Peter Nick Robinson

List of Publications by Year
in descending order

287	23,017	10650 74	13635 134
papers	citations	h-index	g-index

350	350	350	30778
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Curation and expansion of Human Phenotype Ontology for defined groups of inborn errors of immunity. Journal of Allergy and Clinical Immunology, 2022, 149, 369-378.	1.5	16
2	Betacoronavirus-specific alternate splicing. Genomics, 2022, 114, 110270.	1.3	12
3	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	1.1	18
4	Demonstrating an approach for evaluating synthetic geospatial and temporal epidemiologic data utility: results from analyzing >1.8 million SARS-CoV-2 tests in the United States National COVID Cohort Collaborative (N3C). Journal of the American Medical Informatics Association: JAMIA, 2022, 29, 1350-1365.	2.2	8
5	Association of Early Aspirin Use With In-Hospital Mortality in Patients With Moderate COVID-19. JAMA Network Open, 2022, 5, e223890.	2.8	31
6	Phenotypeâ€driven approaches to enhance variant prioritization and diagnosis of rare disease. Human Mutation, 2022, 43, 1071-1081.	1.1	17
7	PhenoRerank: A re-ranking model for phenotypic concept recognition pre-trained on human phenotype ontology. Journal of Biomedical Informatics, 2022, 129, 104059.	2.5	2
8	The Clinical Variant Analysis Tool: Analyzing the evidence supporting reported genomic variation in clinical practice. Genetics in Medicine, 2022, 24, 1512-1522.	1.1	4
9	Evaluation of phenotype-driven gene prioritization methods for Mendelian diseases. Briefings in Bioinformatics, 2022, 23, .	3.2	6
10	FABIAN-variant: predicting the effects of DNA variants on transcription factor binding. Nucleic Acids Research, 2022, 50, W322-W329.	6.5	12
11	PDXNet portal: patient-derived Xenograft model, data, workflow and tool discovery. NAR Cancer, 2022, 4, zcac014.	1.6	7
12	SvAnna: efficient and accurate pathogenicity prediction of coding and regulatory structural variants in long-read genome sequencing. Genome Medicine, 2022, 14, 44.	3.6	7
13	Deep phenotyping: symptom annotation made simple with SAMS. Nucleic Acids Research, 2022, 50, W677-W681.	6.5	5
14	Risk of newâ€onset psychiatric sequelae of <scp>COVID</scp>â€19 in the early and late postâ€acute phase. World Psychiatry, 2022, 21, 319-320.	4.8	15
15	NSAID use and clinical outcomes in COVID-19 patients: a 38-center retrospective cohort study. Virology Journal, 2022, 19, 84.	1.4	19
16	The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820.	9.4	38
17	The National COVID Cohort Collaborative (N3C): Rationale, design, infrastructure, and deployment. Journal of the American Medical Informatics Association: JAMIA, 2021, 28, 427-443.	2.2	342
18	KG-COVID-19: A Framework to Produce Customized Knowledge Graphs for COVID-19 Response. Patterns, 2021, 2, 100155.	3.1	62

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19	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	6.5	652
20	PhenoTagger: a hybrid method for phenotype concept recognition using human phenotype ontology. Bioinformatics, 2021, 37, 1884-1890.	1.8	18
21	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	1.4	4
22	Modeling seizures in the Human Phenotype Ontology according to contemporary ILAE concepts makes big phenotypic data tractable. Epilepsia, 2021, 62, 1293-1305.	2.6	15
23	E2F6 initiates stable epigenetic silencing of germline genes during embryonic development. Nature Communications, 2021, 12, 3582.	5.8	21
24	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
25	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
26	HEMDAG: a family of modular and scalable hierarchical ensemble methods to improve Gene Ontology term prediction. Bioinformatics, 2021, 37, 4526-4533.	1.8	2
27	Comprehensive characterization of 536 patient-derived xenograft models prioritizes candidates for targeted treatment. Nature Communications, 2021, 12, 5086.	5.8	58
28	Interpretable prioritization of splice variants in diagnostic next-generation sequencing. American Journal of Human Genetics, 2021, 108, 1564-1577.	2.6	36
29	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
30	A CRISPR-Cas9â€"engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	8
31	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
32	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
33	The GA4GH Variation Representation Specification: A computational framework for variation representation and federated identification. Cell Genomics, 2021, 1, 100027.	3.0	18
34	Abdominal Computed Tomography Imaging Findings in Hospitalized COVID-19 Patients: A Year-Long Experience and Associations Revealed by Explainable Artificial Intelligence. Journal of Imaging, 2021, 7, 258.	1.7	2
35	Characterizing Long COVID: Deep Phenotype of a Complex Condition. EBioMedicine, 2021, 74, 103722.	2.7	127
36	Supervised learning with word embeddings derived from PubMed captures latent knowledge about protein kinases and cancer. NAR Genomics and Bioinformatics, 2021, 3, lqab113.	1.5	4

