Matthew E Hurles

List of Publications by Citations

187 papers	43,823 citations	86 h-index	205 g-index
205 ext. papers	53,412 ext. citations	18.5 avg, IF	6.44 L-index

#	Paper	IF	Citations
187	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
186	Global variation in copy number in the human genome. *Nature*, **2006**, 444, 444-54	50.4	3306
185	Cerebral organoids model human brain development and microcephaly. *Nature*, **2013**, 501, 373-9	50.4	2621
184	Accurate whole human genome sequencing using reversible terminator chemistry. *Nature*, **2008**, 456, 53-9	50.4	2615
183	Origins and functional impact of copy number variation in the human genome. *Nature*, **2010**, 464, 704-12	50.4	1467
182	Relative impact of nucleotide and copy number variation on gene expression phenotypes. *Science*, **2007**, 315, 848-53	33.3	1361
181	Paired-end mapping reveals extensive structural variation in the human genome. *Science*, **2007**, 318, 420-6	33.3	895
180	A systematic survey of loss-of-function variants in human protein-coding genes. *Science*, **2012**, 335, 823-8	33.3	880
179	Mapping copy number variation by population-scale genome sequencing. *Nature*, **2011**, 470, 59-65	50.4	833
178	The DNA sequence of the human X chromosome. *Nature*, **2005**, 434, 325-37	50.4	822
177	Copy number variation in human health, disease, and evolution. *Annual Review of Genomics and Human Genetics*, **2009**, 10, 451-81	9.7	804
176	The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90	50.4	776
175	Identification of somatically acquired rearrangements in cancer using genome-wide massively parallel paired-end sequencing. *Nature Genetics*, **2008**, 40, 722-9	36.3	666
174	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
173	Copy number variation: new insights in genome diversity. *Genome Research*, **2006**, 16, 949-61	9.7	580
172	A high-resolution survey of deletion polymorphism in the human genome. *Nature Genetics*, **2006**, 38, 75-81	36.3	539
171	Y-chromosomal diversity in Europe is clinal and influenced primarily by geography, rather than by language. *American Journal of Human Genetics*, **2000**, 67, 1526-43	11	471

170	Characterising and predicting haploinsufficiency in the human genome. *PLoS Genetics*, **2010**, 6, e1001154	6	460
169	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. *Lancet, The*, **2015**, 385, 1305-14	40	451
168	Large, rare chromosomal deletions associated with severe early-onset obesity. *Nature*, **2010**, 463, 666-70	50.4	417
167	The genetic legacy of the Mongols. *American Journal of Human Genetics*, **2003**, 72, 717-21	11	414
166	Variation in genome-wide mutation rates within and between human families. *Nature Genetics*, **2011**, 43, 712-4	36.3	404
165	Bayesian refinement of association signals for 14 loci in 3 common diseases. *Nature Genetics*, **2012**, 44, 1294-301	36.3	347
164	Timing, rates and spectra of human germline mutation. *Nature Genetics*, **2016**, 48, 126-133	36.3	338
163	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. *Nature Biotechnology*, **2011**, 29, 512-20	44.5	333
162	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. *Nature Neuroscience*, **2016**, 19, 571-7	25.5	284
161	The Matchmaker Exchange: a platform for rare disease gene discovery. *Human Mutation*, **2015**, 36, 915-21	4.7	280
160	Compound inheritance of a low-frequency regulatory SNP and a rare null mutation in exon-junction complex subunit RBM8A causes TAR syndrome. *Nature Genetics*, **2012**, 44, 435-9, S1-2	36.3	279
159	Challenges and standards in integrating surveys of structural variation. *Nature Genetics*, **2007**, 39, S7-15	36.3	279
158	Prenatal exome sequencing analysis in fetal structural anomalies detected by ultrasonography (PAGE): a cohort study. *Lancet, The*, **2019**, 393, 747-757	40	249
157	Gene duplication: the genomic trade in spare parts. *PLoS Biology*, **2004**, 2, E206	9.7	245
156	Loss-of-function mutations in MICU1 cause a brain and muscle disorder linked to primary alterations in mitochondrial calcium signaling. *Nature Genetics*, **2014**, 46, 188-93	36.3	242
155	Mitochondrial DNA and the origins of the domestic horse. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 10905-10	11.5	238
154	Germline rates of de novo meiotic deletions and duplications causing several genomic disorders. *Nature Genetics*, **2008**, 40, 90-5	36.3	235
153	Genome-wide SNP and CNV analysis identifies common and low-frequency variants associated with severe early-onset obesity. *Nature Genetics*, **2013**, 45, 513-7	36.3	231

