Johan Gille

List of Publications by Year in descending order

87 papers	6,954 citations	81900 39 h-index	58581 82 g-index
88 all docs	88 docs citations	88 times ranked	8604 citing authors

#	Article	IF	Citations
1	Frequency and Spectrum of Cancers in the Peutz-Jeghers Syndrome. Clinical Cancer Research, 2006, 12, 3209-3215.	7.0	746
2	Dysplastic changes in prophylactically removed Fallopian tubes of women predisposed to developing ovarian cancer. Journal of Pathology, 2001, 195, 451-456.	4.5	681
3	The Cowden syndrome: a clinical and genetic study in 21 patients. Clinical Genetics, 1986, 29, 222-233.	2.0	478
4	Expression cloning of a cDNA for the major Fanconi anaemia gene, FAA. Nature Genetics, 1996, 14, 320-323.	21.4	401
5	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
6	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
7	Large genomic deletions and duplications in the BRCA1 gene identified by a novel quantitative method. Cancer Research, 2003, 63, 1449-53.	0.9	233
8	Risk of colorectal and endometrial cancers in EPCAM deletion-positive Lynch syndrome: a cohort study. Lancet Oncology, The, 2011, 12, 49-55.	10.7	232
9	Relative frequency and morphology of cancers in STK11 mutation carriers1 â˜†. Gastroenterology, 2004, 126, 1788-1794.	1.3	228
10	Molecular Evidence Linking Primary Cancer of the Fallopian Tube to BRCA1 Germline Mutations. Gynecologic Oncology, 2000, 76, 45-50.	1.4	186
11	TBX4mutations (small patella syndrome) are associated with childhood-onset pulmonary arterial hypertension. Journal of Medical Genetics, 2013, 50, 500-506.	3.2	171
12	Cell culture models for oxidative stress: superoxide and hydrogen peroxide versus normobaric hyperoxia. Mutation Research - DNAging, 1992, 275, 405-414.	3.2	166
13	Genomic deletions of MSH2 and MLH1 in colorectal cancer families detected by a novel mutation detection approach. British Journal of Cancer, 2002, 87, 892-897.	6.4	155
14	Renal cancer and pneumothorax risk in Birtâ€“Hoggâ€“DubÃ© syndrome; an analysis of 115 FLCN mutation carriers from 35 BHD families. British Journal of Cancer, 2011, 105, 1912-1919.	6.4	142
15	Recurrence and variability of germline EPCAM deletions in Lynch syndrome. Human Mutation, 2011, 32, 407-414.	2.5	137
16	Origin, functional role, and clinical impact of Fanconi anemia FANCA mutations. Blood, 2011, 117, 3759-3769.	1.4	108
17	A new locus-specific database (LSDB) for mutations in the folliculin (FLCN) gene. Human Mutation, 2010, 31, E1043-E1051.	2.5	93
18	Birt-Hogg-DubÃ© Syndrome: Clinical and Genetic Studies of 20 Families. Journal of Investigative Dermatology, 2008, 128, 45-49.	0.7	88

