Leslie G Biesecker

List of Publications by Year
in descending order

150	15,007	38742 50	19749 117
papers	citations	h-index	g-index

153	153	153	18591
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	6.2	2,325
2	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in Medicine, 2013, 15, 565-574.	2.4	2,186
3	A Mosaic Activating Mutation inAKT1Associated with the Proteus Syndrome. New England Journal of Medicine, 2011, 365, 611-619.	27.0	800
4	A genomic view of mosaicism and human disease. Nature Reviews Genetics, 2013, 14, 307-320.	16.3	527
5	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. Human Mutation, 2018, 39, 1517-1524.	2.5	511
6	Diagnostic Clinical Genome and Exome Sequencing. New England Journal of Medicine, 2014, 370, 2418-2425.	27.0	488
7	PIK3CAâ€related overgrowth spectrum (PROS): Diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. American Journal of Medical Genetics, Part A, 2015, 167, 287-295.	1.2	399
8	Proteus syndrome: Diagnostic criteria, differential diagnosis, and patient evaluation. American Journal of Medical Genetics Part A, 1999, 84, 389-395.	2.4	374
9	Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework. Genetics in Medicine, 2018, 20, 1054-1060.	2.4	366
10	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
11	Elevated basal serum tryptase identifies a multisystem disorder associated with increased TPSAB1 copy number. Nature Genetics, 2016, 48, 1564-1569.	21.4	279
12	Mosaic overgrowth with fibroadipose hyperplasia is caused by somatic activating mutations in PIK3CA. Nature Genetics, 2012, 44, 928-933.	21.4	269
13	Clinical delineation and natural history of the PIK3CAâ€related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 1713-1733.	1.2	249
14	The ClinSeq Project: Piloting large-scale genome sequencing for research in genomic medicine. Genome Research, 2009, 19, 1665-1674.	5.5	236
15	The challenges of Proteus syndrome: diagnosis and management. European Journal of Human Genetics, 2006, 14, 1151-1157.	2.8	231
16	Newly delineated syndrome of congenital lipomatous overgrowth, vascular malformations, and epidermal nevi (CLOVE syndrome) in seven patients. American Journal of Medical Genetics, Part A, 2007, 143A, 2944-2958.	1.2	201
17	Reassessment of the Proteus syndrome literature: Application of diagnostic criteria to published cases. American Journal of Medical Genetics Part A, 2004, 130A, 111-122.	2.4	195
18	Strategic vision for improving human health at The Forefront of Genomics. Nature, 2020, 586, 683-692.	27.8	192

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19	Mosaic RAS/MAPK variants cause sporadic vascular malformations which respond to targeted therapy. Journal of Clinical Investigation, 2018, 128, 1496-1508.	8.2	191
20	Secondary Variants in Individuals Undergoing Exome Sequencing: Screening of 572 Individuals Identifies High-Penetrance Mutations in Cancer-Susceptibility Genes. American Journal of Human Genetics, 2012, 91, 97-108.	6.2	190
21	High incidence of unrecognized visceral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. Genetics in Medicine, 2016, 18, 41-48.	2.4	171
22	Intentions to receive individual results from whole-genome sequencing among participants in the ClinSeq study. European Journal of Human Genetics, 2013, 21, 261-265.	2.8	156
23	Systematic Evaluation of Sanger Validation of Next-Generation Sequencing Variants. Clinical Chemistry, 2016, 62, 647-654.	3.2	154
24	Opportunities and challenges for the integration of massively parallel genomic sequencing into clinical practice: lessons from the ClinSeq project. Genetics in Medicine, 2012, 14, 393-398.	2.4	145
25	VarSifter: Visualizing and analyzing exome-scale sequence variation data on a desktop computer. Bioinformatics, 2012, 28, 599-600.	4.1	137
26	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
27	Motivators for participation in a whole-genome sequencing study: implications for translational genomics research. European Journal of Human Genetics, 2011, 19, 1213-1217.	2.8	129
28	Nationwide survey of extended newborn screening by tandem mass spectrometry in Taiwan. Journal of Inherited Metabolic Disease, 2010, 33, 295-305.	3.6	128
29	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
30	Personal utility in genomic testing: a systematic literature review. European Journal of Human Genetics, 2017, 25, 662-668.	2.8	122
31	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. Genetics in Medicine, 2019, 21, 1189-1198.	2.4	115
32	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines. Human Mutation, 2020, 41, 1734-1737.	2.5	105
33	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. Cell, 2017, 169, 6-12.	28.9	103
34	Updated recommendation for the benign standâ€alone ACMG/AMP criterion. Human Mutation, 2018, 39, 1525-1530.	2.5	102
35	Elements of morphology: Introduction. American Journal of Medical Genetics, Part A, 2009, 149A, 2-5.	1.2	98
36	Clinical differentiation between proteus syndrome and hemihyperplasia: Description of a distinct form of hemihyperplasia. , 1998, 79, 311-318.		97

