

Leslie G Biesecker

List of Publications by Year in descending order

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Version: 2024-02-01

150
papers

15,007
citations

38742

50
h-index

19749

117
g-index

153
all docs

153
docs citations

153
times ranked

18591
citing authors

#	ARTICLE	IF	CITATIONS
1	Effectively communicating comprehensive tumor genomic profiling results: Mitigating uncertainty for advanced cancer patients. <i>Patient Education and Counseling</i> , 2022, 105, 452-459.	2.2	5
2	A standard of care for individuals with <scp><i>PIK3CA</i></scp>-related disorders: An international expert consensus statement. <i>Clinical Genetics</i> , 2022, 101, 32-47.	2.0	21
3	Invited Commentary on "My Research Results: a program to facilitate return of clinically actionable genomic research findings" by Willis et al.. <i>European Journal of Human Genetics</i> , 2022, 30, 256-257.	2.8	2
4	Preferences for return of germline genome sequencing results for cancer patients and their genetic relatives in a research setting. <i>European Journal of Human Genetics</i> , 2022, 30, 930-937.	2.8	6
5	Validation of the multidimensional impact of Cancer Risk Assessment Questionnaire to assess impact of waiting for genome sequencing results. <i>Psycho-Oncology</i> , 2022, , .	2.3	1
6	<scp>Late-onset</scp> Proteus syndrome with cerebriform connective tissue nevus and subsequent development of intraductal papilloma. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	1.2	0
7	Development of the Clinical Gestalt Assessment: a visual clinical global impression scale for Proteus syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 173.	2.7	0
8	A commentary on actionable secondary findings in the 73 ACMG-recommended genes in 1559 Thai exomes. <i>Journal of Human Genetics</i> , 2022, , .	2.3	1
9	Further validation of the Perceptions of Uncertainties in Genome Sequencing scale among patients with cancer undergoing tumor sequencing. <i>Clinical Genetics</i> , 2022, 102, 110-116.	2.0	0
10	Genetic regulation of OAS1 nonsense-mediated decay underlies association with COVID-19 hospitalization in patients of European and African ancestries. <i>Nature Genetics</i> , 2022, 54, 1103-1116.	21.4	54
11	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
12	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
13	An open-source python library for detection of known and novel Kell, Duffy and Kidd variants from exome sequencing. <i>Vox Sanguinis</i> , 2021, 116, 451-463.	1.5	5
14	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
15	Cardiothoracic imaging findings of Proteus syndrome. <i>Scientific Reports</i> , 2021, 11, 6577.	3.3	4
16	Cancer Patient Experience of Uncertainty While Waiting for Genome Sequencing Results. <i>Frontiers in Psychology</i> , 2021, 12, 647502.	2.1	8
17	Dyadic concordance and associations of beliefs with intentions to learn carrier results from genomic sequencing. <i>Journal of Behavioral Medicine</i> , 2021, 44, 860-866.	2.1	2
18	Correspondence on: "Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features". <i>Atherosclerosis</i> , 2021, 326, 63-64.	0.8	3

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19	Response to Hamosh et al. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
20	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
21	Mosaicism and the taxonomy of human disease. Journal of Physical Education and Sports Management, 2021, 7, a006163.	1.2	1
22	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
23	Advancing <sc>RAS/RASopathy</sc> therapies: An NCI-sponsored intramural and extramural collaboration for the study of <sc>RASopathies</sc>. American Journal of Medical Genetics, Part A, 2020, 182, 866-876.	1.2	40
24	Acne following Blaschko's lines in Proteus syndrome. JAAD Case Reports, 2020, 6, 1072-1074.	0.8	0
25	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
26	A multi-dimensional analysis of genotype-phenotype discordance in malignant hyperthermia susceptibility. British Journal of Anaesthesia, 2020, 125, 995-1001.	3.4	5
27	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
28	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines. Human Mutation, 2020, 41, 1734-1737.	2.5	105
29	Strategic vision for improving human health at The Forefront of Genomics. Nature, 2020, 586, 683-692.	27.8	192
30	Prophylactic anticoagulation of individuals with Proteus syndrome and <sc>COVID</sc>. American Journal of Medical Genetics, Part A, 2020, 182, 2829-2831.	1.2	1
31	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
32	Ubiquitous expression of <i>Akt1</i> p.(E17K) results in vascular defects and embryonic lethality in mice. Human Molecular Genetics, 2020, 29, 3350-3360.	2.9	1
33	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
34	Allelic heterogeneity of Proteus syndrome. Journal of Physical Education and Sports Management, 2020, 6, a005181.	1.2	8
35	Response to Mendelsohn and Sabbadini. Genetics in Medicine, 2019, 21, 763-763.	2.4	1
36	Secondary findings in exome slices, virtual panels, and anticipatory sequencing. Genetics in Medicine, 2019, 21, 41-43.	2.4	13

