## Alberto Cascon

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	International initiative for a curated <i>SDHB</i> variant database improving the diagnosis of hereditary paraganglioma and pheochromocytoma. Journal of Medical Genetics, 2022, 59, 785-792.	1.5	5
2	<scp>CIBERER</scp> : Spanish national network for research on rare diseases: A highly productive collaborative initiative. Clinical Genetics, 2022, 101, 481-493.	1.0	9
3	Pheochromocytoma and Paraganglioma. , 2021, , 101-137.		0
4	Analysis of Telomere Maintenance Related Genes Reveals NOP10 as a New Metastatic-Risk Marker in Pheochromocytoma/Paraganglioma. Cancers, 2021, 13, 4758.	1.7	14
5	Prevalence of pathogenic germline variants in patients with metastatic renal cell carcinoma. Genetics in Medicine, 2021, 23, 698-704.	1.1	9
6	Hsaâ€miRâ€139â€5p is a prognostic thyroid cancer marker involved in HNRNPFâ€mediated alternative splicing. International Journal of Cancer, 2020, 146, 521-530.	2.3	29
7	PTEN expression and mutations in TSC1 , TSC2 and MTOR are associated with response to rapalogs in patients with renal cell carcinoma. International Journal of Cancer, 2020, 146, 1435-1444.	2.3	14
8	Novel DNMT3A Germline Variant in a Patient with Multiple Paragangliomas and Papillary Thyroid Carcinoma. Cancers, 2020, 12, 3304.	1.7	5
9	A Novel Approach for the Identification of Pharmacogenetic Variants in MT-RNR1 through Next-Generation Sequencing Off-Target Data. Journal of Clinical Medicine, 2020, 9, 2082.	1.0	0
10	CD133 Expression in Medullary Thyroid Cancer Cells Identifies Patients with Poor Prognosis. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3548-3561.	1.8	5
11	Molecular characterization of chromophobe renal cell carcinoma reveals mTOR pathway alterations in patients with poor outcome. Modern Pathology, 2020, 33, 2580-2590.	2.9	29
12	Metabolome-guided genomics to identify pathogenic variants in isocitrate dehydrogenase, fumarate hydratase, and succinate dehydrogenase genes in pheochromocytoma and paraganglioma. Genetics in Medicine, 2019, 21, 705-717.	1.1	60
13	Integrative multi-omics analysis identifies a prognostic miRNA signature and a targetable miR-21-3p/TSC2/mTOR axis in metastatic pheochromocytoma/paraganglioma. Theranostics, 2019, 9, 4946-4958.	4.6	54
14	Pheochromocytomas and Paragangliomas: Bypassing Cellular Respiration. Cancers, 2019, 11, 683.	1.7	22
15	Recurrent Germline DLST Mutations in Individuals with Multiple Pheochromocytomas and Paragangliomas. American Journal of Human Genetics, 2019, 104, 651-664.	2.6	51
16	Concomitant Medications and Risk of Chemotherapy-Induced Peripheral Neuropathy. Oncologist, 2019, 24, e784-e792.	1.9	20
17	Biallelic <i>TSC2</i> Mutations in a Patient With Chromophobe Renal Cell Carcinoma Showing Extraordinary Response to Temsirolimus. Journal of the National Comprehensive Cancer Network: JNCCN, 2018, 16, 352-358.	2.3	18
18	Advanced sporadic renal epithelioid angiomyolipoma: case report of an extraordinary response to sirolimus linked to TSC2 mutation. BMC Cancer, 2018, 18, 561.	1.1	13

