Heidi L Rehm

List of Publications by Year in Descending Order

241 papers	28,787 citations	63 h-index	168 g-index
286 ext. papers	37,850 ext. citations	10.4 avg, IF	6.93 L-index

#	Paper	IF	Citations
241	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	31.7	0
240	Mitochondrial DNA variation across 56,434 individuals in gnomAD.. *Genome Research*, **2022**,	9.7	3
239	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines.. *Genome Medicine*, **2022**, 14, 6	14.4	5
238	Time to make rare disease diagnosis accessible to all.. *Nature Medicine*, **2022**,	50.5	2
237	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
236	An Investigation of the Knowledge Overlap between Pharmacogenomics and Disease Genetics. *Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing*, **2022**, 27, 385-396	1.3	0
235	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	14.4	0
234	seqr: a web-based analysis and collaboration tool for rare disease genomics.. *Human Mutation*, **2022** ,	4.7	1
233	Best practices for the interpretation and reporting of clinical whole genome sequencing.. *Npj Genomic Medicine*, **2022**, 7, 27	6.2	1
232	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases.. *JAMA Oncology*, **2022**,	13.4	2
231	Lumping versus splitting: How to approach defining a disease to enable accurate genomic curation. *Cell Genomics*, **2022**, 2, 100131		0
230	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking.. *Human Mutation*, **2022**, 43, 659-667	4.7	0
229	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. *Genetics in Medicine*, **2022**,	8.1	4
228	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. *American Journal of Human Genetics*, **2021**, 108, 2224-2237	11	3
227	GA4GH: International policies and standards for data sharing across genomic research and healthcare.. *Cell Genomics*, **2021**, 1, 100029-100029		20
226	The GA4GH Variation Representation Specification: A computational framework for variation representation and federated identification.. *Cell Genomics*, **2021**, 1, 100027-100027		4
225	International federation of genomic medicine databases using GA4GH standards.. *Cell Genomics*, **2021**, 1, 100032-100032		7

224	TMPRSS3 Gene Variants With Implications for Auditory Treatment and Counseling. *Frontiers in Genetics*, **2021**, 12, 780874	4.5	3
223	Reanalysis of eMERGE phase III sequence variants in 10,500 participants and infrastructure to support the automated return of knowledge updates.. *Genetics in Medicine*, **2021**,	8.1	1
222	Utilizing ClinGen gene-disease validity and dosage sensitivity curations to inform variant classification. *Human Mutation*, **2021**,	4.7	2
221	Universal newborn genetic screening for pediatric cancer predisposition syndromes: model-based insights. *Genetics in Medicine*, **2021**, 23, 1366-1371	8.1	3
220	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. *Genetics in Medicine*, **2021**, 23, 1372-1375	8.1	11
219	The intersection of genetics and COVID-19 in 2021: preview of the 2021 Rodney Howell Symposium. *Genetics in Medicine*, **2021**, 23, 1001-1003	8.1	3
218	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	8.1	13
217	Randomized prospective evaluation of genome sequencing versus standard-of-care as a first molecular diagnostic test. *Genetics in Medicine*, **2021**, 23, 1689-1696	8.1	3
216	Recontacting registry participants with genetic updates through GenomeConnect, the ClinGen patient registry. *Genetics in Medicine*, **2021**, 23, 1738-1745	8.1	1
215	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. *Frontiers in Genetics*, **2021**, 12, 674295	4.5	2
214	Genomic considerations for FHIR ; eMERGE implementation lessons. *Journal of Biomedical Informatics*, **2021**, 118, 103795	10.2	5
213	Primary care providersPresponses to unsolicited Lynch syndrome secondary findings of varying clinical significance. *Genetics in Medicine*, **2021**, 23, 1977-1983	8.1	0
212	Neptune: an environment for the delivery of genomic medicine. *Genetics in Medicine*, **2021**, 23, 1838-1848	8.1	1
211	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. *Genetics in Medicine*, **2021**, 23, 2208-2212	8.1	0
210	KCND2 variants associated with global developmental delay differentially impair Kv4.2 channel gating. *Human Molecular Genetics*, **2021**, 30, 2300-2314	5.6	0
209	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
208	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. *Kidney International Reports*, **2021**, 6, 460-471	4.1	2
207	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. *Human Mutation*, **2021**, 42, 3-7	4.7	4