152	Genome-wide mapping and assembly of structural variant breakpoints in the mouse genome. *Genome Research*, **2010**, 20, 623-35	9.7	217
151	Recessive HYDIN mutations cause primary ciliary dyskinesia without randomization of left-right body asymmetry. *American Journal of Human Genetics*, **2012**, 91, 672-84	11	212
150	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. *American Journal of Human Genetics*, **2017**, 100, 695-705	11	200
149	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. *Nature Genetics*, **2016**, 48, 1060-5	36.3	200
148	Mutation spectrum revealed by breakpoint sequencing of human germline CNVs. *Nature Genetics*, **2010**, 42, 385-91	36.3	192
147	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. *Nature Genetics*, **2011**, 43, 23-6	36.3	185
146	A brief history of human disease genetics. *Nature*, **2020**, 577, 179-189	50.4	181
145	The dual origin of the Malagasy in Island Southeast Asia and East Africa: evidence from maternal and paternal lineages. *American Journal of Human Genetics*, **2005**, 76, 894-901	11	179
144	Copy number variation and evolution in humans and chimpanzees. *Genome Research*, **2008**, 18, 1698-710	9.7	178
143	Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. *Nature Genetics*, **2010**, 42, 400-5	36.3	167
142	Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of α-dystroglycan. *American Journal of Human Genetics*, **2013**, 93, 29-41	11	162
141	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	8.1	161
140	Somatic mutations reveal asymmetric cellular dynamics in the early human embryo. *Nature*, **2017**, 543, 714-718	50.4	157
139	Genome-wide detection of human copy number variations using high-density DNA oligonucleotide arrays. *Genome Research*, **2006**, 16, 1575-84	9.7	156
138	Genome-wide screen for metabolic syndrome susceptibility Loci reveals strong lipid gene contribution but no evidence for common genetic basis for clustering of metabolic syndrome traits. *Circulation: Cardiovascular Genetics*, **2012**, 5, 242-9		153
137	Male demography in East Asia: a north-south contrast in human population expansion times. *Genetics*, **2006**, 172, 2431-9	4	152
136	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. *Nature*, **2018**, 562, 268-271	50.4	149
135	De novo mutations in regulatory elements in neurodevelopmental disorders. *Nature*, **2018**, 555, 611-616	50.4	146

134	A robust statistical method for case-control association testing with copy number variation. *Nature Genetics*, **2008**, 40, 1245-52	36.3	143
133	Genetic analysis of completely sequenced disease-associated MHC haplotypes identifies shuffling of segments in recent human history. *PLoS Genetics*, **2006**, 2, e9	6	140
132	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
131	Mutations in B3GALNT2 cause congenital muscular dystrophy and hypoglycosylation of α-dystroglycan. *American Journal of Human Genetics*, **2013**, 92, 354-65	11	139
130	DECIPHER: database for the interpretation of phenotype-linked plausibly pathogenic sequence and copy-number variation. *Nucleic Acids Research*, **2014**, 42, D993-D1000	20.1	138
129	The population genetics of structural variation. *Nature Genetics*, **2007**, 39, S30-6	36.3	138
128	Genome assembly comparison identifies structural variants in the human genome. *Nature Genetics*, **2006**, 38, 1413-8	36.3	133
127	Mutations in BICD2 cause dominant congenital spinal muscular atrophy and hereditary spastic paraplegia. *American Journal of Human Genetics*, **2013**, 92, 965-73	11	131
126	Exome sequencing improves genetic diagnosis of structural fetal abnormalities revealed by ultrasound. *Human Molecular Genetics*, **2014**, 23, 3269-77	5.6	128
125	The functional impact of structural variation in humans. *Trends in Genetics*, **2008**, 24, 238-45	8.5	128
124	Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. *Nature Genetics*, **2014**, 46, 1283-1292	36.3	125
123	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, 21-9	5.3	124
122	Genome-wide transcriptome profiling reveals the functional impact of rare de novo and recurrent CNVs in autism spectrum disorders. *American Journal of Human Genetics*, **2012**, 91, 38-55	11	123
121	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
120	Accurate and reliable high-throughput detection of copy number variation in the human genome. *Genome Research*, **2006**, 16, 1566-74	9.7	122
119	DeNovoGear: de novo indel and point mutation discovery and phasing. *Nature Methods*, **2013**, 10, 985-7	21.6	120
118	Fast-evolving noncoding sequences in the human genome. *Genome Biology*, **2007**, 8, R118	18.3	116
117	Rare variants in NR2F2 cause congenital heart defects in humans. *American Journal of Human Genetics*, **2014**, 94, 574-85	11	115

