Frederik J Hes

List of Publications by Year
in descending order

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Version: 2024-02-01

68	2,492	159358	205818
papers	citations	30 h-index	48 g-index

68	68	68	4181
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Use of sanger and next-generation sequencing to screen for mosaic and intronic APC variants in unexplained colorectal polyposis patients. Familial Cancer, 2022, 21, 79-83.	0.9	2
2	Genotype-phenotype correlations for pancreatic cancer risk in Dutch melanoma families with pathogenic CDKN2A variants. Journal of Medical Genetics, 2021, 58, 264-269.	1.5	13
3	Germline DLST Variants Promote Epigenetic Modifications in Pheochromocytoma-Paraganglioma. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 459-471.	1.8	6
4	Declining detection rates for APC and biallelic MUTYH variants in polyposis patients, implications for DNA testing policy. European Journal of Human Genetics, 2020, 28, 222-230.	1.4	12
5	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.	0.6	7
6	The complexity of screening PMS2 in DNA isolated from formalin-fixed paraffin-embedded material. European Journal of Human Genetics, 2020, 28, 333-338.	1.4	10
7	Variant type is associated with disease characteristics in SDHB, SDHC and SDHD-linked phaeochromocytomaâ€“paraganglioma. Journal of Medical Genetics, 2020, 57, 96-103.	1.5	16
8	Disease expression in juvenile polyposis syndrome: a retrospective survey on a cohort of 221 European patients and comparison with a literature-derived cohort of 473 SMAD4/BMPR1A pathogenic variant carriers. Genetics in Medicine, 2020, 22, 1524-1532.	1.1	44
9	Mathematical Models for Tumor Growth and the Reduction of Overtreatment. Journal of Neurological Surgery, Part B: Skull Base, 2019, 80, 072-078.	0.4	6
10	Estimating the penetrance of pathogenic gene variants in families with missing pedigree information. Statistical Methods in Medical Research, 2019, 28, 2924-2936.	0.7	2
11	Increased Mortality in SDHB but Not in SDHD Pathogenic Variant Carriers. Cancers, 2019, 11, 103.	1.7	16
12	Low frequency of POLD1 and POLE exonuclease domain variants in patients with multiple colorectal polyps. Molecular Genetics & Genomic Medicine, 2019, 7, e00603.	0.6	8
13	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. Cancer Cell, 2019, 35, 256-266.e5.	7.7	123
14	Surveillance for familial melanoma: recommendations from a national centre of expertise. British Journal of Dermatology, 2019, 181, 594-596.	1.4	7
15	Multigene panel sequencing of established and candidate melanoma susceptibility genes in a large cohort of Dutch nonâ€CDKN2A/CDK4 melanoma families. International Journal of Cancer, 2019, 144, 2453-2464.	2.3	33
16	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1 Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 277-284.	1.8	22
17	CM-Score: a validated scoring system to predict CDKN2A germline mutations in melanoma families from Northern Europe. Journal of Medical Genetics, 2018, 55, 661-668.	1.5	13
18	Clinical Aspects of SDHA-Related Pheochromocytoma and Paraganglioma: A Nationwide Study. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 438-445.	1.8	62

