

# Joan Anton Puig-Butille

## List of Publications by Year in descending order

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Version: 2024-02-01

65  
papers

2,391  
citations

249298

26  
h-index

242451

47  
g-index

68  
all docs

68  
docs citations

68  
times ranked

5241  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	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
3	Oncogenic properties via <i>MAPK</i> signaling of the <i>SOX5</i> â€ <i>RAF1</i> fusion gene identified in a <i>wild-type</i> <i>NRAS</i> / <i>BRAF</i> giant congenital nevus. Pigment Cell and Melanoma Research, 2022, 35, 450-460.	1.5	1
4	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
5	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
6	Transcriptomic and Genetic Associations between Alzheimerâ€™s Disease, Parkinsonâ€™s Disease, and Cancer. Cancers, 2021, 13, 2990.	1.7	26
7	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
8	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
9	Gene Expression Microarray: Technical Fundamentals and Data Analysis. , 2021, , 291-312.		3
10	Influence of germline genetic variants on dermoscopic features of melanoma. Pigment Cell and Melanoma Research, 2021, 34, 618-628.	1.5	2
11	Detection of cellâ€free circulating <i>BRAF</i> <sup>V</sup> <i>600E</i> 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
12	Melanocortinâ€1 receptor ( <i>MC1R</i> ) genotypes do not correlate with size in two cohorts of mediumâ€giant congenital melanocytic nevi. Pigment Cell and Melanoma Research, 2020, 33, 685-694.	1.5	5
13	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
14	Melanocortin 1 receptor: Parkinson's disease, melanoma risk, and neuroprotection. , 2020, , 189-203.		1
15	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
16	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
17	Genetic Abnormalities in Large to Giant Congenital Nevi: Beyond <i>NRAS</i> Mutations. Journal of Investigative Dermatology, 2019, 139, 900-908.	0.3	67
18	Association of the <i>POT1</i> Germline Missense Variant p.I78T With Familial Melanoma. JAMA Dermatology, 2019, 155, 604.	2.0	34

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19	<i>PTEN</i> germline mutations but not <i>TERT</i> promoter mutations are implicated in melanoma susceptibility in a large cohort of Spanish melanoma families. <i>British Journal of Dermatology</i> , 2019, 181, 105-113.	1.4	37
20	Clinical, Epidemiological, and Molecular Heterogeneity in Acral Melanoma. <i>Journal of Investigative Dermatology</i> , 2018, 138, 254-255.	0.3	10
21	Genome-wide linkage analysis in Spanish melanoma-prone families identifies a new familial melanoma susceptibility locus at 11q. <i>European Journal of Human Genetics</i> , 2018, 26, 1188-1193.	1.4	4
22	Melanocortin 1 receptor ( <i>MC1R</i> ) polymorphisms influence on size and dermoscopic features of nevi. <i>Pigment Cell and Melanoma Research</i> , 2018, 31, 39-50.	1.5	28
23	<i>IRF4</i> rs12203592 functional variant and melanoma survival. <i>International Journal of Cancer</i> , 2017, 140, 1845-1849.	2.3	11
24	A Common Variant in the <i>MC1R</i> Gene (p.V92M) is associated with Alzheimer's Disease Risk. <i>Journal of Alzheimer's Disease</i> , 2017, 56, 1065-1074.	1.2	5
25	<i>AURKA</i> Overexpression Is Driven by <i>FOXM1</i> and <i>MAPK/ERK</i> Activation in Melanoma Cells Harboring <i>BRAF</i> or <i>NRAS</i> Mutations: Impact on Melanoma Prognosis and Therapy. <i>Journal of Investigative Dermatology</i> , 2017, 137, 1297-1310.	0.3	40
26	The p. R151C Polymorphism in <i>MC1R</i> Gene Modifies the Age of Onset in Spanish Huntington's Disease Patients. <i>Molecular Neurobiology</i> , 2017, 54, 3906-3910.	1.9	5
27	Association between dermoscopic and reflectance confocal microscopy features of cutaneous melanoma with <i>BRAF</i> mutational status. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2017, 31, 643-649.	1.3	15
28	Genomic expression differences between cutaneous cells from red hair color individuals and black hair color individuals based on bioinformatic analysis. <i>Oncotarget</i> , 2017, 8, 11589-11599.	0.8	5
29	Discrepant mutational status between naevi and melanomas in naevus-associated melanomas: about mutation-specific immunohistochemistry: reply from the authors. <i>British Journal of Dermatology</i> , 2016, 175, 435-435.	1.4	0
30	Inherited functional variants of the lymphocyte receptor <i>CD5</i> influence melanoma survival. <i>International Journal of Cancer</i> , 2016, 139, 1297-1302.	2.3	14
31	Time and tumor type (primary or metastatic) do not influence the detection of <i>BRAF/NRAS</i> mutations in formalin fixed paraffin embedded samples from melanomas. <i>Clinical Chemistry and Laboratory Medicine</i> , 2016, 54, 1733-1738.	1.4	2
32	Genetic susceptibility to cutaneous melanoma in southern Switzerland: role of <i>CDKN2A</i> , <i>MC1R</i> and <i>MITF</i> . <i>British Journal of Dermatology</i> , 2016, 175, 1030-1037.	1.4	17
33	Reply. <i>Annals of Neurology</i> , 2016, 79, 161-163.	2.8	3
34	Reply. <i>Annals of Neurology</i> , 2016, 79, 868-868.	2.8	0
35	Prevalence of <i>MITF</i> p.E318K in Patients With Melanoma Independent of the Presence of <i>CDKN2A</i> Causative Mutations. <i>JAMA Dermatology</i> , 2016, 152, 405.	2.0	41
36	Characterization of individuals at high risk of developing melanoma in Latin America: bases for genetic counseling in melanoma. <i>Genetics in Medicine</i> , 2016, 18, 727-736.	1.1	31

