

Doris Steinemann

List of Publications by Year in descending order

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Version: 2024-02-01

110
papers

4,389
citations

159358

30
h-index

114278

63
g-index

110
all docs

110
docs citations

110
times ranked

9499
citing authors

#	ARTICLE	IF	CITATIONS
1	Gene Therapy for Wiskott-Aldrich Syndrome—Long-Term Efficacy and Genotoxicity. <i>Science Translational Medicine</i> , 2014, 6, 227ra33.	5.8	460
2	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.	3.8	390
3	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.	9.4	356
4	Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. <i>Human Mutation</i> , 2018, 39, 593-620.	1.1	224
5	Childhood cancer predisposition syndromes—A concise review and recommendations by the Cancer Predisposition Working Group of the Society for Pediatric Oncology and Hematology. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1017-1037.	0.7	200
6	<i>KZF1</i> Defines a New Minimal Residual Disease-Dependent Very-Poor Prognostic Profile in Pediatric B-Cell Precursor Acute Lymphoblastic Leukemia. <i>Journal of Clinical Oncology</i> , 2018, 36, 1240-1249.	0.8	194
7	Direct Reprogramming of Hepatic Myofibroblasts into Hepatocytes In Vivo Attenuates Liver Fibrosis. <i>Cell Stem Cell</i> , 2016, 18, 797-808.	5.2	181
8	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250.	0.8	152
9	Cooperativity of <i>RUNX1</i> and <i>CSF3R</i> mutations in severe congenital neutropenia: a unique pathway in myeloid leukemogenesis. <i>Blood</i> , 2014, 123, 2229-2237.	0.6	135
10	Lobular breast cancer: Clinical, molecular and morphological characteristics. <i>Pathology Research and Practice</i> , 2016, 212, 583-597.	1.0	109
11	Combined genetic and splicing analysis of <i>BRCA1</i> c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. <i>Human Molecular Genetics</i> , 2016, 25, 2256-2268.	1.4	106
12	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	1.1	102
13	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375.	5.8	93
14	Male breast cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers: pathology data from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> . <i>Breast Cancer Research</i> , 2016, 18, 15.	2.2	88
15	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of <i>BRCA1</i> and <i>BRCA2</i> pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666.	1.1	82
16	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. <i>Nature Communications</i> , 2020, 11, 1044.	5.8	81
17	TALEN-mediated functional correction of X-linked chronic granulomatous disease in patient-derived induced pluripotent stem cells. <i>Biomaterials</i> , 2015, 69, 191-200.	5.7	76
18	Phenotypic and molecular insights into <i>CASK</i> -related disorders in males. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 44.	1.2	68