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19	Role of germline aberrations affecting CTNNA1, MAP3K6 and MYD88 in gastric cancer susceptibility. Journal of Medical Genetics, 2018, 55, 669-674.	1.5	37
20	RNA analysis of cancer predisposing genes in formalin-fixed paraffin-embedded tissue determines aberrant splicing. European Journal of Human Genetics, 2018, 26, 1143-1150.	1.4	6
21	The penetrance of paraganglioma and pheochromocytoma in <scp>SDHB</scp> germline mutation carriers. Clinical Genetics, 2018, 93, 60-66.	1.0	51
22	Excluding Lynch syndrome in a female patient with metachronous DNA mismatch repair deficient colon- and ovarian cancer. Familial Cancer, 2018, 17, 415-420.	0.9	2
23	Cancer Risks for PMS2-Associated Lynch Syndrome. Journal of Clinical Oncology, 2018, 36, 2961-2968.	0.8	147
24	Clinical progression and metachronous paragangliomas in a large cohort of SDHD germline variant carriers. European Journal of Human Genetics, 2018, 26, 1339-1347.	1.4	8
25	Molecular Background of Colorectal Tumors From Patients With Lynch Syndrome Associated With Germline Variants in PMS2. Gastroenterology, 2018, 155, 844-851.	0.6	38
26	High Growth Rate of Pancreatic Ductal Adenocarcinoma in CDKN2A-p16-Leiden Mutation Carriers. Cancer Prevention Research, 2018, 11, 551-556.	0.7	5
27	The phenotype of SDHB germline mutation carriers: a nationwide study. European Journal of Endocrinology, 2017, 177, 115-125.	1.9	38
28	CDC73-Related Disorders: Clinical Manifestations and Case Detection in Primary Hyperparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4534-4540.	1.8	65
29	Age and Tumor Volume Predict Growth of Carotid and Vagal Body Paragangliomas. Journal of Neurological Surgery, Part B: Skull Base, 2017, 78, 497-505.	0.4	18
30	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	1.4	34
31	Evidence for genetic association between chromosome 1q loci and predisposition to colorectal neoplasia. British Journal of Cancer, 2017, 117, 1215-1223.	2.9	10
32	Distinct Patterns of Somatic Mosaicism in the APC GeneÂinÂNeoplasms From Patients With Unexplained AdenomatousÂPolyposis. Gastroenterology, 2017, 152, 546-549.e3.	0.6	27
33	A novel keratin 13 variant in a fourâ€generation family with white sponge nevus. Clinical Case Reports (discontinued), 2017, 5, 1503-1509.	0.2	12
34	Loss of maternal chromosome 11 is a signature event in SDHAF2, SDHD, and VHL-related paragangliomas, but less significant in SDHB-related paragangliomas. Oncotarget, 2017, 8, 14525-14536.	0.8	21
35	Whole Gene Capture Analysis of 15 CRC Susceptibility Genes in Suspected Lynch Syndrome Patients. PLoS ONE, 2016, 11, e0157381.	1.1	12
36	Measurement of head and neck paragangliomas: is volumetric analysis worth the effort? A method comparison study. Clinical Otolaryngology, 2016, 41, 571-578.	0.6	4

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37	Characterization of endolymphatic sac tumors and von Hippelâ€“Lindau disease in the International Endolymphatic Sac Tumor Registry. Head and Neck, 2016, 38, E673-9.	0.9	48
38	Parent-of-origin tumourigenesis is mediated by an essential imprinted modifier inSDHD-linked paragangliomas:SLC22A18andCDKN1Care candidate tumour modifiers. Human Molecular Genetics, 2016, 25, 3715-3728.	1.4	15
39	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.	1.1	50
40	Combined mismatch repair and POLE/POLD1 defects explain unresolved suspected Lynch syndrome cancers. European Journal of Human Genetics, 2016, 24, 1089-1092.	1.4	110
41	The effect of genotypes and parent of origin on cancer risk and age of cancer development in PMS2 mutation carriers. Genetics in Medicine, 2016, 18, 405-409.	1.1	15
42	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. Scientific Reports, 2015, 5, 16286.	1.6	24
43	Accuracy of Hereditary Diffuse Gastric Cancer Testing Criteria and Outcomes in Patients With a Germline Mutation in CDH1. Gastroenterology, 2015, 149, 897-906.e19.	0.6	70
44	High-resolution melting (HRM) re-analysis of a polyposis patients cohort reveals previously undetected heterozygous and mosaic APC gene mutations. Familial Cancer, 2015, 14, 247-257.	0.9	14
45	No evidence for increased mortality in SDHD variant carriers compared with the general population. European Journal of Human Genetics, 2015, 23, 1713-1716.	1.4	10
46	Pancreatic cancer-associated gene polymorphisms in a nation-wide cohort of p16-Leiden germline mutation carriers; a caseâ€“control study. BMC Research Notes, 2015, 8, 264.	0.6	10
47	Succinate Dehydrogenase (SDH)-Deficient Pancreatic Neuroendocrine Tumor Expands the SDH-Related Tumor Spectrum. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1386-E1393.	1.8	68
48	Lynch Syndrome Caused by Germline PMS2 Mutations: Delineating the Cancer Risk. Journal of Clinical Oncology, 2015, 33, 319-325.	0.8	177
49	Germline variants in POLE are associated with early onset mismatch repair deficient colorectal cancer. European Journal of Human Genetics, 2015, 23, 1080-1084.	1.4	101
50	Phenotype of SDHB mutation carriers in the Netherlands. Familial Cancer, 2014, 13, 651-657.	0.9	9
51	Paraganglioma and pheochromocytoma upon maternal transmission of SDHDmutations. BMC Medical Genetics, 2014, 15, 111.	2.1	38
52	Colorectal cancer risk variants on 11q23 and 15q13 are associated with unexplained adenomatous polyposis. Journal of Medical Genetics, 2014, 51, 55-60.	1.5	21
53	High prevalence of occult paragangliomas in asymptomatic carriers of SDHD and SDHB gene mutations. European Journal of Human Genetics, 2013, 21, 469-470.	1.4	37
54	Risk of malignant paraganglioma in SDHB-mutation and SDHD-mutation carriers: a systematic review and meta-analysis. Journal of Medical Genetics, 2012, 49, 768-776.	1.5	124

