

# Ian M Frayling

## List of Publications by Year in descending order

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Version: 2024-02-01

56  
papers

5,205  
citations

172457

29  
h-index

175258

52  
g-index

62  
all docs

62  
docs citations

62  
times ranked

6532  
citing authors

#	ARTICLE	IF	CITATIONS
1	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. <i>Gut</i> , 2013, 62, 812-823.	12.1	630
2	Autosomal recessive colorectal adenomatous polyposis due to inherited mutations of MYH. <i>Lancet, The</i> , 2003, 362, 39-41.	13.7	421
3	Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. <i>Gut</i> , 2017, 66, 464-472.	12.1	411
4	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. <i>Nature Genetics</i> , 2014, 46, 107-115.	21.4	410
5	Cancer risk and survival in <i>path_MMR</i> carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. <i>Gut</i> , 2018, 67, 1306-1316.	12.1	410
6	Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. <i>Genetics in Medicine</i> , 2020, 22, 15-25.	2.4	365
7	The type of somatic mutation at APC in familial adenomatous polyposis is determined by the site of the germline mutation: a new facet to Knudson's 'two-hit' hypothesis. <i>Nature Medicine</i> , 1999, 5, 1071-1075.	30.7	339
8	The NF1 somatic mutational landscape in sporadic human cancers. <i>Human Genomics</i> , 2017, 11, 13.	2.9	203
9	DNA mismatch repair deficiency in sporadic colorectal cancer and Lynch syndrome. <i>Histopathology</i> , 2010, 56, 167-179.	2.9	198
10	The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. <i>Genetics in Medicine</i> , 2019, 21, 2390-2400.	2.4	153
11	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotype-Phenotype Correlation. <i>Human Mutation</i> , 2015, 36, 1052-1063.	2.5	143
12	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. <i>Gut</i> , 2017, 66, 1657-1664.	12.1	127
13	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. <i>Clinical Cancer Research</i> , 2008, 14, 1015-1024.	7.0	119
14	A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome. <i>Health Technology Assessment</i> , 2014, 18, 1-406.	2.8	98
15	Molecular pathology of Lynch syndrome. <i>Journal of Pathology</i> , 2020, 250, 518-531.	4.5	96
16	Inherited susceptibility to colorectal adenomas and carcinomas: Evidence for a new predisposition gene on 15q14-q22. <i>Gastroenterology</i> , 1999, 116, 789-795.	1.3	92
17	Consensus for genes to be included on cancer panel tests offered by UK genetics services: guidelines of the UK Cancer Genetics Group. <i>Journal of Medical Genetics</i> , 2018, 55, 372-377.	3.2	88
18	Molecular testing for Lynch syndrome in people with colorectal cancer: systematic reviews and economic evaluation. <i>Health Technology Assessment</i> , 2017, 21, 1-238.	2.8	83

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19	Molecular diagnosis of neurofibromatosis type 1: 20 years experience. <i>Familial Cancer</i> , 2007, 6, 21-34.	1.9	74
20	The proportion of endometrial tumours associated with Lynch syndrome (PETALS): A prospective cross-sectional study. <i>PLoS Medicine</i> , 2020, 17, e1003263.	8.4	58
21	Colorectal cancer incidence in path_MLH1 carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. <i>Hereditary Cancer in Clinical Practice</i> , 2017, 15, 18.	1.5	49
22	Gastric tumours in FAP. <i>Familial Cancer</i> , 2017, 16, 363-369.	1.9	48
23	Multilocus Inherited Neoplasia Alleles Syndrome. <i>JAMA Oncology</i> , 2016, 2, 373.	7.1	43
24	Attenuated adenomatous polyposis coli. <i>Diseases of the Colon and Rectum</i> , 1999, 42, 1078-1080.	1.3	42
25	Lack of association between screening interval and cancer stage in Lynch syndrome may be accounted for by over-diagnosis; a prospective Lynch syndrome database report. <i>Hereditary Cancer in Clinical Practice</i> , 2019, 17, 8.	1.5	42
26	Prevalence of the APC E1317Q variant in colorectal cancer patients. <i>Cancer Letters</i> , 2000, 149, 203-206.	7.2	41
27	A model-based assessment of the cost-effectiveness of strategies to identify Lynch syndrome in early-onset colorectal cancer patients. <i>BMC Cancer</i> , 2015, 15, 313.	2.6	37
28	Allele loss in colorectal cancer at the Cowden disease/Juvenile Polyposis locus on 10q. <i>Cancer Genetics and Cytogenetics</i> , 1997, 97, 64-69.	1.0	36
29	Universal screening for Lynch syndrome in a large consecutive cohort of Chinese colorectal cancer patients: High prevalence and unique molecular features. <i>International Journal of Cancer</i> , 2019, 144, 2161-2168.	5.1	34
30	Lynch syndrome - cancer pathways, heterogeneity and immune escape. <i>Journal of Pathology</i> , 2018, 246, 129-133.	4.5	30
31	Breast cancer risk in neurofibromatosis type 1 is a function of the type of <i>NF1</i> gene mutation: a new genotype-phenotype correlation. <i>Journal of Medical Genetics</i> , 2019, 56, 209-219.	3.2	26
32	Evidence for the simultaneous expression of alternatively spliced alkylpurine N-glycosylase transcripts in human tissues and cells. <i>Carcinogenesis</i> , 1994, 15, 2957-2960.	2.8	24
33	$\beta$ -Catenin expression and allelic loss at APC in sporadic colorectal carcinogenesis. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2002, 440, 362-366.	2.8	22
34	Cost-effectiveness analysis of reflex testing for Lynch syndrome in women with endometrial cancer in the UK setting. <i>PLoS ONE</i> , 2019, 14, e0221419.	2.5	22
35	The predicted impact and cost-effectiveness of systematic testing of people with incident colorectal cancer for Lynch syndrome. <i>Medical Journal of Australia</i> , 2020, 212, 72-81.	1.7	22
36	Urgent improvements needed to diagnose and manage Lynch syndrome. <i>BMJ: British Medical Journal</i> , 2017, 356, j1388.	2.3	20

