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List of Publications by Year in descending order

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516710 752698 2,314 19 16 20 citations g-index h-index papers 20 20 20 4851 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Inherited Mutations in 17 Breast Cancer Susceptibility Genes Among a Large Triple-Negative Breast Cancer Cohort Unselected for Family History of Breast Cancer. Journal of Clinical Oncology, 2015, 33, 304-311.	1.6	521
2	Association of Type and Location of <i>BRCA1</i> And <i>BRCA2</i> Nutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
3	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor–negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	21.4	309
4	A review of a multifactorial probability-based model for classification of BRCA1 and BRCA2 variants of uncertain significance (VUS). Human Mutation, 2012, 33, 8-21.	2.5	190
5	Evaluation of ACMG-Guideline-Based Variant Classification of Cancer Susceptibility and Non-Cancer-Associated Genes in Families Affected by Breast Cancer. American Journal of Human Genetics, 2016, 98, 801-817.	6.2	113
6	Functional Assays for Analysis of Variants of Uncertain Significance in <i>BRCA2</i> . Human Mutation, 2014, 35, 151-164.	2.5	107
7	A Classification Model for <i>BRCA2</i> DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. Cancer Research, 2013, 73, 265-275.	0.9	103
8	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. Journal of Medical Genetics, 2012, 49, 525-532.	3.2	97
9	<i>BRCA2</i> Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
10	Splicing and multifactorial analysis of intronic BRCA1 and BRCA2 sequence variants identifies clinically significant splicing aberrations up to 12 nucleotides from the intron/exon boundary. Human Mutation, 2011, 32, 678-687.	2.5	74
11	BRCA2 Localization to the Midbody by Filamin A Regulates CEP55 Signaling and Completion of Cytokinesis. Developmental Cell, 2012, 23, 137-152.	7.0	74
12	Targeted exome analysis identifies the genetic basis of disease in over 50% of patients with a wide range of ataxia-related phenotypes. Genetics in Medicine, 2019, 21, 195-206.	2.4	65
13	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. American Journal of Human Genetics, 2018, 102, 233-248.	6.2	64
14	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	3.5	47
15	A yeast recombination assay to characterize human <i>BRCA1</i> missense variants of unknown pathological significance. Human Mutation, 2009, 30, 123-133.	2.5	39
16	A novel mutation in the promoter of RARS2 causes pontocerebellar hypoplasia in two siblings. Journal of Human Genetics, 2015, 60, 363-369.	2.3	26
17	Characterisation of gene expression profiles of yeast cells expressing BRCA1 missense variants. European Journal of Cancer, 2009, 45, 2187-2196.	2.8	6
18	A recombination-based method to characterize human BRCA1 missense variants. Breast Cancer Research and Treatment, 2011, 125, 265-272.	2.5	6

 #	Article	IF	CITATIONS
19	Effects on human transcriptome of mutated BRCA1 BRCT domain: A microarray study. BMC Cancer, 2012, 12, 207.	2.6	5