116	Adaptive evolution of UGT2B17 copy-number variation. *American Journal of Human Genetics*, **2008**, 83, 337-46	11	112
115	Recent male-mediated gene flow over a linguistic barrier in Iberia, suggested by analysis of a Y-chromosomal DNA polymorphism. *American Journal of Human Genetics*, **1999**, 65, 1437-48	11	111
114	Deletion of TOP3β, a component of FMRP-containing mRNPs, contributes to neurodevelopmental disorders. *Nature Neuroscience*, **2013**, 16, 1228-1237	25.5	110
113	Deletions of chromosomal regulatory boundaries are associated with congenital disease. *Genome Biology*, **2014**, 15, 423	18.3	108
112	European Y-chromosomal lineages in Polynesians: a contrast to the population structure revealed by mtDNA. *American Journal of Human Genetics*, **1998**, 63, 1793-806	11	103
111	Evidence for 28 genetic disorders discovered by combining healthcare and research data. *Nature*, **2020**, 586, 757-762	50.4	103
110	Structural variation on the short arm of the human Y chromosome: recurrent multigene deletions encompassing Amelogenin Y. *Human Molecular Genetics*, **2007**, 16, 307-16	5.6	102
109	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. *Brain*, **2013**, 136, 3618-24	11.2	100
108	Human spermatogenic failure purges deleterious mutation load from the autosomes and both sex chromosomes, including the gene DMRT1. *PLoS Genetics*, **2013**, 9, e1003349	6	99
107	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 7974-81	11.5	94
106	Cis and trans effects of human genomic variants on gene expression. *PLoS Genetics*, **2014**, 10, e1004461	6	92
105	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. *Nature Genetics*, **2015**, 47, 1363-9	36.3	91
104	Untangling Oceanic settlement: the edge of the knowable. *Trends in Ecology and Evolution*, **2003**, 18, 531-540	10.9	91
103	Combined NGS approaches identify mutations in the intraflagellar transport gene IFT140 in skeletal ciliopathies with early progressive kidney Disease. *Human Mutation*, **2013**, 34, 714-24	4.7	89
102	A chromosomal rearrangement hotspot can be identified from population genetic variation and is coincident with a hotspot for allelic recombination. *American Journal of Human Genetics*, **2006**, 79, 890-902	11	87
101	Mutations in the gene encoding IFT dynein complex component WDR34 cause Jeune asphyxiating thoracic dystrophy. *American Journal of Human Genetics*, **2013**, 93, 932-44	11	84
100	The genome-wide effects of ionizing radiation on mutation induction in the mammalian germline. *Nature Communications*, **2015**, 6, 6684	17.4	83
99	Phenotype-specific effect of chromosome 1q21.1 rearrangements and GJA5 duplications in 2436 congenital heart disease patients and 6760 controls. *Human Molecular Genetics*, **2012**, 21, 1513-20	5.6	83

