

# Frederik J Hes

## List of Publications by Year in descending order

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68  
papers

2,492  
citations

159358

30  
h-index

205818

48  
g-index

68  
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68  
docs citations

68  
times ranked

4181  
citing authors

#	ARTICLE	IF	CITATIONS
1	Lynch Syndrome Caused by Germline <i>PMS2</i> Mutations: Delineating the Cancer Risk. <i>Journal of Clinical Oncology</i> , 2015, 33, 319-325.	0.8	177
2	Cancer Risks for <i>PMS2</i> -Associated Lynch Syndrome. <i>Journal of Clinical Oncology</i> , 2018, 36, 2961-2968.	0.8	147
3	Germline mutations in APC and MUTYH are responsible for the majority of families with attenuated familial adenomatous polyposis. <i>Clinical Genetics</i> , 2007, 71, 427-433.	1.0	134
4	Risk of malignant paraganglioma in SDHB-mutation and SDHD-mutation carriers: a systematic review and meta-analysis. <i>Journal of Medical Genetics</i> , 2012, 49, 768-776.	1.5	124
5	Mutational Signature Analysis Reveals NTHL1 Deficiency to Cause a Multi-tumor Phenotype. <i>Cancer Cell</i> , 2019, 35, 256-266.e5.	7.7	123
6	Combined mismatch repair and POLE/POLD1 defects explain unresolved suspected Lynch syndrome cancers. <i>European Journal of Human Genetics</i> , 2016, 24, 1089-1092.	1.4	110
7	Germline variants in POLE are associated with early onset mismatch repair deficient colorectal cancer. <i>European Journal of Human Genetics</i> , 2015, 23, 1080-1084.	1.4	101
8	Pheochromocytoma in Von Hippel-Lindau Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 969-974.	1.8	78
9	Accuracy of Hereditary Diffuse Gastric Cancer Testing Criteria and Outcomes in Patients With a Germline Mutation in CDH1. <i>Gastroenterology</i> , 2015, 149, 897-906.e19.	0.6	70
10	Succinate Dehydrogenase (SDH)-Deficient Pancreatic Neuroendocrine Tumor Expands the SDH-Related Tumor Spectrum. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E1386-E1393.	1.8	68
11	CDC73-Related Disorders: Clinical Manifestations and Case Detection in Primary Hyperparathyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 4534-4540.	1.8	65
12	Enrichment of Low Penetrance Susceptibility Loci in a Dutch Familial Colorectal Cancer Cohort. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2009, 18, 3062-3067.	1.1	64
13	Clinical Aspects of SDHA-Related Pheochromocytoma and Paraganglioma: A Nationwide Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 438-445.	1.8	62
14	Frequency of Von Hippel-Lindau germline mutations in classic and non-classic Von Hippel-Lindau disease identified by DNA sequencing, Southern blot analysis and multiplex ligation-dependent probe amplification. <i>Clinical Genetics</i> , 2007, 72, 122-129.	1.0	58
15	Quantification of sequence exchange events between PMS2 and PMS2CL provides a basis for improved mutation scanning of Lynch syndrome patients. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	1.1	53
16	Low penetrance of a SDHB mutation in a large Dutch paraganglioma family. <i>BMC Medical Genetics</i> , 2010, 11, 92.	2.1	52
17	Identification of Patients with (Atypical) <i>MUTYH</i> -Associated Polyposis by <i>KRAS2</i> c.34G &gt; T Prescreening Followed by <i>MUTYH</i> Hotspot Analysis in Formalin-Fixed Paraffin-Embedded Tissue. <i>Clinical Cancer Research</i> , 2008, 14, 139-142.	3.2	51
18	The penetrance of paraganglioma and pheochromocytoma in <i>SDHB</i> germline mutation carriers. <i>Clinical Genetics</i> , 2018, 93, 60-66.	1.0	51