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37	A taxonomy of medical uncertainties in clinical genome sequencing. Genetics in Medicine, 2017, 19, 918-925.	2.4	91
38	Elements of morphology: Standard terminology for the hands and feet. American Journal of Medical Genetics, Part A, 2009, 149A, 93-127.	1.2	89
39	Exome sequencing identifies ACSF3 as a cause of combined malonic and methylmalonic aciduria. Nature Genetics, 2011, 43, 883-886.	21.4	89
40	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. Current Protocols in Human Genetics, 2019, 103, e93.	3.5	88
41	Validation of My Family Health Portrait for six common heritable conditions. Genetics in Medicine, 2010, 12, 370-375.	2.4	72
42	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
43	Hypothesis-generating research and predictive medicine. Genome Research, 2013, 23, 1051-1053.	5.5	66
44	Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. American Journal of Human Genetics, 2015, 96, 913-925.	6.2	66
45	A Point Mutation in PDGFRB Causes Autosomal-Dominant Penttinen Syndrome. American Journal of Human Genetics, 2015, 97, 465-474.	6.2	64
46	Web Platform vs In-Person Genetic Counselor for Return of Carrier Results From Exome Sequencing. JAMA Internal Medicine, 2018, 178, 338.	5.1	64
47	Social and behavioral research in genomic sequencing: approaches from the Clinical Sequencing Exploratory Research Consortium Outcomes and Measures Working Group. Genetics in Medicine, 2014, 16, 727-735.	2.4	60
48	Longâ€term survival in TARP syndrome and confirmation of RBM10 as the diseaseâ€causing gene. American Journal of Medical Genetics, Part A, 2011, 155, 2516-2520.	1.2	56
49	Molecular characterization of a cohort of 73 patients with infantile spasms syndrome. European Journal of Medical Genetics, 2015, 58, 51-58.	1.3	56
50	Participant use and communication of findings from exome sequencing: a mixed-methods study. Genetics in Medicine, 2016, 18, 577-583.	2.4	56
51	Pharmacodynamic Study of Miransertib in Individuals with Proteus Syndrome. American Journal of Human Genetics, 2019, 104, 484-491.	6.2	56
52	Genetic susceptibility testing for Alzheimer disease: Motivation to obtain information and control as precursors to coping with increased risk. Patient Education and Counseling, 2006, 64, 259-267.	2.2	54
53	Genetic regulation of OAS1 nonsense-mediated decay underlies association with COVID-19 hospitalization in patients of European and African ancestries. Nature Genetics, 2022, 54, 1103-1116.	21.4	54
54	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

