

# Eladio Andr s Velasco

## List of Publications by Year in descending order

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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	Molecular Analysis of the SMN and NAIP Genes in Spanish Spinal Muscular Atrophy (SMA) Families and Correlation between Number of Copies of <sup>c</sup> BCD541 and SMA Phenotype. <i>Human Molecular Genetics</i> , 1996, 5, 257-263.	2.9	185
2	Analysis of BRCA1 and BRCA2 genes in Spanish breast/ovarian cancer patients: A high proportion of mutations unique to Spain and evidence of founder effects. <i>Human Mutation</i> , 2003, 22, 301-312.	2.5	154
3	A High Proportion of DNA Variants of <i>BRCA1</i> and <i>BRCA2</i> Is Associated with Aberrant Splicing in Breast/Ovarian Cancer Patients. <i>Clinical Cancer Research</i> , 2010, 16, 1957-1967.	7.0	122
4	The Average Cumulative Risks of Breast and Ovarian Cancer for Carriers of Mutations in <i>BRCA1</i> and <i>BRCA2</i> Attending Genetic Counseling Units in Spain. <i>Clinical Cancer Research</i> , 2008, 14, 2861-2869.	7.0	90
5	Genomic Rearrangements at the BRCA1 Locus in Spanish Families with Breast/Ovarian Cancer. <i>Clinical Chemistry</i> , 2006, 52, 1480-1485.	3.2	60
6	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , 2010, 119, 221-232.	2.5	56
7	Functional classification of DNA variants by hybrid minigenes: Identification of 30 spliceogenic variants of BRCA2 exons 17 and 18. <i>PLoS Genetics</i> , 2017, 13, e1006691.	3.5	53
8	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 307-315.	2.5	50
9	Functional Classification of <i>BRCA2</i> DNA Variants by Splicing Assays in a Large Minigene with 9 Exons. <i>Human Mutation</i> , 2015, 36, 210-221.	2.5	50
10	Identification of de novo deletions at the NF1 gene: no preferential paternal origin and phenotypic analysis of patients. <i>Human Genetics</i> , 1997, 99, 720-726.	3.8	49
11	Comprehensive splicing functional analysis of DNA variants of the BRCA2 gene by hybrid minigenes. <i>Breast Cancer Research</i> , 2012, 14, R87.	5.0	48
12	Heteroduplex analysis by capillary array electrophoresis for rapid mutation detection in large multiexon genes. <i>Nature Protocols</i> , 2007, 2, 237-246.	12.0	47
13	Characterization of four mutations in the neurofibromatosis type 1 gene by denaturing gradient gel electrophoresis (DGGE). <i>Human Molecular Genetics</i> , 1994, 3, 639-641.	2.9	45
14	Analysis of PALB2 Gene in BRCA1/BRCA2 Negative Spanish Hereditary Breast/Ovarian Cancer Families with Pancreatic Cancer Cases. <i>PLoS ONE</i> , 2013, 8, e67538.	2.5	44
15	Severe alpha-1 antitrypsin deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. <i>Respiratory Research</i> , 2014, 15, 125.	3.6	38
16	A haplotype containing the <i>p53</i> polymorphisms Ins16bp and Arg72Pro modifies cancer risk in <i>BRCA2</i> mutation carriers. <i>Human Mutation</i> , 2006, 27, 242-248.	2.5	35
17	High proportion of novel mutations of BRCA1 and BRCA2 in breast/ovarian cancer patients from Castilla-La Mancha (central Spain). <i>Journal of Human Genetics</i> , 2006, 51, 611-617.	2.3	33
18	Frequency of Rearrangements in Lynch Syndrome Cases Associated with <i>MSH2</i> : Characterization of a New Deletion Involving both <i>EPCAM</i> and the 5' Part of <i>MSH2</i> . <i>Cancer Prevention Research</i> , 2011, 4, 1556-1562.	1.5	30