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37	The Monarch Initiative in 2019: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2020, 48, D704-D715.	6.5	178
38	The case for open science: rare diseases. JAMIA Open, 2020, 3, 472-486.	1.0	33
39	HBA-DEALS: accurate and simultaneous identification of differential expression and splicing using hierarchical Bayesian analysis. Genome Biology, 2020, 21, 171.	3.8	7
40	parSMURF, a high-performance computing tool for the genome-wide detection of pathogenic variants. GigaScience, 2020, 9, .	3.3	11
41	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	1.2	21
42	Modelling kidney disease using ontology: insights from the Kidney Precision Medicine Project. Nature Reviews Nephrology, 2020, 16, 686-696.	4.1	45
43	Ontologies, Knowledge Representation, and Machine Learning for Translational Research: Recent Contributions. Yearbook of Medical Informatics, 2020, 29, 159-162.	0.8	14
44	Explainable Machine Learning for Early Assessment of COVID-19 Risk Prediction in Emergency Departments. IEEE Access, 2020, 8, 196299-196325.	2.6	55
45	Interpretable Clinical Genomics with a Likelihood Ratio Paradigm. American Journal of Human Genetics, 2020, 107, 403-417.	2.6	56
46	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. Genetics in Medicine, 2020, 22, 1391-1400.	1.1	2
47	Significantly different clinical phenotypes associated with mutations in synthesis and transamidase+remodeling glycosylphosphatidylinositol (GPI)-anchor biosynthesis genes. Orphanet Journal of Rare Diseases, 2020, 15, 40.	1.2	21
48	Pervasive and CpG-dependent promoter-like characteristics of transcribed enhancers. Nucleic Acids Research, 2020, 48, 5306-5317.	6.5	24
49	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	1.0	42
50	Supplementation of the ESID registry working definitions for the clinical diagnosis of inborn errors of immunity with encoded human phenotype ontology (HPO) terms. Journal of Allergy and Clinical Immunology: in Practice, 2020, 8, 1778.	2.0	8
51	How many rare diseases are there?. Nature Reviews Drug Discovery, 2020, 19, 77-78.	21.5	204
52	Encoding Clinical Data with the Human Phenotype Ontology for Computational Differential Diagnostics. Current Protocols in Human Genetics, 2019, 103, e92.	3.5	29
53	Computational Processing and Quality Control of Hi-C, Capture Hi-C and Capture-C Data. Genes, 2019, 10, 548.	1.0	5
54	An integrative systems approach identifies novel candidates in Marfan syndromeâ€related pathophysiology. Journal of Cellular and Molecular Medicine, 2019, 23, 2526-2535.	1.6	17