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55	Variant curation expert panel recommendations for RYR1 pathogenicity classifications in malignant hyperthermia susceptibility. Genetics in Medicine, 2021, 23, 1288-1295.	2.4	46
56	Assessing the reproducibility of exome copy number variations predictions. Genome Medicine, 2016, 8, 82.	8.2	44
57	Progressive overgrowth of the cerebriform connective tissue nevus in patients with Proteus syndrome. Journal of the American Academy of Dermatology, 2010, 63, 799-804.	1.2	43
58	Characterizing Participants in the ClinSeq Genome Sequencing Cohort as Early Adopters of a New Health Technology. PLoS ONE, 2015, 10, e0132690.	2.5	42
59	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
60	Associations between risk perceptions and worry about common diseases: A between- and within-subjects examination. Psychology and Health, 2013, 28, 434-449.	2.2	39
61	Incidental Variants Are Critical for Genomics. American Journal of Human Genetics, 2013, 92, 648-651.	6.2	37
62	Assessment and management of the orthopedic and other complications of Proteus syndrome. Journal of Children's Orthopaedics, 2011, 5, 319-327.	1.1	33
63	Considerations for a multiaxis nomenclature system for medical genetics. Genetics in Medicine, 2001, 3, 290-293.	2.4	32
64	Integrative DNA, RNA, and Protein Evidence Connects TREML4 to Coronary Artery Calcification. American Journal of Human Genetics, 2014, 95, 66-76.	6.2	30
65	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
66	Preferences for learning different types of genome sequencing results among young breast cancer patients: Role of psychological and clinical factors. Translational Behavioral Medicine, 2018, 8, 71-79.	2.4	29
67	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
68	Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism. American Journal of Medical Genetics, Part A, 2014, 164, 120-128.	1.2	28
69	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
70	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
71	Characterization of thrombosis in patients with Proteus syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2359-2365.	1.2	26
72	Quantifying survival in patients with Proteus syndrome. Genetics in Medicine, 2017, 19, 1376-1379.	2.4	25

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73	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
74	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
75	A dyadic genotypeâ€“phenotype approach to diagnostic criteria for Proteus syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 565-570.	1.6	24
76	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
77	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
78	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	2.0	21
79	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
80	Outcomes of Counseling after Education about Carrier Results: A Randomized Controlled Trial. American Journal of Human Genetics, 2018, 102, 540-546.	6.2	18
81	Exome sequencing in clinical settings: preferences and experiences of parents of children with rare diseases (SEQUAPRE study). European Journal of Human Genetics, 2019, 27, 701-710.	2.8	18
82	Severe anomalies associated with ring chromosome 7. American Journal of Medical Genetics Part A, 1991, 40, 429-431.	2.4	16
83	Elements of morphology: Standard terminology for the external genitalia. American Journal of Medical Genetics, Part A, 2013, 161, 1238-1263.	1.2	16
84	A Limited Form of Proteus Syndrome With Bilateral Plantar Cerebriform Collagenomas and Varicose Veins Secondary to a MosaicAKT1Mutation. JAMA Dermatology, 2014, 150, 990.	4.1	16
85	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
86	Epigenetic profiling of the H19 differentially methylated region and comprehensive whole genome arrayâ€based analysis in Silverâ€“Russell syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2521-2528.	1.2	14
87	A patient with CantÃº syndrome associated with fatal bronchopulmonary dysplasia and pulmonary hypertension. American Journal of Medical Genetics, Part A, 2014, 164, 2118-2120.	1.2	14
88	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
89	The PiGeOn project: protocol of a longitudinal study examining psychosocial and ethical issues and outcomes in germline genomic sequencing for cancer. BMC Cancer, 2018, 18, 454.	2.6	14
90	Reactions to clinical reinterpretation of a gene variant by participants in a sequencing study. Genetics in Medicine, 2018, 20, 337-345.	2.4	14

