Eladio AndrÃ©s Velasco

List of Publications by Year
in descending order

56 papers	1,706 citations	257450 24 h-index	289244 40 g-index
60 all docs	60 docs citations	60 times ranked	2362 citing authors

#	Article	IF	Citations
1	Splicing predictions, minigene analyses, and <scp>ACMG</scp>â€<scp>AMP</scp> clinical classification of 42 germline <scp>PALB2</scp> spliceâ€site variants. Journal of Pathology, 2022, 256, 321-334.	4.5	16
2	Germline Variants of CYBA and TRPM4 Predispose to Familial Colorectal Cancer. Cancers, 2022, 14, 670.	3.7	11
3	Minigeneâ€based splicing analysis and <scp>ACMG</scp>/<scp>AMP</scp>â€based tentative classification of 56 <scp>ATM</scp> variants. Journal of Pathology, 2022, 258, 83-101.	4.5	5
4	Minigene Splicing Assays Identify 20 Spliceogenic Variants of the Breast/Ovarian Cancer Susceptibility Gene RAD51C. Cancers, 2022, 14, 2960.	3.7	3
5	RAD51D Aberrant Splicing in Breast Cancer: Identification of Splicing Regulatory Elements and Minigene-Based Evaluation of 53 DNA Variants. Cancers, 2021, 13, 2845.	3.7	10
6	Identification of a truncated Î²1-chimaerin variant that inactivates nuclear Rac1. Journal of Biological Chemistry, 2020, 295, 1300-1314.	3.4	3
7	Comprehensive Functional Characterization and Clinical Interpretation of 20 Splice-Site Variants of the RAD51C Gene. Cancers, 2020, 12, 3771.	3.7	10
8	UGT1A1 Variants c.864+5G>T and c.996+2_996+5del of a Crigler-Najjar Patient Induce Aberrant Splicing in Minigene Assays. Frontiers in Genetics, 2020, 11, 169.	2.3	9
9	Identification of a truncated Î²1-chimaerin variant that inactivates nuclear Rac1. Journal of Biological Chemistry, 2020, 295, 1300-1314.	3.4	2
10	Association Between Bat Vitamin D Receptor 3â€² Haplotypes and Vitamin D Levels at Baseline and a Lower Response After Increased Vitamin D Supplementation and Exposure to Sunlight. International Journal for Vitamin and Nutrition Research, 2020, 90, 290-294.	1.5	2
11	Minigene Splicing Assays Identify 12 Spliceogenic Variants of BRCA2 Exons 14 and 15. Frontiers in Genetics, 2019, 10, 503.	2.3	23
12	Alternative splicing and ACMG-AMP-2015-based classification of PALB2 genetic variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 453-460.	3.2	30
13	Misâ€splicing in breast cancer: identification of pathogenic BRCA2 variants by systematic minigene assays. Journal of Pathology, 2019, 248, 409-420.	4.5	26
14	Genetic dissection of the BRCA2 promoter and transcriptional impact of DNA variants. Breast Cancer Research and Treatment, 2018, 171, 53-63.	2.5	8
15	Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing:BRCA2c.7976+5GÂ>ÂT as a case study. Human Mutation, 2018, 39, 1155-1160.	2.5	12
16	Functional Analyses of a Novel Splice Variant in the CHD7 Gene, Found by Next Generation Sequencing, Confirm Its Pathogenicity in a Spanish Patient and Diagnose Him with CHARGE Syndrome. Frontiers in Genetics, 2018, 9, 7.	2.3	19
17	Identification of Eight Spliceogenic Variants in BRCA2 Exon 16 by Minigene Assays. Frontiers in Genetics, 2018, 9, 188.	2.3	21
18	The Effect of Genistein Supplementation on Vitamin D Levels and Bone Turnover Markers during the Summer in Healthy Postmenopausal Women: Role of Genotypes of Isoflavone Metabolism. Journal of Nutrigenetics and Nutrigenomics, 2017, 10, 139-145.	1.3	9

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19	Functional classification of DNA variants by hybrid minigenes: Identification of 30 spliceogenic variants of BRCA2 exons 17 and 18. PLoS Genetics, 2017, 13, e1006691.	3.5	53
20	Functional Classification of BRCA2 DNA Variants by Splicing Assays in a Large Minigene with 9 Exons. Human Mutation, 2015, 36, 210-221.	2.5	50
21	Severe alpha-1 antitrypsin deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. Respiratory Research, 2014, 15, 125.	3.6	38
22	Capillary Electrophoresis Analysis of Conventional Splicing Assays: IARC Analytical and Clinical Classification of 31BRCA2Genetic Variants. Human Mutation, 2014, 35, 53-57.	2.5	25
23	Genotypeâ€“phenotype correlation in MMR mutation-positive families with Lynch syndrome. International Journal of Colorectal Disease, 2013, 28, 1195-1201.	2.2	19
24	Evaluating the Effect of Unclassified Variants Identified in MMR Genes Using Phenotypic Features, Bioinformatics Prediction, and RNA Assays. Journal of Molecular Diagnostics, 2013, 15, 380-390.	2.8	7
25	The highly prevalent BRCA2 mutation c.2808_2811del (3036delACAA) is located in a mutational hotspot and has multiple origins. Carcinogenesis, 2013, 34, 2505-2511.	2.8	16
26	Analysis of PALB2 Gene in BRCA1/BRCA2 Negative Spanish Hereditary Breast/Ovarian Cancer Families with Pancreatic Cancer Cases. PLoS ONE, 2013, 8, e67538.	2.5	44
27	Comprehensive splicing functional analysis of DNA variants of the BRCA2 gene by hybrid minigenes. Breast Cancer Research, 2012, 14, R87.	5.0	48
28	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. Breast Cancer Research and Treatment, 2012, 132, 307-315.	2.5	50
29	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	2.5	27
30	Characterization of New Founder Alu-Mediated Rearrangements in MSH2 Gene Associated with a Lynch Syndrome Phenotype. Cancer Prevention Research, 2011, 4, 1546-1555.	1.5	17
31	Frequency of Rearrangements in Lynch Syndrome Cases Associated with MSH2: Characterization of a New Deletion Involving both EPCAM and the 5â€² Part of MSH2. Cancer Prevention Research, 2011, 4, 1556-1562.	1.5	30
32	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2010, 119, 221-232.	2.5	56
33	Two founder BRCA2 mutations predispose to breast cancer in young women. Breast Cancer Research and Treatment, 2010, 122, 567-571.	2.5	7
34	BRCA1 5272â€¦1G>A and BRCA2 5374delTATG are founder mutations of high relevance for genetic counselling in breast/ovarian cancer families of Spanish origin. Clinical Genetics, 2010, 77, 60-69.	2.0	12
35	A High Proportion of DNA Variants ofBRCA1andBRCA2Is Associated with Aberrant Splicing in Breast/Ovarian Cancer Patients. Clinical Cancer Research, 2010, 16, 1957-1967.	7.0	122
36	A new strategy to screen MMR genes in Lynch Syndrome: HA-CAE, MLPA and RT-PCR. European Journal of Cancer, 2009, 45, 1485-1493.	2.8	16