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37	High endothelial venules are associated with microsatellite instability, hereditary background and immune evasion in colorectal cancer. <i>British Journal of Cancer</i> , 2019, 121, 395-404.	6.4	20
38	Lynch syndrome screening in gynaecological cancers: results of an international survey with recommendations for uniform reporting terminology for mismatch repair immunohistochemistry results. <i>Histopathology</i> , 2019, 75, 813-824.	2.9	19
39	Identification of two novel SMCHD1 sequence variants in families with FSHD-like muscular dystrophy. <i>European Journal of Human Genetics</i> , 2015, 23, 67-71.	2.8	17
40	Unusual presentation of Lynch Syndrome. <i>Hereditary Cancer in Clinical Practice</i> , 2009, 7, 12.	1.5	12
41	Screening for exonic copy number mutations at MSH2 and MLH1 by MAPH. <i>Familial Cancer</i> , 2005, 4, 145-149.	1.9	10
42	A Distinct Genotype of XP Complementation Group A: Surprisingly Mild Phenotype Highly Prevalent in Northern India/Pakistan/Afghanistan. <i>Journal of Investigative Dermatology</i> , 2016, 136, 869-872.	0.7	10
43	A systematic review of test accuracy studies evaluating molecular micro-satellite instability testing for the detection of individuals with lynch syndrome. <i>BMC Cancer</i> , 2017, 17, 836.	2.6	9
44	Colorectal Cancer Stratification in the Routine Clinical Pathway: A District General Hospital Experience. <i>Applied Immunohistochemistry and Molecular Morphology</i> , 2019, 27, e54-e62.	1.2	7
45	How can histopathologists help clinical genetics in the investigation of suspected hereditary gastrointestinal cancer?. <i>Diagnostic Histopathology</i> , 2015, 21, 137-146.	0.4	6
46	Gene panel testing for breast cancer should not be used to confirm syndromic gene associations. <i>Npj Genomic Medicine</i> , 2018, 3, 32.	3.8	6
47	Searching for Mutations: Familial Adenomatous Polyposis as a Case Study. , 1996, 5, 63-98.		5
48	Cerebral primitive neuroectodermal tumor in an adult with a heterozygous MSH2 mutation. <i>Nature Reviews Clinical Oncology</i> , 2009, 6, 295-299.	27.6	4
49	MYH polyposis: A new autosomal recessive form of familial adenomatous polyposis due to defective base excision repair-reappraisal of genetic risk and family management. <i>Gastroenterology</i> , 2003, 124, A46.	1.3	2
50	Pathways to a cancer-free future: a protocol for modelled evaluations to minimise the future burden of colorectal cancer in Australia. <i>BMJ Open</i> , 2020, 10, e036475.	1.9	1
51	Universal consent form might help. <i>BMJ: British Medical Journal</i> , 2004, 328, 1203.5.	2.3	1
52	Application of Molecular Diagnostics to Hereditary Nonpolyposis Colorectal Cancer. , 2006, , 375-392.		0
53	Implementation and auditing of new genetics and tests: translating genetic tests into practice in the NHS. , 0, , 193-198.		0
54	Establishing pathogenicity of germline mismatch repair gene mutations: A Bayesian model. <i>European Journal of Surgical Oncology</i> , 2009, 35, 1208.	1.0	0

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55	Getting It Right with Lynch Syndrome Genetic and Phenotypic Diagnosis. Human Mutation, 2015, 36, iii-iii.	2.5	0
56	Genomic Applications in Clinical Pediatrics. , 2014, , 603-622.		0