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91	Identification of candidate genes involved in coronary artery calcification by transcriptome sequencing of cell lines. BMC Genomics, 2014, 15, 198.	2.8	13
92	Somatic mosaicism for the p.His1047Arg mutation in PIK3CA in a girl with mesenteric lipomatosis. American Journal of Medical Genetics, Part A, 2014, 164, 2360-2364.	1.2	13
93	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
94	Secondary findings in exome slices, virtual panels, and anticipatory sequencing. Genetics in Medicine, 2019, 21, 41-43.	2.4	13
95	Dizygotic twins concordant for truncus arteriosus. Clinical Genetics, 1991, 39, 75-79.	2.0	12
96	A mouse model of Proteus syndrome. Human Molecular Genetics, 2019, 28, 2920-2936.	2.9	11
97	Genomic screening and genomic diagnostic testingâ€”two very different kettles of fish. Genome Medicine, 2019, 11, 75.	8.2	11
98	Macrocerebellum, epilepsy, intellectual disability, and gut malrotation in a child with a 16q24.1â€“q24.2 contiguous gene deletion. American Journal of Medical Genetics, Part A, 2014, 164, 2062-2068.	1.2	10
99	West syndrome caused by homozygous variant in the evolutionary conserved gene encoding the mitochondrial elongation factor GUF1. European Journal of Human Genetics, 2016, 24, 1001-1008.	2.8	10
100	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
101	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
102	The PiGeOn project: protocol for a longitudinal study examining psychosocial, behavioural and ethical issues and outcomes in cancer tumour genomic profiling. BMC Cancer, 2018, 18, 389.	2.6	10
103	DENND5B Regulates Intestinal Triglyceride Absorption and Body Mass. Scientific Reports, 2019, 9, 3597.	3.3	10
104	The Nirvana Fallacy and the Return of Results. American Journal of Bioethics, 2013, 13, 43-44.	0.9	9
105	Orthopaedic Management of Leg-length Discrepancy in Proteus Syndrome: A Case Series. Journal of Pediatric Orthopaedics, 2018, 38, e138-e144.	1.2	9
106	Ethnic identity and engagement with genome sequencing research. Genetics in Medicine, 2019, 21, 1735-1743.	2.4	9
107	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
108	Factors affecting breast cancer patients' need for genetic risk information: From information insufficiency to information need. Journal of Genetic Counseling, 2019, 28, 543-557.	1.6	8

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109	Cancer Patient Experience of Uncertainty While Waiting for Genome Sequencing Results. Frontiers in Psychology, 2021, 12, 647502.	2.1	8
110	Allelic heterogeneity of Proteus syndrome. Journal of Physical Education and Sports Management, 2020, 6, a005181.	1.2	8
111	Recessive congenital methemoglobinemia caused by a rare mechanism: Maternal uniparental heterodisomy with segmental isodisomy of a chromosome 22. Blood Cells, Molecules, and Diseases, 2012, 49, 114-117.	1.4	7
112	Challenges to informed consent for exome sequencing: A bestâ€"worst scaling experiment. Journal of Genetic Counseling, 2019, 28, 1189-1197.	1.6	7
113	Case Report: Five-Year Experience of AKT Inhibition with Miransertib (MK-7075) in an Individual with Proteus Syndrome. Journal of Physical Education and Sports Management, 2021, 7, mcs.a006134.	1.2	7
114	Complex mosaic CDKL5 deletion with two distinct mutant alleles in a 4â€yearâ€old girl. American Journal of Medical Genetics, Part A, 2014, 164, 2025-2028.	1.2	6
115	Mosaic disorders and the Taxonomy of Human Disease. Genetics in Medicine, 2018, 20, 800-801.	2.4	6
116	Ability of Patients to Distinguish Among Cardiac Genomic Variant Subclassifications. Circulation Genomic and Precision Medicine, 2018, 11, e001975.	3.6	6
117	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
118	Preferences for return of germline genome sequencing results for cancer patients and their genetic relatives in a research setting. European Journal of Human Genetics, 2022, 30, 930-937.	2.8	6
119	Genomic screening for monogenic forms of diabetes. BMC Medicine, 2018, 16, 25.	5.5	5
120	A multi-dimensional analysis of genotypeâ€"phenotype discordance in malignant hyperthermia susceptibility. British Journal of Anaesthesia, 2020, 125, 995-1001.	3.4	5
121	An openâ€source python library for detection of known and novel Kell, Duffy and Kidd variants from exome sequencing. Vox Sanguinis, 2021, 116, 451-463.	1.5	5
122	Effectively communicating comprehensive tumor genomic profiling results: Mitigating uncertainty for advanced cancer patients. Patient Education and Counseling, 2022, 105, 452-459.	2.2	5
123	Implementing the elements of morphology in the American Journal of Medical Genetics. American Journal of Medical Genetics, Part A, 2011, 155, 969-971.	1.2	4
124	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
125	Cardiothoracic imaging findings of Proteus syndrome. Scientific Reports, 2021, 11, 6577.	3.3	4
126	Engagement and communication among participants in the ClinSeq Genomic Sequencing Study. Genetics in Medicine, 2017, 19, 98-103.	2.4	3

