

Ada Hamosh

List of Publications by Year in descending order

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Version: 2024-02-01

63
papers

12,488
citations

147566

31
h-index

133063

59
g-index

66
all docs

66
docs citations

66
times ranked

20356
citing authors

#	ARTICLE	IF	CITATIONS
1	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	1.1	44
2	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	1.1	11
3	The impact of GeneMatcher on international data sharing and collaboration. <i>Human Mutation</i> , 2022, , .	1.1	7
4	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking. <i>Human Mutation</i> , 2022, 43, 659-667.	1.1	11
5	The Gene Curation Coalition: A global effort to harmonize gene disease evidence resources. <i>Genetics in Medicine</i> , 2022, 24, 1732-1742.	1.1	56
6	The GA4GH Phenopacket schema defines a computable representation of clinical data. <i>Nature Biotechnology</i> , 2022, 40, 817-820.	9.4	38
7	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	0.7	17
8	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
9	Orthopaedic Problems in 35 Patients With Organic Acid Disorders. <i>Journal of Pediatric Orthopaedics</i> , 2021, 41, e457-e463.	0.6	0
10	Next-generation sequencing and the evolution of data sharing. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2633-2635.	0.7	4
11	Viewing Victor McKusick's legacy through the lens of his bibliography. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3212-3223.	0.7	1
12	Online Mendelian Inheritance in Man (OMIM®): Victor McKusick's magnum opus. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3259-3265.	0.7	63
13	Memories of Victor A. McKusick. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3377-3383.	0.7	1
14	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 365.	1.2	24
15	Festschrift for Victor A. McKusick on the Centenary of his Birth: Introduction. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3189-3192.	0.7	0
16	Response to Biesecker et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1807-1808.	2.6	3
17	Three decades of the Human Genome Organization. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3314-3321.	0.7	4
18	GA4GH: International policies and standards for data sharing across genomic research and healthcare. <i>Cell Genomics</i> , 2021, 1, 100029.	3.0	94

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19	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
20	<i>KMT2B</i>-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. <i>Brain</i> , 2020, 143, 3242-3261.	3.7	57
21	Whatâ€™s in a name? Issues to consider when naming Mendelian disorders. <i>Genetics in Medicine</i> , 2020, 22, 1573-1575.	1.1	13
22	The utility of exome sequencing for fetal pleural effusions. <i>Prenatal Diagnosis</i> , 2020, 40, 590-595.	1.1	9
23	Genomic Data Sharing for Novel Mendelian Disease Gene Discovery: The Matchmaker Exchange. <i>Annual Review of Genomics and Human Genetics</i> , 2020, 21, 305-326.	2.5	36
24	How many rare diseases are there?. <i>Nature Reviews Drug Discovery</i> , 2020, 19, 77-78.	21.5	204
25	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
26	Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 105, 48-64.	2.6	29
27	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	1.1	161
28	OMIM.org: leveraging knowledge across phenotypeâ€•gene relationships. <i>Nucleic Acids Research</i> , 2019, 47, D1038-D1043.	6.5	562
29	Professional responsibilities regarding the provision, publication, and dissemination of patient phenotypes in the context of clinical genetic and genomic testing: points to considerâ€•a statement of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2018, 20, 169-171.	1.1	13
30	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. <i>Genetics in Medicine</i> , 2017, 19, 45-52.	1.1	94
31	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	6.5	699
32	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	2.6	305
33	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. <i>Human Mutation</i> , 2017, 38, 1281-1285.	1.1	13
34	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. <i>European Journal of Human Genetics</i> , 2017, 25, 946-951.	1.4	33
35	Matchmaker Exchange. <i>Current Protocols in Human Genetics</i> , 2017, 95, 9.31.1-9.31.15.	3.5	47
36	Searching Online Mendelian Inheritance in Man (OMIM): A Knowledgebase of Human Genes and Genetic Phenotypes. <i>Current Protocols in Bioinformatics</i> , 2017, 58, 1.2.1-1.2.12.	25.8	378