206	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	5.3	3
205	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	11	1
204	Rare Coding Variants Associated With Electrocardiographic Intervals Identify Monogenic Arrhythmia Susceptibility Genes: A Multi-Ancestry Analysis. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003300	5.2	0
203	Problems with Using Polygenic Scores to Select Embryos. *New England Journal of Medicine*, **2021**, 385, 78-86	59.2	23
202	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project: A Randomized Clinical Trial. *JAMA Pediatrics*, **2021**, 175, 1132-1141	8.3	8
201	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. *Brain*, **2021**, 144, 2659-2669	11.2	2
200	Creation of an Expert Curated Variant List for Clinical Genomic Test Development and Validation: A ClinGen and GeT-RM Collaborative Project. *Journal of Molecular Diagnostics*, **2021**, 23, 1500-1505	5.1	1
199	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	2.5	1
198	Evaluating the impact of in silico predictors on clinical variant classification.. *Genetics in Medicine*, **2021**,	8.1	2
197	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. *American Journal of Human Genetics*, **2020**, 107, 727-742	11	2
196	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
195	The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. *Genome Medicine*, **2020**, 12, 48	14.4	17
194	Frequency of genomic secondary findings among 21,915 eMERGE network participants. *Genetics in Medicine*, **2020**, 22, 1470-1477	8.1	23
193	Variant Interpretation for Dilated Cardiomyopathy: Refinement of the American College of Medical Genetics and Genomics/ClinGen Guidelines for the DCM Precision Medicine Study. *Circulation Genomic and Precision Medicine*, **2020**, 13, e002480	5.2	27
192	Management of Secondary Genomic Findings. *American Journal of Human Genetics*, **2020**, 107, 3-14	11	9
191	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. *Value in Health*, **2020**, 23, 559-565	3.3	4
190	Diagnoses of uncertain significance: kidney genetics in the 21st century. *Nature Reviews Nephrology* , **2020**, 16, 616-618	14.9	8
189	Genetic variation in the Middle East-an opportunity to advance the human genetics field. *Genome Medicine*, **2020**, 12, 116	14.4	9

188	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	1	4
187	A brief history of human disease genetics. *Nature*, **2020**, 577, 179-189	50.4	181
186	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. *European Journal of Human Genetics*, **2020**, 28, 587-596	5.3	21
185	From Theory to Reality: Establishing a Successful Kidney Genetics Clinic in the Outpatient Setting.. *Kidney360*, **2020**, 1, 1099-1106	1.8	3
184	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	7.4	22
183	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
182	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	8.1	16
181	Introduction of genomics into prenatal diagnostics. *Lancet, The*, **2019**, 393, 719-721	40	9
180	A Rigorous Interlaboratory Examination of the Need to Confirm Next-Generation Sequencing-Detected Variants with an Orthogonal Method in Clinical Genetic Testing. *Journal of Molecular Diagnostics*, **2019**, 21, 318-329	5.1	28
179	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	8.1	22
178	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. *Genetics in Medicine*, **2019**, 21, 2496-2503	8.1	29
177	ClinGen expert clinical validity curation of 164 hearing loss gene-disease pairs. *Genetics in Medicine*, **2019**, 21, 2239-2247	8.1	39
176	The Responsibility to Recontact Research Participants after Reinterpretation of Genetic and Genomic Research Results. *American Journal of Human Genetics*, **2019**, 104, 578-595	11	50
175	Development of a consent resource for genomic data sharing in the clinical setting. *Genetics in Medicine*, **2019**, 21, 81-88	8.1	12
174	Overview of Specifications to the ACMG/AMP Variant Interpretation Guidelines. *Current Protocols in Human Genetics*, **2019**, 103, e93	3.2	14
173	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	11	2
172	Analyzing and Reanalyzing the Genome: Findings from the MedSeq Project. *American Journal of Human Genetics*, **2019**, 105, 177-188	11	22
171	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. *Journal of the American College of Cardiology*, **2019**, 74, 2623-2634	15.1	17

