Doris Steinemann

List of Publications by Year
in descending order

110 papers	4,389 citations	159585 30 h-index	114465 63 g-index
110 all docs	110 docs citations	110 times ranked	9499 citing authors

#	Article	IF	Citations
1	Gene Therapy for Wiskott-Aldrich Syndromeâ€"Long-Term Efficacy and Genotoxicity. Science Translational Medicine, 2014, 6, 227ra33.	12.4	460
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 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
4	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
5	Childhood cancer predisposition syndromesâ€"A concise review and recommendations by the Cancer Predisposition Working Group of the Society for Pediatric Oncology and Hematology. American Journal of Medical Genetics, Part A, 2017, 173, 1017-1037.	1.2	200
6	IKZF1^{plus} Defines a New Minimal Residual Diseaseâ€"Dependent Very-Poor Prognostic Profile in Pediatric B-Cell Precursor Acute Lymphoblastic Leukemia. Journal of Clinical Oncology, 2018, 36, 1240-1249.	1.6	194
7	Direct Reprogramming of Hepatic Myofibroblasts into Hepatocytes InÂVivo Attenuates Liver Fibrosis. Cell Stem Cell, 2016, 18, 797-808.	11.1	181
8	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
9	Cooperativity of RUNX1 and CSF3R mutations in severe congenital neutropenia: a unique pathway in myeloid leukemogenesis. Blood, 2014, 123, 2229-2237.	1.4	135
10	Lobular breast cancer: Clinical, molecular and morphological characteristics. Pathology Research and Practice, 2016, 212, 583-597.	2.3	109
11	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. Human Molecular Genetics, 2016, 25, 2256-2268.	2.9	106
12	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
13	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
14	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	5.0	88
15	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	2.4	82
16	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nature Communications, 2020, 11, 1044.	12.8	81
17	TALEN-mediated functional correction of X-linked chronic granulomatous disease in patient-derived induced pluripotent stem cells. Biomaterials, 2015, 69, 191-200.	11.4	76
18	Phenotypic and molecular insights into CASK-related disorders in males. Orphanet Journal of Rare Diseases, 2015, 10, 44.	2.7	68

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19	A novel murine model of myeloproliferative disorders generated by overexpression of the transcription factor NF-E2. Journal of Experimental Medicine, 2012, 209, 35-50.	8.5	67
20	Mitotic recombination and compound-heterozygous mutations are predominant NF1-inactivating mechanisms in children with juvenile myelomonocytic leukemia and neurofibromatosis type 1. Haematologica, 2010, 95, 320-323.	3.5	58
21	Glycomic Characterization of Induced Pluripotent Stem Cells Derived from a Patient Suffering from Phosphomannomutase 2 Congenital Disorder of Glycosylation (PMM2-CDG). Molecular and Cellular Proteomics, 2016, 15, 1435-1452.	3.8	51
22	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
23	The identification of pathogenic variants in BRCA1/2 negative, high risk, hereditary breast and/or ovarian cancer patients: High frequency of FANCM pathogenic variants. International Journal of Cancer, 2019, 144, 2683-2694.	5.1	45
24	Assessment of Differentiation and Progression of Hepatic Tumors Using Array-Based Comparative Genomic Hybridization. Clinical Gastroenterology and Hepatology, 2006, 4, 1283-1291.	4.4	42
25	On metabolic reprogramming and tumor biology: A comprehensive survey of metabolism in breast cancer. Oncotarget, 2016, 7, 67626-67649.	1.8	42
26	MDS1 and EVI1 complex locus (MECOM): a novel candidate gene for hereditary hematological malignancies. Haematologica, 2018, 103, e55-e58.	3.5	41
27	Myb-like, SWIRM, and MPN domains 1 (MYSM1) deficiency: Genotoxic stress-associated bone marrow failure and developmental aberrations. Journal of Allergy and Clinical Immunology, 2017, 140, 1112-1119.	2.9	40
28	Identification of candidate tumor-suppressor genes in 6q27 by combined deletion mapping and electronic expression profiling in lymphoid neoplasms. Genes Chromosomes and Cancer, 2003, 37, 421-426.	2.8	39
29	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
30	The Clinical Utility of Optical Genome Mapping for the Assessment of Genomic Aberrations in Acute Lymphoblastic Leukemia. Cancers, 2021, 13, 4388.	3.7	37
31	Sustained Knockdown of a Disease-Causing Gene in Patient-Specific Induced Pluripotent Stem Cells Using Lentiviral Vector-Based Gene Therapy. Stem Cells Translational Medicine, 2013, 2, 641-654.	3.3	36
32	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
33	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
34	Improved bi-allelic modification of a transcriptionally silent locus in patient-derived iPSC by Cas9 nickase. Scientific Reports, 2016, 6, 38198.	3.3	29
35	CDKN2A loss and PIK3CA mutation in myoepithelialâ€like metaplastic breast cancer. Journal of Pathology, 2018, 245, 373-383.	4.5	28
36	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

