Joan Anton Puig-Butille

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Genome-wide association study identifies three new melanoma susceptibility loci. Nature Genetics, 2011, 43, 1108-1113.	9.4	230
2	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. Nature Genetics, 2015, 47, 987-995.	9.4	218
3	TERT Promoter Mutation Status as an Independent Prognostic Factor in Cutaneous Melanoma. Journal of the National Cancer Institute, 2014, 106, .	3.0	204
4	Benefits of total body photography and digital dermatoscopy ("two-step method of digital) Tj ETQq0 0 0 rgB American Academy of Dermatology, 2012, 67, e17-e27.	T /Overlocl 0.6	x 10 Tf 50 62 176
5	Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility. Nature Genetics, 2020, 52, 494-504.	9.4	138
6	A variant in FTO shows association with melanoma risk not due to BMI. Nature Genetics, 2013, 45, 428-432.	9.4	111
7	The Effect on Melanoma Risk of Genes Previously Associated With Telomere Length. Journal of the National Cancer Institute, 2014, 106, .	3.0	109
8	Update in genetic susceptibility in melanoma. Annals of Translational Medicine, 2015, 3, 210.	0.7	100
9	Genetic Abnormalities in Large to Giant Congenital Nevi: Beyond NRAS Mutations. Journal of Investigative Dermatology, 2019, 139, 900-908.	0.3	67
10	Increased prevalence of lung, breast, and pancreatic cancers in addition to melanoma risk in families bearing the cyclin-dependent kinase inhibitor 2A mutation: Implications for genetic counseling. Journal of the American Academy of Dermatology, 2014, 71, 888-895.	0.6	52
11	The <scp><i>MC1R</i></scp> melanoma risk variant p. <scp>R160W</scp> is associated with <scp>P</scp> arkinson disease. Annals of Neurology, 2015, 77, 889-894.	2.8	52
12	Dermoscopic features of melanomas associated with <i>MC1R</i> variants in Spanish <i>CDKN2A</i> mutation carriers. British Journal of Dermatology, 2009, 160, 48-53.	1.4	48
13	Development and validation of the new HER2DX assay for predicting pathological response and survival outcome in early-stage HER2-positive breast cancer. EBioMedicine, 2022, 75, 103801.	2.7	47
14	Fibroblast activation and abnormal extracellular matrix remodelling as common hallmarks in three cancerâ€prone genodermatoses. British Journal of Dermatology, 2019, 181, 512-522.	1.4	46
15	Mutational status of naevus-associated melanomas. British Journal of Dermatology, 2015, 173, 671-680.	1.4	42
16	Prevalence of <i>MITF</i> p.E318K in Patients With Melanoma Independent of the Presence of <i>CDKN2A</i> Causative Mutations. JAMA Dermatology, 2016, 152, 405.	2.0	41
17	AURKA Overexpression Is Driven byÂFOXM1 and MAPK/ERK Activation inÂMelanoma Cells Harboring BRAF orÂNRASÂMutations: Impact on MelanomaÂPrognosis and Therapy. Journal of Investigative Dermatology, 2017, 137, 1297-1310.	0.3	40
18	<i> <scp>POT</scp> 1 </i> germline mutations but not <i> <scp>TERT</scp> </i> promoter mutations are implicated in melanoma susceptibility in a large cohort of Spanish melanoma families. British Journal of Dermatology, 2019, 181, 105-113.	1.4	37