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37	Prevalence of CYP2C9 polymorphisms in the south of Europe. Pharmacogenomics Journal, 2009, 9, 306-310.	2.0	28
38	Twenty-three novel BRCA1 and BRCA2 sequence alterations in breast and/or ovarian cancer families of Eastern Spain. Breast Cancer Research and Treatment, 2008, 112, 69-73.	2.5	6
39	The Average Cumulative Risks of Breast and Ovarian Cancer for Carriers of Mutations in BRCA1 and BRCA2 Attending Genetic Counseling Units in Spain. Clinical Cancer Research, 2008, 14, 2861-2869.	7.0	90
40	Heteroduplex analysis by capillary array electrophoresis for rapid mutation detection in large multiexon genes. Nature Protocols, 2007, 2, 237-246.	12.0	47
41	High proportion of novel mutations of BRCA1 and BRCA2 in breast/ovarian cancer patients from Castilla-LeÃ³n (central Spain). Journal of Human Genetics, 2006, 51, 611-617.	2.3	33
42	Calcium-sensing Receptor Gene A986S Polymorphism and Bone Mass in Hypertensive Women. Archives of Medical Research, 2006, 37, 607-611.	3.3	6
43	A haplotype containing thep53polymorphisms Ins16bp and Arg72Pro modifies cancer risk inBRCA2mutation carriers. Human Mutation, 2006, 27, 242-248.	2.5	35
44	Genomic Rearrangements at the BRCA1 Locus in Spanish Families with Breast/Ovarian Cancer. Clinical Chemistry, 2006, 52, 1480-1485.	3.2	60
45	Rapid mutation detection in complex genes by heteroduplex analysis with capillary array electrophoresis. Electrophoresis, 2005, 26, 2539-2552.	2.4	25
46	High-Throughput Mutation Detection Method to Scan BRCA1 and BRCA2 Based on Heteroduplex Analysis by Capillary Array Electrophoresis. Clinical Chemistry, 2004, 50, 313-320.	3.2	28
47	Analysis ofBRCA1andBRCA2genes in Spanish breast/ovarian cancer patients: A high proportion of mutations unique to Spain and evidence of founder effects. Human Mutation, 2003, 22, 301-312.	2.5	154
48	Mutational analysis ofBRCA2in Spanish breast cancer patients from Castilla-Leon: Identification of four novel truncating mutations. Human Mutation, 2003, 21, 448-448.	2.5	19
49	Analysis of the monomeric alphoid sequences in the pericentromeric region of human chromosome 7. Cytogenetic and Genome Research, 1998, 83, 176-181.	1.1	11
50	Identification of de novo deletions at the NF1 gene: no preferential paternal origin and phenotypic analysis of patients. Human Genetics, 1997, 99, 720-726.	3.8	49
51	Molecular Analysis of the SMN and NAIP Genes in Spanish Spinal Muscular Atrophy (SMA) Families and Correlation between Number of Copies of^cBCD541 and SMA Phenotype. Human Molecular Genetics, 1996, 5, 257-263.	2.9	185
52	Linkage disequilibrium between four intragenic polymorphic microsatellites of the NF1 gene and its implications for genetic counselling.. Journal of Medical Genetics, 1996, 33, 590-593.	3.2	9
53	Dinucleotide repeat polymorphism at the D4S2458 locus close to the PKD2 locus on human chromosome 4q. Human Genetics, 1995, 95, 601-2.	3.8	0
54	Dinucleotide repeat polymorphisms at the D5S1356, D5S1357 and D7S1480 loci. Human Molecular Genetics, 1994, 3, 1441-1441.	2.9	2

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55	Characterization of four mutations in the neurofibromatosis type 1 gene by denaturing gradient gel electrophoresis (DGGE). Human Molecular Genetics, 1994, 3, 639-641.	2.9	45
56	Dinucleotide repeat polymorphism between the human C4BPA and C4BPB gene loci (1q32). Human Molecular Genetics, 1992, 1, 552-552.	2.9	3