Marta Pineda Riu

List of Publications by Year
in descending order

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100	5,688	136740	82410
papers	citations	32 h-index	72 g-index

102	102	102	6827
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	6.1	411
2	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.	9.4	410
3	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.	6.1	410
4	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.	1.1	365
5	Identification of a Membrane Protein, LAT-2, That Co-expresses with 4F2 Heavy Chain, an L-type Amino Acid Transport Activity with Broad Specificity for Small and Large Zwitterionic Amino Acids. Journal of Biological Chemistry, 1999, 274, 19738-19744.	1.6	356
6	Identification and Characterization of a Membrane Protein (y+L Amino Acid Transporter-1) That Associates with 4F2hc to Encode the Amino Acid Transport Activity y+L. Journal of Biological Chemistry, 1998, 273, 32437-32445.	1.6	304
7	Identification of SLC7A7, encoding y+LAT-1, as the lysinuric protein intolerance gene. Nature Genetics, 1999, 21, 293-296.	9.4	286
8	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	9.4	280
9	POLE and POLD1 mutations in 529 kindred with familial colorectal cancer and/or polyposis: review of reported cases and recommendations for genetic testing and surveillance. Genetics in Medicine, 2016, 18, 325-332.	1.1	209
10	Cancer Risks for PMS2-Associated Lynch Syndrome. Journal of Clinical Oncology, 2018, 36, 2961-2968.	0.8	147
11	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. Human Molecular Genetics, 2014, 23, 3506-3512.	1.4	135
12	Identification and characterisation of human xCT that co-expresses, with 4F2 heavy chain, the amino acid transport activity system x c -. Pflugers Archiv European Journal of Physiology, 2001, 442, 286-296.	1.3	133
13	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.	6.1	127
14	The Genetics of Heteromeric Amino Acid Transporters. Physiology, 2005, 20, 112-124.	1.6	112
15	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	1.1	102
16	Next-generation sequencing meets genetic diagnostics: development of a comprehensive workflow for the analysis of BRCA1 and BRCA2 genes. European Journal of Human Genetics, 2013, 21, 864-870.	1.4	94
17	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. Gastroenterology, 2015, 149, 563-566.	0.6	94
18	MLH1 promoter hypermethylation in the analytical algorithm of Lynch syndrome: a cost-effectiveness study. European Journal of Human Genetics, 2012, 20, 762-768.	1.4	76

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19	Role of POLE and POLD1 in familial cancer. Genetics in Medicine, 2020, 22, 2089-2100.	1.1	76
20	Evaluation of CNV detection tools for NGS panel data in genetic diagnostics. European Journal of Human Genetics, 2020, 28, 1645-1655.	1.4	67
21	Functional analysis of novel mutations in y+LAT-1 amino acid transporter gene causing lysinuric protein intolerance (LPI). Human Molecular Genetics, 2000, 9, 431-438.	1.4	66
22	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. European Journal of Cancer, 2014, 50, 2241-2250.	1.3	66
23	New perspectives on screening and early detection of endometrial cancer. International Journal of Cancer, 2019, 145, 3194-3206.	2.3	58
24	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	5.1	58
25	Computational Tools for Splicing Defect Prediction in Breast/Ovarian Cancer Genes: How Efficient Are They at Predicting RNA Alterations?. Frontiers in Genetics, 2018, 9, 366.	1.1	53
26	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.	0.6	49
27	Cystinuria-specific rBAT(R365W) mutation reveals two translocation pathways in the amino acid transporter rBAT-b0,+AT. Biochemical Journal, 2004, 377, 665-674.	1.7	47
28	Refining the role ofpms2in Lynch syndrome: germline mutational analysis improved by comprehensive assessment of variants. Journal of Medical Genetics, 2013, 50, 552-563.	1.5	47
29	A comprehensive custom panel design for routine hereditary cancer testing: preserving control, improving diagnostics and revealing a complex variation landscape. Scientific Reports, 2017, 7, 39348.	1.6	45
30	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.	0.6	42
31	Exploring the Role of Mutations in Fanconi Anemia Genes in Hereditary Cancer Patients. Cancers, 2020, 12, 829.	1.7	41
32	MLH1 methylation screening is effective in identifying epimutation carriers. European Journal of Human Genetics, 2012, 20, 1256-1264.	1.4	36
33	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. Scientific Reports, 2017, 7, 37984.	1.6	35
34	Association Between Germline Mutations in BRF1, a Subunit of the RNA Polymerase III Transcription Complex, and Hereditary Colorectal Cancer. Gastroenterology, 2018, 154, 181-194.e20.	0.6	32
35	Comprehensive functional assessment ofMLH1variants of unknown significance. Human Mutation, 2012, 33, 1576-1588.	1.1	30
36	Opportunistic testing of BRCA1, BRCA2 and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. International Journal of Cancer, 2019, 145, 2682-2691.	2.3	30

