Karl Heinimann

List of Publications by Year
in descending order

30 papers	1,961 citations	516710 16 h-index	434195 31 g-index
31 all docs	31 docs citations	31 times ranked	3723 citing authors

#	Article	IF	Citations
1	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
2	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
3	Exome sequencing of osteosarcoma reveals mutation signatures reminiscent of BRCA deficiency. Nature Communications, 2015, 6, 8940.	12.8	242
4	Immunohistochemical Analysis Reveals High Frequency of PMS2 Defects in Colorectal Cancer. Gastroenterology, 2005, 128, 1160-1171.	1.3	166
5	The evolutionary landscape of colorectal tumorigenesis. Nature Ecology and Evolution, 2018, 2, 1661-1672.	7.8	99
6	Exome sequencing of fetal anomaly syndromes: novel phenotypeâ€“genotype discoveries. European Journal of Human Genetics, 2019, 27, 730-737.	2.8	44
7	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.	2.4	44
8	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
9	Target gene mutational pattern in Lynch syndrome colorectal carcinomas according to tumour location and germline mutation. British Journal of Cancer, 2015, 113, 686-692.	6.4	30
10	Cancer Predisposition Cascade Screening for Hereditary Breast/Ovarian Cancer and Lynch Syndromes in Switzerland: Study Protocol. JMIR Research Protocols, 2017, 6, e184.	1.0	30
11	Risk-reducing hysterectomy and bilateral salpingo-oophorectomy in female heterozygotes of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. Genetics in Medicine, 2021, 23, 705-712.	2.4	28
12	Toward a Molecular Classification of Colorectal Cancer: The Role of Microsatellite Instability Status. Frontiers in Oncology, 2013, 3, 272.	2.8	23
13	Somatic alterations in juvenile polyps from <scp>BMPR1A</scp> and <scp>SMAD</scp>4 mutation carriers. Genes Chromosomes and Cancer, 2015, 54, 575-582.	2.8	23
14	Skeletal muscle MRI of the lower limbs in congenital muscular dystrophy patients with novel POMT1 and POMT2 mutations. Neuromuscular Disorders, 2014, 24, 321-324.	0.6	20
15	Challenges and Opportunities for Cancer Predisposition Cascade Screening for Hereditary Breast and Ovarian Cancer and Lynch Syndrome in Switzerland: Findings from an International Workshop. Public Health Genomics, 2018, 21, 121-132.	1.0	20
16	Congenital muscular dystrophy with dropped head phenotype and cognitive impairment due to a novel mutation in the LMNA gene. Neuromuscular Disorders, 2014, 24, 529-532.	0.6	17
17	Wide Spectrum of DUOX2 Deficiency: From Life-Threatening Compressive Goiter in Infancy to Lifelong Euthyroidism. Thyroid, 2019, 29, 1018-1022.	4.5	16
18	Swiss cost-effectiveness analysis of universal screening for Lynch syndrome of patients with colorectal cancer followed by cascade genetic testing of relatives. Journal of Medical Genetics, 2022, 59, 924-930.	3.2	16

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19	TRPS1 gene alterations in human subependymoma. Journal of Neuro-Oncology, 2017, 134, 133-138.	2.9	13
20	Risk-Reducing Gynecological Surgery in Lynch Syndrome: Results of an International Survey from the Prospective Lynch Syndrome Database. Journal of Clinical Medicine, 2020, 9, 2290.	2.4	12
21	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. Genetics in Medicine, 2019, 21, 2706-2712.	2.4	11
22	Uptake of hysterectomy and bilateral salpingo-oophorectomy in carriers of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. European Journal of Cancer, 2021, 148, 124-133.	2.8	11
23	No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in MLH1 and MSH2: A Prospective Lynch Syndrome Database Study. Journal of Clinical Medicine, 2021, 10, 2856.	2.4	11
24	SH2D4A is frequently downregulated in hepatocellular carcinoma and cirrhotic nodules. European Journal of Cancer, 2014, 50, 731-738.	2.8	9
25	Systematic immunohistochemical screening for Lynch syndrome in colorectal cancer: a single centre experience of 486 patients. Swiss Medical Weekly, 2016, 146, w14315.	1.6	8
26	Intention to Inform Relatives, Rates of Cascade Testing, and Preference for Patient-Mediated Communication in Families Concerned with Hereditary Breast and Ovarian Cancer and Lynch Syndrome: The Swiss CASCADE Cohort. Cancers, 2022, 14, 1636.	3.7	8
27	Germline RET variants underlie a subset of paediatric osteosarcoma. Journal of Medical Genetics, 2021, 58, 20-24.	3.2	7
28	How the â€œcontrol-fate continuumâ€•helps explain the genetic testing decision-making process: a grounded theory study. European Journal of Human Genetics, 2020, 28, 1010-1019.	2.8	5
29	Genetic Literacy and Communication of Genetic Information in Families Concerned with Hereditary Breast and Ovarian Cancer: A Cross-Study Comparison in Two Countries and within a Timeframe of More Than 10 Years. Cancers, 2021, 13, 6254.	3.7	5
30	Convergent Evolution of Copy Number Alterations in Multi-Centric Hepatocellular Carcinoma. Scientific Reports, 2019, 9, 4611.	3.3	2