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19	A novel murine model of myeloproliferative disorders generated by overexpression of the transcription factor NF-E2. <i>Journal of Experimental Medicine</i> , 2012, 209, 35-50.	4.2	67
20	Mitotic recombination and compound-heterozygous mutations are predominant NF1-inactivating mechanisms in children with juvenile myelomonocytic leukemia and neurofibromatosis type 1. <i>Haematologica</i> , 2010, 95, 320-323.	1.7	58
21	Glycomic Characterization of Induced Pluripotent Stem Cells Derived from a Patient Suffering from Phosphomannomutase 2 Congenital Disorder of Glycosylation (PMM2-CDC). <i>Molecular and Cellular Proteomics</i> , 2016, 15, 1435-1452.	2.5	51
22	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS Genetics</i> , 2014, 10, e1004256.	1.5	47
23	The identification of pathogenic variants in <i>BRCA1/2</i> negative, high risk, hereditary breast and/or ovarian cancer patients: High frequency of <i>FANCM</i> pathogenic variants. <i>International Journal of Cancer</i> , 2019, 144, 2683-2694.	2.3	45
24	Assessment of Differentiation and Progression of Hepatic Tumors Using Array-Based Comparative Genomic Hybridization. <i>Clinical Gastroenterology and Hepatology</i> , 2006, 4, 1283-1291.	2.4	42
25	On metabolic reprogramming and tumor biology: A comprehensive survey of metabolism in breast cancer. <i>Oncotarget</i> , 2016, 7, 67626-67649.	0.8	42
26	<i>MDS1</i> and <i>EVI1</i> complex locus (MECOM): a novel candidate gene for hereditary hematological malignancies. <i>Haematologica</i> , 2018, 103, e55-e58.	1.7	41
27	Myb-like, SWIRM, and MPN domains 1 (MYSM1) deficiency: Genotoxic stress-associated bone marrow failure and developmental aberrations. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 140, 1112-1119.	1.5	40
28	Identification of candidate tumor-suppressor genes in 6q27 by combined deletion mapping and electronic expression profiling in lymphoid neoplasms. <i>Genes Chromosomes and Cancer</i> , 2003, 37, 421-426.	1.5	39
29	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638.	0.4	39
30	The Clinical Utility of Optical Genome Mapping for the Assessment of Genomic Aberrations in Acute Lymphoblastic Leukemia. <i>Cancers</i> , 2021, 13, 4388.	1.7	37
31	Sustained Knockdown of a Disease-Causing Gene in Patient-Specific Induced Pluripotent Stem Cells Using Lentiviral Vector-Based Gene Therapy. <i>Stem Cells Translational Medicine</i> , 2013, 2, 641-654.	1.6	36
32	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. <i>PLoS ONE</i> , 2015, 10, e0120020.	1.1	34
33	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. <i>Breast Cancer Research</i> , 2016, 18, 64.	2.2	31
34	Improved bi-allelic modification of a transcriptionally silent locus in patient-derived iPSC by Cas9 nickase. <i>Scientific Reports</i> , 2016, 6, 38198.	1.6	29
35	<i>CDKN2A</i> loss and <i>PIK3CA</i> mutation in myoepithelial-like metaplastic breast cancer. <i>Journal of Pathology</i> , 2018, 245, 373-383.	2.1	28
36	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2015, 17, 61.	2.2	26

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37	E-cadherin to P-cadherin switching in lobular breast cancer with tubular elements. <i>Modern Pathology</i> , 2020, 33, 2483-2498.	2.9	26
38	Haploinsufficiency of ETV6 and CDKN1B in patients with acute myeloid leukemia and complex karyotype. <i>BMC Genomics</i> , 2014, 15, 784.	1.2	25
39	Modified Lentiviral LTRs Allow Flp Recombinase-mediated Cassette Exchange and In Vivo Tracing of Factor-free-Induced Pluripotent Stem Cells. <i>Molecular Therapy</i> , 2014, 22, 919-928.	3.7	24
40	Altered NFE2 activity predisposes to leukemic transformation and myelosarcoma with AML-specific aberrations. <i>Blood</i> , 2019, 133, 1766-1777.	0.6	23
41	Implementation of RNA sequencing and array CGH in the diagnostic workflow of the AIEOP-BFM ALL 2017 trial on acute lymphoblastic leukemia. <i>Annals of Hematology</i> , 2020, 99, 809-818.	0.8	23
42	Quantitative microsatellite analysis to delineate the commonly deleted region 1p22.3 in mantle cell lymphomas. <i>Genes Chromosomes and Cancer</i> , 2006, 45, 883-892.	1.5	22
43	A child with Fraumeni syndrome: Modes to inactivate the second allele of TP53 in three different malignancies. <i>Pediatric Blood and Cancer</i> , 2015, 62, 1481-1484.	0.8	22
44	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 308-316.	1.1	22
45	Copy number alterations in childhood acute lymphoblastic leukemia and their association with minimal residual disease. <i>Genes Chromosomes and Cancer</i> , 2008, 47, 471-480.	1.5	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. <i>British Journal of Cancer</i> , 2019, 121, 180-192.	2.9	19
47	A selectable all-in-one CRISPR prime editing piggyBac transposon allows for highly efficient gene editing in human cell lines. <i>Scientific Reports</i> , 2021, 11, 22154.	1.6	19
48	Clinical and genetic characteristics of children with acute lymphoblastic leukemia and Fraumeni syndrome. <i>Leukemia</i> , 2021, 35, 1475-1479.	3.3	17
49	Frequency and prognostic impact of PAX5 p.P80R in pediatric acute lymphoblastic leukemia patients treated on an AIEOP-BFM acute lymphoblastic leukemia protocol. <i>Genes Chromosomes and Cancer</i> , 2020, 59, 667-671.	1.5	16
50	Optical Genome Mapping as a Diagnostic Tool in Pediatric Acute Myeloid Leukemia. <i>Cancers</i> , 2022, 14, 2058.	1.7	16
51	Clonal heterogeneity in childhood myelodysplastic syndromes—Challenge for the detection of chromosomal imbalances by array-CGH. <i>Genes Chromosomes and Cancer</i> , 2010, 49, 885-900.	1.5	15
52	A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 1773-1783.	1.4	15
53	Venetoclax and dexamethasone synergize with inotuzumab ozogamicin-induced DNA damage signaling in B-lineage ALL. <i>Blood</i> , 2021, 137, 2657-2661.	0.6	15
54	NCAM2 deletion in a boy with macrocephaly and autism: Cause, association or predisposition?. <i>European Journal of Medical Genetics</i> , 2016, 59, 493-498.	0.7	13