170	Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2019**, 30, 2338-2353	12.7	11
169	Is P likely pathogenic really 90% likely? Reclassification data in ClinVar. *Genome Medicine*, **2019**, 11, 72	14.4	37
168	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	8.1	7
167	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	8.1	13
166	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. *American Journal of Human Genetics*, **2019**, 104, 76-93	11	86
165	A whole genome approach for discovering the genetic basis of blood group antigens: independent confirmation for P1 and Xg. *Transfusion*, **2019**, 59, 908-915	2.9	9
164	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
163	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
162	Research Directions in the Clinical Implementation of Pharmacogenomics: An Overview of US Programs and Projects. *Clinical Pharmacology and Therapeutics*, **2018**, 103, 778-786	6.1	63
161	Association of Racial/Ethnic Categories With the Ability of Genetic Tests to Detect a Cause of Cardiomyopathy. *JAMA Cardiology*, **2018**, 3, 341-345	16.2	40
160	Analysis of intragenic USH2A copy number variation unveils broad spectrum of unique and recurrent variants. *European Journal of Medical Genetics*, **2018**, 61, 621-626	2.6	6
159	Points to consider for sharing variant-level information from clinical genetic testing with ClinVar. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	17
158	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	5.5	13
157	Peter Bauer, Ellen Karges, Gabriela Oprea and Arndt Rolfs. *Genetics in Medicine*, **2018**, 20, 378-379	8.1	
156	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
155	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	8.1	17
154	Response to Biesecker and Harrison. *Genetics in Medicine*, **2018**, 20, 1689-1690	8.1	4
153	Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations. *Prenatal Diagnosis*, **2018**, 38, 26-32	3.2	36

152	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	8.1	20
151	Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. *Genetics in Medicine*, **2018**, 20, 536-544	8.1	14
150	and Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2123-2138	12.7	21
149	Lack Of Diversity In Genomic Databases Is A Barrier To Translating Precision Medicine Research Into Practice. *Health Affairs*, **2018**, 37, 780-785	7	99
148	Registered access: authorizing data access. *European Journal of Human Genetics*, **2018**, 26, 1721-1731	5.3	17
147	Reclassification of the p.Ile208Val variant by case-level data sharing. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	2
146	The BabySeq project: implementing genomic sequencing in newborns. *BMC Pediatrics*, **2018**, 18, 225	2.6	55
145	Reconciling newborn screening and a novel splice variant in associated with partial biotinidase deficiency: a BabySeq Project case report. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	4
144	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	12.7	75
143	Curating Clinically Relevant Transcripts for the Interpretation of Sequence Variants. *Journal of Molecular Diagnostics*, **2018**, 20, 789-801	5.1	18
142	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
141	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. *PLoS Genetics*, **2018**, 14, e1007752	6	90
140	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. *Human Mutation*, **2018**, 39, 1827-1834	4.7	13
139	ClinGen GenomeConnect registry enables patient-centered data sharing. *Human Mutation*, **2018**, 39, 1668-1676	4.7	14
138	ClinGen advancing genomic data-sharing standards as a GA4GH driver project. *Human Mutation*, **2018**, 39, 1686-1689	4.7	12
137	Updated recommendation for the benign stand-alone ACMG/AMP criterion. *Human Mutation*, **2018**, 39, 1525-1530	4.7	48
136	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and gene-level specification of the ACMG/AMP guidelines for sequence variant interpretation. *Human Mutation*, **2018**, 39, 1614-1622	4.7	83
135	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