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19	Non-invasive prenatal detection of achondroplasia in size-fractionated cell-free DNA by MALDI-TOF MS assay. Prenatal Diagnosis, 2007, 27, 11-17.	2.3	87
20	Improved prenatal detection of a fetal point mutation for achondroplasia by the use of size-fractionated circulatory DNA in maternal plasma?case report. Prenatal Diagnosis, 2004, 24, 896-898.	2.3	86
21	Heterogeneous spectrum of mutations in the Fanconi anaemia group A gene. European Journal of Human Genetics, 1999, 7, 52-59.	2.8	84
22	Investigation of the Birt-Hogg-Dube tumour suppressor gene (FLCN) in familial and sporadic colorectal cancer. Journal of Medical Genetics, 2010, 47, 385-390.	3.2	77
23	Mutagenicity of metabolic oxygen radicals in mammalian cell cultures. Carcinogenesis, 1994, 15, 2695-2699.	2.8	76
24	A DGGE system for comprehensive mutation screening ofBRCA1andBRCA2: application in a Dutch cancer clinic setting. Human Mutation, 2006, 27, 654-666.	2.5	75
25	A Homozygous MSH6 Mutation in a Child with CafÃ©-au-Lait Spots, Oligodendroglioma and Rectal Cancer. Familial Cancer, 2002, 3, 123-127.	1.9	74
26	Contribution of CYLN2 and GTF2IRD1 to neurological and cognitive symptoms in Williams Syndrome. Neurobiology of Disease, 2007, 26, 112-124.	4.4	67
27	CHEK2*1100delC homozygosity is associated with a high breast cancer risk in women. Journal of Medical Genetics, 2011, 48, 860-863.	3.2	66
28	Copy number variation analysis identifies novel CAKUT candidate genes in children with a solitary functioning kidney. Kidney International, 2015, 88, 1402-1410.	5.2	65
29	Histopathological characteristics of BRCA1- and BRCA2-associated intraperitoneal cancer: a clinic-based study. Familial Cancer, 2003, 2, 73-78.	1.9	60
30	Novel strategies in newborn screening for cystic fibrosis: a prospective controlled study. Thorax, 2012, 67, 289-295.	5.6	53
31	Variable phenotypes associated with 10q23 microdeletions involving the PTEN and BMPR1A genes. Clinical Genetics, 2008, 74, 145-154.	2.0	52
32	Comprehensive Mutation Analysis ofPMS2in a Large Cohort of Probands Suspected of Lynch Syndrome or Constitutional Mismatch Repair Deficiency Syndrome. Human Mutation, 2016, 37, 1162-1179.	2.5	50
33	An Autosomal Dominant High Bone Mass Phenotype in Association With Craniosynostosis in an Extended Family Is Caused by an LRP5 Missense Mutation. Journal of Bone and Mineral Research, 2005, 20, 1254-1260.	2.8	46
34	A method to assess the clinical significance of unclassified variants in the BRCA1 and BRCA2genes based on cancer family history. Breast Cancer Research, 2009, 11, R8.	5.0	45
35	PALB2 analysis in BRCA2-like families. Breast Cancer Research and Treatment, 2011, 127, 357-362.	2.5	45
36	STK11 status and intussusception risk in Peutz-Jeghers syndrome. Journal of Medical Genetics, 2006, 43, e41-e41.	3.2	44