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37	Response to Nakaguma et al.. Genetics in Medicine, 2019, 21, 261.	2.4	1
38	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
39	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. Current Protocols in Human Genetics, 2019, 103, e93.	3.5	88
40	Challenges to informed consent for exome sequencing: A bestâ€“worst scaling experiment. Journal of Genetic Counseling, 2019, 28, 1189-1197.	1.6	7
41	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
42	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
43	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
44	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
45	A mouse model of Proteus syndrome. Human Molecular Genetics, 2019, 28, 2920-2936.	2.9	11
46	Genome Sequencing and Individual Responses to Results. , 2019, , 17-30.		0
47	Judgment and Decision Making in Genome Sequencing. , 2019, , 57-73.		1
48	Uncertainties in Genome Sequencing. , 2019, , 75-88.		0
49	DENND5B Regulates Intestinal Triglyceride Absorption and Body Mass. Scientific Reports, 2019, 9, 3597.	3.3	10
50	Pharmacodynamic Study of Miransertib in Individuals with Proteus Syndrome. American Journal of Human Genetics, 2019, 104, 484-491.	6.2	56
51	Genomic screening and genomic diagnostic testingâ€“two very different kettles of fish. Genome Medicine, 2019, 11, 75.	8.2	11
52	Ethnic identity and engagement with genome sequencing research. Genetics in Medicine, 2019, 21, 1735-1743.	2.4	9
53	Response to Esplin et al.. Genetics in Medicine, 2019, 21, 1252-1253.	2.4	1
54	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. Genetics in Medicine, 2019, 21, 1189-1198.	2.4	115

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55	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. <i>Genetics in Medicine</i> , 2018, 20, 1175-1185.	2.4	133
56	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
57	Preferences for learning different types of genome sequencing results among young breast cancer patients: Role of psychological and clinical factors. <i>Translational Behavioral Medicine</i> , 2018, 8, 71-79.	2.4	29
58	Orthopaedic Management of Leg-length Discrepancy in Proteus Syndrome: A Case Series. <i>Journal of Pediatric Orthopaedics</i> , 2018, 38, e138-e144.	1.2	9
59	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
60	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
61	Mosaic disorders and the Taxonomy of Human Disease. <i>Genetics in Medicine</i> , 2018, 20, 800-801.	2.4	6
62	Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework. <i>Genetics in Medicine</i> , 2018, 20, 1054-1060.	2.4	366
63	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
64	The PiGeOn project: protocol for a longitudinal study examining psychosocial, behavioural and ethical issues and outcomes in cancer tumour genomic profiling. <i>BMC Cancer</i> , 2018, 18, 389.	2.6	10
65	The PiGeOn project: protocol of a longitudinal study examining psychosocial and ethical issues and outcomes in germline genomic sequencing for cancer. <i>BMC Cancer</i> , 2018, 18, 454.	2.6	14
66	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
67	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
68	Reactions to clinical reinterpretation of a gene variant by participants in a sequencing study. <i>Genetics in Medicine</i> , 2018, 20, 337-345.	2.4	14
69	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
70	Updated recommendation for the benign stand-alone ACMG/AMP criterion. <i>Human Mutation</i> , 2018, 39, 1525-1530.	2.5	102
71	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. <i>Human Mutation</i> , 2018, 39, 1517-1524.	2.5	511
72	Genomic screening for monogenic forms of diabetes. <i>BMC Medicine</i> , 2018, 16, 25.	5.5	5

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73	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
74	Ability of Patients to Distinguish Among Cardiac Genomic Variant Subclassifications. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e001975.	3.6	6
75	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
76	Mosaic RAS/MAPK variants cause sporadic vascular malformations which respond to targeted therapy. <i>Journal of Clinical Investigation</i> , 2018, 128, 1496-1508.	8.2	191
77	A taxonomy of medical uncertainties in clinical genome sequencing. <i>Genetics in Medicine</i> , 2017, 19, 918-925.	2.4	91
78	Characterization of thrombosis in patients with Proteus syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2359-2365.	1.2	26
79	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. <i>Cell</i> , 2017, 169, 6-12.	28.9	103
80	Personal utility in genomic testing: a systematic literature review. <i>European Journal of Human Genetics</i> , 2017, 25, 662-668.	2.8	122
81	Quantifying survival in patients with Proteus syndrome. <i>Genetics in Medicine</i> , 2017, 19, 1376-1379.	2.4	25
82	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
83	Engagement and communication among participants in the ClinSeq Genomic Sequencing Study. <i>Genetics in Medicine</i> , 2017, 19, 98-103.	2.4	3
84	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
85	Recommendations for the integration of genomics into clinical practice. <i>Genetics in Medicine</i> , 2016, 18, 1075-1084.	2.4	125
86	Assessing the reproducibility of exome copy number variations predictions. <i>Genome Medicine</i> , 2016, 8, 82.	8.2	44
87	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
88	Elevated basal serum tryptase identifies a multisystem disorder associated with increased TPSAB1 copy number. <i>Nature Genetics</i> , 2016, 48, 1564-1569.	21.4	279
89	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
90	High incidence of unrecognized visceral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. <i>Genetics in Medicine</i> , 2016, 18, 41-48.	2.4	171