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37	E-cadherin to P-cadherin switching in lobular breast cancer with tubular elements. Modern Pathology, 2020, 33, 2483-2498.	5.5	26
38	Haploinsufficiency of ETV6 and CDKN1B in patients with acute myeloid leukemia and complex karyotype. BMC Genomics, 2014, 15, 784.	2.8	25
39	Modified Lentiviral LTRs Allow Flp Recombinaseâ€“mediated Cassette Exchange and In Vivo Tracing of â€œFactor-freeâ€•Induced Pluripotent Stem Cells. Molecular Therapy, 2014, 22, 919-928.	8.2	24
40	Altered NFE2 activity predisposes to leukemic transformation and myelosarcoma with AML-specific aberrations. Blood, 2019, 133, 1766-1777.	1.4	23
41	Implementation of RNA sequencing and array CGH in the diagnostic workflow of the AIEOP-BFM ALL 2017 trial on acute lymphoblastic leukemia. Annals of Hematology, 2020, 99, 809-818.	1.8	23
42	Quantitative microsatellite analysis to delineate the commonly deleted region 1p22.3 in mantle cell lymphomas. Genes Chromosomes and Cancer, 2006, 45, 883-892.	2.8	22
43	A child with Liâ€“Fraumeni syndrome: Modes to inactivate the second allele of TP53 in three different malignancies. Pediatric Blood and Cancer, 2015, 62, 1481-1484.	1.5	22
44	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	2.5	22
45	Copy number alterations in childhood acute lymphoblastic leukemia and their association with minimal residual disease. Genes Chromosomes and Cancer, 2008, 47, 471-480.	2.8	21
46	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2019, 121, 180-192.	6.4	19
47	A selectable all-in-one CRISPR prime editing piggyBac transposon allows for highly efficient gene editing in human cell lines. Scientific Reports, 2021, 11, 22154.	3.3	19
48	Clinical and genetic characteristics of children with acute lymphoblastic leukemia and Liâ€“Fraumeni syndrome. Leukemia, 2021, 35, 1475-1479.	7.2	17
49	Frequency and prognostic impact of <scp>PAX5</scp> p.<scp>P80R</scp> in pediatric acute lymphoblastic leukemia patients treated on an <scp>AIEOPâ€BFM</scp> acute lymphoblastic leukemia protocol. Genes Chromosomes and Cancer, 2020, 59, 667-671.	2.8	16
50	Optical Genome Mapping as a Diagnostic Tool in Pediatric Acute Myeloid Leukemia. Cancers, 2022, 14, 2058.	3.7	16
51	Clonal heterogeneity in childhood myelodysplastic syndromesâ€”Challenge for the detection of chromosomal imbalances by arrayâ€CGH. Genes Chromosomes and Cancer, 2010, 49, 885-900.	2.8	15
52	A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency. European Journal of Human Genetics, 2018, 26, 1773-1783.	2.8	15
53	Venetoclax and dexamethasone synergize with inotuzumab ozogamicinâ€“induced DNA damage signaling in B-lineage ALL. Blood, 2021, 137, 2657-2661.	1.4	15
54	NCAM2 deletion in a boy with macrocephaly and autism: Cause, association or predisposition?. European Journal of Medical Genetics, 2016, 59, 493-498.	1.3	13

