Ian M Frayling

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4033252/publications.pdf

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	Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Genetics in Medicine, 2020, 22, 15-25.	2.4	365
2	The predicted impact and costâ€effectiveness of systematic testing of people with incident colorectal cancer for Lynch syndrome. Medical Journal of Australia, 2020, 212, 72-81.	1.7	22
3	Pathways to a cancer-free future: a protocol for modelled evaluations to minimise the future burden of colorectal cancer in Australia. BMJ Open, 2020, 10, e036475.	1.9	1
4	The proportion of endometrial tumours associated with Lynch syndrome (PETALS): A prospective cross-sectional study. PLoS Medicine, 2020, 17, e1003263.	8.4	58
5	Molecular pathology of Lynch syndrome. Journal of Pathology, 2020, 250, 518-531.	4.5	96
6	Lynch syndrome screening in gynaecological cancers: results of an international survey with recommendations for uniform reporting terminology for mismatch repair immunohistochemistry results. Histopathology, 2019, 75, 813-824.	2.9	19
7	High endothelial venules are associated with microsatellite instability, hereditary background and immune evasion in colorectal cancer. British Journal of Cancer, 2019, 121, 395-404.	6.4	20
8	Cost-effectiveness analysis of reflex testing for Lynch syndrome in women with endometrial cancer in the UK setting. PLoS ONE, 2019, 14, e0221419.	2.5	22
9	The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. Genetics in Medicine, 2019, 21, 2390-2400.	2.4	153
10	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. Hereditary Cancer in Clinical Practice, 2019, 17, 8.	1.5	42
11	Colorectal Cancer Stratification in the Routine Clinical Pathway: A District General Hospital Experience. Applied Immunohistochemistry and Molecular Morphology, 2019, 27, e54-e62.	1.2	7
12	Universal screening for Lynch syndrome in a large consecutive cohort of Chinese colorectal cancer patients: High prevalence and unique molecular features. International Journal of Cancer, 2019, 144, 2161-2168.	5.1	34
13	Breast cancer risk in neurofibromatosis type 1 is a function of the type of NF1 gene mutation: a new genotype-phenotype correlation. Journal of Medical Genetics, 2019, 56, 209-219.	3.2	26
14	Consensus for genes to be included on cancer panel tests offered by UK genetics services: guidelines of the UK Cancer Genetics Group. Journal of Medical Genetics, 2018, 55, 372-377.	3.2	88
15	Cancer risk and survival in path_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Gut, 2018, 67, 1306-1316.	12.1	410
16	Gene panel testing for breast cancer should not be used to confirm syndromic gene associations. Npj Genomic Medicine, 2018, 3, 32.	3.8	6
17	Lynch syndrome â€“ cancer pathways, heterogeneity and immune escape. Journal of Pathology, 2018, 246, 129-133.	4.5	30
18	Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. Gut, 2017, 66, 464-472.	12.1	411

#	Article	IF	Citations
19	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. Gut, 2017, 66, 1657-1664.	12.1	127
20	Urgent improvements needed to diagnose and manage Lynch syndrome. BMJ: British Medical Journal, 2017, 356, j1388.	2.3	20
21	Gastric tumours in FAP. Familial Cancer, 2017, 16, 363-369.	1.9	48
22	The NF1 somatic mutational landscape in sporadic human cancers. Human Genomics, 2017, 11, 13.	2.9	203
23	A systematic review of test accuracy studies evaluating molecular micro-satellite instability testing for the detection of individuals with lynch syndrome. BMC Cancer, 2017, 17, 836.	2.6	9
24	Colorectal cancer incidence in path_MLH1 carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. Hereditary Cancer in Clinical Practice, 2017, 15, 18.	1.5	49
25	Molecular testing for Lynch syndrome in people with colorectal cancer: systematic reviews and economic evaluation. Health Technology Assessment, 2017, 21, 1-238.	2.8	83
26	A Distinct Genotype of XP Complementation Group A: Surprisingly Mild Phenotype HighlyÂPrevalent in Northern India/Pakistan/Afghanistan. Journal of Investigative Dermatology, 2016, 136, 869-872.	0.7	10
27	Multilocus Inherited Neoplasia Alleles Syndrome. JAMA Oncology, 2016, 2, 373.	7.1	43
28	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
29	How can histopathologists help clinical genetics in the investigation of suspected hereditary gastrointestinal cancer?. Diagnostic Histopathology, 2015, 21, 137-146.	0.4	6
30	A model-based assessment of the costâ€“utility of strategies to identify Lynch syndrome in early-onset colorectal cancer patients. BMC Cancer, 2015, 15, 313.	2.6	37
31	Getting It Right with Lynch Syndrome Genetic and Phenotypic Diagnosis. Human Mutation, 2015, 36, iii-iii.	2.5	0
32	Identification of two novel SMCHD1 sequence variants in families with FSHD-like muscular dystrophy. European Journal of Human Genetics, 2015, 23, 67-71.	2.8	17
33	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
34	A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome. Health Technology Assessment, 2014, 18, 1-406.	2.8	98
35	Genomic Applications in Clinical Pediatrics. , 2014, , 603-622.		0
36	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. Gut, 2013, 62, 812-823.	12.1	630