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19	Role of MDH2 pathogenic variant in pheochromocytoma and paraganglioma patients. Genetics in Medicine, 2018, 20, 1652-1662.	1.1	45
20	Gain-of-function mutations in DNMT3A in patients with paraganglioma. Genetics in Medicine, 2018, 20, 1644-1651.	1.1	73
21	Molecular Genetics of Pheochromocytoma and Paraganglioma. , 2017, , 15-45.		0
22	Targeted Exome Sequencing of Krebs Cycle Genes Reveals Candidate Cancer–Predisposing Mutations in Pheochromocytomas and Paragangliomas. Clinical Cancer Research, 2017, 23, 6315-6324.	3.2	73
23	Exceptional Response to Temsirolimus in a Metastatic Clear Cell Renal Cell Carcinoma With an Early Novel MTOR -Activating Mutation. Journal of the National Comprehensive Cancer Network: JNCCN, 2017, 15, 1310-1315.	2.3	16
24	PheoSeq. Journal of Molecular Diagnostics, 2017, 19, 575-588.	1.2	63
25	Pathological and Genetic Characterization of Bilateral Adrenomedullary Hyperplasia in a Patient with Germline MAX Mutation. Endocrine Pathology, 2017, 28, 302-307.	5.2	25
26	Consensus Statement on next-generation-sequencing-based diagnostic testing of hereditary phaeochromocytomas and paragangliomas. Nature Reviews Endocrinology, 2017, 13, 233-247.	4.3	198
27	Targeted Sequencing Reveals Low-Frequency Variants in <i>EPHA</i> Genes as Markers of Paclitaxel-Induced Peripheral Neuropathy. Clinical Cancer Research, 2017, 23, 1227-1235.	3.2	16
28	ATRX driver mutation in a composite malignant pheochromocytoma. Cancer Genetics, 2016, 209, 272-277.	0.2	24
29	Alkali Pretreatment for Producing Ethanol from Lignocellulosics. Journal of the Japan Petroleum Institute, 2015, 58, 119-127.	0.4	1
30	Whole-Exome Sequencing Identifies MDH2 as a New Familial Paraganglioma Gene. Journal of the National Cancer Institute, 2015, 107, .	3.0	143
31	Functional and in silico assessment of MAX variants of unknown significance. Journal of Molecular Medicine, 2015, 93, 1247-1255.	1.7	25
32	DNA Methylation Profiling in Pheochromocytoma and Paraganglioma Reveals Diagnostic and Prognostic Markers. Clinical Cancer Research, 2015, 21, 3020-3030.	3.2	53
33	Recommendations for somatic and germline genetic testing of single pheochromocytoma and paraganglioma based on findings from a series of 329 patients. Journal of Medical Genetics, 2015, 52, 647-656.	1.5	102
34	Whole-Exome Sequencing Reveals Defective <i>CYP3A4</i> Variants Predictive of Paclitaxel Dose-Limiting Neuropathy. Clinical Cancer Research, 2015, 21, 322-328.	3.2	61
35	High frequency and founder effect of the CYP3A4*20 loss-of-function allele in the Spanish population classifies CYP3A4 as a polymorphic enzyme. Pharmacogenomics Journal, 2015, 15, 288-292.	0.9	48
36	VEGF, VEGFR3, and PDGFRB Protein Expression Is Influenced by <i>RAS</i> Mutations in Medullary Thyroid Carcinoma. Thyroid, 2014, 24, 1251-1255.	2.4	18

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37	Germline mutations in FH confer predisposition to malignant pheochromocytomas and paragangliomas. Human Molecular Genetics, 2014, 23, 2440-2446.	1.4	316
38	Differential Gene Expression of Medullary Thyroid Carcinoma Reveals Specific Markers Associated with Genetic Conditions. American Journal of Pathology, 2013, 182, 350-362.	1.9	35
39	Usefulness of Negative and Weak–Diffuse Pattern of SDHB Immunostaining in Assessment of SDH Mutations in Paragangliomas and Pheochromocytomas. Endocrine Pathology, 2013, 24, 199-205.	5.2	42
40	Tumoral EPAS1 (HIF2A) mutations explain sporadic pheochromocytoma and paraganglioma in the absence of erythrocytosis. Human Molecular Genetics, 2013, 22, 2169-2176.	1.4	142
41	Genetics of pheochromocytoma and paraganglioma in Spanish pediatric patients. Endocrine-Related Cancer, 2013, 20, L1-L6.	1.6	44
42	Integrative analysis of miRNA and mRNA expression profiles in pheochromocytoma and paraganglioma identifies genotype-specific markers and potentially regulated pathways. Endocrine-Related Cancer, 2013, 20, 477-493.	1.6	52
43	Influence of RET mutations on the expression of tyrosine kinases in medullary thyroid carcinoma. Endocrine-Related Cancer, 2013, 20, 611-619.	1.6	17
44	<i>MAX</i> Mutations Cause Hereditary and Sporadic Pheochromocytoma and Paraganglioma. Clinical Cancer Research, 2012, 18, 2828-2837.	3.2	277
45	Regulatory Polymorphisms in β-Tubulin IIa Are Associated with Paclitaxel-Induced Peripheral Neuropathy. Clinical Cancer Research, 2012, 18, 4441-4448.	3.2	61
46	Hematologic β-Tubulin VI Isoform Exhibits Genetic Variability That Influences Paclitaxel Toxicity. Cancer Research, 2012, 72, 4744-4752.	0.4	26
47	MAX and MYC: A Heritable Breakup. Cancer Research, 2012, 72, 3119-3124.	0.4	144
48	From Transcriptional Profiling to Tumor Biology in Pheochromocytoma and Paraganglioma. Endocrine Pathology, 2012, 23, 15-20.	5.2	16
49	Exome sequencing identifies MAX mutations as a cause of hereditary pheochromocytoma. Nature Genetics, 2011, 43, 663-667.	9.4	478
50	Detection of the first gross CDC73 germline deletion in an HPTâ€JT syndrome family. Genes Chromosomes and Cancer, 2011, 50, 922-929.	1.5	41
51	Polymorphisms in cytochromes P450 2C8 and 3A5 are associated with paclitaxel neurotoxicity. Pharmacogenomics Journal, 2011, 11, 121-129.	0.9	112
52	Tumoral and tissueâ€specific expression of the major human βâ€ŧubulin isotypes. Cytoskeleton, 2010, 67, 214-223.	1.0	221
53	Are we overestimating the penetrance of mutations in SDHB?. Human Mutation, 2010, 31, 761-762.	1.1	64
54	Research Resource: Transcriptional Profiling Reveals Different Pseudohypoxic Signatures in SDHB and VHL-Related Pheochromocytomas. Molecular Endocrinology, 2010, 24, 2382-2391.	3.7	179

