Dietmar R Lohmann

List of Publications by Year
in descending order

91 papers	8,104 citations	81900 39 h-index	51608 86 g-index
101 all docs	101 docs citations	101 times ranked	11351 citing authors

#	Article	IF	Citations
1	DNA methylation-based classification of central nervous system tumours. Nature, 2018, 555, 469-474.	27.8	1,872
2	The landscape of genomic alterations across childhood cancers. Nature, 2018, 555, 321-327.	27.8	1,068
3	Exome sequencing identifies recurrent somatic mutations in EIF1AX and SF3B1 in uveal melanoma with disomy 3. Nature Genetics, 2013, 45, 933-936.	21.4	436
4	Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. Nature Genetics, 2011, 43, 20-22.	21.4	308
5	Characterisation of retinoblastomas without RB1 mutations: genomic, gene expression, and clinical studies. Lancet Oncology, The, 2013, 14, 327-334.	10.7	304
6	Tumor classification based on gene expression profiling shows that uveal melanomas with and without monosomy 3 represent two distinct entities. Cancer Research, 2003, 63, 2578-84.	0.9	233
7	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
8	RB1 gene mutations in retinoblastoma. Human Mutation, 1999, 14, 283-288.	2.5	195
9	Haploinsufficiency of a Spliceosomal GTPase Encoded by EFTUD2 Causes Mandibulofacial Dysostosis with Microcephaly. American Journal of Human Genetics, 2012, 90, 369-377.	6.2	180
10	Retinoblastoma: Revisiting the model prototype of inherited cancer. American Journal of Medical Genetics Part A, 2004, 129C, 23-28.	2.4	166
11	Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. European Journal of Human Genetics, 2004, 12, 879-890.	2.8	149
12	Constitutional RB1-Gene Mutations in Patients with Isolated Unilateral Retinoblastoma. American Journal of Human Genetics, 1997, 61, 282-294.	6.2	148
13	Genotype-phenotype correlation in a series of 167 deletion and non-deletion patients with Prader-Willi syndrome. Human Genetics, 1995, 96, 638-643.	3.8	147
14	Frequency and parental origin of hypermethylated RB1 alleles in retinoblastoma. Human Genetics, 1994, 94, 491-6.	3.8	143
15	Conservative management of retinoblastoma: Challenging orthodoxy without compromising the state of metastatic grace. â€œAlive, with good vision and no comorbidityâ€• Progress in Retinal and Eye Research, 2019, 73, 100764.	15.5	123
16	A novel real-time PCR assay for quantitative analysis of methylated alleles (QAMA): analysis of the retinoblastoma locus. Nucleic Acids Research, 2004, 32, e125-e125.	14.5	120
17	The Human Retinoblastoma Gene Is Imprinted. PLoS Genetics, 2009, 5, e1000790.	3.5	110
18	Gains and overexpression identify DEK and E2F3 as targets of chromosome 6p gains in retinoblastoma. Oncogene, 2005, 24, 6441-6449.	5.9	108

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19	Detection of chromosomal imbalances in retinoblastoma by matrixâ€based comparative genomic hybridization. Genes Chromosomes and Cancer, 2005, 43, 294-301.	2.8	101
20	Identification of chromosomes 3, 6, and 8 aberrations in uveal melanoma by microsatellite analysis in comparison to comparative genomic hybridization. Cancer Genetics and Cytogenetics, 2000, 122, 13-17.	1.0	99
21	Incidence of second cancers after radiotherapy and systemic chemotherapy in heritable retinoblastoma survivors: A report from the German reference center. Pediatric Blood and Cancer, 2017, 64, 71-80.	1.5	90
22	Genotypeâ€“phenotype correlations in patients with retinoblastoma and interstitial 13q deletions. European Journal of Human Genetics, 2011, 19, 947-958.	2.8	83
23	Comparing the Prognostic Value of BAP1 Mutation Pattern, Chromosome 3 Status, and BAP1 Immunohistochemistry in Uveal Melanoma. American Journal of Surgical Pathology, 2016, 40, 796-805.	3.7	79
24	Marked differences in unilateral isolated retinoblastomas from young and older children studied by comparative genomic hybridization. Human Genetics, 2001, 108, 98-104.	3.8	74
25	Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction. American Journal of Human Genetics, 2015, 96, 765-774.	6.2	67
26	Patterns of missplicing caused byRB1gene mutations in patients with retinoblastoma and association with phenotypic expression. Human Mutation, 2008, 29, 475-484.	2.5	66
27	Loss of heterozygosity of 1p in uveal melanomas with monosomy 3. International Journal of Cancer, 2005, 116, 909-913.	5.1	64
28	A Parent-of-Origin Effect in Two Families with Retinoblastoma Is Associated with a Distinct Splice Mutation in the RB1 Gene. American Journal of Human Genetics, 2002, 71, 174-179.	6.2	63
29	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	2.4	63
30	Retinoblastoma. Advances in Experimental Medicine and Biology, 2010, 685, 220-227.	1.6	61
31	Identification of p53 gene mutations in gastrointestinal and pancreatic carcinoids by nonradioisotopic SSCA. Vigiliae Christianae, 1993, 64, 293-296.	0.1	58
32	Detection of small RB1 gene deletions in retinoblastoma by multiplex PCR and high-resolution gel electrophoresis. Human Genetics, 1992, 89, 49-53.	3.8	56
33	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	6.2	55
34	Molecular subgrouping of primary pineal parenchymal tumors reveals distinct subtypes correlated with clinical parameters and genetic alterations. Acta Neuropathologica, 2020, 139, 243-257.	7.7	50
35	Multiple lipomas linked to an RB1 gene mutation in a large pedigree with low penetrance retinoblastoma. European Journal of Human Genetics, 2001, 9, 690-694.	2.8	49
36	Human TRHâ€degrading ectoenzyme. FEBS Journal, 1999, 265, 415-422.	0.2	48

