Gudmundur A Thorisson

List of Publications by Year
in descending order

27 papers	13,513 citations	430874 18 h-index	642732 23 g-index
27 all docs	27 docs citations	27 times ranked	20144 citing authors

#	Article	IF	Citations
1	A homozygous loss-of-function mutation leading to CYBC1 deficiency causes chronic granulomatous disease. Nature Communications, 2018, 9, 4447.	12.8	95
2	Compound heterozygous mutations in UBA5 causing early-onset epileptic encephalopathy in two sisters. BMC Medical Genetics, 2017, 18, 103.	2.1	28
3	COPA syndrome in an Icelandic family caused by a recurrent missense mutation in COPA. BMC Medical Genetics, 2017, 18, 129.	2.1	47
4	ODIN: the ORCID and DataCite interoperability network. International Journal of Knowledge and Learning, 2014, 9, 305.	0.2	3
5	Quantifying the use of bioresources for promoting their sharing in scientific research. GigaScience, 2013, 2, 7.	6.4	38
6	Applying ontologies and exploring nanopublishing in a genome-wide association study database. , 2012, , .		0
7	VarioML framework for comprehensive variation data representation and exchange. BMC Bioinformatics, 2012, 13, 254.	2.6	17
8	Semantically enabling a genome-wide association study database. Journal of Biomedical Semantics, 2012, 3, 9.	1.6	8
9	Observ-OM and Observ-TAB: Universal syntax solutions for the integration, search, and exchange of phenotype and genotype information. Human Mutation, 2012, 33, 867-873.	2.5	18
10	The BRIF (Bioresource Research Impact Factor) as a tool for improving bioresource sharing in biomedical research. Nature Precedings, 2011, , .	0.1	0
11	Identity in research infrastructure and scientific communication: Report from the 1st IRISC workshop, Helsinki Sep 12-13, 2011. Nature Precedings, 2011, , .	0.1	0
12	The role of a bioresource research impact factor as an incentive to share human bioresources. Nature Genetics, 2011, 43, 503-504.	21.4	66
13	An informatics project and online â€œKnowledge Centreâ€•supporting modern genotype-to-phenotype research. Human Mutation, 2011, 32, 543-550.	2.5	35
14	The MOLGENIS toolkit: rapid prototyping of biosoftware at the push of a button. BMC Bioinformatics, 2010, 11, S12.	2.6	102
15	Finding and sharing: new approaches to registries of databases and services for the biomedical sciences. Database: the Journal of Biological Databases and Curation, 2010, 2010, baq014-baq014.	3.0	12
16	HGVbaseG2P: a central genetic association database. Nucleic Acids Research, 2009, 37, D797-D802.	14.5	60
17	BioMart â€“ biological queries made easy. BMC Genomics, 2009, 10, 22.	2.8	738
18	The phenotype and genotype experiment object model (PaGE-OM): a robust data structure for information related to DNA variation. Human Mutation, 2009, 30, 968-977.	2.5	18

#	Article	IF	Citations
19	Accreditation and attribution in data sharing. Nature Biotechnology, 2009, 27, 984-985.	17.5	21
20	Genotypeâ€“phenotype databases: challenges and solutions for the post-genomic era. Nature Reviews Genetics, 2009, 10, 9-18.	16.3	87
21	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	27.8	1,788
22	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	27.8	4,137
23	A haplotype map of the human genome. Nature, 2005, 437, 1299-1320.	27.8	5,440
24	The International HapMap Project Web site: Figure 1.. Genome Research, 2005, 15, 1592-1593.	5.5	497
25	The SNP Consortium website: past, present and future. Nucleic Acids Research, 2003, 31, 124-127.	14.5	150
26	WormBase: a cross-species database for comparative genomics. Nucleic Acids Research, 2003, 31, 133-137.	14.5	107
27	Identity in research infrastructure and scientific communication: Report from the 1st IRISC workshop, Helsinki Sep 12-13, 2011. Nature Precedings, 0, , .	0.1	1