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37	Mutational status of naevus-associated melanomas. British Journal of Dermatology, 2015, 173, 671-680.	1.4	42
38	Reply. Annals of Neurology, 2015, 78, 153-154.	2.8	1
39	The <i>MC1R</i> melanoma risk variant p.R160W is associated with Parkinson disease. Annals of Neurology, 2015, 77, 889-894.	2.8	52
40	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. Nature Genetics, 2015, 47, 987-995.	9.4	218
41	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
42	Update in genetic susceptibility in melanoma. Annals of Translational Medicine, 2015, 3, 210.	0.7	100
43	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
44	Clinical and Histopathological Characteristics between Familial and Sporadic Melanoma in Barcelona, Spain. Journal of Clinical & Experimental Dermatology Research, 2014, 05, 231.	0.1	7
45	Multiple Primary Acral Melanomas in Two Young Caucasian Patients. Dermatology, 2014, 228, 307-310.	0.9	10
46	An inherited variant in the gene coding for vitamin D-binding protein and survival from cutaneous melanoma: a BIOGENO-MEL study. Pigment Cell and Melanoma Research, 2014, 27, 234-243.	1.5	25
47	Dermoscopic criteria associated with <i>BRAF</i> and <i>NRAS</i> mutation status in primary cutaneous melanoma. British Journal of Dermatology, 2014, 171, 754-759.	1.4	26
48	The Effect on Melanoma Risk of Genes Previously Associated With Telomere Length. Journal of the National Cancer Institute, 2014, 106, .	3.0	109
49	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
50	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
51	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
52	TERT Promoter Mutation Status as an Independent Prognostic Factor in Cutaneous Melanoma. Journal of the National Cancer Institute, 2014, 106, .	3.0	204
53	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
54	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

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55	Benefits of oral <i>Polypodium Leucotomos</i> extract in MM high-risk patients. Journal of the European Academy of Dermatology and Venereology, 2013, 27, 1095-1100.	1.3	34
56	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
57	A variant in FTO shows association with melanoma risk not due to BMI. Nature Genetics, 2013, 45, 428-432.	9.4	111
58	Role of <i>CPI-17</i> in restoring skin homeostasis in cutaneous field of cancerization: effects of topical application of a film-forming medical device containing photolyase and UV filters. Experimental Dermatology, 2013, 22, 494-496.	1.4	19
59	Benefits of total body photography and digital dermatoscopy (two-step method of digital) Tj ETQq1 1 0.784314 rgBT /Overlock 10 American Academy of Dermatology, 2012, 67, e17-e27.	0.6	176
60	Molecular characterization of human cutaneous melanoma-derived cell lines. Anticancer Research, 2012, 32, 1245-51.	0.5	7
61	Genome-wide association study identifies three new melanoma susceptibility loci. Nature Genetics, 2011, 43, 1108-1113.	9.4	230
62	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
63	<i>CDKN2A</i> mutations in melanoma families from Uruguay. British Journal of Dermatology, 2009, 161, 536-541.	1.4	20
64	Novedades en genética del melanoma. Piel, 2006, 21, 272-274.	0.0	7
65	Mutation of the tumour suppressor p33 ING1 b is rare in melanoma. British Journal of Dermatology, 2006, 155, 94-99.	1.4	12