Ada Hamosh

List of Publications by Year
in descending order

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	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
2	OMIM.org: Online Mendelian Inheritance in Man (OMIMÂ®), an online catalog of human genes and genetic disorders. Nucleic Acids Research, 2015, 43, D789-D798.	6.5	1,713
3	GeneMatcher: A Matching Tool for Connecting Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 928-930.	1.1	1,153
4	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
5	McKusick's Online Mendelian Inheritance in Man (OMIM(R)). Nucleic Acids Research, 2009, 37, D793-D796.	6.5	623
6	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
7	OMIM.org: leveraging knowledge across phenotypeâ€“gene relationships. Nucleic Acids Research, 2019, 47, D1038-D1043.	6.5	562
8	Online Mendelian Inheritance in Man (OMIM), a knowledgebase of human genes and genetic disorders. Nucleic Acids Research, 2002, 30, 52-55.	6.5	528
9	Online Mendelian Inheritance In Man (OMIM). Human Mutation, 2000, 15, 57-61.	1.1	503
10	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
11	Searching Online Mendelian Inheritance in Man (OMIM): A Knowledgebase of Human Genes and Genetic Phenotypes. Current Protocols in Bioinformatics, 2017, 58, 1.2.1-1.2.12.	25.8	378
12	A new face and new challenges for Online Mendelian Inheritance in Man (OMIMÂ®). Human Mutation, 2011, 32, 564-567.	1.1	309
13	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
14	How many rare diseases are there?. Nature Reviews Drug Discovery, 2020, 19, 77-78.	21.5	204
15	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
16	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
17	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. Human Mutation, 2015, 36, 425-431.	1.1	141
18	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	2.6	134

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19	The Centers for Mendelian Genomics: A new largeâ€scale initiative to identify the genes underlying rare Mendelian conditions. American Journal of Medical Genetics, Part A, 2012, 158A, 1523-1525.	0.7	110
20	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	3.7	99
21	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	1.1	94
22	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
23	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. Bioinformatics, 2014, 30, 3215-3222.	1.8	91
24	<scp>P</scp> heno <scp>DB</scp> : A New Webâ€Based Tool for the Collection, Storage, and Analysis of Phenotypic Features. Human Mutation, 2013, 34, 566-571.	1.1	64
25	Online Mendelian Inheritance in Man (<scp>OMIM</scp>Â®): Victor <scp>McKusick</scp>'s magnum opus. American Journal of Medical Genetics, Part A, 2021, 185, 3259-3265.	0.7	63
26	Sensitivity of the denaturing gradient gel electrophoresis technique in detection of known mutations and novel Asian mutations in theCFTR gene. , 1997, 9, 136-147.		58
27	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	3.7	57
28	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	1.1	56
29	KIF5A mutations cause an infantile onset phenotype including severe myoclonus with evidence of mitochondrial dysfunction. Annals of Neurology, 2016, 80, 633-637.	2.8	47
30	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47
31	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
32	Assessment of incidental findings in 232 whole-exome sequences from the Baylorâ€“Hopkins Center for Mendelian Genomics. Genetics in Medicine, 2015, 17, 782-788.	1.1	41
33	Genotypeâ€“phenotype correlation of congenital anomalies in multiple congenital anomalies hypotonia seizures syndrome (MCAHS1)/PIGNâ€related epilepsy. American Journal of Medical Genetics, Part A, 2016, 170, 77-86.	0.7	41
34	The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820.	9.4	38
35	Genomic Data Sharing for Novel Mendelian Disease Gene Discovery: The Matchmaker Exchange. Annual Review of Genomics and Human Genetics, 2020, 21, 305-326.	2.5	36
36	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34

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37	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. European Journal of Human Genetics, 2017, 25, 946-951.	1.4	33
38	Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. American Journal of Human Genetics, 2019, 105, 48-64.	2.6	29
39	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	1.2	24
40	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
41	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
42	Pachygyria in Weaver Syndrome. American Journal of Medical Genetics Part A, 1999, 86, 395-397.	2.4	14
43	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
44	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	1.1	13
45	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). Genetics in Medicine, 2018, 20, 169-171.	1.1	13
46	Whatâ€™s in a name? Issues to consider when naming Mendelian disorders. Genetics in Medicine, 2020, 22, 1573-1575.	1.1	13
47	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	1.1	11
48	Seven years since the launch of the Matchmaker Exchange: The evolution of genomic matchmaking. Human Mutation, 2022, 43, 659-667.	1.1	11
49	The utility of exome sequencing for fetal pleural effusions. Prenatal Diagnosis, 2020, 40, 590-595.	1.1	9
50	The management of pregnancy and delivery in 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. , 2016, 170, 1600-1602.		7
51	The impact of GeneMatcher on international data sharing and collaboration. Human Mutation, 2022, , .	1.1	7
52	Maternal Hyperphenylalaninemia: Rapid achievement of metabolic control predicts overall control throughout pregnancy. Molecular Genetics and Metabolism, 2013, 109, 3-8.	0.5	6
53	Horizontal integration of OMIM across the medical school preclinical curriculum for early reinforcement of clinical genetics principles. Genetics in Medicine, 2015, 17, 158-163.	1.1	6
54	Nonstop treatment of cystic fibrosis. Nature Medicine, 1996, 2, 608-608.	15.2	4

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55	Nextâ€generation sequencing and the evolution of data sharing. American Journal of Medical Genetics, Part A, 2021, 185, 2633-2635.	0.7	4
56	Three decades of the Human Genome Organization. American Journal of Medical Genetics, Part A, 2021, 185, 3314-3321.	0.7	4
57	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
58	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
59	Integrating the microbiome into precision medicine. Expert Review of Precision Medicine and Drug Development, 2016, 1, 475-477.	0.4	2
60	Viewing Victor McKusick' s legacy through the lens of his bibliography. American Journal of Medical Genetics, Part A, 2021, 185, 3212-3223.	0.7	1
61	Memories of Victor A. <scp>McKusick</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3377-3383.	0.7	1
62	Orthopaedic Problems in 35 Patients With Organic Acid Disorders. Journal of Pediatric Orthopaedics, 2021, 41, e457-e463.	0.6	0
63	Festschrift for Victor A. <scp>McKusick</scp> on the Centenary of his Birth: Introduction. American Journal of Medical Genetics, Part A, 2021, 185, 3189-3192.	0.7	0