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55	An ontological foundation for ocular phenotypes and rare eye diseases. Orphanet Journal of Rare Diseases, 2019, 14, 8.	1.2	18
56	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	1.1	58
57	Semantic integration of clinical laboratory tests from electronic health records for deep phenotyping and biomarker discovery. Npj Digital Medicine, 2019, 2, .	5.7	39
58	Ensembling Descendant Term Classifiers to Improve Gene - Abnormal Phenotype Predictions. Lecture Notes in Computer Science, 2019, , 70-80.	1.0	2
59	Estimating heritability and genetic correlations from large health datasets in the absence of genetic data. Nature Communications, 2019, 10, 5508.	5.8	17
60	Representing glycophenotypes: semantic unification of glycobiology resources for disease discovery. Database: the Journal of Biological Databases and Curation, 2019, 2019, .	1.4	5
61	Assessment of Bones Deficient in Fibrillin-1 Microfibrils Reveals Pronounced Sex Differences. International Journal of Molecular Sciences, 2019, 20, 6059.	1.8	8
62	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
63	GOPHER: Generator Of Probes for capture Hi-C Experiments at high Resolution. BMC Genomics, 2019, 20, 40.	1.2	10
64	An Integrated Understanding of the Molecular Mechanisms of How Adipose Tissue Metabolism Affects Long-term Body Weight Maintenance. Diabetes, 2019, 68, 57-65.	0.3	23
65	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	9.4	28
66	Transcriptional profiling of murine osteoblast differentiation based on RNA-seq expression analyses. Bone, 2018, 113, 29-40.	1.4	13
67	Harmonising phenomics information for a better interoperability in the rare disease field. European Journal of Medical Genetics, 2018, 61, 706-714.	0.7	29
68	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
69	Improving service delivery for neuromuscular diseases: a survey of consumers at a tertiary Australian hospital. Internal Medicine Journal, 2018, 48, 1520-1524.	0.5	0
70	Classification, Ontology, and Precision Medicine. New England Journal of Medicine, 2018, 379, 1452-1462.	13.9	220
71	A Census of Disease Ontologies. Annual Review of Biomedical Data Science, 2018, 1, 305-331.	2.8	29
72	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	3.6	67

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73	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
74	Once doesn't count: Phenotype-driven gene hunting in cohorts. Human Mutation, 2017, 38, 469-469.	1.1	0
75	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
76	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	0.7	36
77	Imbalance-Aware Machine Learning for Predicting Rare and Common Disease-Associated Non-Coding Variants. Scientific Reports, 2017, 7, 2959.	1.6	63
78	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. Cell, 2017, 169, 6-12.	13.5	103
79	Interpreting Phenotypic Features of Bicuspid Aortic Valve Disease: From Simplification to Complexity to Simplicity?. American Journal of Medicine, 2017, 130, e315-e316.	0.6	3
80	The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2017, 45, D712-D722.	6.5	306
81	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
82	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Advances in Experimental Medicine and Biology, 2017, 1031, 55-94.	0.8	20
83	AP-1 Oligodeoxynucleotides Reduce Aortic Elastolysis in a Murine Model of Marfan Syndrome. Molecular Therapy - Nucleic Acids, 2017, 9, 69-79.	2.3	15
84	A likelihood ratio-based method to predict exact pedigrees for complex families from next-generation sequencing data. Bioinformatics, 2017, 33, 72-78.	1.8	8
85	Defining Disease, Diagnosis, and Translational Medicine within a Homeostatic Perturbation Paradigm: The National Institutes of Health Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2017, 4, 62.	1.2	23
86	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. BioMed Research International, 2017, 2017, 1-13.	0.9	28
87	Biometric and structural ocular manifestations of Marfan syndrome. PLoS ONE, 2017, 12, e0183370.	1.1	54
88	Genome-Wide Binding of Posterior HOXA/D Transcription Factors Reveals Subgrouping and Association with CTCF. PLoS Genetics, 2017, 13, e1006567.	1.5	38
89	Prediction of Human Phenotype Ontology terms by means of hierarchical ensemble methods. BMC Bioinformatics, 2017, 18, 449.	1.2	22
90	Identification of a molecular defect in a stillborn fetus with perinatal lethal hypophosphatasia using a disease-associated genome sequencing approach. Polish Journal of Pathology, 2016, 1, 78-83.	0.1	3