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55	iPSC modeling of stage-specific leukemogenesis reveals BAALC as a key oncogene in severe congenital neutropenia. Cell Stem Cell, 2021, 28, 906-922.e6.	11.1	13
56	The heteromeric transcription factor GABP activates the ITGAM/CD11b promoter and induces myeloid differentiation. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2015, 1849, 1145-1154.	1.9	12
57	A tandem duplication of BRCA1 exons 1â€“19 through DHX8 exon 2 in four families with hereditary breast and ovarian cancer syndrome. Breast Cancer Research and Treatment, 2018, 172, 561-569.	2.5	12
58	Functional classification of RUNX1 variants in familial platelet disorder with associated myeloid malignancies. Leukemia, 2021, 35, 3304-3308.	7.2	11
59	Precise ERBB2 copy number assessment in breast cancer by means of molecular inversion probe array analysis. Oncotarget, 2016, 7, 82733-82740.	1.8	11
60	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS ONE, 2016, 11, e0158801.	2.5	10
61	Breast cancer patients suggestive of Li-Fraumeni syndrome: mutational spectrum, candidate genes, and unexplained heredity. Breast Cancer Research, 2018, 20, 87.	5.0	9
62	GT198 (PSMC3IP) germline variants in early-onset breast cancer patients from hereditary breast and ovarian cancer families. Genes and Cancer, 2017, 8, 472-483.	1.9	9
63	Analysis of Array-CGH Data Using the R and Bioconductor Software Suite. Comparative and Functional Genomics, 2009, 2009, 1-8.	2.0	8
64	The stem cell zinc finger 1 (SZF1)/ZNF589 protein has a human-specific evolutionary nucleotide DNA change and acts as a regulator of cell viability inÂthe hematopoietic system. Experimental Hematology, 2016, 44, 257-268.	0.4	8
65	Looking for the hidden mutation: Bannayanâ€“Rileyâ€“Ruvalcaba syndrome caused by constitutional and mosaic 10q23 microdeletions involving PTEN and BMPR1A. American Journal of Medical Genetics, Part A, 2019, 179, 1383-1389.	1.2	8
66	Lentiviral gene therapy and vitamin B3 treatment enable granulocytic differentiation of G6PC3-deficient induced pluripotent stem cells. Gene Therapy, 2020, 27, 297-306.	4.5	8
67	12q14 microdeletion syndrome: A family with short stature and Silver-Russell syndrome (SRS)-like phenotype and review of the literature. European Journal of Medical Genetics, 2018, 61, 421-427.	1.3	7
68	GABP is necessary for stem/progenitor cell maintenance and myeloid differentiation in human hematopoiesis and chronic myeloid leukemia. Stem Cell Research, 2016, 16, 677-681.	0.7	6
69	KBG syndrome patient due to 16q24.3 microdeletion presenting with a paratesticular rhabdoid tumor: Coincidence or cancer predisposition?. American Journal of Medical Genetics, Part A, 2018, 176, 1449-1454.	1.2	6
70	Copy Number Analysis in a Large Cohort Suggestive of Inborn Errors of Immunity Indicates a Wide Spectrum of Relevant Chromosomal Losses and Gains. Journal of Clinical Immunology, 2022, 42, 1083-1092.	3.8	6
71	Expression of the ETS transcription factor GABPÎ± is positively correlated toÂthe BCR-ABL1/ABL1 ratio in CML patients and affects imatinib sensitivity inÂvitro. Experimental Hematology, 2015, 43, 880-890.	0.4	5
72	Mechanism of allele specific assembly and disruption of master regulator transcription factor complexes of NF- K Bp50, NF- K Bp65 and HIF1a on a non-coding FAS SNP. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2016, 1859, 1411-1428.	1.9	5