98	Quantifying the contribution of recessive coding variation to developmental disorders. *Science*, **2018**, 362, 1161-1164	33.3	83
97	High incidence of recurrent copy number variants in patients with isolated and syndromic Müllerian aplasia. *Journal of Medical Genetics*, **2011**, 48, 197-204	5.8	80
96	Independent and population-specific association of risk variants at the IRGM locus with Crohn's disease. *Human Molecular Genetics*, **2010**, 19, 1828-39	5.6	79
95	Y chromosomal evidence for the origins of oceanic-speaking peoples. *Genetics*, **2002**, 160, 289-303	4	79
94	Heterozygous loss-of-function mutations in YAP1 cause both isolated and syndromic optic fissure closure defects. *American Journal of Human Genetics*, **2014**, 94, 295-302	11	74
93	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, 618-24	11	74
92	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. *Nature*, **2020**, 583, 90-95	50.4	69
91	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. *PLoS Genetics*, **2016**, 12, e1005963	6	67
90	The Y-chromosome landscape of the Philippines: extensive heterogeneity and varying genetic affinities of Negrito and non-Negrito groups. *European Journal of Human Genetics*, **2011**, 19, 224-30	5.3	66
89	Monoallelic and biallelic mutations in MAB21L2 cause a spectrum of major eye malformations. *American Journal of Human Genetics*, **2014**, 94, 915-23	11	64
88	Quantifying single nucleotide variant detection sensitivity in exome sequencing. *BMC Bioinformatics*, **2013**, 14, 195	3.6	63
87	Extreme growth failure is a common presentation of ligase IV deficiency. *Human Mutation*, **2014**, 35, 76-85	4.7	63
86	Dynamics of a human interparalog gene conversion hotspot. *Genome Research*, **2004**, 14, 835-44	9.7	63
85	Independent estimation of the frequency of rare CNVs in the UK population confirms their role in schizophrenia. *Schizophrenia Research*, **2012**, 135, 1-7	3.6	62
84	Deciphering past human population movements in Oceania: provably optimal trees of 127 mtDNA genomes. *Molecular Biology and Evolution*, **2006**, 23, 1966-75	8.3	59
83	High level of male-biased Scandinavian admixture in Greenlandic Inuit shown by Y-chromosomal analysis. *Human Genetics*, **2003**, 112, 353-63	6.3	57
82	Identification of a human synaptotagmin-1 mutation that perturbs synaptic vesicle cycling. *Journal of Clinical Investigation*, **2015**, 125, 1670-8	15.9	57
81	Analysis of deletion breakpoints from 1,092 humans reveals details of mutation mechanisms. *Nature Communications*, **2015**, 6, 7256	17.4	56

80	Geographical, linguistic, and cultural influences on genetic diversity: Y-chromosomal distribution in Northern European populations. *Molecular Biology and Evolution*, **2001**, 18, 1077-87	8.3	56
79	Patterns of inter- and intra-group genetic diversity in the Vlax Roma as revealed by Y chromosome and mitochondrial DNA lineages. *European Journal of Human Genetics*, **2001**, 9, 97-104	5.3	56
78	DECIPHER: web-based, community resource for clinical interpretation of rare variants in developmental disorders. *Human Molecular Genetics*, **2012**, 21, R37-44	5.6	55
77	Reciprocal duplication of the Williams-Beuren syndrome deletion on chromosome 7q11.23 is associated with schizophrenia. *Biological Psychiatry*, **2014**, 75, 371-7	7.9	54
76	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. *Cell*, **2017**, 168, 830-842.e7	36.2	53
75	TRAIP promotes DNA damage response during genome replication and is mutated in primordial dwarfism. *Nature Genetics*, **2016**, 48, 36-43	36.3	53
74	B56-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. *Journal of Clinical Investigation*, **2015**, 125, 3051-62	15.9	53
73	Consent Codes: Upholding Standard Data Use Conditions. *PLoS Genetics*, **2016**, 12, e1005772	6	51
72	Genomic pathology of SLE-associated copy-number variation at the FCGR2C/FCGR3B/FCGR2B locus. *American Journal of Human Genetics*, **2013**, 92, 28-40	11	48
71	Biallelic Variants in UBA5 Link Dysfunctional UFM1 Ubiquitin-like Modifier Pathway to Severe Infantile-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 683-694	11	43
70	Genomic analysis of hepatitis B virus reveals antigen state and genotype as sources of evolutionary rate variation. *Viruses*, **2011**, 3, 83-101	6.2	42
69	Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations of WDR73. *Brain*, **2015**, 138, 2173-90	11.2	40
68	Absence of heterozygosity due to template switching during replicative rearrangements. *American Journal of Human Genetics*, **2015**, 96, 555-64	11	39
67	Mosaic structural variation in children with developmental disorders. *Human Molecular Genetics*, **2015**, 24, 2733-45	5.6	39
66	A novel method for detecting uniparental disomy from trio genotypes identifies a significant excess in children with developmental disorders. *Genome Research*, **2014**, 24, 673-87	9.7	38
65	Similarities and differences in patterns of germline mutation between mice and humans. *Nature Communications*, **2019**, 10, 4053	17.4	35
64	Haploid chromosomes in molecular ecology: lessons from the human Y. *Molecular Ecology*, **2001**, 10, 1599-613	5.7	35
63	Prenatal whole exome sequencing: the views of clinicians, scientists, genetic counsellors and patient representatives. *Prenatal Diagnosis*, **2016**, 36, 935-941	3.2	35