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37	Diagnosis of Fanconi Anemia: Mutation Analysis by Next-Generation Sequencing. Anemia, 2012, 2012, 1-7.	1.7	44
38	Antioxidant status of Fanconi anemia fibroblasts. Human Genetics, 1987, 77, 28-31.	3.8	43
39	Clinical and genetic evaluation of thirty ovarian cancer families. American Journal of Obstetrics and Gynecology, 1998, 178, 85-90.	1.3	43
40	Diagnosis of Fanconi Anemia: Mutation Analysis by Multiplex Ligation-Dependent Probe Amplification and PCR-Based Sanger Sequencing. Anemia, 2012, 2012, 1-13.	1.7	39
41	Expression of differentiation and proliferation related proteins in epithelium of prophylactically removed ovaries from women with a hereditary female adnexal cancer predisposition. Histopathology, 2003, 43, 26-32.	2.9	38
42	Low prevalence of (pre) malignant lesions in the breast and high prevalence in the ovary and Fallopian tube in women at hereditary high risk of breast and ovarian cancer. International Journal of Cancer, 2006, 119, 1412-1418.	5.1	33
43	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.	2.9	32
44	Hereditary causes of kidney tumours. European Journal of Clinical Investigation, 2010, 40, 433-439.	3.4	30
45	Serum CA-125 in Relation to Adnexal Dysplasia and Cancer in Women at Hereditary High Risk of Ovarian Cancer. Journal of Clinical Oncology, 2007, 25, 1383-1389.	1.6	29
46	Early onset of renal cancer in a family with Birtâ€“Hoggâ€“DubÃ© syndrome. Clinical Genetics, 2009, 75, 537-543.	2.0	28
47	Cost-effectiveness of newborn screening for cystic fibrosis determined with real-life data. Journal of Cystic Fibrosis, 2015, 14, 194-202.	0.7	28
48	Newborn blood spot screening for cystic fibrosis with a four-step screening strategy in the Netherlands. Journal of Cystic Fibrosis, 2019, 18, 54-63.	0.7	28
49	Inactivating Mutations in GT198 in Familial and Early-Onset Breast and Ovarian Cancers. Genes and Cancer, 2013, 4, 15-25.	1.9	25
50	New mutations and an updated database for the patchedâ€1 (<scp>PTCH</scp>1) gene. Molecular Genetics & Genomic Medicine, 2018, 6, 409-415.	1.2	25
51	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Results from The BRCA1 and BRCA2 Cohort Consortium. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 368-378.	2.5	24
52	Survival analysis in familial ovarian cancer, a case control study. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2001, 98, 219-223.	1.1	21
53	A novel pathogenic MLH1 missense mutation, c.112A > C, p.Asn38His, in six families with Lynch syndrome. Hereditary Cancer in Clinical Practice, 2010, 8, 7.	1.5	21
54	Heterozygote FANCD2 mutations associated with childhood T Cell ALL and testicular seminoma. Familial Cancer, 2012, 11, 661-665.	1.9	21

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55	Analysis of the Novel Fanconi Anemia GeneSLX4/FANCPin Familial Breast Cancer Cases. Human Mutation, 2013, 34, 70-73.	2.5	21
56	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2009, 101, 1456-1460.	6.4	19
57	Familial multiple discoid fibromas: AÂlook-alike of Birt-Hogg-DubÃ© syndrome not linked to the FLCN locus. Journal of the American Academy of Dermatology, 2012, 66, 259.e1-259.e9.	1.2	19
58	A de novo FLCN mutation in a patient with spontaneous pneumothorax and renal cancer; a clinical and molecular evaluation. Familial Cancer, 2013, 12, 373-379.	1.9	19
59	Pediatric Diamondâ€Blackfan anemia in the Netherlands: An overview of clinical characteristics and underlying molecular defects. European Journal of Haematology, 2018, 100, 163-170.	2.2	19
60	CFTR Mutations in Turkish and North African Cystic Fibrosis Patients in Europe: Implications for Screening. Genetic Testing and Molecular Biomarkers, 2008, 12, 25-35.	1.7	18
61	TP53 germline mutation testing in early-onset breast cancer: findings from a nationwide cohort. Familial Cancer, 2019, 18, 273-280.	1.9	18
62	Effect of iron chelators on the cytotoxic and genotoxic action of hyperoxia in Chinese hamster ovary cells. Mutation Research - DNAging, 1992, 275, 31-39.	3.2	17
63	Renal imaging in 199 Dutch patients with Birt-Hogg-DubÃ© syndrome: Screening compliance and outcome. PLoS ONE, 2019, 14, e0212952.	2.5	17
64	Mechanism of hyperoxia-induced chromosomal breakage in chinese hamster cells. Environmental and Molecular Mutagenesis, 1993, 22, 264-270.	2.2	16
65	The frequent BRCA1 mutation 1135insA has multiple origins: a haplotype study in different populations. BMC Medical Genetics, 2006, 7, 15.	2.1	15
66	Effect of normobaric hyperoxia on antioxidant defenses of hela and CHO cells. Free Radical Biology and Medicine, 1988, 4, 85-91.	2.9	14
67	Comparing Two Diagnostic Laboratory Tests for Williams Syndrome: Fluorescent In Situ Hybridization versus Multiplex Ligation-Dependent Probe Amplification. Genetic Testing and Molecular Biomarkers, 2007, 11, 321-327.	1.7	14
68	No evidence that GATA3 rs570613 SNP modifies breast cancer risk. Breast Cancer Research and Treatment, 2009, 117, 371-379.	2.5	12
69	Genetic Evaluation in a Cohort of 126 Dutch Pulmonary Arterial Hypertension Patients. Genes, 2020, 11, 1191.	2.4	12
70	Breast Cancer Risk and 6q22.33: Combined Results from Breast Cancer Association Consortium and Consortium of Investigators on Modifiers of BRCA1/2. PLoS ONE, 2012, 7, e35706.	2.5	11
71	Hereditary breast cancer syndromes in a Turkish population. Results of molecular germline analysis. Cancer Genetics and Cytogenetics, 2005, 160, 164-168.	1.0	10
72	Germline mutation in the STK11 gene in a girl with an ovarian Sertoli cell tumour. European Journal of Pediatrics, 2007, 166, 1083-1085.	2.7	10

