Heidi L Rehm

List of Publications by Year
in descending order

244 papers	45,081 citations	12597 71 h-index	2750 198 g-index
286 all docs	286 docs citations	286 times ranked	52118 citing authors

#	Article	IF	Citations
1	Utilizing ClinGen geneâ€disease validity and dosage sensitivity curations to inform variant classification. Human Mutation, 2022, 43, 1031-1040.	1.1	20
2	Reanalysis of eMERGE phase III sequence variants in 10,500 participants and infrastructure to support the automated return of knowledge updates. Genetics in Medicine, 2022, 24, 454-462.	1.1	6
3	Variant interpretation using population databases: Lessons from gnomAD. Human Mutation, 2022, 43, 1012-1030.	1.1	184
4	JAK inhibition in a patient with a STAT1 gain-of-function variant reveals STAT1 dysregulation as a common feature of aplastic anemia. Med, 2022, 3, 42-57.e5.	2.2	11
5	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	2.4	59
6	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.	3.6	34
7	Time to make rare disease diagnosis accessible to all. Nature Medicine, 2022, 28, 241-242.	15.2	19
8	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
9	An Investigation of the Knowledge Overlap between Pharmacogenomics and Disease Genetics. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2022, 27, 385-396.	0.7	1
10	Whole-genome sequencing as an investigational device for return of hereditary disease risk and pharmacogenomic results as part of the All of Us Research Program. Genome Medicine, 2022, 14, 34.	3.6	27
11	seqr : A webâ€based analysis and collaboration tool for rare disease genomics. Human Mutation, 2022, , .	1.1	31
12	Evaluating the impact of in silico predictors on clinical variant classification. Genetics in Medicine, 2022, 24, 924-930.	1.1	20
13	Monogenic and Polygenic Contributions to QTc Prolongation in the Population. Circulation, 2022, 145, 1524-1533.	1.6	14
14	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	1.7	48
15	Harmonizing variant classification for return of results in the All of Us Research Program. Human Mutation, 2022, 43, 1114-1121.	1.1	7
16	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	3.4	25
17	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. Cell Genomics, 2022, 2, 100131.	3.0	11
18	Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1392-1406.	1.1	18

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19	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking. Human Mutation, 2022, 43, 659-667.	1.1	11
20	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	1.1	56
21	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. Genetics in Medicine, 2022, 24, 1697-1707.	1.1	14
22	Abstract 1192: The Clinical Genome Resource (ClinGen) somatic cancer clinical domain working group. Cancer Research, 2022, 82, 1192-1192.	0.4	0
23	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	3.6	65
24	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.	1.1	1
25	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.4	2
26	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	1.1	10
27	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. European Journal of Human Genetics, 2021, 29, 988-997.	1.4	8
28	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	2.6	14
29	Universal newborn genetic screening for pediatric cancer predisposition syndromes: model-based insights. Genetics in Medicine, 2021, 23, 1366-1371.	1.1	16
30	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	1.1	47
31	The intersection of genetics and COVID-19 in 2021: preview of the 2021 Rodney Howell Symposium. Genetics in Medicine, 2021, 23, 1001-1003.	1.1	6
32	Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1399-1415.	1.1	64
33	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. Genetics in Medicine, 2021, 23, 1689-1696.	1.1	17
34	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. Genetics in Medicine, 2021, 23, 1738-1745.	1.1	7
35	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. Frontiers in Genetics, 2021, 12, 674295.	1.1	23
36	Genomic considerations for FHIRÂ®; eMERGE implementation lessons. Journal of Biomedical Informatics, 2021, 118, 103795.	2.5	15

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37	Primary care providersâ€™ responses to unsolicited Lynch syndrome secondary findings of varying clinical significance. Genetics in Medicine, 2021, 23, 1977-1983.	1.1	4
38	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	1.1	3
39	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	1.1	18
40	KCND2 variants associated with global developmental delay differentially impair Kv4.2 channel gating. Human Molecular Genetics, 2021, 30, 2300-2314.	1.4	12
41	Rare Coding Variants Associated With Electrocardiographic Intervals Identify Monogenic Arrhythmia Susceptibility Genes: A Multi-Ancestry Analysis. Circulation Genomic and Precision Medicine, 2021, 14, e003300.	1.6	7
42	Problems with Using Polygenic Scores to Select Embryos. New England Journal of Medicine, 2021, 385, 78-86.	13.9	105
43	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	3.3	35
44	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	3.7	19
45	Creation of an Expert Curated Variant List for Clinical Genomic Test Development and Validation. Journal of Molecular Diagnostics, 2021, 23, 1500-1505.	1.2	2
46	Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792.	0.7	6
47	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. American Journal of Human Genetics, 2021, 108, 2224-2237.	2.6	34
48	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
49	The GA4GH Variation Representation Specification: A computational framework for variation representation and federated identification. Cell Genomics, 2021, 1, 100027.	3.0	18
50	International federation of genomic medicine databases using GA4GH standards. Cell Genomics, 2021, 1, 100032.	3.0	22
51	An Investigation of the Knowledge Overlap between Pharmacogenomics and Disease Genetics. , 2021, , .		1
52	TMPRSS3 Gene Variants With Implications for Auditory Treatment and Counseling. Frontiers in Genetics, 2021, 12, 780874.	1.1	10
53	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	13.7	441
54	Keeping up with the genomes: scaling genomic variant interpretation. Genome Medicine, 2020, 12, 5.	3.6	13

