinical genome and exome sequencing. *New England Journal of Medicine*, **2014**, 370, 2418-25	59.2	404
3	An approach to pediatric exome and genome sequencing. *Current Opinion in Pediatrics*, **2014**, 26, 639-45	3.2	21
2	PROTEUS SYNDROME763		0
1	Communicating Scientific Uncertainty About the COVID-19 Pandemic: Online Experimental Study of an Uncertainty-Normalizing Strategy (Preprint)		1