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37	A variant allele of Growth Factor Independence 1 (GFI1) is associated with acute myeloid leukemia. Blood, 2010, 115, 2462-2472.	1.4	46
38	Genomic gains on chromosome 1q in retinoblastoma: Consequences on gene expression and association with clinical manifestation. International Journal of Cancer, 2005, 116, 555-563.	5.1	44
39	Ultradeep sequencing detects GNAQ and GNA11 mutations in cellâ€free DNA from plasma of patients with uveal melanoma. Cancer Medicine, 2013, 2, 208-215.	2.8	44
40	RB1 Gene Mutations in Peripheral Blood DNA of Patients with Isolated Unilateral Retinoblastoma. American Journal of Human Genetics, 1999, 64, 667-668.	6.2	40
41	Microcephaly, microtia, preauricular tags, choanal atresia and developmental delay in three unrelated patients: A mandibulofacial dysostosis distinct from Treacher Collins syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 837-843.	1.2	39
42	How Eye-Preserving Therapy Affects Long-Term Overall Survival in Heritable Retinoblastoma Survivors. Journal of Clinical Oncology, 2016, 34, 3183-3188.	1.6	39
43	Imprinting of RB1 (the new kid on the block). Briefings in Functional Genomics, 2010, 9, 347-353.	2.7	36
44	EFS shows biallelic methylation in uveal melanoma with poor prognosis as well as tissue-specific methylation. BMC Cancer, 2011, 11, 380.	2.6	32
45	Allelic Loss in a Minimal Region on Chromosome 16q24 Is Associated with Vitreous Seeding of Retinoblastoma. Cancer Research, 2007, 67, 408-416.	0.9	29
46	Multiple subcutaneous granular-cell tumours in a patient with Noonan syndrome. Clinical Dysmorphology, 2000, 9, 301-302.	0.3	28
47	Pediatric second primary malignancies after retinoblastoma treatment. Pediatric Blood and Cancer, 2015, 62, 1799-1804.	1.5	27
48	BAP1germline mutation in two first grade family members with uveal melanoma. British Journal of Ophthalmology, 2014, 98, 224-227.	3.9	25
49	Treacher Collins syndrome: clinical implications for the paediatricianâ€"a new mutation in a severely affected newborn and comparison with three further patients with the same mutation, and review of the literature. European Journal of Pediatrics, 2012, 171, 1611-1618.	2.7	23
50	Chemoreduction improves eye retention in patients with retinoblastoma: a report from the German Retinoblastoma Reference Centre. British Journal of Ophthalmology, 2013, 97, 1277-1283.	3.9	23
51	Loss of Heterozygosity of the Retinoblastoma (RB1) Gene in Lipomas From a Retinoblastoma Patient. Journal of the National Cancer Institute, 1998, 90, 324-326.	6.3	22
52	Establishment and characterization of two uveal melanoma cell lines derived from tumors with loss of one chromosome 3. Experimental Eye Research, 2006, 83, 858-864.	2.6	22
53	The Origin of the RB1 Imprint. PLoS ONE, 2013, 8, e81502.	2.5	21
54	Methylation Analysis of Several Tumour Suppressor Genes Shows a Low Frequency of Methylation ofCDKN2AandRARBin Uveal Melanomas. Comparative and Functional Genomics, 2003, 4, 329-336.	2.0	19

