Helen V Firth

List of Publications by Year
in descending order

62 papers	7,535 citations	136950 32 h-index	123424 61 g-index
75 all docs	75 docs citations	75 times ranked	13752 citing authors

#	Article	IF	Citations
1	DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources. American Journal of Human Genetics, 2009, 84, 524-533.	6.2	1,614
2	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	13.7	651
3	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
4	Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 2413-2421.	2.4	378
5	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	16.3	369
6	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
7	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
8	Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1,133 families with developmental disorders. Genetics in Medicine, 2018, 20, 1216-1223.	2.4	255
9	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246
10	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
11	Consensus Statement on next-generation-sequencing-based diagnostic testing of hereditary phaeochromocytomas and paragangliomas. Nature Reviews Endocrinology, 2017, 13, 233-247.	9.6	198
12	DECIPHER: database for the interpretation of phenotype-linked plausibly pathogenic sequence and copy-number variation. Nucleic Acids Research, 2014, 42, D993-D1000.	14.5	195
13	Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research. European Journal of Human Genetics, 2016, 24, 21-29.	2.8	161
14	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
15	The Deciphering Developmental Disorders (DDD) study. Developmental Medicine and Child Neurology, 2011, 53, 702-703.	2.1	153
16	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
17	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
18	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108

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19	De Novo Loss-of-Function Mutations in SETD5, Encoding a Methyltransferase in a 3p25 Microdeletion Syndrome Critical Region, Cause Intellectual Disability. American Journal of Human Genetics, 2014, 94, 618-624.	6.2	96
20	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
21	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	12.8	86
22	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
23	DECIPHER: web-based, community resource for clinical interpretation of rare variants in developmental disorders. Human Molecular Genetics, 2012, 21, R37-R44.	2.9	74
24	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
25	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
26	Chorion villus sampling and limb deficiencyâ€”cause or coincidence?. Prenatal Diagnosis, 1997, 17, 1313-1330.	2.3	53
27	The phenotypic spectrum of WWOX-related disorders: 20 additional cases of WOREE syndrome and review of the literature. Genetics in Medicine, 2019, 21, 1308-1318.	2.4	48
28	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
29	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
30	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
31	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	12.8	43
32	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42
33	Facilitating Collaboration in Rare Genetic Disorders Through Effective Matchmaking in DECIPHER. Human Mutation, 2015, 36, 941-949.	2.5	38
34	Potential research participants support the return of raw sequence data. Journal of Medical Genetics, 2015, 52, 571-574.	3.2	38
35	HeterozygousKIDINS220/ARMSnonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity. Human Molecular Genetics, 2016, 25, 2158-2167.	2.9	37
36	Heterozygous mutations affecting the protein kinase domain of CDK13 cause a syndromic form of developmental delay and intellectual disability. Journal of Medical Genetics, 2018, 55, 28-38.	3.2	36

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37	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.5	34
38	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	2.8	33
39	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
40	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	1.8	31
41	Principle of proportionality in genomic data sharing. Nature Reviews Genetics, 2016, 17, 1-2.	16.3	26
42	Integrating population variation and protein structural analysis to improve clinical interpretation of missense variation: application to the WD40 domain. Human Molecular Genetics, 2016, 25, 927-935.	2.9	26
43	Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. PLoS Genetics, 2020, 16, e1008916.	3.5	22
44	No expectation to share incidental findings in genomic research. Lancet, The, 2015, 385, 1289-1290.	13.7	19
45	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	2.4	16
46	The NHS England 100,000 Genomes Project: feasibility and utility of centralised genome sequencing for children with cancer. British Journal of Cancer, 2022, 127, 137-144.	6.4	16
47	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	2.5	13
48	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. American Journal of Human Genetics, 2021, 108, 2186-2194.	6.2	12
49	Chromosome 22q12.1 microdeletions: confirmation of the MN1 gene as a candidate gene for cleft palate. European Journal of Human Genetics, 2016, 24, 51-58.	2.8	10
50	DECIPHER: Supporting the interpretation and sharing of rare disease phenotypeâ€linked variant data to advance diagnosis and research. Human Mutation, 2022, , .	2.5	10
51	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
52	ADA2 deficiency complicated by EBV-driven lymphoproliferative disease. Clinical Immunology, 2020, 215, 108443.	3.2	9
53	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
54	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	1.8	7

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55	Genomics: the power, potential and pitfalls of the new technologies and how they are transforming healthcare. Clinical Medicine, 2019, 19, 269-272.	1.9	6
56	Malta (MYH9 Associated Elastin Aggregation) Syndrome: Germline Variants in MYH9 Cause RareÂSweat Duct Proliferations and Irregular ElastinÂAggregations. Journal of Investigative Dermatology, 2019, 139, 2238-2241.e6.	0.7	5
57	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
58	Craniomicromelic syndrome: Report of a third case. , 1999, 87, 360-361.		3
59	The genetics of developmental disorders. Paediatrics and Child Health (United Kingdom), 2019, 29, 422-430.	0.4	3
60	Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349.	6.2	3
61	Novel compound heterozygous STN1 variants are associated with Coats Plus syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1708.	1.2	3
62	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	6.2	3