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55	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. European Journal of Human Genetics, 2020, 28, 587-596.	1.4	38
56	From Theory to Reality: Establishing a Successful Kidney Genetics Clinic in the Outpatient Setting. Kidney360, 2020, 1, 1099-1106.	0.9	18
57	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	2.6	25
58	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.	2.6	51
59	The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. Genome Medicine, 2020, 12, 48.	3.6	40
60	Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477.	1.1	61
61	Variant Interpretation for Dilated Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e002480.	1.6	70
62	Management of Secondary Genomic Findings. American Journal of Human Genetics, 2020, 107, 3-14.	2.6	29
63	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.1	6
64	Diagnoses of uncertain significance: kidney genetics in the 21st century. Nature Reviews Nephrology, 2020, 16, 616-618.	4.1	16
65	How many rare diseases are there?. Nature Reviews Drug Discovery, 2020, 19, 77-78.	21.5	204
66	LB-11. Comparison of Viral Loads in Individuals With or Without Symptoms At Time of COVID-19 Testing Among 32,480 Residents and Staff of Nursing Homes and Assisted Living Facilities in Massachusetts. Open Forum Infectious Diseases, 2020, 7, S848-S849.	0.4	7
67	Genetic variation in the Middle Eastâ€"an opportunity to advance the human genetics field. Genome Medicine, 2020, 12, 116.	3.6	27
68	Development of a consent resource for genomic data sharing in the clinical setting. Genetics in Medicine, 2019, 21, 81-88.	1.1	20
69	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. Current Protocols in Human Genetics, 2019, 103, e93.	3.5	88
70	Rates of Actionable Genetic Findings in Individuals with Colorectal Cancer or Polyps Ascertained from a Community Medical Setting. American Journal of Human Genetics, 2019, 105, 526-533.	2.6	4
71	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. American Journal of Human Genetics, 2019, 105, 177-188.	2.6	38
72	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. Journal of the American College of Cardiology, 2019, 74, 2623-2634.	1.2	27

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73	Returning a Genomic Result for an Adult-Onset Condition to the Parents of a Newborn: Insights From the BabySeq Project. Pediatrics, 2019, 143, S37-S43.	1.0	45
74	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	2.6	99
75	A survey assessing adoption of the ACMG-AMP guidelines for interpreting sequence variants and identification of areas for continued improvement. Genetics in Medicine, 2019, 21, 1699-1701.	1.1	35
76	Introduction of genomics into prenatal diagnostics. Lancet, The, 2019, 393, 719-721.	6.3	13
77	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencingâ€“Detected Variants with an Orthogonal MethodÂin Clinical Genetic Testing. Journal of Molecular Diagnostics, 2019, 21, 318-329.	1.2	49
78	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	1.1	56
79	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	1.1	45
80	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
81	The Responsibility to Recontact Research Participants after Reinterpretation of Genetic and Genomic Research Results. American Journal of Human Genetics, 2019, 104, 578-595.	2.6	91
82	TBC1D8B Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2019, 30, 2338-2353.	3.0	25
83	Is â€˜likely pathogenicâ€™ really 90% likely? Reclassification data in ClinVar. Genome Medicine, 2019, 11, 72.	3.6	78
84	Misattributed parentage as an unanticipated finding during exome/genome sequencing: current clinical laboratory practices and an opportunity for standardization. Genetics in Medicine, 2019, 21, 861-866.	1.1	14
85	Development of Clinical Domain Working Groups for the Clinical Genome Resource (ClinGen): lessons learned and plans for the future. Genetics in Medicine, 2019, 21, 987-993.	1.1	17
86	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	2.6	176
87	A whole genome approach for discovering the genetic basis of blood group antigens: independent confirmation for P1 and Xg^a. Transfusion, 2019, 59, 908-915.	0.8	13
88	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
89	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.	1.1	111
90	Research Directions in the Clinical Implementation of Pharmacogenomics: An Overview of US Programs and Projects. Clinical Pharmacology and Therapeutics, 2018, 103, 778-786.	2.3	110

