Sebastian KÃ¶hler

List of Publications by Year
in descending order

55 papers	8,770 citations	147801 31 h-index	161849 54 g-index
72 all docs	72 docs citations	72 times ranked	11465 citing authors

#	Article	IF	Citations
1	Walking the Interactome for Prioritization of Candidate Disease Genes. American Journal of Human Genetics, 2008, 82, 949-958.	6.2	1,111
2	The Human Phenotype Ontology: A Tool for Annotating and Analyzing Human Hereditary Disease. American Journal of Human Genetics, 2008, 83, 610-615.	6.2	797
3	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
4	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
5	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	14.5	652
6	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
7	Clinical Diagnostics in Human Genetics with Semantic Similarity Searches in Ontologies. American Journal of Human Genetics, 2009, 85, 457-464.	6.2	444
8	The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2017, 45, D712-D722.	14.5	306
9	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	5.5	300
10	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	12.0	296
11	Identity-by-descent filtering of exome sequence data identifies PIGV mutations in hyperphosphatasia mental retardation syndrome. Nature Genetics, 2010, 42, 827-829.	21.4	286
12	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. Science Translational Medicine, 2014, 6, 252ra123.	12.4	223
13	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	6.2	223
14	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. American Journal of Human Genetics, 2015, 97, 111-124.	6.2	203
15	Finding Our Way through Phenotypes. PLoS Biology, 2015, 13, e1002033.	5.6	178
16	The Monarch Initiative in 2019: an integrative data and analytic platform connecting phenotypes to genotypes across species. Nucleic Acids Research, 2020, 48, D704-D715.	14.5	178
17	Deletions of chromosomal regulatory boundaries are associated with congenital disease. Genome Biology, 2014, 15, 423.	8.8	144
18	Characterizing Long COVID: Deep Phenotype of a Complex Condition. EBioMedicine, 2021, 74, 103722.	6.1	127

#	Article	IF	Citations
19	PhenoDigm: analyzing curated annotations to associate animal models with human diseases. Database: the Journal of Biological Databases and Curation, 2013, 2013, bat025-bat025.	3.0	115
20	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. Bioinformatics, 2014, 30, 3215-3222.	4.1	91
21	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. Genetics in Medicine, 2016, 18, 608-617.	2.4	85
22	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30.	1.6	72
23	Entity/quality-based logical definitions for the human skeletal phenome using PATO. , 2009, 2009, 7069-72.		67
24	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	8.2	67
25	Navigating the Phenotype Frontier: The Monarch Initiative. Genetics, 2016, 203, 1491-1495.	2.9	65
26	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. F1000Research, 2013, 2, 30.	1.6	64
27	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
28	Interpretable Clinical Genomics with a Likelihood Ratio Paradigm. American Journal of Human Genetics, 2020, 107, 403-417.	6.2	56
29	Bayesian ontology querying for accurate and noise-tolerant semantic searches. Bioinformatics, 2012, 28, 2502-2508.	4.1	55
30	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.	3.0	55
31	MouseFinder: Candidate disease genes from mouse phenotype data. Human Mutation, 2012, 33, 858-866.	2.5	53
32	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.	2.4	43
33	Semantic integration of clinical laboratory tests from electronic health records for deep phenotyping and biomarker discovery. Npj Digital Medicine, 2019, 2, .	10.9	39
34	Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. Human Mutation, 2015, 36, 979-984.	2.5	36
35	Improving ontologies by automatic reasoning and evaluation of logical definitions. BMC Bioinformatics, 2011, 12, 418.	2.6	29
36	Harmonising phenomics information for a better interoperability in the rare disease field. European Journal of Medical Genetics, 2018, 61, 706-714.	1.3	29

#	Article	IF	Citations
37	Encoding Clinical Data with the Human Phenotype Ontology for Computational Differential Diagnostics. Current Protocols in Human Genetics, 2019, 103, e92.	3.5	29
38	Ontological phenotype standards for neurogenetics. Human Mutation, 2012, 33, 1333-1339.	2.5	28
39	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	21.4	28
40	Clinical interpretation of CNVs with cross-species phenotype data. Journal of Medical Genetics, 2014, 51, 766-772.	3.2	23
41	Integrative analysis of genomic, functional and protein interaction data predicts long-range enhancer-target gene interactions. Nucleic Acids Research, 2011, 39, 2492-2502.	14.5	22
42	Disease insights through cross-species phenotype comparisons. Mammalian Genome, 2015, 26, 548-555.	2.2	19
43	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	14.5	17
44	Exact score distribution computation for ontological similarity searches. BMC Bioinformatics, 2011, 12, 441.	2.6	15
45	Short ultraconserved promoter regions delineate a class of preferentially expressed alternatively spliced transcripts. Genomics, 2009, 94, 308-316.	2.9	11
46	Improved ontology-based similarity calculations using a study-wise annotation model. Database: the Journal of Biological Databases and Curation, 2018, 2018, .	3.0	11
47	The influence of disease categories on gene candidate predictions from model organism phenotypes. Journal of Biomedical Semantics, 2014, 5, S4.	1.6	9
48	A Collection of Benchmark Data Sets for Knowledge Graph-based Similarity in the Biomedical Domain. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	3.0	9
49	A Hierarchical Ensemble Method for DAG-Structured Taxonomies. Lecture Notes in Computer Science, 2015, , 15-26.	1.3	8
50	Representing glycophenotypes: semantic unification of glycobiology resources for disease discovery. Database: the Journal of Biological Databases and Curation, 2019, 2019, .	3.0	5
51	Prediction of Human Gene - Phenotype Associations by Exploiting the Hierarchical Structure of the Human Phenotype Ontology. Lecture Notes in Computer Science, 2015, , 66-77.	1.3	3
52	Phenotero: Annotate as you write. Clinical Genetics, 2019, 95, 287-292.	2.0	3
53	Exact Score Distribution Computation for Similarity Searches in Ontologies. Lecture Notes in Computer Science, 2009, , 298-309.	1.3	3
54	A Collection of Benchmark Data Sets for Knowledge Graph-Based Similarity in the Biomedical Domain. Lecture Notes in Computer Science, 2020, , 50-55.	1.3	1

#	Article	IF	Citations
55	Use of animal models for exome prioritization of rare disease genes. Orphanet Journal of Rare Diseases, 2014, 9, O19.	2.7	0