134	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. *Human Mutation*, **2018**, 39, 1593-1613	4.7	167
133	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. *Human Mutation*, **2018**, 39, 1641-1649	4.7	28
132	ClinGen and ClinVar Enabling Genomics in Precision Medicine. *Human Mutation*, **2018**, 39, 1473-1475	4.7	10
131	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. *Human Mutation*, **2018**, 39, 1517-1524	4.7	215
130	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	2.3	10
129	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. *Lancet Haematology,the*, **2018**, 5, e241-e251	14.6	35
128	ClinVar Miner: Demonstrating utility of a Web-based tool for viewing and filtering ClinVar data. *Human Mutation*, **2018**, 39, 1051-1060	4.7	38
127	ClinGen Pathogenicity Calculator: a configurable system for assessing pathogenicity of genetic variants. *Genome Medicine*, **2017**, 9, 3	14.4	47
126	Newborn Sequencing in Genomic Medicine and Public Health. *Pediatrics*, **2017**, 139,	7.4	109
125	Evolving health care through personal genomics. *Nature Reviews Genetics*, **2017**, 18, 259-267	30.1	69
124	A curated gene list for reporting results of newborn genomic sequencing. *Genetics in Medicine*, **2017**, 19, 809-818	8.1	47
123	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705	11	200
122	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	8.1	33
121	"Matching" consent to purpose: The example of the Matchmaker Exchange. *Human Mutation*, **2017**, 38, 1281-1285	4.7	10
120	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	11	242
119	Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar. *Genetics in Medicine*, **2017**, 19, 1096-1104	8.1	137
118	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	5.1	13
117	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. *Circulation: Cardiovascular Genetics*, **2017**, 10,		45

116	Matchmaker Exchange. *Current Protocols in Human Genetics*, **2017**, 95, 9.31.1-9.31.15	3.2	32
115	Toward Genetics-Driven Early Intervention in Dilated Cardiomyopathy: Design and Implementation of the DCM Precision Medicine Study. *Circulation: Cardiovascular Genetics*, **2017**, 10,		25
114	The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients: A Pilot Randomized Trial. *Annals of Internal Medicine*, **2017**, 167, 159-169	8	112
113	A new era in the interpretation of human genomic variation. *Genetics in Medicine*, **2017**, 19, 1092-1095	8.1	27
112	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. *Genetics in Medicine*, **2017**, 19, 575-582	8.1	52
111	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. *Genetics in Medicine*, **2017**, 19, 496-504	8.1	11
110	Standardizing terms for clinical pharmacogenetic test results: consensus terms from the Clinical Pharmacogenetics Implementation Consortium (CPIC). *Genetics in Medicine*, **2017**, 19, 215-223	8.1	284
109	Creating a data resource: what will it take to build a medical information commons?. *Genome Medicine*, **2017**, 9, 84	14.4	27
108	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. *BMJ Paediatrics Open*, **2017**, 1, e000119	2.4	12
107	Genetic Misdiagnoses and the Potential for Health Disparities. *New England Journal of Medicine*, **2016**, 375, 655-65	59.2	394
106	Aggregate penetrance of genomic variants for actionable disorders in European and African Americans. *Science Translational Medicine*, **2016**, 8, 364ra151	17.5	41
105	CDH23 Related Hearing Loss: A New Genetic Risk Factor for Semicircular Canal Dehiscence?. *Otology and Neurotology*, **2016**, 37, 1583-1588	2.6	15
104	Using ClinVar as a Resource to Support Variant Interpretation. *Current Protocols in Human Genetics*, **2016**, 89, 8.16.1-8.16.23	3.2	70
103	Improving hearing loss gene testing: a systematic review of gene evidence toward more efficient next-generation sequencing-based diagnostic testing and interpretation. *Genetics in Medicine*, **2016** , 18, 545-53	8.1	44
102	VisCap: inference and visualization of germ-line copy-number variants from targeted clinical sequencing data. *Genetics in Medicine*, **2016**, 18, 712-9	8.1	46
101	Evaluation: A Qualitative Pilot Study of Novel Information Technology Infrastructure to Communicate Genetic Variant Updates. *Applied Clinical Informatics*, **2016**, 7, 461-76	3.1	10
100	Information Technology Support for Clinical Genetic Testing within an Academic Medical Center. *Journal of Personalized Medicine*, **2016**, 6,	3.6	7
99	The Changing Landscape of Molecular Diagnostic Testing: Implications for Academic Medical Centers. *Journal of Personalized Medicine*, **2016**, 6,	3.6	13