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55	Brick1 Is an Essential Regulator of Actin Cytoskeleton Required for Embryonic Development and Cell Transformation. Cancer Research, 2010, 70, 9349-9359.	0.4	31
56	Overexpression and activation of EGFR and VEGFR2 in medullary thyroid carcinomas is related to metastasis. Endocrine-Related Cancer, 2010, 17, 7-16.	1.6	108
57	Systematic comparison of sporadic and syndromic pancreatic islet cell tumors. Endocrine-Related Cancer, 2010, 17, 875-883.	1.6	29
58	Spectrum and Prevalence of <i>FP/TMEM127</i> Gene Mutations in Pheochromocytomas and Paragangliomas. JAMA - Journal of the American Medical Association, 2010, 304, 2611.	3.8	174
59	SDHAF2 mutations in familial and sporadic paraganglioma and phaeochromocytoma. Lancet Oncology, The, 2010, 11, 366-372.	5.1	256
60	Head and Neck Paragangliomas in Von Hippel-Lindau Disease and Multiple Endocrine Neoplasia Type 2. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1938-1944.	1.8	112
61	Genetics of Pheochromocytoma and Paraganglioma in Spanish Patients. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1701-1705.	1.8	120
62	Rationalization of Genetic Testing in Patients with Apparently Sporadic Pheochromocytoma/Paraganglioma. Hormone and Metabolic Research, 2009, 41, 672-675.	0.7	41
63	Clinical Predictors for Germline Mutations in Head and Neck Paraganglioma Patients: Cost Reduction Strategy in Genetic Diagnostic Process as Fall-Out. Cancer Research, 2009, 69, 3650-3656.	0.4	178
64	Determination of CYP2D6 gene copy number by multiplex polymerase chain reaction analysis. Analytical Biochemistry, 2009, 389, 74-76.	1.1	14
65	Cellular senescence bypass screen identifies new putative tumor suppressor genes. Oncogene, 2008, 27, 1961-1970.	2.6	59
66	<i>SDHC</i> mutation in an elderly patient without familial antecedents. Clinical Endocrinology, 2008, 69, 906-910.	1.2	37
67	Gene expression analysis in pheochromocytoma – searching for new pathways involved in the hereditary susceptibility and the malignant outcome. European Journal of Cancer, Supplement, 2008, 6, 160-161.	2.2	Ο
68	Extra-adrenal and adrenal pheochromocytomas associated with a germline SDHC mutation. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 111-115.	2.9	95
69	Evaluation of a functional epigenetic approach to identify promoter region methylation in phaeochromocytoma and neuroblastoma. Endocrine-Related Cancer, 2008, 15, 777-786.	1.6	25
70	PPP1CA contributes to the senescence program induced by oncogenic Ras. Carcinogenesis, 2007, 29, 491-499.	1.3	61
71	Molecular characterisation of a common SDHB deletion in paraganglioma patients. Journal of Medical Genetics, 2007, 45, 233-238.	1.5	69
72	GermlineNF1Mutational Spectra and Loss-of-Heterozygosity Analyses in Patients with Pheochromocytoma and Neurofibromatosis Type 1. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2784-2792.	1.8	126