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37	Detection of genetic alterations in hereditary colorectal cancer screeningâ˜†. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2010, 693, 19-31.	0.4	29
38	MLH1 Founder Mutations with Moderate Penetrance in Spanish Lynch Syndrome Families. Cancer Research, 2010, 70, 7379-7391.	0.4	29
39	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.	1.1	28
40	Survival by colon cancer stage and screening interval in Lynch syndrome: a prospective Lynch syndrome database report. Hereditary Cancer in Clinical Practice, 2019, 17, 28.	0.6	27
41	Elucidating the molecular basis of MSH2â€deficient tumors by combined germline and somatic analysis. International Journal of Cancer, 2017, 141, 1365-1380.	2.3	26
42	The amino acid transporter asc-1 is not involved in cystinuria. Kidney International, 2004, 66, 1453-1464.	2.6	25
43	Mutational Heterogeneity in APC and KRAS Arises at the Crypt Level and Leads to Polyclonality in Early Colorectal Tumorigenesis. Clinical Cancer Research, 2017, 23, 5936-5947.	3.2	25
44	GALNT12is Not a Major Contributor of Familial Colorectal Cancer Type X. Human Mutation, 2014, 35, 50-52.	1.1	22
45	Primary constitutional MLH1 epimutations: a focal epigenetic event. British Journal of Cancer, 2018, 119, 978-987.	2.9	22
46	Telomere Length and Genetic Anticipation in Lynch Syndrome. PLoS ONE, 2013, 8, e61286.	1.1	21
47	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. European Journal of Human Genetics, 2013, 21, 511-516.	1.4	20
48	Usefulness of epithelial cell adhesion molecule expression in the algorithmic approach to Lynch syndrome identification. Human Pathology, 2013, 44, 412-416.	1.1	20
49	High-sensitivity microsatellite instability assessment for the detection of mismatch repair defects in normal tissue of biallelic germline mismatch repair mutation carriers. Journal of Medical Genetics, 2020, 57, 269-273.	1.5	20
50	Characteristics of Adrenocortical Carcinoma Associated With Lynch Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 318-325.	1.8	20
51	Germline mutations in the spindle assembly checkpoint genes BUB1 and BUB3 are infrequent in familial colorectal cancer and polyposis. Molecular Cancer, 2018, 17, 23.	7.9	19
52	Clonal relationship and directionality of progression of synchronous endometrial and ovarian carcinomas in patients with DNA mismatch repair-deficiency associated syndromes. Modern Pathology, 2021, 34, 994-1007.	2.9	19
53	Nonâ€Hodgkin lymphoma related to hereditary nonpolyposis colorectal cancer in a patient with a novel heterozygous complex deletion in the MSH2 gene. Genes Chromosomes and Cancer, 2008, 47, 326-332.	1.5	18
54	Characterization of New Founder Alu-Mediated Rearrangements in MSH2 Gene Associated with a Lynch Syndrome Phenotype. Cancer Prevention Research, 2011, 4, 1546-1555.	0.7	17

