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: Supporting the interpretation and sharing of rare disease phenotypeâ€linked variant data to advance diagnosis and research. Human Mutation, 2022, , .	2.5	10
2	Structural mapping of GABRB3 variants reveals genotypeâ€“phenotype correlations. Genetics in Medicine, 2022, 24, 681-693.	2.4	10
3	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
4	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
5	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	2.4	56
6	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
7	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	2.4	16
8	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
9	Novel compound heterozygous STN1 variants are associated with Coats Plus syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1708.	1.2	3
10	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
11	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	6.2	3
12	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
13	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
14	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
15	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
16	Trappc9 deficiency causes parent-of-origin dependent microcephaly and obesity. PLoS Genetics, 2020, 16, e1008916.	3.5	22
17	Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349.	6.2	3
18	ADA2 deficiency complicated by EBV-driven lymphoproliferative disease. Clinical Immunology, 2020, 215, 108443.	3.2	9

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19	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
20	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
21	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
22	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	12.8	43
23	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
24	The genetics of developmental disorders. Paediatrics and Child Health (United Kingdom), 2019, 29, 422-430.	0.4	3
25	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.5	34
26	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	12.8	86
27	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
28	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
29	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
30	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	1.8	31
31	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	16.3	369
32	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
33	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
34	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
35	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
36	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246

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37	Registered access: authorizing data access. European Journal of Human Genetics, 2018, 26, 1721-1731.	2.8	33
38	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
39	â€œMatchingâ€•consent to purpose: The example of the Matchmaker Exchange. Human Mutation, 2017, 38, 1281-1285.	2.5	13
40	Consensus Statement on next-generation-sequencing-based diagnostic testing of hereditary phaeochromocytomas and paragangliomas. Nature Reviews Endocrinology, 2017, 13, 233-247.	9.6	198
41	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
42	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
43	Principle of proportionality in genomic data sharing. Nature Reviews Genetics, 2016, 17, 1-2.	16.3	26
44	HeterozygousKIDINS220/ARMSnonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity. Human Molecular Genetics, 2016, 25, 2158-2167.	2.9	37
45	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
46	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
47	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
48	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
49	Facilitating Collaboration in Rare Genetic Disorders Through Effective Matchmaking in DECIPHER. Human Mutation, 2015, 36, 941-949.	2.5	38
50	Potential research participants support the return of raw sequence data. Journal of Medical Genetics, 2015, 52, 571-574.	3.2	38
51	No expectation to share incidental findings in genomic research. Lancet, The, 2015, 385, 1289-1290.	13.7	19
52	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
53	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
54	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

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55	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
56	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
57	DECIPHER: web-based, community resource for clinical interpretation of rare variants in developmental disorders. Human Molecular Genetics, 2012, 21, R37-R44.	2.9	74
58	The Deciphering Developmental Disorders (DDD) study. Developmental Medicine and Child Neurology, 2011, 53, 702-703.	2.1	153
59	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
60	Craniomicromelic syndrome: Report of a third case. , 1999, 87, 360-361.		3
61	Chorion villus sampling and limb deficiencyâ€"cause or coincidence?. Prenatal Diagnosis, 1997, 17, 1313-1330.	2.3	53
62	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	1.8	7