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37	Integrating the microbiome into precision medicine. <i>Expert Review of Precision Medicine and Drug Development</i> , 2016, 1, 475-477.	0.4	2
38	<i>KIF5A</i> mutations cause an infantile onset phenotype including severe myoclonus with evidence of mitochondrial dysfunction. <i>Annals of Neurology</i> , 2016, 80, 633-637.	2.8	47
39	Genotype-phenotype correlation of congenital anomalies in multiple congenital anomalies hypotonia seizures syndrome (MCAHS1)/ <i>PIGN</i> -related epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 77-86.	0.7	41
40	The management of pregnancy and delivery in 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. , 2016, 170, 1600-1602.		7
41	<i>EPG5</i> -related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 2016, 139, 765-781.	3.7	99
42	GeneMatcher: A Matching Tool for Connecting Investigators with an Interest in the Same Gene. <i>Human Mutation</i> , 2015, 36, 928-930.	1.1	1,153
43	Assessment of incidental findings in 232 whole-exome sequences from the Baylor-Hopkins Center for Mendelian Genomics. <i>Genetics in Medicine</i> , 2015, 17, 782-788.	1.1	41
44	New Tools for Mendelian Disease Gene Identification: PhenoDB Variant Analysis Module; and GeneMatcher, a Web-Based Tool for Linking Investigators with an Interest in the Same Gene. <i>Human Mutation</i> , 2015, 36, 425-431.	1.1	141
45	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015, 97, 199-215.	2.6	574
46	OMIM.org: Online Mendelian Inheritance in Man (OMIM®), an online catalog of human genes and genetic disorders. <i>Nucleic Acids Research</i> , 2015, 43, D789-D798.	6.5	1,713
47	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 915-921.	1.1	390
48	Mutations in <i>SPATA5</i> Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. <i>American Journal of Human Genetics</i> , 2015, 97, 457-464.	2.6	134
49	Horizontal integration of OMIM across the medical school preclinical curriculum for early reinforcement of clinical genetics principles. <i>Genetics in Medicine</i> , 2015, 17, 158-163.	1.1	6
50	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. <i>Bioinformatics</i> , 2014, 30, 3215-3222.	1.8	91
51	Maternal Hyperphenylalaninemia: Rapid achievement of metabolic control predicts overall control throughout pregnancy. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 3-8.	0.5	6
52	PhenoDB: A New Web-Based Tool for the Collection, Storage, and Analysis of Phenotypic Features. <i>Human Mutation</i> , 2013, 34, 566-571.	1.1	64
53	The Centers for Mendelian Genomics: A new large-scale initiative to identify the genes underlying rare Mendelian conditions. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1523-1525.	0.7	110
54	A new face and new challenges for Online Mendelian Inheritance in Man (OMIM®). <i>Human Mutation</i> , 2011, 32, 564-567.	1.1	309

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55	McKusick's Online Mendelian Inheritance in Man (OMIM(R)). Nucleic Acids Research, 2009, 37, D793-D796.	6.5	623
56	Online Mendelian Inheritance in Man (OMIM), a knowledgebase of human genes and genetic disorders. Nucleic Acids Research, 2004, 33, D514-D517.	6.5	2,523
57	Online Mendelian Inheritance in Man (OMIM), a knowledgebase of human genes and genetic disorders. Nucleic Acids Research, 2002, 30, 52-55.	6.5	528
58	Online Mendelian Inheritance In Man (OMIM). Human Mutation, 2000, 15, 57-61.	1.1	503
59	Pachygyria in Weaver Syndrome. American Journal of Medical Genetics Part A, 1999, 86, 395-397.	2.4	14
60	Sensitivity of the denaturing gradient gel electrophoresis technique in detection of known mutations and novel Asian mutations in the CFTR gene. , 1997, 9, 136-147.		58
61	Sensitivity of the denaturing gradient gel electrophoresis technique in detection of known mutations and novel Asian mutations in the CFTR gene. Human Mutation, 1997, 9, 136-147.	1.1	4
62	Nonstop treatment of cystic fibrosis. Nature Medicine, 1996, 2, 608-608.	15.2	4
63	Identification of a novel nonsense mutation (L88X) in exon 3 of the cystic fibrosis transmembrane conductance regulator gene in a native Korean cystic fibrosis chromosome. Human Mutation, 1992, 1, 501-502.	1.1	15