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	Prevalence of medulloblastoma in basal cell nevus syndrome patients with a PTCH1 mutation. Neuro-Oncology, 2021, 23, 1035-1036.	1.2	3
2	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
3	Genetic Evaluation in a Cohort of 126 Dutch Pulmonary Arterial Hypertension Patients. Genes, 2020, 11, 1191.	2.4	12
4	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
5	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
6	Renal imaging in 199 Dutch patients with Birt-Hogg-DubÃ© syndrome: Screening compliance and outcome. PLoS ONE, 2019, 14, e0212952.	2.5	17
7	TP53 germline mutation testing in early-onset breast cancer: findings from a nationwide cohort. Familial Cancer, 2019, 18, 273-280.	1.9	18
8	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
9	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
10	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
11	SNP association study in PMS2-associated Lynch syndrome. Familial Cancer, 2018, 17, 507-515.	1.9	7
12	Possible hints and pitfalls in diagnosing Peutz-Jeghers syndrome. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1381-1386.	0.9	5
13	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
14	A strategy for molecular diagnostics of Fanconi anemia in Brazilian patients. Molecular Genetics & Genomic Medicine, 2017, 5, 360-372.	1.2	8
15	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
16	Are lung cysts in renal cell cancer (RCC) patients an indication for FLCN mutation analysis?. Familial Cancer, 2016, 15, 297-300.	1.9	3
17	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
18	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

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19	Cost-effectiveness of newborn screening for cystic fibrosis determined with real-life data. Journal of Cystic Fibrosis, 2015, 14, 194-202.	0.7	28
20	Analysis of the Novel Fanconi Anemia GeneSLX4/FANCPin Familial Breast Cancer Cases. Human Mutation, 2013, 34, 70-73.	2.5	21
21	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
22	TBX4mutations (small patella syndrome) are associated with childhood-onset pulmonary arterial hypertension. Journal of Medical Genetics, 2013, 50, 500-506.	3.2	171
23	Inactivating Mutations in GT198 in Familial and Early-Onset Breast and Ovarian Cancers. Genes and Cancer, 2013, 4, 15-25.	1.9	25
24	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
25	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
26	Novel strategies in newborn screening for cystic fibrosis: a prospective controlled study. Thorax, 2012, 67, 289-295.	5.6	53
27	Diagnosis of Fanconi Anemia: Mutation Analysis by Next-Generation Sequencing. Anemia, 2012, 2012, 1-7.	1.7	44
28	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
29	Heterozygote FANCD2 mutations associated with childhood T Cell ALL and testicular seminoma. Familial Cancer, 2012, 11, 661-665.	1.9	21
30	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
31	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
32	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
33	CHEK2*1100delC homozygosity is associated with a high breast cancer risk in women. Journal of Medical Genetics, 2011, 48, 860-863.	3.2	66
34	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
35	Origin, functional role, and clinical impact of Fanconi anemia FANCA mutations. Blood, 2011, 117, 3759-3769.	1.4	108
36	Genetic counselling for pulmonary arterial hypertension: a matter of variable variability. Netherlands Heart Journal, 2011, 19, 89-92.	0.8	7

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37	PALB2 analysis in BRCA2-like families. Breast Cancer Research and Treatment, 2011, 127, 357-362.	2.5	45
38	Recurrence and variability of germline EPCAM deletions in Lynch syndrome. Human Mutation, 2011, 32, 407-414.	2.5	137
39	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
40	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
41	A new locus-specific database (LSDB) for mutations in the folliculin (FLCN) gene. Human Mutation, 2010, 31, E1043-E1051.	2.5	93
42	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
43	Hereditary causes of kidney tumours. European Journal of Clinical Investigation, 2010, 40, 433-439.	3.4	30
44	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
45	No evidence that GATA3 rs570613 SNP modifies breast cancer risk. Breast Cancer Research and Treatment, 2009, 117, 371-379.	2.5	12
46	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
47	Early onset of renal cancer in a family with Birtâ€“Hoggâ€“DubÃ© syndrome. Clinical Genetics, 2009, 75, 537-543.	2.0	28
48	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
49	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
50	Birt-Hogg-DubÃ© Syndrome: Clinical and Genetic Studies of 20 Families. Journal of Investigative Dermatology, 2008, 128, 45-49.	0.7	88
51	Variable phenotypes associated with 10q23 microdeletions involving the PTEN and BMPR1A genes. Clinical Genetics, 2008, 74, 145-154.	2.0	52
52	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
53	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
54	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