98	Consent Codes: Upholding Standard Data Use Conditions. *PLoS Genetics*, **2016**, 12, e1005772	6	51
97	Development and Validation of a Mass Spectrometry-Based Assay for the Molecular Diagnosis of Mucin-1 Kidney Disease. *Journal of Molecular Diagnostics*, **2016**, 18, 566-71	5.1	20
96	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. *Human Mutation*, **2016**, 37, 119-26	4.7	29
95	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	11	271
94	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. *American Journal of Human Genetics*, **2016**, 98, 1051-1066	11	107
93	Recommendations for the integration of genomics into clinical practice. *Genetics in Medicine*, **2016**, 18, 1075-1084	8.1	92
92	Health Care Infrastructure for Financially Sustainable Clinical Genomics. *Journal of Molecular Diagnostics*, **2016**, 18, 697-706	5.1	9
91	Comprehensive red blood cell and platelet antigen prediction from whole genome sequencing: proof of principle. *Transfusion*, **2016**, 56, 743-54	2.9	55
90	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, 405-24	8.1	11964
89	Good laboratory practice for clinical next-generation sequencing informatics pipelines. *Nature Biotechnology*, **2015**, 33, 689-93	44.5	115
88	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, 1133-6	24.4	35
87	The Matchmaker Exchange: a platform for rare disease gene discovery. *Human Mutation*, **2015**, 36, 915-21	4.7	280
86	Building the foundation for genomics in precision medicine. *Nature*, **2015**, 526, 336-42	50.4	290
85	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. *Genome Research*, **2015**, 25, 305-15	9.7	252
84	Summarizing polygenic risks for complex diseases in a clinical whole-genome report. *Genetics in Medicine*, **2015**, 17, 536-44	8.1	30
83	Global implementation of genomic medicine: We are not alone. *Science Translational Medicine*, **2015**, 7, 290ps13	17.5	112
82	ClinGen--the Clinical Genome Resource. *New England Journal of Medicine*, **2015**, 372, 2235-42	59.2	635
81	A one-page summary report of genome sequencing for the healthy adult. *Public Health Genomics*, **2015**, 18, 123-9	1.9	32

80	GenomeConnect: matchmaking between patients, clinical laboratories, and researchers to improve genomic knowledge. *Human Mutation*, **2015**, 36, 974-8	4.7	43
79	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, 388-96	3.7	56
78	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. *Genetics in Medicine*, **2015**, 17, 880-8	8.1	236
77	Return of genomic results to research participants: the floor, the ceiling, and the choices in between. *American Journal of Human Genetics*, **2014**, 94, 818-26	11	283
76	Guidelines for investigating causality of sequence variants in human disease. *Nature*, **2014**, 508, 469-76	50.4	910
75	Analysis of two Arab families reveals additional support for a DFNB2 nonsyndromic phenotype of MYO7A. *Molecular Biology Reports*, **2014**, 41, 193-200	2.8	8
74	Comprehensive diagnostic testing for stereocilin: an approach for analyzing medically important genes with high homology. *Journal of Molecular Diagnostics*, **2014**, 16, 639-47	5.1	41
73	The MedSeq Project: a randomized trial of integrating whole genome sequencing into clinical medicine. *Trials*, **2014**, 15, 85	2.8	103
72	The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. *Genetics in Medicine*, **2014**, 16, 601-8	8.1	215
71	American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. *Genetics in Medicine*, **2014**, 16, 347-55	8.1	147
70	A systematic approach to the reporting of medically relevant findings from whole genome sequencing. *BMC Medical Genetics*, **2014**, 15, 134	2.1	66
69	A novel clinician interface to improve clinician access to up-to-date genetic results. *Journal of the American Medical Informatics Association: JAMIA*, **2014**, 21, e117-21	8.6	16
68	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. *Genome Biology*, **2014**, 15, R53	18.3	86
67	Homozygosity for the V37I GJB2 mutation in fifteen probands with mild to moderate sensorineural hearing impairment: further confirmation of pathogenicity and haplotype analysis in Asian populations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2148-57	2.5	11
66	ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genetics in Medicine*, **2013**, 15, 565-74	8.1	1787
65	ACMG clinical laboratory standards for next-generation sequencing. *Genetics in Medicine*, **2013**, 15, 733-47	8.1	640
64	New approaches to molecular diagnosis. *JAMA - Journal of the American Medical Association*, **2013**, 309, 1511-21	27.4	91
63	PECONPI: a novel software for uncovering pathogenic copy number variations in non-syndromic sensorineural hearing loss and other genetically heterogeneous disorders. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2134-47	2.5	5

