

# Sebastian Kähler

## List of Publications by Year in descending order

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Version: 2024-02-01

55  
papers

8,770  
citations

168829

31  
h-index

182931

54  
g-index

72  
all docs

72  
docs citations

72  
times ranked

12649  
citing authors

#	ARTICLE	IF	CITATIONS
1	The Human Phenotype Ontology in 2021. <i>Nucleic Acids Research</i> , 2021, 49, D1207-D1217.	6.5	652
2	Characterizing Long COVID: Deep Phenotype of a Complex Condition. <i>EBioMedicine</i> , 2021, 74, 103722.	2.7	127
3	The Monarch Initiative in 2019: an integrative data and analytic platform connecting phenotypes to genotypes across species. <i>Nucleic Acids Research</i> , 2020, 48, D704-D715.	6.5	178
4	Interpretable Clinical Genomics with a Likelihood Ratio Paradigm. <i>American Journal of Human Genetics</i> , 2020, 107, 403-417.	2.6	56
5	A Collection of Benchmark Data Sets for Knowledge Graph-based Similarity in the Biomedical Domain. <i>Database: the Journal of Biological Databases and Curation</i> , 2020, 2020, .	1.4	9
6	A Collection of Benchmark Data Sets for Knowledge Graph-Based Similarity in the Biomedical Domain. <i>Lecture Notes in Computer Science</i> , 2020, , 50-55.	1.0	1
7	Encoding Clinical Data with the Human Phenotype Ontology for Computational Differential Diagnostics. <i>Current Protocols in Human Genetics</i> , 2019, 103, e92.	3.5	29
8	RegulationSpotter: annotation and interpretation of extratranscriptomic DNA variants. <i>Nucleic Acids Research</i> , 2019, 47, W106-W113.	6.5	17
9	PEDIA: prioritization of exome data by image analysis. <i>Genetics in Medicine</i> , 2019, 21, 2807-2814.	1.1	58
10	Semantic integration of clinical laboratory tests from electronic health records for deep phenotyping and biomarker discovery. <i>Npj Digital Medicine</i> , 2019, 2, .	5.7	39
11	Representing glycophenotypes: semantic unification of glycobiology resources for disease discovery. <i>Database: the Journal of Biological Databases and Curation</i> , 2019, 2019, .	1.4	5
12	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. <i>Nucleic Acids Research</i> , 2019, 47, D1018-D1027.	6.5	539
13	Phenotero: Annotate as you write. <i>Clinical Genetics</i> , 2019, 95, 287-292.	1.0	3
14	Plain-language medical vocabulary for precision diagnosis. <i>Nature Genetics</i> , 2018, 50, 474-476.	9.4	28
15	Harmonising phenomics information for a better interoperability in the rare disease field. <i>European Journal of Medical Genetics</i> , 2018, 61, 706-714.	0.7	29
16	Improved ontology-based similarity calculations using a study-wise annotation model. <i>Database: the Journal of Biological Databases and Curation</i> , 2018, 2018, .	1.4	11
17	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. <i>Genome Medicine</i> , 2018, 10, 3.	3.6	67
18	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	6.5	699