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55	Quantification of sequence exchange events betweenPMS2andPMS2CLprovides a basis for improved mutation scanning of Lynch syndrome patients. Human Mutation, 2010, 31, n/a-n/a.	1.1	53
56	Low penetrance of a SDHB mutation in a large Dutch paraganglioma family. BMC Medical Genetics, 2010, 11, 92.	2.1	52
57	Psychosocial impact of Von Hippelâ€“Lindau disease: levels and sources of distress. Clinical Genetics, 2010, 77, 483-491.	1.0	35
58	Enrichment of Low Penetrance Susceptibility Loci in a Dutch Familial Colorectal Cancer Cohort. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 3062-3067.	1.1	64
59	Multiple genomic aberrations in a patient with mental retardation and hypogonadism: 45,X/46,X,psu dic(Y) karyotype, thyroid hormone receptor beta (THRB) mutation and heterozygosity for Wilson disease. American Journal of Medical Genetics, Part A, 2009, 149A, 2231-2235.	0.7	2
60	Where genetics and pathology meet: mulibrey nanism. Journal of Pathology, 2009, 218, 143-145.	2.1	3
61	High frequency of copyâ€neutral LOH in MUTYHâ€associated polyposis carcinomas. Journal of Pathology, 2008, 216, 25-31.	2.1	38
62	Lynch syndrome: still not a familiar picture. World Journal of Surgical Oncology, 2008, 6, 21.	0.8	4
63	Identification of Patients with (Atypical) MUTYH-Associated Polyposis by KRAS2 c.34G > T Prescreening Followed by MUTYH Hotspot Analysis in Formalin-Fixed Paraffin-Embedded Tissue. Clinical Cancer Research, 2008, 14, 139-142.	3.2	51
64	Germline mutations in APC and MUTYH are responsible for the majority of families with attenuated familial adenomatous polyposis. Clinical Genetics, 2007, 71, 427-433.	1.0	134
65	Frequency of Von Hippelâ€Lindau germline mutations in classic and nonâ€classic Von Hippelâ€Lindau disease identified by DNA sequencing, Southern blot analysis and multiplex ligationâ€dependent probe amplification. Clinical Genetics, 2007, 72, 122-129.	1.0	58
66	The natural history of a combined defect in MSH6 and MUTYH in a HNPCC family. Familial Cancer, 2007, 6, 43-51.	0.9	21
67	Von Hippel-Lindau Disease. Hereditary Cancer in Clinical Practice, 2005, 3, 171-8.	0.6	21
68	Pheochromocytoma in Von Hippel-Lindau Disease. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 969-974.	1.8	78