#	Article	IF	Citations
37	DNA mismatch repair deficiency in sporadic colorectal cancer and Lynch syndrome. Histopathology, 2010, 56, 167-179.	2.9	198
38	Cerebral primitive neuroectodermal tumor in an adult with a heterozygous MSH2 mutation. Nature Reviews Clinical Oncology, 2009, 6, 295-299.	27.6	4
39	Establishing pathogenicity of germline mismatch repair gene mutations: A Bayesian model. European Journal of Surgical Oncology, 2009, 35, 1208.	1.0	0
40	Unusual presentation of Lynch Syndrome. Hereditary Cancer in Clinical Practice, 2009, 7, 12.	1.5	12
41	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. Clinical Cancer Research, 2008, 14, 1015-1024.	7.0	119
42	Molecular diagnosis of neurofibromatosis type 1: 2Âyears experience. Familial Cancer, 2007, 6, 21-34.	1.9	74
43	Application of Molecular Diagnostics to Hereditary Nonpolyposis Colorectal Cancer. , 2006, , 375-392.		0
44	Screening for exonic copy number mutations at MSH2 and MLH1 by MAPH. Familial Cancer, 2005, 4, 145-149.	1.9	10
45	Universal consent form might help. BMJ: British Medical Journal, 2004, 328, 1203.5.	2.3	1
46	MYH polyposis: A new autosomal recessive form of familial adenomatous polyposis due to defective base excision repair-reappraisal of genetic risk and family management. Gastroenterology, 2003, 124, A46.	1.3	2
47	Autosomal recessive colorectal adenomatous polyposis due to inherited mutations of MYH. Lancet, The, 2003, 362, 39-41.	13.7	421
48	Î²-Catenin expression and allelic loss at APC in sporadic colorectal carcinogenesis. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2002, 440, 362-366.	2.8	22
49	Prevalence of the APC E1317Q variant in colorectal cancer patients. Cancer Letters, 2000, 149, 203-206.	7.2	41
50	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. Nature Medicine, 1999, 5, 1071-1075.	30.7	339
51	Attenuated adenomatous polyposis coli. Diseases of the Colon and Rectum, 1999, 42, 1078-1080.	1.3	42
52	Inherited susceptibility to colorectal adenomas and carcinomas: Evidence for a new predisposition gene on 15q14-q22. Gastroenterology, 1999, 116, 789-795.	1.3	92
53	Allele loss in colorectal cancer at the Cowden disease/Juvenile Polyposis locus on 10q. Cancer Genetics and Cytogenetics, 1997, 97, 64-69.	1.0	36
54	Searching for Mutations: Familial Adenomatous Polyposis as a Case Study. , 1996, 5, 63-98.		5

#	Article	IF	Citations
55	Evidence for the simultaneous expression of alternatively spliced alkylpurine N-glycosylase transcripts in human tissues and cells. Carcinogenesis, 1994, 15, 2957-2960.	2.8	24
56	Implementation and auditing of new genetics and tests: translating genetic tests into practice in the NHS. , 0, , 193-198.		0