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127	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
128	Correspondence on: â€œHomozygous familial hypercholesterolemia in Italy: Clinical and molecular featuresâ€• Atherosclerosis, 2021, 326, 63-64.	0.8	3
129	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
130	Dyadic concordance and associations of beliefs with intentions to learn carrier results from genomic sequencing. Journal of Behavioral Medicine, 2021, 44, 860-866.	2.1	2
131	Invited Commentary on â€œMy Research Results: a program to facilitate return of clinically actionable genomic research findingsâ€•by Willis et al.. European Journal of Human Genetics, 2022, 30, 256-257.	2.8	2
132	Hepatomegaly and hyperammonemia in a girl with Silverâ€“Russell syndrome caused by maternal uniparental isodisomy of chromosome 7. American Journal of Medical Genetics, Part A, 2014, 164, 2114-2117.	1.2	1
133	Response to Mendelsohn and Sabbadini. Genetics in Medicine, 2019, 21, 763-763.	2.4	1
134	Response to Nakaguma et al.. Genetics in Medicine, 2019, 21, 261.	2.4	1
135	Judgment and Decision Making in Genome Sequencing. , 2019, , 57-73.		1
136	Response to Esplin et al.. Genetics in Medicine, 2019, 21, 1252-1253.	2.4	1
137	Roles of attitudes and injunctive norms in decisional conflict and disclosure following receipt of genome sequencing results. Social Science and Medicine, 2020, 262, 113147.	3.8	1
138	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
139	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
140	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
141	Validation of the multidimensional impact of Cancer Risk Assessment Questionnaire to assess impact of waiting for genome sequencing results. Psycho-Oncology, 2022, , .	2.3	1
142	Mosaicism and the taxonomy of human disease. Journal of Physical Education and Sports Management, 2021, 7, a006163.	1.2	1
143	A commentary on actionable secondary findings in the 73 ACMG-recommended genes in 1559 Thai exomes. Journal of Human Genetics, 2022, , .	2.3	1
144	Genome Sequencing and Individual Responses to Results. , 2019, , 17-30.		0

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145	Uncertainties in Genome Sequencing. , 2019, , 75-88.		0
146	Acne following Blaschko's lines in Proteus syndrome. JAAD Case Reports, 2020, 6, 1072-1074.	0.8	0
147	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
148	<scp>Lateâ€onset</scp> Proteus syndrome with cerebriform connective tissue nevus and subsequent development of intraductal papilloma. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
149	Development of the Clinical Gestalt Assessment: a visual clinical global impression scale for Proteus syndrome. Orphanet Journal of Rare Diseases, 2022, 17, 173.	2.7	0
150	Further validation of the Perceptions of Uncertainties in Genome Sequencing scale among patients with cancer undergoing tumor sequencing. Clinical Genetics, 2022, 102, 110-116.	2.0	0