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91	Association of Racial/Ethnic Categories With the Ability of Genetic Tests to Detect a Cause of Cardiomyopathy. JAMA Cardiology, 2018, 3, 341.	3.0	83
92	Analysis of intragenic USH2A copy number variation unveils broad spectrum of unique and recurrent variants. European Journal of Medical Genetics, 2018, 61, 621-626.	0.7	9
93	Points to consider for sharing variant-level information from clinical genetic testing with ClinVar. Journal of Physical Education and Sports Management, 2018, 4, a002345.	0.5	23
94	Allele-Specific Droplet Digital PCR Combined with a Next-Generation Sequencing-Based Algorithm for Diagnostic Copy Number Analysis in Genes with High Homology: Proof of Concept Using Stereocilin. Clinical Chemistry, 2018, 64, 705-714.	1.5	24
95	Peter Bauer, Ellen Karges, Gabriela Oprea and Arndt Rolfs. Genetics in Medicine, 2018, 20, 378-379.	1.1	0
96	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.	1.1	13
97	The Lifespan of Genetic Testing. American Journal of Medicine, 2018, 131, 991-992.	0.6	0
98	Short-term costs of integrating whole-genome sequencing into primary care and cardiology settings: a pilot randomized trial. Genetics in Medicine, 2018, 20, 1544-1553.	1.1	25
99	Response to Biesecker and Harrison. Genetics in Medicine, 2018, 20, 1689-1690.	1.1	7
100	Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations. Prenatal Diagnosis, 2018, 38, 26-32.	1.1	47
101	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). Genetics in Medicine, 2018, 20, 294-302.	1.1	27
102	Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. Genetics in Medicine, 2018, 20, 536-544.	1.1	18
103	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in Medicine, 2018, 20, 855-866.	1.1	22
104	Rapid communication of efforts to resolve differences or update variant interpretations in ClinVar through case-level data sharing. Journal of Physical Education and Sports Management, 2018, 4, a003467.	0.5	2
105	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148
106	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	1.1	20
107	ClinGen's GenomeConnect registry enables patientâ€centered data sharing. Human Mutation, 2018, 39, 1668-1676.	1.1	25
108	ClinGen advancing genomic dataâ€sharing standards as a GA4GH driver project. Human Mutation, 2018, 39, 1686-1689.	1.1	15

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109	Updated recommendation for the benign standâ€alone ACMG/AMP criterion. Human Mutation, 2018, 39, 1525-1530.	1.1	102
110	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	1.1	132
111	Distinguishing Variant Pathogenicity From Genetic Diagnosis. JAMA - Journal of the American Medical Association, 2018, 320, 1929.	3.8	32
112	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	1.1	312
113	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. Human Mutation, 2018, 39, 1641-1649.	1.1	50
114	ClinGen and ClinVar â€“ Enabling Genomics in Precision Medicine. Human Mutation, 2018, 39, 1473-1475.	1.1	14
115	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. Human Mutation, 2018, 39, 1517-1524.	1.1	511
116	Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. Molecular Genetics & Genomic Medicine, 2018, 6, 898-909.	0.6	15
117	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. Lancet Haematology,the, 2018, 5, e241-e251.	2.2	70
118	The Ancestral Pace of Variant Reclassification. Journal of the National Cancer Institute, 2018, 110, 1133-1134.	3.0	7
119	ClinVar Miner: Demonstrating utility of a Web-based tool for viewing and filtering ClinVar data. Human Mutation, 2018, 39, 1051-1060.	1.1	81
120	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138.	3.0	42
121	Lack Of Diversity In Genomic Databases Is A Barrier To Translating Precision Medicine Research Into Practice. Health Affairs, 2018, 37, 780-785.	2.5	213
122	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	1.4	33
123	Reclassification of the BRAF p.Ile208Val variant by case-level data sharing. Journal of Physical Education and Sports Management, 2018, 4, a002675.	0.5	4
124	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	0.7	115
125	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	0.5	7
126	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361.	3.0	147

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127	Curating Clinically Relevant Transcripts for the Interpretation of Sequence Variants. Journal of Molecular Diagnostics, 2018, 20, 789-801.	1.2	25
128	ClinGen Pathogenicity Calculator: a configurable system for assessing pathogenicity of genetic variants. Genome Medicine, 2017, 9, 3.	3.6	59
129	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	1.0	174
130	Evolving health care through personal genomics. Nature Reviews Genetics, 2017, 18, 259-267.	7.7	98
131	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	1.1	79
132	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
133	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. Genetics in Medicine, 2017, 19, 1245-1252.	1.1	43
134	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	1.1	13
135	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. American Journal of Human Genetics, 2017, 100, 895-906.	2.6	403
136	Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar. Genetics in Medicine, 2017, 19, 1096-1104.	1.1	200
137	Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings. Journal of Molecular Diagnostics, 2017, 19, 417-426.	1.2	19
138	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	62
139	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47
140	Toward Genetics-Driven Early Intervention in Dilated Cardiomyopathy. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	41
141	The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients. Annals of Internal Medicine, 2017, 167, 159.	2.0	145
142	A new era in the interpretation of human genomic variation. Genetics in Medicine, 2017, 19, 1092-1095.	1.1	34
143	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	1.1	68
144	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. Genetics in Medicine, 2017, 19, 496-504.	1.1	15