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91	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
92	Systematic Evaluation of Sanger Validation of Next-Generation Sequencing Variants. <i>Clinical Chemistry</i> , 2016, 62, 647-654.	3.2	154
93	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
94	West syndrome caused by homozygous variant in the evolutionary conserved gene encoding the mitochondrial elongation factor GUF1. <i>European Journal of Human Genetics</i> , 2016, 24, 1001-1008.	2.8	10
95	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
96	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
97	Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. <i>American Journal of Human Genetics</i> , 2015, 96, 913-925.	6.2	66
98	<i>PIK3CA</i> -related overgrowth spectrum (PROS): Diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 287-295.	1.2	399
99	Molecular characterization of a cohort of 73 patients with infantile spasms syndrome. <i>European Journal of Medical Genetics</i> , 2015, 58, 51-58.	1.3	56
100	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
101	A Point Mutation in <i>PDGFRB</i> Causes Autosomal-Dominant Penttinen Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 465-474.	6.2	64
102	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. <i>Genome Research</i> , 2015, 25, 305-315.	5.5	313
103	Social and behavioral research in genomic sequencing: approaches from the Clinical Sequencing Exploratory Research Consortium Outcomes and Measures Working Group. <i>Genetics in Medicine</i> , 2014, 16, 727-735.	2.4	60
104	Identification of candidate genes involved in coronary artery calcification by transcriptome sequencing of cell lines. <i>BMC Genomics</i> , 2014, 15, 198.	2.8	13
105	A patient with Cantu syndrome associated with fatal bronchopulmonary dysplasia and pulmonary hypertension. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2118-2120.	1.2	14
106	Somatic mosaicism for the p.His1047Arg mutation in <i>PIK3CA</i> in a girl with mesenteric lipomatosis. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2360-2364.	1.2	13
107	Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 120-128.	1.2	28
108	Complex mosaic <i>CDKL5</i> deletion with two distinct mutant alleles in a 4-year-old girl. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2025-2028.	1.2	6

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109	Macrocephallism, epilepsy, intellectual disability, and gut malrotation in a child with a 16q24.1â€“q24.2 contiguous gene deletion. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2062-2068.	1.2	10
110	Hepatomegaly and hyperammonemia in a girl with Silverâ€“Russell syndrome caused by maternal uniparental isodisomy of chromosome 7. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2114-2117.	1.2	1
111	Clinical delineation and natural history of the <i>PIK3CA</i>-related overgrowth spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1713-1733.	1.2	249
112	Integrative DNA, RNA, and Protein Evidence Connects TREML4 to Coronary Artery Calcification. <i>American Journal of Human Genetics</i> , 2014, 95, 66-76.	6.2	30
113	Diagnostic Clinical Genome and Exome Sequencing. <i>New England Journal of Medicine</i> , 2014, 370, 2418-2425.	27.0	488
114	A Limited Form of Proteus Syndrome With Bilateral Plantar Cerebriform Collagenomas and Varicose Veins Secondary to a Mosaic<i>AKT1</i> Mutation. <i>JAMA Dermatology</i> , 2014, 150, 990.	4.1	16
115	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. <i>Genetics in Medicine</i> , 2013, 15, 565-574.	2.4	2,186
116	Incidental Variants Are Critical for Genomics. <i>American Journal of Human Genetics</i> , 2013, 92, 648-651.	6.2	37
117	Intentions to receive individual results from whole-genome sequencing among participants in the ClinSeq study. <i>European Journal of Human Genetics</i> , 2013, 21, 261-265.	2.8	156
118	A genomic view of mosaicism and human disease. <i>Nature Reviews Genetics</i> , 2013, 14, 307-320.	16.3	527
119	Elements of morphology: Standard terminology for the external genitalia. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1238-1263.	1.2	16
120	The Nirvana Fallacy and the Return of Results. <i>American Journal of Bioethics</i> , 2013, 13, 43-44.	0.9	9
121	Associations between risk perceptions and worry about common diseases: A between- and within-subjects examination. <i>Psychology and Health</i> , 2013, 28, 434-449.	2.2	39
122	Hypothesis-generating research and predictive medicine. <i>Genome Research</i> , 2013, 23, 1051-1053.	5.5	66
123	VarSifter: Visualizing and analyzing exome-scale sequence variation data on a desktop computer. <i>Bioinformatics</i> , 2012, 28, 599-600.	4.1	137
124	Mosaic overgrowth with fibroadipose hyperplasia is caused by somatic activating mutations in PIK3CA. <i>Nature Genetics</i> , 2012, 44, 928-933.	21.4	269
125	Secondary Variants in Individuals Undergoing Exome Sequencing: Screening of 572 Individuals Identifies High-Penetrance Mutations in Cancer-Susceptibility Genes. <i>American Journal of Human Genetics</i> , 2012, 91, 97-108.	6.2	190
126	Opportunities and challenges for the integration of massively parallel genomic sequencing into clinical practice: lessons from the ClinSeq project. <i>Genetics in Medicine</i> , 2012, 14, 393-398.	2.4	145

