

# 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

293460

24  
h-index

355658

38  
g-index

41  
all docs

41  
docs citations

41  
times ranked

10878  
citing authors

#	ARTICLE	IF	CITATIONS
1	Improved Gene Fusion Detection in Childhood Cancer Diagnostics Using RNA Sequencing. <i>JCO Precision Oncology</i> , 2022, 6, e2000504.	1.5	9
2	Epigenetic regulator genes direct lineage switching in <i>MLL/AF4</i> leukemia. <i>Blood</i> , 2022, 140, 1875-1890.	0.6	26
3	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. <i>European Journal of Human Genetics</i> , 2021, 29, 1418-1423.	1.4	12
4	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology. <i>Npj Precision Oncology</i> , 2021, 5, 15.	2.3	30
5	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.	2.6	58
6	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.	1.0	8
7	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. <i>Genome Medicine</i> , 2019, 11, 38.	3.6	49
8	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.	2.6	34
9	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2019, 21, 1719-1725.	1.1	34
10	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.	1.5	8
11	BRCA Testing by Single-Molecule Molecular Inversion Probes. <i>Clinical Chemistry</i> , 2017, 63, 503-512.	1.5	46
12	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. <i>European Journal of Human Genetics</i> , 2017, 25, 591-599.	1.4	104
13	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.	1.4	90
14	Copy number variations as potential diagnostic and prognostic markers for CNS melanocytic neoplasms in neurocutaneous melanosis. <i>Acta Neuropathologica</i> , 2017, 133, 333-335.	3.9	3
15	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.	1.1	45
16	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.	1.1	143
17	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.	0.8	12
18	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. <i>European Journal of Human Genetics</i> , 2016, 24, 1707-1714.	1.4	14

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19	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. <i>Nature Communications</i> , 2016, 7, 12989.	5.8	99
20	Reply to Sajantila and Budowle. <i>European Journal of Human Genetics</i> , 2016, 24, 330-330.	1.4	1
21	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.	1.4	108
22	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , 2015, 23, 1601-1606.	1.4	85
23	Exome sequencing and whole genome sequencing for the detection of copy number variation. <i>Expert Review of Molecular Diagnostics</i> , 2015, 15, 1023-1032.	1.5	73
24	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.	1.5	14
25	The clustering of functionally related genes contributes to CNV-mediated disease. <i>Genome Research</i> , 2015, 25, 802-813.	2.4	31
26	Characteristics of de novo structural changes in the human genome. <i>Genome Research</i> , 2015, 25, 792-801.	2.4	115
27	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.	9.4	311
28	Clinical interpretation of CNVs with cross-species phenotype data. <i>Journal of Medical Genetics</i> , 2014, 51, 766-772.	1.5	23
29	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over	1.4	109
30	The Genome of the Netherlands: design, and project goals. <i>European Journal of Human Genetics</i> , 2014, 22, 221-227.	1.4	246
31	Refining analyses of copy number variation identifies specific genes associated with developmental delay. <i>Nature Genetics</i> , 2014, 46, 1063-1071.	9.4	583
32	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. <i>Genomics Data</i> , 2014, 2, 144-146.	1.3	13
33	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	13.7	996
34	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. <i>Human Mutation</i> , 2013, 34, 1439-1448.	1.1	105
35	Clinical Significance of De Novo and Inherited Copy-Number Variation. <i>Human Mutation</i> , 2013, 34, 1679-1687.	1.1	100
36	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.	1.5	95

#	ARTICLE	IF	CITATIONS
37	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. PLoS Computational Biology, 2010, 6, e1000752.	1.5	46
38	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. DNA Research, 2007, 14, 1-11.	1.5	91