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19	Comprehensive Mutation Analysis of PMS2 in a Large Cohort of Proband Suspected of Lynch Syndrome or Constitutional Mismatch Repair Deficiency Syndrome. <i>Human Mutation</i> , 2016, 37, 1162-1179.	1.1	50
20	Characterization of endolymphatic sac tumors and von Hippel-Lindau disease in the International Endolymphatic Sac Tumor Registry. <i>Head and Neck</i> , 2016, 38, E673-9.	0.9	48
21	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. <i>Genetics in Medicine</i> , 2020, 22, 1524-1532.	1.1	44
22	High frequency of copy-neutral LOH in MUTYH-associated polyposis carcinomas. <i>Journal of Pathology</i> , 2008, 216, 25-31.	2.1	38
23	Paraganglioma and pheochromocytoma upon maternal transmission of SDHD mutations. <i>BMC Medical Genetics</i> , 2014, 15, 111.	2.1	38
24	The phenotype of SDHB germline mutation carriers: a nationwide study. <i>European Journal of Endocrinology</i> , 2017, 177, 115-125.	1.9	38
25	Molecular Background of Colorectal Tumors From Patients With Lynch Syndrome Associated With Germline Variants in PMS2. <i>Gastroenterology</i> , 2018, 155, 844-851.	0.6	38
26	High prevalence of occult paragangliomas in asymptomatic carriers of SDHD and SDHB gene mutations. <i>European Journal of Human Genetics</i> , 2013, 21, 469-470.	1.4	37
27	Role of germline aberrations affecting CTNNA1, MAP3K6 and MYD88 in gastric cancer susceptibility. <i>Journal of Medical Genetics</i> , 2018, 55, 669-674.	1.5	37
28	Psychosocial impact of Von Hippel-Lindau disease: levels and sources of distress. <i>Clinical Genetics</i> , 2010, 77, 483-491.	1.0	35
29	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 1246-1252.	1.4	34
30	Multigene panel sequencing of established and candidate melanoma susceptibility genes in a large cohort of Dutch non-CDKN2A/CDK4 melanoma families. <i>International Journal of Cancer</i> , 2019, 144, 2453-2464.	2.3	33
31	Distinct Patterns of Somatic Mosaicism in the APC Gene in Neoplasms From Patients With Unexplained Adenomatous Polyposis. <i>Gastroenterology</i> , 2017, 152, 546-549.e3.	0.6	27
32	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. <i>Scientific Reports</i> , 2015, 5, 16286.	1.6	24
33	Clinical and Molecular Characteristics May Alter Treatment Strategies of Thyroid Malignancies in DICER1 Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 277-284.	1.8	22
34	Von Hippel-Lindau Disease. <i>Hereditary Cancer in Clinical Practice</i> , 2005, 3, 171-8.	0.6	21
35	The natural history of a combined defect in MSH6 and MUTYH in a HNPCC family. <i>Familial Cancer</i> , 2007, 6, 43-51.	0.9	21
36	Colorectal cancer risk variants on 11q23 and 15q13 are associated with unexplained adenomatous polyposis. <i>Journal of Medical Genetics</i> , 2014, 51, 55-60.	1.5	21

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37	Loss of maternal chromosome 11 is a signature event in SDHAF2, SDHD, and VHL-related paragangliomas, but less significant in SDHB-related paragangliomas. <i>Oncotarget</i> , 2017, 8, 14525-14536.	0.8	21
38	Age and Tumor Volume Predict Growth of Carotid and Vagal Body Paragangliomas. <i>Journal of Neurological Surgery, Part B: Skull Base</i> , 2017, 78, 497-505.	0.4	18
39	Increased Mortality in SDHB but Not in SDHD Pathogenic Variant Carriers. <i>Cancers</i> , 2019, 11, 103.	1.7	16
40	Variant type is associated with disease characteristics in SDHB, SDHC and SDHD-linked pheochromocytoma/paraganglioma. <i>Journal of Medical Genetics</i> , 2020, 57, 96-103.	1.5	16
41	Parent-of-origin tumorigenesis is mediated by an essential imprinted modifier in SDHD-linked paragangliomas: SLC22A18 and CDKN1C are candidate tumour modifiers. <i>Human Molecular Genetics</i> , 2016, 25, 3715-3728.	1.4	15
42	The effect of genotypes and parent of origin on cancer risk and age of cancer development in PMS2 mutation carriers. <i>Genetics in Medicine</i> , 2016, 18, 405-409.	1.1	15
43	High-resolution melting (HRM) re-analysis of a polyposis patients cohort reveals previously undetected heterozygous and mosaic APC gene mutations. <i>Familial Cancer</i> , 2015, 14, 247-257.	0.9	14
44	CM-Score: a validated scoring system to predict CDKN2A germline mutations in melanoma families from Northern Europe. <i>Journal of Medical Genetics</i> , 2018, 55, 661-668.	1.5	13
45	Genotype-phenotype correlations for pancreatic cancer risk in Dutch melanoma families with pathogenic CDKN2A variants. <i>Journal of Medical Genetics</i> , 2021, 58, 264-269.	1.5	13
46	Whole Gene Capture Analysis of 15 CRC Susceptibility Genes in Suspected Lynch Syndrome Patients. <i>PLoS ONE</i> , 2016, 11, e0157381.	1.1	12
47	A novel keratin 13 variant in a four-generation family with white sponge nevus. <i>Clinical Case Reports (discontinued)</i> , 2017, 5, 1503-1509.	0.2	12
48	Declining detection rates for APC and biallelic MUTYH variants in polyposis patients, implications for DNA testing policy. <i>European Journal of Human Genetics</i> , 2020, 28, 222-230.	1.4	12
49	No evidence for increased mortality in SDHD variant carriers compared with the general population. <i>European Journal of Human Genetics</i> , 2015, 23, 1713-1716.	1.4	10
50	Pancreatic cancer-associated gene polymorphisms in a nation-wide cohort of p16-Leiden germline mutation carriers; a case-control study. <i>BMC Research Notes</i> , 2015, 8, 264.	0.6	10
51	Evidence for genetic association between chromosome 1q loci and predisposition to colorectal neoplasia. <i>British Journal of Cancer</i> , 2017, 117, 1215-1223.	2.9	10
52	The complexity of screening PMS2 in DNA isolated from formalin-fixed paraffin-embedded material. <i>European Journal of Human Genetics</i> , 2020, 28, 333-338.	1.4	10
53	Phenotype of SDHB mutation carriers in the Netherlands. <i>Familial Cancer</i> , 2014, 13, 651-657.	0.9	9
54	Clinical progression and metachronous paragangliomas in a large cohort of SDHD germline variant carriers. <i>European Journal of Human Genetics</i> , 2018, 26, 1339-1347.	1.4	8