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55	iPSC modeling of stage-specific leukemogenesis reveals BAALC as a key oncogene in severe congenital neutropenia. <i>Cell Stem Cell</i> , 2021, 28, 906-922.e6.	5.2	13
56	The heteromeric transcription factor GABP activates the ITGAM/CD11b promoter and induces myeloid differentiation. <i>Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms</i> , 2015, 1849, 1145-1154.	0.9	12
57	A tandem duplication of BRCA1 exons 19 through DHX8 exon 2 in four families with hereditary breast and ovarian cancer syndrome. <i>Breast Cancer Research and Treatment</i> , 2018, 172, 561-569.	1.1	12
58	Functional classification of RUNX1 variants in familial platelet disorder with associated myeloid malignancies. <i>Leukemia</i> , 2021, 35, 3304-3308.	3.3	11
59	Precise ERBB2 copy number assessment in breast cancer by means of molecular inversion probe array analysis. <i>Oncotarget</i> , 2016, 7, 82733-82740.	0.8	11
60	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS ONE</i> , 2016, 11, e0158801.	1.1	10
61	Breast cancer patients suggestive of Li-Fraumeni syndrome: mutational spectrum, candidate genes, and unexplained heredity. <i>Breast Cancer Research</i> , 2018, 20, 87.	2.2	9
62	GT198 (PSMC3IP) germline variants in early-onset breast cancer patients from hereditary breast and ovarian cancer families. <i>Genes and Cancer</i> , 2017, 8, 472-483.	0.6	9
63	Analysis of Array-CGH Data Using the R and Bioconductor Software Suite. <i>Comparative and Functional Genomics</i> , 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 in the hematopoietic system. <i>Experimental Hematology</i> , 2016, 44, 257-268.	0.2	8
65	Looking for the hidden mutation: Bannayan-Ruvalcaba syndrome caused by constitutional and mosaic 10q23 microdeletions involving PTEN and BMPRIA. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1383-1389.	0.7	8
66	Lentiviral gene therapy and vitamin B3 treatment enable granulocytic differentiation of G6PC3-deficient induced pluripotent stem cells. <i>Gene Therapy</i> , 2020, 27, 297-306.	2.3	8
67	12q14 microdeletion syndrome: A family with short stature and Silver-Russell syndrome (SRS)-like phenotype and review of the literature. <i>European Journal of Medical Genetics</i> , 2018, 61, 421-427.	0.7	7
68	GABP is necessary for stem/progenitor cell maintenance and myeloid differentiation in human hematopoiesis and chronic myeloid leukemia. <i>Stem Cell Research</i> , 2016, 16, 677-681.	0.3	6
69	KBG syndrome patient due to 16q24.3 microdeletion presenting with a paratesticular rhabdoid tumor: Coincidence or cancer predisposition?. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1449-1454.	0.7	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. <i>Journal of Clinical Immunology</i> , 2022, 42, 1083-1092.	2.0	6
71	Expression of the ETS transcription factor GABP is positively correlated to the BCR-ABL1/ABL1 ratio in CML patients and affects imatinib sensitivity in vitro. <i>Experimental Hematology</i> , 2015, 43, 880-890.	0.2	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. <i>Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms</i> , 2016, 1859, 1411-1428.	0.9	5