62	Inherited cardiomyopathies: molecular genetics and clinical genetic testing in the postgenomic era. *Journal of Molecular Diagnostics*, **2013**, 15, 158-70	5.1	139
61	Clinical Genome Sequencing **2013**, 102-122		24
60	Disease-targeted sequencing: a cornerstone in the clinic. *Nature Reviews Genetics*, **2013**, 14, 295-300	30.1	306
59	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 4667-72	11.5	151
58	Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. *Genetics in Medicine*, **2013**, 15, 860-7	8.1	95
57	Genome-wide SNP genotyping identifies the Stereocilin (STRC) gene as a major contributor to pediatric bilateral sensorineural hearing impairment. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 298-308	2.5	67
56	Usability of a novel clinician interface for genetic results. *Journal of Biomedical Informatics*, **2012**, 45, 950-7	10.2	28
55	Exploring concordance and discordance for return of incidental findings from clinical sequencing. *Genetics in Medicine*, **2012**, 14, 405-10	8.1	136
54	Genetic testing for dilated cardiomyopathy in clinical practice. *Journal of Cardiac Failure*, **2012**, 18, 296-303	3.3	118
53	Molecular diagnosis of hearing loss. *Current Protocols in Human Genetics*, **2012**, Chapter 9, Unit 9.16	3.2	14
52	Burden of rare sarcomere gene variants in the Framingham and Jackson Heart Study cohorts. *American Journal of Human Genetics*, **2012**, 91, 513-9	11	88
51	Assuring the quality of next-generation sequencing in clinical laboratory practice. *Nature Biotechnology*, **2012**, 30, 1033-6	44.5	372
50	Norrie disease: extraocular clinical manifestations in 56 patients. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1909-17	2.5	31
49	Communicating new knowledge on previously reported genetic variants. *Genetics in Medicine*, **2012**, 14, 713-719	8.1	77
48	A public resource facilitating clinical use of genomes. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 11920-7	11.5	154
47	Targeted sequencing using Affymetrix CustomSeq Arrays. *Current Protocols in Human Genetics*, **2011**, Chapter 7, Unit7.18	3.2	8
46	Allelic hierarchy of CDH23 mutations causing non-syndromic deafness DFNB12 or Usher syndrome USH1D in compound heterozygotes. *Journal of Medical Genetics*, **2011**, 48, 767-75	5.8	73
45	Retinal disease course in Usher syndrome 1B due to MYO7A mutations **2011**, 52, 7924-36		58

