Marco Montagna

List of Publications by Year
in descending order

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121 papers	9,349 citations	41258 49 h-index	43802 91 g-index
126 all docs	126 docs citations	126 times ranked	12368 citing authors

#	Article	IF	Citations
1	Association Between <emph type="ital">BRCA1</emph> and <emph type="ital">BRCA2</emph> Mutations and Survival in Women With Invasive Epithelial Ovarian Cancer. JAMA - Journal of the American Medical Association, 2012, 307, 382.	3.8	546
2	Pathology of Breast and Ovarian Cancers among BRCA1 and BRCA2 Mutation Carriers: Results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 134-147.	1.1	513
3	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	9.4	493
4	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	3.8	390
5	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
6	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	9.4	309
7	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
8	ENIGMA-Evidence-based network for the interpretation of germline mutant alleles: An international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. Human Mutation, 2012, 33, 2-7.	1.1	269
9	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
10	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
11	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, .	3.0	242
12	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	1.1	224
13	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	9.4	221
14	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Implications for Risk Prediction. Cancer Research, 2010, 70, 9742-9754.	0.4	169
15	Genomic rearrangements account for more than one-third of the BRCA1 mutations in northern Italian breast/ovarian cancer families. Human Molecular Genetics, 2003, 12, 1055-1061.	1.4	159
16	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.	0.8	152
17	Germline Mutation in BRCA1 or BRCA2 and Ten-Year Survival for Women Diagnosed with Epithelial Ovarian Cancer. Clinical Cancer Research, 2015, 21, 652-657.	3.2	138
18	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	9.4	125

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19	Association Between MDM2â€“SNP309 and Age at Colorectal Cancer Diagnosis According to p53 Mutation Status. Journal of the National Cancer Institute, 2006, 98, 285-288.	3.0	123
20	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
21	The human plakoglobin gene localizes on chromosome 17q21 and is subjected to loss of heterozygosity in breast and ovarian cancers.. Proceedings of the National Academy of Sciences of the United States of America, 1995, 92, 6384-6388.	3.3	111
22	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.	1.1	102
23	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. American Journal of Human Genetics, 2000, 67, 207-212.	2.6	100
24	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2009, 18, 4442-4456.	1.4	99
25	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	5.8	98
26	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	2.2	97
27	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. Clinical Chemistry, 2014, 60, 341-352.	1.5	95
28	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
29	Identification of a 3â€‰kb Alu-mediated BRCA1 gene rearrangement in two breast/ovarian cancer families. Oncogene, 1999, 18, 4160-4165.	2.6	91
30	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	1.4	91
31	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
32	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.	2.2	88
33	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	5.8	88
34	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	1.5	85
35	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.	1.1	82
36	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33.	2.2	78

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37	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	5.8	78
38	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	3.0	77
39	Clinical and pathologic characteristics of BRCA-positive and BRCA-negative male breast cancer patients: results from a collaborative multicenter study in Italy. Breast Cancer Research and Treatment, 2012, 134, 411-418.	1.1	73
40	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110.	2.2	71
41	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	1.4	68
42	Color bar coding theBRCA1 gene on combed DNA: A useful strategy for detecting large gene rearrangements. Genes Chromosomes and Cancer, 2001, 31, 75-84.	1.5	64
43	Altered tumor formation and evolutionary selection of genetic variants in the human MDM4 oncogene. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 10236-10241.	3.3	62
44	Role of Î²-Defensin-1 Polymorphisms in Mother-to-Child Transmission of HIV-1. Journal of Acquired Immune Deficiency Syndromes (1999), 2009, 51, 13-19.	0.9	59
45	Large genomic deletions inactivate the BRCA2 gene in breast cancer families. Journal of Medical Genetics, 2005, 42, e64-e64.	1.5	57
46	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	2.2	57
47	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. Breast Cancer Research and Treatment, 2012, 132, 1009-1023.	1.1	56
48	Evaluation of widely used models for predicting BRCA1 and BRCA2 mutations. Journal of Medical Genetics, 2004, 41, 278-285.	1.5	55
49	Penetrances of breast and ovarian cancer in a large series of families tested for BRCA1/2 mutations. European Journal of Human Genetics, 2004, 12, 899-906.	1.4	55
50	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. Cancer Research, 2018, 78, 5419-5430.	0.4	54
51	Prevalence ofBRCA1 genomic rearrangements in a large cohort of Italian breast and breast/ovarian cancer families without detectableBRCA1 andBRCA2 point mutations. Genes Chromosomes and Cancer, 2006, 45, 791-797.	1.5	50
52	Lack of association between androgen receptor CAG polymorphism and familial breast/ovarian cancer. Cancer Letters, 2001, 168, 31-36.	3.2	49
53	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	3.4	48
54	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. Clinical Cancer Research, 2011, 17, 3742-3750.	3.2	47