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55	Low frequency of POLD1 and POLE exonuclease domain variants in patients with multiple colorectal polyps. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e00603.	0.6	8
56	Surveillance for familial melanoma: recommendations from a national centre of expertise. <i>British Journal of Dermatology</i> , 2019, 181, 594-596.	1.4	7
57	Lack of genotype-phenotype correlation in basal cell nevus syndrome: A Dutch multicenter retrospective cohort study. <i>Journal of the American Academy of Dermatology</i> , 2020, 83, 604-607.	0.6	7
58	RNA analysis of cancer predisposing genes in formalin-fixed paraffin-embedded tissue determines aberrant splicing. <i>European Journal of Human Genetics</i> , 2018, 26, 1143-1150.	1.4	6
59	Mathematical Models for Tumor Growth and the Reduction of Overtreatment. <i>Journal of Neurological Surgery, Part B: Skull Base</i> , 2019, 80, 072-078.	0.4	6
60	Germline <i>DLST</i> Variants Promote Epigenetic Modifications in Pheochromocytoma-Paraganglioma. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 459-471.	1.8	6
61	High Growth Rate of Pancreatic Ductal Adenocarcinoma in <i>CDKN2A-p16-Leiden</i> Mutation Carriers. <i>Cancer Prevention Research</i> , 2018, 11, 551-556.	0.7	5
62	Lynch syndrome: still not a familiar picture. <i>World Journal of Surgical Oncology</i> , 2008, 6, 21.	0.8	4
63	Measurement of head and neck paragangliomas: is volumetric analysis worth the effort? A method comparison study. <i>Clinical Otolaryngology</i> , 2016, 41, 571-578.	0.6	4
64	Where genetics and pathology meet: mulibrey nanism. <i>Journal of Pathology</i> , 2009, 218, 143-145.	2.1	3
65	Multiple genomic aberrations in a patient with mental retardation and hypogonadism: 45,X/46,X,psu dic(Y) karyotype, thyroid hormone receptor beta ( <i>THRβ</i> ) mutation and heterozygosity for Wilson disease. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2231-2235.	0.7	2
66	Excluding Lynch syndrome in a female patient with metachronous DNA mismatch repair deficient colon- and ovarian cancer. <i>Familial Cancer</i> , 2018, 17, 415-420.	0.9	2
67	Estimating the penetrance of pathogenic gene variants in families with missing pedigree information. <i>Statistical Methods in Medical Research</i> , 2019, 28, 2924-2936.	0.7	2
68	Use of sanger and next-generation sequencing to screen for mosaic and intronic APC variants in unexplained colorectal polyposis patients. <i>Familial Cancer</i> , 2022, 21, 79-83.	0.9	2