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91	Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. Human Mutation, 2016, 37, 737-744.	1.1	46
92	Alternate-locus aware variant calling in whole genome sequencing. Genome Medicine, 2016, 8, 130.	3.6	16
93	A systematic, large-scale comparison of transcription factor binding site models. BMC Genomics, 2016, 17, 388.	1.2	15
94	An expanded evaluation of protein function prediction methods shows an improvement in accuracy. Genome Biology, 2016, 17, 184.	3.8	308
95	The digital revolution in phenotyping. Briefings in Bioinformatics, 2016, 17, 819-830.	3.2	41
96	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	2.6	223
97	Navigating the Phenotype Frontier: The Monarch Initiative. Genetics, 2016, 203, 1491-1495.	1.2	65
98	Q-nexus: a comprehensive and efficient analysis pipeline designed for ChIP-nexus. BMC Genomics, 2016, 17, 873.	1.2	12
99	Animal-based studies will be essential for precision medicine. Science Translational Medicine, 2016, 8, 352ed12.	5.8	19
100	Tools for exploring mouse models of human disease. Drug Discovery Today: Disease Models, 2016, 20, 21-26.	1.2	0
101	NT-proBNP and diastolic left ventricular function in patients with Marfan syndrome. IJC Heart and Vasculature, 2016, 12, 15-20.	0.6	7
102	Clinical utility gene card for: Hereditary thoracic aortic aneurysm and dissection including next-generation sequencing-based approaches. European Journal of Human Genetics, 2016, 24, 146-150.	1.4	28
103	Marfanoidâ€“progeroidâ€“lipodystrophy syndrome: a newly recognized fibrillinopathy. European Journal of Human Genetics, 2016, 24, 1244-1247.	1.4	29
104	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. Genetics in Medicine, 2016, 18, 608-617.	1.1	85
105	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	1.1	107
106	The Genomic Birthday Paradox: How Much Is Enough?. Human Mutation, 2015, 36, 989-997.	1.1	13
107	Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. Human Mutation, 2015, 36, 979-984.	1.1	36
108	Crowdsourced direct-to-consumer genomic analysis of a family quartet. BMC Genomics, 2015, 16, 910.	1.2	20

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109	Phenotype-driven strategies for exome prioritization of human Mendelian disease genes. Genome Medicine, 2015, 7, 81.	3.6	97
110	PhenoMiner: from text to a database of phenotypes associated with OMIM diseases. Database: the Journal of Biological Databases and Curation, 2015, 2015, bav104.	1.4	29
111	Automatic concept recognition using the Human Phenotype Ontology reference and test suite corpora. Database: the Journal of Biological Databases and Curation, 2015, 2015, bav005-bav005.	1.4	55
112	Analysis of Strengths, Weaknesses, Opportunities, and Threats as a Tool for Translating Evidence into Individualized Medical Strategies (I-SWOT). Aorta, 2015, 03, 98-107.	0.1	24
113	Perspectives on the revised Ghent criteria for the diagnosis of Marfan syndrome. The Application of Clinical Genetics, 2015, 8, 137.	1.4	120
114	IMSEQâ€"a fast and error aware approach to immunogenetic sequence analysis. Bioinformatics, 2015, 31, 2963-2971.	1.8	98
115	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	1.4	85
116	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. Genome Medicine, 2015, 7, 36.	3.6	119
117	Capturing phenotypes for precision medicine. Journal of Physical Education and Sports Management, 2015, 1, a000372.	0.5	32
118	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	5.5	296
119	Missense variant in CCDC22 causes X-linked recessive intellectual disability with features of Ritscher-Schinzel/3C syndrome. European Journal of Human Genetics, 2015, 23, 633-638.	1.4	42
120	Finding Our Way through Phenotypes. PLoS Biology, 2015, 13, e1002033.	2.6	178
121	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. American Journal of Human Genetics, 2015, 97, 111-124.	2.6	203
122	FGFR2 mutation in a patient without typical features of Pfeiffer syndrome â€“ The emerging role of combined NGS and phenotype based strategies. European Journal of Medical Genetics, 2015, 58, 376-380.	0.7	9
123	Phenolyzer: phenotype-based prioritization of candidate genes for human diseases. Nature Methods, 2015, 12, 841-843.	9.0	327
124	Saturation analysis of ChIP-seq data for reproducible identification of binding peaks. Genome Research, 2015, 25, 1391-1400.	2.4	24
125	Somatic neurofibromatosis type 1 (NF1) inactivation events in cutaneous neurofibromas of a single NF1 patient. European Journal of Human Genetics, 2015, 23, 870-873.	1.4	20
126	Capturing domain knowledge from multiple sources: the rare bone disorders use case. Journal of Biomedical Semantics, 2015, 6, 21.	0.9	2