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19	Alternative splicing and ACMG-AMP-2015-based classification of PALB2 genetic variants: an ENIGMA report. <i>Journal of Medical Genetics</i> , 2019, 56, 453-460.	3.2	30
20	High-Throughput Mutation Detection Method to Scan BRCA1 and BRCA2 Based on Heteroduplex Analysis by Capillary Array Electrophoresis. <i>Clinical Chemistry</i> , 2004, 50, 313-320.	3.2	28
21	Prevalence of CYP2C9 polymorphisms in the south of Europe. <i>Pharmacogenomics Journal</i> , 2009, 9, 306-310.	2.0	28
22	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. <i>Breast Cancer Research and Treatment</i> , 2011, 127, 671-679.	2.5	27
23	Mis-splicing in breast cancer: identification of pathogenic BRCA2 variants by systematic minigene assays. <i>Journal of Pathology</i> , 2019, 248, 409-420.	4.5	26
24	Rapid mutation detection in complex genes by heteroduplex analysis with capillary array electrophoresis. <i>Electrophoresis</i> , 2005, 26, 2539-2552.	2.4	25
25	Capillary Electrophoresis Analysis of Conventional Splicing Assays: IARC Analytical and Clinical Classification of 31 BRCA2 Genetic Variants. <i>Human Mutation</i> , 2014, 35, 53-57.	2.5	25
26	Minigene Splicing Assays Identify 12 Spliceogenic Variants of BRCA2 Exons 14 and 15. <i>Frontiers in Genetics</i> , 2019, 10, 503.	2.3	23
27	Identification of Eight Spliceogenic Variants in BRCA2 Exon 16 by Minigene Assays. <i>Frontiers in Genetics</i> , 2018, 9, 188.	2.3	21
28	Mutational analysis of BRCA2 in Spanish breast cancer patients from Castilla-Leon: Identification of four novel truncating mutations. <i>Human Mutation</i> , 2003, 21, 448-448.	2.5	19
29	Genotype-phenotype correlation in MMR mutation-positive families with Lynch syndrome. <i>International Journal of Colorectal Disease</i> , 2013, 28, 1195-1201.	2.2	19
30	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. <i>Frontiers in Genetics</i> , 2018, 9, 7.	2.3	19
31	Characterization of New Founder Alu-Mediated Rearrangements in MSH2 Gene Associated with a Lynch Syndrome Phenotype. <i>Cancer Prevention Research</i> , 2011, 4, 1546-1555.	1.5	17
32	A new strategy to screen MMR genes in Lynch Syndrome: HA-CAE, MLPA and RT-PCR. <i>European Journal of Cancer</i> , 2009, 45, 1485-1493.	2.8	16
33	The highly prevalent BRCA2 mutation c.2808_2811del (3036delACAA) is located in a mutational hotspot and has multiple origins. <i>Carcinogenesis</i> , 2013, 34, 2505-2511.	2.8	16
34	Splicing predictions, minigene analyses, and ACMG-AMP clinical classification of 42 germline PALB2 splice-site variants. <i>Journal of Pathology</i> , 2022, 256, 321-334.	4.5	16
35	BRCA1 5272G>A and BRCA2 5374delTATG are founder mutations of high relevance for genetic counselling in breast/ovarian cancer families of Spanish origin. <i>Clinical Genetics</i> , 2010, 77, 60-69.	2.0	12
36	Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing: BRCA2 c.7976+5G>A as a case study. <i>Human Mutation</i> , 2018, 39, 1155-1160.	2.5	12

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37	Analysis of the monomeric alphoid sequences in the pericentromeric region of human chromosome 7. <i>Cytogenetic and Genome Research</i> , 1998, 83, 176-181.	1.1	11
38	Germline Variants of CYBA and TRPM4 Predispose to Familial Colorectal Cancer. <i>Cancers</i> , 2022, 14, 670.	3.7	11
39	Comprehensive Functional Characterization and Clinical Interpretation of 20 Splice-Site Variants of the RAD51C Gene. <i>Cancers</i> , 2020, 12, 3771.	3.7	10
40	RAD51D Aberrant Splicing in Breast Cancer: Identification of Splicing Regulatory Elements and Minigene-Based Evaluation of 53 DNA Variants. <i>Cancers</i> , 2021, 13, 2845.	3.7	10
41	Linkage disequilibrium between four intragenic polymorphic microsatellites of the NF1 gene and its implications for genetic counselling. <i>Journal of Medical Genetics</i> , 1996, 33, 590-593.	3.2	9
42	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. <i>Journal of Nutrigenetics and Nutrigenomics</i> , 2017, 10, 139-145.	1.3	9
43	UGT1A1 Variants c.864+5G>T and c.996+2_996+5del of a Crigler-Najjar Patient Induce Aberrant Splicing in Minigene Assays. <i>Frontiers in Genetics</i> , 2020, 11, 169.	2.3	9
44	Genetic dissection of the BRCA2 promoter and transcriptional impact of DNA variants. <i>Breast Cancer Research and Treatment</i> , 2018, 171, 53-63.	2.5	8
45	Two founder BRCA2 mutations predispose to breast cancer in young women. <i>Breast Cancer Research and Treatment</i> , 2010, 122, 567-571.	2.5	7
46	Evaluating the Effect of Unclassified Variants Identified in MMR Genes Using Phenotypic Features, Bioinformatics Prediction, and RNA Assays. <i>Journal of Molecular Diagnostics</i> , 2013, 15, 380-390.	2.8	7
47	Calcium-sensing Receptor Gene A986S Polymorphism and Bone Mass in Hypertensive Women. <i>Archives of Medical Research</i> , 2006, 37, 607-611.	3.3	6
48	Twenty-three novel BRCA1 and BRCA2 sequence alterations in breast and/or ovarian cancer families of Eastern Spain. <i>Breast Cancer Research and Treatment</i> , 2008, 112, 69-73.	2.5	6
49	Minigene-based splicing analysis and ACMG/AMP-based tentative classification of 56 ATM variants. <i>Journal of Pathology</i> , 2022, 258, 83-101.	4.5	5
50	Dinucleotide repeat polymorphism between the human C4BPA and C4BPB gene loci (1q32). <i>Human Molecular Genetics</i> , 1992, 1, 552-552.	2.9	3
51	Identification of a truncated $\beta$ 21-chimaerin variant that inactivates nuclear Rac1. <i>Journal of Biological Chemistry</i> , 2020, 295, 1300-1314.	3.4	3
52	Minigene Splicing Assays Identify 20 Spliceogenic Variants of the Breast/Ovarian Cancer Susceptibility Gene RAD51C. <i>Cancers</i> , 2022, 14, 2960.	3.7	3
53	Dinucleotide repeat polymorphisms at the D5S1356, D5S1357 and D7S1480 loci. <i>Human Molecular Genetics</i> , 1994, 3, 1441-1441.	2.9	2
54	Identification of a truncated $\beta$ 21-chimaerin variant that inactivates nuclear Rac1. <i>Journal of Biological Chemistry</i> , 2020, 295, 1300-1314.	3.4	2

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55	Association Between Bat Vitamin D Receptor 3â€™ 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
56	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