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19	Capturing the biological impact of CDKN2A and MC1R genes as an early predisposing event in melanoma and non melanoma skin cancer. Oncotarget, 2014, 5, 1439-1451.	0.8	35
20	TERT gene amplification is associated with poor outcome in acral lentiginous melanoma. Journal of the American Academy of Dermatology, 2014, 71, 839-841.	0.6	35
21	Benefits of oral <i>Polypodium Leucotomos</i> extract in MM highâ€risk patients. Journal of the European Academy of Dermatology and Venereology, 2013, 27, 1095-1100.	1.3	34
22	Association of the <i>POT1</i> Germline Missense Variant p.178T With Familial Melanoma. JAMA Dermatology, 2019, 155, 604.	2.0	34
23	Characterization of individuals at high risk of developing melanoma in Latin America: bases for genetic counseling in melanoma. Genetics in Medicine, 2016, 18, 727-736.	1.1	31
24	Fine mapping of genetic susceptibility loci for melanoma reveals a mixture of single variant and multiple variant regions. International Journal of Cancer, 2015, 136, 1351-1360.	2.3	30
25	TERT and AURKA Gene Copy Number Gains Enhance the Detection of Acral Lentiginous Melanomas by Fluorescence in Situ Hybridization. Journal of Molecular Diagnostics, 2014, 16, 198-206.	1.2	28
26	Melanocortin 1 receptor (<i><scp>MC</scp>1R</i>) polymorphisms' influence on size and dermoscopic features of nevi. Pigment Cell and Melanoma Research, 2018, 31, 39-50.	1.5	28
27	Dermoscopic criteria associated with <i> <scp>BRAF</scp> </i> and <i> <scp>NRAS</scp> </i> mutation status in primary cutaneous melanoma. British Journal of Dermatology, 2014, 171, 754-759.	1.4	26
28	Transcriptomic and Genetic Associations between Alzheimer's Disease, Parkinson's Disease, and Cancer. Cancers, 2021, 13, 2990.	1.7	26
29	Distribution of <i>MC1R</i> variants among melanoma subtypes: p.R163Q is associated with lentigo maligna melanoma in a Mediterranean population. British Journal of Dermatology, 2013, 169, 804-811.	1.4	25
30	An inherited variant in the gene coding for vitamin <scp>D</scp> â€binding protein and survival from cutaneous melanoma: a <scp>B</scp> io <scp>G</scp> eno <scp>MEL</scp> study. Pigment Cell and Melanoma Research, 2014, 27, 234-243.	1.5	25
31	Serum 25-hydroxyvitamin D3 levels and vitamin D receptor variants in melanoma patients from the Mediterranean area of Barcelona. BMC Medical Genetics, 2013, 14, 26.	2.1	24
32	CDKN2Amutations in melanoma families from Uruguay. British Journal of Dermatology, 2009, 161, 536-541.	1.4	20
33	Role of <i><scp>CPI</scp>â€17</i> in restoring skin homoeostasis in cutaneous field of cancerization: effects of topical application of a filmâ€forming medical device containing photolyase and <scp>UV</scp> filters. Experimental Dermatology, 2013, 22, 494-496.	1.4	19
34	Genetic susceptibility to cutaneous melanoma in southern Switzerland: role of <i>CDKN2A</i> , <i>MC1R</i> and <i>MITF</i> . British Journal of Dermatology, 2016, 175, 1030-1037.	1.4	17
35	Implementation of an open-source robotic platform for SARS-CoV-2 testing by real-time RT-PCR. PLoS ONE, 2021, 16, e0252509.	1.1	17
36	Association between dermoscopic and reflectance confocal microscopy features of cutaneous melanoma with <scp>BRAF</scp> mutational status. Journal of the European Academy of Dermatology and Venereology, 2017, 31, 643-649.	1.3	15