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127	Recessive congenital methemoglobinemia caused by a rare mechanism: Maternal uniparental heterodisomy with segmental isodisomy of a chromosome 22. <i>Blood Cells, Molecules, and Diseases</i> , 2012, 49, 114-117.	1.4	7
128	A Mosaic Activating Mutation in <i>AKT1</i> Associated with the Proteus Syndrome. <i>New England Journal of Medicine</i> , 2011, 365, 611-619.	27.0	800
129	Exome sequencing identifies ACSF3 as a cause of combined malonic and methylmalonic aciduria. <i>Nature Genetics</i> , 2011, 43, 883-886.	21.4	89
130	Assessment and management of the orthopedic and other complications of Proteus syndrome. <i>Journal of Children's Orthopaedics</i> , 2011, 5, 319-327.	1.1	33
131	Implementing the elements of morphology in the <i>American Journal of Medical Genetics</i> . <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 969-971.	1.2	4
132	Long-term survival in TARP syndrome and confirmation of <i>RBM10</i> as the disease-causing gene. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2516-2520.	1.2	56
133	Motivators for participation in a whole-genome sequencing study: implications for translational genomics research. <i>European Journal of Human Genetics</i> , 2011, 19, 1213-1217.	2.8	129
134	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2010, 86, 749-764.	6.2	2,325
135	Nationwide survey of extended newborn screening by tandem mass spectrometry in Taiwan. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 295-305.	3.6	128
136	Epigenetic profiling of the <i>H19</i> differentially methylated region and comprehensive whole genome array-based analysis in Silver-Russell syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2521-2528.	1.2	14
137	Validation of My Family Health Portrait for six common heritable conditions. <i>Genetics in Medicine</i> , 2010, 12, 370-375.	2.4	72
138	Progressive overgrowth of the cerebriform connective tissue nevus in patients with Proteus syndrome. <i>Journal of the American Academy of Dermatology</i> , 2010, 63, 799-804.	1.2	43
139	Elements of morphology: Standard terminology for the hands and feet. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 93-127.	1.2	89
140	Elements of morphology: Introduction. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2-5.	1.2	98
141	The ClinSeq Project: Piloting large-scale genome sequencing for research in genomic medicine. <i>Genome Research</i> , 2009, 19, 1665-1674.	5.5	236
142	Newly delineated syndrome of congenital lipomatous overgrowth, vascular malformations, and epidermal nevi (CLOVE syndrome) in seven patients. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2944-2958.	1.2	201
143	The challenges of Proteus syndrome: diagnosis and management. <i>European Journal of Human Genetics</i> , 2006, 14, 1151-1157.	2.8	231
144	Genetic susceptibility testing for Alzheimer disease: Motivation to obtain information and control as precursors to coping with increased risk. <i>Patient Education and Counseling</i> , 2006, 64, 259-267.	2.2	54

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145	Reassessment of the Proteus syndrome literature: Application of diagnostic criteria to published cases. <i>American Journal of Medical Genetics Part A</i> , 2004, 130A, 111-122.	2.4	195
146	Considerations for a multi-axis nomenclature system for medical genetics. <i>Genetics in Medicine</i> , 2001, 3, 290-293.	2.4	32
147	Proteus syndrome: Diagnostic criteria, differential diagnosis, and patient evaluation. <i>American Journal of Medical Genetics Part A</i> , 1999, 84, 389-395.	2.4	374
148	Clinical differentiation between proteus syndrome and hemihyperplasia: Description of a distinct form of hemihyperplasia. , 1998, 79, 311-318.		97
149	Severe anomalies associated with ring chromosome 7. <i>American Journal of Medical Genetics Part A</i> , 1991, 40, 429-431.	2.4	16
150	Dizygotic twins concordant for truncus arteriosus. <i>Clinical Genetics</i> , 1991, 39, 75-79.	2.0	12