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73	Influence of the antioxidant N-acetylcysteine and its metabolites on damage induced by bleomycin in PM2 bacteriophage DNA. Carcinogenesis, 1996, 17, 327-331.	2.8	9
74	Biological consequences of DNA damage introduced in bacteriophage PM2 DNA by hydrogen peroxide-mediated free radial reactions. Carcinogenesis, 1996, 17, 5-11.	2.8	8
75	Variable phenotypic manifestation of IRF6 mutations in the Van der Woude syndrome and popliteal pterygium syndrome: implications for genetic counseling. Clinical Dysmorphology, 2009, 18, 225-227.	0.3	8
76	A strategy for molecular diagnostics of Fanconi anemia in Brazilian patients. Molecular Genetics & Genomic Medicine, 2017, 5, 360-372.	1.2	8
77	Genetic counselling for pulmonary arterial hypertension: a matter of variable variability. Netherlands Heart Journal, 2011, 19, 89-92.	0.8	7
78	Improving test properties for neonatal cystic fibrosis screening in the Netherlands before the nationwide start by May 1st 2011. Journal of Inherited Metabolic Disease, 2012, 35, 635-640.	3.6	7
79	SNP association study in PMS2-associated Lynch syndrome. Familial Cancer, 2018, 17, 507-515.	1.9	7
80	Lack of genotype-phenotype correlation in basal cell nevus syndrome: A Dutch multicenter retrospective cohort study. Journal of the American Academy of Dermatology, 2020, 83, 604-607.	1.2	7
81	Possible hints and pitfalls in diagnosing Peutz-Jeghers syndrome. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1381-1386.	0.9	5
82	Direct-to-consumer carrier screening for cystic fibrosis via a hospital website: a 6-year evaluation. Journal of Community Genetics, 2019, 10, 249-257.	1.2	5
83	Are lung cysts in renal cell cancer (RCC) patients an indication for FLCN mutation analysis?. Familial Cancer, 2016, 15, 297-300.	1.9	3
84	Prevalence of medulloblastoma in basal cell nevus syndrome patients with a PTCH1 mutation. Neuro-Oncology, 2021, 23, 1035-1036.	1.2	3
85	Feasibility and outcomes of multiplex ligation-dependent probe amplification on buccal smears as a screening method for microdeletions and duplications among 300 adults with an intellectual disability of unknown aetiology. Journal of Intellectual Disability Research, 2007, 52, 070808045409004-???.	2.0	2
86	Multifocal and microscopic chromophobe renal cell carcinomatous lesions associated with â€˜capsulomasâ€™ without <scp>FCLN</scp> gene abnormality. Pathology International, 2013, 63, 510-515.	1.3	0
87	PULMONARY BIRT-HOGG-DUBE SYNDROME: A GOOD REASON TO ROUTINELY PERFORM A LOW DOSE CHEST CT SCAN IN PATIENTS WITH PRIMARY SPONTANEOUS PNEUMOTHORAX?. Chest, 2018, 154, 576A.	0.8	0