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	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
2	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
3	Germline RET variants underlie a subset of paediatric osteosarcoma. Journal of Medical Genetics, 2021, 58, 20-24.	3.2	7
4	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
5	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
6	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
7	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
8	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
9	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
10	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
11	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
12	Exome sequencing of fetal anomaly syndromes: novel phenotypeâ€“genotype discoveries. European Journal of Human Genetics, 2019, 27, 730-737.	2.8	44
13	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. Genetics in Medicine, 2019, 21, 2706-2712.	2.4	11
14	Wide Spectrum of DUOX2 Deficiency: From Life-Threatening Compressive Goiter in Infancy to Lifelong Euthyroidism. Thyroid, 2019, 29, 1018-1022.	4.5	16
15	Convergent Evolution of Copy Number Alterations in Multi-Centric Hepatocellular Carcinoma. Scientific Reports, 2019, 9, 4611.	3.3	2
16	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
17	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
18	The evolutionary landscape of colorectal tumorigenesis. Nature Ecology and Evolution, 2018, 2, 1661-1672.	7.8	99

#	Article	IF	Citations
19	TRPS1 gene alterations in human subependymoma. Journal of Neuro-Oncology, 2017, 134, 133-138.	2.9	13
20	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
21	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
22	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
23	Exome sequencing of osteosarcoma reveals mutation signatures reminiscent of BRCA deficiency. Nature Communications, 2015, 6, 8940.	12.8	242
24	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
25	SH2D4A is frequently downregulated in hepatocellular carcinoma and cirrhotic nodules. European Journal of Cancer, 2014, 50, 731-738.	2.8	9
26	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
27	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
28	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
29	Toward a Molecular Classification of Colorectal Cancer: The Role of Microsatellite Instability Status. Frontiers in Oncology, 2013, 3, 272.	2.8	23
30	Immunohistochemical Analysis Reveals High Frequency of PMS2 Defects in Colorectal Cancer. Gastroenterology, 2005, 128, 1160-1171.	1.3	166