62	Origins of chromosomal rearrangement hotspots in the human genome: evidence from the AZFa deletion hotspots. *Genome Biology*, **2004**, 5, R55	18.3	34
61	Managing clinically significant findings in research: the UK10K example. *European Journal of Human Genetics*, **2014**, 22, 1100-4	5.3	33
60	TM4SF20 ancestral deletion and susceptibility to a pediatric disorder of early language delay and cerebral white matter hyperintensities. *American Journal of Human Genetics*, **2013**, 93, 197-210	11	32
59	Facilitating collaboration in rare genetic disorders through effective matchmaking in DECIPHER. *Human Mutation*, **2015**, 36, 941-9	4.7	32
58	Native American Y chromosomes in Polynesia: the genetic impact of the Polynesian slave trade. *American Journal of Human Genetics*, **2003**, 72, 1282-7	11	31
57	Molecular autopsy by trio exome sequencing (ES) and postmortem examination in fetuses and neonates with prenatally identified structural anomalies. *Genetics in Medicine*, **2019**, 21, 1065-1073	8.1	31
56	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. *Genetics in Medicine*, **2017**, 19, 900-908	8.1	30
55	Gene conversion homogenizes the CMT1A paralogous repeats. *BMC Genomics*, **2001**, 2, 11	4.5	30
54	Global haplotype diversity in the human insulin gene region. *Genome Research*, **2003**, 13, 2101-11	9.7	29
53	Pathogenicity and selective constraint on variation near splice sites. *Genome Research*, **2019**, 29, 159-170	9.7	29
52	Evidence for widespread reticulate evolution within human duplicons. *American Journal of Human Genetics*, **2005**, 77, 824-40	11	28
51	Assaying chromosomal inversions by single-molecule haplotyping. *Nature Methods*, **2006**, 3, 439-45	21.6	27
50	Detection of structural mosaicism from targeted and whole-genome sequencing data. *Genome Research*, **2017**, 27, 1704-1714	9.7	26
49	Exome sequencing identifies rare variants in multiple genes in atrioventricular septal defect. *Genetics in Medicine*, **2016**, 18, 189-98	8.1	25
48	Contribution of retrotransposition to developmental disorders. *Nature Communications*, **2019**, 10, 4630	17.4	25
47	Harnessing genomics to identify environmental determinants of heritable disease. *Mutation Research - Reviews in Mutation Research*, **2013**, 752, 6-9	7	25
46	Potential research participants support the return of raw sequence data. *Journal of Medical Genetics*, **2015**, 52, 571-4	5.8	24
45	COX10 mutations resulting in complex multisystem mitochondrial disease that remains stable into adulthood. *JAMA Neurology*, **2013**, 70, 1556-61	17.2	24