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55	Quality of Colonoscopy Is Associated With Adenoma Detection and Postcolonoscopy Colorectal Cancer Prevention in Lynch Syndrome. Clinical Gastroenterology and Hepatology, 2022, 20, 611-621.e9.	2.4	17
56	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. Cancers, 2020, 12, 1799.	1.7	15
57	Exome sequencing identifiesMUTYHmutations in a family with colorectal cancer and an atypical phenotype. Gut, 2015, 64, 355-356.	6.1	14
58	A Collaborative Effort to Define Classification Criteria forATMVariants in Hereditary Cancer Patients. Clinical Chemistry, 2021, 67, 518-533.	1.5	14
59	Longer Telomeres Are Associated with Cancer Risk in MMR-Proficient Hereditary Non-Polyposis Colorectal Cancer. PLoS ONE, 2014, 9, e86063.	1.1	13
60	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. Clinical Genetics, 2014, 85, 260-266.	1.0	12
61	Germline variation in O6-methylguanine-DNA methyltransferase (MGMT) as cause of hereditary colorectal cancer. Cancer Letters, 2019, 447, 86-92.	3.2	12
62	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.	1.0	12
63	Founder effect of a pathogenic MSH2 mutation identified in Spanish families with Lynch syndrome. Clinical Genetics, 2010, 78, 186-190.	1.0	11
64	Does multilocus inherited neoplasia alleles syndrome have severe clinical expression?. Journal of Medical Genetics, 2019, 56, 521-525.	1.5	11
65	Assessing Effectiveness of Colonic and Gynecological Risk Reducing Surgery in Lynch Syndrome Individuals. Cancers, 2020, 12, 3419.	1.7	11
66	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.	1.3	11
67	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.	1.0	11
68	Comprehensive analysis and ACMGâ€based classification of CHEK2 variants in hereditary cancer patients. Human Mutation, 2020, 41, 2128-2142.	1.1	10
69	TP53, a gene for colorectal cancer predisposition in the absence of Li-Fraumeni-associated phenotypes. Gut, 2021, 70, 1139-1146.	6.1	10
70	First international workshop of the ATM and cancer risk group (4-5 December 2019). Familial Cancer, 2022, 21, 211-227.	0.9	10
71	Paired Somatic-Germline Testing of 15 Polyposis and Colorectal Cancerâ€“Predisposing Genes Highlights the Role of APC Mosaicism in de Novo Familial Adenomatous Polyposis. Journal of Molecular Diagnostics, 2021, 23, 1452-1459.	1.2	10
72	Scarce evidence of the causal role of germline mutations in UNC5C in hereditary colorectal cancer and polyposis. Scientific Reports, 2016, 6, 20697.	1.6	9

