

Christine R Beck

List of Publications by Year in descending order

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Version: 2024-02-01

33
papers

3,123
citations

279798

23
h-index

395702

33
g-index

36
all docs

36
docs citations

36
times ranked

5721
citing authors

#	ARTICLE	IF	CITATIONS
1	A comprehensive long-read isoform analysis platform and sequencing resource for breast cancer. <i>Science Advances</i> , 2022, 8, eabg6711.	10.3	30
2	SvAnna: efficient and accurate pathogenicity prediction of coding and regulatory structural variants in long-read genome sequencing. <i>Genome Medicine</i> , 2022, 14, 44.	8.2	7
3	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders. <i>Cell</i> , 2022, 185, 1986-2005.e26.	28.9	67
4	Hotspots of Human Mutation. <i>Trends in Genetics</i> , 2021, 37, 717-729.	6.7	62
5	Structural variant identification and characterization. <i>Chromosome Research</i> , 2020, 28, 31-47.	2.2	13
6	Inter-Strain Epigenomic Profiling Reveals a Candidate IAP Master Copy in C3H Mice. <i>Viruses</i> , 2020, 12, 783.	3.3	9
7	Extrachromosomal DNA is associated with oncogene amplification and poor outcome across multiple cancers. <i>Nature Genetics</i> , 2020, 52, 891-897.	21.4	273
8	Megabase Length Hypermethylation Accompanies Human Structural Variation at 17p11.2. <i>Cell</i> , 2019, 176, 1310-1324.e10.	28.9	73
9	Distinct patterns of complex rearrangements and a mutational signature of microhomeology are frequently observed in PLP1 copy number gain structural variants. <i>Genome Medicine</i> , 2019, 11, 80.	8.2	24
10	Predicting human genes susceptible to genomic instability associated with Alu-mediated rearrangements. <i>Genome Research</i> , 2018, 28, 1228-1242.	5.5	74
11	Spliced integrated retrotransposed element (SplRE) formation in the human genome. <i>PLoS Biology</i> , 2018, 16, e2003067.	5.6	11
12	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 232-245.	2.9	261
13	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. <i>Genome Medicine</i> , 2016, 8, 105.	8.2	20
14	The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. <i>Genetics in Medicine</i> , 2016, 18, 443-451.	2.4	18
15	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. <i>PLoS Genetics</i> , 2015, 11, e1005686.	3.5	21
16	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. <i>Human Molecular Genetics</i> , 2015, 24, 4061-4077.	2.9	83
17	Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. <i>PLoS Genetics</i> , 2015, 11, e1005050.	3.5	57
18	PacBio-LITS: a large-insert targeted sequencing method for characterization of human disease-associated chromosomal structural variations. <i>BMC Genomics</i> , 2015, 16, 214.	2.8	63

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19	Assessing structural variation in a personal genome towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	2.8	153
20	Mus81 and converging forks limit the mutagenicity of replication fork breakage. Science, 2015, 349, 742-747.	12.6	162
21	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707.	6.2	33
22	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	8.2	136
23	Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia. European Journal of Human Genetics, 2014, 22, 1145-1148.	2.8	19
24	Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D. Genetics in Medicine, 2014, 16, 386-394.	2.4	30
25	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161.	6.2	87
26	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	6.2	148
27	Transduction-Specific ATLAS Reveals a Cohort of Highly Active L1 Retrotransposons in Human Populations. Human Mutation, 2013, 34, 974-985.	2.5	38
28	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. Human Mutation, 2013, 34, 1415-1423.	2.5	40
29	Curcumin facilitates a transitory cellular stress response in Trembler-J mice. Human Molecular Genetics, 2013, 22, 4698-4705.	2.9	43
30	LINE-1 Elements in Structural Variation and Disease. Annual Review of Genomics and Human Genetics, 2011, 12, 187-215.	6.2	471
31	Mutation screening of the IL-1 receptor antagonist gene in chronic non-bacterial osteomyelitis of childhood and adolescence. Clinical and Experimental Rheumatology, 2011, 29, 1040-3.	0.8	26
32	LINE-1 Retrotransposition Activity in Human Genomes. Cell, 2010, 141, 1159-1170.	28.9	531
33	Optimization of Feline Immunodeficiency Virus Vectors for RNA Interference. Journal of Virology, 2006, 80, 9371-9380.	3.4	29