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55	Neurotrophin receptor expression in human primary retinoblastomas and retinoblastoma cell lines. Pediatric Blood and Cancer, 2008, 50, 218-222.	1.5	17
56	Neoadjuvant/adjuvant treatment of high-risk retinoblastoma: a report from the German Retinoblastoma Referral Centre. British Journal of Ophthalmology, 2015, 99, 949-953.	3.9	17
57	The impact of RB1 genotype on incidence of second tumours in heritable retinoblastoma. European Journal of Cancer, 2020, 133, 47-55.	2.8	16
58	Sporadic unilateral retinoblastoma or first sign of bilateral disease?. British Journal of Ophthalmology, 2013, 97, 475-480.	3.9	15
59	GNAQ Q209R Mutations Are Highly Specific for Circumscribed Choroidal Hemangioma. Cancers, 2019, 11, 1031.	3.7	15
60	Two brothers with Burn-McKeown syndrome. Clinical Dysmorphology, 2003, 12, 171-174.	0.3	14
61	The Interdisciplinary Diagnosis and Treatment of Intraocular Tumors. Deutsches Ärzteblatt International, 2018, 115, 106-111.	0.9	13
62	Detection of HER-2 oncogene amplification in breast cancer by differential polymerase chain reaction from single cryosections. Vigiliae Christianae, 1993, 64, 209-212.	0.1	12
63	Clinical utility gene card for: Retinoblastoma. European Journal of Human Genetics, 2011, 19, 3-3.	2.8	12
64	Loss at chromosome arm 16q in retinoblastoma: Confirmation of the association with diffuse vitreous seeding and refinement of the recurrently deleted region. Genes Chromosomes and Cancer, 2011, 50, 327-337.	2.8	12
65	Early detection of metastatic uveal melanoma by the analysis of tumorâ€specific mutations in cellâ€free plasma DNA. Cancer Medicine, 2021, 10, 5974-5982.	2.8	11
66	Molecular analysis and predictive testing in retinoblastoma. Ophthalmic Genetics, 1995, 16, 135-142.	1.2	10
67	Retinoblastoma tumorigenesis: genetic and epigenetic changes walk hand in hand. Future Oncology, 2012, 8, 525-528.	2.4	10
68	Genetic testing in Tunisian families with heritable retinoblastoma using a low cost approach permits accurate risk prediction in relatives and reveals incomplete penetrance in adults. Experimental Eye Research, 2014, 124, 48-55.	2.6	10
69	Psychosocial impact of prognostic genetic testing in uveal melanoma patients: a controlled prospective clinical observational study. BMC Psychology, 2020, 8, 8.	2.1	10
70	TFF1 in Aqueous Humorâ€”A Potential New Biomarker for Retinoblastoma. Cancers, 2022, 14, 677.	3.7	10
71	Identification of a mutation in exon 27 of the RB1 gene associated with incomplete penetrance retinoblastoma. Familial Cancer, 2009, 8, 55-58.	1.9	9
72	Hereditary Diffuse Infiltrating Retinoblastoma. Ophthalmic Genetics, 2014, 37, 1-3.	1.2	9

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73	Differentiation Protocol for 3D Retinal Organoids, Immunostaining and Signal Quantitation. Current Protocols in Stem Cell Biology, 2020, 55, e120.	3.0	8
74	Eye Tumors in Childhood as First Sign of Tumor Predisposition Syndromes: Insights from an Observational Study Conducted in Germany and Austria. Cancers, 2021, 13, 1876.	3.7	7
75	RB1-Negative Retinal Organoids Display Proliferation of Cone Photoreceptors and Loss of Retinal Differentiation. Cancers, 2022, 14, 2166.	3.7	6
76	Trilateral Retinoblastoma in a Patient With Peutzâ€“<scp>J</scp>eghers Syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1096-1100.	1.2	4
77	Psychosocial impact of prognostic genetic testing in the care of uveal melanoma patients: protocol of a controlled prospective clinical observational study. BMC Cancer, 2016, 16, 408.	2.6	4
78	Chromosome 3 is a valid marker for prognostic testing of biopsy material from uveal melanoma later treated by brachytherapy. Biomarkers, 2019, 24, 134-140.	1.9	4
79	13q deletion syndrome resulting from balanced chromosomal rearrangement in father: the significance of parental karyotyping. Molecular Cytogenetics, 2020, 13, 31.	0.9	4
80	Introduction of a Variant Classification System for Analysis of Genotype-Phenotype Relationships in Heritable Retinoblastoma. Cancers, 2021, 13, 1605.	3.7	4
81	Human PPP1R26P1 Functions as cis-Repressive Element in Mouse Rb1. PLoS ONE, 2013, 8, e74159.	2.5	4
82	Ectopic intracranial retinoblastoma in a 3.5â€monthâ€old infant without eye involvement and without evidence of heritability. Pediatric Blood and Cancer, 2019, 66, e27599.	1.5	3
83	Comparing efficacy and side effects of two systemic chemotherapy regimens for eyeâ€preserving therapy in children with retinoblastoma. Pediatric Blood and Cancer, 2022, 69, e29362.	1.5	3
84	Two brothers with Burn???McKeown syndrome. Clinical Dysmorphology, 2003, 12, 171-174.	0.3	1
85	Retinoblastoma with late metastatic spreadâ€”a case report. Pediatric Blood and Cancer, 2019, 66, e27656.	1.5	1
86	RB1 gene mutations in retinoblastoma. , 1999, 14, 283.		1
87	Prognostic Testing in Uveal Melanoma. , 2013, , 79-96.		1
88	Minimal Region of Deletion on Chromosomal Arm 3p25.1-p25.2 in Uveal Melanoma. Cancer Research, 2005, 65, 10634-10634.	0.9	0
89	Retinoblastom. , 2001, , 23-41.		0
90	Genetische Einflussfaktoren des Krebsrisikos. Springer Reference Medizin, 2020, , 1-7.	0.0	0

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91	OTHR-34. Identifying mechanisms of microglia-tumor cell interactions in retinoblastoma. Neuro-Oncology, 2022, 24, i154-i154.	1.2	0