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19	The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. <i>Nucleic Acids Research</i> , 2017, 45, D712-D722.	6.5	306
20	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. <i>American Journal of Human Genetics</i> , 2016, 99, 595-606.	2.6	223
21	Navigating the Phenotype Frontier: The Monarch Initiative. <i>Genetics</i> , 2016, 203, 1491-1495.	1.2	65
22	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. <i>Genetics in Medicine</i> , 2016, 18, 608-617.	1.1	85
23	Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 979-984.	1.1	36
24	Automatic concept recognition using the Human Phenotype Ontology reference and test suite corpora. <i>Database: the Journal of Biological Databases and Curation</i> , 2015, 2015, bav005-bav005.	1.4	55
25	Next-generation diagnostics and disease-gene discovery with the Exomiser. <i>Nature Protocols</i> , 2015, 10, 2004-2015.	5.5	296
26	Finding Our Way through Phenotypes. <i>PLoS Biology</i> , 2015, 13, e1002033.	2.6	178
27	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. <i>American Journal of Human Genetics</i> , 2015, 97, 111-124.	2.6	203
28	Prediction of Human Gene - Phenotype Associations by Exploiting the Hierarchical Structure of the Human Phenotype Ontology. <i>Lecture Notes in Computer Science</i> , 2015, , 66-77.	1.0	3
29	Disease insights through cross-species phenotype comparisons. <i>Mammalian Genome</i> , 2015, 26, 548-555.	1.0	19
30	A Hierarchical Ensemble Method for DAG-Structured Taxonomies. <i>Lecture Notes in Computer Science</i> , 2015, , 15-26.	1.0	8
31	Clinical interpretation of CNVs with cross-species phenotype data. <i>Journal of Medical Genetics</i> , 2014, 51, 766-772.	1.5	23
32	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. <i>Bioinformatics</i> , 2014, 30, 3215-3222.	1.8	91
33	Improved exome prioritization of disease genes through cross-species phenotype comparison. <i>Genome Research</i> , 2014, 24, 340-348.	2.4	300
34	Use of animal models for exome prioritization of rare disease genes. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, O19.	1.2	0
35	Deletions of chromosomal regulatory boundaries are associated with congenital disease. <i>Genome Biology</i> , 2014, 15, 423.	3.8	144
36	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. <i>Science Translational Medicine</i> , 2014, 6, 252ra123.	5.8	223

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37	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. <i>Nucleic Acids Research</i> , 2014, 42, D966-D974.	6.5	698
38	The influence of disease categories on gene candidate predictions from model organism phenotypes. <i>Journal of Biomedical Semantics</i> , 2014, 5, S4.	0.9	9
39	PhenoDigm: analyzing curated annotations to associate animal models with human diseases. <i>Database: the Journal of Biological Databases and Curation</i> , 2013, 2013, bat025-bat025.	1.4	115
40	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. <i>DMM Disease Models and Mechanisms</i> , 2013, 6, 358-72.	1.2	43
41	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. <i>F1000Research</i> , 2013, 2, 30.	0.8	72
42	Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. <i>F1000Research</i> , 2013, 2, 30.	0.8	64
43	Bayesian ontology querying for accurate and noise-tolerant semantic searches. <i>Bioinformatics</i> , 2012, 28, 2502-2508.	1.8	55
44	Ontological phenotype standards for neurogenetics. <i>Human Mutation</i> , 2012, 33, 1333-1339.	1.1	28
45	MouseFinder: Candidate disease genes from mouse phenotype data. <i>Human Mutation</i> , 2012, 33, 858-866.	1.1	53
46	Improving ontologies by automatic reasoning and evaluation of logical definitions. <i>BMC Bioinformatics</i> , 2011, 12, 418.	1.2	29
47	Exact score distribution computation for ontological similarity searches. <i>BMC Bioinformatics</i> , 2011, 12, 441.	1.2	15
48	Integrative analysis of genomic, functional and protein interaction data predicts long-range enhancer-target gene interactions. <i>Nucleic Acids Research</i> , 2011, 39, 2492-2502.	6.5	22
49	Identity-by-descent filtering of exome sequence data identifies PIGV mutations in hyperphosphatasia mental retardation syndrome. <i>Nature Genetics</i> , 2010, 42, 827-829.	9.4	286
50	Entity/quality-based logical definitions for the human skeletal phenome using PATO. , 2009, 2009, 7069-72.		67
51	Clinical Diagnostics in Human Genetics with Semantic Similarity Searches in Ontologies. <i>American Journal of Human Genetics</i> , 2009, 85, 457-464.	2.6	444
52	Short ultraconserved promoter regions delineate a class of preferentially expressed alternatively spliced transcripts. <i>Genomics</i> , 2009, 94, 308-316.	1.3	11
53	Exact Score Distribution Computation for Similarity Searches in Ontologies. <i>Lecture Notes in Computer Science</i> , 2009, , 298-309.	1.0	3
54	Walking the Interactome for Prioritization of Candidate Disease Genes. <i>American Journal of Human Genetics</i> , 2008, 82, 949-958.	2.6	1,111

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55	The Human Phenotype Ontology: A Tool for Annotating and Analyzing Human Hereditary Disease. American Journal of Human Genetics, 2008, 83, 610-615.	2.6	797