Kim De Leeneer

List of Publications by Year
in descending order

50 papers	3,332 citations	257450 24 h-index	223800 46 g-index
51 all docs	51 docs citations	51 times ranked	6954 citing authors

#	Article	IF	Citations
1	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
2	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
3	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
4	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
5	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
6	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	1.6	152
7	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	21.4	125
8	ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. Genetics in Medicine, 2019, 21, 1761-1771.	2.4	111
9	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. Clinical Chemistry, 2014, 60, 341-352.	3.2	95
10	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
11	Applying massive parallel sequencing to molecular diagnosis of Marfan and Loeys-Dietz syndromes. Human Mutation, 2011, 32, 1053-1062.	2.5	71
12	Rapid and Sensitive Detection of BRCA1/2 Mutations in a Diagnostic Setting: Comparison of Two High-Resolution Melting Platforms. Clinical Chemistry, 2008, 54, 982-989.	3.2	65
13	Massive parallel amplicon sequencing of the breast cancer genes BRCA1 and BRCA2: opportunities, challenges, and limitations. Human Mutation, 2011, 32, 335-344.	2.5	58
14	Novel pathogenic COL11A1/COL11A2 variants in Stickler syndrome detected by targeted NGS and exome sequencing. Molecular Genetics and Metabolism, 2014, 113, 230-235.	1.1	48
15	Mutation analysis of RAD51D in non-BRCA1/2 ovarian and breast cancer families. British Journal of Cancer, 2012, 106, 1460-1463.	6.4	43
16	Flexible, Scalable, and Efficient Targeted Resequencing on a Benchtop Sequencer for Variant Detection in Clinical Practice. Human Mutation, 2015, 36, 379-387.	2.5	43
17	BRCA1, BRCA2 and PALB2 mutations and CHEK2 c.1100delC in different South African ethnic groups diagnosed with premenopausal and/or triple negative breast cancer. BMC Cancer, 2015, 15, 912.	2.6	41
18	Practical Tools to Implement Massive Parallel Pyrosequencing of PCR Products in Next Generation Molecular Diagnostics. PLoS ONE, 2011, 6, e25531.	2.5	40

#	Article	IF	Citations
19	Non Coding RNA Molecules as Potential Biomarkers in Breast Cancer. Advances in Experimental Medicine and Biology, 2015, 867, 263-275.	1.6	32
20	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
21	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. Breast Cancer Research, 2016, 18, 64.	5.0	31
22	Prevalence of BRCA1/2 mutations in sporadic breast/ovarian cancer patients and identification of a novel de novo BRCA1 mutation in a patient diagnosed with late onset breast and ovarian cancer: implications for genetic testing. Breast Cancer Research and Treatment, 2012, 132, 87-95.	2.5	29
23	Variant Ataxia Telangiectasia: Clinical and Molecular Findings and Evaluation of Radiosensitive Phenotypes in a Patient and Relatives. NeuroMolecular Medicine, 2013, 15, 447-457.	3.4	29
24	Dealing with Pseudogenes in Molecular Diagnostics in the Next-Generation Sequencing Era. Methods in Molecular Biology, 2014, 1167, 303-315.	0.9	29
25	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61.	5.0	26
26	Analysis of chromosomal radiosensitivity of healthy BRCA2 mutation carriers and non-carriers in BRCA families with the G2 micronucleus assay. Oncology Reports, 2017, 37, 1379-1386.	2.6	26
27	Novel FRMD7 Mutations and Genomic Rearrangement Expand the Molecular Pathogenesis of X-Linked Idiopathic Infantile Nystagmus. Investigative Ophthalmology and Visual Science, 2015, 56, 1701-1710.	3.3	25
28	Increased chromosomal radiosensitivity in asymptomatic carriers of a heterozygous BRCA1 mutation. Breast Cancer Research, 2016, 18, 52.	5.0	25
29	Evaluation of RAD51C as cancer susceptibility gene in a large breast-ovarian cancer patient population referred for genetic testing. Breast Cancer Research and Treatment, 2012, 133, 393-398.	2.5	23
30	Genotyping of Frequent BRCA1/2 SNPs with Unlabeled Probes. Journal of Molecular Diagnostics, 2009, 11, 415-419.	2.8	21
31	Analysis of the Novel Fanconi Anemia GeneSLX4/FANCPin Familial Breast Cancer Cases. Human Mutation, 2013, 34, 70-73.	2.5	21
32	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
33	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	2.5	18
34	Analysing 454 amplicon resequencing experiments using the modular and database oriented Variant Identification Pipeline. BMC Bioinformatics, 2010, 11, 269.	2.6	15
35	BRCA1 and BRCA2 5â€² noncoding region variants identified in breast cancer patients alter promoter activity and protein binding. Human Mutation, 2018, 39, 2025-2039.	2.5	15
36	Evaluation of relative quantification of alternatively spliced transcripts using droplet digital PCR. Biomolecular Detection and Quantification, 2017, 13, 40-48.	7.0	12

#	Article	IF	Citations
37	Accurate detection and quantification of epigenetic and genetic second hits in BRCA1 and BRCA2 -associated hereditary breast and ovarian cancer reveals multiple co-acting second hits. Cancer Letters, 2018, 425, 125-133.	7.2	12
38	Diagnosis of Fanconi Anaemia by ionising radiation- or mitomycin C-induced micronuclei. DNA Repair, 2018, 61, 17-24.	2.8	12
39	Next Generation Sequencing to Determine the Cystic Fibrosis Mutation Spectrum in Palestinian Population. Disease Markers, 2015, 2015, 1-6.	1.3	11
40	Prevalence of Germline Pathogenic Variants in Cancer Predisposing Genes in Czech and Belgian Pancreatic Cancer Patients. Cancers, 2021, 13, 4430.	3.7	8
41	CRISPR-SID: Identifying EZH2 as a druggable target for desmoid tumors via inÂvivo dependency mapping. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	6
42	Thorough in silico and in vitro cDNA analysis of 21 putativeBRCA1andBRCA2splice variants and a complex tandem duplication inBRCA2allowing the identification of activated cryptic splice donor sites inBRCA2exon 11. Human Mutation, 2018, 39, 515-526.	2.5	5
43	Germline Genetic Findings Which May Impact Therapeutic Decisions in Families with a Presumed Predisposition for Hereditary Breast and Ovarian Cancer. Cancers, 2020, 12, 2151.	3.7	5
44	Targeted resequencing and variant validation using pxlence PCR assays. Biomolecular Detection and Quantification, 2016, 6, 22-26.	7.0	2
45	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.4	1
46	Dealing with Pseudogenes in Molecular Diagnostics in the Next Generation Sequencing Era. Methods in Molecular Biology, 2021, 2324, 363-381.	0.9	1
47	Second hit landscape in BRCA1/2-associated breast cancer. Annals of Oncology, 2017, 28, i9.	1.2	0
48	miRNA expression profiles in BRCA1-associated breast cancers reveal upregulation of specific miRNAs in tumors lacking a clear second hit in a large proportion of the tumour. Annals of Oncology, 2019, 30, iii72.	1.2	0
49	Novel and known FRMD7 mutations and copy number variation in Belgian patients with X-linked idiopathic infantile nystagmus. Acta Ophthalmologica, 2014, 92, 0-0.	1.1	0
50	Performance Evaluation of Three DNA Sample Tracking Tools in a Whole Exome Sequencing Workflow. Molecular Diagnosis and Therapy, 2022, 26, 411-419.	3.8	0