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73	Analysis in the Prospective Lynch Syndrome Database identifies sarcoma as part of the Lynch syndrome tumor spectrum. International Journal of Cancer, 2021, 148, 512-513.	2.3	9
74	The Challenge of Diagnosing Constitutional Mismatch Repair Deficiency Syndrome in Brain Malignancies from Young Individuals. International Journal of Molecular Sciences, 2021, 22, 4629.	1.8	9
75	Improving Genetic Testing in Hereditary Cancer by RNA Analysis. Journal of Molecular Diagnostics, 2020, 22, 1453-1468.	1.2	9
76	Studies on the function of TM20, a transmembrane protein present in cereal embryos. Planta, 2005, 222, 80-90.	1.6	8
77	Comprehensive molecular characterisation of hereditary non-polyposis colorectal tumours with mismatch repair proficiency. European Journal of Cancer, 2014, 50, 1964-1972.	1.3	8
78	ERCC3, a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2020, 141, 1-8.	1.3	8
79	Defining a mutational signature for endometrial cancer screening and early detection. Cancer Epidemiology, 2019, 61, 129-132.	0.8	7
80	Highly sensitive MLH1 methylation analysis in blood identifies a cancer patient with low-level mosaic MLH1 epimutation. Clinical Epigenetics, 2019, 11, 171.	1.8	7
81	Splicing analyses for variants in MMR genes: best practice recommendations from the European Mismatch Repair Working Group. European Journal of Human Genetics, 2022, 30, 1051-1059.	1.4	7
82	Detailed characterization of <scp>MLH1</scp> p.<scp>D41H</scp> and p.<scp>N710D</scp> variants coexisting in a Lynch syndrome family with conserved <scp>MLH1</scp> expression tumors. Clinical Genetics, 2015, 87, 543-548.	1.0	6
83	The Apparent Genetic Anticipation in PMS2-Associated Lynch Syndrome Families Is Explained by Birth-cohort Effect. Cancer Epidemiology Biomarkers and Prevention, 2019, 28, 1010-1014.	1.1	6
84	Sensitivity of cervical cytology in endometrial cancer detection in a tertiary hospital in Spain. Cancer Medicine, 2021, 10, 6762-6766.	1.3	6
85	Night work, chronotype and risk of endometrial cancer in the Screenwide caseâ€“control study. Occupational and Environmental Medicine, 2022, , oemed-2021-108080.	1.3	6
86	An Integrated Approach for the Early Detection of Endometrial and Ovarian Cancers (Screenwide) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.1	6
87	Approaches to functionally validate candidate genetic variants involved in colorectal cancer predisposition. Molecular Aspects of Medicine, 2019, 69, 27-40.	2.7	5
88	Validation of an inÂVitro Mismatch Repair Assay Used in the Functional Characterization of Mismatch Repair Variants. Journal of Molecular Diagnostics, 2020, 22, 376-385.	1.2	5
89	Complete Loss of EPCAM Immunoexpression Identifies EPCAM Deletion Carriers in MSH2-Negative Colorectal Neoplasia. Cancers, 2020, 12, 2803.	1.7	4
90	Variants of uncertain significance (VUS) in cancer predisposing genes: What are we learning from multigene panels?. European Journal of Medical Genetics, 2022, 65, 104400.	0.7	4

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91	Predicting Ovarian-Cancer Burden in Catalonia by 2030: An Ageâ€“Periodâ€“Cohort Modelling. International Journal of Environmental Research and Public Health, 2022, 19, 1404.	1.2	4
92	Colorectal Cancer Incidence in Lynch Syndrome Patients: First Report of a Multicenter Nation-Wide Study. Gastroenterology, 2017, 152, S552.	0.6	3
93	Elucidating the clinical significance of two PMS2 missense variants coexisting in a family fulfilling hereditary cancer criteria. Familial Cancer, 2017, 16, 501-507.	0.9	3
94	Screening of CNVs using NGS data improves mutation detection yield and decreases costs in genetic testing for hereditary cancer. Journal of Medical Genetics, 2020, , jmedgenet-2020-107366.	1.5	3
95	ICO Amplicon NGS Data Analysis: A Web Tool for Variant Detection in Common High-Risk Hereditary Cancer Genes Analyzed by Amplicon GS Junior Next-Generation Sequencing. Human Mutation, 2014, 35, 271-277.	1.1	2
96	The Molecular Basis of Lynch-like Syndrome. , 2018, , 21-41.		2
97	CNVfilteR: an R/Bioconductor package to identify false positives produced by germline NGS CNV detection tools. Bioinformatics, 2021, 37, 4227-4229.	1.8	1
98	Classification of genetic variants in hereditary cancer genes. , 2021, , 349-387.		0
99	Abstract 4445: Defining a pipeline to use next generation sequencing for genetic testing in hereditary cancer. , 2012, , .		0
100	RNA assay identifies a previous misclassification of BARD1 c.1977A>G variant. Scientific Reports, 2021, 11, 22948.	1.6	0