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73	Clinical utility gene card for: Familial platelet disorder with associated myeloid malignancies. European Journal of Human Genetics, 2016, 24, 3-4.	2.8	5
74	From a variant of unknown significance to pathogenic: Reclassification of a large novel duplication in BRCA2 by highâ€throughput sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1045.	1.2	5
75	De novo missense variants in the <scp>RAP1B</scp> gene identified in two patients with syndromic thrombocytopenia. Clinical Genetics, 2020, 98, 374-378.	2.0	5
76	Genetic Correction of IL-10RB Deficiency Reconstitutes Anti-Inflammatory Regulation in iPSC-Derived Macrophages. Journal of Personalized Medicine, 2021, 11, 221.	2.5	5
77	Generation of three induced pluripotent cell lines (iPSCs) from an Aicardiâ€“GoutiÃ¨res syndrome (AGS) patient harboring a deletion in the genomic locus of the sterile alpha motif and HD domain containing protein 1 (SAMHD1). Stem Cell Research, 2020, 43, 101697.	0.7	4
78	Cryptic <scp>TCF3</scp> fusions in childhood leukemia: Detection by <scp>RNA</scp> sequencing. Genes Chromosomes and Cancer, 2022, 61, 22-26.	2.8	4
79	Poor Prognosis in Children with ABL-Class Fusion Positive B-Cell Acute Lymphoblastic Leukemia Treated According to AIEOP-BFM Protocols. Blood, 2019, 134, 1351-1351.	1.4	4
80	Mutation analysis of the HAX1 gene in childhood myelodysplastic syndrome. British Journal of Haematology, 2009, 145, 533-534.	2.5	3
81	BCR-ABL1 positive AML or CML in blast crisis? A pediatric case report with inv(3) and t(9;22) in the initial clone. Cancer Genetics, 2021, 254-255, 70-74.	0.4	3
82	Chromosome 2q gain and epigenetic silencing of <scp>GATA3</scp> in microglandular adenosis of the breast. Journal of Pathology: Clinical Research, 2021, 7, 220-232.	3.0	3
83	Unbalanced translocation der(5;17) resulting in a TP53 loss as recurrent aberration in myelodysplastic syndrome and acute myeloid leukemia with complex karyotype. Genes Chromosomes and Cancer, 2021, 60, 452-457.	2.8	2
84	BRCA1/2 mutation prevalence in triple-negative breast cancer patients without family history of breast and ovarian cancer.. Journal of Clinical Oncology, 2016, 34, 1090-1090.	1.6	2
85	Array-CGH in Childhood MDS. Methods in Molecular Biology, 2013, 973, 267-278.	0.9	1
86	Octasomy 21 in a patient with secondary AML after CMML: the role of acquired NRAS mutations in triggering aneuploidy. Leukemia and Lymphoma, 2018, 59, 2478-2481.	1.3	1
87	Induced pluripotent stem cells (iPSCs) derived from a renpenning syndrome patient with c.459_462delAGAG mutation in PQBP1 (PEIi001-A). Stem Cell Research, 2019, 41, 101592.	0.7	1
88	Induced pluripotent stem cell line (PEIi003-A) derived from an apparently healthy male individual. Stem Cell Research, 2020, 42, 101679.	0.7	1
89	Cooperativity Of RUNX1 and CSF3R Mutations In The Development Of Leukemia In Severe Congenital Neutropenia: A Unique Pathway In Myeloid Leukemogenesis. Blood, 2013, 122, 444-444.	1.4	1
90	Clinical Heterogeneity in RUNX1-Associated Familial Myelodysplastic Syndrome - Report of Two Novel Pedigrees with Childhoodleukemia. Blood, 2016, 128, 5509-5509.	1.4	1

