

Jayne Y Hehir-Kwa

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

3,977
citations

257450

24
h-index

315739

38
g-index

41
all docs

41
docs citations

41
times ranked

9959
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	27.8	996
2	Refining analyses of copy number variation identifies specific genes associated with developmental delay. <i>Nature Genetics</i> , 2014, 46, 1063-1071.	21.4	583
3	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. <i>Nature Genetics</i> , 2015, 47, 668-671.	21.4	311
4	The Genome of the Netherlands: design, and project goals. <i>European Journal of Human Genetics</i> , 2014, 22, 221-227.	2.8	246
5	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. <i>Genetics in Medicine</i> , 2017, 19, 667-675.	2.4	143
6	Characteristics of de novo structural changes in the human genome. <i>Genome Research</i> , 2015, 25, 792-801.	5.5	115
7	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over 2.8 109	2.8	109
8	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016, 24, 652-659.	2.8	108
9	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. <i>Human Mutation</i> , 2013, 34, 1439-1448.	2.5	105
10	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. <i>European Journal of Human Genetics</i> , 2017, 25, 591-599.	2.8	104
11	Clinical Significance of De Novo and Inherited Copy-Number Variation. <i>Human Mutation</i> , 2013, 34, 1679-1687.	2.5	100
12	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. <i>Nature Communications</i> , 2016, 7, 12989.	12.8	99
13	De novo copy number variants associated with intellectual disability have a paternal origin and age bias. <i>Journal of Medical Genetics</i> , 2011, 48, 776-778.	3.2	95
14	Genome-wide Copy Number Profiling on High-density Bacterial Artificial Chromosomes, Single-nucleotide Polymorphisms, and Oligonucleotide Microarrays: A Platform Comparison based on Statistical Power Analysis. <i>DNA Research</i> , 2007, 14, 1-11.	3.4	91
15	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. <i>European Journal of Human Genetics</i> , 2017, 25, 308-314.	2.8	90
16	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , 2015, 23, 1601-1606.	2.8	85
17	Exome sequencing and whole genome sequencing for the detection of copy number variation. <i>Expert Review of Molecular Diagnostics</i> , 2015, 15, 1023-1032.	3.1	73
18	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 338-355.	6.2	58

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19	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. <i>Genome Medicine</i> , 2019, 11, 38.	8.2	49
20	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. <i>PLoS Computational Biology</i> , 2010, 6, e1000752.	3.2	46
21	BRCA Testing by Single-Molecule Molecular Inversion Probes. <i>Clinical Chemistry</i> , 2017, 63, 503-512.	3.2	46
22	Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia. <i>Human Mutation</i> , 2017, 38, 1592-1605.	2.5	45
23	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2019, 104, 758-766.	6.2	34
24	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2019, 21, 1719-1725.	2.4	34
25	The clustering of functionally related genes contributes to CNV-mediated disease. <i>Genome Research</i> , 2015, 25, 802-813.	5.5	31
26	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology. <i>Npj Precision Oncology</i> , 2021, 5, 15.	5.4	30
27	Epigenetic regulator genes direct lineage switching in MLL/AF4 leukemia. <i>Blood</i> , 2022, 140, 1875-1890.	1.4	26
28	Clinical interpretation of CNVs with cross-species phenotype data. <i>Journal of Medical Genetics</i> , 2014, 51, 766-772.	3.2	23
29	Gene Networks Underlying Convergent and Pleiotropic Phenotypes in a Large and Systematically-Phenotyped Cohort with Heterogeneous Developmental Disorders. <i>PLoS Genetics</i> , 2015, 11, e1005012.	3.5	14
30	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. <i>European Journal of Human Genetics</i> , 2016, 24, 1707-1714.	2.8	14
31	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. <i>Genomics Data</i> , 2014, 2, 144-146.	1.3	13
32	A molecular inversion probe-based next-generation sequencing panel to detect germline mutations in Chinese early-onset colorectal cancer patients. <i>Oncotarget</i> , 2017, 8, 24533-24547.	1.8	12
33	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. <i>European Journal of Human Genetics</i> , 2021, 29, 1418-1423.	2.8	12
34	Improved Gene Fusion Detection in Childhood Cancer Diagnostics Using RNA Sequencing. <i>JCO Precision Oncology</i> , 2022, 6, e2000504.	3.0	9
35	The clinical implementation of copy number detection in the age of next-generation sequencing. <i>Expert Review of Molecular Diagnostics</i> , 2018, 18, 907-915.	3.1	8
36	Clonal Relationship Between Lichen Sclerosus, Differentiated Vulvar Intra-epithelial Neoplasia and Non HPV-related Vulvar Squamous Cell Carcinoma. <i>Cancer Genomics and Proteomics</i> , 2020, 17, 151-160.	2.0	8

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37	Copy number variations as potential diagnostic and prognostic markers for CNS melanocytic neoplasms in neurocutaneous melanosis. <i>Acta Neuropathologica</i> , 2017, 133, 333-335.	7.7	3
38	Reply to Sajantila and Budowle. <i>European Journal of Human Genetics</i> , 2016, 24, 330-330.	2.8	1