

Lucia Guidugli

List of Publications by Year in descending order

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Version: 2024-02-01

19
papers

2,314
citations

516710

16
h-index

752698

20
g-index

20
all docs

20
docs citations

20
times ranked

4851
citing authors

#	ARTICLE	IF	CITATIONS
1	Targeted exome analysis identifies the genetic basis of disease in over 50% of patients with a wide range of ataxia-related phenotypes. <i>Genetics in Medicine</i> , 2019, 21, 195-206.	2.4	65
2	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. <i>American Journal of Human Genetics</i> , 2018, 102, 233-248.	6.2	64
3	<i>BRCA2</i> Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , 2017, 77, 2789-2799.	0.9	75
4	Evaluation of ACMG-Guideline-Based Variant Classification of Cancer Susceptibility and Non-Cancer-Associated Genes in Families Affected by Breast Cancer. <i>American Journal of Human Genetics</i> , 2016, 98, 801-817.	6.2	113
5	Inherited Mutations in 17 Breast Cancer Susceptibility Genes Among a Large Triple-Negative Breast Cancer Cohort Unselected for Family History of Breast Cancer. <i>Journal of Clinical Oncology</i> , 2015, 33, 304-311.	1.6	521
6	Association of Type and Location of <i>BRCA1</i> and <i>BRCA2</i> Mutations With Risk of Breast and Ovarian Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2015, 313, 1347.	7.4	390
7	A novel mutation in the promoter of <i>RARS2</i> causes pontocerebellar hypoplasia in two siblings. <i>Journal of Human Genetics</i> , 2015, 60, 363-369.	2.3	26
8	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>PLoS Genetics</i> , 2014, 10, e1004256.	3.5	47
9	Functional Assays for Analysis of Variants of Uncertain Significance in <i>BRCA2</i> . <i>Human Mutation</i> , 2014, 35, 151-164.	2.5	107
10	A Classification Model for <i>BRCA2</i> DNA Binding Domain Missense Variants Based on Homology-Directed Repair Activity. <i>Cancer Research</i> , 2013, 73, 265-275.	0.9	103
11	<i>BRCA1</i> R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. <i>Journal of Medical Genetics</i> , 2012, 49, 525-532.	3.2	97
12	Effects on human transcriptome of mutated <i>BRCA1</i> BRCT domain: A microarray study. <i>BMC Cancer</i> , 2012, 12, 207.	2.6	5
13	<i>BRCA2</i> Localization to the Midbody by Filamin A Regulates CEP55 Signaling and Completion of Cytokinesis. <i>Developmental Cell</i> , 2012, 23, 137-152.	7.0	74
14	A review of a multifactorial probability-based model for classification of <i>BRCA1</i> and <i>BRCA2</i> variants of uncertain significance (VUS). <i>Human Mutation</i> , 2012, 33, 8-21.	2.5	190
15	A recombination-based method to characterize human <i>BRCA1</i> missense variants. <i>Breast Cancer Research and Treatment</i> , 2011, 125, 265-272.	2.5	6
16	Splicing and multifactorial analysis of intronic <i>BRCA1</i> and <i>BRCA2</i> sequence variants identifies clinically significant splicing aberrations up to 12 nucleotides from the intron/exon boundary. <i>Human Mutation</i> , 2011, 32, 678-687.	2.5	74
17	A locus on 19p13 modifies risk of breast cancer in <i>BRCA1</i> mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. <i>Nature Genetics</i> , 2010, 42, 885-892.	21.4	309
18	A yeast recombination assay to characterize human <i>BRCA1</i> missense variants of unknown pathological significance. <i>Human Mutation</i> , 2009, 30, 123-133.	2.5	39

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19	Characterisation of gene expression profiles of yeast cells expressing BRCA1 missense variants. European Journal of Cancer, 2009, 45, 2187-2196.	2.8	6