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73	Clinical utility gene card for: Familial platelet disorder with associated myeloid malignancies. <i>European Journal of Human Genetics</i> , 2016, 24, 3-4.	1.4	5
74	From a variant of unknown significance to pathogenic: Reclassification of a large novel duplication in <i>BRCA2</i> by high-throughput sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1045.	0.6	5
75	<i>De novo</i> missense variants in the <i>RAP1B</i> gene identified in two patients with syndromic thrombocytopenia. <i>Clinical Genetics</i> , 2020, 98, 374-378.	1.0	5
76	Genetic Correction of IL-10RB Deficiency Reconstitutes Anti-Inflammatory Regulation in iPSC-Derived Macrophages. <i>Journal of Personalized Medicine</i> , 2021, 11, 221.	1.1	5
77	Generation of three induced pluripotent cell lines (iPSCs) from an Aicardi-Goutières syndrome (AGS) patient harboring a deletion in the genomic locus of the sterile alpha motif and HD domain containing protein 1 (SAMHD1). <i>Stem Cell Research</i> , 2020, 43, 101697.	0.3	4
78	Cryptic <i>TCF3</i> fusions in childhood leukemia: Detection by RNA sequencing. <i>Genes Chromosomes and Cancer</i> , 2022, 61, 22-26.	1.5	4
79	Poor Prognosis in Children with ABL-Class Fusion Positive B-Cell Acute Lymphoblastic Leukemia Treated According to AIEOP-BFM Protocols. <i>Blood</i> , 2019, 134, 1351-1351.	0.6	4
80	Mutation analysis of the <i>HAX1</i> gene in childhood myelodysplastic syndrome. <i>British Journal of Haematology</i> , 2009, 145, 533-534.	1.2	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. <i>Cancer Genetics</i> , 2021, 254-255, 70-74.	0.2	3
82	Chromosome 2q gain and epigenetic silencing of <i>GATA3</i> in microglandular adenosis of the breast. <i>Journal of Pathology: Clinical Research</i> , 2021, 7, 220-232.	1.3	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. <i>Genes Chromosomes and Cancer</i> , 2021, 60, 452-457.	1.5	2
84	<i>BRCA1/2</i> mutation prevalence in triple-negative breast cancer patients without family history of breast and ovarian cancer. <i>Journal of Clinical Oncology</i> , 2016, 34, 1090-1090.	0.8	2
85	Array-CGH in Childhood MDS. <i>Methods in Molecular Biology</i> , 2013, 973, 267-278.	0.4	1
86	Octasomy 21 in a patient with secondary AML after CMML: the role of acquired <i>NRAS</i> mutations in triggering aneuploidy. <i>Leukemia and Lymphoma</i> , 2018, 59, 2478-2481.	0.6	1
87	Induced pluripotent stem cells (iPSCs) derived from a reppening syndrome patient with c.459_462delAGAG mutation in PQBP1 (PEIi001-A). <i>Stem Cell Research</i> , 2019, 41, 101592.	0.3	1
88	Induced pluripotent stem cell line (PEIi003-A) derived from an apparently healthy male individual. <i>Stem Cell Research</i> , 2020, 42, 101679.	0.3	1
89	Cooperativity Of RUNX1 and CSF3R Mutations In The Development Of Leukemia In Severe Congenital Neutropenia: A Unique Pathway In Myeloid Leukemogenesis. <i>Blood</i> , 2013, 122, 444-444.	0.6	1
90	Clinical Heterogeneity in RUNX1-Associated Familial Myelodysplastic Syndrome - Report of Two Novel Pedigrees with Childhoodleukemia. <i>Blood</i> , 2016, 128, 5509-5509.	0.6	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. <i>Anticancer Research</i> , 2016, 36, 1507-18.	0.5	1