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127	Prediction of Human Gene - Phenotype Associations by Exploiting the Hierarchical Structure of the Human Phenotype Ontology. Lecture Notes in Computer Science, 2015, , 66-77.	1.0	3
128	Disease insights through cross-species phenotype comparisons. Mammalian Genome, 2015, 26, 548-555.	1.0	19
129	Phenotyping: Targeting genotype's rich cousin for diagnosis. Journal of Paediatrics and Child Health, 2015, 51, 381-386.	0.4	29
130	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
131	Strategies to improve the performance of rare variant association studies by optimizing the selection of controls. Bioinformatics, 2015, 31, btv457.	1.8	0
132	Human genotypeâ€“phenotype databases: aims, challenges and opportunities. Nature Reviews Genetics, 2015, 16, 702-715.	7.7	100
133	MiR-497â^¼195 Cluster MicroRNAs Regulate Osteoblast Differentiation by Targeting BMP Signaling. Journal of Bone and Mineral Research, 2015, 30, 796-808.	3.1	65
134	A Hierarchical Ensemble Method for DAG-Structured Taxonomies. Lecture Notes in Computer Science, 2015, , 15-26.	1.0	8
135	Differential effect of cataract-associated mutations in MAF on transactivation of MAF target genes. Molecular and Cellular Biochemistry, 2014, 396, 137-145.	1.4	11
136	The main pulmonary artery in adults: a controlled multicenter study with assessment of echocardiographic reference values, and the frequency of dilatation and aneurysm in Marfan syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 203.	1.2	34
137	Clinical interpretation of CNVs with cross-species phenotype data. Journal of Medical Genetics, 2014, 51, 766-772.	1.5	23
138	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. Bioinformatics, 2014, 30, 3215-3222.	1.8	91
139	Phenotype Ontologies and Cross-Species Analysis for Translational Research. PLoS Genetics, 2014, 10, e1004268.	1.5	63
140	Genomic data sharing for translational research and diagnostics. Genome Medicine, 2014, 6, 78.	3.6	8
141	Total Serum Transforming Growth Factorâ€î²1 Is Elevated in the Entire Spectrum of Genetic Aortic Syndromes. Clinical Cardiology, 2014, 37, 672-679.	0.7	36
142	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	2.4	300
143	Screening for single nucleotide variants, small indels and exon deletions with a nextâ€generation sequencing based gene panel approach for <scp>U</scp> sher syndrome. Molecular Genetics & Genomic Medicine, 2014, 2, 393-401.	0.6	22
144	Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. European Journal of Human Genetics, 2014, 22, 762-767.	1.4	39

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145	Use of animal models for exome prioritization of rare disease genes. Orphanet Journal of Rare Diseases, 2014, 9, O19.	1.2	0
146	Deletions of chromosomal regulatory boundaries are associated with congenital disease. Genome Biology, 2014, 15, 423.	3.8	144
147	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. Science Translational Medicine, 2014, 6, 252ra123.	5.8	223
148	Neonatal progeroid variant of Marfan syndrome with congenital lipodystrophy results from mutations at the 3â€² end of FBN1 gene. European Journal of Medical Genetics, 2014, 57, 230-234.	0.7	41
149	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	6.5	698
150	Jannovar: A Java Library for Exome Annotation. Human Mutation, 2014, 35, 548-555.	1.1	63
151	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. Journal of General Internal Medicine, 2014, 29, 780-787.	1.3	159
152	Clinical phenotype-based gene prioritization: an initial study using semantic similarity and the human phenotype ontology. BMC Bioinformatics, 2014, 15, 248.	1.2	48
153	The influence of disease categories on gene candidate predictions from model organism phenotypes. Journal of Biomedical Semantics, 2014, 5, S4.	0.9	9
154	Comprehensive analysis of dural ectasia in 150 patients with a causative <scp>FBN1</scp> mutation. Clinical Genetics, 2014, 86, 238-245.	1.0	24
155	Mutations in PGAP3 Impair GPI-Anchor Maturation, Causing a Subtype of Hyperphosphatasia with Mental Retardation. American Journal of Human Genetics, 2014, 94, 278-287.	2.6	88
156	First description of a patient with Vici syndrome due to a mutation affecting the penultimate exon of EPG5 and review of the literature. American Journal of Medical Genetics, Part A, 2014, 164, 3170-3175.	0.7	33
157	When Should Surgery Be Performed in Marfan Syndrome and Other Connective Tissue Disorders to Protect Against Type A Dissection?. , 2014, , 17-47.		10
158	Computational Phenotype Analysis in Human Medicine. , 2014, , 8-23.		3
159	Estimating exome genotyping accuracy by comparing to data from large scale sequencing projects. Genome Medicine, 2013, 5, 69.	3.6	23
160	TCR Repertoire Analysis by Next Generation Sequencing Allows Complex Differential Diagnosis of T Cellâ€“Related Pathology. American Journal of Transplantation, 2013, 13, 2842-2854.	2.6	131
161	A case of paroxysmal nocturnal hemoglobinuria caused by a germline mutation and a somatic mutation in PIGT. Blood, 2013, 122, 1312-1315.	0.6	77
162	FBN1 gene mutation characteristics and clinical features for the prediction of mitral valve disease progression. International Journal of Cardiology, 2013, 168, 953-959.	0.8	15