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91	Establishment and Characterization of a Pair of Patient-derived Human Non-small Cell Lung Cancer Cell Lines from a Primary Tumor and Corresponding Lymph Node Metastasis. Anticancer Research, 2016, 36, 1507-18.	1.1	1
92	Donorâ€transmitted extramedullary acute myeloid leukaemia after living donor kidney transplantation. British Journal of Haematology, 2022, , .	2.5	1
93	Two cancerâ€predisposing variants in one family: Incidental finding of a fumarate hydrogenase (FH) germline variant in a family with Liâ€“Fraumeni syndrome. Pediatric Blood and Cancer, 2018, 65, e27254.	1.5	0
94	Drug-Response Signature Predicts Outcome in Adult Acute Myeloid Leukemia and Associates Poor Response with Molecular Characteristics of Hematopoietic Stem Cells.. Blood, 2004, 104, 2024-2024.	1.4	0
95	Fluorescence In Situ Hybridization (FISH) Using a Subtelomeric 11q Probe as a New Diagnostic Tool for Congenital Thrombocytopenia Caused by Deletions in 11q.. Blood, 2004, 104, 3035-3035.	1.4	0
96	Characterization of Secondary Alterations in Mantle Cell Lymphomas Using Matrix-/Array CGH.. Blood, 2004, 104, 4349-4349.	1.4	0
97	Gain of Chromosome 21 Is Associated with Early Treatment Sensitivity in Childhood Acute Lymphoblastic Leukemia.. Blood, 2005, 106, 1440-1440.	1.4	0
98	Genome-Wide Single Nucleotide Polymorphism Analysis in Juvenile Myelomonocytic Leukemia Uncovers Long-Range Uniparental Disomy Surrounding the NF1 Locus in Cases Associated with Type 1 Neurofibromatosis but Not in Cases with Mutant RAS or PTPN11.. Blood, 2006, 108, 1453-1453.	1.4	0
99	Significance of Copy Number Alterations for Molecular Treatment Response in Childhood Acute Lymphoblastic Leukemia.. Blood, 2007, 110, 1434-1434.	1.4	0
100	Gain of the Centromeric Region of Chromosome 8, a New Germline Alteration In Juvenile Myelomonocytic Leukemia. Blood, 2010, 116, 4830-4830.	1.4	0
101	Evidence for Cooperation of Receptor Tyrosine Kinases and Activating NOTCH Mutations to Hyperactivate mTOR in T-Cell Leukemia: A Rationale Basis for Targeted Therapy. Blood, 2011, 118, 1381-1381.	1.4	0
102	Lentiviral Vector Induced Insertional Haploinsufficiency of Ebf1 Causes Leukemia in a Murine Bone Marrow Transplantation Model. Blood, 2011, 118, 671-671.	1.4	0
103	Receptor Tyrosine Kinases, Notch Mutations and PTEN Loss Converge On mTOR in T-ALL and Cause Addiction to Cap Dependent mRNA Translation.. Blood, 2012, 120, 2415-2415.	1.4	0
104	RUNX1 Mutations Are the Most Frequent Leukemia Associated Mutations in Congenital Neutropenia Patients. Blood, 2012, 120, 7-7.	1.4	0
105	Lentivirally Transduced Human Cord Blood CD34+FLT3-ITD+ Cells Induce Murine Acute Leukemia in the NOD/SCID Transplantation Model.. Blood, 2012, 120, 2984-2984.	1.4	0
106	BCR-ABL Cooperates With a â€œTelomere-Associated Secretory Phenotypeâ€•(TASP) To Facilitate Malignant Proliferation Of Hematopoietic Stem Cells. Blood, 2013, 122, 3976-3976.	1.4	0
107	Familial AML With Germline CEBPA Mutations: Extended Clinical Outcomes and Analysis Of Secondary Mutations Using Whole Exome Sequencing. Blood, 2013, 122, 740-740.	1.4	0
108	Unbalanced Translocation t(5;17) Resulting In a TP53 Loss As Recurrent Aberration In Myelodysplastic Syndrome With Complex Karyotype. Blood, 2013, 122, 4949-4949.	1.4	0

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109	Expression of the ETS Transcription Factor GABPÎ± Is Correlated to BCR-ABL/ABL ratio in Human CML and Mediates Imatinib Sensitivity. Blood, 2014, 124, 1787-1787.	1.4	0
110	Clonal Evolution at First Sight: A Combined Visualization of Diverse Diagnostic Methods Improves Understanding of Leukemia Progression. Blood, 2021, 138, 1293-1293.	1.4	0