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55	Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657.	1.1	47
56	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	1.5	47
57	Epstein-Barr virus-associated post-transplant lympho-proliferative disease of donor origin in liver transplant recipients. Journal of Hepatology, 1997, 26, 926-934.	1.8	45
58	BRCA1 p.Val1688del Is a Deleterious Mutation That Recurs in Breast and Ovarian Cancer Families From Northeast Italy. Journal of Clinical Oncology, 2008, 26, 26-31.	0.8	44
59	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	2.2	42
60	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2011, 103, 105-116.	3.0	40
61	Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy. International Journal of Cancer, 2019, 145, 390-400.	2.3	40
62	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. Human Mutation, 2004, 24, 100-101.	1.1	39
63	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
64	Toll-like receptor 9 polymorphisms influence mother-to-child transmission of human immunodeficiency virus type 1. Journal of Translational Medicine, 2010, 8, 49.	1.8	36
65	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	1.1	34
66	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	1.1	34
67	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. Human Molecular Genetics, 2011, 20, 4732-4747.	1.4	32
68	Association of low-penetrance alleles with male breast cancer risk and clinicopathological characteristics: results from a multicenter study in Italy. Breast Cancer Research and Treatment, 2013, 138, 861-868.	1.1	32
69	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	0.6	32
70	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.	2.2	31
71	Wholeâ€exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. Cancer, 2017, 123, 210-218.	2.0	31
72	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. Journal of the National Cancer Institute, 2019, 111, 350-364.	3.0	30

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73	DNA copy number alterations correlate with survival of esophageal adenocarcinoma patients. Modern Pathology, 2009, 22, 58-65.	2.9	29
74	Occasional loss of constitutive heterozygosity at 11p15.5 and imprinting relaxation of theIGFII maternal allele in hepatoblastoma. Journal of Cancer Research and Clinical Oncology, 1994, 120, 732-736.	1.2	28
75	Establishment and characterization of xenografts and cancer cell cultures derived from BRCA1 â˜'/â˜' epithelial ovarian cancers. European Journal of Cancer, 2006, 42, 1475-1483.	1.3	28
76	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
77	Methyl group metabolism gene polymorphisms as modifier of breast cancer risk in Italian BRCA1/2 carriers. Breast Cancer Research and Treatment, 2007, 103, 29-36.	1.1	27
78	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.	2.2	26
79	Evaluation of copy-number variants as modifiers of breast and ovarian cancer risk for BRCA1 pathogenic variant carriers. European Journal of Human Genetics, 2017, 25, 432-438.	1.4	26
80	Novel and known genetic variants for male breast cancer risk at 8q24.21, 9p21.3, 11q13.3 and 14q24.1: Results from a multicenter study in Italy. European Journal of Cancer, 2015, 51, 2289-2295.	1.3	25
81	Contribution of MUTYH Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. Frontiers in Oncology, 2018, 8, 583.	1.3	25
82	A Nonsynonymous Polymorphism in IRS1 Modifies Risk of Developing Breast and Ovarian Cancers in BRCA1 and Ovarian Cancer in BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1362-1370.	1.1	23
83	A possible role of FANCM mutations in male breast cancer susceptibility: Results from a multicenter study in Italy. Breast, 2018, 38, 92-97.	0.9	23
84	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	1.4	23
85	Different Expressivity of BRCA1 and BRCA2: Analysis of 179 Italian Pedigrees with Identified Mutation. Breast Cancer Research and Treatment, 2003, 81, 71-79.	1.1	22
86	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	1.1	22
87	Performance of BOADICEA and BRCAPRO genetic models and of empirical criteria based on cancer family history for predicting BRCA mutation carrier probabilities: A retrospective study in a sample of Italian cancer genetics clinics. Breast, 2013, 22, 1130-1135.	0.9	21
88	Anomalous Transcripts and Allelic Deletions of the FHIT Gene in Human Esophageal Cancer. Cancer Genetics and Cytogenetics, 2000, 119, 56-61.	1.0	19
89	The BRCA2 c.68-7TÂ>ÂA variant is not pathogenic: A model for clinical calibration of spliceogenicity. Human Mutation, 2018, 39, 729-741.	1.1	19
90	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.	2.9	19