44	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. *Nature Communications*, **2019**, 10, 2373	17.4	22
43	Reduced burden of very large and rare CNVs in bipolar affective disorder. *Bipolar Disorders*, **2013**, 15, 893-8	3.8	22
42	Genetic basis of Y-linked hearing impairment. *American Journal of Human Genetics*, **2013**, 92, 301-6	11	21
41	Are 100,000 "SNPs" useless?. *Science*, **2002**, 298, 1509; author reply 1509	33.3	20
40	Exome Sequencing in Fetuses with Structural Malformations. *Journal of Clinical Medicine*, **2014**, 3, 747-62	5.1	19
39	Copy number variation in the human Y chromosome in the UK population. *Human Genetics*, **2015**, 134, 789-800	6.3	19
38	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. *Genome Research*, **2019**, 29, 1047-1056	9.7	18
37	Principle of proportionality in genomic data sharing. *Nature Reviews Genetics*, **2016**, 17, 1-2	30.1	18
36	How homologous recombination generates a mutable genome. *Human Genomics*, **2005**, 2, 179-86	6.8	18
35	Integrating healthcare and research genetic data empowers the discovery of 28 novel developmental disorders		18
34	Loss of ADAMTS19 causes progressive non-syndromic heart valve disease. *Nature Genetics*, **2020**, 52, 40-47	36.3	18
33	Registered access: authorizing data access. *European Journal of Human Genetics*, **2018**, 26, 1721-1731	5.3	17
32	A genome-wide assessment of the role of untagged copy number variants in type 1 diabetes. *PLoS Genetics*, **2014**, 10, e1004367	6	16
31	High-throughput haplotype determination over long distances by haplotype fusion PCR and ligation haplotyping. *Nature Protocols*, **2009**, 4, 1771-83	18.8	13
30	Long-range, high-throughput haplotype determination via haplotype-fusion PCR and ligation haplotyping. *Nucleic Acids Research*, **2008**, 36, e82	20.1	13
29	Don't mix radiocarbon and calendar years. *Nature*, **2005**, 434, 697	50.4	13
28	The rate of nonallelic homologous recombination in males is highly variable, correlated between monozygotic twins and independent of age. *PLoS Genetics*, **2014**, 10, e1004195	6	12
27	Older males beget more mutations. *Nature Genetics*, **2012**, 44, 1174-6	36.3	12

26	Empirical research on the ethics of genomic research. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2099-101	2.5	12
25	Detection and correction of artefacts in estimation of rare copy number variants and analysis of rare deletions in type 1 diabetes. *Human Molecular Genetics*, **2015**, 24, 1774-90	5.6	11
24	A direct multi-generational estimate of the human mutation rate from autozygous segments seen in thousands of parentally related individuals		11
23	"Matching" consent to purpose: The example of the Matchmaker Exchange. *Human Mutation*, **2017**, 38, 1281-1285	4.7	10
22	High throughput exome coverage of clinically relevant cardiac genes. *BMC Medical Genomics*, **2014**, 7, 67	3.7	8
21	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	11	8
20	Exome Sequencing for Prenatal Detection of Genetic Abnormalities in Fetal Ultrasound Anomalies: An Economic Evaluation. *Fetal Diagnosis and Therapy*, **2020**, 47, 554-564	2.4	7
19	Recombination Hotspots in Nonallelic Homologous Recombination **2006**, 341-355		6
18	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. *Genetics in Medicine*, **2021**, 23, 571-575	8.1	6
17	Y chromosomal STRs haplotypes in two populations from Bolivia. *Legal Medicine*, **2007**, 9, 43-7	1.9	5
16	Shotgun haplotyping: a novel method for surveying allelic sequence variation. *Nucleic Acids Research*, **2005**, 33, e152	20.1	5
15	The contribution of X-linked coding variation to severe developmental disorders. *Nature Communications*, **2021**, 12, 627	17.4	5
14	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. *American Journal of Human Genetics*, **2019**, 105, 933-946	11	4
13	Analysis of novel missense ATR mutations reveals new splicing defects underlying Seckel syndrome. *Human Mutation*, **2018**, 39, 1847-1853	4.7	4
12	Validity of the family-based association test for copy number variant data in the case of non-linear intensity-genotype relationship. *Genetic Epidemiology*, **2012**, 36, 895-8	2.6	3
11	Genetic and pharmacological causes of germline hypermutation		3
10	Germline variants in HEY2 functional domains lead to congenital heart defects and thoracic aortic aneurysms. *Genetics in Medicine*, **2021**, 23, 103-110	8.1	3
9	DECIPHER: Supporting the interpretation and sharing of rare disease phenotype-linked variant data to advance diagnosis and research.. *Human Mutation*, **2022**,	4.7	2

8	Genetic correlates of phenotypic heterogeneity in autism		2
7	Contribution of Retrotransposition to Developmental Disorders		2
6	Sex-biased reduction in reproductive success drives selective constraint on human genes		1
5	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. *PLoS Genetics*, **2021**, 17, e1009679	6	1
4	Y-Chromosomal Rearrangements and Azoospermia **2006**, 273-288		0
3	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	11	0
2	Reduced reproductive success is associated with selective constraint on human genes.. *Nature*, **2022**, 603, 858-863	50.4	0
1	Genetic and chemotherapeutic influences on germline hypermutation.. *Nature*, **2022**, 605, 503-508	50.4	0