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73	Association Study of 69 Genes in the Ret Pathway Identifies Low-penetrance Loci in Sporadic Medullary Thyroid Carcinoma. Cancer Research, 2007, 67, 9561-9567.	0.4	36
74	Evidence of MEN-2 in the Original Description of Classic Pheochromocytoma. New England Journal of Medicine, 2007, 357, 1311-1315.	13.9	95
75	Cytochrome P450 3A5 is highly expressed in normal prostate cells but absent in prostate cancer. Endocrine-Related Cancer, 2007, 14, 645-654.	1.6	34
76	Loss of the actin regulator HSPC300 results in clear cell renal cell carcinoma protection in Von Hippel-Lindau patients. Human Mutation, 2007, 28, 613-621.	1.1	41
77	Molecular pathways linking the pheochromocytoma susceptibility genes—response. Pediatric Blood and Cancer, 2007, 49, 1052-1053.	0.8	0
78	Immunohistochemical classification of non-BRCA1/2 tumors identifies different groups that demonstrate the heterogeneity of BRCAX families. Modern Pathology, 2007, 20, 1298-1306.	2.9	48
79	Pediatric paraganglioma: An early manifestation of an adult disease secondary to germline mutations. Pediatric Blood and Cancer, 2006, 47, 785-789.	0.8	20
80	RFLP-PCR analysis of the aroA gene as a taxonomic tool for the genus Aeromonas. FEMS Microbiology Letters, 2006, 156, 199-204.	0.7	10
81	GrossSDHB deletions in patients with paraganglioma detected by multiplex PCR: A possible hot spot?. Genes Chromosomes and Cancer, 2006, 45, 213-219.	1.5	73
82	About the origin and development of hereditary conventional renal cell carcinoma in a four-generation t(3;8)(p14.1;q24.23) family. European Journal of Human Genetics, 2005, 13, 570-578.	1.4	15
83	A novel candidate region linked to development of both pheochromocytoma and head/neck paraganglioma. Genes Chromosomes and Cancer, 2005, 42, 260-268.	1.5	33
84	Novel Pheochromocytoma Susceptibility Loci Identified by Integrative Genomics. Cancer Research, 2005, 65, 9651-9658.	0.4	88
85	Succinate Dehydrogenase D Variants Do Not Constitute a Risk Factor for Developing C Cell Hyperplasia or Sporadic Medullary Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 2127-2130.	1.8	12
86	Epigenetic analysis of HIC1, CASP8, FLIP, TSP1, DCR1, DCR2, DR4, DR5, KvDMR1, H19 and preferential 11p15.5 maternal-allele loss in von Hippel-Lindau and sporadic phaeochromocytomas. Endocrine-Related Cancer, 2005, 12, 161-172.	1.6	56
87	Expression Profiling of T-Cell Lymphomas Differentiates Peripheral and Lymphoblastic Lymphomas and Defines Survival Related Genes. Clinical Cancer Research, 2004, 10, 4971-4982.	3.2	88
88	Genetic and epigenetic profile of sporadic pheochromocytomas. Journal of Medical Genetics, 2004, 41, 30e-30.	1.5	42
89	Genetic characterization and structural analysis of VHL Spanish families to define genotype-phenotype correlations. Human Mutation, 2004, 23, 160-169.	1.1	28
90	G12S and H50R variations are polymorphisms in the SDHD gene. Genes Chromosomes and Cancer, 2003, 37, 220-221.	1.5	31

6

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91	Mutational and gross deletion study of the MEN1 gene and correlation with clinical features in Spanish patients. Journal of Medical Genetics, 2003, 40, 72e-72.	1.5	48
92	SDHB mutation analysis in familial and sporadic phaeochromocytoma identifies a novel mutation. Journal of Medical Genetics, 2002, 39, 64e-64.	1.5	15
93	A rapid and easy method for multiple endocrine neoplasia type 1 mutation detection using conformation-sensitive gel electrophoresis. Journal of Human Genetics, 2002, 47, 190-195.	1.1	9
94	Identification of novel SDHD mutations in patients with phaeochromocytoma and/or paraganglioma. European Journal of Human Genetics, 2002, 10, 457-461.	1.4	60
95	Cloning, characterization, and insertional inactivation of a major extracellular serine protease gene with elastolytic activity from Aeromonas hydrophila. Journal of Fish Diseases, 2000, 23, 49-59.	0.9	19
96	A Major Secreted Elastase Is Essential for Pathogenicity of Aeromonas hydrophila. Infection and Immunity, 2000, 68, 3233-3241.	1.0	136
97	RFLP-PCR analysis of the aroA gene as a taxonomic tool for the genus Aeromonas. FEMS Microbiology Letters, 1997, 156, 199-204.	0.7	19