44	MutaDATABASE: a centralized and standardized DNA variation database. *Nature Biotechnology*, **2011**, 29, 117-8	44.5	29
43	Development and validation of a computational method for assessment of missense variants in hypertrophic cardiomyopathy. *American Journal of Human Genetics*, **2011**, 88, 183-92	11	60
42	Temporal bone abnormalities in children with GJB2 mutations. *Laryngoscope*, **2011**, 121, 630-5	3.6	16
41	The GeneInsight Suite: a platform to support laboratory and provider use of DNA-based genetic testing. *Human Mutation*, **2011**, 32, 532-6	4.7	69
40	Phased whole-genome genetic risk in a family quartet using a major allele reference sequence. *PLoS Genetics*, **2011**, 7, e1002280	6	112
39	Reply to Clarity and claims in variation/mutation databasing. *Nature Biotechnology*, **2011**, 29, 792-794	44.5	
38	Short communication: the cardiac myosin binding protein C Arg502Trp mutation: a common cause of hypertrophic cardiomyopathy. *Circulation Research*, **2010**, 106, 1549-52	15.7	56
37	A novel custom resequencing array for dilated cardiomyopathy. *Genetics in Medicine*, **2010**, 12, 268-78	8.1	62
36	Translating research discoveries into clinical tests. *Seminars in Nephrology*, **2010**, 30, 426-30	4.8	
35	Use and interpretation of genetic tests in cardiovascular genetics. *Heart*, **2010**, 96, 1669-75	5.1	34
34	Evaluation of second-generation sequencing of 19 dilated cardiomyopathy genes for clinical applications. *Journal of Molecular Diagnostics*, **2010**, 12, 818-27	5.1	37
33	Audiologic phenotype and progression in GJB2 (Connexin 26) hearing loss. *JAMA Otolaryngology*, **2010**, 136, 81-7		54
32	High-throughput detection of mutations responsible for childhood hearing loss using resequencing microarrays. *BMC Biotechnology*, **2010**, 10, 10	3.5	41
31	Disease boundaries in the retina of patients with Usher syndrome caused by MYO7A gene mutations **2009**, 50, 1886-94		75
30	An overview of custom array sequencing. *Current Protocols in Human Genetics*, **2009**, Chapter 7, Unit 7.17	3.2	23
29	Implications of hypertrophic cardiomyopathy transmitted by sperm donation. *JAMA - Journal of the American Medical Association*, **2009**, 302, 1681-4	27.4	29
28	Filter-based hybridization capture of subgenomes enables resequencing and copy-number detection. *Nature Methods*, **2009**, 6, 507-10	21.6	51
27	Shared genetic causes of cardiac hypertrophy in children and adults. *New England Journal of Medicine*, **2008**, 358, 1899-908	59.2	288

26	Additional clinical manifestations in children with sensorineural hearing loss and biallelic GJB2 mutations: who should be offered GJB2 testing?. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1560-6	2.5	21
25	Short-term effects of intravitreal bevacizumab for subfoveal choroidal neovascularization in pathologic myopia. *Genetics in Medicine*, **2007**, 27, 707-12	8.1	108
24	A multicenter study of the frequency and distribution of GJB2 and GJB6 mutations in a large North American cohort. *Genetics in Medicine*, **2007**, 9, 413-26	8.1	33
23	GJB2 mutations and degree of hearing loss: a multicenter study. *American Journal of Human Genetics*, **2005**, 77, 945-57	11	372
22	A genetic approach to the child with sensorineural hearing loss. *Seminars in Perinatology*, **2005**, 29, 173-81	3.3	19
21	The PTPN11 gene is not implicated in nonsyndromic hypertrophic cardiomyopathy **2005**, 132A, 333-4		9
20	Audiologic features of Norrie disease. *Annals of Otology, Rhinology and Laryngology*, **2005**, 114, 533-8	2.1	23
19	TRPA1 is a candidate for the mechanosensitive transduction channel of vertebrate hair cells. *Nature*, **2004**, 432, 723-30	50.4	576
18	Molecular diagnosis of hearing loss. *Current Protocols in Human Genetics*, **2004**, Chapter 9, Unit9.16	3.2	
17	Genetics and the genome project. *Ear and Hearing*, **2003**, 24, 270-4	3.4	16
16	Early childhood hearing loss: clinical and molecular genetics. An educational slide set of the American College of Medical Genetics. *Genetics in Medicine*, **2003**, 5, 338-41	8.1	2
15	Vascular defects and sensorineural deafness in a mouse model of Norrie disease. *Journal of Neuroscience*, **2002**, 22, 4286-92	6.6	116
14	Connexin 26 studies in patients with sensorineural hearing loss. *JAMA Otolaryngology*, **2001**, 127, 1037-42		115
13	Mutation of a gene encoding a protein with extracellular matrix motifs in Usher syndrome type IIa. *Science*, **1998**, 280, 1753-7	33.3	321
12	Norrie disease gene mutation in a large Costa Rican kindred with a novel phenotype including venous insufficiency. *Human Mutation*, **1997**, 9, 402-8	4.7	28
11	Mono- and bi-allelic effects of coding variants on disease in 176,899 Finns		2
10	Curating clinically relevant transcripts for the interpretation of sequence variants		1
9	Consensus interpretation of the Met34Thr and Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel		1

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