92	Donor-transmitted extramedullary acute myeloid leukaemia after living donor kidney transplantation. <i>British Journal of Haematology</i> , 2022, , .	1.2	1
93	Two cancer-predisposing variants in one family: Incidental finding of a fumarate hydrogenase (<i>FH</i>) germline variant in a family with Li-Fraumeni syndrome. <i>Pediatric Blood and Cancer</i> , 2018, 65, e27254.	0.8	0
94	Drug-Response Signature Predicts Outcome in Adult Acute Myeloid Leukemia and Associates Poor Response with Molecular Characteristics of Hematopoietic Stem Cells.. <i>Blood</i> , 2004, 104, 2024-2024.	0.6	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.. <i>Blood</i> , 2004, 104, 3035-3035.	0.6	0
96	Characterization of Secondary Alterations in Mantle Cell Lymphomas Using Matrix-/Array CGH.. <i>Blood</i> , 2004, 104, 4349-4349.	0.6	0
97	Gain of Chromosome 21 Is Associated with Early Treatment Sensitivity in Childhood Acute Lymphoblastic Leukemia.. <i>Blood</i> , 2005, 106, 1440-1440.	0.6	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.. <i>Blood</i> , 2006, 108, 1453-1453.	0.6	0
99	Significance of Copy Number Alterations for Molecular Treatment Response in Childhood Acute Lymphoblastic Leukemia.. <i>Blood</i> , 2007, 110, 1434-1434.	0.6	0
100	Gain of the Centromeric Region of Chromosome 8, a New Germline Alteration In Juvenile Myelomonocytic Leukemia. <i>Blood</i> , 2010, 116, 4830-4830.	0.6	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. <i>Blood</i> , 2011, 118, 1381-1381.	0.6	0
102	Lentiviral Vector Induced Insertional Haploinsufficiency of Ebf1 Causes Leukemia in a Murine Bone Marrow Transplantation Model. <i>Blood</i> , 2011, 118, 671-671.	0.6	0
103	Receptor Tyrosine Kinases, Notch Mutations and PTEN Loss Converge On mTOR in T-ALL and Cause Addiction to Cap Dependent mRNA Translation.. <i>Blood</i> , 2012, 120, 2415-2415.	0.6	0
104	RUNX1 Mutations Are the Most Frequent Leukemia Associated Mutations in Congenital Neutropenia Patients. <i>Blood</i> , 2012, 120, 7-7.	0.6	0
105	Lentivirally Transduced Human Cord Blood CD34+FLT3-ITD+ Cells Induce Murine Acute Leukemia in the NOD/SCID Transplantation Model.. <i>Blood</i> , 2012, 120, 2984-2984.	0.6	0
106	BCR-ABL Cooperates With a Telomere-Associated Secretory Phenotype (TASP) To Facilitate Malignant Proliferation Of Hematopoietic Stem Cells. <i>Blood</i> , 2013, 122, 3976-3976.	0.6	0
107	Familial AML With Germline CEBPA Mutations: Extended Clinical Outcomes and Analysis Of Secondary Mutations Using Whole Exome Sequencing. <i>Blood</i> , 2013, 122, 740-740.	0.6	0
108	Unbalanced Translocation t(5;17) Resulting In a TP53 Loss As Recurrent Aberration In Myelodysplastic Syndrome With Complex Karyotype. <i>Blood</i> , 2013, 122, 4949-4949.	0.6	0

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109	Expression of the ETS Transcription Factor GABP \hat{I} ± Is Correlated to BCR-ABL/ABL ratio in Human CML and Mediates Imatinib Sensitivity. <i>Blood</i> , 2014, 124, 1787-1787.	0.6	0
110	Clonal Evolution at First Sight: A Combined Visualization of Diverse Diagnostic Methods Improves Understanding of Leukemia Progression. <i>Blood</i> , 2021, 138, 1293-1293.	0.6	0