JOAN ANTON PUIG-BUTILLE

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37	Inherited functional variants of the lymphocyte receptor CD5 influence melanoma survival. International Journal of Cancer, 2016, 139, 1297-1302.	2.3	14
38	Monitoring of Donorâ€Derived Cellâ€Free DNA by Short Tandem Repeats: Concentration of Total Cellâ€Free DNA and Fragment Size for Acute Rejection Risk Assessment in Liver Transplantation. Liver Transplantation, 2022, 28, 257-268.	1.3	13
39	Mutation of the tumour suppressor p33 ING1 b is rare in melanoma. British Journal of Dermatology, 2006, 155, 94-99.	1.4	12
40	<i>IRF4</i> rs12203592 functional variant and melanoma survival. International Journal of Cancer, 2017, 140, 1845-1849.	2.3	11
41	Multiple Primary Acral Melanomas in Two Young Caucasian Patients. Dermatology, 2014, 228, 307-310.	0.9	10
42	Clinical, Epidemiological, and Molecular Heterogeneity in AcralÂMelanoma. Journal of Investigative Dermatology, 2018, 138, 254-255.	0.3	10
43	Genetic variations of patients with familial or multiple melanoma in Southern Brazil. Journal of the European Academy of Dermatology and Venereology, 2013, 27, e179-85.	1.3	8
44	Novedades en genética del melanoma. Piel, 2006, 21, 272-274.	0.0	7
45	Clinical and Histopathological Characteristics between Familial and Sporadic Melanoma in Barcelona, Spain. Journal of Clinical & Experimental Dermatology Research, 2014, 05, 231.	0.1	7
46	Detection of cellâ€free circulating <scp> <i>BRAF</i> ^V </scp> ^{600E} by droplet digital polymerase chain reaction in patients with and without melanoma under dermatological surveillance. British Journal of Dermatology, 2020, 182, 382-389.	1.4	7
47	Molecular characterization of human cutaneous melanoma-derived cell lines. Anticancer Research, 2012, 32, 1245-51.	0.5	7
48	Novel P397S <i>MAPT</i> variant associated with late onset and slow progressive frontotemporal dementia. Annals of Clinical and Translational Neurology, 2019, 6, 1559-1565.	1.7	6
49	Cell-free DNA concentration and fragment size fraction correlate with FDG PET/CT-derived parameters in NSCLC patients. European Journal of Nuclear Medicine and Molecular Imaging, 2021, 48, 3631-3642.	3.3	6
50	A Common Variant in the MC1R Gene (p.V92M) is associated with Alzheimer's Disease Risk. Journal of Alzheimer's Disease, 2017, 56, 1065-1074.	1.2	5
51	The p. R151C Polymorphism in MC1R Gene Modifies the Age of Onset in Spanish Huntington's Disease Patients. Molecular Neurobiology, 2017, 54, 3906-3910.	1.9	5
52	Melanocortinâ€1 receptor (<i>MC1R</i>) genotypes do not correlate with size in two cohorts of mediumâ€toâ€giant congenital melanocytic nevi. Pigment Cell and Melanoma Research, 2020, 33, 685-694.	1.5	5
53	Genomic expression differences between cutaneous cells from red hair color individuals and black hair color individuals based on bioinformatic analysis. Oncotarget, 2017, 8, 11589-11599.	0.8	5
54	Genome-wide linkage analysis in Spanish melanoma-prone families identifies a new familial melanoma susceptibility locus at 11q. European Journal of Human Genetics, 2018, 26, 1188-1193.	1.4	4

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55	Molecular characterization of advanced non-small cell lung cancer patients by cfDNA analysis: experience from routine laboratory practice. Journal of Thoracic Disease, 2021, 13, 1658-1670.	0.6	4
56	Reply. Annals of Neurology, 2016, 79, 161-163.	2.8	3
57	Technical Evaluation of the COBAS EGFR Semiquantitative Index (SQI) for Plasma cfDNA Testing in NSCLC Patients with EGFR Exon 19 Deletions. Diagnostics, 2021, 11, 1319.	1.3	3
58	Gene Expression Microarray: Technical Fundamentals and Data Analysis. , 2021, , 291-312.		3
59	Time and tumor type (primary or metastatic) do not influence the detection of BRAF/NRAS mutations in formalin fixed paraffin embedded samples from melanomas. Clinical Chemistry and Laboratory Medicine, 2016, 54, 1733-1738.	1.4	2
60	Influence of germline genetic variants on dermoscopic features of melanoma. Pigment Cell and Melanoma Research, 2021, 34, 618-628.	1.5	2
61	Reply. Annals of Neurology, 2015, 78, 153-154.	2.8	1
62	Melanocortin 1 receptor: Parkinson's disease, melanoma risk, and neuroprotection. , 2020, , 189-203.		1
63	Oncogenic properties via <scp>MAPK</scp> signaling of the <scp>SOX5â€RAF1</scp> fusion gene identified in a <i>wildâ€type</i> <scp>NRAS</scp> / <scp>BRAF</scp> giant congenital nevus. Pigment Cell and Melanoma Research, 2022, 35, 450-460.	1.5	1
64	Discrepant mutational status between naevi and melanomas in naevus-associated melanomas: about mutation-specific immunohistochemistry: reply from the authors. British Journal of Dermatology, 2016, 175, 435-435.	1.4	0
65	Reply. Annals of Neurology, 2016, 79, 868-868.	2.8	Ο