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163	miR-181a promotes osteoblastic differentiation through repression of TGF-Î² signaling molecules. International Journal of Biochemistry and Cell Biology, 2013, 45, 696-705.	1.2	120
164	PGAP2 Mutations, Affecting the GPI-Anchor-Synthesis Pathway, Cause Hyperphosphatasia with Mental Retardation Syndrome. American Journal of Human Genetics, 2013, 92, 584-589.	2.6	98
165	Regulation of fibrillin-1 gene expression by Sp1. Gene, 2013, 527, 448-455.	1.0	7
166	Ascending aortic aneurysm and aortic valve dysfunction in bicuspid aortic valve disease. International Journal of Cardiology, 2013, 164, 301-305.	0.8	24
167	The fibrillin-1 hypomorphic mgR/mgR murine model of Marfan syndrome shows severe elastolysis in all segments of the aorta. Journal of Vascular Surgery, 2013, 57, 1628-1636.e3.	0.6	36
168	Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome. Journal of Experimental Medicine, 2013, 210, 433-443.	4.2	186
169	Doubly heterozygous LMNA and TTN mutations revealed by exome sequencing in a severe form of dilated cardiomyopathy. European Journal of Human Genetics, 2013, 21, 1105-1111.	1.4	86
170	Antagonism of GxxPG fragments ameliorates manifestations of aortic disease in Marfan syndrome mice. Human Molecular Genetics, 2013, 22, 433-443.	1.4	33
171	PhenoDigm: analyzing curated annotations to associate animal models with human diseases. Database: the Journal of Biological Databases and Curation, 2013, 2013, bat025-bat025.	1.4	115
172	Distinct global shifts in genomic binding profiles of limb malformation-associated HOXD13 mutations. Genome Research, 2013, 23, 2091-2102.	2.4	31
173	Phenotypic overlap in the contribution of individual genes to CNV pathogenicity revealed by cross-species computational analysis of single-gene mutations in humans, mice and zebrafish. DMM Disease Models and Mechanisms, 2013, 6, 358-72.	1.2	43
174	Indomethacin Prevents the Progression of Thoracic Aortic Aneurysm in Marfan Syndrome Mice. Aorta, 2013, 1, 5-12.	0.1	14
175	Filtering for Compound Heterozygous Sequence Variants in Non-Consanguineous Pedigrees. PLoS ONE, 2013, 8, e70151.	1.1	41
176	Observational Cohort Study of Ventricular Arrhythmia in Adults with Marfan Syndrome Caused by FBN1 Mutations. PLoS ONE, 2013, 8, e81281.	1.1	45
177	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30.	0.8	72
178	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30.	0.8	64
179	Integrating the human phenotype ontology into HeTOP terminology-ontology server. Studies in Health Technology and Informatics, 2013, 192, 961.	0.2	1
180	Bayesian ontology querying for accurate and noise-tolerant semantic searches. Bioinformatics, 2012, 28, 2502-2508.	1.8	55

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181	The allele distribution in next-generation sequencing data sets is accurately described as the result of a stochastic branching process. Nucleic Acids Research, 2012, 40, 2426-2431.	6.5	40
182	Mechanism for Release of Alkaline Phosphatase Caused by Glycosylphosphatidylinositol Deficiency in Patients with Hyperphosphatasia Mental Retardation Syndrome. Journal of Biological Chemistry, 2012, 287, 6318-6325.	1.6	82
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