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145	Standardizing terms for clinical pharmacogenetic test results: consensus terms from the Clinical Pharmacogenetics Implementation Consortium (CPIC). Genetics in Medicine, 2017, 19, 215-223.	1.1	410
146	Creating a data resource: what will it take to build a medical information commons?. Genome Medicine, 2017, 9, 84.	3.6	36
147	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. BMJ Paediatrics Open, 2017, 1, e000119.	0.6	16
148	Rapid access to genetic discoveries underlying human disease. Journal of Physical Education and Sports Management, 2016, 2, a001545.	0.5	0
149	Evaluation: A Qualitative Pilot Study of Novel Information Technology Infrastructure to Communicate Genetic Variant Updates. Applied Clinical Informatics, 2016, 07, 461-476.	0.8	10
150	Information Technology Support for Clinical Genetic Testing within an Academic Medical Center. Journal of Personalized Medicine, 2016, 6, 4.	1.1	7
151	The Changing Landscape of Molecular Diagnostic Testing: Implications for Academic Medical Centers. Journal of Personalized Medicine, 2016, 6, 8.	1.1	15
152	Consent Codes: Upholding Standard Data Use Conditions. PLoS Genetics, 2016, 12, e1005772.	1.5	65
153	Development and Validation of a Mass Spectrometryâ€“Based Assay for the Molecular Diagnosis of Mucin-1 Kidney Disease. Journal of Molecular Diagnostics, 2016, 18, 566-571.	1.2	25
154	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126.	1.1	37
155	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. American Journal of Human Genetics, 2016, 98, 1067-1076.	2.6	432
156	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	2.6	137
157	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	1.1	125
158	Health Care Infrastructure for Financially Sustainable Clinical Genomics. Journal of Molecular Diagnostics, 2016, 18, 697-706.	1.2	15
159	Comprehensive red blood cell and platelet antigen prediction from whole genome sequencing: proof of principle. Transfusion, 2016, 56, 743-754.	0.8	81
160	Genetic Misdiagnoses and the Potential for Health Disparities. New England Journal of Medicine, 2016, 375, 655-665.	13.9	602
161	Aggregate penetrance of genomic variants for actionable disorders in European and African Americans. Science Translational Medicine, 2016, 8, 364ra151.	5.8	55
162	CDH23 Related Hearing Loss. Otology and Neurotology, 2016, 37, 1583-1588.	0.7	17

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163	Using ClinVar as a Resource to Support Variant Interpretation. Current Protocols in Human Genetics, 2016, 89, 8.16.1-8.16.23.	3.5	89
164	Improving hearing loss gene testing: a systematic review of gene evidence toward more efficient next-generation sequencingâ€“based diagnostic testing and interpretation. Genetics in Medicine, 2016, 18, 545-553.	1.1	63
165	VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. Genetics in Medicine, 2016, 18, 712-719.	1.1	61
166	Global implementation of genomic medicine: We are not alone. Science Translational Medicine, 2015, 7, 290ps13.	5.8	146
167	ClinGen â€” The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	13.9	1,016
168	A One-Page Summary Report of Genome Sequencing for the Healthy Adult. Public Health Genomics, 2015, 18, 123-129.	0.6	37
169	GenomeConnect: Matchmaking Between Patients, Clinical Laboratories, and Researchers to Improve Genomic Knowledge. Human Mutation, 2015, 36, 974-978.	1.1	56
170	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	0.5	76
171	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. Genetics in Medicine, 2015, 17, 880-888.	1.1	344
172	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine, 2015, 17, 405-424.	1.1	20,455
173	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	9.4	134
174	All the World's a Stage: Facilitating Discovery Science and Improved Cancer Care through the Global Alliance for Genomics and Health. Cancer Discovery, 2015, 5, 1133-1136.	7.7	45
175	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
176	Building the foundation for genomics in precision medicine. Nature, 2015, 526, 336-342.	13.7	376
177	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	2.4	313
178	Summarizing polygenic risks for complex diseases in a clinical whole-genome report. Genetics in Medicine, 2015, 17, 536-544.	1.1	34
179	American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. Genetics in Medicine, 2014, 16, 347-355.	1.1	207
180	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. BMC Medical Genetics, 2014, 15, 134.	2.1	84

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