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91	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	5.8	19
92	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19
93	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. Human Genetics, 2011, 130, 685-699.	1.8	18
94	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	0.6	18
95	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.	1.1	18
96	CDKN2AUnclassified Variants in Familial Malignant Melanoma: Combining Functional and Computational Approaches for Their Assessment. Human Mutation, 2014, 35, 828-840.	1.1	17
97	The predictive ability of the 313 variantâ€“based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. Genetics in Medicine, 2021, 23, 1726-1737.	1.1	16
98	Evaluation of a candidate breast cancer associated SNP in ERCC4 as a risk modifier in BRCA1 and BRCA2 mutation carriers. Results from the Consortium of Investigators of Modifiers of BRCA1/BRCA2 (CIMBA). British Journal of Cancer, 2009, 101, 2048-2054.	2.9	15
99	DNA copy number profile discriminates between esophageal adenocarcinoma and squamous cell carcinoma and represents an independent prognostic parameter in esophageal adenocarcinoma. Cancer Letters, 2011, 310, 84-93.	3.2	15
100	Functional impairment of p16INK4A due to CDKN2A p.Gly23Asp missense mutation. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2009, 671, 26-32.	0.4	14
101	Co-existence of cutaneous T-cell lymphoma and B hairy cell leukemia. American Journal of Hematology, 2000, 64, 197-202.	2.0	13
102	The Role of Genetic Variants of Stromal Cell-Derived Factor 1 in Pediatric HIV-1 Infection and Disease Progression. PLoS ONE, 2012, 7, e44460.	1.1	13
103	Contribution of susceptibility gene variants to melanoma risk in families from the Veneto region of Italy. Pigment Cell and Melanoma Research, 2011, 24, 728-730.	1.5	12
104	Differential sensitivity of BRCA1-mutated HCC1937 human breast cancer cells to microtubule-interfering agents. International Journal of Oncology, 2005, 26, 1257.	1.4	11
105	Family History of Cancer Rather Than p53 Status Predicts Efficacy of Pegylated Liposomal Doxorubicin and Oxaliplatin in Relapsed Ovarian Cancer. International Journal of Gynecological Cancer, 2009, 19, 1022-1028.	1.2	10
106	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.	1.1	10
107	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants. Genetics in Medicine, 2022, 24, 119-129.	1.1	10
108	Dominance of a single Epstein-Barr virus strain in SCID-mouse tumors induced by injection of peripheral blood mononuclear cells from healthy human donors. Virus Research, 1995, 36, 215-231.	1.1	9

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109	Identification of BRCA1 and BRCA2 carriers by allele-specific gene expression (AGE) analysis. International Journal of Cancer, 2002, 98, 732-736.	2.3	9
110	Analysis of Epsteinâ€“Barr virus (EBV) type and variant in spontaneous lymphoblastoid cells and Hu-SCID mouse tumours. Molecular and Cellular Probes, 1996, 10, 453-461.	0.9	7
111	Association of SULT1A1 Arg213His polymorphism with male breast cancer risk: results from a multicenter study in Italy. Breast Cancer Research and Treatment, 2014, 148, 623-628.	1.1	7
112	Synchronous and Metachronous Breast and Ovarian Cancer: Experience From Two Large Cancer Center. Frontiers in Oncology, 2020, 10, 608783.	1.3	7
113	Localization of the human phosphotyrosine phosphatase-related genes (h-PRL-1) to chromosome bands 1p35-p34, 17q12-q21, 11q24-q25 and 12q24. Human Genetics, 1996, 98, 738-740.	1.8	6
114	Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk. Endocrine Connections, 2019, 8, 1224-1229.	0.8	6
115	The BRCA2 sequence variant IVS19+1Gâ†'A leads to an aberrant transcript lacking exon 19. Cancer Genetics and Cytogenetics, 2003, 141, 175-176.	1.0	5
116	Segregation analysis of the BRCA2 c.9227G>T variant in multiple families suggests a pathogenic role in breast and ovarian cancer predisposition. Scientific Reports, 2020, 10, 13987.	1.6	5
117	Evaluation of chromosome 6p22 as a breast cancer risk modifier locus in a follow-up study of BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2012, 136, 295-302.	1.1	4
118	Identification of a human endogenous LTR-like sequence using HIV-1 LTR specific primers. Molecular and Cellular Probes, 1996, 10, 443-451.	0.9	3
119	TP53 gene mutations in gastric carcinoma detected by polymerase chain reaction/single-strand conformation polymorphism analysis of archival material. Journal of Cancer Research and Clinical Oncology, 1995, 121, 79-83.	1.2	2
120	Abstract 1236: Insight into genetic susceptibility toBRCA-negative male breast cancer by multigene panel testing: Results from a multicenter study in Italy. , 2018, , .		1
121	Myasthenic syndrome and oligoclonal lymphocytosis: evolution into chronic lymphocytic leukemia. Annals of Hematology, 1998, 76, 45-47.	0.8	0