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55	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
56	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
57	Contribution of CYLN2 and GTF2IRD1 to neurological and cognitive symptoms in Williams Syndrome. Neurobiology of Disease, 2007, 26, 112-124.	4.4	67
58	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
59	The frequent BRCA1 mutation 1135insA has multiple origins: a haplotype study in different populations. BMC Medical Genetics, 2006, 7, 15.	2.1	15
60	A DGGE system for comprehensive mutation screening ofBRCA1andBRCA2: application in a Dutch cancer clinic setting. Human Mutation, 2006, 27, 654-666.	2.5	75
61	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
62	STK11 status and intussusception risk in Peutz-Jeghers syndrome. Journal of Medical Genetics, 2006, 43, e41-e41.	3.2	44
63	Frequency and Spectrum of Cancers in the Peutz-Jeghers Syndrome. Clinical Cancer Research, 2006, 12, 3209-3215.	7.0	746
64	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
65	Hereditary breast cancer syndromes in a Turkish population. Results of molecular germline analysis. Cancer Genetics and Cytogenetics, 2005, 160, 164-168.	1.0	10
66	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
67	Relative frequency and morphology of cancers in STK11 mutation carriers1 â˜†. Gastroenterology, 2004, 126, 1788-1794.	1.3	228
68	Histopathological characteristics of BRCA1- and BRCA2-associated intraperitoneal cancer: a clinic-based study. Familial Cancer, 2003, 2, 73-78.	1.9	60
69	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
70	Large genomic deletions and duplications in the BRCA1 gene identified by a novel quantitative method. Cancer Research, 2003, 63, 1449-53.	0.9	233
71	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
72	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

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73	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
74	Dysplastic changes in prophylactically removed Fallopian tubes of women predisposed to developing ovarian cancer. Journal of Pathology, 2001, 195, 451-456.	4.5	681
75	Molecular Evidence Linking Primary Cancer of the Fallopian Tube to BRCA1 Germline Mutations. Gynecologic Oncology, 2000, 76, 45-50.	1.4	186
76	Heterogeneous spectrum of mutations in the Fanconi anaemia group A gene. European Journal of Human Genetics, 1999, 7, 52-59.	2.8	84
77	Clinical and genetic evaluation of thirty ovarian cancer families. American Journal of Obstetrics and Gynecology, 1998, 178, 85-90.	1.3	43
78	Expression cloning of a cDNA for the major Fanconi anaemia gene, FAA. Nature Genetics, 1996, 14, 320-323.	21.4	401
79	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
80	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
81	Mutagenicity of metabolic oxygen radicals in mammalian cell cultures. Carcinogenesis, 1994, 15, 2695-2699.	2.8	76
82	Mechanism of hyperoxia-induced chromosomal breakage in chinese hamster cells. Environmental and Molecular Mutagenesis, 1993, 22, 264-270.	2.2	16
83	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
84	Cell culture models for oxidative stress: superoxide and hydrogen peroxide versus normobaric hyperoxia. Mutation Research - DNAging, 1992, 275, 405-414.	3.2	166
85	Effect of normobaric hyperoxia on antioxidant defenses of hela and CHO cells. Free Radical Biology and Medicine, 1988, 4, 85-91.	2.9	14
86	Antioxidant status of Fanconi anemia fibroblasts. Human Genetics, 1987, 77, 28-31.	3.8	43
87	The Cowden syndrome: a clinical and genetic study in 21 patients. Clinical Genetics, 1986, 29, 222-233.	2.0	478