Jayne Y Hehir-Kwa

List of Publications by Year
in descending order

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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	Improved Gene Fusion Detection in Childhood Cancer Diagnostics Using RNA Sequencing. JCO Precision Oncology, 2022, 6, e2000504.	3.0	9
2	Epigenetic regulator genes direct lineage switching inÂMLL/AF4 leukemia. Blood, 2022, 140, 1875-1890.	1.4	26
3	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. European Journal of Human Genetics, 2021, 29, 1418-1423.	2.8	12
4	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology. Npj Precision Oncology, 2021, 5, 15.	5.4	30
5	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 338-355.	6.2	58
6	Clonal Relationship Between Lichen Sclerosus, Differentiated Vulvar Intra-epithelial Neoplasia and Non HPV-related Vulvar Squamous Cell Carcinoma. Cancer Genomics and Proteomics, 2020, 17, 151-160.	2.0	8
7	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	8.2	49
8	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	6.2	34
9	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	2.4	34
10	The clinical implementation of copy number detection in the age of next-generation sequencing. Expert Review of Molecular Diagnostics, 2018, 18, 907-915.	3.1	8
11	BRCA Testing by Single-Molecule Molecular Inversion Probes. Clinical Chemistry, 2017, 63, 503-512.	3.2	46
12	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599.	2.8	104
13	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. European Journal of Human Genetics, 2017, 25, 308-314.	2.8	90
14	Copy number variations as potential diagnostic and prognostic markers for CNS melanocytic neoplasms in neurocutaneous melanosis. Acta Neuropathologica, 2017, 133, 333-335.	7.7	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. Human Mutation, 2017, 38, 1592-1605.	2.5	45
16	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	2.4	143
17	A molecular inversion probe-based next-generation sequencing panel to detect germline mutations in Chinese early-onset colorectal cancer patients. Oncotarget, 2017, 8, 24533-24547.	1.8	12
18	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. European Journal of Human Genetics, 2016, 24, 1707-1714.	2.8	14

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19	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. Nature Communications, 2016, 7, 12989.	12.8	99
20	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	2.8	1
21	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
22	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
23	Exome sequencing and whole genome sequencing for the detection of copy number variation. Expert Review of Molecular Diagnostics, 2015, 15, 1023-1032.	3.1	73
24	Gene Networks Underlying Convergent and Pleiotropic Phenotypes in a Large and Systematically-Phenotyped Cohort with Heterogeneous Developmental Disorders. PLoS Genetics, 2015, 11, e1005012.	3.5	14
25	The clustering of functionally related genes contributes to CNV-mediated disease. Genome Research, 2015, 25, 802-813.	5.5	31
26	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	5.5	115
27	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. Nature Genetics, 2015, 47, 668-671.	21.4	311
28	Clinical interpretation of CNVs with cross-species phenotype data. Journal of Medical Genetics, 2014, 51, 766-772.	3.2	23
29	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over	2.8	109
30	The Genome of the Netherlands: design, and project goals. European Journal of Human Genetics, 2014, 22, 221-227.	2.8	246
31	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
32	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 2014, 2, 144-146.	1.3	13
33	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	27.8	996
34	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	2.5	105
35	Clinical Significance of De Novo and Inherited Copy-Number Variation. Human Mutation, 2013, 34, 1679-1687.	2.5	100
36	De novo copy number variants associated with intellectual disability have a paternal origin and age bias. Journal of Medical Genetics, 2011, 48, 776-778.	3.2	95

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37	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. PLoS Computational Biology, 2010, 6, e1000752.	3.2	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.	3.4	91